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AUTOSOMAL DNA AND GENEALOGY: HOW CAN DNA AND TRIANGULATION BE USED TO IDENTIFY, EVALUATE AND PRESENT CONCLUSIONS OF RELATEDNESS?

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How Can DNA and Triangulation Be Used to Identify, Evaluate and Present Conclusions of Relatedness?

by
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Acknowledgements:

Producing a dissertation is a monumental production. This one has been no exception. I have been the beneficiary of many miracles—great and small—in my journey to discover, synthesize, and present this subject matter.

I give thanks to my cousins who have invested in and shared their genetic heritage, enabling my journey in the wilderness of genetic genealogy. I acknowledge the expertise of citizen scientists and academics who have made their findings accessible—oases enabling the journey. I thank family, my supervisor, and friends who have given of their time to read and comment on my work as I have sought the paths others have followed and attempted to blazed my own trails in this wilderness.

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ABSTRACT

DNA genotyping allows consumers to examine their genetic heritage, including (within limits) a record of their ancestry. Computer algorithms can associate genotyped individuals who appear to share a genetic past, but appearances cannot be the basis for declaring genealogical relationships. While parent/child relationships can be established with certainty, other genealogical relationships can only be estimated—even in cases where a genetic relationship is sure to exist. How, then, can autosomal DNA (atDNA) be used to reveal, confirm, or even prove genealogical relationships?

This text presents a methodology for identifying, evaluating, and presenting a conclusion of relatedness utilizing atDNA. This methodology fits firmly within the framework of the genealogical research process, enabling atDNA to be used in proof arguments about relationships through the processes of question asking, information gathering, hypothesis testing, conclusion accepting, and proof explained.

The genetic genealogical community has not been definitive about acceptance criteria for atDNA-based conclusions (i.e., required elements, standards of evaluation, etc.), and more especially triangulated conclusions. It is the author's opinion that triangulation ought to be central to most genetic genealogical proof arguments, yet the literature explaining triangulation fails to make plain the reasons why triangulation is effective. These deficiencies can be addressed by decomposing triangulation into its fundamental building blocks and re-presenting it in the context of the genealogical research process.

This text focuses particularly on how hypothesis testing is used to determine strengths and weaknesses of atDNA-based conclusions. This discussion includes important heuristics that help the genetic genealogist understand the capabilities and limits that accompany this biotechnical genealogical record.

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DEFINITIONS

Allele In the context of a SNP, an allele is one of two or more alternative values that may be found when assaying a SNP.[1]

Assay A procedure for qualitatively assessing or quantitatively measuring the presence or amount of a target entity.[2]

atDNA Autosomal DNA. The autosomes are all of the chromosomes (numbered 1 through 22 from longest to shortest) that are not the sex chromosomes (the X and Y chromosomes).

Base pair A pair of nucleotides (bases). DNA molecules incorporate four nucleotides: adenine (A), guanine (G), cytosine (C) and thymine (T). In DNA, these nucleotides always form up in pairs—A always pairs with T, G always pairs with C—and thus the designation "base pair".

Chr1, …, Chr22 A shorthand for referring to the numbered autosomes—i.e., Chr1 is Chromosome 1, and so on.

cM Centimorgan. A unit of genetic distance. 1 cM corresponds to a 1% chance of a recombination (crossover) event occurring between two locations on the chromosome.[3]

Diploid Regarding cells, refers to the fact that each contains two complete copies of the genome—one copy from each biological parent.[4] See also: *haploid*.

DNA Deoxyribonucleic acid.

Genome A complete haploid set (a single copy) of the chromosomes (genetic information) held by a gamete, microorganism, or multi-cellular organism.[5,6] Humans are diploid and, as such, have two haploid copies of the human genome in each somatic cell—one copy from each biological parent.

Genotype A set of genetic markers (e.g., SNPs or STRs) selected for their likelihood of variation and used to study the genetic makeup of individuals—usually used as a proxy for the whole genome of an individual.[7,8]

HIR Half-identical region. In a diploid subject, a region along a homologous chromosome pair where only one of the two alleles from that pair matches only one of the two alleles from another subject's pair across an entire region (segment), though allowances for differences due to a mutation are generally included. See also: *match*, *segment*.

Haplotype The genotype data for a single chromosome. See also: *genotype*.

Haploid Regarding cells, refers to having a single set of unpaired chromosomes.[9] See also: *genotype*.

IBC Identical by chance. A characterization applied to a *match* when at least one of the sequences used in the comparison does not actually correspond to a real sequence found on either haploid chromosome of the haplotype from which it originated—usually the result of a computer algorithm processing unphased haplotype data. Though this concept seems useful, it is not used extensively in the literature; it is generally lumped into the more generic IBS concept. See also: *IBS*.

IBD Identical by descent. Also: identity by descent. A characterization applied to a *match* when the sequence shared between two or more haplotypes is identical because it was inherited from a recent common ancestor.

IBS Identical by state. A characterization applied to a *match* when the haplotype sequence is identical for reasons other than inheritance by descent; not IBD. Some reasons a *match* may be IBS are because it is IBC, or because it is ancient DNA (common to a population and no longer distinguishable as IBD from a recent ancestor).

ICW group In Common With (ICW) group. Given two persons who match each other, and the list of matches associated with each person, it is the set of matches that is common to both match lists.

Locus (loci) A specific location or position.[10] Each SNP is situated at a known locus. The plural form of the word is *loci*.

Mash-up A mixture or fusion of disparate elements; something created by combining elements from two or more sources.[11,12]

Match Also called: matching segment; shared segment; identical segment. When comparing two haplotypes, an assertion that at least one of the two alleles from one haplotype matches at least one of the two alleles from the other haplotype across an entire region (segment), though such assertions generally include an allowance for mutations. See also: *HIR*, *segment*.

Mbp Megabase pairs or million base pairs. A unit of physical length along a DNA strand.

Meiosis A type of cell division that produces reproductive cells (egg or sperm) in which the diploid chromosome is reduced to a haploid.[13,14]

MRCA Most recent common ancestor.

NPE Several different expansions of this acronym are present in the literature. Here are a few:[15]

- non-paternity event
- non-paternal event
- non-parental event
- not the parent expected

In all cases, it refers to cases of misattributed paternity.

Nucleotide A molecular building block (monomeric component) of the polymers DNA or RNA.[16] Also called a "base".

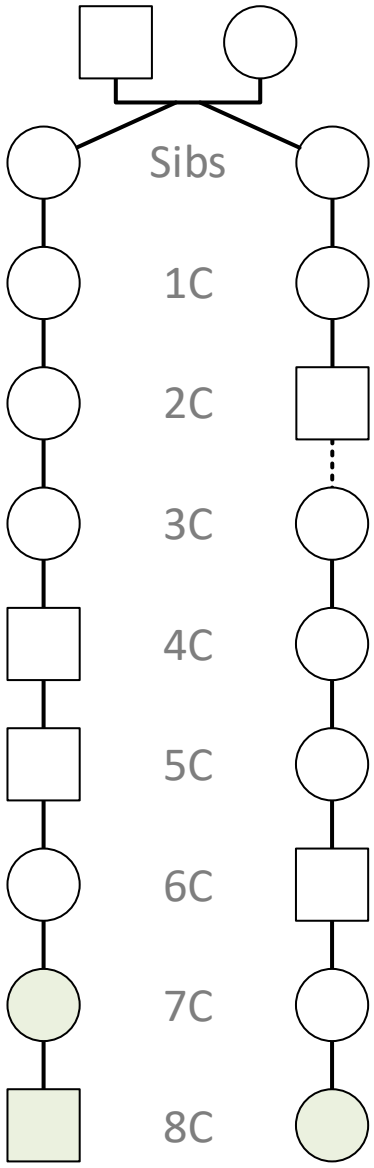
Segment A region of a homologous chromosome pair for diploid cells, or a region of a single chromosome in the case of a haploid cell. See also: *match*, *HIR*.

SNP Single nucleotide polymorphism. A single nucleotide variation that occurs at a defined location within the genome.

STR Short tandem repeat.

Triangulation A technique that involves finding three (and preferably more) persons that have a portion(s) of their DNA that is identical in a way that suggests they share a common ancestor, and then correlating this suggested commonality with genealogical research that shows a common ancestor is uniquely shared among these same persons.

KEY FOR LINEAGE DIAGRAMS



Sibs	Siblings [Often spelled out, but abbreviated in a few cases.]
1C - 8C	1st cousin, 2nd cousin, ..., 8th cousin, ...
	couple relationship
or	parent/child relationship (dashed is proposed)
	male relation
	female relation
	shading indicates genotyped individuals

INTRODUCTION

DNA genotyping gives consumers an opportunity to discover their genetic heritage, giving them a personal profile—a genotype—made up of hundreds of thousands of genetic markers.[17] In genetic genealogy, genotypes are used to associate individuals that may share a genetic relationship. Yet many of the suggested relationships are mirages—a by-product of data and algorithm limitations. Even in cases where a genetic relationship is sure to exist, the exact nature of that relationship can only be estimated. For example, one genotyping company estimates the relationship with a grandmother and her grandson as a range that includes siblings, aunts/uncles, nieces/nephews, grandparents/grandchildren, half-siblings, first cousins, great aunts/uncles, great grandparents/grandchildren and half aunts/uncles.[18] To what extent, then, can autosomal DNA (atDNA) genotypes be used to reveal and confirm (even prove) genealogical relationships?

This text investigates core concepts and methods that enable genetic genealogists to make use of atDNA to reveal and/or corroborate genealogical relationships—adding another type of genealogical record to the repertoire that researchers can use to solve genealogical problems.

This text is addressed to experts in the genetic genealogical community, and more particularly to those experts who promote the use of atDNA in genealogical research and who teach genealogical enthusiasts how to make use of their genotypes to address genealogical research questions. It is the author's hope that these experts will seek standard ways to identify, evaluate and present genetic genealogical conclusions, and that the methodological framework presented herein can be instrumental in bringing the community toward a consensus in these matters. It is the author's belief that genealogists with a serious interest in using atDNA to further their genealogical research will also benefit from this text.

GENOTYPING

Genotyping atDNA has been available direct-to-consumers since 2008.[19] In 2016, three genotyping companies have large consumer genotype databases: AncestryDNA™, 23andMe™ and Family Tree DNA™. Most of their genotypes are from US-based consumers.

Genotypes from AncestryDNA™, 23andMe™ and Family Tree DNA™ are proprietary compilations of the assayed values of single nucleotide polymorphisms (SNPs). Genotypes from these companies are based on the examination of 570,000-700,000 SNPs.[20]

So-called "next-generation" sequencing services are moving toward broad availability, making complete genome sequencing a reality.[21] Genetic genealogists do not have broad access to such data yet, so this text does not focus on the potential usefulness of such data.

Milestones in the Development of atDNA Genotyping	
1871	Friedrich Miescher published a paper that identified the presence of DNA in the cell nucleus.[22]
1904	Walter Sutton and Theodor Boveri had independently proposed that chromosomes were involved in inheritance and behaved in accordance with Mendelian laws, but it took until 1915 for their theories to be fully accepted.[23]
1953	James Watson and Francis Crick proposed the double helix structure of DNA along with the idea that it could be separated into strands to be copied.[24]
1977	Walter Gilbert and Frederick Sanger independently developed rapid DNA sequencing techniques that could be used to sequence genes.[25]
1983	Kary Mullis developed the polymerase chain reaction (PCR) to amplify DNA.[26]
1985	Alec Jeffreys developed a mechanism for profiling a genome.[27]
1990	The Human Genome Project was launched to sequence the entire human genome.[28]
2000 (March)	Family Tree DNA™ began offering a service to produce a 12-marker Y-chromosome haplotype.[29]
2000 (June)	An announcement was made concerning the initial completion of a full sequence of the human genome.[30]
2008	Genotyping companies (including 23andMe™) began selling direct-to-consumer (DTC) atDNA genotyping services.[31]
2010	Family Tree DNA™ began a phased roll-out of their atDNA genotyping service.[32]
2012	AncestryDNA™ launched their atDNA genotyping service.[33]

Figure 1: Milestones in the development of atDNA genotyping.

GENETICS AND GENOMICS

Genetic genealogy has its foundations in genetics—a specialty within molecular biology that is concerned with heredity, its mechanisms, and variation of inherited

characteristics.[34] Geneticists tend to focus on genes with known functions.[35] As computing and genome sequencing have advanced, a new specialty—genomics—has emerged: the study of the totality of an organism's genetic material.[36]

Understanding the fundamentals of genetic inheritance is critical to making use of genomic information for genealogical purposes. Figure 2 depicts the pattern of inheritance for the recombining (autosomal pairs) and non-recombining (mtDNA and Y chromosome) parts of the genome.

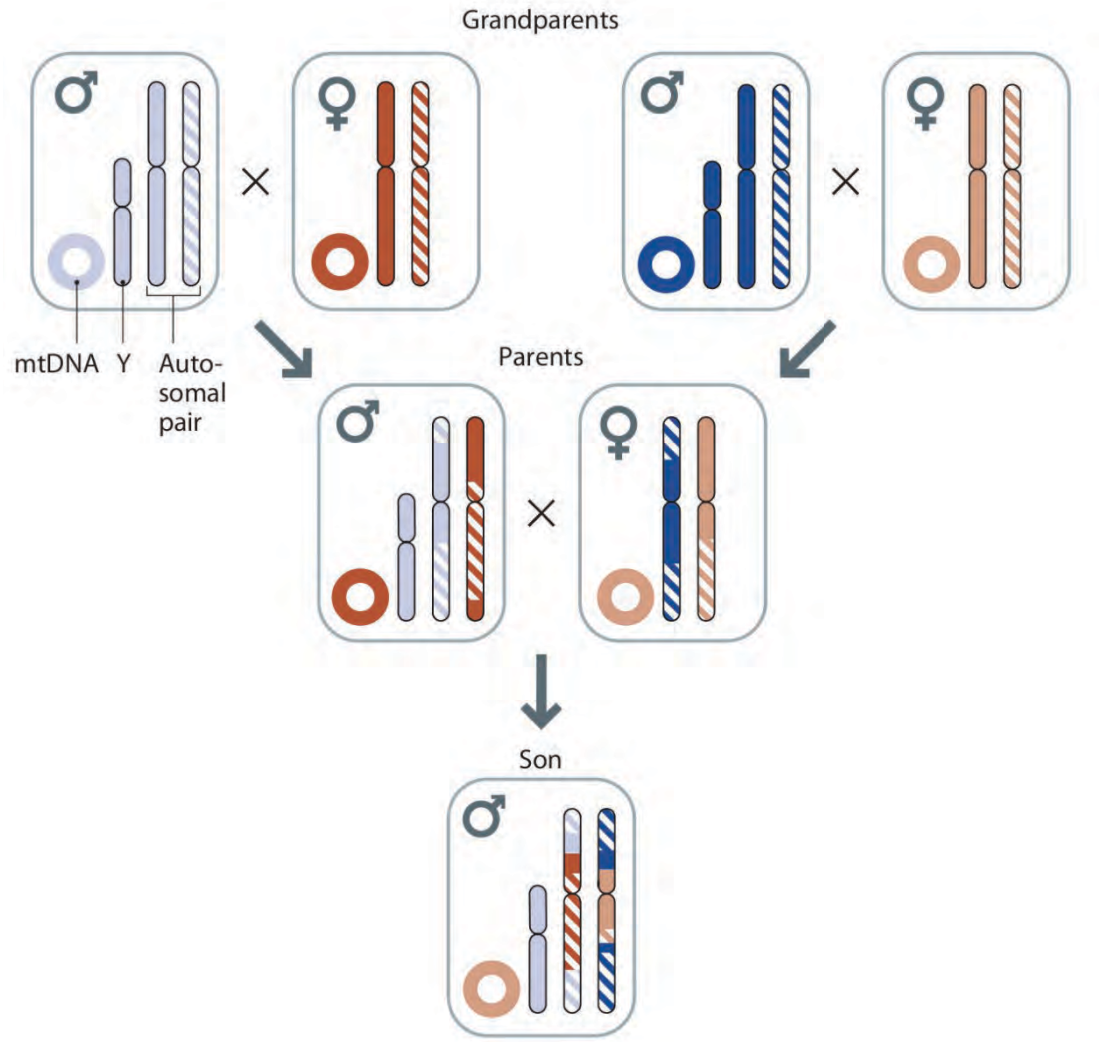


Figure 2: Inheritance of recombining and non-recombining portions of the genome.[37]

Mitochondrial DNA (mtDNA) is passed without recombination from a mother to her children (male or female). When considering an individual, it is known that they received their mtDNA from their mother, who received it from her mother, and so on, back to Mitochondrial Eve.

Most of the Y chromosome (or yDNA)—about 90% of it—is passed without recombination from a father to his son(s).[38] When considering an individual male, he received his yDNA from his father, who received it from his father, and so on, back to Y-Adam.

The autosomes—a focus of this text—are all the chromosomes (numbered 1 through 22 from longest to shortest) that are not the sex chromosomes (the X and Y chromosomes). Autosomes are paired—diploid. Given an individual, one chromosome in each pair was received from their father (the haploid paternal chromosome), and one from their mother (the haploid maternal chromosome). During meiosis, material from both chromosomes in a diploid pair can be reshuffled (a process called recombination) to form a unique, new haploid copy of the chromosome—a mash-up of material from the paternal and maternal haploid chromosomes. The result of this reshuffling has been represented in Figure 2.

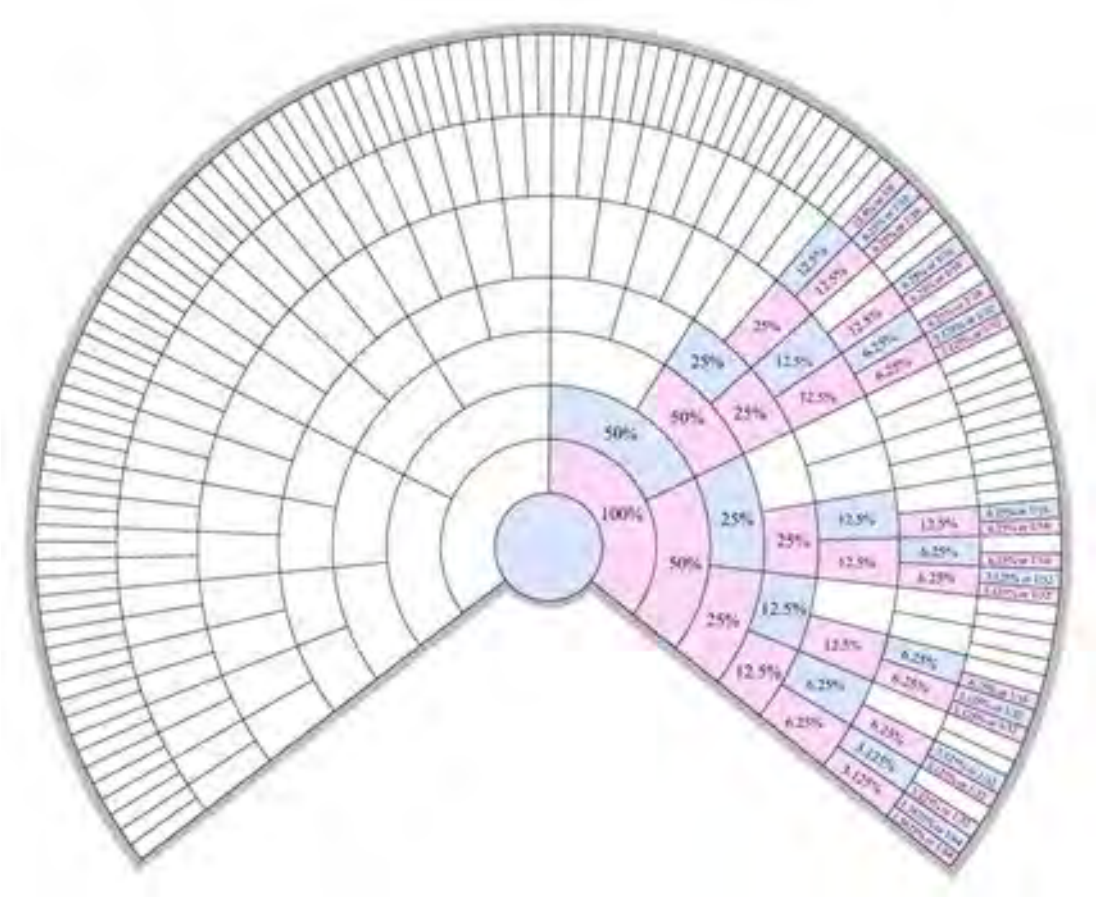


Figure 3: Male inheritance of X DNA.[39]

The X chromosome is not specifically depicted in the Figure 2 diagram (and many diagrams like it). It is another portion of the genome that undergoes recombination.

While it is possible that some recombination occurs when an X and Y are paired (the sex chromosome configuration in males), it is the reshuffling that occurs when two X chromosomes are paired (the sex chromosome configuration in females) that is generally responsible for variation in a haploid X chromosome.[40] Thus, the paternal copy of the X chromosome received by a daughter is received essentially unmodified from her father. On the other hand, the maternal copy of the X chromosome received by a child (son or daughter) from their mother is a reshuffled copy of their mother's X DNA. *Appendix E* examines an X chromosome match.

The primary source of variation in the autosomes (and X chromosomes) is recombination. Recombination rates are higher in women than men—researchers reporting about 41 crossovers per meiosis in women compared to about 27 crossovers in men.[41,42] There are recombination hotspots—loci on chromosomes where crossover events are more likely. Other types of variation in the autosomes are rare; the mechanisms that replicate and maintain DNA are very resistant to mistakes.

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LITERATURE

This text makes heavy uses of material from blogs and online forums. Proving genealogical relationships using autosomal DNA in general, and triangulation in particular, has not received attention in peer-reviewed journals. Much of the discussion around these topics plays out online in these mediums, among citizen scientists and community pundits alike.

PERSONAL GENOMICS

Also called recreational genomics, personal genomics is an area of emerging consumer interest. People are curious about what their DNA says about their health, physical characteristics, and ethnicity.[43] Media and advertising around DNA genotyping (particularly atDNA genotyping) caters directly to these common interests.[44,45]

DNA genotyping promises to help clinicians optimize health care, to answer questions about ancestral origins and give insights into personal physiology.[46,47] However, interpretation of genomic information is in its infancy. Science and its ability to prognosticate are always ahead of its ability to intervene or affect outcomes.[48]

As the cost of sequencing individual genomes falls, studies that incorporate full genome sequencing are on the rise.[49] With consumer confidence reasonably high, research groups are seeking to engage consumers interested in personal genomics, asking them to share their genomes to accelerate genetic research objectives.[50,51]

Social issues can affect consumer confidence. Of particular concern is information protection—keeping personal genomic details from being sold, misused, and out of unfriendly hands.[52] One company suspended access to personal genomic data when a firestorm of media attention became a public relations nightmare.[53,54]

DNA INHERITANCE

Solving genealogical problems using atDNA has its foundation in genetics. Genealogists desiring to make use of atDNA require fundamental knowledge of DNA inheritance—yDNA, mtDNA, and atDNA (including the X-chromosome

inheritance)—and a sense of the types and sources of variation in the genome (particularly in the autosomes). This knowledge is central to reasoning about DNA and its relevance to genealogical problems. The volume of literature exploring and explaining these topics is enormous and not a focus of this text.

The International Society of Genetic Genealogy (ISOGG) hosts a wiki encyclopedia detailing many of the topics important to DNA inheritance.[55] This wiki is a recommended resource in the genetic genealogy community. Although ISOGG requires wiki contributors to make application for wiki edit privileges, content is susceptible to the same foibles common to all crowd-sourced content: inaccuracies, lack of depth/detail, missing attribution, and/or lack of curation by subject matter experts.[56,57] Information collected there is valuable but should be interpreted with appropriate caution. Researchers should consult the work of credentialed experts (e.g., Jobling, et al, or Dudley and Karczewski) to fully understand these topics.[58,59] The deeper understanding gained by forays into the academic literature will help them as they make decisions and form conclusions using genetic principles.

Success Stories

In a study published in 1998, DNA was used to show that US President Thomas Jefferson fathered at least one of the children of his slave Sally Hemings.[60] In 2009, DNA genotyping (including atDNA genotyping) helped confirm the murder of two missing children of Russian Tsar Nicholas II.[61] Archaeologists, excavating an abbey found under a car park in 2012, found a skeleton that was later shown to be Richard III, King of England.[62,63] These peer-reviewed findings demonstrate DNA's power to prove relationships. Others, such as Richard Hill, share their personal journey to finding their biological roots.[64,65] ISOGG has created a space for members to post such success stories.[66] Plenty of literature gives efficacy to the use of DNA in establishing genealogical relationships.

Proof

As relationships are identified using atDNA, the question that naturally follows is: Does atDNA prove the relationship?

Questions of proof using DNA dwell within the broader topic of genealogical proof. The Board for Certification of Genealogists (BCG) has been, perhaps, the most

vigorous proponent for implementing genealogical standards. Its first attempt to codify a Genealogical Proof Standard (GPS) was *The BCG Genealogical Standards Manual* published in 2000.[67,68] Others, particularly Mills, have done much to bring this GPS out of obscurity and into the collective consciousness of genealogical practitioners.[69,70,71] Jones wrote a clarifying volume focused on the core elements of the GPS.[72] Further refinements have since been published in the *Genealogy Standards* manual.[73] The GPS defines foundational concepts (i.e., sources, information, evidence, hypotheses, conclusions, and proof) and describes fundamental processes and conditions—standards—by which genealogists achieve valid and acceptable outcomes. It will be shown that these concepts can be applied to atDNA in making genealogical conclusions.

When considering genealogical relationships determined by DNA analysis, the genetic genealogy community continues to debate what is required to demonstrate proof. Discussion can be found in a variety of settings (e.g., blog posts, forum discussions, conferences, mailing lists).[74,75,76,77] Some seem to want to impose so many conditions as to make conclusions an impossibility.[78] Others are quick to point out how tenuous any conclusion can be.[79] For example, in a contribution to the community's on-going discussion of triangulation, Jim Bartlett posted an article on his blog about intermediate common ancestors having a role in triangulation that sparked a number of discussions.[80] Jason Lee posted a three-part article on triangulation in response to Bartlett's post, but his discussion of triangulation does not include the concepts presented by Bartlett—giving Bartlett's ideas no credibility.[81] In a post to the ISOGG Facebook group, Blaine Bettinger lists several triangulation reliability factors important in his thinking and adds Bartlett's concept as possibly having merit.[82] Kathy Johnson seems much more adamant about the need for intermediate common ancestors, discounting triangulation solutions that do not include them.[83] Principles that can guide genetic genealogists in forming and qualifying their conclusions are developed in the *Methodology* portion of this text.

Social Concerns

Genotyping brings with it a host of social issues. Scientists are now able to modify DNA to achieve a desired outcome (i.e., genetic engineering).[84] Even if only to prevent a known genetic disorder, such capabilities raise many ethical questions.[85] The notion that genetics are the primary contributor to personal characteristics,

including intelligence, behavior and health—sometimes called genetic determinism—led high-profile actress Angelina Jolie to choose elective surgery in hopes of avoiding cancer.[86] Movies such as *Gattaca* highlight fears that misuse of DNA information will lead to discrimination and other social problems.[87] Trepidation about privacy and custodianship are among a host of other concerns.[88] The Genetic Genealogy Standards Committee is proactively creating standards that include the need to address social and ethical concerns.[89]

Alongside these broader concerns are anxieties and consequences affecting individuals and/or families. DNA can reveal secrets that individuals involved may wish had remained hidden. Birth parents may wish to remain anonymous or may be unwilling to admit their role in a child's birth. These unexpected results can cause turmoil and pain for those involved.[90] One should consider potential social pitfalls, and even implement pre-emptive measures, as they undertake genetic genealogical research.

QUANTITATIVE ANALYSIS

Analyzing the total amount of atDNA shared between two individuals compared to an "expected" amount—something Tim Janzen calls quantitative analysis—is important both as a means of estimating relationships and as a means of supporting conclusions.[91] Tables and diagrams expressing the mathematical halving of the expected amount of atDNA received by each succeeding generation abound.[92] This technique is effective in identifying close family relationships—to fourth degree relationships on Bettinger's scale (see Figure 10).[93,94]

There is a clear disconnect between the small "expected" amounts and "observed" amounts as relationships become more distant.[95] Geneticists have identified this as an expected result.[96,97,98,99,100] Yet, many genetic genealogists struggle to understand this disconnect. Blaine Bettinger's *Shared cM Project* gives important insight to this variance but has only published data for relationships closer than fourth cousins.[101] Speed and Balding's findings give important insight into this variance, particularly as it pertains to more distant relationships; however, a genetic genealogist will wish their work had been presented in centimorgans, and with more granularity, when considering distant relationships.[102]

Luke Jostins gives an oft-quoted answer to a related question: "What percentage, on average, of an individual's genealogical tree at X generations is part of their genetic tree?"—estimating that the probability that one will share atDNA with all of their ancestors in a given generation starts dropping in the fifth generation.[103] Jostins now explicitly defers to Graham Coop's work answering this same question—that the probability starts dropping in the seventh generation.[104] Jostins' fifth-generation answer is still prominent in genetic genealogical literature.

TRIANGULATION

This text focuses particularly on a technique referred to herein as *triangulation*. Triangulation is accepted by many as a method for proving relationships and is especially relevant to establishing distant relationships.[105] At its core, triangulation involves finding three (or more) persons that share an identical atDNA segment (HIR) and that also have genealogies that show a single common ancestor is uniquely shared among them.[106,107] As most of a person's atDNA matches will be distant cousins, triangulation is an essential tool in the genetic genealogist's repertoire.

In the body of literature produced by the genetic genealogical community, *triangulation* is used in a few different ways, making it necessary to disambiguate this term as used in this text from other uses common in the community. For some, triangulation is a set of mechanical genotype comparisons (no genealogy required) relative to a specific matching segment used to confirm membership in a triangulated group (see *Identifying Triangulation Building Blocks* and its subsections starting on p. 55). These rote comparisons—sometimes disambiguated as *segment triangulation*—are a necessary part of the technique that is the focus of this text but are not *triangulation* as used in this text.[108]

Sometimes genealogists seem to equate *In Common With (ICW)* groups (see *Definitions*) with triangulated groups—associating *triangulation* with attempting to identify relationships using ICW comparisons.[109] Researchers, particularly in the adoption community, have used ICW groups to great effect. One of the reasons for this success is that many of these groups would, in fact, triangulate if the relevant details of these groups could be known. ICW analysis can function as a proxy (of sorts) for triangulation, a tool to use when the information required for triangulation is

not available. However, ICW comparisons are not sufficient to assure triangulation. Because there is more than one possible genetic relationship within an ICW group, what can be concluded by using these groups seems ill-defined.

Genotyping companies provide varying levels of support for triangulation, including providing no support.[110] Deficiencies inherent in genotyping company tools can be overcome using third-party tools (e.g., GEDmatch.com) if one's matches will cooperate.[111]

Triangulation begins with identifying a triangulated group (TG) and culminates with a conclusion about a common ancestor (CA) shared among members of the TG.[112,113] Factors that affect the reliability of triangulated conclusions are constantly under discussion. In one such discussion:

- Bettinger advocates the need to understand the coverage and accuracy of the trees contributing the common ancestor.
- Bettinger, and also Johnston, expect a matching segment to be sized such that it is probable it is identical-by-descent (IBD; see *IBD vs. IBS vs. IBC* on page 40 for more information).
- Bettinger expects quantitative analysis of total shared atDNA to fit anticipated ranges.
- Bettinger gives Bartlett's intermediate CA concept possible merit, while Johnston seems wholeheartedly in favor of declaring it required.
- Both Bartlett and Johnston believe that the matching segments will fit (without conflict) within the chromosome maps that have been established for the genotyped individuals.
- Johnston wishes to impose a number of other constraints including genotyped individuals knowing their chromosome crossover locations, and grandparent phasing.[114,115,116]

It is unlikely that all solutions will satisfy all demands. Genetic genealogists will need to be able to evaluate their solutions against a number of criteria and convey information about strengths and weaknesses inherent in their solutions.

Evaluating Matching Segments

Another type of quantitative analysis that is important to identifying matching atDNA segments as IBD is the analysis of the length/size of the matching segment. In one

definition, IBD matches are characterized by their frequency (or rather their rarity)—the likelihood that an identical sequence in the same position could be observed in two separate individuals at the same time.[117] Segment size (given in centimorgans) becomes a proxy measure for segment frequency: the longer the shared sequence, the smaller the likelihood that it could be observed by chance in separate individuals. In fact, a centimorgan is defined in terms of likelihood.[118]

Much attention is given to whether a small half identical region (HIR)—often called a matching segment in the literature—can be used in making genealogical inferences (i.e., can be shown to be IBD).[119,120] Janzen published data about two genomes he studied extensively showing that nearly 80% of 5 cM HIRs could not be IBD.[121] Prairielad, an author in the Family Tree DNA™ forums, posted similar data from 14 genomes studied in his family.[122] In these instances, the small numbers of genomes involved leave unresolved questions about the statistical validity of their conclusions. Walden shared data from a private study of 9000 genomes, showing that even small, phased matches are largely IBS (not IBD).[123] This study has received criticism for its lack of disclosure and peer-review.[124] The relevance of small HIRs remains a critical topic; experts continue to advocate extreme caution when considering these regions.[125]

Quantitative evaluation of HIRs is also tied to the nature and quality of genotype data. With two measured allele values per SNP (and with possibly missing/uncalled values), there is a likelihood that a match will be declared where there should be no match—a false-positive. At times, this likelihood is quite high.[126] For this reason, long HIRs are required for confidence in IBD conclusions.[127] Even phasing (discussed below) cannot eliminate the possibility of false matching as HIRs get shorter. While thought leaders warn against trusting small HIRs, not all possible sources of error are included in their reasoning.[128,129]

PHASING

An atDNA genotype is a collection of SNP assay results. The SNPs examined are selected to show common variation—to highlight areas in the human genome where one is likely to find differences, not areas that are always the same.[130] SNPs are located at well-known sites (loci) in the genome. Because a human genome is diploid, every autosome SNP that is measured generates two values—one allele

(base value) coming from the paternal haploid and one from the maternal haploid. There is no mechanism in the assay process to distinguish which of the two base values reported for a given SNP is paternal or maternal—but one surely is paternal and the other maternal. The process of assigning alleles to a parent-specific chromosome is called phasing.[131] Phased genotype data is very valuable to many aspects of genetic research.[132] For the genetic genealogist, phasing greatly assists in the identification of IBD matches—or perhaps more accurately, the elimination of a great number of matches that are IBC.[133,134] Phasing does not ensure that all remaining matches are IBD, as some would like to believe.[135,136] Using his own genome, Janzen reports that only 23% of his "matches" remained when using phased data for comparison at a 7 cM threshold.[137]

CHROMOSOME MAPPING

Genetic genealogists undertake chromosome mapping to associate specific matching atDNA segments (HIRs) with specific ancestors.[138] It can also be used to associate specific HIRs with ethnic populations.[139] By way of example, Janzen has mapped 95% of his mother's genome as to whether it came from her father or her mother, but he has mapped only 30% of his mother's genome to specific grandparents.[140] Because triangulation associates HIRs with known ancestors, chromosome mapping and triangulation are integrally connected.[141,142] When a matching segment does not fit properly within a mapped region of a chromosome, it can signal that the matching segment is IBS.[143,144] In the genetic genealogical community, Janzen's use of this technique is well known and his expertise often sought.[145] Janzen, sometimes collaborating with others, has been forthcoming in explaining his processes.[146,147] Others have provided tools for representing maps. Kitty Cooper has built and published several on-line tools to assist in the graphical representation of chromosome maps.[148] Griffiths has provided several spreadsheets for representing chromosome maps.[149]

RESEARCH PROCESS

This text's focus on methodology was born out of the author's own desire to make use of his genotypes for genealogical purposes. His first genotype (one representing his aging grandmother) was made available April 6th, 2014.[150] His own genotype was available starting February 8th, 2015.[151] Yet, the author had not made use of these important resources. He had examined the information provided by the genotype services, had read various published materials, and had attended numerous presentations about genetic genealogy. Even so, there was no clear place to start or path forward.

The author was very obviously not alone in this feeling. Most of the people sitting with him in presentations seemed to face a similar dilemma. The hands of most present would be up when asked if they had been genotyped, but most hands were down when asked about doing more with their results than reviewing their ethnicity estimates. People in the genealogical community are very interested in using their genomes to further their genealogy but, from novice to expert, genealogists seem to feel confusion and a lack direction as they seek to make use of their genotypes.

When proposing a solution to a cousin's "brick wall" (detailed in *Appendix F*), the cousin was unwilling to believe the solution as initially proposed because the literature and materials common in the community do not address the nuances that make the solution viable. As the author sought to understand the necessary details and to address the concerns raised, it became apparent that the elements important to the process were not well identified and often only discussed in isolation. Even after months of exploring genetic genealogical literature, a vision of what the ultimate goals ought to be and a path to reach those goals had not emerged. The needed roadmap was missing. A methodological framework was needed.

The framework presented in this text is grounded in the genealogical research process and the genealogical proof standard. The author first encountered these topics as evangelized by Elizabeth Shown Mills at a National Genealogical Society (NGS) conference in 2010.[152] He has continued exploring these topics in subsequent years—in Mills' book *Evidence Explained*, in conference talks, and in other literature (see the section titled *Proof* above). In 2013, the author participated in the Salt Lake Institute of Genealogy (SLIG), completing the *Advanced*

Genealogical Methods course (taught by Thomas W. Jones)—a deep dive into genealogical research methodology.[153] It is Jones' explanations of methodology that have most resonated with the author and that are the foundation of the genealogical research process and genealogical proof concepts presented herein. Subsequent collaboration in the development of the GEDCOM X project (including collaboration with Mr. Jones) has served to further deepen and refine the author's knowledge and understanding of these concepts.[154]

The author spent time reading a wide variety of blogs and other published materials seeking to understand the use of atDNA for genealogical purposes. The author looked for methodological material, and then for answers to specific questions relative to his research issues. Methodological material tended to be step-by-step directions (the *how* without much content as to *why*). The author also read a wide variety of material published in academia (e.g., Speed) and by expert members from the genetic genealogy community (e.g., Bettinger, Janzen). Addressing the concerns raised in the case detailed in *Appendix F* was particularly important; this focused the research and served to highlight the most important information.

As the author explored the various concepts strewn around the landscape of the genetic genealogical literature and tried to synthesize these concepts into an integrated whole, he began to see how the fundamental elements could be mapped into the conceptual framework and methods of the genealogical research process. The needed methodological framework already existed! Mapping the concepts and processes from the genetic genealogical community into that context caused a genetic genealogical methodology to emerge.

The information (i.e., genotypes and compiled genealogies) used in this text has been gathered using methods typical of genetic genealogists (e.g., vender-specific messaging, email, phone calls, published genealogies, etc.). The author sought to experience genetic genealogy in the way it would be experienced by any other researcher. Because genotyping services are not providing all the tools required to execute the methodology detailed herein, the author focused on genotypes available in GEDmatch (a service that does provide tools sufficient to execute this methodology). In many cases, genotype administrators were willing to make their genotypes available in GEDmatch when asked.

The author worked with genotypes that match the familial genotypes that he administers. AncestryDNA™ had matched the author with about 3600 genotypes, and 23andMe™ with about 900, at the time he began work on this text.[155] Contacting the administrators for each of these genotypes in a systematic way was never a priority for this project. Rather, the author was opportunistic in his approach—interacting first with genealogists actively researching their connection with the genotypes administered by the author, then seeking the cooperation of the administrators of related genotypes. The author also sought the cooperation of close cousins that were already genotyped.

Given a cooperating genotype administrator, the first goals were to perform any testing that could eliminate the match as IBC and to establish the existence of a common ancestor. If the genotype was not likely IBC and if a common ancestor was identified, other genotypes that shared this same matching segment were identified—i.e., the "triangulated group" for the matching segment was identified. At this point, an analysis of genotype independence was performed to identify any family groups within the triangulated group. Using compiled genealogies that represented the individuals (or family groups) in the triangulated group, common ancestors (if any) were identified with these additional genotypes.

Over the course of this research, the correspondence with genotype administrators surpassed 700 messages. A common ancestor was identified with more than 50 genotypes.

To anonymize the data presented in this text, genotypes have been assigned an identifier specific to this text. The identifier is in the form of GT000—"GT" for genotype, followed by three randomly generated digits. The mapping of these identifiers to vender-specific identifiers will be submitted with the text, but not published. Except in one case, the actual details of persons (living or deceased) have been omitted from lineages, etc. The exception is a case where a "brick wall" necessitated a discussion of the genealogy of a few specific ancestors (*Appendix F*). In this case, only identifying details of deceased individuals are included.

METHODOLOGY

GENEALOGICAL RESEARCH PRINCIPLES AND PRACTICE

A *conclusion* is a *hypothesis* that passes scrutiny. To form a *hypothesis*, the researcher correlates at least two independent instances of *evidence*. *Evidence* is the result of analyzing and correlating *information* to answer a question. *Information* comes from a *source*.

The genealogical research process begins with question asking. The question should not be too broad (where more than one correct answer is possible) nor too narrow (where an answer is not possible because the sources available are not capable of producing an answer).[156]

Armed with a proper question, the genealogist shifts to *information* gathering. A thorough search is planned, identifying all *sources* that might reasonably contain *information* that could answer the question.[157] Each *source* is examined and the results (positive or negative) are noted. If the examination of a *source* leads to additional *sources*, the genealogist repeats the process for each additional *source*.

The genealogist analyses all relevant *information* for answers to their research question—each tentative answer an item of *evidence*.[158] A minimum of two independent items of *evidence*—two independent answers to the research question—must correlate (agree) to form a credible *hypothesis*.[159] All relevant items of evidence must be synthesized to form an integrated whole.

As the researcher synthesizes the relevant evidence to the answer the research question, the *evidence* forming their answer is tested—the tentative answer becoming a *hypothesis*.[160] *Hypothesis* testing considers whether items of *evidence* are suitable and able to answer the research question. Each building block used to establish the *hypothesis* (each *source*, *information* item, and/or item of *evidence*) is examined. Tests of analysis and tests of correlation are considered. Tests of analysis evaluate the likelihood a building block is what it was assumed to be. Tests of correlation examine whether independent items of *evidence* agree.

A *hypothesis* is accepted as a *conclusion* when it passes all applicable tests of

analysis and correlation.[161] In the GPS, a proven *conclusion* requires a written *conclusion*.[162]

The genealogical research process is summarized in Figure 4 as follows: *Sources* (people and artifacts available in our present day) are interrogated for *information* that, through mental analysis and manipulation—i.e., the genealogical research process of question asking, information gathering, hypothesis testing, conclusion accepting and proof explained—are transformed into *evidence* that is used to answer questions about the facts of the past.

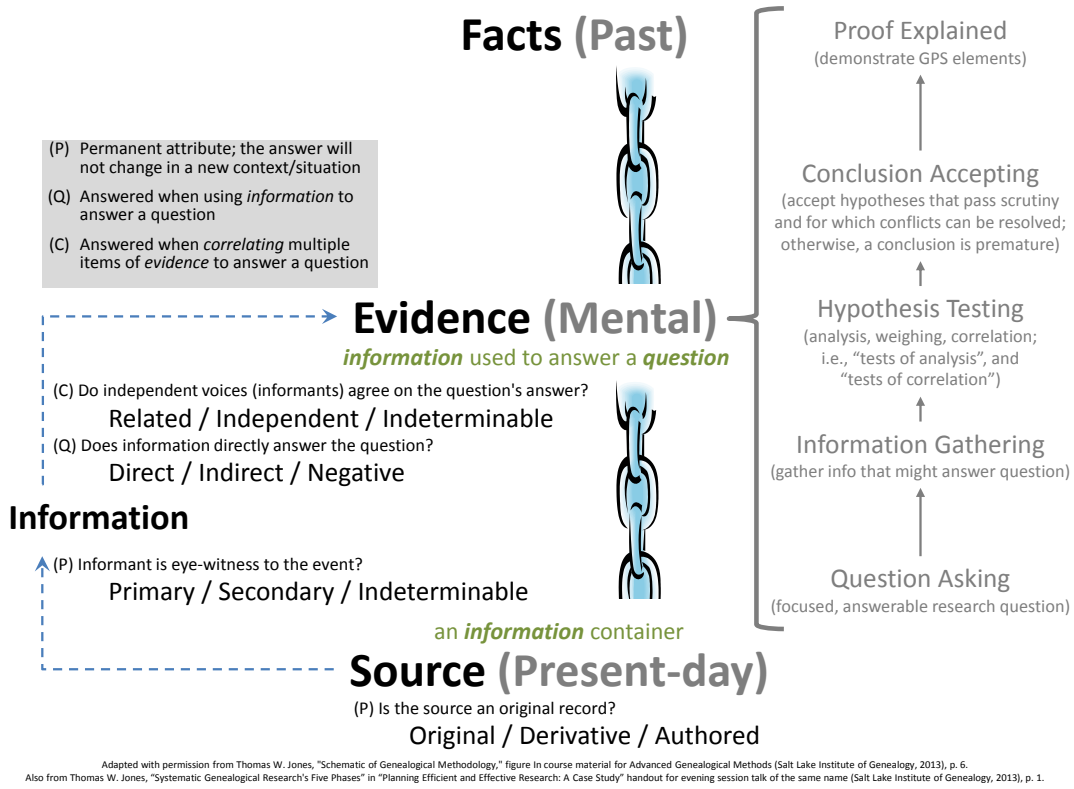


Figure 4: A representation of the genealogical research process.[163]

Methods used in genetic genealogy should (and do) fit into the genealogical research process. As genetic genealogical practice is considered in what follows, the methodology is grounded in the genealogical research process:

1. question asking
2. information gathering
3. hypothesis testing
4. conclusion accepting
5. and proof explained

Considering the methodology in this way integrates genetic genealogical practice within the broader genealogical research practice.

QUESTION ASKING

A genetic genealogist, like any researcher, needs focused, answerable questions—questions suited to the unique characteristics and capabilities inherent in the human genome. Yet would-be genetic genealogists are sometimes unrealistic in their expectations. It is appropriate, therefore, to consider the possibilities and limitations before one engages in a genetic genealogy research project.

Genetic genealogists have found genotyping useful for the following:

- finding cousins
- confirming relationships (e.g., when paper trails are weak)
- disproving relationships
- breaking through brick walls
    - results can suggest relationships where none were previously known
    - results can reveal/correct misattributed paternity
- finding biological parents (for someone living, or for a deceased ancestor)
- exploring ethnic origins
- exploring deep ancestry (requires mtDNA or yDNA haplotypes)

New genetic genealogists often feel overwhelmed as they attempt to make sense of the information provided by genotyping companies. They express surprise at the amount of work and learning required to get the expected benefits. Yet, these benefits are real and achievable in many instances.

INFORMATION GATHERING

With a research question in hand that may be answerable using atDNA, the genetic genealogist takes up the information gathering aspects of the research process.

CORE SOURCES

Information is found in and gathered from *sources*.

In genetic genealogy, the primary sources of genetic *information* are human

subjects—oneself, parents, siblings, children, cousins, grandparents, etc. These primary *sources* are living records—each unique (except in cases of monozygotic twins, triplets, etc.), and each with information about several generations of ancestors. Access to the *information* in these records comes via genotyping. Genotyping requires that one submit biological material to be assayed. Current direct-to-consumer genotyping services request a saliva sample or cheek swab to collect the necessary biological material.

Genetic *information* is necessarily interpreted in the context of pedigrees when used for genealogical purposes. Compiled genealogies become critical *sources* of pedigree *information* as research proceeds. These *sources* are authored works—presenting many *conclusions*, possibly containing information available nowhere else, often based on primary *information* from original *sources*. These *sources* (compiled genealogies) may be susceptible to author bias or mistakes made in interpretation. It is not practical for genetic genealogists to personally create and curate all of the pedigrees necessary to unlock the *information* available in these genomic records. Collaboration and sharing are critical to genetic genealogy success.

CORE INFORMATION

The core *information* item from a human *source* is their genotype. The core *information* item from a compiled genealogy is a pedigree extract that details a person's lineal relationship to a common ancestor.

GENOTYPES

Genotyping studies the genetic makeup of individuals by examining the genetic variants they possess.[164] Genotyping differs from genome sequencing (which attempts to measure every base value) in that it measures a small subset of alleles selected for their likelihood of variation.[165] The resulting data, as an aggregate entity, is called a *genotype*. In many research situations, a genotype stands as a proxy for an individual's entire genome.

In a typical atDNA genotype, the proxy data represents only about 700,000 SNPs of more than 3 billion sequenceable loci that make up a human genome—representing less than 1/5,000th of the total information that could have been measured.[166,167]

Despite its clearly sparse representation, a genotype can effectively represent the entire genome because an individual human genome is 99.9% identical to all other human genomes.[168] Though sparse, genetic genealogists still consider atDNA genotype data in a sequenced way.

With the raw SNP data, genotyping companies provide at least two other *information* products: a list of DNA matches (persons identified as having genotypes with identical regions in common with the given genotype), and ethnicity estimates.

What is an identical region? Who can I expect to find in my match list? How can an ethnicity estimate be useful? Why do ethnicity estimates vary from company to company and from tool to tool for the same genotype? Exploring these topics further will facilitate the use and prevent the misuse of this important *information*.

IBD vs. IBS vs. IBC

Genetic genealogy has its foundation in the principles of genetic inheritance. Genetic genealogy becomes possible when two genomes have one or more identical regions—matching segments—that are identical-by-descent (IBD). In an atDNA genotype, a segment is the allele values from a specific region of a single haplotype (chromosome). Two segments are IBD if they are identical and if they were inherited from a *recent* common ancestor.

A segment is a region of a single chromosome—its allele values all a part of a single haplotype, and each haplotype representing a single chromosome (e.g., Chr1). Because each chromosome is diploid, each SNP assay results in two measured base values—one from the father, and one from the mother. The assay is not haploid-specific for parent origin; it does not report which base value came from which parent.

Assayed:	C/A	A/C	C/C	A/T	G/T	C/C	C/A	G/C	G/T	G/A	A/T	A/G
Father:	A	A	C	T	G	C	C	C	T	G	T	G
Mother	C	C	C	A	T	C	A	G	G	A	A	A

Figure 5: Un-phased haplotype is not haploid-specific.

Now consider asking whether two haplotypes are identical. Haplotypes are identical if, at a given SNP, they have at least one allele in common.

Identical-By-Chance (a false match)												
Assayed:	A/G	A/T	C/A	T/C	G/C	A/C	C/T	C/T	C/T	G/C	T/G	G/C
Father:	A	T	C	C	G	A	C	T	T	C	T	C
Mother	G	A	A	T	C	C	T	C	C	G	G	G

Identical-By-State (a paternal match; potentially IBD)												
Assayed:	C/A	C/A	C/C	A/T	T/G	C/C	C/G	A/C	A/T	T/G	G/T	G/C
Father	C	C	C	A	T	C	G	A	A	T	G	C
Mother:	A	A	C	T	G	C	C	C	T	G	T	G

Figure 6: Two genotypes that match with the Figure 5 genotype: the first is identical-by-chance; the second is at least identical-by-state and matches the paternal haploid.

Matching algorithms will declare both haplotypes in Figure 6 to match the haplotype in Figure 5. However, the first match has nothing to do with inheritance. It is an artefact of the data and its lack of haploid-specificity. The match is declared using a mix of alleles from both the paternal and maternal haploids. Because the matching algorithm does not know from which parent (haploid chromosome) the allele originated, it ends up declaring a match that is not biologically meaningful—a false-positive. The author designates matches of this type as identical-by-chance (IBC).

The second haplotype in Figure 6 has an allele sequence along its maternal haploid that matches the sequence along the paternal haploid in the Figure 5 haplotype. This match is, at a minimum, identical-by-state (IBS). It could be IBD, but this designation requires that other factors be considered.

When comparing two sets of unphased allele values from two haplotypes, if an allele from each set is identical, the data are said to be half-identical at that location. If a shared region is composed of half-identical alleles, it is said to be a half-identical region (HIR). HIRs may be IBC, IBS or IBD. When one considers the HIR that is the result of comparing the paternal haplotype from Figure 5 with the first haplotype in Figure 6, one can see that the HIR in Figure 6 is IBC because the matching alleles from Figure 6 are a mix of alleles from both parents.

It is possible, especially in the case of siblings, that regions are identical simultaneously on both chromosomes. Such a region is designated a fully-identical-region (FIR). The result of comparing the allele values from the Figure 5 haplotype with the allele values in the second haplotype in Figure 6 is an FIR across the first half of the region, and an HIR across the second half.

The genetic genealogy community generally does not differentiate the terms IBS and IBC, using them interchangeably.[169,170] A few in the community seem to use IBC in the manner it has been employed in this text.[171,172] In at least one instance, this author found a use of IBC that may have been better characterized as IBS.[173] It is true that the above IBC match is IBS (making IBC matches a subset of all IBS matches). However, the author believes that it is useful to consider matches that are a mash-up of the alleles from both parents separately from other types of IBS matches.

By definition, every base pair (and, therefore, every allele) in one's genome was inherited from an ancestor (except where mutation has modified a base pair after inheritance has played its role). This is why the notion of *recent* is part of the IBD definition. A timeframe for *recent* is not formally defined, but it would be difficult to classify an ancestor as *recent* if the common ancestor is outside of "genealogical time" (the timeframe in which it is reasonable to expect it is possible to document the genealogy of one's ancestors). The *recent* designation is also important in that we need to be able to identify the common ancestor (in time and space, and our relationship to them) to show inheritance. However, Speed and Balding have shown the possibility of sizeable IBD segments from ancestors more than 20 generations from the subject—well outside the possibility of documented genealogies for most.[174] Sometimes, shared atDNA is identical for historical reasons (e.g., endogamy or ethnicity)—IBS, but no longer distinctly inherited from a single common ancestor—separating *recent* by contrast from *historical* atDNA. Therefore, we might define *recent* by contrast as *not ancient*.

Match Lists

In addition to the *information* from SNP assays, genotyping services that enable genetic genealogy provide a list of matches—other genotypes in their database that share regions that are IBS with regions in one's own genotype. Matches in these lists are generally assigned an estimated relationship (e.g., "2nd Cousin – 3rd Cousin" or "5th Cousin – Remote Cousin") and grouped according to these estimates. Typically, these lists are ordered by the amount of IBS sharing in centimorgans (with those sharing the most listed first).

Unless one has recruited (or been recruited by) close relatives, it is not typical to find

many close relatives in their list of matches. The bulk of one's matches will be (predicted) distant relatives. This is because the more likely sharing is IBD, the rarer (lower frequency) the matches will be. (See also *Matching Segment* Size on p. 64.)

Consider also that the number of distant cousins one has is much greater than the number of close cousins. Henn et al estimated one might have 4,700 fifth cousins, 23,000 sixth cousins, and 120,000 seventh cousins.[175] As Steve Mount puts it: "…if you have many more distant cousins, as would be expected if your ancestors had large families, then someone who shares a single IBD segment is more likely to be a distant cousin, because you have so many more distant cousins."[176]

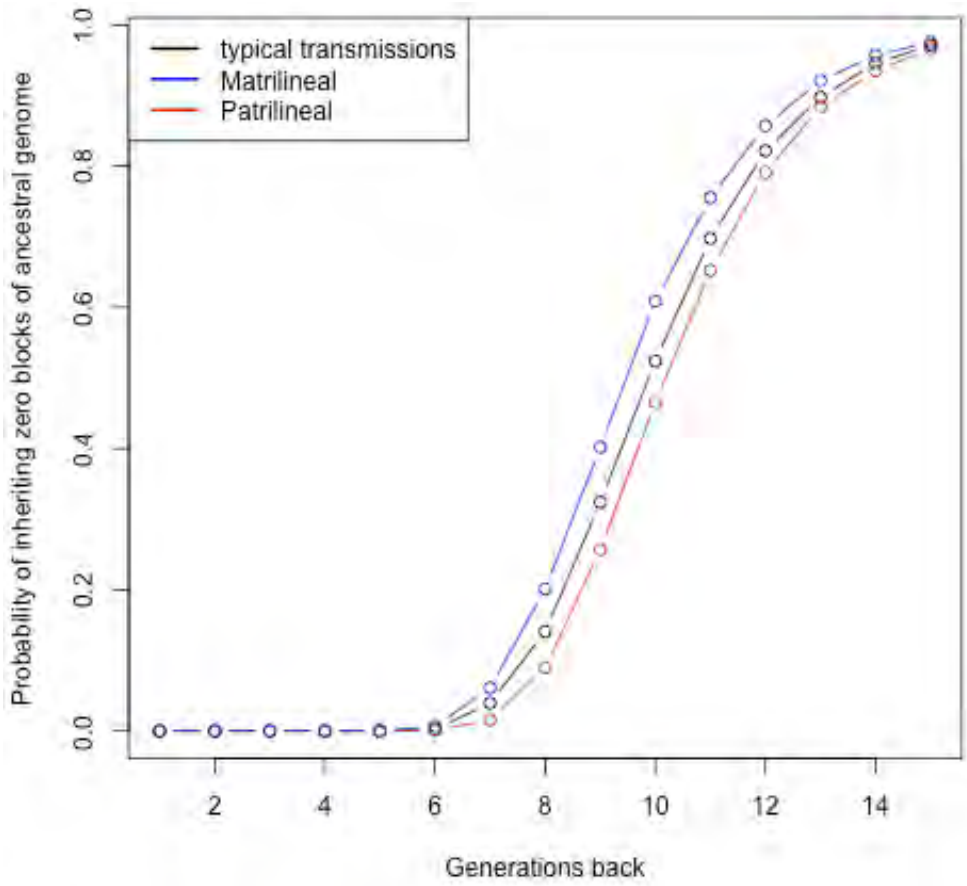


Figure 7: Probability of inheriting zero (large) blocks of ancestral atDNA.[177]

How many generations of ancestors on all of one's ancestral lines are detectable in one's atDNA? The genetic genealogy community's answer has been five generations.[178] Donnelly (1983) and Jostins (2009) are cited in support of this notion.[179,180] However, in 2013, Jostins explicitly deferred to the work of Graham Coop. Coop predicts that the likelihood of having zero detectable atDNA from an ancestor will not rise significantly until the 8th generation—giving researchers a high

likelihood of carrying detectable atDNA regions for nearly all of their 7th generation ancestors (see Figure 7). Beyond the 7th generation, the number of genetic ancestors (persons with whom one shares atDNA) is no longer the same as the number of genealogical ancestors (persons in one's pedigree). Bartlett affectionately calls a fan-chart plot of genetic ancestors a porcupine chart (see Figure 8).[181]

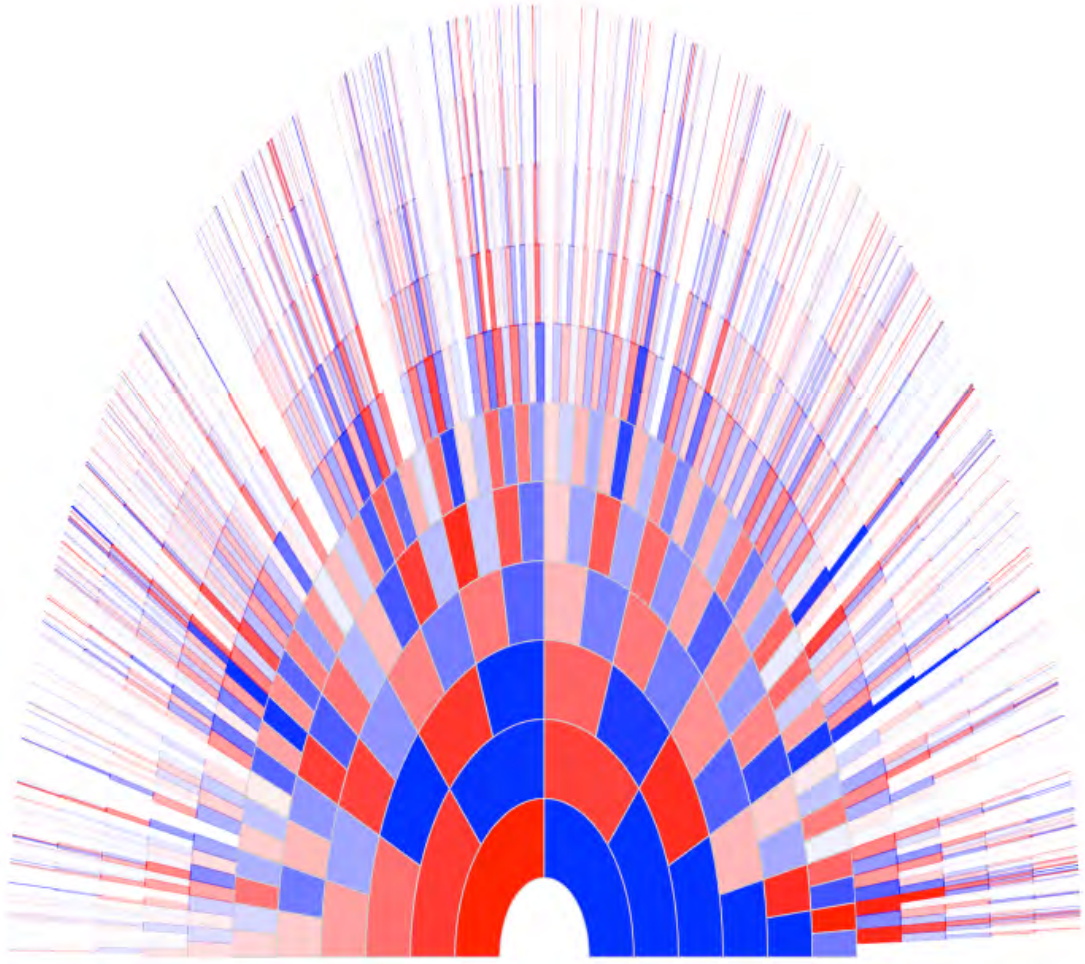


Figure 8: A fan-chart plot of 11 generations of genetic ancestors (simulated)—a so-called porcupine chart.[182]

Ethnicity Estimates

Along with assayed results and match lists, genotyping services provide *information* about ethnicity. Ethnicity estimates involve comparing a genotype to a reference population—a set of genotypes selected to represent a given ethnicity.[183] Algorithms seek a genotype's "best fit" with the collected reference populations.[184]

Each genotyping service's ethnicity reports are different, and individual results will differ between services. Hence, it has been said that estimating ethnicity is "part interpretive art and part science."[185]

Many express frustrations because their estimates do not correlate with what they know of their ancestry. The process involves comparing genotypes; it is susceptible to all the problems of such comparisons (e.g., being IBC). Reference populations may not actually represent the actual population of one's ancestor. One may no longer carry atDNA from the ancestor that was part of that population. Roberta Estes' article *Ethnicity Testing – A Conundrum* explains many of the pitfalls associated with ethnicity *information*.[186]

Ethnicity *information* can be used for genetic genealogical purposes, but it requires knowing which segment(s) are associated with the ethnicity of interest. Except for 23andMe™, genotyping companies do not provide this information. Triangulation can also be used to identify the segments of interest. The GEDmatch *Admixture/Oracle Population Search Utility* has reference populations that can be used to assist in this process.[187] Estes' article touches briefly on these methods.

LINEAGES

Genotypes cannot be converted into genealogical *information* without knowing the individual *source* subjects and their pedigrees. The *information* needed about these subjects is the relationship(s) (if any) that exists between them. Genetic genealogists seek the ancestors common among individuals—the point(s) at which two pedigrees intersect. Compiled genealogies are the *sources* that contain the *information* that is searched to identify these common ancestors and the lineages that link these *source* subjects to these common ancestors.

Ideally, the search for a common ancestor involves pedigrees that are complete (all ancestors identified) to at least the generation that includes the common ancestor, that are sourced with original records, and that exhibit only one ancestor in common. Practically speaking, there will be many incomplete pedigrees in various stages of development. Each compiler possesses different capabilities and different access to the genealogical records. Searching these pedigrees may lead the genetic genealogist to additional activities such as filling in pedigree gaps, looking at

descendants, correcting erroneous relationships, etc. Searches may also reveal more than one common ancestor—the reason for desiring a complete pedigree. *Conclusions* are susceptible to error/conflict when the search for a common ancestor does not include complete pedigrees for all individuals involved.

Once the common ancestor has been identified and the strengths and weaknesses of the compiled genealogies have been characterized, researchers generally extract and present enough *information* to document lineages from each subject to the common ancestor.

DEVELOPING A RESEARCH PLAN

In genealogical research practice, developing a research plan is about enumerating the *sources* that are likely to contain *information* that can be used to answer a research question, then developing a plan to exhaustively search those *sources* to gather the *information* that answers the research question. Genetic genealogists answer questions like the following:

- Which persons, if genotyped, are likely to possess the atDNA needed to answer the research question?
- If these persons are genotyped, how can I access the genotypic and genealogical *information* necessary to answer the research question?
- If these persons have not been genotyped already, what can be done to obtain their genotypes?
- What can be done to prepare to make use of these *sources* when they become available?

IDENTIFYING GENOTYPE SOURCES

Genetic genealogy might be easier if everyone was genotyped. This is not a realistic option. Who, then, are the priorities?

If research questions address distant generations, the highest priority genotypes are the genotypes separated from these distant generations by the fewest meioses. If one is looking for the parents of a 3rd great-grandparent, it would be better to genotype one's parent, aunt/uncle, or grandparent (who also share a relationship with that 3rd grandparent) than oneself. It may be that a great-uncle or a second cousin twice removed is available to be genotyped. Seeking the youngest child of

the youngest child, etc., might yield a living descendent that could be genotyped with a smaller generational gap than one's own close family.

If one's research interest is broad (not focused on a particular ancestor) but interest is high for distant generations, start with members of the oldest living generation (e.g., grandparents).

If seeking to access the genome of a parent not available for genotyping, gather the genotypes of their children.

In many cases (e.g., seeking biological parents), the persons with the needed genotypes are not known in advance. The only option is to wait for the right person(s) to become interested in exploring their genotype. There is plenty of reason to remain hopeful. Bartlett estimates that genotype databases are doubling every fourteen months.[188] Today's missing genotype may be available next year.

Phased genotypes help eliminate matches with genotypes that are IBC. Parent genotypes are the easiest path to a phased genotype—making parent genotypes invaluable to researching with one's own genotype.

For some, a dearth of matching genotypes inhibits research. Making one's genotype available in all the major genotype databases ("fishing in all of the ponds") could help in finding the needed matches. Figure 9 shows the value of genotyping other relatives to uncover genotypes that did not match one's own. Some of the values in Figure 9 might be surprising; for instance, genotyping an aunt/uncle is more likely to reveal a new fourth cousin than testing a parent.

Distant Relationship	Sibling	Uncle/Aunt	Niece/Nephew	Parent	Grandparent	First Cousin	Second Cousin
3rd cousins	87%	99%	64%	96%	100%	94%	97%
4th cousins	42%	78%	25%	63%	92%	57%	64%
5th cousins	15%	37%	8%	26%	58%	22%	26%
6th cousins	4%	13%	2%	9%	23%	7%	8%
7th cousins	1.5%	4%	0.8%	3%	8%	2%	3%
8th cousins	0.43%	1.23%	0.23%	0.81%	2.3%	0.65%	0.77%

Figure 9: The likelihood that a genotype of a close relative from one side of the family will match a genotyped distant cousin from the same side of the family that does not match one's own genotype.[189]

RECRUITING SOURCES

Genetic genealogy is collaborative—requiring lots of participation and sharing.

Access to the right genotypes often requires recruiting relatives to be genotyped (which often includes paying for the creation of those genotypes). Some will not be willing to participate. Others might be willing but will find the sampling mechanism to be difficult (e.g., older persons often have difficulty providing a saliva sample), necessitating a different sampling technique (e.g., a cheek swab) and different genotyping service. Recruiting will likely require education—i.e., helping relatives understand the ramifications of being genotyped, helping them understand the results expected, helping them interpret the results received, etc.

Genotyping services do not provide all the information and tools necessary to use genotypes genealogically. Genetic genealogists need to know the following for each region shared with another genotype: the chromosome on which it is located, the start and end positions, the number of SNPs, and the segment size. Some of the required genotype-to-genotype comparisons cannot be made—in some services (e.g., AncestryDNA™), not at all; in others (e.g., Family Tree DNA™ and 23andMe™), they can be managed but only with the cooperation of other matching individuals. Recruiting genotypes of interest to other services (e.g., GEDmatch) that provide the necessary tools and information will maximize genotype utility. Again, this can require education—i.e., assisting these individuals with the technical challenges of doing so, helping them to understand privacy issues, etc.

Genetic genealogists require collaboration in the form of shared genealogies—i.e., published (public or private) genealogies that give information about one's ancestors. Researchers need to be prepared to share the genealogy of their genotyped person-of-interest. They will be requesting access to and examining genealogies compiled by others. They may need to assist others in creating or expanding pedigrees.

PREPARING YOUR OWN FAMILY TREE

If one's genealogy is known (or knowable), collect and document that genealogy. How much of one's genealogy is needed? Speed and Balding's work suggests an ideal tree might include twenty generations of ancestors and fully populated descendant trees for each ancestor—an impossible standard.[190,191] While the

reasons for these statements have yet to be discussed, the author wishes to note that no one has (or can even create) such an ideal family tree. The essential work, then, is to gather the knowable genealogy—both ancestors and their descendants.

INFORMATION GATHERING IS ON-GOING

Information gathering is an on-going effort. Genotyping services continue to expand their databases which results in continual expansion of genotype match lists. New matches mean new cousins and new opportunities to collaborate. It will be necessary to revisit match lists, triangulated groups, research *conclusions*, etc.—keeping information current, adding individuals to circles of collaboration, and updating research *conclusions* to incorporate new findings.

HYPOTHESIS TESTING

With a critical mass of *information* gathered (i.e., matches to be analyzed, and compiled genealogies relevant to those matches), the *information* can be used to answer genealogical questions.

Consider, again, the genealogical research process. *Information* becomes *evidence* as it is used to answer a research question. *Evidence* suggests a tentative answer to a research question. At least two independent pieces of *evidence* must be correlated to have a testable *hypothesis*. Each *hypothesis* must be scrutinized—i.e., tested. A *hypothesis* that stands up to scrutiny is accepted as a *conclusion*. Testing safeguards the researcher from erroneous *conclusions* and helps to shine a spotlight on the strengths and/or weaknesses of those *conclusions*.

TENTATIVE ANSWERS

In genetic genealogy, researchers work with two kinds of *information*—genome *information* and relationship *information*—to find tentative answers to research questions, these answers becoming *evidence* of genealogical relationships.

EVIDENCE FROM QUANTITATIVE INFORMATION

The first *information* item used as evidence of relationships is the total amount of atDNA shared between the two individuals—a result of comparing two genotypes. This total can be (and has been) used to estimate the relationship between two individuals. This estimate can be very good for close relationships but breaks down

as relationships become more distant. The following two figures illustrate the use of this basic metric.

The first comes from Bettinger's *Shared cM Project* (Figure 10). Researchers report the total amount of shared atDNA between two individuals along with the relationship believed to exist between them. Bettinger classifies each relationship within a "degree" of relatedness. Outliers for a given "degree" most likely represent errors in the reported relationship.

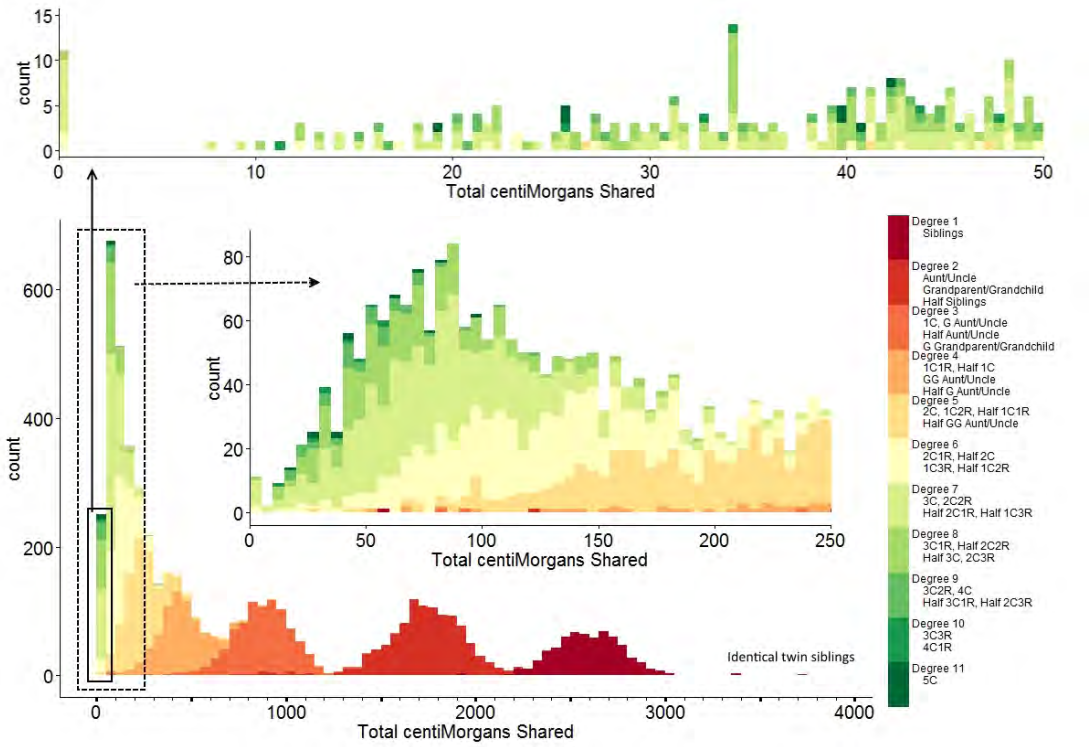


Figure 10: Distributions of shared cM by relationship type from the Shared cM Project.[192]

When considering these distributions, note that the distributions for the first several degrees on Bettinger's scale are very distinct from their neighboring degrees—with almost no overlap. If the total amount of shared atDNA between two individuals falls in the range of a first- or second-degree relationship, it would be illogical to argue that the relationship is somehow a third or fourth degree relationship. Contrast that to two individuals that share 100 cM of atDNA where it is much more difficult to discriminate relatedness. 100 cM is still in range with fifth-degree relationships, yet may also be in the ninth- or even tenth-degree ranges on Bettinger's scale.

Henn et al published a plot of data from a 23andMe™ data set (Figure 11).

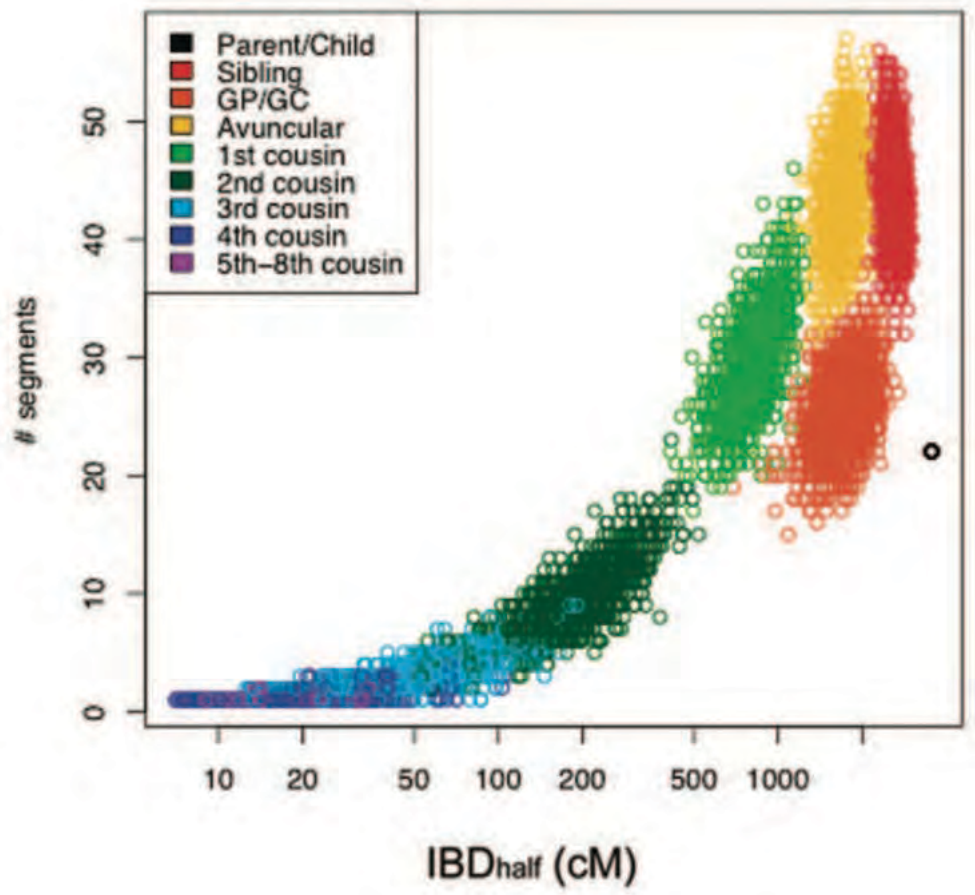


Figure 11: Total shared IBD_{half} atDNA (x-axis) and number of segments shared IBD_{half} (y-axis).[193]

Note the distinct data groupings for parent/child, sibling, grandparent/grandchild, avuncular and 1st cousin relationships in Figure 11. For these close relationships, each grouping seems to have little overlap with neighboring groupings. As groupings for distant relationships are considered, boundaries are no longer distinct. If one considers two genomes sharing five IBD segments totaling 100 cM, it is difficult to definitively identify the relationship shared between them.

As shown above, using the total amount of atDNA sharing as evidence of a particular relationship can be viable for close relationships. The data in Figure 11 suggests it would be safe to do so for first cousins and other closer relationships. The distributions from the *Shared cM Project* seems to suggest there may even be trouble with first cousin relationships and that the safe realm ends one degree closer than first cousin. The *Genetic Genealogy Standards* limit conclusions based solely on total sharing to first degree relationships.[194]

Consider the case of an adoptee—identified as an8181—searching for her birth

parents via AncestryDNA™. Some point after receiving her initial results, AncestryDNA™ reported a match with a man designated M.G.[195] AncestryDNA™ reported that an8181 and M.G. shared 454 cM across 22 segments and estimated their relationship to be 1st-2nd cousins. Using the distributions in Figure 10, 454 cM of shared atDNA falls near the peak of Bettinger's Degree 4 relationship distribution. It would appear that the upper end of the Degree 5 distribution might overlap slightly with the Degree 4 distribution at 454 cM, but a Degree 4 relationship is clearly the highest probability answer.

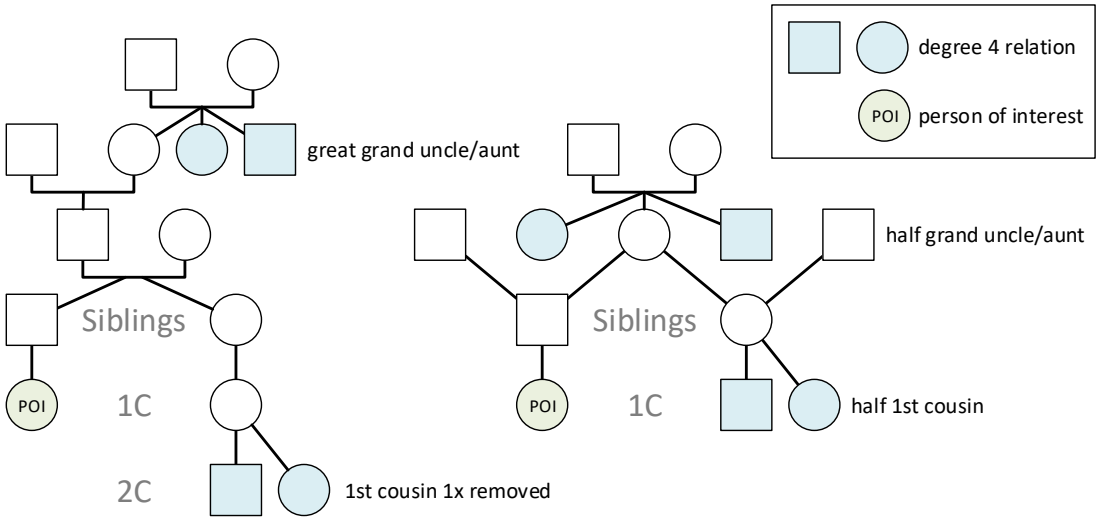


Figure 12: Relationships consistent with Bettinger's Degree 4 relatedness for a person of interest.[196]

Bettinger gives the following as Degree 4 relationships: 1st cousin 1x removed, half 1st cousin, great grand uncle/aunt, or half grand uncle/aunt.[197] When an8181's biological father was identified (see *Appendix A*), M.G. was shown to be her half 1st cousin—a perfect fit with a Degree 4 relationship.[198]

A NOTE ABOUT QUANTITIES

In genetics, segment size can be expressed in terms of physical length (typically in million base pairs—or Mbp) or in terms of genetic distance (centimorgans—cM). The total amount of sharing (the sum of the size of all the shared regions) is typically expressed in genetic distance (cM). When the genetic genealogist encounters information about size in the literature, tools, and reports that they use, sizes are generally expressed in cM—but not always.

While one might be tempted to consider the centimorgan to be a unit of length, it can be misleading when needing to consider an actual physical length. One can find

many instances in the genetic genealogical community where a conversion is given between genetic distance (cM) and physical length (Mbp)—$1\ cM\ \cong 1\ Mbp$.[199] Given all of the matching segments m for GT999 where $10\ cM\ \geq size(m) < 11\ cM$, the smallest physical length encountered among these segments was 2.2 Mbp, and the largest was 39.9 Mbp.[200] So when considering heuristics that are given with genetic distances, seek to use sizes given in cM; and when considering heuristics given with physical lengths, seek to use sizes given in terms of base pairs.

EVIDENCE FROM TRIANGULATION

Genetic genealogists use triangulation to reliably attribute a matching segment (HIR) as coming from a particular ancestor. However, the literature explaining triangulation often fails to make plain the reasons why triangulation is effective. Pundits in the genetic genealogy community do not seem to agree broadly as to the elements required for a triangulated *conclusion*. Moreover, the community does not seem to apply uniform methods and criteria when evaluating the strengths and/or weaknesses of a triangulated conclusion. These deficiencies can be addressed if triangulation is decomposed into its fundamental building blocks and re-presented in the context of the genealogical research process.

AN AXIOM AND A THEOREM

Triangulation is born out of the following principle:

> A segment of atDNA shared by two individuals and received from a common ancestor is IBD.

This definition can be restated as follows:

> A segment shared by two individuals that is IBD was received from a common ancestor.

This principle is axiomatic. It is an axiom because it is true by definition. In mathematics, axioms are the premise for all further reasoning. This axiom is the cornerstone upon which genetic genealogists build—the basis for further reasoning—as they work to deduce identities and relationships from the genetic *information* related to their research interest.

The axiom above leads to the following triangulation theorem:

$$m \wedge a \wedge t \rightarrow c$$

where

m: a matching segment of atDNA, that might be IBD, that is shared by the individuals being considered

a: a recent ancestor, believed to be unique (the only one), that is shared in common among the individuals being considered

t: testing that supports the IBD and uniqueness suppositions in m and a (i.e., that supports a conclusion that the matching segment m was received IBD by the individuals being considered from the recent common ancestor a)

c: the contributor of the matching segment m for the individuals being considered was the common ancestor a.

In prose, it reads:

If m and a and t, then c.

In other words, the triangulation theorem can be expressed as follows:

If, for the individuals being considered, there is matching segment of atDNA (m) that might be IBD and a recent ancestor (a), believed to be uniquely common among them, and if testing (t) supports that the segment shared among them is IBD from the specific common ancestor, we can deduce that the recent common ancestor was the contributor (c) of the identified matching segment.

THE FUNDAMENTAL BUILDING BLOCK OF TRIANGULATION

The question that is being asked when triangulating is this: Can the presence of a shared segment (HIR) between two genotyped *source* individuals be attributed to a particular ancestor of those persons?

To answer this question, the genetic genealogist seeks *information* to answer two intermediate questions:

1. Does the *information* that resulted from genotyping the two *source* individuals substantially match (i.e., in an IBD-consistent manner) over the sequence identified by the segment in question?
2. Is there *information* in one or more compiled genealogies to suggest that the two *source* individuals share a single common ancestor?

If the answer to both of these intermediate questions is affirmative, there is *evidence* that the atDNA in common between the identified *source* individuals was contributed by the ancestor in common between them.

Notice that m and a in the triangulation theorem are answers to these intermediate questions. If the answers to these questions, together, are a piece of evidence that a particular ancestor contributed a particular segment (HIR) to one's genome, it is useful to refactor the triangulation theorem as follows:

$$\mathcal{E} \wedge t \rightarrow c$$

where

m, a, t, and c: as specified in the theorem above

$\mathcal{E}$: the conjunction of m and a—that is: $m \wedge a \rightarrow \mathcal{E}$

In prose, $m \wedge a \rightarrow \mathcal{E}$ says that if there is *information* indicating a shared match (m), and *information* documenting a common ancestor (a), there is *evidence* ($\mathcal{E}$)—a tentative answer—that the common ancestor (a) could be the contributor (c) of the shared match (m).

> Often, the common ancestor between two donors is actually given as two ancestors—a father and a mother. Biology dictates that only one person in the couple could have actually contributed the matching segment (HIR) because an IBD segment is part of a haploid chromosome, and a haploid chromosome is received from only one parent.

In prose, $\mathcal{E} \wedge t \rightarrow c$ says that if the *evidence* ($\mathcal{E}$) withstands testing (t), there is a tentative answer suggesting that (c) is the contributor.

Considered as an entity, $\mathcal{E}$ is the fundamental building block in triangulation. A single instance of $\mathcal{E}$ is an item of *evidence* that becomes a tentative answer to the triangulation question. If two (or more) items of *evidence* (instances of $\mathcal{E}$) give the same answer (i.e., the items correlate), the genetic genealogist has a hypothesis that can be tested.

IDENTIFYING TRIANGULATION BUILDING BLOCKS

Given $\mathcal{E}$ as the fundamental building block and given that $\mathcal{E}$ is the result of m and a, an examination of m and a is merited.

ISOLATING m

m is the result of comparing two genotypes. In its simplest form, two individuals—considered *sources* in the context of the genealogical research process—submit biological material for genotyping. The genotype gives *information* about selected

SNPs dispersed throughout the autosomes. Because humans are diploid (each somatic cell containing two complete copies of the human haploid genome), a SNP measurement yields two allele values—one allele from each haploid copy. In the case of a SNP, each allele represents a nucleotide in a base pair; the second nucleotide in the base pair can be inferred from the value of the first—each nucleotide having a known complimentary base. Thus, the genotype is a collection of SNP allele values, each representing a nucleotide in a base pair at a known location on a known chromosome.

Matches (m) are determined by comparing two genotypes to discover matching segments (SNPs in sequence along a haploid chromosome) that might be IBD. Currently, all companies that offer a genome sequencing service also provide a list of genotypes that match the given genotype. Knowing that a match exists is not sufficient. A precise description of the match is required for triangulation. It is necessary to know:

- the chromosome that contains the segment
- the segment begin and end locations
- the size of the match (in cM)
- the number of SNPs present in the segment.

Genotyping companies are not consistent in describing matches. Vendors do not universally provide the necessary elements described above. To overcome this deficiency, genetic genealogists must persuade matches to add their genotypes into other databases—databases with tools sufficient to fully characterize each match.

Comparing two genotypes to identify shared regions is a form of *information* correlation. It transforms *information* about allele values into *information* about shared regions.

ISOLATING a

a comes from the *information* available in compiled genealogies. In the context of triangulation, compiled genealogies take on the role of *sources* in the genealogical research process. These genealogies are most useful when the persons and relationships contained in them are, themselves, documented using the GPS. A lineage extracted from such a genealogy showing an individual's relationship to the common ancestor is the *information* sought from these *source* genealogies.

A common ancestor is identified by consulting one or more genealogies and identifying the lineages within those genealogies that show how individuals share a biological relationship with a single ancestor (or ancestral couple). A common ancestor is "theoretical" in that the biological relationship is presumed and susceptible to question. In this text, a statement that a common ancestor exists is a statement that lineages exist that show this shared relationship to a presumed biological ancestor (or ancestral couple). Each such statement must be analyzed, evaluated, and weighed on its merits.

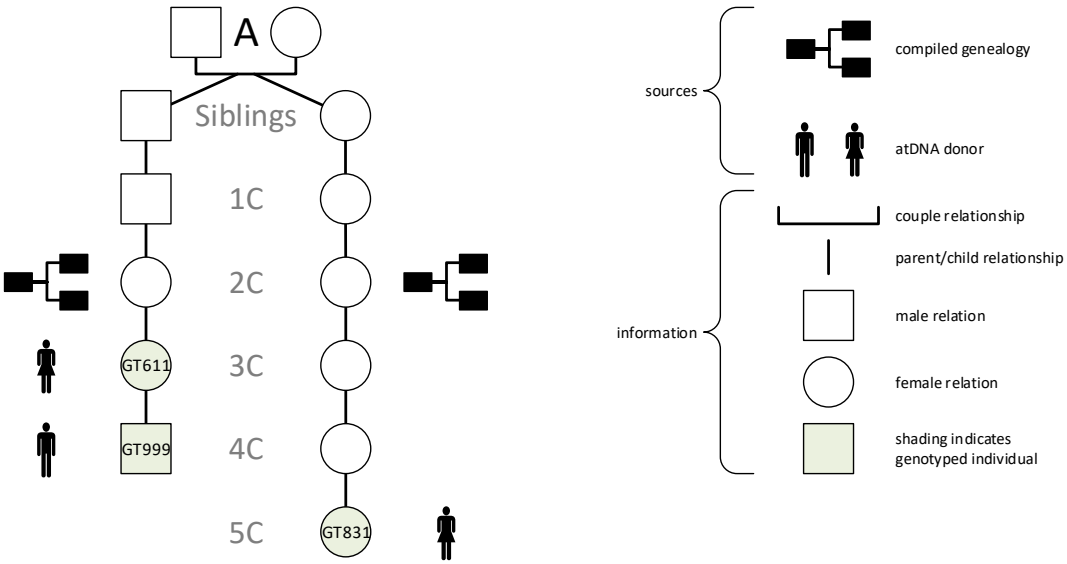
WHAT'S NEXT?

Having isolated and correlated m and a that are true for the two *source* individuals, the genetic genealogist has the first piece of *evidence*—the first instance of $\mathcal{E}$—needed to answer the triangulation question.

A single instance of $\mathcal{E}$ is not sufficient for a conclusion. It is not sufficient to form a *hypothesis* that can be tested. This is only one piece of *evidence* that the matching segment in common was contributed by their common ancestor. At least one more instance of $\mathcal{E}$ is needed—an $\mathcal{E}'$ that correlates with $\mathcal{E}$—to have a *hypothesis* that can be tested. A second genotype matching the person of interest on the same segment and with a lineage to the same common ancestor is needed.

A CONCRETE EXAMPLE

Consider the following example from research about GT999's Chr1.



GEDmatch One-to-One Comparison
Comparing GT999 and GT831

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
1	158,609,568	178,443,889	23.5	5,205

Chr 1

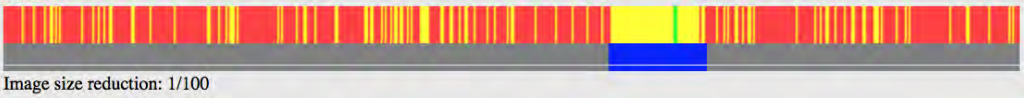
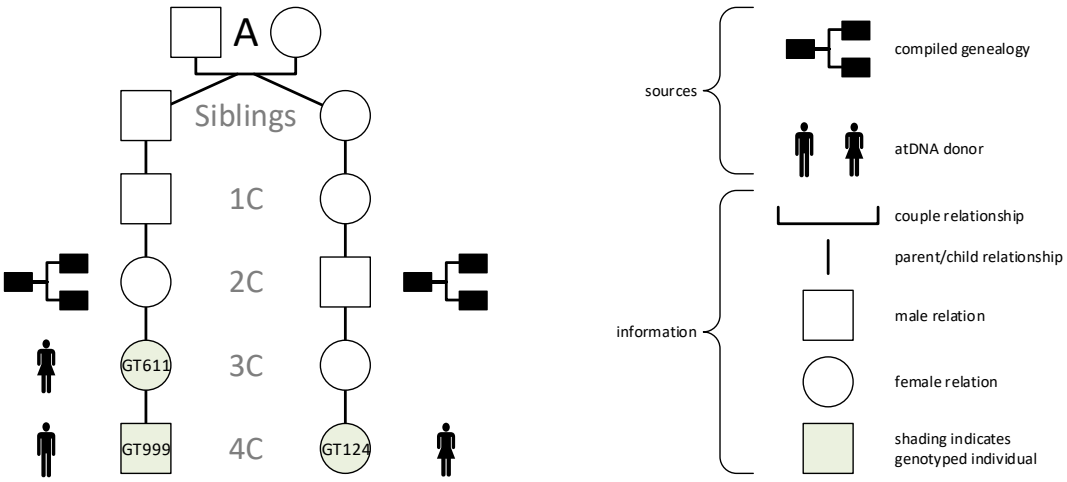


Image size reduction: 1/100

Figure 13: Graph representing the common ancestor between GT999 and GT831, and their GEDmatch comparison.[201,202,203]

In Figure 13, there is a person—identified as GT831—who matches GT999 on Chr1. The match is located between 159M and 178M, is 23.5 cM, and is represented by 5,205 SNPs. There is *information* to support supposition m. An examination of the compiled genealogies provided by GT999 and GT831 reveals that the couple—labeled *A*—contains the ancestor in common between GT999 and GT831. GT999 and GT831 are 4th cousins 1x removed. There is *information* to support supposition a. Because $m \wedge a \rightarrow \mathcal{E}$, one instance of $\mathcal{E}$—one piece of evidence—now exists to suggest that GT999 (and GT831) received the matching segment on Chr1 between 159M and 178M from one of their ancestors in the couple designated *A*.

But one item of *evidence* is not enough to have a valid *hypothesis*. A second instance of $\mathcal{E}$ is needed.



GEDmatch One-to-One Comparison
Comparing GT999 and GT124

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
1	108,512,791	119,996,565	12.9	3,009
1	155,348,641	166,653,409	20.3	3,358

Image size reduction: 1/100

Figure 14: Graph representing the common ancestor between GT999 and GT124, and their GEDmatch comparison.[204,205,206]

In Figure 14, a person identified as GT124 matches GT999 on Chr1 in two locations. One of these locations—the segment located between 155M and 167M—overlaps with the location in common with GT831. If the segment of interest is changed to be the overlapping region shared between the three genotypes—the segment between 159M and 167M (14.0 cM)—there is *information* to support supposition m.[207] An examination of the compiled genealogies provided by GT999 and GT124 reveals that the same couple *A* is common between GT999 and GT124; they are 4th cousins. There is information to support supposition a. Again $m \wedge a \rightarrow \mathcal{E}$, and there is now a second instance of $\mathcal{E}$ suggesting that GT999 (and GT124) received the matching segment on Chr1 between 159M and 167M from an ancestor in the couple designated *A*.

CORRELATING THE INSTANCES OF $\mathcal{E}$

With two instances of $\mathcal{E}$, there is still one more condition that must be met to have a *hypothesis* that can be tested. Both instances of $\mathcal{E}$ must correlate with each other.

CORRELATING m

The correlation of m between the two instances of $\mathcal{E}$ is crucial. I cannot over-emphasize this point. Failing to do this correlation is the source of many errors and will render any conclusions invalid.

Genetic genealogists usually work with a group of genotypes that match on a single segment—herein called a triangulated group. This group is often identified prior to identifying a common ancestor. All the individuals in the triangulated group need to match the same segment (HIR).

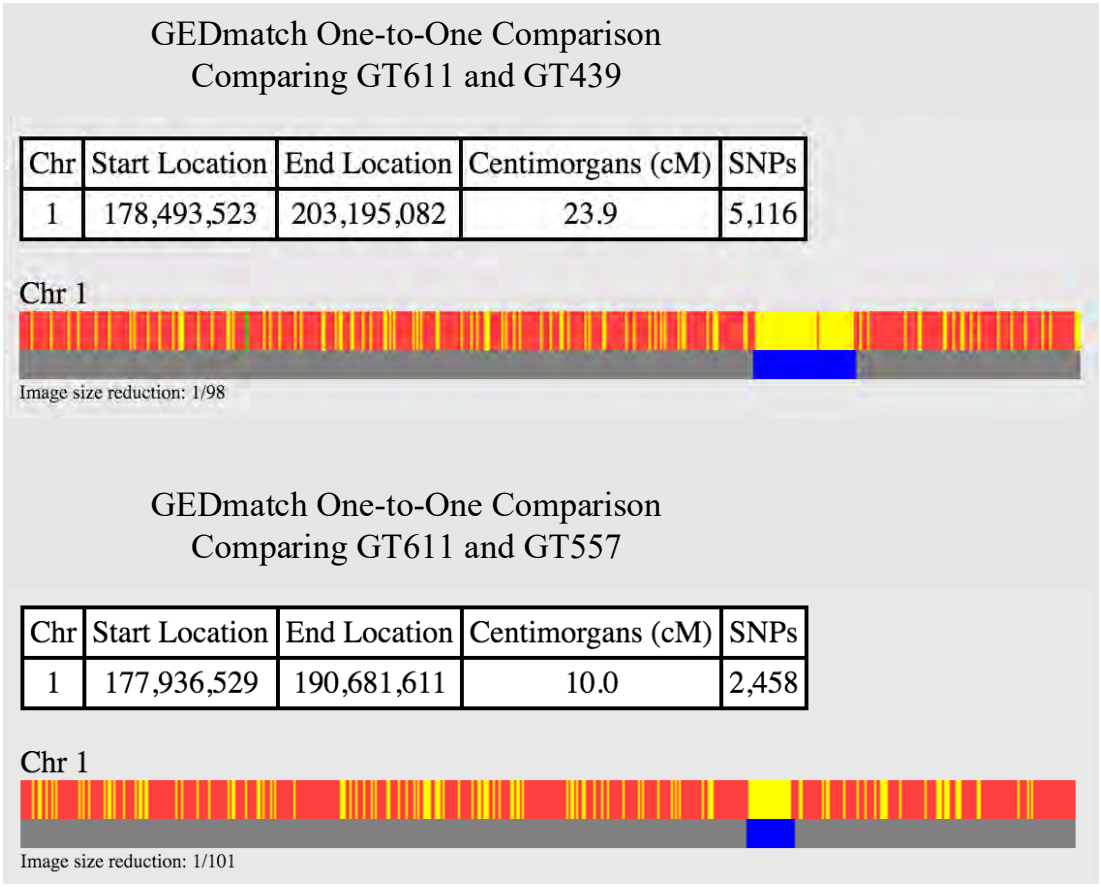
The question that needs to be answered is this: Is the atDNA in each piece of evidence from the same ancestor (even if we cannot identify which ancestor)? When considering a list of matches in a particular region, there are three possible matching scenarios. The segment could:

1. match a sequence of alleles along a person's paternal chromosome
2. match a sequence of alleles along a person's maternal chromosome
3. match a mix of maternal and paternal alleles in the region of interest, making it a false (IDC) match.

Without additional conclusions in place (like knowing one of the individuals is a paternal match), it is not possible to know which case is being encountered. However, *a third comparison—a correlation comparison—will indicate whether the three individuals match each other in the same way*.

If the genotype A matches genotype B and genotype C along a shared region of interest, a comparison that shows genotype B and genotype C matching each other over that same region ensures that all three individuals are matching the region of interest in the same way; it would also be a strong indication that the matching segment belongs to a parent haploid (case 1 or case 2 above). On the other hand, if genotype B and genotype C do not match each other, it cannot be true that the genotype A matches with genotype B and genotype C on the same chromosome. It must be that genotype B or genotype C each match separate parent chromosomes (one maternal and one paternal), or that one or both of the genotypes is falsely (IBC) matching genotype A.

As an example, consider GT611 who matches both GT557 and GT439 on Chr1 with an apparent overlap between 178M and 191M.



Chr	Start Location	End Location	Centimorgans (cM)	SNPs
1	178,493,523	203,195,082	23.9	5,116

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
1	177,936,529	190,681,611	10.0	2,458

Figure 15: Comparisons showing GT611's shared segments with GT439 and GT557 respectively.[208,209]

The comparisons in Figure 15 show the matches between GT611 and GT439 and GT557 respectively.

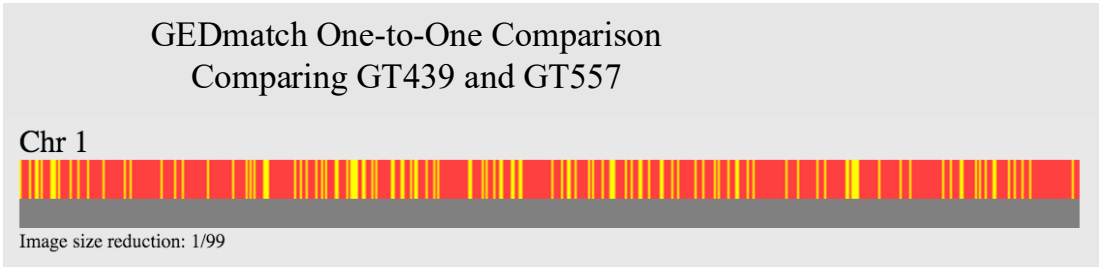


Figure 16: Correlation comparison between GT439 and GT557 showing no shared match.[210]

The correlation comparison—the comparison between GT439 and GT557—did not show a match (Figure 16). Thus, the instances of m do not correlate. [Research later showed that GT439 was a maternal match and GT557 was a paternal match for GT611.]

GEDmatch One-to-One Comparison
Comparing GT831 and GT124

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
1	119,596,656	155,734,186	10.6	2,359
1	158,879,805	166,653,409	13.2	2,373

Figure 17: Correlation comparison between GT831 and GT124 showing that correlating match exists for the segment 159M to 167M.[211]

Returning to the case of GT999 matching GT831 and GT124 on Chr1 over the segment between 159M and 167M (see Figure 13 and Figure 14), the correlation comparison—comparing GT831 and GT124—reveals a match over the same segment (see Figure 17). The instances of m do correlate.

CORRELATING a

It is intuitive that both instances of ε correlate with regard to the common ancestor a. The same couple *A* was identified as the common ancestor for both instances.

TRIANGULATION IN TERMS OF ε

How is it that two or more instances of ε are the building blocks of triangulation?

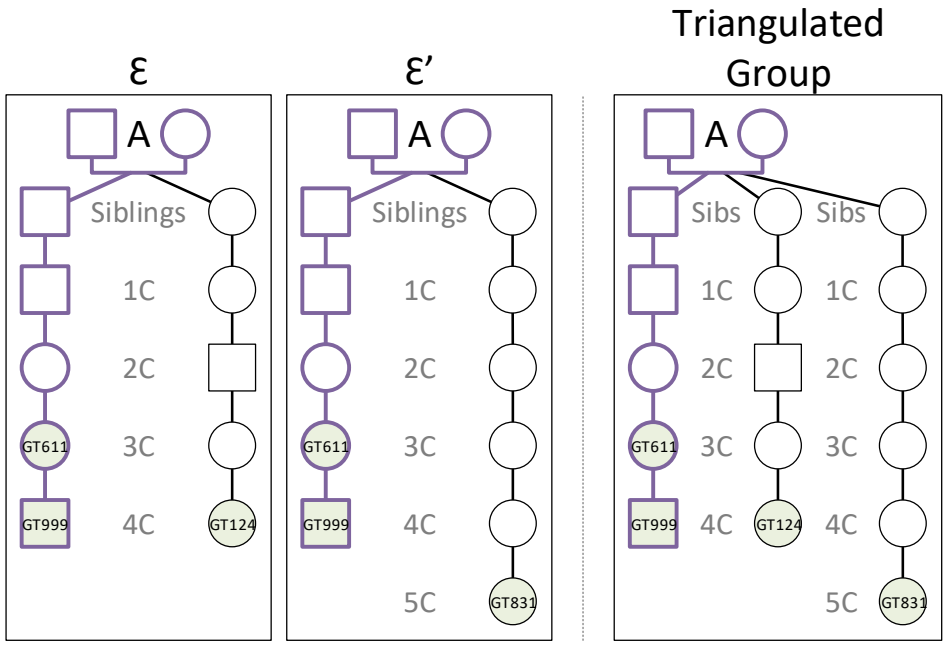


Figure 18: Two instances of ε are combined to form a triangulated group.

In the example of GT999 matching GT831 and GT124, two instances of *evidence* ($\mathcal{E}$ and $\mathcal{E}'$) have been identified. Note that the GT999's lineage is repeated in each representation of $\mathcal{E}$ (highlighted in lavender in Figure 18). If the representations of each instance of $\mathcal{E}$ are combined and the duplication removed, the result is a representation of a triangulated group, and the reason it is called triangulation begins to be apparent. *Appendix B* describes the testing of this triangulated group.

TESTING

With both instances of m correlating, and both instances of a correlating, it is safe to declare that both instances of $\mathcal{E}$ correlate. A *hypothesis* exists that can be tested.

For a *hypothesis* to become a *conclusion*, it must be scrutinized. No one *source* is foolproof. *Information* items from a *source* could be all right, all wrong, or a mix of the two (a common scenario in genealogical sources). It follows that an item of *evidence* based on *information* from a single *source* could be either right or wrong. Testing employs both analysis and correlation to evaluate the reliability of *sources*, *information*, and *evidence* used to form the *hypothesis* under scrutiny. Alone, analysis and correlation are insufficient; both types of testing must both be applied.

TESTS OF ANALYSIS

Tests of analysis examine the characteristics that affect the reliability of *sources*, items of *information*, and items of *evidence* in isolation. Tests will not prove correctness but will give insight into the likelihood of errors or misinterpretations. In genetic genealogy, tests of analysis may require an examination of biological possibilities, genome sequencing technologies and techniques, result reporting, feasibility to answer a particular question, donor motives, sample provenance, etc. Not all of these will be explored in this text. In typical scenarios, it is not necessary to examine such things as biological anomalies, provenance, and motives. The following tests of analysis should always be considered.

QUANTITATIVE CONSIDERATIONS

Given two genotypes, what can be said about the likelihood that sharing between them is IBD? What can be said about the likelihood that the sharing between them fits the relationship being examined? Some quantitative factors that affect the reliability of *conclusions* should be considered.

Matching Segment Size

Sharon and Brian Browning defined IBD in terms of shared haplotype frequency (i.e., rarity).[212] They point out that the amount of sharing (the size of the shared region) is generally used as a proxy measure of frequency—the principle being that the larger the shared region, the rarer it must be. The size of the region is important to one's confidence that the region is consistent with IBD-sharing.

Attempts to quantify the likelihood a matching segment is IBD use genotyped parent-child trios and measure the likelihood a segment shared with the child is also shared with at least one parent. As stated previously, a few have proffered anecdotal data, and John Walden has published findings from a larger dataset. A. J. Levin, using the published findings from Walden's dataset and regression, created the chart in Figure 19 to show the likelihood of IBD given matching segment sizes in cM.

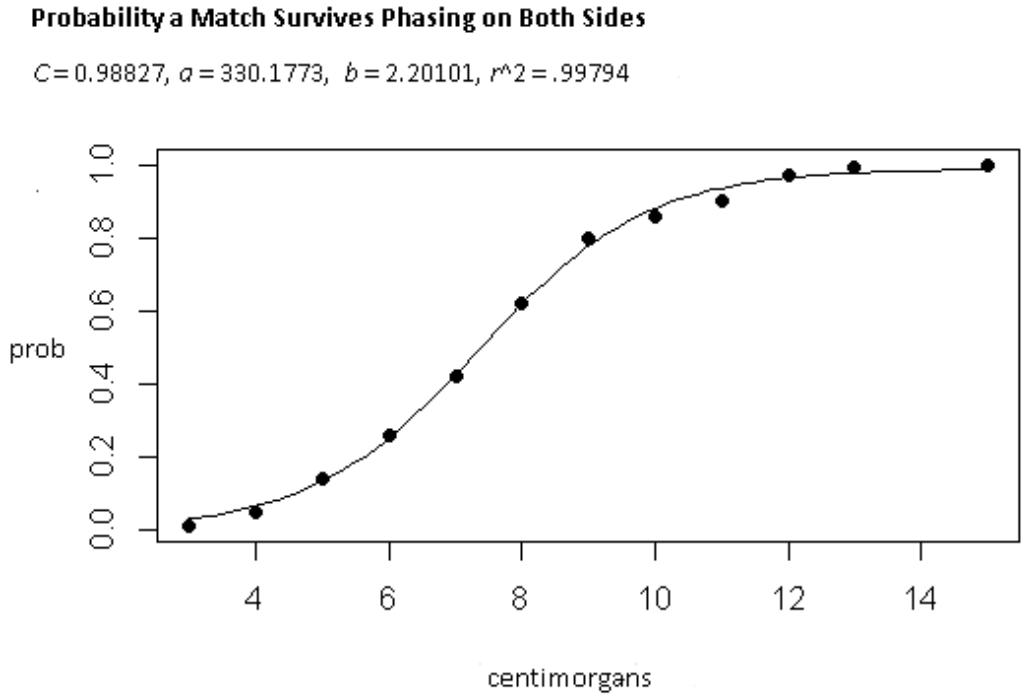


Figure 19: Probability a match survives when compared to a genotype phased with both maternal and paternal haplotypes.[213]

While Walden's findings have received criticism for lack of peer review, anecdotal evidence certainly fits well with the result. Figure 20 shows the number of matching segments, phased and un-phased, of a given size for GT999.

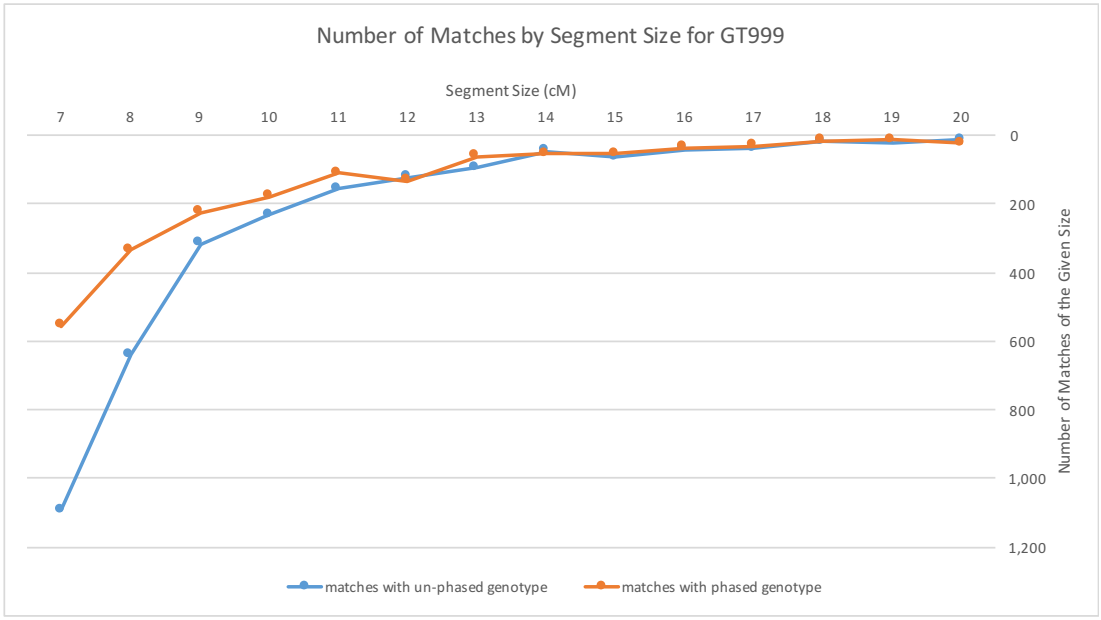


Figure 20: Number of matches by segment size for GT999.[214]

As match sizes grow larger, the number of segments of that size decreases—getting rarer. This aligns with the likelihood prediction of Figure 19. At the 7 cM size, nearly half of the segments that matched the un-phased genotype do not match the phased genotype. As match size increases, perhaps somewhere between 14-16 cM, the phased and un-phased genotypes produce roughly the same set of matches.

Even in cases where phased genotypes are used, current genotype data cannot be reliably used to detect IBD segments smaller than 5 cM. Durrand et al studied 2,952 parent-child-trios in a set of 25,432 genotyped individuals.[215] In their study, matching segments were declared IBD if a haplotype matching the child in a trio had at least an 80% segment overlap with at least one parent haplotype [perhaps a too-lenient standard?]. They state the following:

> "Most 2–3 cM segments are erroneous, and only segments longer than 5 cM have a negligible number of false positives. Indeed, when filtering solely by genetic length, all segments shorter than 5 cM must be discarded to achieve a precision value of 0.8."

As a quantitative consideration, matching segment size is important. If match size is not in a highly IBD-consistent range, conclusion reliability is poor. As Bettinger puts it: "…it is the responsibility of the genealogist to place a VERY high burden on any

argument that utilizes … small segments…."[216]

Matching segment size is not just problematic for the genetic genealogist. Genotyping companies face a dilemma when selecting matching thresholds. Should the threshold be set high (matches reported out are mostly IBD-consistent; but many real matches are not reported—i.e., many false negatives), or set low (increases the number of real matches reported; but reports many non-cousins as potential matches—i.e., many false positives). Different research objectives might benefit from being able to tune matching segment size thresholds, but the reality is that genotyping companies control initial thresholds and genetic genealogists are left to operate with the data reported forward by these thresholds. Genotyping companies seem to target a threshold that balances the number of false-negatives and false-positives for matches being reported at the threshold size.[217]

Total Shared IBD

A second quantitative question asks: Is the size of the match a reasonable fit with the relationship being tested with the *hypothesis*? This question can be answered by comparing the measured amount of sharing to the expected amount of sharing.

Relationship	#
1C	662
1C1R	606
1C2R	134
2C	598
2C1R	590
2C2R	101
3C	517
3C1R	264
3C2R	36
Aunt/Uncle	722
Grandparent/Grandchild	195
Great Aunt/Uncle	123
Great Grandparent/Grandchild	25
Half Aunt/Uncle	43
Half Siblings	125

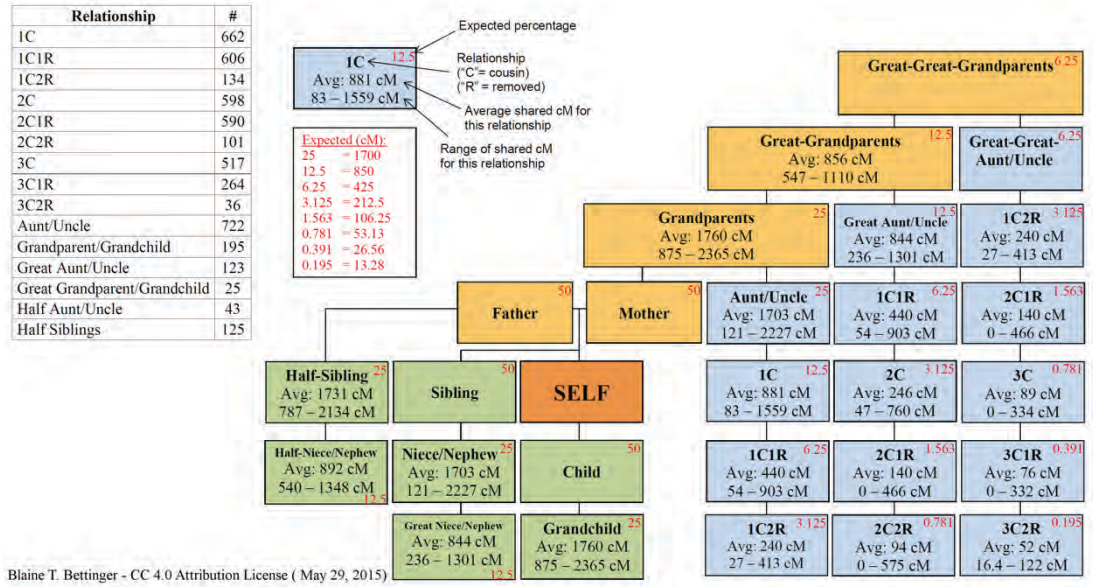


Figure 21: Data from Blaine Bettinger's Shared cM Project.[218]

As discussed in *Evidence from Quantitative Information*, the amount of sharing for close relationships should fall in an expected range. While these ranges are not exactly defined, the ranges from Bettinger's *Shared cM Project* (Figure 10 and Figure 21) can provide practical guidance for these close-in relationships. In

particular, the shapes of the distributions evident in Figure 10 will help exclude outliers that are reported in Figure 21. These outliers most likely represent data entry errors, or misidentified relationships.[219]

Tim Janzen continues to analyze expected amounts of sharing for relationships that are more distant—relationships involving 5th generation ancestors.[220] Janzen makes comparisons using 5 cM and 4 cM match thresholds and creates separate totals for matching segments that appear in these reports. If there are family groups involved (see Figure 26 and Figure 27 and the discussion related to independence), Janzen gets these same totals for each individual in the family group, then averages the totals for all of the individuals in the group (in the same generation) to create totals that represent the group. He then compares these totals with the expected amount. (See Figure 36 in *Appendix B* for an example.) Often his totals seem high, but this would be expected as some of the smaller matches included by the 5 cM and 4 cM thresholds will not be IBD. The *Shared cM Project* averages can be higher than expected for this same reason. Another reason that the averages can be higher than expected is due to additional background genealogical relationships that may not be known to the genetic genealogist. This can cause additional IBD segments to be included in the totals. Sometimes accurate chromosome mapping can identify these shared segments and they can then be excluded from the averages.

As research moves to common ancestors beyond the 5th generation, the expected amount of sharing is not so readily identified. The halving of the expected amount of shared atDNA would predict sharing at 3.32 cM for a fifth cousin—an amount that has already been flagged as untenable given current genotyping.[221] In contrast, GT999 appears to share 29.6 cM with his fifth cousin GT880.[222,223,224] Is this unreasonable because it more closely approximates the expected amount of sharing for a third cousin once removed? A researcher might find this match hard to accept because it seems incongruous to the expectations. Additionally, it is clear from the data in Figure 21 (and Figure 11) that some cousins in the third cousin range (and beyond) do not share any atDNA. This raises two questions. What is the probability that two people will share any atDNA IBD through a given ancestor? If sharing exists, how much sharing is expected?

Table 1 | **Properties of genomic regions shared IBD by two individuals from *G* generations in the past**

Relationship	*G*	*A*	$\theta = E[\theta']$	95% CI of θ'	$P[\theta' > 0]$	E[#SR]	μ_G (SD)
Sibling	1	2	$0.25 = (1/2)^2$	(0.204, 0.296)	1.000	85.9	31.1 (35.2)
Half-sibling	1	1	$0.125 = (1/2)^3$	(0.092, 0.158)	1.000	42.9	31.1 (35.2)*
First cousin	2	2	$0.062 = (1/2)^4$	(0.038, 0.089)	1.000	37.5	17.8 (21.5)
Half-cousin	2	1	$0.031 = (1/2)^5$	(0.012, 0.055)	1.000	18.8	17.8 (21.5)*
Second cousin	3	2	$0.016 = (1/2)^6$	(0.004, 0.031)	1.000	13.3	12.5 (15.4)
Half-second cousin	3	1	$0.008 = (1/2)^7$	(0.001, 0.020)	0.995	6.7	12.5 (15.4)*
Third cousin	4	2	$0.004 = (1/2)^8$	(0.000, 0.012)	0.970	4.3	9.6 (12.0)
Half-third cousin	4	1	$0.002 = (1/2)^9$	(0.000, 0.008)	0.834	2.2	9.6 (12.0)*
	5	1	$(1/2)^{11}$	(0.000, 0.004)	0.431	0.7	7.9 (9.9)
	6	1	$(1/2)^{13}$	(0.000, 0.001)	0.160	0.2	6.6 (8.4)
	8	1	$(1/2)^{17}$	(0.000, 0.000)	0.015	0.0	5.1 (6.5)
	10	1	$(1/2)^{21}$	(0.000, 0.000)	0.001	0.0	4.1 (5.3)

CI, credible interval; SR, shared region. We consider only IBD (identity-by-descent) sharing that results from the direct lineage path of length *G* from each ancestor to each individual. *A* denotes the number of common ancestors: if $A = 2$, then these ancestors are mates, and the two individuals descend from distinct offspring of this union. θ' is the realized IBD genomic fraction from the indicated common ancestors, for which we show the expected (E) value (which is equal to the coancestry (θ)), the equal-tailed 95% CI and $P[\theta' > 0]$, the probability that the two individuals share any genomic region IBD from those ancestors. Also shown are the average number of SRs and, conditional on $SR > 0$, the expected region length in megabase pairs (μ_G) and its standard deviation (SD). Estimates are based on 10^5 Type A simulations (see Supplementary information S1 (box)). *The value shown is the same as the one above by definition.

Figure 22: Properties of genomic regions shared IBD by two individuals G generations in the past.[225]

Speed and Balding address these two questions. In Figure 22, column "P[θ' > 0]" gives the probability that two individuals will share any region IBD from the ancestor *G* generations in the past that is common between them. If a region is shared IBD, column "μ_G (SD)" gives the average size of that shared region with its standard deviation in parenthesis.

Be sure to note that all of the size information given by Speed and Balding is in Mbp.

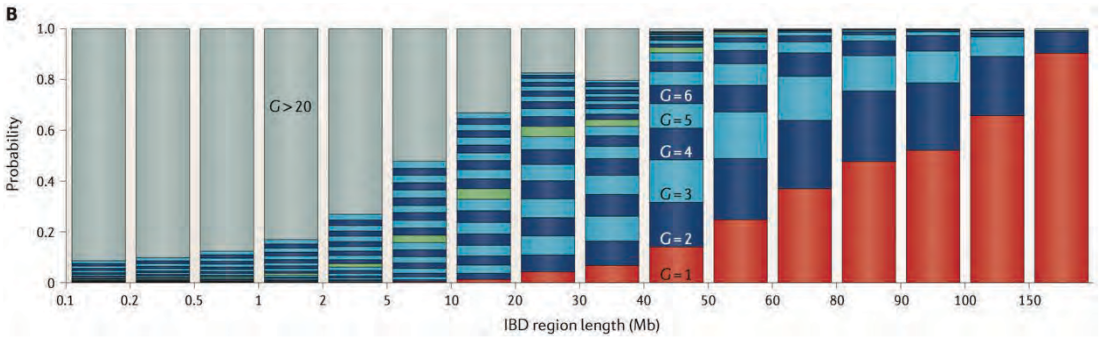


Figure 23: Statistics of IBD genomic regions.[226] *For all IBD regions arising from a common ancestor within the last 50 generations, the bars show how the distribution of the generation of the common ancestor depends on the length of the region. From bottom to top in the graph, the tranches correspond to G = 1 (red), G = 2...9 (alternating dark and light blue), G = 10 (green), G = 11...20 (alternating dark and light blue) and G >20 (grey).*

Figure 23 is a visual representation of Speed and Balding's findings regarding the possible common ancestors that might be associated with IBD segments of a given

size. In the case of GT999 and GT880, the shared segment is 23,685,019 Mbp in length. Using the graph in Figure 23, there is about a 40% chance that the common ancestor associated with a shared segment of this size is more than 10 generations away from these two cousins, so it is possible for cousins at *G*=6 to share a segment of this size.

EVALUATING COMPILED GENEALOGIES

A compiled genealogy is typically an authored *source*. In some cases, however, genealogies are derivatives—copies of someone else's genealogy—and an in-depth analysis might require hunting down the original authored *source*. Questions that might be considered when examining a compiled genealogy include:

1. What motivated the creation of the compiled genealogy? Hobby? Society membership? Something else? Does this motivation make the research susceptible to bias?
2. Was the compiler careful and professional in their work?
3. Is the genealogy sourced? Were the best and most accurate sources used?
4. Was/is the genealogy open to challenge and correction?
5. How complete is the genealogy? Are there gaps? How many generations are complete?

The question of completeness is of particular importance in the hunt for the most recent common ancestor (MRCA).

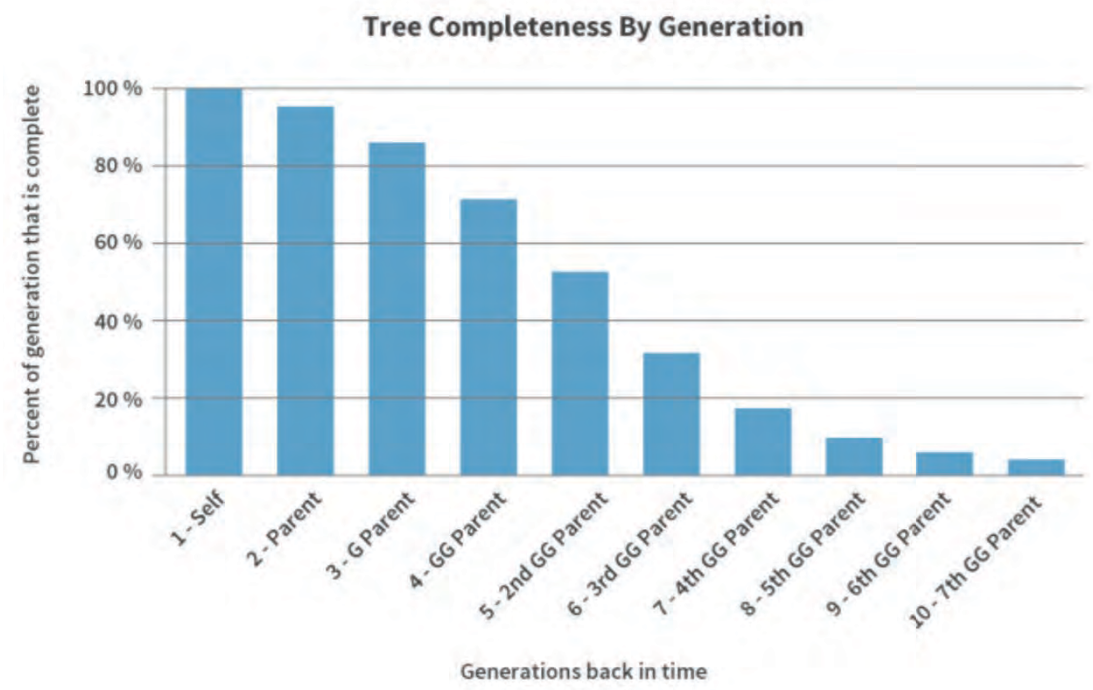


Figure 24: Sample of genealogies studied in AncestryDNA™ DNA Circles™ White Paper.[227] *Bars show the proportion of ancestors whom, for that generation of a pedigree, are known and documented (averaged over all studied pedigrees).*

Data about pedigree completeness provided by AncestryDNA™ (Figure 24) highlights this issue. The 512 seventh great-grandparents that need to be identified when seeking a MRCA in the 10th generation (Figure 25) is a hefty requirement and is, in many cases, impossible to satisfy. Being able to evaluate only 500 of the possible 1,023 pedigree slots makes a declaration of a single MRCA susceptible to doubt. *Conclusions* based on the *information* from such genealogies are made weaker by their incompleteness.

Generation	Ancestors in Generation	Number of Ancestors
15th	16,384	32,767
14th	8,192	16,383
13th	4,096	8,191
12th	2,048	4,095
11th	1,024	2,047
10th	512	1,023
9th	256	511
8th	128	255
7th	64	127
6th	32	63
5th	16	31
4th	8	15
3rd	4	7
2nd	2	3
1st	1	1

Figure 25: Number of ancestors (pedigree slots) in each generation, and the total number of ancestors that need to be searched (by generation) when seeking to identify a MRCA at that generation.

TESTS OF CORRELATION

Tests of correlation examine whether independent items are in agreement. Items in agreement may become *conclusions*. Items in disagreement are in conflict. Conflicts must be resolved before *conclusions* are possible.

INDEPENDENCE VS. RELATEDNESS

Correlation is only valid if the items being compared are independent. Independence connotes separate and distinct informants. Related items come from the same informant. It does not make sense to correlate related items; these items are just alternate representations of the original, and the original will always correlate with itself.

Independence is a key consideration when triangulating. The lineages to the common ancestor need to be independent. Having three (or more) independent lineages to the common ancestor are what gives triangulation its power to prove relationships.

Value shown is cM total of matching segments over minimum threshold.

Kit	name	GT999	GT163	GT625	GT177	GT381	GT136	GT978	GT789	GT606	GT491
GT999			3585.3	36.0	36.0	19.4	15.1	13.4	12.7	13.4	18.9
GT163		3585.3		36.2	36.5	24.9	17.3	12.0	17.4	17.1	17.8
GT625		36.0	36.2		1058.7	2074.5	20.7	26.7	26.4	11.9	27.2
GT177		36.0	36.5	1058.7		1095.3	17.6	11.4	12.0	11.7	11.7
GT381		19.4	24.9	2074.5	1095.3		15.5	12.3	12.2	12.2	12.2
GT136		15.1	17.3	20.7	17.6	15.5		12.6	18.1	27.0	17.9
GT978		13.4	12.0	26.7	11.4	12.3	12.6		3587.0	1565.9	2061.4
GT789		12.7	17.4	26.4	12.0	12.2	18.1	3587.0		3587.1	3587.1
GT606		13.4	17.1	11.9	11.7	12.2	27.0	1565.9	3587.1		2550.6
GT491		18.9	17.8	27.2	11.7	12.2	17.9	2061.4	3587.1	2550.6	

Figure 26: Total amount of atDNA sharing between members of the group that triangulates on a matching Chr4 segment from 187M to 191M.[228]

Figure 26 shows the comparison of ten individuals that match a segment on Chr4 (187M to 191M). The question is: How many independent answers to the triangulation question—instances of ε—are possible from this group? One might be tempted to say nine—one for each individual that can be paired with the researcher's person-of-interest. However, only three independent answers are possible because there are family groups (identifiable in Figure 26 as rows with green clusters on the diagonal) that contribute related information and cannot be considered independently.

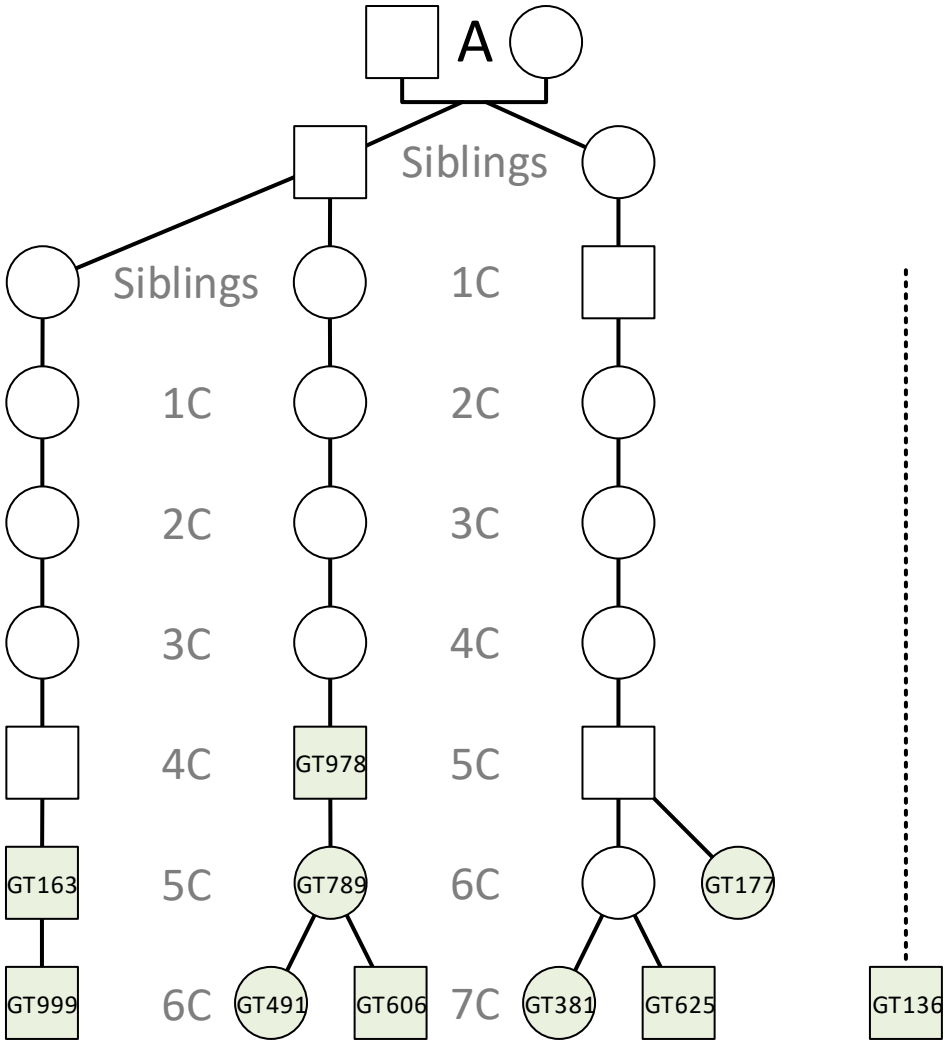


Figure 27: Representation of the lineages (known and unknown) in a triangulated group for a Chr4 segment (187M to 191M).[229,230,231]

The first group (GT163 and GT999) are a father/son pair (see Figure 27). The son received his copy of the matching segment from his father. The son cannot add any independent information about the lineage between the father and the common ancestor because he shares the same lineage (and the same set of meiosis events). The son's answer is derived from the father's answer and is, therefore, related.

The second group (GT978, GT789, GT491 and GT606) is a father, his daughter, and two of her children (see Figure 27). The mother and her children in this family group received their copy of the matching segment from GT978. The answers that GT789, GT606 and GT491 would provide would all be related to the answer that GT978 provides.

The third group (GT177, GT625 and GT381) received their copy of the matching segment from their father/grandfather (see Figure 27). For the subsequent six generations that lead to the common ancestor from their father/grandfather, these three can add no independent information; their answers are identical—from the same source.

The only other possibly independent answer could come from GT136 who has no close familial relationships with other members of the group. His exact relationship to the group remains unknown.

Even though this triangulated group has ten people that match the segment being considered, only three independent instances of ε are possible. *Appendix C* describes the testing for this triangulated group.

TESTING THE IBD ASSERTION

One of the primary assertions that needs to be accepted is that the matching segment being considered was received IBD. Tests of correlation are crucial in establishing such claims. If a match stands up to quantitative testing (size is consistent with IBD, shared total is plausible), there are a number of ways that correlation is used to show/deny plausibility that a matching segment was received IBD.

PHASED MATCHING

One of the best mechanisms for eliminating IBC matches is to make comparisons with phased chromosome data. For example, GT999 has 7933 matches over the default GEDmatch thresholds (matching segments of at least 7.0 cM and made up of at least 700 SNPs); using his phased genotypes (created with genotypes from both parents) reduced the number of matches to 2660.[232] Two thirds of the default matches were eliminated as IBC matches.

AncestryDNA™ uses phasing to help with their matching algorithm.[233] GEDmatch allows the users to generate and use phased data.[234] Tim Janzen, David Pike and Felix Immanuel have published utilities to phase genotypes.[235]

The best phased genotypes are based on child-parent trios; phasing is still possible

with a child-parent duo. It is possible to phase data without a parent; Janzen has published information about this process.[236]

Using phased data ensures that the allele sequence used in comparisons is an allele sequence that actually exists on a single haploid chromosome received from a parent, not just a random mash-up of alleles from both parental chromosomes.

Generational Matching

If genotype has parents and/or grandparents in line with the lineage of the proposed common ancestor that are also genotyped, comparing the match with each related genotype in line with the match can be telling.

In Figure 27, consider the family group made up of GT978, GT789, GT491 and GT606. The fact that GT491 and GT606 can show they received the matching segment IBD from their mother, and that GT789 can show she received it IBD from her father gives credence to the claim that GT978 received it IBD from his ancestors.

If, instead, the grandfather and a grandchild both share the match, but the mother does not, how could it be that the matching segment was received IBD? This is a biological impossibility. The grandchild could not have received it IBD unless the mother also shares the match.

Intermediate Common Ancestors

The concept of an intermediate common ancestor is related to generational matching. Jim Bartlett wrote a notable blog on this topic.[237] In Figure 28, consider a hypothesis that GT654 received the identical segment under consideration IBD.

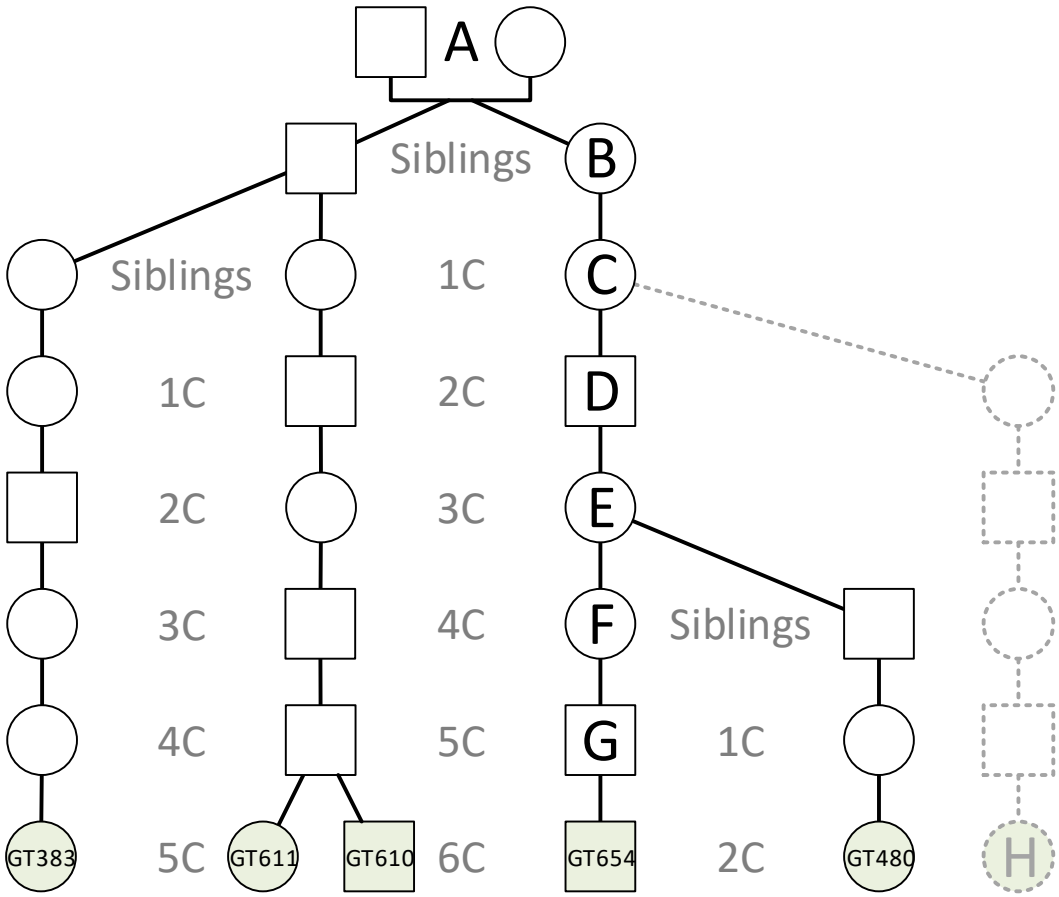


Figure 28: Representation of the lineages in a triangulated group for a Chr1 segment (177M to 191M).[238,239,240,241,242,243] *See also* Appendix D.

GT654 has a second cousin GT480 that shares the match in consideration. Considered in isolation, this match and the associated lineage that relates GT654 to GT480 qualifies as an instance of ε. This intermediate instance of ε gives *evidence* that GT654 and GT480 received this matching segment IBD from their great-grandfather (labeled *E*). What if another independent cousin (*H*) existed that shared this match with a lineage that related GT654 and *H* via their ancestor *C*—an instance of ε giving *evidence* that GT654 and *H* received this matching segment IBD? These intermediate instances of ε would give considerable strength to the supposition that GT654 received the matching segment IBD from the triangulated group's common ancestor *A*. What if additional instances of ε existed for B, D, F and G? It would be like knowing the provenance of that matching segment through every generation from *A* to GT654. Jim Bartlett likened this concept to "[walking] the segment back" through the generations to the common ancestor.[244]

Does a lack of intermediate common ancestor matches invalidate the *hypothesis*

being tested? No. But they are desirable. Without them, there may be more risk in declaring a *conclusion*, but no more risk than declaring a *conclusion* based on the *evidence* that initiated testing.

Can intermediate common ancestors be sought out? Yes…and no. Yes, because it is sometimes possible to identify that a match may be in common with the person of interest because they may have a particular intermediate common ancestor in common with the person of interest. Given a list of all your n^{th} cousins, the probability of picking the cousin(s) that share the matching segment is small; but if the person of interest has a large number n^{th} cousins contributing genotypes, the probability of finding a match increases.[245] In other words, the probability of finding one by picking one is small, whereas the probability of finding one by comparing one's genotype to a whole database of genotypes is much greater.

Close Relative Matching

Close relatives that are related to the common ancestor—siblings, aunts, uncles, close cousins, etc.—can give *evidence* that a matching segment was received IBD.

In Figure 27, consider the family group identified by GT177, GT625 and GT381. The fact that GT381 has a sibling and an aunt that share the match in this group is *evidence* supporting the claim that she received her copy of the matching segment by descent from her more distant ancestors.

Conversely, GT136 has four relations that descend from his paternal grandfather—GT357, GT787, GT502 and GT241— that do not share the match being considered in the Figure 27 triangulated group.[246] Additionally, searching for a common ancestor among his paternal grandfather's ancestors has not produced any ancestors in common with the other members of the group. This becomes *evidence* that GT136 did not receive this matching segment IBD from his paternal grandfather.

Testing by matching with close relatives is just a special case of the *intermediate common ancestor* test. It is, perhaps, not natural to consider them as such because, often, no thought is given to "proving" the genetic relationship because it is "known" as such.

Match Stability

When considering generational matches, the stability of the match needs to be considered. Stability has to do with whether the matches conform to biological principles as it passes from generation to generation.

Consider the match shared with GT208 as it passes through four generations—from GT118 to her son GT163 to his son GT999 to his son GT186:

Kit #	Chr	Start Location	End Location	cM	SNPs
GT118 with GT208	1	63,175,608	76,807,359	12.1	3,132
GT163 with GT208	1	63,496,685	74,958,148	10.0	2,553
GT999 with GT208	1	63,336,657	74,958,148	10.2	2,609
GT186 with GT208	1	63,592,864	77,595,616	12.4	3,133

Figure 29: Unstable generational match with GT208.[247]

If the matching segment is IBD, biology would dictate that GT118 has the longest matching segment with GT208, and that successive generations are the same or smaller. Yet, the longest matching segment is with the most recent generation, and match size increases as it is transmitted from the 2nd generation to the 3rd and 4th generations—biological impossibilities. This match is apparently not stable.

Alternatively, it could be that the matching segment considered in Figure 29 is partially IBD and partially IBC. Perhaps the region between positions 63,592,864 and 74,958,148 is mostly (or completely) IBD, while the regions between positions 63,175,608 and 63,592,864 and between positions 74,958,148 and 77,595,616 are IBC. This may, in fact, be a more likely explanation for the example given in Figure 29. Perhaps additional questions could be asked. Perhaps other tests will assuage (or accentuate) the concerns.

Now consider the same generational matching with GT293:

Kit #	Chr	Start Location	End Location	cM	SNPs
GT118 with GT293	12	90,999,825	100,870,206	12.8	2,690
GT163 with GT293	12	90,999,825	100,870,206	12.8	2,691
GT999 with GT293	12	90,999,825	100,870,206	12.8	2,690
GT186 with GT293	12	90,999,825	100,870,206	12.8	2,668

Figure 30: Stable generational matching with GT293.[248]

With GT293, the matching segments are almost identical across all four generations. The only differences in the comparisons are in the number of identical SNPs in the matching sequence. This matching segment seems likely to be mostly or entirely IBD.

CHROMOSOME MAP CORRELATION

A chromosome map associates matching segments of atDNA with specific ancestors. If the chromosome in consideration has been mapped for the person of interest, the matching segment in consideration must fit within the map without conflict. The matching segment cannot inappropriately span known crossover locations. The common ancestor must fit as a relation of the known ancestors already associated with that location on the chromosome. If these conditions are not true, there is a conflict that must be resolved before a *conclusion* can be declared.

No one mapping technique will fully populate a given chromosome map. This means that chromosome maps are created over time and are a compilation of many *conclusions* accumulated over time. It follows that not every *hypothesis* can be tested against a map. But every *conclusion* could be added to a map and used in future evaluations.

In cases where the genotypes of three or more siblings are available, it is possible to jumpstart the creation of a chromosome map that shows matching segments (HIRs) received from their grandparents without accumulating a set of triangulated *conclusions* to do so. Kathy Johnston describes this method.[249] The process involves comparing the sibling sequence data to identify crossover locations, assigning crossovers to specific siblings, and then using logical inferences regarding FIRs, HIRs, and regions with no matching to work out HIRs received from each grandparent for each sibling. Figure 31 shows the Chr1 comparisons for four siblings and the maps that resulted. Note there are still portions of the map which could not be assigned. Also note that without knowing at least two *conclusions* (one about a paternal ancestor, and one about a maternal ancestor) that fit into the map, it is impossible to know which color belongs to which grandparent.

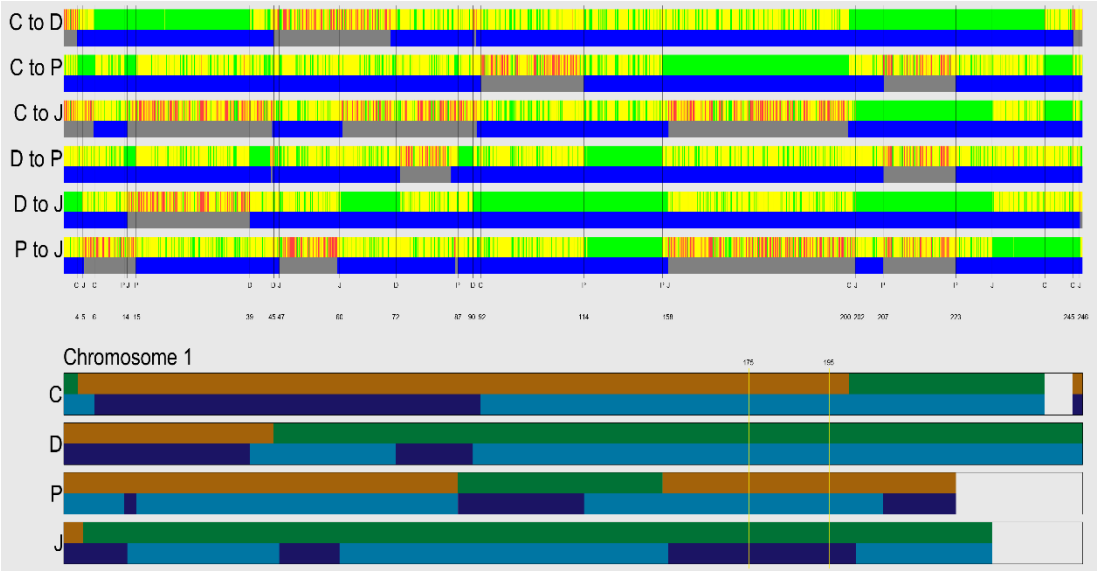


Figure 31: Chromosome 1 comparisons among four siblings (six bands at the top) and resulting maps showing half-identical regions inherited from grandparents (four maps at the bottom).[250,251,252]

The segment shared by the triangulated group represented in Figure 28 represents a paternal ancestor of GT611. Her Chr1 map is labeled *C* in Figure 31. It turns out her brother GT610 (labeled *P*) also matches this segment. With this information, it can be inferred that the top lines in these maps represent the chromosomes inherited from their father, and that the brown bands represent their paternal grandfather and the green bands their paternal grandmother.

Now consider the shared segment from Figure 28 in the context of the maps. The segment's position falls between the yellow lines (overlaid onto the maps). As such, the segment does not straddle any crossover locations. Only two of the siblings matched the segment, and the map predicted the second match (meaning that GT610 had not been compared to the triangulated group until after the map had been created and the maternal and paternal chromosomes had already been identified). The segment shared in Figure 28 fits without conflict into GT611's Chr1 grandparent map.

Excessive Matching

Some regions of the genome tend to match other genomes at a higher-than-expected rate—they are prone to excessive matching.[253] These regions have lots of identical matches. These matches are not identical because of recent shared ancestry but are considered the result of demographic or historic factors. In the genetic genealogy community vernacular, these groupings of excessive matches

are termed *pile-ups*. The "Excess IBD sharing" section of the *Identical by descent* article on the ISOGG wiki gives several references to research about specific pile-up regions.[254]

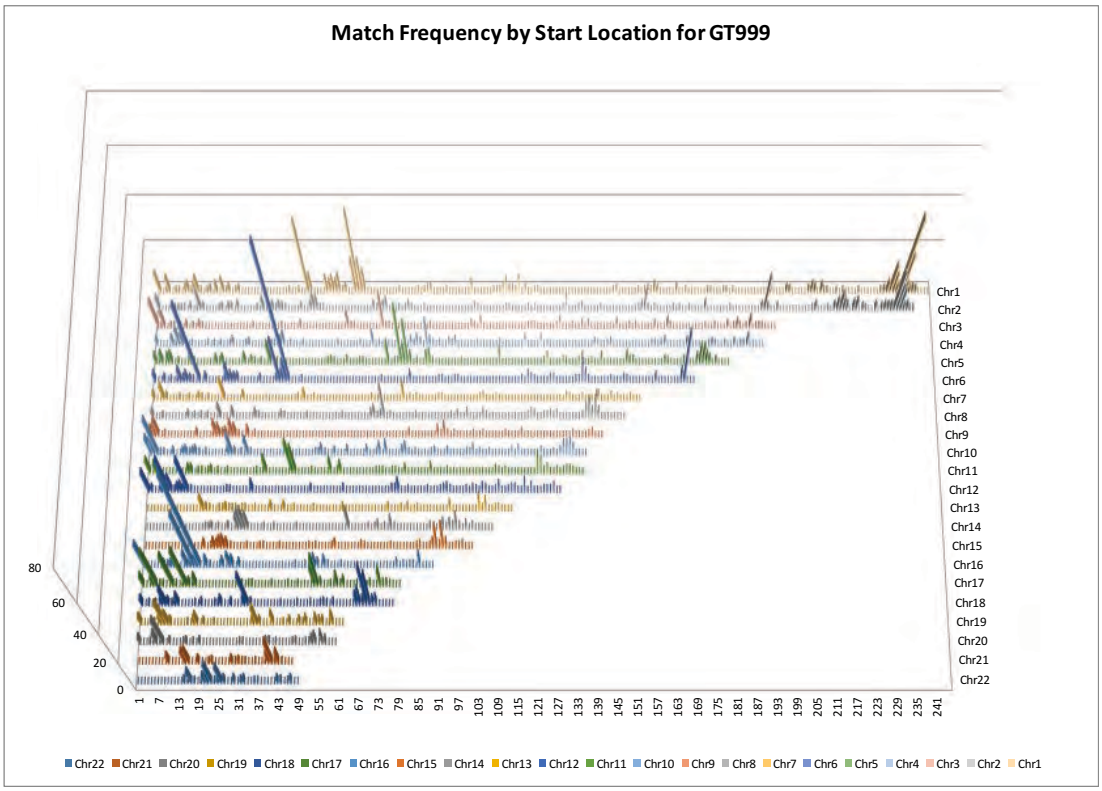


Figure 32: Plot of match frequency (1 Mbp tranches) to show potential pile-up regions for GT999.[255]

Figure 32 plots GT999's match frequency by chromosome and start location. There are several locations with potential pile-ups. Chr1, Chr2, Chr5, Chr6 and Chr16 (and others) all have small regions with significantly higher numbers of matches compared to the rest of the genotype, made evident by tall spikes in the number of matches at locations along those chromosomes.

If one finds there are a lot of matches for a particular location in the genome, can it be assumed to be excessively matching? There could be reasons to answer no. It may be that there are several family groups (clusters of related genotypes) that make the frequency higher than expected. Sometimes these clusters include duplicate genotypes. Each cluster needs to be evaluated separately.

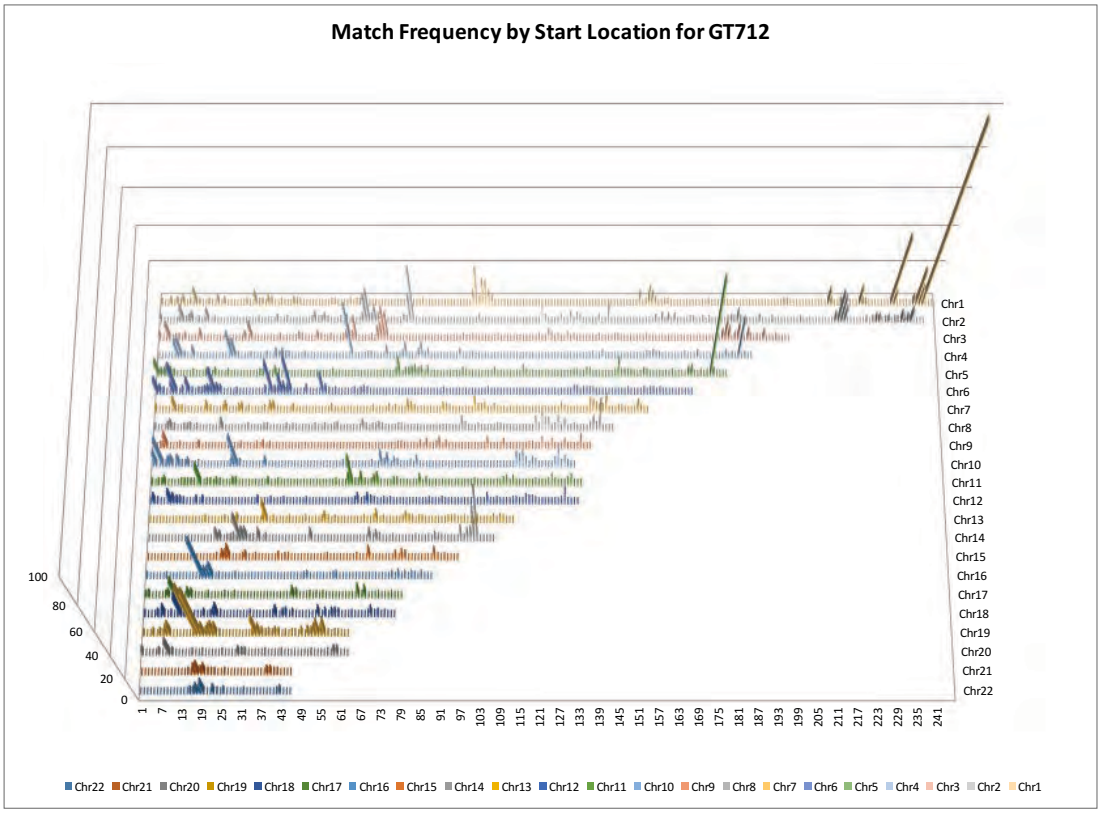


Figure 33: Plot of match frequency (1 Mbp tranches) to show potential pile-up regions for GT712.[256]

These pile-up regions are not necessarily consistent from genome to genome (note both the similarities and differences between Figure 32 and Figure 33), nor is there perfect agreement about what should be done about them. For example, AncestryDNA™ has added the so-called "Timber" algorithm to its processing to de-emphasize regions with excessive matching.[257] Some angst has been expressed about this addition, with many asserting that the algorithm eliminates useful matches.[258] Yet current limitations—both in genotyping and in the timeframes that genealogical records can viably cover—seem to prevent correlation or inference involving these matches. Dan Edwards asserts that incorporating these matches into a genealogical research process is futile—genealogically intractable.[259]

If a region seems to be excessively matched, one needs to consider whether such matches can even be considered IBD. Sharon and Brian Browning define IBD in terms of shared haplotype frequency.[260] If sharing exists between two haplotypes but is not likely with any other haplotypes, it is more likely IBD. The frequency that other haplotypes match a given region can indicate a likelihood that the region was received IBD. Excessive (i.e., high frequency) matching dilutes the supposition that region was received IBD.

Given a *hypothesis*, how many independent instances of ε ought to be present in the *hypothesis*? Ideally, each match capable of giving an independent answer to the triangulation question should have an instance of ε in the *hypothesis*. If the *hypothesis* should have four instances of ε and only has three, the *hypothesis* likely remains tenable. If the *hypothesis* should have sixty-five instances of ε and only has three, there is very likely reason for caution. If a *conclusion* is declared in this latter case, is there a basis to expect that further instances of ε will not alter that *conclusion*? It seems likely to be an untenable position; the author's own foray into researching such a match (e.g., a triangulated group needing 25 instances of ε) ran afoul almost immediately with multiple, conflicting common ancestors.[261,262,263,264]

SOLUTION PREDICTS RELATIONSHIPS

Bartlett claims that match databases have doubled every fourteen months.[265] This means that new matches are continuously being added to match lists. From time to time, new matches will associate with triangulated groups that have previously *concluded* common ancestors. These matches are an opportunity to add additional instances of ε in support of the existing *conclusion*. If the existing *conclusion* is sound, one would expect that the existing *conclusion* would predict the ancestor (or lineage) that would be found in common with the new match.

Finding the predicted ancestor (or lineage) in the compiled genealogy of the new match does not mean that the new instance of ε does not need testing; all applicable tests of analysis and correlation should still be applied. If the predicted ancestor (or lineage) is still part of the resulting *conclusion* when testing is complete, it also stands to reason that the pre-existing *conclusion* was successful in predicting the new match's relationship to the triangulated group. This success is additional evidence that the matching segment being considered was received IBD.

COMMON ANCESTOR UNIQUENESS

Concluding that a particular matching segment of atDNA was received IBD from an ancestor is difficult if there is more than one viable candidate ancestor. Ideally, the compiled genealogies being searched would be completely known to the generation of the candidate ancestor, and the only ancestor common to these genealogies would be the candidate ancestor. The practicalities of reality merit consideration.

It is not uncommon for two pedigrees to have more than one candidate in common. If this is the case, additional instances of $\mathcal{E}$ are needed to narrow the field. Generally, the more independent instances of $\mathcal{E}$ that are present in the hypothesis, the more likely there will be only one candidate in common.

Sometimes, more instances of $\mathcal{E}$ result in conflicting candidates for a common ancestor. Is the common ancestor from an older generation? Perhaps there is an issue in the genealogies themselves—e.g., the genealogies are not complete, the genealogies are not accurate, or an unidentified NPE exists in the genealogies. Additional matches may be able to help sort out such conflicts.

Another reason for conflict is that the matching segment is not IBD. Should the match be categorized as excessively matching—IBS due to demographic or historical reasons? Is the match IBC?

It is common to encounter incomplete genealogies when searching for common ancestors. Often, a common ancestor cannot be identified in such cases—an incomplete instance of $\mathcal{E}$ (an m without an a). Sometimes, like in the case of an adoptee, parentage is unknown, and a complete genealogy cannot exist until the parentage problem is solved. In some cases, the individual may withhold such information. In cases where the information is withheld, it may be possible to compile a genealogy as a surrogate of the missing information—though the privacy and protection of this information must be considered, and it may raise ethical concerns. An incomplete genealogy is probably not a reason to “fail” this test. It does marginalize the match’s contribution to the hypothesis.

It is possible that a common ancestor will emerge even though one or more of the genealogies are incomplete. Incomplete genealogies mean incomplete data is used to determine the MRCA—leaving any identified MRCA open to question. It should be very rare, however, to have two separate instances of $\mathcal{E}$ (each with independent instances of m and independent lineages to a). Therefore, having a number of independent instances of $\mathcal{E}$ in the hypothesis reduces the likelihood that an incomplete tree will result in an error due to incomplete genealogy.

LOOKING FORWARD

In considering hypothesis testing, this text has considered a number of the most common *tests of analysis* and *tests of correlation* to be used in validating and characterizing *conclusions* supported by atDNA *evidence*. Is this list exhaustive? No. Will additional tests be identified? Absolutely. This is an emerging practice in the genealogical community. Genetics and genomics are evolving fields of study. Further refinement of the tests and heuristics detailed herein are needed and expected; for example, it would be helpful to re-present the information reported by Speed and Balding in terms of genetic distance rather than physical distance so that it is more readily interpreted within the context that genetic genealogists use. Next-generation sequencing is sure to enable further innovation. This text presents a framework and methodology for identifying, evaluating, and presenting the strengths and weaknesses of a *conclusion* involving atDNA *evidence*.

CONCLUSION ACCEPTING

The desired outcome of *hypothesis* testing is a *conclusion*. If the *hypothesis* holds up to scrutiny—if it passes testing and all conflicts can be resolved—it becomes a *conclusion*. Testing safeguards the researcher from erroneous *conclusions*, and spotlights the strengths and/or weaknesses of the resulting *conclusion*.

For some tests, a failure will invalidate the *hypothesis*. For example, a *hypothesis* that does not pass a generational matching test cannot be accepted as a *conclusion*. Other tests only cast/remove doubt. An incomplete genealogy introduces doubt but does not invalidate. Doubt can be alleviated on the strength of other testing. If doubts mount, a *conclusion* may become untenable.

Testing may spotlight conflicts. Conflicts must be resolved or no *conclusion* can be made. For example, if a matching segment inappropriately spans a crossover location in a chromosome map, a *conclusion* is premature. It may be that the map itself is flawed and that fixing the map will resolve the conflict; if so, a *conclusion* may be possible; if not, the *hypothesis* must be discarded.

Extraordinary *conclusions* require extraordinary *evidence*.[266] In other words, the burden of proof is very high for *conclusions* that push the edges of possibility—*conclusions* that are unusual and/or improbable. At the same time, *conclusions*

should not be discarded because all tests could not be applied, or because doubt remains. As much as possible, tests should be applied, and the outcomes discussed and weighed. As an exhaustive search and only two items of evidence in traditional genealogy can result in a conclusion, it follows that two scrutinized instances of ε with no conflicts and no invalidating tests is sufficient to *conclude*—if explained.

Appendix F details two *conclusions* based on the triangulation of a segment shared by GT999 on Chr1 with two distant cousins. The author *concludes* that members of the triangulated group received the specified matching segment IBD from a particular common ancestor. He also *concludes* about the identity a cousin's 5th great-grandparents, breaking down a "brick wall." It is not a perfect example of triangulation. Yet, a reasonably exhaustive search has been executed, available evidence has been integrated, and no conflicts remain. One might wish for (and eventually find) additional records (atDNA matches, or otherwise) that will bolster (or debunk) the *conclusion*, but there is a *conclusion* that is viable now—a *conclusion* the author accepts now. The information bears up under scrutiny and leaves the author confident that the *conclusion* will stand as additional *information* becomes available.

PROOF EXPLAINED

Proof exists only if the *conclusion* has been recorded for others to examine. This documentation should include the matches (m) involved, the lineages relating the individuals to the common ancestor (a), and details of the testing (t) applied. In presenting results, areas of doubt should be highlighted and reasons for acceptance explained. Conflicts encountered and their resolutions must be explained. Appendices B, C, D and F document *conclusions* (or not)—proof explained.

Standards for presenting this documentation are not well developed. An area of particular interest is how to reference genotype information. The Genetic Genealogy Standards Committee promises to provide guidance on this issue.[267]

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SUMMARY

This text presents a methodology for identifying, evaluating, and presenting a *conclusion* involving atDNA *evidence*. It places this methodology firmly within the framework of the genealogical research process—question asking, information gathering, hypothesis testing, conclusion accepting, and proof explained—rooting genetic genealogical practice within the processes that lead to genealogical proof.

This text focuses particularly on *hypothesis* testing as the means of evaluating the strengths and weaknesses of a *conclusion* based on atDNA *evidence*, including important heuristics that define the capabilities and limits that accompany the use of the atDNA record. Testing scrutinizes and refines *hypotheses*, ultimately making it possible to confidently use atDNA to confirm genealogical relationships.

Not all possible topics have been discussed. Instead, this text has focused on core topics used regularly in genetic genealogy (e.g., triangulation).

Genetic genealogical practice continues to evolve. New technologies and more detailed information (e.g., next-generation sequence data) are already working their way toward broad availability. Scientists continue to explain behaviors and refine heuristics that both enable and limit the use of atDNA for genealogical purposes. Even so, the methodology presented herein, and the framework within which it resides, will continue to be relevant as the genetic genealogical community of practice transforms to take advantage of these developments.

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APPENDIX A: FINDING AN ADOPTEE'S BIOLOGICAL FAMILY

An adoptee—identified here as an8181—decided to pursue the identity of her biological parents. Her adoptive parents had been provided with a document from the adoption agency that contained information about her birth parents.[268] This information included various genealogical clues that could be helpful in vetting candidate parents including: information about her own birth, her birth parent's ages, marital status at the time of the adoption, the marital status of each of her biological grandparents, physical descriptions of her biological parents and grandparents, occupational information about her biological parents and grandparents.

an8181 submitted a sample for genotyping to AncestryDNA™. When she received her genotyping results at the end of December 2015, her closest match (designated C.S.) was estimated to be a 3rd - 4th cousin—sharing 153 cM across 7 segments.[269] This match had limited access to their published genealogy, and they were unresponsive to requests for access.

On January 23rd, 2016, an8181's match list was revisited. A new, closer match (designated M.G.) had been added to her match list. This match was given as a 1st - 2nd cousin—sharing 454 cM across 22 segments. Using the distributions published from Bettinger's *Shared cM Project* (Figure 10), the 454 cM of shared atDNA falls in the peak of the distribution of Degree 4 relationships.

The administrator of M.G.'s genotype had associated his genotype with a publicly accessible genealogy. Browsing close relatives of M.G., an8181's husband was able to find a candidate family with a son that could have been a sibling to one of an8181's parents. The administrator of M.G.'s genotype was contacted, and the administrator was able to confirm that M.G.'s half first cousin had given up a child for adoption on the day an8181 was born. M.G.'s half first cousin was an8181's biological father.

Genealogical research about an8181's biological father was able to reveal an8181's mother.[270] Many of the details given in the document from the adoption agency about an8181's biological parents could be identified in the genealogical records found about her parents and their families.

an8181's search for her biological parents turned out to be a relatively straight forward process. It is typical for an adoptee to have to wait for the right match, but often the wait is much longer than occurred here. There are strategies for actively seeking better matches, but she did not have to employ any of these. Additional triangulation is often required, but the nature of the match combined with the details available from the adoption agency and from members of the biological family rendered this level of proof unnecessary.

For more information on methodology around seeking biological parents as an adoptee, the genetic genealogical community recommends starting with the resources published at DNAadoption.com.[271,272,273]

APPENDIX B: TRIANGULATION FOR GT999 ON CHR1 FROM 159M TO 167M

The following triangulated group was identified with the phased maternal kit for GT999 on Chr1:

KIT #	START POS	END POS	cM	SNPs
GT611	72,017	247,169,190	281.5	54,743
GT861	99,175,785	194,953,541	73.8	16,350
GT709	111,732,674	194,928,236	60.8	13,486
GT610	114,435,751	200,890,245	64.9	13,883
GT901	152,790,212	194,928,236	45.5	9,809
GT124	155,348,641	166,650,792	20.3	3,348
GT732	158,328,558	166,650,792	14.8	2,522
GT831	158,879,805	177,283,021	22.3	4,848

Figure 34: Triangulated group with GT999 on Chr1 for a matching segment from 159M to 167M.[274]

All the individuals in the group share a common ancestor as shown in Figure 35.

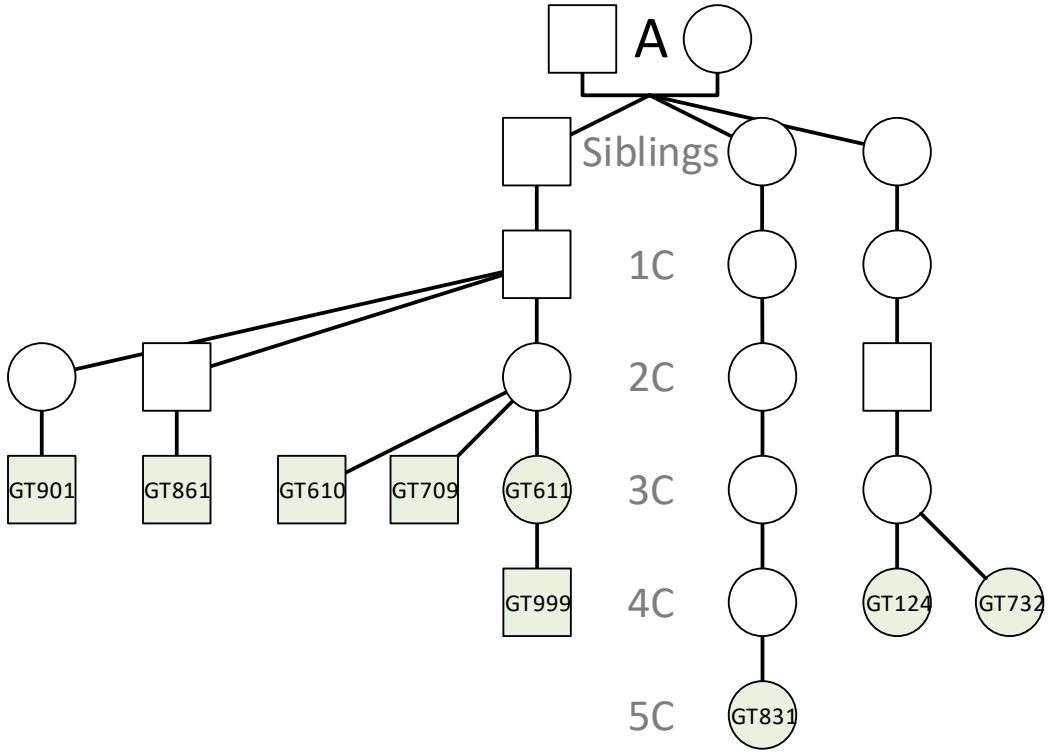

Figure 35: Representation of the lineages connecting members of a triangulated group for Chr1 (159M to 167M).[275,276,277] *See also Figure 13, Figure 14, and Figure 18.*

This triangulated group has a large family group (to be called Group 1 in this appendix) composed of GT999, GT611, GT709, GT610, GT861, and GT901. There is another family group (to be called Group 2 in this appendix) composed of two sisters: GT124 and GT732. A third individual GT831 is also a member of the triangulated group.

There is another individual—GT681—who matches the group, but whose common ancestor with the group has not been identified. Therefore, GT681 has not been represented in Figure 34 or Figure 35; GT681 is present in Figure 37. Several candidate common ancestors with GT999 have been identified that are ninth great-grandparent generation for GT999, suggesting that GT681 has a much more distant relationship with members of this group. This group may be candidate for an intermediate MRCA if another match is found that will triangulate with GT681.

Using the matches listed in Figure 34, the segment shared by this group is positioned from 158,879,805 (159M) to 166,650,792 (167M) with a physical length of 7,770,987 bps (7.8 Mbp) and a genetic size of 13.2096 cM.[278]

The *hypothesis* is that Group 1, Group 2 and GT831 all received the shared segment IBD from the common ancestor identified for the triangulated group. Before accepting the *hypothesis*, factors affecting the likelihood of reliability, errors or misinterpretations must be considered; also, the items of *evidence* should agree with each other and with the principles of inheritance. The testing that follows will help determine whether the *hypothesis* can be accepted as a *conclusion*.

TESTS OF ANALYSIS

MATCHING SEGMENT SIZE

The size of the individual matches shared with GT999 are all greater than 20 cM except for GT732 at 14.8 cM. Based on Figure 19, the probability any of these matches will not survive phasing is negligible. As the comparisons were all made with GT999's phased maternal genotype, it is very unlikely that these matches are IBC. All are well above the minimum 5.0 cM threshold suggested for phased matches, the threshold necessary to achieve an acceptable level of false positives.[279] The matches are certainly all good candidates to be IBD according to these tests.

The physical length of the matches that GT999 shares with Group 2 and GT831 range from 8,322,234 bps (8.3 Mbp) to 18,403,216 bps (18.4 Mbp). Considering these matches in the context of Speed and Balding's work (Figure 23), both relationship types are considered to have *G*=5. The smallest match is in the tranche from 5-10 Mbp, and the largest is in the tranche from 10-20 Mbp. For 10-20 Mbp

tranche, there is more than a 10% chance that a match of the given size came from an ancestor with *G*=5 or closer. For the 5-10 Mbp tranche, the likelihood has fallen to roughly 5%. In all cases, the probability is not negligible; the matches are candidates to be IBD.

TOTAL SHARED IBD

The theoretical average sharing for fourth cousins (the relationship between GT999 in Group 1, and Group 2) is 13.28 cM.[280] For fourth cousins once removed (GT999 in Group 1 and GT831), it is 6.64 cM. In all cases, the match from each individual that is being evaluated in this triangulated group already exceed these values. This comparison is not meaningful.

	Threshold	
Comparison	**5 cM**	**4 cM**
GT999 (Group 1) → GT124 (Group 2)	12.9	12.9
	20.3	20.3
	10.0	4.8
		10.0
		4.0
		4.9
GT999 (Group 1) → GT732 (Group 2)	14.8	4.1
	8.4	14.8
	15.6	4.8
	5.0	8.4
		4.2
		15.6
		5.0
Total Shared w/ Group 2 (Avg)	43.5	56.9
GT999 (Group 1) → GT831	23.5	23.5
		4.1
Total Shared w/ GT831	23.5	27.6

Figure 36: GT999's total atDNA sharing with Group 2 (an average) and with GT831.[281]

GT999's total amount of sharing with members of Group 2 (calculated as an average per individual in the group) and with GT831 (given in Figure 36) was calculated using Janzen's method.[282] Both relationships are outside the relationships reported in Figure 21 (from the *Shared cM Project*). The shape and bounds of the distributions from the *Shared cM Project* for these relationship types are difficult to discern in Figure 10, but the associated distributions seem to cover

the calculated totals for both relationships being considered here. The totals are high, presumably because they include some amount of IBC (false) sharing or because the relatives share more atDNA that would be expected by chance. The difference between the amounts shared matches the expected difference—the fourth cousins sharing roughly twice as much atDNA than the fourth cousin once removed.

The total amount of sharing is consistent with the relationships specified in the *hypothesis*.

TESTS OF CORRELATION

INDEPENDENCE

Value shown is cM total of matching segments over minimum threshold.

Kit	name	PGT999M1	GT611	GT861	GT709	GT610	GT901	GT124	GT681	GT732	GT831
PGT999M1			3587.1	568.2	1747.7	1791.6	465.9	41.3	12.8	37.2	22.3
GT611		3587.1		930.3	2711.0	2732.0	691.3	63.3	12.8	39.2	27.3
GT861		568.2	930.3		851.2	813.1	963.1	33.7	18.7	49.3	23.4
GT709		1747.7	2711.0	851.2		2697.4	815.6	36.9	12.8	42.1	48.2
GT610		1791.6	2732.0	813.1	2697.4		852.3	48.4	12.8	25.1	27.6
GT901		465.9	691.3	963.1	815.6	852.3		34.8	12.8	20.9	54.8
GT124		41.3	63.3	33.7	36.9	48.4	34.8		12.8	2274.7	23.8
GT681		12.8	12.8	18.7	12.8	12.8	12.8	12.8		10.4	13.5
GT732		37.2	39.2	49.3	42.1	25.1	20.9	2274.7	10.4		14.0
GT831		22.3	27.3	23.4	48.2	27.6	54.8	23.8	13.5	14.0	

Figure 37: Total amount of atDNA sharing between members of the group that triangulates on a matching Chr1 segment from 159M to 167M.[283]

The report shown in Figure 37 helps evaluate the independence within the triangulated group. There are two family groups (Group 1 includes the first six individuals; Group 2 includes the seventh and ninth individuals), and two individuals (GT831 and GT732). Only three independent answers to the triangulation question are possible. So far, only two answers have been identified.

A common ancestor has not been identified for GT681. Note that GT681's matches with the group are generally smaller (her row and column in the table is a much darker red). As already mentioned, the candidate common ancestors that have been identified thus far for GT681 are much more distant than the ancestor identified for this group. The smaller size of her matches with the group seems to be indicative of this more distant relationship.

EXCESSIVE MATCHING

With only three possible instances of ε in the database thus far, excessive matching is not a worry for this triangulated group.

CHROMOSOME MAP CORRELATION

Figure 31 is a Chr1 map for four siblings, three of which are members of this triangulated group: GT611, GT709 and GT610. The segment under consideration lies somewhere to the left of the yellow line marking 175M. The next numeric reference point along the map marks 158M which corresponds to the crossover from green to brown on the person labeled *P*. The segment in question lies somewhere between 158M and 175M, but on the regions colored light blue. There are no crossover events within this range. Within this range, there is only one possible configuration of siblings that could match the light-blue region—and the expected three are the ones with matching segments. The chromosome map agrees with the IBD claim.

INTERMEDIATE COMMON ANCESTORS

GT999 has intermediate common ancestors that share this segment at his grandmother (two uncles: GT709 and GT610), and at his great-grandfather (two of his mother's cousins: GT861 and GT901). The intermediate common ancestors are strong *evidence* that this segment was received IBD.

PHASED MATCHING

The phased maternal genotype of GT999 was used to identify members of this triangulated group, essentially eliminating the risk that IBC matches were included in the group—a boost to the IBD claim.

GENERATIONAL MATCHING

Both GT999 and his mother GT611 match with Group 2 and with GT831. The expected generational match is present—*evidence* that the matching segment was received IBD.

GT999 also has a son GT186, but the son does not match any of the members of Group 2, or GT831.[284] This does not cast doubt on our hypothesis because it is plausible that the son did not receive this segment.

Close Relative Matching

GT999 has several close relatives that match the individuals in Group 2, and that match GT831: two uncles (GT709 and GT610), and two of his mother's cousins (GT861 and GT901). These matches strengthen the claim that GT999 received this segment IBD.

The individuals in Group 2 (GT124 and GT732) are siblings and they both match this shared segment—*evidence* that they received this segment IBD.

Match Stability

Match stability can only be considered in the context of GT999 and his mother GT611. Comparing mother and son to Group 2 and to GT831, the match is stable—genetically correct; i.e., the newest generation received a segment that was the same size as or smaller than the older generation, and the newer generation's segment was bounded at or within the boundaries of the older generation—as it was passed from mother (GT611) to son (GT999).[285,286]

A lack of stability would have cast doubt on an IBD claim. There is no lack of stability in this case.

ε (1 of 2)

This instance of ε is between Group 1 and Group 2.

COMMON ANCESTOR UNIQUENESS

		GT999		GT124 & GT732	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	28	88%
Total	63	63	100%	59	94%

Figure 38: Compiled genealogy completeness evaluation for GT999 and Group 2 (GT124 & GT732).[287,288]

The compiled genealogy representing Group 1 is complete to the generation of the proposed common ancestor. The compiled genealogy representing Group 2 is only missing the identities of four ancestors, all of them missing in the last generation. There is little risk that a different MRCA will be identified between these two family groups.

CONCLUSION FOR $\mathcal{E}$ (1 OF 2)

All the testing for match (m) strongly supports that this shared segment is consistent with IBD segments, and that it was received IBD. The search for a common ancestor (a) between these two groups has considered all but four ancestors from Group 2—a very small residual risk.

$\mathcal{E}$ (2 OF 2)

This instance of $\mathcal{E}$ is between Group 1 and GT831.

COMMON ANCESTOR UNIQUENESS

		GT999		GT831	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	28	88%
7	64			32	50%
Total	127	63	100%	91	72%

Figure 39: Compiled genealogy completeness evaluation for GT999 and GT831.

The compiled genealogy representing Group 1 is complete to the generation of the proposed common ancestor. The compiled genealogy representing GT831 is missing half of the identities of in the last generation and four identities in the preceding generation. There are definitely gaps in this search.

CONCLUSION FOR $\mathcal{E}$ (2 OF 2)

All of the testing for match (m) strongly supports that this shared segment is consistent with IBD segments, and that it was received IBD. The search for a

common ancestor (a) between these two groups has gaps. Some mitigation is had in the fact that there are two instances of $\mathcal{E}$ in this hypothesis (each with independent instances of m and independent lineages to a), which is rare in and of itself; this tends to offset the risk that lingers due to the lack of coverage in the MRCA search.

CONCLUSION ACCEPTING

There is every reason to believe that the specified segment was received IBD by members of this triangulated group. The proposed common ancestor is unique with little residual risk for one instance of $\mathcal{E}$, but the genealogy of GT831 is incomplete and exposes the *hypothesis* to additional risk. This risk is mitigated by the fact that there are two independent instances of $\mathcal{E}$ in the hypothesis. It is therefore *concluded* that one person in the ancestral couple identified as common to this triangulated group must have contributed this shared segment (Chr1 from 159M to 167M) to all of the members of this triangulated group.

APPENDIX C: TRIANGULATION FOR GT999 ON CHR4 FROM 187M TO 191M

The following triangulated group was identified with the phased paternal genotype for GT999 on Chr4:

KIT #	START POS	END POS	cM	SNPs
GT163	61,566	191,117,403	214.4	37,262
GT625	175,696,978	190,696,128	36.0	4,166
GT177	175,740,345	190,696,128	35.9	4,128
GT381	184,520,405	190,922,297	19.1	1,819
GT136	185,592,550	190,568,137	14.9	1,426
GT978	186,651,620	191,117,403	12.0	1,207
GT789	186,651,620	191,117,403	12.0	1,207
GT606	186,651,620	191,117,403	12.0	1,212
GT491	186,651,620	191,117,403	12.0	1,199

Figure 40: Triangulated group with GT999 on Chr4 for a matching segment from 187M to 191M.[289]

With the exception of GT136 (whose connection with the group is currently undetermined), all share a common ancestor as shown in Figure 27.

The match is positioned from 186,651,620 (187M) to 190,568,137 (191M). It has a physical length of 3,916,517 bps (3.9 Mbp) and a genetic size of 11.0543 cM.[290]

TESTS OF ANALYSIS

MATCHING SEGMENT SIZE

Based on Figure 19, the probability that any of the matches is IBC is less than 5%. As the comparisons were made with a phased genotype, it is already known that these matches are not IBC. All are well above the minimum 5.0 cM threshold suggested for phased matches, the threshold necessary to achieve an acceptable level of false positives.[291] The matches are certainly good candidates to be IBD.

TOTAL SHARED IBD

For all of the genotypes in the group (except GT163—father of GT999), there is only one shared segment with GT999. A comparison with a theoretical average for total sharing is rendered meaningless because the closest relationship with GT999 (that is not his father) is expected to share just 3.32 cM which is below the segment reliability thresholds already mentioned, and it is outside the relationships reported by the Shared cM Project.[292,293]

Instead, whether the match size is IBD-consistent is considered within context of Speed and Balding's work.[294] A 3.9 Mbp shared region falls in the tranche between 2 and 5 Mbp (Figure 23)—a small, shared region by IBD-consistent standards. Roughly 10% of shared segments this size belong to ancestors in the closest 10 generations. While it is considered rare to share such a small segment IBD from a G=9 ancestor, it is not an impossible occurrence. The shared segment's genetic size (11.1 cM) suggests that an IBD segment in this region will generally be quite small physically.

TESTS OF CORRELATION

INDEPENDENCE

The report shown in Figure 26 helps evaluate independence within the group.[295] It shows that there are three family groups and one individual. GT999 is in a group with his father (GT163). There can be three independent answers to the triangulation question between GT999 and the other independent groups/individuals.

EXCESSIVE MATCHING

Based on in Figure 26, the number of independent matches at this location is only three; this does not seem to be an excessive amount of matching.

CHROMOSOME MAP CORRELATION

GT999 does not have an established chromosome map for any other generations on this chromosome, so no correlation with a chromosome map is possible.

INTERMEDIATE COMMON ANCESTORS

At this time, there are no known intermediate common ancestors that correlate with this matching segment.

PHASED MATCHING

The matching was done with the phased paternal genotype of the POI, essentially eliminating the risk that IBC matches were included in the group—giving a boost to the IBD claim.

ε (1 OF 3)

The first instance of *ε* is between the group headed by GT163 (with son GT999) and the group headed by GT978 (including child GT789, and grandchildren GT606 and GT491). All of the members of this group match each other in the expected ways (meaning all of the parents share the expected amount with their children, and the siblings share an expected amount with their sibling).

GENERATIONAL MATCHING

Generationally, the phased matching already guarantees a generational match between GT999 and GT163. GT999 does have a son (GT186) who could have been included in the group; his phased genotype matches the GT978-group in the same stable manner as GT999's genotype.[296] There is no additional generational matching that can be tried between these two family groups. This testing supports the IBD claim.

CLOSE RELATIVE MATCHING

This group did not match any genotypes of other identified close relatives for GT999 (i.e., an aunt, a second cousin and twice-related cousin—3C /3C1R).[297] A match could have strengthened the IBD claim; a lack of match does not cast any appreciable doubt.

MATCH STABILITY

The matches of the phased genotype of GT999 with the genotypes of the GT978-group are very stable—all of them starting and ending at the same position—giving credibility to the *hypothesis* that these matches are IBD-consistent.

COMMON ANCESTOR UNIQUENESS

		GT999		GT978	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	30	94%
7	64	64	100%	60	94%
8	128	126	98%	109	85%
Total	255	253	99%	230	90%

Figure 41: Compiled genealogy completeness evaluation for GT999 and GT978.[298,299]

For eight generations, GT999 and GT978 both have great coverage in their compiled genealogies—GT999 has identified 99% of his ancestors, and GT978 has identified 90% of his ancestors. GT978 does not reference much in the way of original records, but other compiled genealogies for his lineage to the common ancestor do.[300] No other factors were identified that might affect the reliability of the information contributing to the lineages that connect these two cousins. Some risk remains but would be considered limited relative to many other cases.

CONCLUSION FOR $\mathcal{E}$ (1 OF 3)

All the tests of analysis and correlation for the match (m) for this first instance of $\mathcal{E}$ support the IBD claim—none presenting any conflicts, and only the map correlation test being untried. The biggest risk for the match (m) seems to be the physical length of the matching segment—it being relatively smaller than perhaps expected. The risks associated with the compiled genealogies are also relatively small. The first instance of $\mathcal{E}$ seems to bear up well to scrutiny.

$\mathcal{E}$ (2 OF 3)

The second instance of $\mathcal{E}$ is between the group containing GT999 and GT1653 and the group containing GT381, GT625 and GT177. GT381 and GT625 are half-siblings, and GT177 is their half-aunt.[301] GT381 matches her half-sibling and half-aunt at the high-end of their expected total sharing amounts—to be expected given there is no phasing to eliminate IBC matching—supporting the relationships claimed

within this family group.[302]

GENERATIONAL MATCHING

The phased matching already guarantees a generational match GT999 and GT163. GT999's son also matches the members of this family group.[303] There is no additional generational matching that can be tried between these two family groups. This testing supports the IBD claim.

CLOSE RELATIVE MATCHING

GT381 and GT625 did not match any of the genotypes of other close relatives (an aunt, a second cousin and twice-related cousin—3C /3C1R), but GT625 does match the second cousin (GT859) nearby (Chr4 from 176M to 184M).[304] This match strengthens the claim of relatedness, and in so doing, strengthens the claim that the segment under consideration was received IBD.

MATCH STABILITY

The test cannot be applied because the members of the group being tested are not lineally related.

COMMON ANCESTOR UNIQUENESS

		GT999		GT177	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	19	59%
7	64	64	100%	13	20%
8	128	126	98%	16	13%
9	256	218	85%		
Total	511	471	92%	79	31%

Figure 42: Compiled genealogy completeness evaluation for GT999 and GT177.[305,306]

While GT999's genealogy is fairly complete, GT177's genealogy is very incomplete. This does tend to cast doubt on the completeness of the MCRA search. Though

incomplete, the genealogy covering GT177's lineage to the proposed common ancestor does appear to be original research and does reference expected original records. No other factors were identified that might affect the reliability of the information contributing to the lineages that connect these two cousins.

CONCLUSION FOR ε (2 OF 3)

The second instance of ε's main flaw is the coverage of ancestors in the search for the MRCA. The fact that there are two instances of ε is rare in and of itself; this tends to offset the risk that lingers due to the lack of coverage in the MRCA search. The second instance of ε bears up to scrutiny, but there is a bit more risk that future research may introduce conflict.

ε (3 OF 3)

COMMON ANCESTOR UNIQUENESS

The third instance of ε is between the group containing the group headed by GT163 and GT136. Attempts to identify a common ancestor between GT136 and other members of this triangulated group have failed. The administrator of this genotype has only been able to share the genealogy of the paternal grandfather of GT136, leaving 75% of the ancestors of GT136 unidentified. Furthermore, the administrator is aware of four other genotypes that are related to GT136 and that all match GT136 and that are related to this paternal grandfather.[307] None of the members of this triangulated group match any of the other four genotypes—making it much less likely that members of this triangulated group have a relationship with this paternal grandfather.[308] The common ancestor is most likely among the unidentified ancestors of GT136.

GENERATIONAL MATCHING

GT136's match with the GT999 is not IBC because of the comparison with the GT999's phased genotype. This also guarantees a generational match with the GT999's father. The GT999's son is also matches with GT136 on this shared segment.[309] No additional generational matching is possible at this time.

CLOSE RELATIVE MATCHING

GT136 did not match any other close relatives.[310]

CONCLUSION FOR ε (3 OF 3)

GT136 remains a member of the triangulated group, and the match should be considered when evaluating excessive matching; but it could end up in conflict with the group, or as an intermediate MRCA, or even as an instance of ε for an older generation.

OTHER CONSIDERATIONS

One additional risk must be called out. The common ancestor in the first instance of ε is one generation newer that the common ancestor in the second instance of ε. This is not ideal, leaving room for doubt. It could be argued that the first instance of ε should be represented as an intermediate MRCA for the second instance of ε—leaving only one instance of ε in the triangulated group. But lacking other matches, and lacking any conflict, and given the relative independence of these two instances, it seems safe to accept both instances of ε for the purposes of triangulating this shared segment until such time as a proper instance of ε presents itself (at which time the first instance can become an intermediate MRCA), or until a conflict overturns the proposed solution.

CONCLUSION ACCEPTING

Given that two of the three instances of ε in this triangulated group pass testing with minimal residual risk, and given that the third instance of ε (without an identified common ancestor with the group) remains a candidate member of the group (insofar as it could be tested), it is therefore *concluded* that one person in the ancestral couple identified as common to this triangulated group must have contributed the Chr4 matching segment (187M to 191M) to the members of this triangulated group.

APPENDIX D: TRIANGULATION FOR GT611 ON CHR4 FROM 177M TO 191M

Figure 43 shows a set of genotypes that match GT611 on Chr1.

KIT #	CA	START POS	END POS	cM	SNPs
GT610	Y	113,623,623	207,063,451	75.0	15,948
GT116		162,639,701	191,953,018	26.9	2,771
GT341		162,926,661	191,859,017	26.2	2,688
GT654	Y	164,598,102	190,827,077	23.2	5,739
GT383	Y	175,935,925	190,828,095	11.3	1,283
GT480	Y	177,088,988	191,031,443	10.8	2,703
GT938		177,936,529	193,743,320	12.9	3,024
GT196		177,936,529	190,681,611	10.0	2,433
GT557		177,936,529	190,681,611	10.0	2,458
GT554		177,945,266	190,677,903	10.0	1,005
GT338		177,936,529	189,944,474	9.6	2,369
GT369		177,578,725	189,891,367	9.7	2,415
GT728		177,936,529	189,891,367	9.6	2,347
GT167		177,936,529	189,891,367	9.6	2,338
GT820		177,936,529	189,002,080	9.4	2,198
GT633		179,386,458	190,680,905	8.8	2,118
GT966		180,689,290	192,059,107	8.5	781

Figure 43: Group of Chr1 matches for GT611 that also matched GT654.[311]

All of the genotypes used in these comparisons are not phased. To ensure all of the individuals match an identical segment (see *Correlating* $\mathfrak{m}$ on page 60), all of the individuals in the group were also shown to match GT654.

The author has not been able to contact most of the individuals in this list; therefore, for most individuals, a common ancestor remains unidentified. There are few in the group (individuals with a "Y" in the *CA* column in Figure 43) that share a common ancestor as shown in Figure 44. Because all the genotypes in the group passed the correlating comparison, it is unlikely that any are IDC. If all the shared segments turned out to be IBD, we would expect all of the members of this group to have a common ancestor, though the common ancestor may be an ancestor of the common ancestor shared by the four with a known common ancestor.

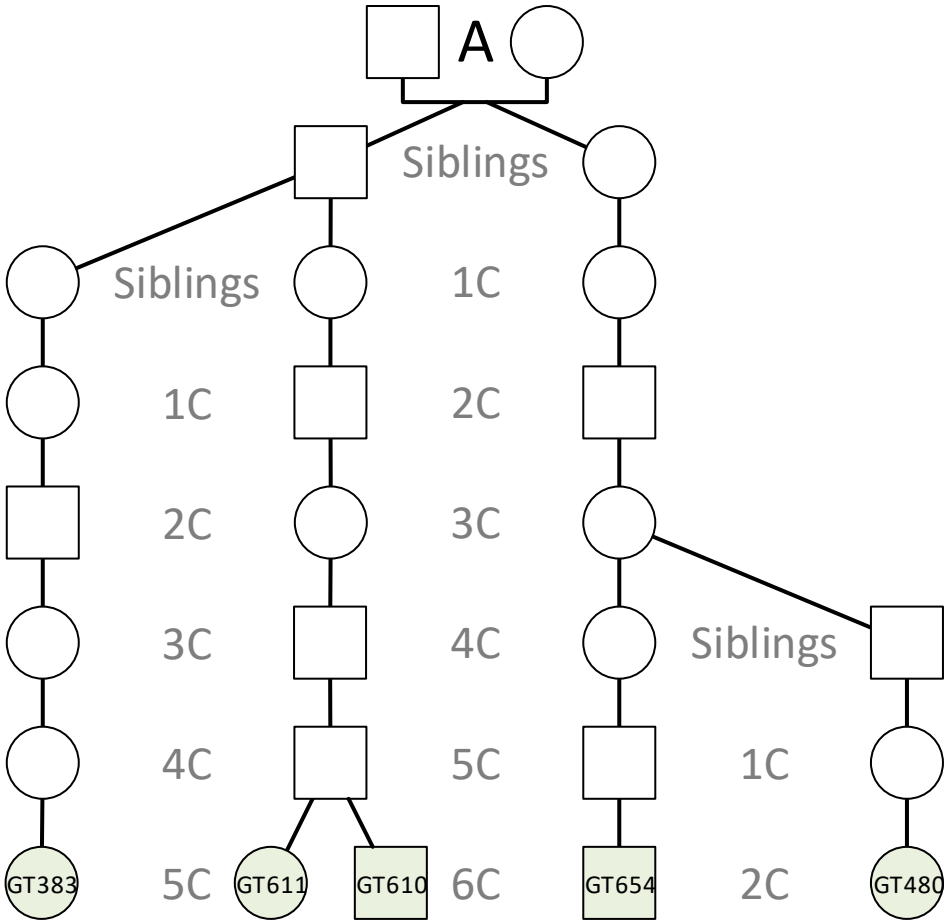

Figure 44: Representation of lineages linking connecting individuals in a triangulated group with GT611 for Chr1 (177M to 191M).[312,313,314,315] *See also Figure 28.*

GT611 and GT610 are siblings and atDNA confirms this relationship. In many instances, GT611 and GT610 can be used interchangeably in the testing detailed in this appendix. Unless GT610 is mentioned explicitly, GT611 can be considered a proxy for GT610 in that test as either sibling's result would be equivalent.

GT654 and GT480 are second cousins. They share 194.1 cM of atDNA.[316] This is consistent with Degree 5 relationship (which includes second cousins) as given in Figure 10.

Using the matches shown in Figure 44, the matching segment for this group is positioned from 177,088,988 (177M) to 190,827,077 (191M) with a physical length of 13,738,089 bps (13.7 Mbp) and a genetic size of 10.5218 cM.[317]

The *hypothesis* is that GT611, GT610, GT654, GT383 and GT480 all received their

matching segment IBD from the common ancestor identified for the triangulated group. Before accepting the *hypothesis*, factors affecting the likelihood of reliability, errors or misinterpretations must be considered; also, the items of *evidence* should agree with each other and with the principles of inheritance. The testing that follows will help determine whether the *hypothesis* can be accepted.

TESTS OF ANALYSIS

MATCHING SEGMENT SIZE

The size of the GT611's match with GT654 is greater than 20 cM and (based on Figure 19) the likelihood it will not survive phasing is negligible. The size of the matches with GT383 and GT480 are much smaller and there is roughly a 5-10% chance that these matches would fail to match if compared to a phased genotype. These matches remain candidates to be IBD, but it would be desirable to have other testing to strengthen this claim.

Using physical length, the segment that GT611 shares with GT654 is 26,228,975 bps (26.2 Mbp), and with GT480 is 13,942,455 bps (13.9 Mbp). GT654 and GT480 are sixth cousins with GT611—*G*=7 in the context of Speed and Balding's work (see Figure 22). Using Figure 23, the smallest shared segment is in the tranche from 10-20 Mbp, and the largest is in the tranche from 20-30 Mbp. For the 10-20 Mbp tranche, there is more than a 20% chance that a shared segment of the given size came from an ancestor with *G*=7 or closer. For the 20-30 Mbp tranche, the likelihood is roughly 45%. It is not unreasonable for shared segments of this size to be IBD.

GT383's shared segment has a length of 14,892,170 bps (14.9 Mbp), but a value of *G*=6. Using Figure 23, there is just under a 20% chance that a shared segment of this size is from an ancestor with G=6 or closer. It is not unreasonable for a shared segment of this size to be IBD.

In all cases, the matches remain likely candidates to be IBD.

TOTAL SHARED IBD

A comparison with a theoretical average for total sharing is rendered meaningless because the closest relationship (GT611 and GT383 are fifth cousins) is expected to

share just 3.32 cM which is well below the segment reliability thresholds.[318] The fifth cousin relationship is outside the set of relationships reported by the *Shared cM Project* in Figure 21. The distribution for fifth cousin relationships is difficult to discern in Figure 10, so it is not particularly useful for this situation.

Instead, total IBD sharing testing degrades to an evaluation of the shared segment within the context of Speed and Balding's work.[319] This has already been considered as part of the matching segment size testing and did not cast any doubt on the IBD claim.

TESTS OF CORRELATION

INDEPENDENCE

Value shown is cM total of matching segments over minimum threshold.

Kit	name	GT611	GT610	GT654	GT383	GT480	GT938	GT557	GT728	GT167	GT338	GT196	GT369	GT341	GT116	GT820	GT554	GT633	GT966
GT611			2732.0	23.2	11.3	10.8	12.9	10.0	9.6	9.6	17.1	10.0	9.7	26.2	26.9	9.4	10.0	8.8	16.6
GT610		2732.0		23.2	11.3	10.8	12.9	15.2	9.6	9.6	9.6	15.2	9.7	26.2	26.9	9.4	10.0	8.8	16.6
GT654		23.2	23.2		17.5	194.1	10.3	10.3	9.6	9.7	9.8	10.1	14.8	29.8	29.5	9.2	9.6	8.8	13.7
GT383		11.3	11.3	17.5		43.5	9.7	10.9	9.8	9.7	10.2	9.5	9.7	36.3	44.6	9.2	9.6	8.6	16.0
GT480		10.8	10.8	194.1	43.5		10.2	19.0	9.6	15.5	9.6	10.3	9.7	17.3	17.4	9.2	18.4	14.4	12.2
GT938		12.9	12.9	10.3	9.7	10.2		12.1	14.7	17.1	20.2	22.2	19.9	10.4	9.6	11.0	12.6	18.5	8.8
GT557		10.0	15.2	10.3	10.9	19.0	12.1		23.2	24.2	11.4	21.3	16.9	11.0	9.6	23.0	16.7	14.8	8.5
GT728		9.6	9.6	9.6	9.8	9.6	14.7	23.2		14.7	14.3	15.0	17.5	15.3	9.6	9.4	10.9	14.2	
GT167		9.6	9.6	9.7	9.7	15.5	17.1	24.2	14.7		14.4	18.0	13.6	10.4	9.6	29.1	17.6	13.2	6.5
GT338		17.1	9.6	9.8	10.2	9.6	20.2	11.4	14.3	14.4		14.5	12.7	11.4	9.6	9.3	13.1	12.8	6.5
GT169		10.0	15.2	10.1	9.5	10.3	22.2	21.3	15.0	18.0	14.5		13.6	18.2	9.5	12.7	15.9	13.5	8.6
GT369		9.7	9.7	14.8	9.7	9.7	19.9	16.9	17.5	13.6	12.7	13.6		10.5	15.3	10.8	24.1	10.6	15.6
GT341		26.2	26.2	29.8	36.3	17.3	10.4	11.0	15.3	10.4	11.4	18.2	10.5		3582.7	9.6	10.1	8.4	19.2
GT116		26.9	26.9	29.5	44.6	17.4	9.6	9.6	9.6	9.6	9.6	9.5	15.3	3582.7		9.1	9.6	8.3	18.9
GT820		9.4	9.4	9.2	9.2	9.2	11.0	23.0	9.4	29.1	9.3	12.7	10.8	9.6	9.1		15.8	8.2	
GT554		10.0	10.0	9.6	9.6	18.4	12.6	16.7	10.9	17.6	13.1	15.9	24.1	10.1	9.6	15.8		9.5	
GT633		8.8	8.8	8.8	8.6	14.4	18.5	14.8	14.2	13.2	12.8	13.5	10.6	8.4	8.3	8.2	9.5		6.9
GT966		16.6	16.6	13.7	16.0	12.2	8.8	8.5		6.5	6.5	8.6	15.6	19.2	18.9			6.9	

Figure 45: Total amount of atDNA sharing between members of the group that triangulates on a matching Chr1 segment from 177M to 191M.[320]

There are seventeen genotypes that match the segment being examined here. Of this seventeen, there are three pairs of closely related individuals: GT611 and GT610 (siblings), GT654 and GT480 (second cousins), and GT116 and GT341 (a parent/child pair). The remaining individuals appear to be more distantly related. This means that thirteen independent answers to the triangulation question (thirteen independent instances of ε) are possible with this set of matches. The current hypothesis has only two of the possible thirteen instances of ε.

EXCESSIVE MATCHING

There are fourteen independent matches with GT611 on this segment—which starts to feel like a large number. This may be indicative of excessive matching, which means some of these matches may be IBS. Many matching segments are on the small side, making them more likely to be IBS, or at least making the associated

individuals more distantly related (and therefore hard to identify as relations because genealogies do not generally reach back far enough in time to make the necessary connections).

Chromosome Map Correlation

Figure 31 is a Chr1 map for two of the members of the triangulated group: GT611 and GT610. The matching segment under consideration lies somewhere in between the yellow line marking 175M and 195M. The matching segment belongs to the brown-colored regions. There are no crossover events within this range. Within this range, there is only one possible configuration of siblings that could match the brown-colored regions—and the expected two siblings are the ones matching this segment. The chromosome map supports the IBD claim.

Intermediate Common Ancestors

GT654 and GT480 each could be considered an intermediate common ancestor of the other. This supports the IBD claim.

Phased Matching

None of the genotypes are phased, so no phased matching was possible.

Generational Matching

There is no lineal descendancy in the triangulated group, so generational matching cannot be tried.

Close Relative Matching

GT654 and GT480 are second cousins, and both share this match, supporting the idea that the matching segment was received IBD. For GT611 and GT610, this is a paternal match, and all their close relatives that are genotyped are maternal relatives; genotyped descendants do not match this segment.

Match Stability

There is no lineal descendancy in the triangulated group, so match stability cannot be evaluated.

ε (1 OF 2)

This instance of ε is between GT611 (and GT610) and GT654 and GT480.

COMMON ANCESTOR UNIQUENESS

		GT611		GT654	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	6	75%
5	16	16	100%	12	75%
6	32	32	100%	13	41%
7	64	64	100%	14	22%
8	128	108	84%	16	13%
Total	255	235	92%	68	27%

Figure 46: Compiled genealogy completeness evaluation for GT611 and GT654.[321,322]

		GT611		GT480	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	32	100%
7	64	64	100%	55	86%
8	128	108	84%	69	54%
Total	255	235	92%	187	73%

Figure 47: Compiled genealogy completeness evaluation for GT611 and GT480.[323,324]

GT654's genealogy is very incomplete. GT480 is better. But because these two are second cousins, the only portion of the tree that needs to be good is the portion beyond their common ancestor. In GT654's genealogy, the genealogy is complete up to the generation prior to the common ancestor and it is missing half (eight) of the identities in the generation of the common ancestor. GT480's is the same.

GT480's appears to have original research up until the last few generations, then has no sources—a surprise given the thorough documentation prior to that point. GT654's is similar.

CONCLUSION FOR ε (1 OF 2)

The size of the match with GT480 is a risk because there are no phased comparisons, but the fact that her cousin GT654 has a very likely IBD match with GT611 mitigates this risk. No other analysis factors cast doubt on this match. The match is helped by its fit with the chromosome maps for GT611 and GT610. It is helped by the fact that GT654 and GT480's second cousin relationship acts as an intermediate MRCA. It is hurt by the potential for excessive matching. It is also hurt somewhat by the incompleteness of the genealogies.

ε (2 OF 2)

This instance of ε is between GT611 (and GT610) and GT383.

COMMON ANCESTOR UNIQUENESS

		GT611		GT383	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	20	63%
7	64	64	100%	23	36%
8	128	108	84%	29	23%
Total	255	235	92%	103	40%

Figure 48: Compiled genealogy completeness evaluation for GT611 and GT383.[325,326]

The mechanism that was used to share GT383's genealogy did not include sources; it is essentially an Ahnentafel ancestors report. Again, the genealogy is quite incomplete.

CONCLUSION FOR ε (2 OF 2)

The size of the match with GT383 is a risk because there are no phased comparisons, so there is a small chance that GT383 is not IBD. No other analysis

factors cast doubt on this match. The match is helped by its fit with the chromosome maps for GT611 and GT610. It is hurt by the potential for excessive matching. The lineage is also hurt by the incompleteness of the genealogy.

OTHER CONSIDERATIONS

The MRCA between GT611 and GT383 is one generation newer than the common ancestor shared with GT654 (and GT480). This is not ideal. It could be argued that the MRCA between GT611 and GT383 should be presented as an intermediate MRCA, but this leaves only one instance of ε—making a conclusion impossible. But lacking other matches with known genealogies, and lacking any conflict, and given the relative independence of these two instances of ε, it seems safe to accept both as instances of ε for the purposes of triangulation until such time as a proper instance of ε presents itself (at which time the MRCA between GT611 and GT383 can become an intermediate MRCA), or until a conflict overturns the proposed solution.

CONCLUSION ACCEPTING

This shared segment is very "matchy"—an excessive matching risk. So far, only two out of the thirteen possible instances of ε have contributed an answer to the triangulation question—leaving eleven opportunities to introduce conflict. The incompleteness of the genealogies also remains a risk.

The main supporting factors are the strength of GT654's match and the fit with the chromosome maps for GT611 and GT610.

An additional instance of ε that fits the hypothesis would establish a stronger pattern that future instances of ε would confirm the *hypothesis*. Without discarding the *hypothesis*, it seems better not to accept it and pursue additional instances of ε.

APPENDIX E: AN X CHROMOSOME MATCH

GT999 shares a large match on the X chromosome with GT793.

KIT #	Chr	START POS	END POS	cM	SNPs
GT793	10	114,905,204	121,216,437	10.6	803
GT793	X	31,676,880	100,037,158	59.7	2,310

Figure 49: Segments that GT999 shares with GT793.[327,328]

The common ancestor shared between GT999 and GT793 was identified as given in Figure 50.

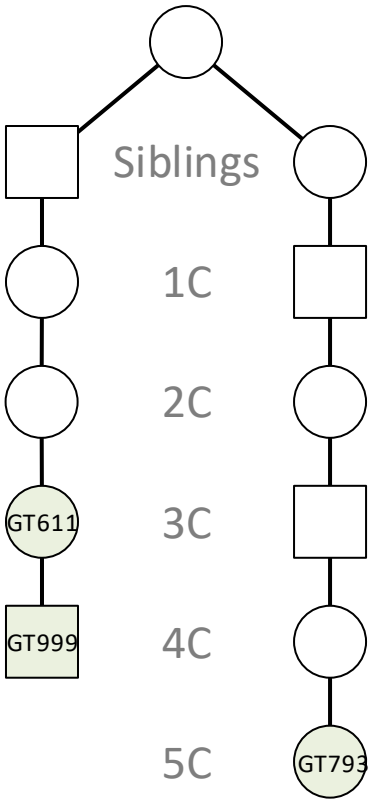

Figure 50: Representation of the lineages that relate GT999 and GT793.[329,330]

Using Figure 3, GT999 is expected to have received (on average) 12.5% of his X chromosome from the specified common ancestor, while GT793 is expected to receive (on average) 6.25% of her X from this ancestor. Using 196.1 cM as the total size of the X chromosome, GT999 is expected to receive about 24.5 cM from this ancestor, and GT793 about 12.3 cM.[331]

So why is the amount of X DNA received by these two cousins from this ancestor—59.7 cM—so high?

The author does not know the answer to this question. Having made this statement, there are anecdotal reports that indicate that the X chromosome may not participate in recombination as often as one might expect, perhaps not reshuffling at all some percentage of the time.[332] In other words, the recombination rate (the number of crossover events expected) is lower for the X chromosome than for the autosomes. Schaffner gives the rate as two thirds of the genome average.[333] The implication of this is that the theoretical percentages given in Figure 3 may be misleading. The percentages are based on the autosome's recombination model, but that model may not fit the realities of the X chromosome.

APPENDIX F: TRIANGULATION FOR GT999 ON CHR1 FROM 180M TO 195M

The following triangulated group was identified for GT999 on Chr1:

KIT #	START POS	END POS	cM	SNPs
GT611	72,017	247,169,190	281.5	54,743
GT709	111,732,674	194,928,236	60.8	13,486
GT610	114,435,751	200,890,245	64.9	13,883
GT861	99,175,785	194,953,541	73.8	16,350
GT901	152,790,212	194,928,236	45.5	9,809
GT553	175,724,793	194,928,236	14.7	3,669
GT439	179,239,541	194,925,386	12.4	2,851
GT436	180,141,290	194,953,541	11.2	2,648

Figure 51: Triangulated group with GT999 for a matching segment on Chr1 from 180M to 195M.[334]

The relationships shared among these genotypes are shown in Figure 52.

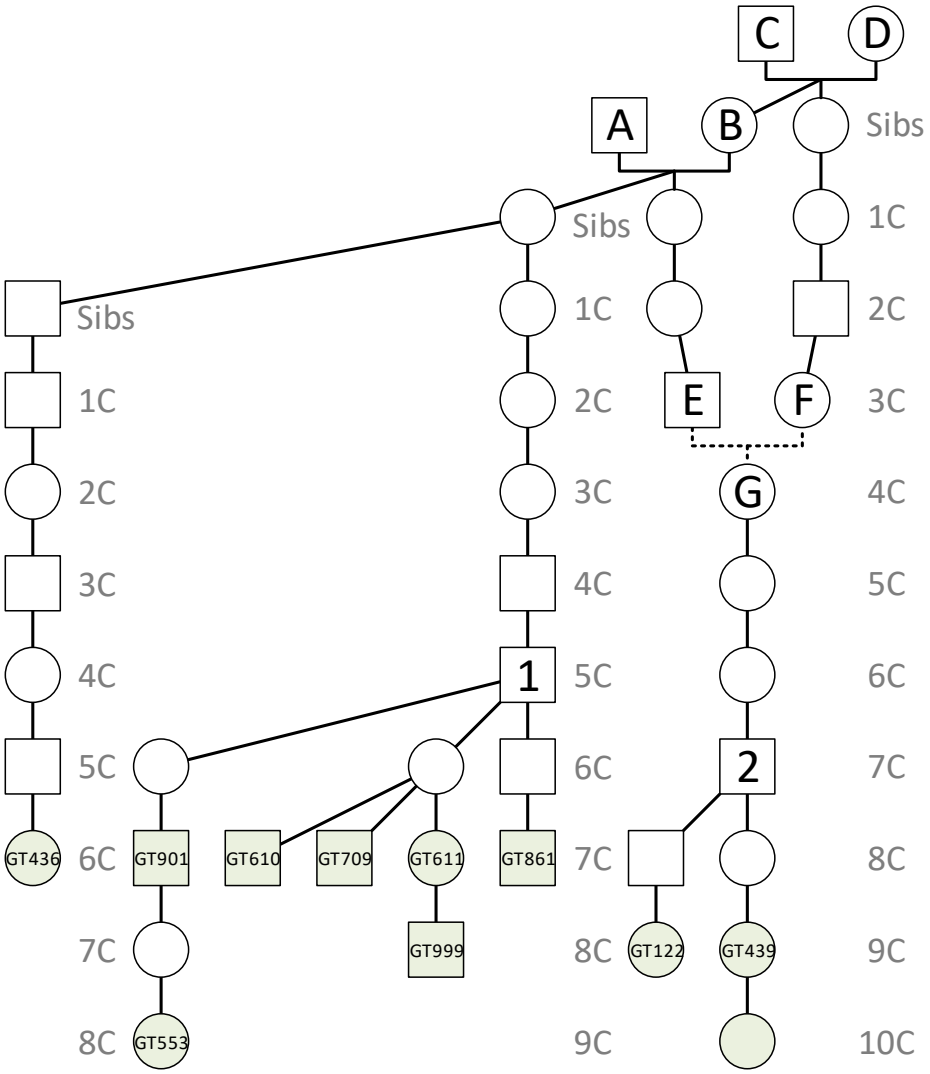

Figure 52: Representation of the known lineages connecting members of a triangulated group for Chr1 (180M to 195M).[335,336,337]

The triangulated group features two family groups and one individual. The first family group (the common ancestor for the family group is marked with a *1*) has representation from seven genotypes (GT611, GT709, GT610, GT861, GT901 and GT553). The second family group (the common ancestor for the family group is marked with a *2*) is represented in the analysis by only one individual: GT439. The GT122 genotype is not part of the triangulated group (because it does not triangulate with GT436).[338] There is another genotype without an identifier—the daughter of GT439—who is reported to match the group, but whose genotype has not been made available for comparisons.[339,340]

There is another genotype from 23andMe™ that has not been represented, but that has been shown to match the triangulated group.[341] A common ancestor with this match has not been identified. Attempts to recruit the cooperation of this match (other than the default genotype sharing on 23andMe™) have been unsuccessful. This genotype is not included in this analysis.

Using the matches listed in Figure 51, the shared match is positioned from 180,141,290 (180M) to 194,925,386 (195M) with a physical length of 14,784,096 bps (14.8 Mbp) and a genetic size of 11.1874 cM.[342]

PROPOSED COMMON ANCESTOR

A search for a common ancestor is usually required to derive the full genealogical benefit of a matching segment. In the case of GT999 and GT439, a lengthy search for a common ancestor ensued.

GT439 had a "brick wall" ancestor identified as Susan SHAW (labeled G in Figure 52) who was married to John J TRAVER and who died in Potsdam, St Lawrence, New York on 18 March 1855.[343] After an extended search of the ancestors of GT999 and GT439, Susan SHAW was identified as an ancestor of interest because GT999 was known to have ancestors that had lived in Potsdam. Continued analysis revealed that Abiel SHURTLEFF (labeled A in Figure 52) and Lydia BARNES (labeled B), the 8th great grandparents of GT999, had a descendant identified as Daniel SHAW (labeled E), who had died in Potsdam and whose age and family might accommodate a daughter that fit what was known about Susan SHAW.[344] To date, all searching for a familial connection between GT999 and GT439, other than

the SHAW connection, has been unsuccessful. With atDNA suggesting a family link and given that extensive searching had not revealed any other options, Abiel and Lydia were proposed as common ancestors.

Later, it was shown that GT436 was also related to Abiel and Lydia (as shown in Figure 52).

TRIANGULATION EVALUATION

Before accepting the *hypothesis* that GT999, GT439 and GT436 received the specified matching segment IBD from Abiel and Lydia, the hypothesis must be scrutinized. Factors that affect reliability or that increase the likelihood of errors or misinterpretations must be considered. The items of *information* and *evidence* should agree with each other and with the principles of inheritance. The testing that follows will help determine whether the *hypothesis* can be accepted as a *conclusion*.

TESTS OF ANALYSIS

MATCHING SEGMENT SIZE

Using Figure 19, the probability that any of these matching segments is IBC is less than 5%. As the comparisons were made with a phased genotype, the likelihood that these matches are IBC has been essentially eliminated. All are well above the minimum 5.0 cM threshold suggested for phased matches, the threshold necessary to achieve an acceptable level of false positives.[345] The matching segments are certainly good candidates to be IBD.

TOTAL SHARED IBD

A comparison with a theoretical average for total sharing is rendered meaningless because the closest relationship (GT436 with GT439) is expected to share just 0.1 cM which is well below the 5 cM segment reliability threshold.[346] The relationship is also outside the set of relationships reported by the *Shared cM Project*.[347,348]

Instead, total IBD sharing testing degrades to an evaluation of the shared segment within the context of Speed and Balding's work.[349] The common ancestor is at G=8 or G=9 (Figure 22) for the member of this triangulated group. A 14.8 Mbp shared region falls in the tranche between 10 and 20 Mbp (Figure 23). There is just under a 40% chance that a segment of this size was shared IBD by an ancestor in the most

recent ten generations, and a roughly 5% chance it came from either a G=8 or G=9 ancestor. It would not be considered impossible, or even improbable, that a shared segment of this size (if present) came from the proposed common ancestor IBD.

TESTS OF CORRELATION

INDEPENDENCE

Value shown is cM total of matching segments over minimum threshold.

Kit	name	PGT999M1	GT611	GT610	GT709	GT861	GT901	GT553	GT439	GT436
PGT999M1			3587.1	1791.6	1747.7	568.2	465.9	133.8	12.5	11.2
GT611		3587.1		2732.0	2711.0	930.3	691.3	178.6	23.9	13.1
GT610		1791.6	2732.0		2697.4	813.1	852.3	213.6	23.9	13.1
GT709		1747.7	2711.0	2697.4		851.2	815.6	186.5	23.3	12.5
GT861		568.2	930.3	813.1	851.2		963.1	245.0	19.5	13.0
GT901		465.9	691.3	852.3	815.6	963.1		1820.4	23.4	12.6
GT553		133.8	178.6	213.6	186.5	245.0	1820.4		18.1	12.1
GT439		12.5	23.9	23.9	23.3	19.5	23.4	18.1		21.8
GT436		11.2	13.1	13.1	12.5	13.0	12.6	12.1	21.8	

Figure 53: Total amount of atDNA sharing between members of the group that triangulates on a matching Chr1 segment from 180M to 195M.[350]

The report shown in Figure 53 helps evaluate independence within the group. The first seven genotypes in the report are all members of Group 1 (Figure 52). There are two other individuals, independent of Group 1 and independent of each other. Only two independent answers to the triangulation question are possible—the minimum required for triangulation.

EXCESSIVE MATCHING

As discussed previously, there are three independent matches in the triangulated group, and there is only one other candidate that has been identified as matching with the group. This shared segment is not at risk of excessive matching. Rarity strengthens the claim that this matching segment could have been received IBD.

CHROMOSOME MAP CORRELATION

Figure 31 is a Chr1 map for three of the members of the triangulated group: GT611, GT709 and GT610. The matching segment lies between the two yellow lines marking the range 175M and 195M. There are no crossover events within this range. There is only one possible configuration where three siblings could match the same region—the lighter-blue region—and it fits with the siblings that match this triangulated group. The chromosome map agrees with the IBD claim.

INTERMEDIATE COMMON ANCESTORS

There are no known intermediate common ancestors as yet. GT999 has recruited several third, fourth and fifth cousins but none have matched this segment. There are also some AncestryDNA™ matches that might be possible matches based on their shared lineage with GT999, but AncestryDNA™ does not give identifying details (chromosome, start and end locations, size, SNP count) for matching segments, and the genotype administrators have not responded to inquiry.

PHASED MATCHING

GT999 has phased haplotypes that were created with the haplotypes of both parents. All of the genotypes in the triangulated group have been matched against GT999's phased maternal haplotype successfully, essentially eliminating the risk that IBC matches were included in the group—giving a boost to the IBD claim.

GENERATIONAL MATCHING

Using GT999's phased genotype guarantees a generational match with his mother GT611. For GT553, her grandfather GT901 shares the segment. For GT439, her daughter shares the segment. Each of these gives evidence that the matching segment might have been received IBD, giving credibility to the IBD claim.

CLOSE RELATIVE MATCHING

From GT999's point of view, he has five other close relatives that match GT439 and GT436—two uncles, two of his mom's first cousins, and the granddaughter of one of those cousins. There is plenty of evidence that members of Group 1 received this matching segment IBD from their common ancestor (labeled *1* in Figure 52) or his spouse, which gives credence to the idea that they received it IBD from the common ancestor for the triangulated group.

Group 2 includes GT439's first cousin GT122. This cousin does not match GT999 or GT436 but does match the other members of the triangulated group. These matches strengthen the claim of relatedness, and in so doing, strengthen the claim that the matching segment was received IBD.

MATCH STABILITY

There are three pairs of individuals who have a lineal relationship with each other that share this matching segment.

Without the data from her daughter, stability cannot be considered for GT439 and her daughter.

The match is stable—genetically correct; i.e., the newest generation received the matching segment that was the same as or smaller than the older generation's, and the newer generation's matching segment was bounded at or within the boundaries of the older generation's—as it is passed from mother (GT611) to son (GT999).[351,352] This was also true for the matching segment as it was passed from GT901 (grandfather) to GT553 (granddaughter).

A lack of stability (not the case here) would have cast doubt on the IBD claim.

ε (1 OF 2)

The first instance of ε is between Group 1 and Group 2. All of the members of these two family groups match each other in the expected ways (as measured by total sharing).[353,354]

COMMON ANCESTOR UNIQUENESS

		GT999		GT439	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	10	63%
6	32	32	100%	16	50%
7	64	64	100%	22	34%
8	128	126	98%	31	24%
9	256	218	85%	56	22%
10	512	389	76%	46	9%
Total	1023	860	84%	196	19%

Figure 54: Compiled genealogy completeness evaluation for GT999 and GT439.[355,356]

GT999 is used as a proxy for Group 1. Because of close-relative matching, the match is known to have come from GT999's maternal grandmother's father's family—narrowing the portion of his pedigree that needs to be searched to 1/8th of his ancestry. In this portion of his pedigree, only ten ancestors have not been identified—just less than 16% of the expected number of ancestors in this part of the pedigree.

GT439 is used as a proxy for Group 2. Using close-relative matching and ethnicity information, it is believed that this match would be a maternal match for GT439. The pedigree for GT439 is quite incomplete, but the biggest gaps in the pedigree are paternal gaps. The incompleteness is further manifested in the fact that the proposed common ancestor would also "break down a brick wall" for GT439. Because of the gaps in this compiled genealogy, the search for a MRCA had to be supplemented heavily with other published genealogies and supplemental research.[357,358,359] These supplemental activities combined with many hours of searching should serve to mitigate the risks presented by the missing genealogy.

CONCLUSION FOR $\mathcal{E}$ (1 OF 2)

The main flaw with the first instance of $\mathcal{E}$ is the incompleteness of the genealogies. Much has been done to mitigate this risk by using other published genealogies and doing supplemental research. The author believes that the remaining risk does not leave the solution any more vulnerable than if the pedigrees had been more acceptably complete.

$\mathcal{E}$ (2 OF 2)

The second instance of $\mathcal{E}$ is between Group 1 and GT436.

COMMON ANCESTOR UNIQUENESS

		GT999		GT436	
Generation	Total # Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors	Total # Identifed Ancestors	Percent of Expected Ancestors
1	1	1	100%	1	100%
2	2	2	100%	2	100%
3	4	4	100%	4	100%
4	8	8	100%	8	100%
5	16	16	100%	16	100%
6	32	32	100%	27	84%
7	64	64	100%	46	72%
8	128	126	98%	66	52%
9	256	218	85%	86	34%
10	512	389	76%		
Total	1023	860	84%	256	50%

Figure 55: Compiled genealogy completeness evaluation for GT999 and GT436.[360,361]

The pedigree for GT436 has a number of gaps beyond the fifth generation. The pedigree seems to be a mix of original research and information copied from other

compiled genealogies—though in cases of reliance on other's research, it is often supported with well-regarded secondary sources, or one or two original records.

CONCLUSION FOR $\mathcal{E}$ (2 OF 2)

Again, the declaration of a common ancestor (a) is exposed to risk relating to an incomplete genealogy. Not as much has been done to directly mitigate the risks introduced by this second genealogy. Some mitigation is had in the fact that there are two instances of $\mathcal{E}$ (each with independent instances of m and independent lineages to a), which is rare in and of itself; this tends to offset the risk that lingers due to the lack of coverage in the MRCA search.

The match (m) in the second instance of $\mathcal{E}$ bears up to scrutiny; and while there is lingering risk regarding common ancestor (a) due to the incomplete genealogies used in the search for the MRCA, the author feels it is an acceptable risk.

SOLUTION PREDICTS RELATIONSHIPS

Attempts to contact GT436 via GEDmatch in order to find a common ancestor with the group initially failed. Needing an additional match to triangulate with Group 1 and Group 2, the author spent time identifying matches that could potentially corroborate the common ancestor that had been identified between Group 1 and Group 2. He identified an AncestryDNA™ match where the common ancestor identified by AncestryDNA™ was one generation short of the common ancestor, and attempted contact. It turned out that the AncestryDNA™ genotype and the GEDmatch genotype were GT436's genotype. In this sense, the common ancestor identified between Group 1 and Group 2 helped predict that the AncestryDNA™ genotype would be a fit in the triangulated group.

OTHER CONSIDERATIONS

The MRCA between Group 1 and GT436 is one generation closer than the common ancestor shared with Group 2. This is not ideal. It could be argued that the MRCA between Group 1 and GT436 should be represented as an intermediate MRCA, but this leaves only one instance of $\mathcal{E}$—making a conclusion impossible. Lacking other matches with known genealogies, and lacking any conflict, and given the relative independence of these two instances of $\mathcal{E}$, it seems safe to accept both as instances of $\mathcal{E}$ for the purposes of triangulation until such time as a proper instance of $\mathcal{E}$

presents itself (at which time the MRCA between Group 1 and GT436 can become an intermediate MRCA), or until a conflict overturns the proposed solution.

The above discussion has considered the MRCA (labeled *A* and *B* in Figure 52). Interestingly, there is a common lineage between members of this group that could go through a different couple (labeled *C* and *D*) back one generation further. Genetic genealogists assume the MRCA to be the ancestor that contributed a matching segment, but additional matches could change aspects of this assumption. For example, an intermediate common ancestor sharing this matching segment along the lineage between Susan SHAW (labeled *G*) and the couple labeled *C* and *D* would cause us to modify our conclusion about how GT439 received this matching segment. [Whether the matching segment was received by GT439 via Abiel and Lydia (the couple labeled *A* and *B*) or by the couple labeled *C* and *D* would not change the conclusion about the parents of Susan SHAW.]

CONCLUSION ACCEPTING

None of the testing of the matches (instances of m) cast any doubt in the IBD claim. Some might see lingering doubts in the deficiencies of the search for a MRCA, but the author feels that the work to mitigate these risks, particularly the work leading to the identification of the MRCA between GT999 (Group 1) and GT439 (Group 2), is sufficient to alleviate these doubts. Two lineages were identified by which GT439 could have received this matching segment IBD, but information accumulated thus far is insufficient to select one over the other; for now, the selection of lineage is based on the prevailing MRCA assumption. It is therefore *concluded* that one person in the ancestral couple identified as common to this triangulated group (Abiel SHURTLEFF and Lydia BARNES—labeled *A* and *B* in Figure 52) must have contributed this matching Chr1 segment (180M to 195M) to the members of this triangulated group.

THE SHAW CONNECTION

The proposed common ancestor's viability rests (in part) on the following conclusion: that Susan SHAW (married to John J TRAVER) is the daughter of Daniel SHAW and Mary BARROWS.

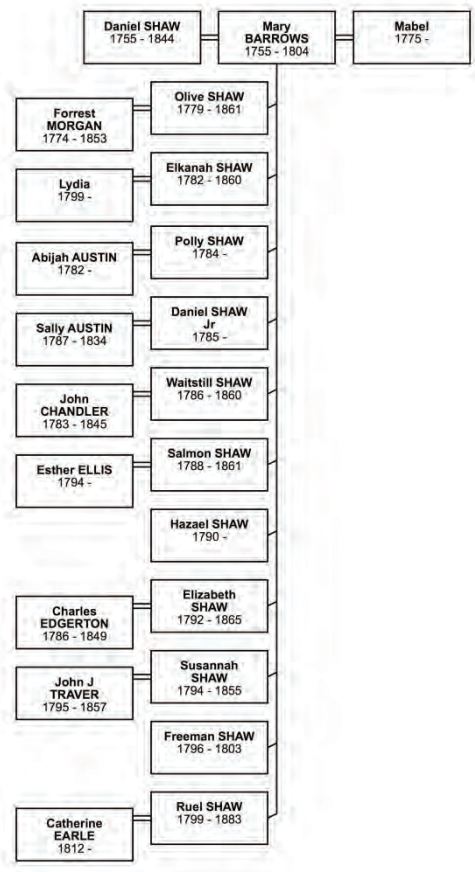

Figure 56: The Daniel SHAW family as discussed in this appendix.

This appendix considers the evidence available about Daniel SHAW and his family and whether Susan is a viable candidate as a daughter in this family. Ultimately, the conclusion outlined here rests on the following:

- A conclusion that the atDNA in common between GT999 and GT439 is shared identical by descent (IBD) through Abiel SHURTLEFF or Lydia BARNES as the proposed common ancestor between GT999 and GT439—a conclusion examined above and incorporated here as the connection to the Daniel SHAW family is considered.
- A conclusion that the persons with the SHAW surname found in the Potsdam arrival record(s)—considered below—are a single family group: Daniel SHAW and his children.
- A conclusion that any persons that resided in Potsdam and bore the SHAW surname in the early 1800s are part of a single family group: Daniel SHAW and his descendants—considered below.

DANIEL SHAW IDENTIFIED

From a Revolutionary War pension file, personal testimony from him and his wife Mabel reveals the following details about Daniel SHAW (unless otherwise noted).[362] Daniel SHAW was born 12 Oct 1753 in Middleborough, Plymouth, Massachusetts. Sometime during the war, he moved to Plympton, Plymouth, Massachusetts—probably about the time he married Mary BARROWS (a native of Plympton) in Plympton on 6 Aug 1778.[363] Daniel lived in Plympton for eight to ten years, then removed from thence to Bridgewater, Windsor, Vermont. After six years in Bridgewater, he moved to Rochester, Windsor, Vermont. His wife Mary died in Rochester 4 May 1804.[364] Daniel was married a second time in late 1804 to Mrs. Mabel Easton.[365] After twenty years in Rochester, Daniel SHAW moved from Vermont to Potsdam, St Lawrence, New York—maybe in late 1811, as his name was published in the *List of Letters* in five successive issues of the newspaper in early 1812.[366] Daniel died 22 Mar 1844.

THE UNION

Daniel's sons Elkanah and Salmon were the first of the SHAW family to move from Vermont to Potsdam.[367] A man named William BULLARD convinced several associates (mostly from Royalton, Windsor, Vermont) to join him in an experiment in communal living. Mr. BULLARD was said to be a student in community theory and had published a pamphlet espousing his ideologies. In 1803, Mr. BULLARD identified lands just north of Potsdam Village as a suitable site for the inauguration of his experiment. On 28 Nov 1804, he and his associates took possession of some 2400+ acres of land. In some sources, Elkanah and Salmon are named among Mr. BULLARD's founding associates.

Mr. BULLARD's cooperative organized itself as "The Union", formally adopting a constitution in 1807.[368] The community is said to have thrived well enough. But problems among community participants soon surfaced and, just three years after its formal organization, "The Union" was amicably dissolved. Union lands were divided among the participants. Both Elkanah and Salmon SHAW were among the families receiving lands as "The Union" dissolved, both taking possession of tracts of land on 30 Nov 1810.[369,370]

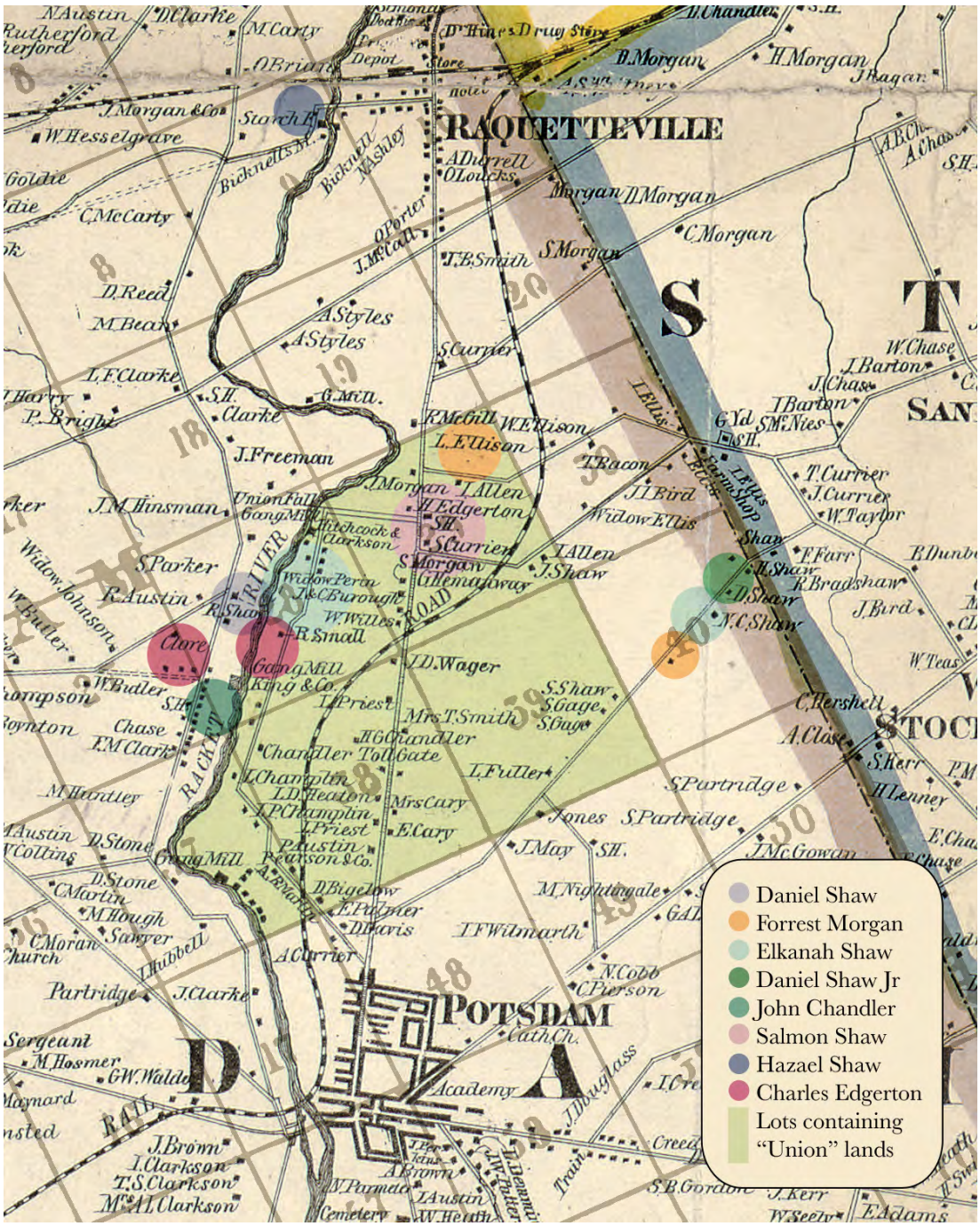

Figure 57: The numbered mile-square lots that contained "The Union" land purchases and the land purchase made by the SHAW family between 1810 and 1818 are overlaid on an 1858 map of St Lawrence county.[371,372,373,374]

Over the next eight years, Daniel, four men believed to be his sons, and three men believed to be his sons-in-law, all purchased lands in the Potsdam area. The above figure shows an approximate location for each of these purchases. The purchases are shown both in the context of the original “Union” land purchase, and in the context of an 1858 map of those same lands. In 1858, the landowners shown are generally contemporaries with Daniel’s grandchildren.

In many cases, the descendants of these purchasers are still resident on the purchaser's lands in 1858. The SHAWs' connection to "The Union" is evident. It is easy to see how the proximity of these families to each other suggest they are a family group. These purchases also establish the SHAW presence in Potsdam in the early 1800s—facts important to conclusions drawn in subsequent discussion.

NY 1815 PORT ARRIVALS

There is an index of *NY 1815 Port Arrivals* that includes entries for nine individuals with the surname SHAW (returned in alphabetical order by the search software): Daniel, Daniel Jr., Elizabeth, Elkanah, Hazel, Olive, Salmon, Susanna, and Waitstill.[375] The location given in each index entry is Potsdam, St Lawrence, New York. Given that the stated record type is an arrival record and given that these nine people are the only entries in this entire index with the surname SHAW, and given their arrival in what must have been a small Potsdam port, it seems reasonable to conclude that these individuals are members of a family group, and that perhaps they may have even been recorded in a single arrival record. The index does not identify the original source record(s) used in creating this index, so these assumptions could not be verified. However, it can be shown that Daniel's family is (in part) composed of individuals with these names.

DANIEL SHAW

It is believed that the Daniel in the *NY 1815 Port Arrivals* is Daniel (b. 1753). In support of this belief, it will be shown that the other persons with the SHAW surname in this index fit as children of Daniel. Also, the presence of a Daniel Jr. in the grouping suggests Daniel to be the senior to at least Daniel Jr.

OLIVE SHAW

Olive MORGAN (born about 1779 in Massachusetts) is enumerated with her husband Forrest MORGAN in 1850.[376] She is enumerated in the household of her son Joseph MORGAN in 1860.[377] The marriage record of her son Joseph identifies his parents a Forest MORGAN and Olive SHAW.[378] Forrest MORGAN, Olive's husband, purchased land near the Potsdam SHAW's in 1816 and 1818.[379,380] She was living near her brother Rewell (Ruel) and her son Joseph in 1850.[381] She was living near her brother Salamon (Salmon) in 1860. Her husband, Forrest, was born in Massachusetts, lived in Rochester, Windsor, Vermont, and then lives in Potsdam,

St Lawrence, New York—following a similar pattern of migration to that of Daniel himself.[382] Olive is a good fit as a daughter of Daniel SHAW.

ELKANAH SHAW

Elkanah SHAW was born in about 1782.[383] As detailed above, Elkanah and his brother Salmon were founding members of "The Union" with William BULLARD. In 1820, Elkanah was enumerated next to his brother Daniel Jr., and was living near his brother Salmon (with Salmon being enumerated next to William BULLARD).[384] The name Elkanah was very likely given in remembrance of Daniel's father, further evidence that Elkanah is the son of Daniel.[385] Elkanah is a good fit as a son of Daniel SHAW.

DANIEL SHAW JR.

Daniel SHAW Jr. was born in about 1785 in Massachusetts.[386] Daniel Jr. married Sally Austin on 18 Mar 1804 in Rochester, Windsor, Vermont.[387] The marriage was performed by his father Daniel SHAW, a Justice of the Peace—a position his father held from at least 1802 to 1811.[388,389] In 1810, Daniel Jr. was enumerated next to an Abijah Austin (believed to be both his wife's brother and his sister Polly's husband).[390,391] Daniel Jr. purchased land in 1818 near his other siblings, with his land being nearest to his brother Elkanah and brother-in-law Forrest MORGAN (as shown above).[392] In 1820, Daniel Jr. was enumerated next to his brother Elkanah and near his brother Salmon.[393] His designation as Daniel SHAW Jr. would also denote him to be the son of a Daniel SHAW. Daniel Jr. is a good fit as a son of Daniel SHAW.

WAITSTILL SHAW

Waitstill CHANDLER (born in about 1786 in Massachusetts) and her son Nelson were enumerated in 1850 in the household of her married daughter Naomi Pero (Perro).[394] Four years earlier, Waitstill was designated the administratrix of John CHANDLER, suggesting her to be his widow.[395] Waitstill's tombstone connects her to her husband John and reveals that her maiden name was SHAW.[396]

John CHANDLER purchased land near the SHAW family in 1811, with his land being nearest to brother-in-law Charles EDGERTON (Waitstill's sister Elizabeth's husband) and father-in-law Daniel.[397] John was enumerated near several SHAW

relations in the 1820, 1830, and 1840 censuses: Charles EDGERTON, Daniel, and Waitstill's brothers Salmon, Daniel Jr., and Elkanah in 1820; Charles EDGERTON, Daniel SHAW, and Forrest MORGAN (Waitstill's sister Olive's husband) in 1830; Charles EDGERTON, Daniel SHAW and Ruel SHAW (Waitstill's brother) in 1840.[398,399,400]

In 1860, Waitstill was again in Naomi's household; a MORGAN (perhaps a relative of her sister Olive's husband?) was a servant in the household.[401]

It is likely that Waitstill received her name in remembrance of her aunt (and father's sister) Waitstill who died 18 Jun 1781 at 11 years of age, approximately 5 years before she was born.[402,403]

The above facts considered together make Waitstill SHAW a solid fit as a daughter of Daniel SHAW.

SALMON SHAW

Salmon SHAW was born in about 1788.[404,405] The sources that give information about his place of birth conflict—one giving Massachusetts, and the other giving Vermont.[406,407] It would seem that his birth is very near the time his family transitioned from Massachusetts to Vermont. An informant without actual knowledge of his birth may have used a knowledge of the family's movements to estimate his birth location and, therefore, introduced this inconsistency.

As detailed above, Salmon SHAW and his brother Elkanah were founding members of "The Union" with William BULLARD. In 1820, Salmon was enumerated next to William BULLARD and near his brothers Elkanah and Daniel Jr.[408] Salmon was enumerated on the same page as his sister Olive's husband Forrest MORGAN in 1830.[409] He was near his nephew Freeman SHAW in 1840.[410] He was living near his sister Olive in 1860.[411]

Salmon named one of his son's Elkanah—a name that connects him to his brother, and his paternal grandfather.[412,413,414]

Considered together, the facts make Salmon SHAW a fit as a son of Daniel SHAW.

HAZAEL SHAW

Hazael SHAW was born in about 1790 at Bridgewater, Windsor, Vermont—a place Daniel was known to have lived.[415] Hazael was among the SHAW men purchasing land between 1810 and 1818, making a purchase in August of 1817.[416]

Daniel SHAW Jr. named one of his sons Hazael in 1819, an indication from Daniel Jr. that he is related to Hazael, and therefore Hazael's connection to the Daniel SHAW family.

The author wonders if Daniel Jr.'s use of the name Hazael in 1819 is an indication that his brother Hazael is recently deceased. In the search for SHAW records, the last record showing Hazael to be living was his land purchase of 23 Aug 1817.[417] Searches for him in subsequent censuses, newspapers, and in other general searches have not been successful. Also, it can be shown that Daniel Jr. had previously named a son after a recently-deceased brother: his son Freeman after his brother of the same name.[418,419,420]

Given these factors together, Hazael fits as a son of Daniel SHAW.

ELIZABETH SHAW

Elizabeth EDGERTON was born in about 1792 in Vermont.[421,422] In both 1850 and 1860, Elizabeth was enumerated in the household of Joseph and Louisa MORGAN.[423] In 1850, the MORGAN household also included Ransom G EDGERTON. In 1860, the MORGAN household included Olive MORGAN (i.e., Olive SHAW, mother of this Joseph MORGAN); the family was also just a few households away from Salmon SHAW.

In the surrogate court's order to grant letters of administration for the estate of Charles EDGERTON, Elizabeth was named Charles's widow, and Ransom his only [living] son.[424] In a similar grant in Ransom's probate proceedings, Louisa MORGAN was named as a sister to Ransom.[425] Elizabeth is the widow of Charles Edgerton and is living in the home of her daughter Louisa.

Elizabeth's presence in the MORGAN household with Olive SHAW is evidence of a family relationship with the SHAW family. If Elizabeth is a daughter of Daniel SHAW

(and it appears that she is), she and Olive are sisters, and Olive's son Joseph and Elizabeth's daughter Louisa are first cousins. It is perhaps unusual to find two mothers-in-law residing in the same household with their children; when considered as sisters, it seems a bit more sensible.

Elizabeth's husband Charles purchased land near the SHAWs—probably adjacent to Daniel SHAW.[426,427] Charles was enumerated next to Daniel SHAW in the 1820, 1830, and 1840 censuses.[428,429,430] Charles was also near brothers-in-law John CHANDLER, Salmon SHAW, Elkanah SHAW and Daniel SHAW Jr. in 1820, John CHANDLER and Forrest MORGAN in 1830, and John CHANDLER and Ruel SHAW in 1840. Based on enumeration position in the 1850 census' non-population agriculture schedule, his property is situated next to Joseph MORGAN who is next to Ruel SHAW.[431,432] His proximity to the SHAWs, and especially his proximity to Daniel SHAW, suggest that Elizabeth EDGERTON is a member of the SHAW family.

The above information considered together makes Elizabeth a good fit as a daughter of Daniel SHAW.

SUSANNAH SHAW

Susanna TRAVER was born in about 1794 in Vermont.[433,434,435] She is enumerated in 1850 with her husband John and three children, including a daughter Elisa R TRAVER. This daughter Elisa's death record states that she was born 6 Mar 1825 in Potsdam, St Lawrence, New York and that her parents are John TRAVER and Susan SHAW.[436] Susan (Susannah) died 18 Mar 1855 and was buried in Potsdam.[437]

In the 1850 census, Susannah's place of birth was given as New York.[438] This seems to conflict with the fact that Daniel SHAW did not live in New York at the time of her birth. However, her daughter Elisa and her daughter Susan both gave Susannah's place of birth as Vermont when asked about their mother's place of birth.[439,440] It is not known who the informant was in the 1850 census. It is easy to find examples in census records where the informant did not have actual knowledge of the person's place of birth. On the other hand, if the informant in this record was aware of Susan's place of birth, perhaps one possible explanation for such an

answer can be found in the dispute between New York and Vermont over jurisdiction of the lands that now make up Vermont—a dispute that was just being resolved within the one or two years prior to Susan's estimated date of birth.[441] With two daughters reporting her place of birth as Vermont, the New York answer becomes the outlier, giving credence to a conclusion that she was born in Vermont.

Susannah's husband John, while in Potsdam, was not enumerated particularly close to any of the other SHAW siblings or parents in the 1830 and 1840 census.[442,443] The nearest SHAW sibling to Susanna in 1850 (in enumeration order) was Waitstill, but there are sixty-seven dwellings enumerated between their entries.[444] The only proximity to suggest a relationship to other Potsdam SHAW families is Susan's own residence in Potsdam. In fact, after many hours of searching, the only record found to link Susan to Daniel SHAW (outside of her being a SHAW in Potsdam) is the *NY 1815 Port Arrivals* record(s).[445]

There is an Elkanah SHAW (b 1766, d. 1850) married to a Susanna that lived in Bridgewater, Windsor, Vermont.[446] This Elkanah is distinct from the Potsdam Elkanah (b. 1782) by both age (about 16 years different) and geography (one being consistently enumerated in Bridgewater—1800 through 1850, while at the same time the other being consistently enumerated in Potsdam—1820 through 1850).[447,448] One must consider whether the Elkanah and Susanna of Bridgewater might be the Elkanah and Susannah in the arrival records. While it may be possible, it does not seem likely. The more likely candidates would be the candidates actually living their lives in the Potsdam area.

Susannah is certainly a candidate to be a daughter of Daniel SHAW.

ARRIVAL RECORD INFORMANT

Another interesting feature of the entries in the *NY 1815 Port Arrivals* when considered as a family group, is that, based on the births of their children, at least Olive and Waitstill are already married in 1815. Why, then, would they be listed with their maiden surname in an arrival record? If one considers a family group traveling together and being recorded together in an arrival record(s), it seems possible that a single informant provided the information in that record(s). That everyone in the record(s) is listed as a SHAW suggests that the informant knew everyone in the

traveling party to be members of the same family group—that they knew everyone to be a SHAW.

ARRIVAL RECORD CONCLUSION

As given above, the index of *NY 1815 Port Arrivals* includes entries for nine individuals with the surname SHAW. It has been show that each entry can be associated with a candidate person resident in the Potsdam area. These same candidates have been shown to be part of a family group as follows: Daniel SHAW (b. 1755) and his children Olive (b. 1779), Elkanah (b. 1782), Daniel Jr. (b. 1785), Waitstill (b. 1786), Salmon (b. 1788), Hazael (b. 1790), Elizabeth (b. 1792) and Susannah (b. 1794). All of these children fit without conflict into a birth order consistent with Daniel's marriage to Mary BARROWS in 1778, that includes children Polly (b. 1784), Freemam (b. 1796) and Ruel (b. 1799), and all were born prior to Mary's last recorded births (sons Freeman and Ruel). Though unable to access the original arrival record(s), these entries, when combined with the evidence presented above, give evidence of a family relationship. This is important because this grouping includes Susannah who has not appeared in other records with her family.

BEING A SHAW IN POTSDAM

In extensive searching, any person known to have been a SHAW and to have spent time as a resident of Potsdam from the time of the foundation of "The Union" through 1850 (and beyond) have all been shown to be related to Daniel SHAW or one of his descendants, or they have not been eliminated as a descendant. In extensive searching, any person known to have been a SHAW, but that cannot be shown to have spent time as a resident of Potsdam in the specified timeframe, has yet to be shown to be a relation of Daniel SHAW. While the search continues for records about the SHAWs of Potsdam, it seems that being a SHAW in Potsdam between 1804 and 1850 is a strong indicator of a relationship with Daniel SHAW.

BRICK WALL CRUMBLING

This appendix has considered evidence about Daniel SHAW, his children, and whether Susan SHAW (who married John J. Traver) can viably be considered a daughter of Daniel SHAW and Mary BARROWS. From a genealogical proof standard point of view, at least two pieces of evidence are needed to make a

conclusion.[449] The author presents the following three evidences in concluding that Susan is, in fact, the daughter of Daniel and Mary:

- evidence of membership in the Daniel SHAW family by virtue of Susannah's association with other members of the Daniel SHAW family in the *NY 1815 Port Arrivals* records,
- evidence of membership in the Daniel SHAW family by virtue of Susannah's status as a SHAW that was resident in Potsdam in the early 1800s,
- and evidence that GT439 received a segment of atDNA that is identical-by-descent (IBD) from Abiel SHURTLEFF or Lydia BARNES, received by GT439 via Susan SHAW.

Some might consider the first two evidences to be sufficient to declare the relationship, and two evidences would be sufficient to declare a conclusion. Adding the atDNA evidence strengthens the argument considerably.

As with any proof argument, this argument stands on the strength and use of all available evidence. As new evidence comes to light, this argument may need to be reconsidered and reformed to include new findings. For now, this argument is stable with both a viable paper trail and confirming atDNA evidence.

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