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DNA Haplotyping and Diversity: An Anthropogenealogical Method for Researching Lineages and Family Ethnicity

Emerging only in the last four to five years, anthropogenealogy combines genealogy and surname studies with DNA analysis and population genetics. Described here is a method for determining the geopolitical connections and deep history of an individual's lineage.

An overview of genetics-and-genealogy begins with Skorecki et al.'s seminal publication on the chromosomes of Jewish priests. In 2000, N. Brent Kennedy became the first person to investigate his full ancestry through DNA haplotyping.

There are two main approaches. One determines the father's lineage by testing DNA material on relevant sites of the Y-chromosome. The other uses a certain region of the maternally inherited mitochondrial DNA to assemble the mutations characteristic of different female lineages. Matches for any given Y chromosomal haplotype can be found in the Y-STR Haplotype Reference Database. Mitochondrial mutations are reported in a similar concordance using the Cambridge Reference Sequence.

Two brief case studies illustrate the steps researchers can take to explore either of these lines. In most regions of the world, males passed surnames as well as property, titles, and socio-economic class. Hence, male haplotyping can be a valuable tool for determining historical relationships. MtDNA haplotyping derives its interest from the fact that it is often the mother who instils values of culture, religion and education.

Combining DNA analysis with the study of Scottish history—particularly records of medieval and early modern guilds and cemeteries of Aberdeen and Glasgow—the authors have used male haplotyping to suggest that many of Scotland's founding lineages from the Norman period were likely Jewish families from the Continent.

In summary, DNA haplotyping opens a new avenue to exploring race and ethnicity in history. In genealogy, it offers a way of overcoming brick walls and supplementing mute records. By revealing the interrelatedness of all human diversity around the world, anthropogenealogy can also be a potent force for promoting tolerance and peace.

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Short Description

Anthropogenealogy combines DNA analysis with knowledge of history, linguistics and anthropology. A step-by-step method for its practice is described and documented.

Keywords

Genetics
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Scottish Clans
Indians of North America

Y-STR Haplotype Reference Database
Cambridge Reference Sequence
Human Genome Project

Nina Jo Grimwood
John F. Kennedy
N. Brent Kennedy
Steven Newberry

Scotland
Appalachia

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The investigation of surnames in genetics goes back to George Darwin, son of the founder of evolutionary science. In 1875, Darwin *filis* used surnames to estimate the frequency of first-cousin marriages and calculated the expected

incidence of marriage between people of the same surname (isonymy). He arrived at a figure between 2.25% and 4.5% for cousin-marriage in the population of Great Britain (Jobling June 2001, p. 353), with the upper classes being on the high and the general rural population on the low end. Admittedly, this was a pretty crude effort by modern scientific standards, but quite innovative for its era. The next stimulus toward using genetics to study family history had to wait until the 1990s, when certain locations on the Y chromosome were identified as being useful for tracing father-to-son inheritance.

The new chapter began when a Canadian nephrologist of Ashkenazi parentage attended synagogue one morning and noticed that a Sephardi congregant with the same surname as his—Cohen—seemed to have completely different physical features (Kleiman 2001; Thomas et al. 1998; Skorecki et al. 1997). According to Jewish tradition, Cohens are descended from the same male ancestor, the priest Aaron, brother of Moses, and as such are regarded as the hereditary Jewish priestly caste, called upon first to come forth and read Torah in temple services. Dr. Skorecki reasoned that if the Kohanim (plural of Cohen) were indeed the descendants of only one man, they should have a common set of genetic markers and should perhaps preserve some family resemblance to each other. To test that hypothesis, he made contact with Professor Michael Hammer of the University of Arizona, a leading researcher in molecular genetics and pioneer in Y chromosome research and population history. The publication of their study in the prestigious British science journal *Nature* in 1997 sent shock waves through the worlds of science and religion. A particular marker (now known as the Cohanim Modal Haplotype, or CMH) did indeed appear in 98.5% of men bearing the surname Cohen (or a variation thereof such as Cone). It was apparently true that awareness of their priestly calling and descent from the Biblical Aaron had been strictly preserved for thousands of years (Skorecki et al. 1997; Thomas et al. 1998). Moreover, the data showed that there were very few “non-paternity events,” testimony as one Jewish scholar put it, to the faithfulness of thousands of Mrs. Cohens down through the ages (Kleiman 2001).

The first to test the new methodology in general surname research was Bryan Sykes, a molecular biologist at Oxford University (Sykes and Irven 2000). His study of the Sykes surname obtained valid results by looking at only four markers on the male chromosome. This use of a “minimal” haplotype pointed the way to genetics becoming a valuable assistant in the service of genealogy, anthropology and history. Sykes wrote the popular book *The Seven Daughters of Eve* and founded Oxford Ancestors, one of the first commercial DNA laboratories for genealogy purposes.

In 1999, Hammer led an effort that was successful in standardizing the phylogenetic nomenclature of lineages, or haplogroups (See Table 1). In 2000, N. Brent Kennedy became the first subject in the Melungeon DNA Project, initiated by co-author Elizabeth Hirschman at Rutgers.¹ Finally, DNA Consulting went on line in March of this year as the first genealogical service to offer historical and anthropological reports for DNA customers. Five years after the genetic tree of

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man came of age (Richards and Macauley 2001), anthropogenealogy was thus brought into the realm of what might be called “popular science.”

Y-STR and mtDNA Tests

“Genetics-and-genealogy” companies emphasize two main approaches. One determines the father’s lineage by testing short tandem repeat (STRs) of DNA material on sites of interest with names like DYS 390 and DYS 285a on the Y-chromosome. The other is a test of maternally inherited mitochondrial DNA that yields mutational motifs characteristic of different female lineages in the tree of humanity. Matches for any given Y chromosomal haplotype can be found in the Y-STR Haplotype Reference Database (YHRD), which now consists of 25,066 haplotypes in a worldwide set of 229 populations (release 13). Global mitochondrial mutations are reported in a concordance that resides at MITOMAP at Emory University, where they are compared with the Cambridge Reference Sequence.

Table 1
Conversion Table for Y-Chromosome Haplogroups.

	Job-ling 2000	Hammer (2001)	Race Archi ves	Karafet (2001)	Semino (2000)	Defining Mutation	FTDN A	Popular Label
R1b	1	1L	HG 1	42	Eu18	R-P25*	R1b	Aurignac, Western Atlantic
Q3	18	1G	--	41	Eu 22	Q-M3*	Q3	Native American
R1a1	3	1D	HG 3	43	Eu16, 18, 19	R-M17*	R1a	Eastern European
J	9	Med	HG 9	23, 24	Eu 9-10	J-12f2a, J- M172	J, J2, J*	Jewish, Semitic
E3a	8	5	HG 8	15	Eu 2	E-M2	E3a	Bantu
E3b	25	4	HG 21	14	Eu4	E-M2	E3b	North African, Moorish
I	--	--	HG 2	21	Eu7, Eu 8 ¹	I-P37-38, 40	I	Viking, Epi- Gravettan
C	10	V	HG 1, 22	16-19	Eu6	C3b=C- P39	C	Old Eurasian
G2	?	1H	?	22	Eu11	G-P15	G	No.Caucasian
K	23	1E, 1U	HG 12, 26	25	Eu15, Eu 16	SRY9138, P27, Tat	K1, K*	Central Asian
N3	12	1I	HG 16	26	Eu13, Eu 14	M178	N3*	NW Europe

Source: DNA Consulting, Santa Fe, N.M. ²

It is important to note that the STR and mtDNA tests offered by commercial firms can only test the so-called “outside” male and female lines, either the

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exclusive father-son heritage or exclusive mother-daughter. Although population geneticists use other methods, these have not become commercially feasible. They include human lymphocyte antigens (Guthrie 2001), polymorphic Alu insertions (Xiao 2002, Novick 1998), and ancient DNA. An economical test to determine the proportions of racial stock in a given DNA sample was developed by DNAPrint Genomics. However, not only are the categories (Caucasian, African, Asian, Native American) problematical but the methodologies for measuring them do not yet seem to be perfected.

The DNA toolkit also uses SNPs. A single nucleotide polymorphism (pronounced "snip") is a small genetic change, or variation, that can occur within a person's DNA sequence. The genetic code is specified by the four nucleotide "letters" A (adenine), C (cytosine), T (thymine), and G (guanine). SNP variation occurs when a single nucleotide, such as an A, replaces one of the other three nucleotide letters—C, G, or T. An example of a SNP is the alteration of the DNA segment AAGGTTA to ATGGTTA, where the second "A" in the first snippet is replaced with a "T." On average, SNPs occur in the human population only a little more than 1 percent of the time among the millions of genes. Because only about 3 to 5 percent of a person's DNA sequence codes for the production of proteins, most SNPs are found outside of "coding sequences." SNPs found within a coding sequence are of particular interest to researchers because they are more likely to alter the biological function of a protein.

Because of the recent advances in technology, coupled with the unique ability of these genetic variations to facilitate gene identification, there has been a recent flurry of SNP discovery and detection. Most haplotypes assigned by commercial testing companies such as Family Tree DNA, however, are determined by statistical inference and extrapolation, not by observation on a case-by-case basis, and some DNA cannot be haplotyped because of its rarity.

In Europe and most other regions of the world, surnames as well as property, titles, and socio-economic class are passed from father to son. This custom makes male haplotyping a valuable tool for determining recent migrations, political fortunes, intermarriage patterns and other historical relationships in a family. In Europe, most surnames were first adopted around 1,000 years ago, the same timeframe for the most recent common ancestor (MRCA) projected by matching haplotypes, so Y-STR haplotyping and surname research are ideal companions.

Mitochondrial haplotyping, on the other hand, derives its utility from the fact that it is often the mother who instills in children their culture, religion and education. In Native societies, the clan is usually passed through the mother. Since existing historical records as well as traditional genealogical researchers have, by default, emphasized male ancestors in their family history, mtDNA opens up exciting new vistas on the otherwise silent role played by women down through the generations.

Two Case Studies: Melungeons and Newberry

To illustrate the potential for haplotyping, let us look first at the Melungeon DNA Surname Project. The authors studied an ethnic group to which we both belong. Melungeons are a people who have been dwelling in the Appalachian Mountains of the southeastern United States for between 300 and 500 years. They have been the subject of intense speculation for at least three centuries (Gallegos 1997; Ball 1984; Bible 1975), though recent attention in the so-called Melungeon Movement has focused more on the history of discrimination against them in the South, together with medical conditions like familial Mediterranean fever (FMF) to which they are subject. Typically, they are described as having dark skin, black or dark-brown straight hair, brown or blue eyes and European features (Ball 1984; Bible 1975). Brent Kennedy's book (1997) renewed interest in investigation of the group's origins and stimulated an abundance of research.

Our biogenetics study supported what Kennedy had earlier proposed: The Melungeons were, in part, a Sephardic Jewish and Moorish community that began as early as 1540 with the De Soto expedition to what became the south-eastern United States (Hirschman 2004). Incoming Sephardic Jews and Moors, who had found refuge in such way stations as the Low Countries, Germany, France, Italy, Greece, and England after fleeing the Iberian Peninsula due to religious persecution, augmented the community over the centuries. (Hirschman's study called *The Melungeons: The Last Lost Tribe in America* is scheduled to appear from Mercer University Press this year).

We tested our thesis by obtaining Y chromosome DNA samples both from known persons of Melungeon roots in our own ancestry (who may or may not have actually emigrated from Scotland) and also from seven specific Scottish clans whose surnames are found among Melungeon populations. To do this, we contacted known relatives (Melungeon) and posted requests on Internet genealogical forums for male persons in a direct line of patrilineal descent from any one of the nine Scottish clans of interest. These were Alexander (associated with Macalister and Macdonald), Bruce, Campbell, Douglas, Forbes, Fraser, Gordon, Leslie, and Stewart/Stuart.

In every case where exact 12-marker matches could be obtained for the individual, the DNA modal center was found to be in Spain and Portugal or former colonies thereof (cf. Bosch et al. 2001; Carvajal-Carmona et al. 2000). Further, some of the Melungeon and Scottish clan donors had exact matches to living Jews; and *all* of the donors had one-off or two-off matches to present-day Jews. A parallel study of French Jews by Lavender (2003) came to comparable conclusions.

How could all these ethnically Jewish people be living in Scotland and no one know about it? The answers to these enigmas, put very simply, appear to be that, first, the Jews originally living in Scotland practiced an underground or secret form of their religion called crypto-Judaism, similar to the Crypto-Jews of New Mexico (Gitlitz 2002; Santos 2000); second, a minority of the descendants of these early Crypto-Jews *did* in some cases revert to the open practice of their faith upon arriving in the American colonies; and third, the majority of the descendants

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of these Jews are presently *unaware* of their ancestors' religious practices—in many cases because their faith was so well dissembled, in others because of conversion and assimilation. Jewish roots took a severe beating during the days of so-called Nativism in the U.S. and the Ku Klux Klan that followed. The 1920s saw many Melungeon families bury their heritage, so that today the Melungeons have been called a mystery even to themselves (Panther-Yates 2003).

A second case study will give readers a better idea of the technical and statistical side of haplotyping. Fig. 1 is a photograph of one of the author's mother-in-law Nina Jo Grimwood nee Newberry with her young son Ken (both now deceased). She looks very Nordic and Viking, doesn't she! We had a Newberry male cousin take the Y chromosome test, but found no exact matches, although Family Tree DNA did identify his haplotype as I, usually thought of as Viking. The family claimed to be English with Spanish or Portuguese and Tuscarora and Cherokee Indian, related to the Jewish Newberrys who owned a large department store chain in the South. Where did the Newberry haplotype come from?



Figure 1

Nina Jo Grimwood, Haplotype I on father's side (Newberry). *Family photo.*

No SNP test was done to determine what kind of I Newberry was. There were no matches at FTDNA, and only two exact matches in the Y-STR Db, Düsseldorf and Finland. But there were 85 matches, quite a large haplotype, if you changed the values at DYS 385ab from 12, 14 to 13, 14 (Figure 2). This is one of the fastest-mutating sites. Berlin had 17/548 (3% of the sample). Blecking Sweden had 6/40 (15%). Sweden as a whole had 44/401 (11%). Based on these matches, it was concluded that Newberry belonged to the most common form of I, I1a, similar to Locklear, another “Viking Indian” we tested.

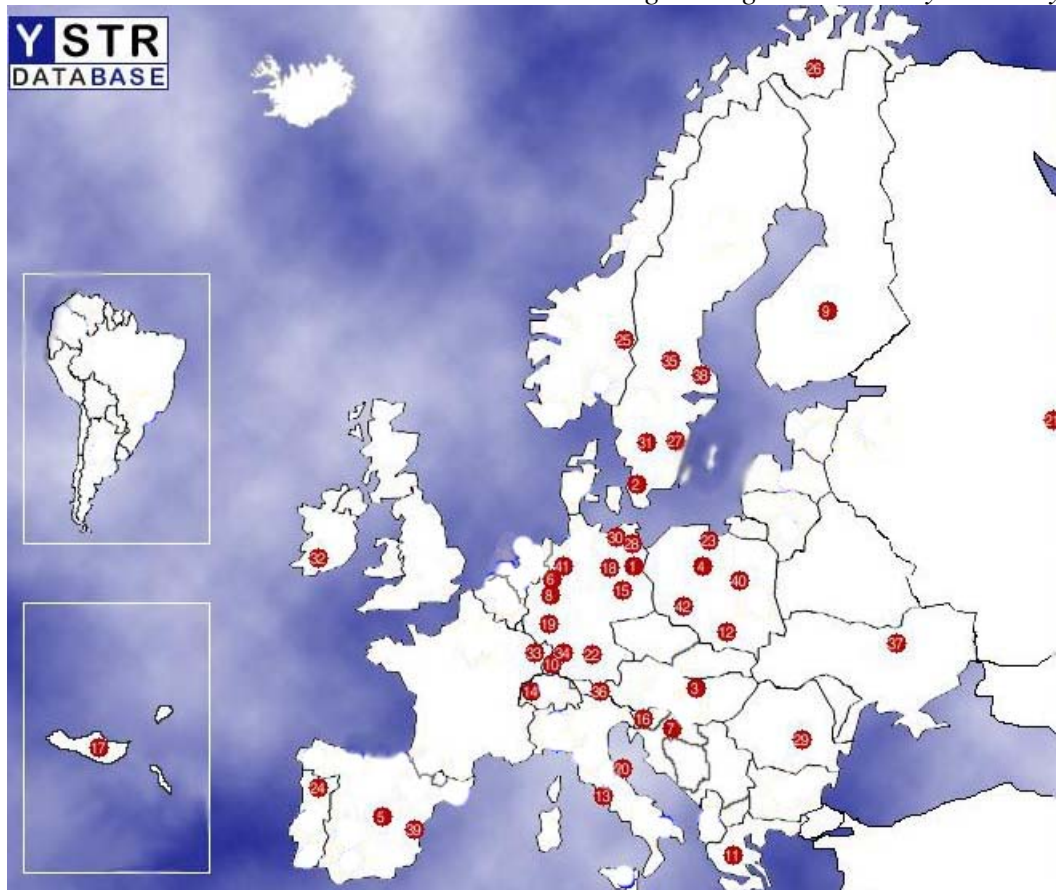


Figure 2
Newberry (I1a) close matches in Europe. *Y-STR Haplotype Reference Database, Berlin.*

Review of frequencies of haplogroup I in select European populations yielded the information that half the French sample from Low Normandy, the land of origin claimed by our Newberry descendants, was of the I1a variety, while there was a single Jewish match, lending credence, again, to family oral traditions of a Jewish heritage (Rootsi et al 2004). Detailed genealogical work by the test subject, Steven Newberry, suggested that the North Carolina Newberry line went back to the Viking warlord Bernard the Dane (de Harcourt, died about 955), through retainers of William the Conqueror, the earls of Warwick and the immigrant Thomas Newberry in colonial Massachusetts (see <http://www.indixie.com/genealogy/newberry>). Genetics thus confirmed orally transmitted genealogy, as the Melungeon study clarified and corroborated Appalachian families' stories of being both Scots-Irish and Jewish.

Steps to Mapping a Y Chromosome Lineage

A typical protocol for researching an individual's patrilineal anthropogenealogy can be summarized as follows:

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1. Verify the raw data for 12 target alleles from the original lab report source.
2. Control for laboratory-specific testing parameters, known validity and reliability issues, and nomenclature.
3. Perform a global search in the Y-STR Database for the haplotype.
 - a. Search for truncated or mutational matches if unsatisfactory yield.
 - b. Compute descriptive statistics for relevant countries.
 - c. Establish bivariate research theses.
 - d. Determine patterns, models, correlations, and other inferential statistics.
 - e. Control for fast-moving markers and different mutation rates and estimate most recent common ancestor.
 - f. Estimate risk factor of non-paternity events in line.
4. Compare search results with the other available sequence databases (Ybase, Sorenson, Family Tree DNA)
 - a. Repeat steps 3a.-3f, if necessary.
5. Search DNA Surname Projects.
 - a. Correlate branches or allonymic (other surname) genealogies.
 - b. Repeat steps 3a.-3f., if necessary.
6. Conduct chronological interactive search in relevant genealogy forums and e-mail discussion list threads at Rootsweb and other sites.
7. Search WorldCat for archival papers, special collection materials, and unique titles.
8. Check unpublished or password-protected databases, including:
 - a. Subscriber-based information at Ancestry.com
 - b. Melungeon DNA Surname Project (unpublished)
 - c. JewishGen and Daitch-Mokotoff Soundex System (need research code)
 - d. Scottish Clans (partially private)
 - e. Rabbinical genealogies (largely private)
 - f. Native American genealogies (usually requires e-mail correspondence with owners)
 - g. Human Genome Project (requires inputting correct gene sequence and glossary term)
 - h. PubMed at the National Library of Medicine (may require publisher's password)
 - i. Linkage literature and deep history of genotype (highly specialized).
9. Read any library articles or order any required extra materials such as interlibrary loan books.
10. Adjust inferences and retest research thesis.
11. Conduct general Internet keyword and natural phrase searches.

Mitochondrial searches involve a different protocol and references, leading to reconstruction of the deep history of the line since the last Ice Age rather than a medieval or modern timeframe (see MITOMAP, Cambridge Reference Sequence, Richards et al.).

Conclusion

As we emphasized, maternal and paternal lines are but two lines that can be measured by the current state of genetics. Each generation you go back, the number of your ancestors doubles, so that for a timeframe of 1500 CE (the beginning of European immigration to Americas), a person alive today would have, in theory, over 34,000,000 lines. In practice, however, populations go through bottlenecks and do not marry randomly, so the number is considerably lower. DNA tests cannot measure "crossover" patterns between the male and female line, only the "outside" male-male and female-female lineages.

In rare cases, your outside lines may not be the dominant ones in your genetic makeup. For instance, a British schoolteacher was found to have a distant sub-Saharan male progenitor, believed to have originated with an African slave in Roman Britain. And about 30 percent of American blacks have a Y chromosome that originated in Europe and is Caucasian. Many Native Americans could never "prove" their American Indian origins by DNA because the transmission of American Indian genes does not fall within the strictly male-male or female-female line.

This introduction to haplotyping cannot address the many theoretical considerations and scientific disputes regarding primers, mutation rates, and statistical inference. (For paternity rates, a good overview is Franklin [1998]). Independent convergence of gene types and mutations is thought to explain some similarities between Native American and Asian DNA; an excellent critique of the limitations of current methods and models in American population studies is a report by the The Bäu Institute (Jones 2002). Indeed, some critics of haplotyping believe large sub-populations such as the Western Atlantic Modal Haplotype, the most widespread haplotype in Europe and the Americas, reflect several converging types and do not even descend from a single recent common ancestor. Zerubavel (2003) among others has demonstrated that ethnicity is a social construct, even if it has a biological basis. Finally, some bioethical issues are raised by Brodwin (2002), Smith (1999), and Hall and du Gay (1996).

Anthropogenealogy holds considerable potential for supplementing traditional records-driven genealogy as well as for enhancing methods of anthropology and palaeontology. In both applications, it can overcome mute historical or archaeological witnesses to family origins, migrations, ethnic contributions and cultural diversity. For the purposes of anthropogenealogy, ethnicity is determined by haplotyping. While this article adopts a genetic definition of lineage, stock and race (and family), once anthropogenealogy matures as a discipline, the experience gained by laboratory haplotyping of large numbers of individuals can be applied to unravelling the complex diversity of families and population groups such as the Sephardic and Ashkenazic Jews, English, Scottish clans, indigenous peoples of the Americas and others. By revealing the interrelatedness of all forms of human genetic diversity around the world, we hope that anthropogenealogy can also be a potent force in fostering tolerance and peace among its peoples.

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continuous submission of Y-STR haplotypes by the [International Forensic Y-User Group](http://www.yhrd.org/index.html).” Available (July 2004) online at: <http://www.yhrd.org/index.html>.

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¹ Analysis of Kennedy’s DNA scores brought some surprising results. Not only did it validate his Melungeon family’s traditions of being French Jews who migrated through Scotland to Appalachia but it also suggested that his more famous Hyannisport namesakes arrived in Ireland from the same origins. Their American descendents were evidently of Sephardic Jewish ancestry from France, where their name was Canady. The original name was very likely Candiani—“from Candy,” the old name for the Turkish capital of Crete (now Iràklion). From the 14th century onward Crete was ruled by the Venetians, who imported sugar from Arabia and gave the world “candy.” The Ottoman Turks wrested it from the Venetians in 1669 after one of the longest sieges on record, an event that must have sent many “Candiani” refugees streaming westward. Another branch went to Ceylon after a generation’s sojourn in Portugal. Genealogies of John F. Kennedy, the U.S. president, do not go farther back than Patrick Kennedy, a prosperous farmer of Dunganstown, County Wexford, Ireland, who was born about 1785 and whose son emigrated to America (Burkes Peerage 1999). However, there is no reason to rule out a possible French origin before the family became Irish. Both Canady and Cassel—a sept of Clan Kennedy pointing to Casale in the Piedmontese region in southern France and northern Italy—appear on a list of refugee French Huguenots to Ireland (See <http://home.talkcity.com/DeckDr/bigjames7/irishhuganoautimigration.html>).

² Key: **FTDNA** = Family Tree DNA, Houston, Texas. **Hammer** (2001) = Hammer, M.F., Karafet, T.M., Redd, A.J., Jarjanazi, H., Santachiara-Benerecetti, S., Soodiyall, H., and Zegura, S.L. 2001. Hierarchical patterns of global human y-chromosome diversity. *Mol. Biol. Evol.* 18: 1189-1203. **Jobling** (2000) = Jobling, M.A. and Tyler-Smith, C. 2000. New uses for new haplotypes: the human Y chromosome, disease and selection. *Trends Genet.* 16: 356-362. **Karafet** (2001) = Karafet, T., Xu, L., Du, R., Wang, W., Feng, S., Wells, R.S., Redd, A.J., Zegura, S.L., and Hammer, M.F. 2001. Paternal population history of East Asia: Sources, patterns, and microevolutionary processes. *Am. J. Hum. Genet.* 69: 615-628. **Race Archives** = <http://www.racearchives.com/>. Based on Rosser et al. (2000), Y-chromosomal diversity in Europe is clinal and influenced primarily by geography, rather than by language, *Am J Hum Genet.* 2000 Dec;67(6):1526-43. **Semino** (2000) = Semino, O., Passarino, G., Oefner, P.J., Lin, A.A., Arbuzova, S., Beckman, L.E., De Benedictis, G., Francalacci, P., Kouvatsi, A., Limborska, S. 2000. The genetic legacy of paleolithic homo sapiens in extant Europeans: A Y chromosome perspective. *Science* 290: 1155-1159. **Su** = Su, B., Xiao, J., Underhill, P., Deka, R., Zhang, W., Akey, J., Huang, W., Shen, D., Lu, D., Luo, J. 1999. Y-Chromosome evidence for a northward migration of modern humans into Eastern Asia during the last Ice Age. *Am. J. Hum. Genet.* 65: 1718-1724.

The popular designations are often false; e.g., J is not always Jewish, and I is not synonymous with Vikings. R1b is included in P*. R1b2 is Semino’s Eu8. After Q, C is the second most common haplotype in Native American males (as much as 30% in certain populations).