

# A Reassessment of the Jewish DNA Evidence



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**The Jewish community has been the focus of extensive genetic study over the past decade in an attempt to better understand the origins of this group. In particular, those descended from Northwestern and Eastern European Jewish groups, known as “Ashkenazim,” have been the subject of numerous DNA studies examining both the Y chromosome and mitochondrial genetic evidence.**

**The focus of the present study is to analyze and reassess Ashkenazi results obtained by DNA researchers and synthesize them into a coherent picture of Jewish genetics, interweaving historical evidence in order to obtain a more accurate depiction of the complex genetic history of this group. Many of the DNA studies on Ashkenazim fail to adequately address the complexity of the genetic evidence, in particular, the significant genetic contribution of European and Central Asian peoples in the makeup of the contemporary Ashkenazi population. One important contribution to Ashkenazi DNA appears to have originated with the Khazars, an ancient people of probable Central Asian stock that lived in southern Russia during the 8th-12th centuries CE. Significant inflow of genes from European host populations over the centuries is also supported by the DNA evidence. The present study analyzes not only the Middle Eastern component of Ashkenazi ancestry, but also the genetic contribution from European and Central Asian sources that appear to have had an important impact on Ashkenazi ancestry.**

**Introduction**

The word “Jew” has a mosaic of meanings: it defines a follower of the Jewish faith, a person who has at least one Jewish parent, or a member of a particular ethnic group (“Jewish”). There are many Jews who do not practice Judaism as a religion but define themselves as “Jewish” by virtue of their family’s heritage and identification with the culture and history of the Jewish people.

Thus, Judaism is a mosaic of culture, religion, ethnicity, and for some, a way of life. It is an identity that is not quite a nationality, but neither is it a simple ethnic or cultural phenomenon either. This unusual combination of characteristics, coupled with Jewish resistance over the centuries to assimilation and strong adherence to their religious faith, has contributed to the intense feelings of curiosity, hatred, admiration, attraction and hostility by the rest of the world.

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Early on, the unique history of the Jews attracted DNA researchers who sought to solve the mystery of the origins of the Jewish people. Researchers had previously relied on linguistic, anthropological and archaeological evidence to try to address this question; genetic genealogical research has opened up a new area for researchers to explore.

One question the DNA studies sought to answer was whether the genetic ancestry of contemporary Jewish populations demonstrated, to any degree, their supposed descent from the ancient Israelites of the Middle East of three thousand years ago. Or rather, did the DNA evidence indicate that Jews were simply a people who came into being in Europe during the Diaspora years, being mainly comprised of those descended from European ancestors? Or, as some historical researchers suggested, did the DNA of Jews mainly reflect ancestry from the Khazars, an ancient tribal people with roots in both Central Asia and Russia who converted to Judaism in the 8th century?

This paper represents a new examination and reassessment of the Jewish DNA studies to date, presenting possible alternative explanations for the origins and distribution of certain genetic markers among Jewish populations, and in particular, among the group of Jews known as “Ashkenazim.”

Recent genetic research has greatly expanded our understanding of the probable origins and distinct geographic patterns of certain groups of people, including Jews. This recent research has superceded some of the earlier studies on Jewish DNA, allowing a reassessment of the theories of Jewish origins in light of this new research.

The new analysis shows that Jewish ancestry reflects a mosaic of genetic sources. While earlier studies focused on the Middle Eastern component of Jewish DNA, new research has revealed that

both Europeans and Central Asians also made significant genetic contributions to Jewish ancestry. Moreover, while the DNA studies have confirmed the close genetic interrelatedness of many Jewish communities, they have also confirmed what many suspected all along: Jews do not constitute a single group distinct from all others. Rather, modern Jews exhibit a diversity of genetic profiles, some reflective of their Semitic/Mediterranean ancestry, but others suggesting an origin in European and Central Asian groups. The blending of European, Semitic, Central Asian and Mediterranean heritage over the centuries has led to today's Jewish populations.

In examining Y chromosomal diversity in this review, two types of data are considered: Single Nucleotide Polymorphisms (SNPs), and Short Tandem Repeat Loci (STRs). STR markers are characterized by mutation rates much higher than those seen with SNPs. SNPs, on the other hand, are derived from rare nucleotide changes along the Y chromosome, so-called unique event polymorphisms (UEP). These UEPs represent a single historical mutational event, occurring only once in the course of human evolution. UEPs have been given a unified nomenclature system by the Y Chromosome Consortium (2002), resulting in the identification of each UEP with a particular haplogroup.

While I examine both types of Y chromosome data, I rely primarily on SNP data due to its increasing use by researchers as a tool in reconstructing the peopling of the world. Research on the diversity and geographic patterns of haplogroups have provided researchers with a greatly expanded understanding of prehistoric movements of people and a means of better understanding the present-day genetic variation among populations. Research with STR "haplotypes" is also occasionally discussed in this paper, particularly in light of its ability to demonstrate a high rate of endogamy, genetic drift, and founder effects among Jewish populations.

Examination of mitochondrial DNA, on the other hand, is based on the combined polymorphisms of the control region (hypervariable segments I and II, or HVSI and HVSII) along with specific SNPs in the coding regions of DNA found in the mitochondria. Both males and females have mtDNA, which they have inherited from their mothers, whereas Y chromosome DNA is found only in males and is inherited directly from their fathers.

Like the Y chromosome data, mtDNA sequences are sorted into major phylogenetic haplogroups as well. Recent analysis on both mtDNA and Y chromosome SNPs have allowed researchers to further divide many haplogroups into sub-branches, known in the DNA literature as "sub-clades." The geographic distribution of mtDNA haplogroups and their sub-clades also adds to our understanding of relationships of groups of people, including Jewish populations.

## **The Birth of European Judaism**

This section is intended to provide the reader with a brief history of the Jews in Europe as well as define terms used frequently in the Jewish DNA studies, such as “Diaspora,” “Sephardim,” and “Ashkenazim.” Furthermore, since Jews appear to have both Israelite/Middle Eastern and European genetic ancestry, an understanding of the Jewish experience in Europe is important in explaining how European ancestry became an integral part of the Jewish genetic makeup. However, this section is not intended to be an extensive recounting of the history of the Ashkenazi people.

The birth of European Judaism begins with the Diaspora. “Diaspora” is a term derived from the Greek work meaning “scattering.” While the word was originally used by ancient peoples to identify any group that was exiled or resettled from their homeland, the term has now become particularly associated with the Jewish exile from ancient Israel and resettlement elsewhere.

The Jews resettled in many distant lands, even as far as China. This work, however, focuses specifically on the Ashkenazi Jewish experience. Jews were subdivided into groups depending on where they resettled. Ashkenazi Jews are the Jews of France, Germany, and Eastern Europe. Sephardic Jews are the Jews of Spain, Portugal and North Africa. Mizrachi/Oriental Jews are the Jews of the Middle East. Certain Jewish communities do not fit into these distinctive groupings – in particular, the Falasha Jews of Ethiopia and the Chinese Jews.

Contemporary Jewry is comprised of approximately 13 million people, of whom 5.7 million live in the United States, 4.7 million live in Israel, and the remainder resides throughout the world (Ostrer 2001). Approximately 90% of the Jews of the U.S. are of Ashkenazi origin, while among the Jews of Israel, 47% are Ashkenazi, 30% are Sephardic, and 23% are of Mizrachi/Oriental origin (Ostrer 2001). Within Jewish groups, membership in three male castes (Cohen, Levi, and Israelite) is determined by paternal descent (Behar et al. 2003).

The history and genetic ancestry of Sephardic Jews is dealt with in only a cursory fashion here. There have been only very limited genetic studies on Jews of Sephardic descent, while in contrast, many DNA studies have explored the genetic ancestry of Ashkenazi Jews. Thus, the primary focus of this work is on Ashkenazim DNA results, but also included is a comparison of Sephardic and Ashkenazi results pertaining to Y chromosome haplogroups J and E.

The word “Ashkenazi” is derived from the Hebrew word for Germany, while “Sephardic” is derived from the Hebrew word for Spain. The word “Ashkenazi” was first used in medieval rabbinical literature to define western European Jews. An interesting story was related by author Arthur Koestler, who noted that the term “Ashkenaz” is also mentioned in the Hebrew bible, referring to a people living somewhere in the vicinity of Armenia. Probably for this reason, the Khazars, a people who lived in and around this area in ancient times and converted to Judaism in the 7th- 8th centuries, came to believe they were the descendants of these biblical people. Some scholars

argue that they began to call themselves “Ashkenazim” when they migrated to Poland in the 13th century. Eventually, perhaps, the term came to describe the community as a whole, not just the Khazarian immigrants (Koestler 1976, pp. 181-182).

While the Jews of today are connected historically and religiously to the Jews of ancient Israel, the DNA evidence also indicates that a significant amount of Jewish ancestry can be traced directly back to their Israelite/Middle Eastern ancestors. However, these ancestors represented a heterogeneous mix of Semitic and Mediterranean groups, even at their very beginnings.

The Israelite Kingdom arose in the 11th century BCE in an area between modern-day Lebanon, Jordan and Saudi Arabia. Current archaeological evidence indicates that the Israelite kingdom arose out of the earlier, Bronze Age Canaanite culture of that region, and displayed significant continuity with the Canaanites in culture, technology, language and ethnicity (Dever 2003, pp. 153-154).

While the Canaanites were a Western Semitic people indigenous to the area, they appear to have consisted of a diverse ethno-cultural mix from the earliest times. It is from this diverse group that the evolution of the Israelites occurred. Although little is known about these groups, they probably included some of the following populations:

1. Amorites: Western Semites like the Canaanites. They were probably the pastoral nomadic component of the Canaanite people.
2. Hittites: A non-Semitic people from Anatolia and Northern Syria.
3. Hurrians (Horites): A non-Semitic people who inhabited parts of Syria and Mesopotamia. Many kings of the early Canaanite city-states had Hurrian names.
4. Amalekites: Nomads from southern Transjordan. Even inimical references to this group in the Hebrew Bible “tacitly” acknowledge that the Israelites and Amalekites shared a common ancestry.
5. Philistines: Referred to in ancient texts as “Sea Peoples.” They invaded and settled along the coasts of ancient Canaan. Their culture appears to stem from that of Mycenae.

(Dever 2003, pp. 219-220).

While the Israelite kingdom clashed with a number of world powers over the centuries, including Egypt, Babylon, and Persia, it was the Romans who would destroy the Second Temple in 70 CE, violently sacking Jerusalem and scattering the Israelite population from their homeland. Many Jews were taken as slaves to Rome and its colonies (Konner 2003, p. 86). This watershed

moment in the history of the Jewish people is often considered by many researchers to represent the true beginnings of the Jewish Diaspora.

Ironically, however, many scholars believe the Ashkenazi population probably had its earliest roots in Rome, where Jews began to establish communities as early as the second century B.C. While some of these Jews were brought to Rome as slaves, others settled there voluntarily. There were as many as 50,000 Jews in and around Rome by the first century CE, most who were “poor, Greek-speaking foreigners” scorned for their poverty and slave status (Konner 2003, p. 86). Eventually, however, many of these slaves gained their freedom, continuing to live in and around Rome.

By the first century, however, the Jewish Diaspora had already spread to a number of regions of the world, many of which may have contributed to the make-up of the early Ashkenazi Jewish community. These include the Aegean Island of Delos, Ostia (a main port of Rome), Alexandria, and other places in Macedonia and Asia Minor (Konner 2003, p. 83). Jews also began to migrate north of the Alps, probably from Italy (Ostrer 2001).

By 600 CE, Jews were present in many parts of Europe, with small settlements in Germany, France and Spain. More to the east, there were also small Jewish settlements along the Black Sea, as well as larger communities in Greece and the Balkans (Konner 2003, p. 110).

By the 12th-13th centuries CE, Jews were expelled from many countries of Western Europe, but were granted charters to settle in Poland and Lithuania (Ostrer 2001). The Ashkenazi Jewish population expanded rapidly in Eastern Europe, growing from an estimated 15,000-25,000 people in the 13th-15th centuries, to two million by 1800 and eight million in 1939 (Ostrer 2001, Behar 2004b). Thus, Jewish settlement in Eastern Europe became the dominant culture of the European Jews, and then of most Jews throughout the world.

### **The DNA Evidence for Israelite Ancestry: The Jewish Priests and Cohanim DNA Study**

The search for Israelite/Middle Eastern DNA among contemporary Jewish populations properly begins with Dr. Karl Skorecki's landmark genetic study of the Cohanim, the priests of the Jewish religion. The study came about based on the following story:

Dr. Skorecki, a Cohen of Eastern European descent (Ashkenazim), was attending synagogue one morning. During the service, a Cohen of Sephardic descent from North Africa was reading from the Hebrew bible. According to Jewish tradition, all Cohanim (plural of “Cohan” or “Cohen”) are direct descendants of Aaron, the brother of Moses, and serve important priestly functions within the Jewish religion. The line of the Cohanim is patrilineal, allegedly being passed from father to son without interruption from Aaron, for 3,300 years, or more than 100 generations. Dr. Skorecki wondered if this claim could actually be tested. Could he find scientific evidence to support the

oral tradition of an ancient priestly lineage? Did he and the Sephardic Cohen possess a set of common genetic markers indicating they shared a common ancestor?

Dr. Skorecki, a nephrologist already involved in molecular genetic research, contacted Dr. Michael Hammer of the University of Arizona, a pioneer in Y chromosome research, and the Cohanim DNA study was born. Their findings clearly indicated that the Cohanim did indeed share a common ancestor. They discovered that a particular haplotype was found in 97 out of the 106 participants tested. This haplotype has come to be known as the “Cohen Modal Haplotype” or “CMH”. According to the study, calculations for dating the CMH yielded a time frame of 106 generations from the ancestral founder of the lineage – approximately 3,300 years ago (Thomas et al. 1998).

Not only did the genetic researchers corroborate the oral history of an ancient Jewish priestly caste, but they also confirmed the genetic link between both Sephardic and Ashkenazi populations, indicating that before the two populations separated, those who shared the CMH also shared common Israelite ancestry. Today, the CMH is considered not only the standard genetic signature of the priestly Cohanim, but also the yardstick by which all Jewish DNA is compared for determination of Israelite genetic ancestry. Thus, if a haplogroup is not shared by both Sephardim and Ashkenazim at a similar frequency, then it is generally not considered to be of Israelite origin.

Skorecki and Hammer reported that the CMH occurred within Y chromosome haplogroup J (Skorecki et al. 1997). We now know significantly more about haplogroup J than when these studies were originally published. Haplogroup J consists of an ancestral form (J\*) and two subgroups – J1 and J2. Although you can have the CMH in either J1 or J2, it is the genetic signature in J1 that is considered the Jewish priestly signature.

What is not widely reported is that only 48% of Ashkenazi Cohanim and 58% of Sephardic Cohanim have the J1 Cohen Modal Haplotype (Skorecki et al. 1997). So nearly half of the Ashkenazi Cohanim results are in haplogroups other than J1. Overall, J1 constitutes 14.6% of the Ashkenazim results and 11.9% of the Sephardic results (Semino et al. 2004). Nor is Cohanim status dependent on a finding of haplogroup J1.

Additionally, many other haplogroups among the Ashkenazim, and among the Cohanim in particular, appear to be of Israelite/Middle Eastern origin. According to Behar (2003), the Cohanim possess an unusually high frequency of haplogroup J in general, reported to comprise nearly 87% of the total Cohanim results. Among the Sephardim, the frequency of 75% is also notably high (Behar 2003). Both groups have dramatically lower percentages of other haplogroups, including haplogroup E. Given the high frequency of haplogroup J among Ashkenazi Cohanim, it appears that J2 may be only slightly less common than J1, perhaps indicating multiple J lineages among the priestly Cohanim dating back to the ancient Israelite kingdom.

However, J1 is the only haplogroup that researchers consider “Semitic” in origin because it is restricted almost completely to Middle Eastern populations, with a very low frequency in Italy and Greece as well (Semino et al. 2004). The group’s origins are thought to be in the southern Levant. Its presence among contemporary Sephardic and Ashkenazi populations indicates the preservation of Israelite Semitic ancestry, despite their long settlement in Europe and North Africa. Further, the CMH is considered the putative ancestral haplotype of haplogroup J1 (Di Giacomo et al. 2004).

Table 1 compares the Jewish J1 CMH to the J1 modal haplotypes of other Middle Eastern populations:

**Table 1**

**Modal Haplotypes\* in J1 Populations**

<b>J1 GROUPS</b>	<b>D Y S 0 1 9</b>	<b>D Y S 3 8 8</b>	<b>D Y S 3 9 0</b>	<b>D Y S 3 9 1</b>	<b>D Y S 3 9 2</b>	<b>D Y S 3 9 3</b>
CMH	14	16	23	10	11	12
Bedouin	14	15	23	10	11	13
Palestinian	14	17	22	11	11	13

\*6-Locus Haplotype.

Researchers believe that marker 388=17 is linked with the later expansion of Arabian tribes in the southern Levant and northern Africa (Di Giacomo et al. 2004). There were two migrations of J1, the first occurring in the Neolithic period, spreading J1 to Ethiopia and Europe (Semino et al. 2004). A second wave of J1 occurred in the 7th century, spread by Arab expansion from the southern Levant into North Africa. This secondary migration is also distinguished by a mutational event at marker YCAII—YCAIIa=22 and YCAIIb=22 (Semino et al. 2004).

The Cohanim study was widely misinterpreted by the public as indicating that all Jews were in haplogroup J and had the CMH. Furthermore, many non-Jews in haplogroup J mistakenly believed that they must have some Jewish ancestry hidden in their past to explain their DNA



results. As it turned out, most non-Jews were in subgroup J2 rather than J1 (Semino et al. 2004). Interestingly, Jews were later found to have as much J2 ancestry as J1.

The misinterpretation of the Cohanim results was damaging in some ways to the wider understanding of Jewish genetic ancestry. For example, one widely published media quote went like this: “This genetic research has clearly refuted the once-current libel that Ashkenazi Jews are not related to the ancient Hebrews, but are descendants of the Kuzar (sic) tribe – a pre-10th century Turko-Asian empire which reportedly converted *en masse* to Judaism.” Further, it was claimed that “[r]esearchers compared the DNA signature of the Ashkenazi Jews against those of Turkish-derived people, and found no correspondence” (Kleinman 1999).

However, it would soon become very clear that Jewish DNA was much more complicated than was presented by the media in their reporting of the Cohanim data. And Jewish Khazarian ancestry would come to the public’s attention yet again when another DNA study was conducted, this time on the Jewish priestly group known as the Levites.

### **The Khazars: A Jewish Kingdom in Europe**

Author Arthur Koestler (1976) is generally credited for bringing the unique history of the Khazars to the attention of the public. The decades that have past since the publication of his book have not dampened its highly controversial nature.

The country of the Khazars lay in the area between the Black and Caspian Seas, between the Caucasus Mountains and the Volga River. There, between the ever-invading Muslim Arabs and the Christian Byzantine Empire, a peculiar thing occurred – a Jewish empire arose. In 740 CE, the Khazarian King, his court and military ruling class all embraced the Jewish faith. This large scale official conversion of an ethnically non-Jewish people is well attested to in Arab, Byzantine, Russian and Hebrew sources (Koestler 1976, pp.13-15).

The rationale behind such conversion continues to both puzzle and fascinate historians – why would a people, despite political pressure from two great powers, chose a religion which had no support from any political power, but was rather persecuted by all? Whatever the reason, the Jewish Khazars continued to rule their kingdom until the 12th-13th century, when their empire finally dissolved. The fate of the Khazars after the fall of their empire remains a subject of great controversy among researchers.

The Khazars are often described as “a people of Turkish stock,” although such description is misleading (Koestler 1976, p. 13). Although the Khazars spoke a Turkish dialect believed to be related to that spoken today by the peoples of the Chuvash Soviet Republic, their ethnic origins remains a matter of debate. Many of the Eurasian tribes driven westward by the Chinese,

including the Huns, were labeled under the generic term of “Turk.” The origin of the word “Khazar” most likely derives from the Turkish root “gaz,” meaning “to wander” or simply “nomad.” (Koestler 1976, p. 21).

Given that the Khazarian kingdom arose in the area of today’s Ukraine, it is likely that there was a significant amount of indigenous Eastern European ancestry among this group. And, in fact, the various descriptions of the Khazars provided by ancient writers attest to the probable heterogeneous ethnic mixture in this group.

According to an 11th century Arab chronicler Ibn-al-Balkhi, the Khazars are

*... to the north of the inhabited earth towards the 7th clime, having over their heads the constellation of the Plough. Their land is cold and wet. Accordingly their complexions are white, their eyes blue, their hair flowing and predominately reddish, their bodies large and their natures cold. Their general aspect is wild” (Koestler 1976, p. 19). An Armenian writer described them as having “insolent, broad, lashless faces and long falling hair, like women. (Koestler 1976, p. 20).*

A slightly more flattering picture is provided by Arab geographer Istakhri:

*The Khazars do not resemble the Turks. They are black-haired, and are of two kinds, one called the Kara-Khazars [Black Khazars] who are swarthy verging on deep black as if they were kind of Indian, and a white kind [Ak-Khazars], who are strikingly handsome. (Koestler 1976, p. 20)*

However, Koestler (1976, p. 22) cautions the reader not to place too much weight on this description, since it was customary among Turkish peoples to refer to the ruling classes as “white” and the lower clans as “black.”

It is clear that the Khazars were closely connected to the Huns, who themselves are an ethnic mystery. The Byzantine rhetorician Priscus, who was part of an embassy to Attila the Hun’s court in 448 CE, reported that a people known as the “Akatzirs” or “White Khazars” were subjects of the Huns. According to Koestler (1976, p. 23), “Priscus’s chronicle confirms that the Khazars appeared on the European scene about the middle of the fifth century as a people under Hunnish sovereignty, and may be regarded, together with the Magyars and other tribes, as a later offspring of Attila’s horde.” After the collapse of the Hunnish Empire following Attila’s death, the confederation of tribes known as the Khazars eventually gained supremacy in the southern half of Eastern Europe, retaining control of this region for nearly four centuries.

What became a matter of dispute among historians was the fate of the Jewish Khazars after the destruction of their empire in the 12th- 13th centuries. Koestler argued that remnants of the Khazar tribes migrated into regions of Eastern Europe where the greatest concentrations of Jews were found, eventually merging with those pre-existing communities. In fact, Koestler’s

controversial argument was that the Khazars emigrated in substantial enough numbers to have had a significant genetic impact on contemporary Jewish ancestry.

With the advent of DNA studies, the question of whether contemporary Jews could trace any part of their ancestry back to the Khazars became a tantalizing mystery to try to solve. While the Cohanim DNA writers attempted to close the book on this question, evidence from another important genetic study, that of the Jewish Levite priests, made it apparent that the Khazarian debate was far from over.

### **The Levites: The DNA of the Jewish Khazarian Priests**

The other Jewish priestly caste is known as the “Levites.” Like the Cohanim, Levites are recorded in the Hebrew Bible as direct descendants of Aaron, Israel’s first High Priest. In fact, the Cohanim are actually a special subsection of the Levites (Telushkin 1997, p. 125).

In the second study published on the Cohanim, researchers reported that despite a priori expectations, Jews who identified themselves as Levites did not share a common set of markers with the Cohanim (Thomas et al. 1998). Unfortunately, the reporting that the Levites did not share a genetic signature from a common patrilineal ancestor with the Cohanim flew in the face of Jewish tradition. This led to some rather bizarre and disparaging explanations, like the following from Rabbi Yaakov Kleiman (1999) in *Jewish Action*:

*It is interesting to note that the tribe of Levi has a history of lack of quantity...After the Babylonian exile, the Levi'im (plural) failed to return en masse to Jerusalem, though urged by Ezra the Scribe to do so (They were therefore fined by losing their exclusive rights to maser.). Though statistically, the Levi'im should be more numerous than Cohanim, in synagogues today it is not unusual to have a minyan with a surplus of Cohanim, yet not one Levi.*

In point of fact, the Levites were shown to have a common set of genetic markers – just not the CMH. These markers were not even part of the same J1 haplogroup as found in the Cohanim. The majority of Levites shared a common haplotype, indicating a shared common ancestor among them, but this haplotype occurred within haplogroup R1a and, more specifically, within subgroup R1a1. Furthermore, this haplogroup was found only in the Ashkenazi Levites; it was not shared with the Sephardic Levite population in the same fashion as the CMH. Given the fact that the Ashkenazi Levites did not share R1a with their Sephardic counterparts, it appeared that this haplogroup had entered the Jewish population sometime during the Diaspora.

In one of the first studies to closely examine the high levels of R1a among Levites, researchers found that R1a1 formed a “tight cluster” within the Ashkenazi Levites (Behar et al. 2003). This

suggested to the researchers a very recent origin of this group from a single common ancestor (Behar et al. 2003).

In a subsequent Levite study, the modal haplotype reported for Ashkenazi R1a1, known as “H6,” was reported to occur twice as often as the second most common R1a1 haplotype among Ashkenazim, known as “H10” (Nebel et al. 2005). Out of a sample of 55 individuals, 25 had haplotype “H6” and 12 had haplotype “H10” (Nebel et al. 2005, Supplementary Material).

**Table 2**

**Haplotypes\* for Ashkenazi R-M17**

<b>HAPLOTYPE</b>	<b>D Y S 0 1 9</b>	<b>D Y S 3 8 8</b>	<b>D Y S 3 9 0</b>	<b>D Y S 3 9 1</b>	<b>D Y S 3 9 2</b>	<b>D Y S 3 9 3</b>
H6	16	12	25	10	11	13
H10	15	12	25	10	11	13

**\*6-Locus Haplotype**

Behar believed that among Ashkenazi Jews, R1a1 was essentially restricted to Levites. However, we know from subsequent research that R1a1 comprises nearly 12% of Ashkenazi results, while the Levites only make up about 4-5 % of the Jewish people (Nebel et al. 2005). Thus, these results extend well beyond the Levite priestly class to approximately 5-8% of the Cohanim and Israelites (the non-priestly Jewish population) as well.

Haplogroup R1a1 is relatively rare within Middle Eastern populations, but very common among Eastern European and Scandinavian populations (Behar et al. 2003). It is found at a frequency of 7% in some Near Eastern groups (Behar et al. 2004b). However, given that Sephardic groups did not share R1a1 frequencies with the Ashkenazim, it was apparent that Jewish R1a1 was probably not of ancient Israelite origin.

Confirmation of the high frequency of Haplogroup R1a1 among Ashkenazim as compared to other Jewish and non-Jewish Middle Eastern populations was found in a genetic study on Samaritan and Israeli groups (Shen et al. 2004). Although population samples were small, consisting of

twenty participants from Ashkenazi Jewish groups, all were Eastern Ashkenazim of Polish ancestry. Ashkenazi results were compared to other Jewish groups from Iraq, Libya, Morocco, Ethiopia and Yemen, as well as to non-Jewish Samaritan, Druze and Palestinian populations. Shen found that haplogroup R was found in 10-30% of all the groups, with the exception of Palestinians and Ethiopian Jews, though the majority belonged to R1b and R\*. In contrast, the Ashkenazim had the highest percentage of haplogroup R (30%), with two-thirds of those results found in haplogroup R1a (Shen et al. 2004).

As for when R1a1 first entered the Jewish community, Behar (2003) estimated a mean TMRCA (time to the most recent common ancestor) of 663 years before the present using the Simple Stepwise Mutation Model and a mean time of 1,000 years before present under the Linear Length-Dependent Stepwise Mutational Model. This calculation was striking because it fit precisely within the time period that Koestler believed the mass migration and absorption of the Khazars by the larger Eastern European Jewish communities occurred.

R1a1 is found in very high frequencies not only in the area of Eastern Europe where the Khazarian kingdom is reported to have existed, but also in many Central Asian populations as well, where some of the Khazarian population may have originated (Nebel et al. 2005). Furthermore, the most common Ashkenazi haplotype, H6, is identical to the most common haplotype found among European R1a1 (YHRD 2003). Ashkenazi H10 is identical to the fifth most common European R1a1 haplotype.[\[1\]](#)

Behar (2003) noted that Ashkenazi R1a1 haplotypes clustered closely with those seen in Sorbian and Belarusian groups in Eastern Europe, yet the haplotypes were dissimilar enough to convince him that these groups were not the original source population for Ashkenazi R1a1. While the Ashkenazi H6 haplotype is also one of the most common haplotypes among the Sorbian and Belarusian populations, the modal haplotypes found among these two Eastern European groups do not appear among Ashkenazim (Behar, 2003). However, it is possible that genetic drift could have led to the loss of other Jewish R1a1 lineages (Behar, 2003).

Nebel (2005) emphasized that the R1a1 haplogroup must have entered the Jewish gene pool from outside sources because the ancestral haplotype (H6) is almost completely absent in Sephardic Jews, Kurdish Jews and Palestinian population samples. He suggested that R1a1 in Ashkenazim "may represent vestiges of the mysterious Khazars." However, he also argued for a single founder event early on in the Jewish Diaspora, proposing that the TMRCA for R1a1 among Ashkenazi was approximately 62.7 generations ago, or 1567 years ago.

However, the proposal that R1a1 originated with a single founder event early in the Diaspora has become increasingly unlikely as research on Jewish DNA progresses. Since R1a1 is spread fairly evenly in haplotype distribution and frequency throughout the Ashkenazi populations from various

countries (Germany, Lithuania, Czechoslovakia, Hungary, Romania, Poland, Russia and the Ukraine), then the founders must have entered the community either before it expanded and spread to Eastern Europe, or merged separately into both eastern and western Ashkenazi groups. However, Nebel (2005) is forced to assert an extremely early TMRCA due to his belief that R1a1 must have originated with a single founder or very small group of founders. In order for R1a1 to reach its high frequency (12%) among the Ashkenazim from a single founder, a very early date must be proposed for the introgression of this haplogroup. Under this scenario, R1a1 entered the Jewish community when it was extremely small and in its formative stage. Gene flow from a single R1a founder at this early stage would likely have a huge impact on the expanding Ashkenazi population.

However, it appears that the most recently revised mutational dating techniques lend support to Behar's (2003) later date when applied to Jewish R1a1 haplotypes. If we assume that R1a1 entered the Jewish community around 1300 CE, then there would need to be enough founders to leave a 12% genetic impact on the population. Given that the Ashkenazi population at that time is estimated to be approximately 25,000 persons, it would be nearly impossible for a single founder to make such a significant genetic impact (Behar et al. 2004b). Adopting this conservative estimate of 25,000 persons, approximately two to three thousand R1a1 males probably entered the Ashkenazi community between the 12th-13th centuries.

Interestingly, there are no historical accounts of any large scale conversions or Eastern European groups entering the Jewish community at this time – except the Khazars.

Additionally, given the relatively late date of introgression and the large number of founders, these males must have already been very closely related to each other, sharing the R1a1 haplotypes that are later reflected in the Levite results. Behar (2003) noted that the lack of Levite R1a1 haplotype diversity suggested that all the founding lineages were very closely related to each other if, in fact, a large number of founding lineages contributed to the Levite R1a1 gene pool. The ancient reports on the Khazars indicate that the majority of the Jewish converts were from the Khazarian royalty and ruling classes (Koestler 1976, p.15). Although speculative, it seems likely this group would have intermarried heavily amongst itself, helping to preserve the group's elite status. Thus, it is probable that they would have already possessed a set of closely related R1a1 haplotypes which they simply passed on to their Levite descendants.

Most importantly, the fact that these R1a1 founders were endowed with Levite status is highly revealing. Behar (2003), in fact, argues against the possibility of a large number of R1a founders because it would involve a breach of "a well-regulated rabbinically controlled barrier" and would "most likely leave some prominent trace in the historical record – which it has not." However, he then suggests that the R1a introgression may indicate a lesser degree of stringency for the

assumption of Levite status than for the assumption of Cohen status. He points to a passage in the Talmud involving a debate over whether Levite status should be accorded to a man whose father was a non-Jew and whose mother was the daughter of a Levite. This suggests that assignment of Levite status other than through patrilineal descent could have been sanctioned by the rabbinical authorities.

However, the Khazars were already Jewish, having converted hundreds of years before. Although of a different ethnic make-up than the Ashkenazim of the 13th century, they were not “non-Jews.” They probably already had their own Levite caste in place who may have simply continued their priestly functions among the Ashkenazim.

Integration into the Levite priesthood would have secured for the Khazarian immigrants a place in their new community while helping them maintain a sense of elite status among a new people. Yet it is clear that the Khazars had become Jews long before they became part of the larger Ashkenazi community. Thus, it should not be surprising that six hundred years after their reported conversion, the Ashkenazim may have accorded them a special role among their Levite priesthood.

### **The Khazars and the Smoking Gun of Haplogroup Q**

With the discovery of haplogroup Q among Ashkenazi Jews, DNA researchers may have found the “smoking gun” of Khazarian ancestry.

In one of the few DNA studies to examine haplogroup Q among Jews, researchers made the surprising declaration that only 5-8% of the Ashkenazi gene pool is comprised of Y chromosomes that originated from non-Jewish European populations (Behar et al. 2004b). But since subsequent research has confirmed that R1a1 alone comprises nearly 12% of the Ashkenazi gene pool, it now appears that Behar’s estimate is much too low. Additionally, Behar’s (2004b, Supplementary Material) own data indicate that haplogroups R1b, R1a and I comprise more than a quarter of Ashkenazi DNA results.

As for haplogroup Q, Behar (2004b) states that it is a “minor founding lineage” among the Ashkenazim, but does not discuss it any further in the study. Haplogroup Q appears in 23 out of 442 Ashkenazi results in Behar’s study, or approximately 5% of the total results (Behar et al. 2004b, Supplementary Material). Interestingly, out of 50 non-Jewish Hungarian results also appearing in this study, haplogroup Q did not appear at all (Behar et al. 2004b, Supplementary Material).

The modal haplotype for Ashkenazi Q is shown in Table 3:

**Table 3**

## Ashkenazi Q-P36 Modal Haplotype\*

D Y S 0 1 9	D Y S 3 8 8	D Y S 3 8 9 i	D Y S 3 8 9 ii	D Y S 3 9 0	D Y S 3 9 1	D Y S 3 9 2	D Y S 3 9 3	D Y S 4 2 6	D Y S 4 3 9
13	12	13	16	22	10	15	13	12	16

### \*10-Locus Haplotype

Approximately 19 out of the 23 Q results exhibited the above haplotype, with 3 additional results being a single step mutation away on DYS marker #393 (Behar et al. 2004b, Supplementary Material). In fact, so many identical haplotypes makes it difficult to accurately date Ashkenazi Q, since using a TMRCA calculation indicates these Ashkenazim, both eastern and western groups, could be related within the last hundred years. This, however, seems highly unlikely, given the separation between these populations over the last few hundred years.

By designating Q a “minor founding lineage,” Behar (2004b) places this group among “those haplogroups likely to be present in the founding Ashkenazi population.” However, given that Haplogroup Q is rarely found in Middle Eastern populations in DNA studies, the likelihood that Q can be attributed to Israelite ancestry seems remote. The presence of Haplogroup Q among all Ashkenazi groups indicates the founders of this group either mixed with a number of separate Ashkenazi populations or, more likely, entered to the Ashkenazi population in western Europe in a similar fashion to Haplogroup R1a1, before the Ashkenazi migrated in large numbers eastward in the 13th-14th centuries.

The extremely low haplotype diversity of Ashkenazi Q supports the argument of a small number of closely-related founders merging with the Ashkenazim while they still resided primarily in Western Europe, but not significantly earlier in their formation, since a longer time span would result in more haplotype diversity. It does not support the contention that Q is Israelite in origin, or that the founders merged into the Jewish population much earlier in the Diaspora. Assuming the Ashkenazi population consisted of approximately 25,000 individuals around 1200-1300 CE, then approximately 1000-1500 Q individuals became part of the Ashkenazi population at that time.

Haplogroup Q is rare in European populations as well. It occurs in low percentages in Hungary (2.6%) and much higher percentages in Siberia (Tambets et al. 2004). It can be found among populations in Norway and the Shetland Islands of Scotland where many Norwegian Vikings settled. The frequency of Haplogroup Q among Scandinavians is comparable to that found in



Ashkenazim (Faux, private correspondence). It appears that Norwegians/Shetlanders and Ashkenazi Jews possess the highest percentages of haplogroup Q of any populations in Europe – a rare link between two very different populations who may share a common ancestor from Central Asia or Eastern Europe. Interestingly, Scandinavians and Shetlanders also possess high levels of haplogroup R1a1 as well, perhaps some of it originating from Central Asian sources (Faux, private correspondence).

David Faux, a researcher examining the Shetlander's DNA and possible Central Asian links, notes the following:

*The best evidence we have to date is that, although not investigated scientifically, that Q and K\* arrived with R1a from the same population source in the Altai region of Russian Siberia. It is likely that what we are seeing with Q and K are very rare Scandinavian haplogroups whose origins were long ago in Asia. If this is true, then it is very unusual that there does not seem to be any Q or K along the overland pathways to Norway (e.g., in Western Russia) – but there is Q, along with R1a, in the region of Kurdistan, and among a significant percentage of Ashkenazi Jews.*

Faux further hypothesized that the homeland of Norse Q lies somewhere in the populations of Siberia, such as with the Selkups (66.4% Q and 19.1% R1a) or the Kets (93.7% Q), or among the populations of the Altai mountain system extending through Mongolia, Kazakhstan and Russia (Tambets et al. 2004).

Haplogroup K\* also appears among Ashkenazim, though this group is rarely discussed in the DNA literature. Behar (2004b, Supplementary Information) found 2-3% among Ashkenazi Jews. Behar identifies this group as K\*-M9, though they may, in fact, be within Haplogroup K2, since they closely match the K2 haplotypes reported among Turkish groups (Cinnioglu 2004). The appearance of Haplogroup K\* only among eastern Ashkenazim may be attributable to Eastern European or Khazarian admixture (Behar 2004b, Supplementary Material). Interestingly, Ashkenazi K\* exhibits more haplotype diversity than haplogroup Q results, perhaps indicating a larger percentage of unrelated K\* founders or genetic drift.

However, Behar (2003) reports finding a significantly higher frequency of haplogroup K\* among Sephardic Levites (23%) and Sephardic Israelites (13%), perhaps the highest frequency of K\* found among any European population. This may indicate that some of Ashkenazi K\* is, in fact, of Israelite origin. Its absence among western Ashkenazim and very low frequency among eastern Ashkenazim suggests that the high frequency of Sephardic K\* may be due to pronounced genetic drift or significantly more K\* founders as part of the original Sephardic population. However, it is also possible that Sephardic K\* is the result of admixture with African or Mediterranean groups. Haplogroup K\* is known to reach a frequency of 10% in Cabo Verde, an east Atlantic island population with ties to Jewish founders from Spain and Portugal (Goncalves et al. 2003).

A comparison of haplogroup Q among Altaians and Ashkenazi Jews was undertaken by Dienekes Pontikos (2004), who operates a respected website dedicated to the examination of anthropological, archaeological and genetic research. He compared the frequency of haplogroups R1a and Q among Altaian Turkic speakers and Ashkenazi Jews. For Altaians, the percentages are 46/17, or a ratio of about 2.7, while in Ashkenazim it is 12/5, or a ratio of about 2.4. Dienekes writes:

*If Proto-Khazars were similar to present-day Altaians minus haplogroup C, then they would have a frequency of about 59% R1a and 22% Q. Therefore, it seems reasonable that an overall 5/22=22% of such Proto-Khazar elements into the Ashkenazi Jewish populations may be likely. But, the Khazars of Khazaria may themselves have been somewhat mixed with Western Eurasian elements, which would decrease their frequency of haplogroup Q.*

Dienekes (2004) also wrote that he found the continued silence of researchers about the presence of haplogroup Q among Ashkenazim “puzzling.”

Haplogroup Q is found in high frequencies in only a few regions of the world. Native American's possess very high percentages of Q, particularly a sub-group known as “Q3” (Zegura et al. 2004). But haplogroup Q did not originate among the Native Americans, nor did this population obtain their Q ancestry from Jewish or Scandinavian ancestors. As previously noted by Faux, its origins probably lie somewhere in northern Eurasia, in Siberia or the Altai, where Q continues to be a common Y chromosome haplogroup. It is from this group after migration to the New World that Native American Haplogroup Q3 originated.

Genetic analysis has allowed researchers to trace Native American haplogroup Q to its probable ancestral homeland – the Altai Mountains of Southwest Siberia (Zegura et al. 2004). The researchers have also pointed out that the Kets and Sekups, who currently inhabit the eastern part of Western Siberia and the Yenisey River Valley, can trace their origin homeland further south, on the slopes of the Altai mountains (Zegura et al. 2004). This region is, of course, where Faux postulated that Scandinavia's Q and K\* ancestors originated. It may also be the homeland of Khazarian Q ancestors whose descendants are found today among Ashkenazi Jewish groups.

In conclusion, it appears that some members of three very distinct populations—Scandinavian-Shetlanders, Native Americans and Ashkenazi Jews—may share common ancestors originating from the Altai regions of southern Siberia. However, the Q ancestors of the Native Americans appears to have departed from their Altai homeland much earlier than the other two groups, migrating to the New World sometime between 10,000 to 17,000 years ago, providing sufficient time for the Native Americans to develop their own unique subgroup of Q, known as Q3 (Zegura et al. 2004).

The migration of R1a and Q groups into Scandinavia is presently unknown, though Faux postulates a group from Central Asia may have moved up into Scandinavia sometime around 400 CE. Only a few hundred years later, the Khazars of southern Russia make their first appearance in the historical record. And it is to the Khazars, who undoubtedly possessed a high frequency of this haplogroup, to which the Jews most likely owe their unique Q ancestry.

### **Possible Other Israelite Y-Haplogroups: J, E and G**

Previously, the presence of Haplogroups J, E3b, and G among Jews was interpreted as additional evidence of Middle Eastern or Israelite ancestry in much the same fashion as the Cohanim Modal Haplotype. However, recent studies demonstrate that their origin is uncertain.

Unfortunately, misinformation about these haplogroups continues to pervade the public and media. Haplogroup E3b is often incorrectly described as “African,” leaving a misimpression regarding the origin and complex history of this haplogroup. Haplogroup J2, as previously discussed, is often incorrectly equated with J1 and described as “Jewish” or “Semitic,” despite the fact that it is present in a variety of non-Jewish Mediterranean and Northern European populations. And haplogroup G is rarely discussed in depth; its origin and distribution remain poorly understood.

#### *Haplogroup G Among Jews*

Lack of reported data regarding haplogroup G is surprising given that it is found in approximately 9% of Ashkenazi Jews, with G-M201\* consisting of the great majority of those results (Behar et al. 2004b, Supplementary Material). Behar (2004b) considers G-M201\* a “minor founder haplogroup” likely to have been present in the founding Ashkenazi population due to its very low frequency among non-Jewish Europeans. It is unclear whether Behar’s G-M201\* indicates G\* results rather than sub-group G1, though this seems unlikely given the lack of G\* reported in the Middle East and southern Europe (Cinnioglu et al. 2004). Haplogroup G-M201\* is distributed among both western and eastern Ashkenazi groups (Behar et al. 2004b, Supplementary Material). Unfortunately, so little has been reported about the distribution of this haplogroup among European and Middle Eastern populations that its origins among the Ashkenazim remain unclear. Haplogroup G-M201 is found at high frequencies among populations of the Caucasus and Georgia and may have originated in that region (Cinnioglu et al. 2004). The modal haplotype shown in Table 4 was found in 14 out of 34 Ashkenazi results, with an additional 5 results only a single-step mutation away:

#### **Table 4**

##### **Modal Haplotype\* of Ashkenazi G-M201\***

<b>D Y S O 1 9</b>	<b>D Y S 3 8 8</b>	<b>D Y S 3 8 9 i</b>	<b>D Y S 3 8 9 ii</b>	<b>D Y S 3 9 0</b>	<b>D Y S 3 9 1</b>	<b>D Y S 3 9 2</b>	<b>D Y S 3 9 3</b>	<b>D Y S 4 2 6</b>	<b>D Y S 4 3 9</b>
15	12	14	18	23	10	11	13	11	15

\*10-Locus Haplotype (Behar et al. 2004b, Supplementary Material)

Haplogroup G2 (G-P15) is present in both Jewish and non-Jewish European groups (Behar et al. 2004b). Although G2 is found in Turkey, it may be less common in Middle Eastern populations as compared to European groups. Haplogroup G2 appears almost exclusively in eastern Ashkenazim, comprising approximately 2% of the results (Behar et al. 2004b, Supplementary Material). The restriction to eastern Ashkenazim argues in favor of admixture with Eastern European or Khazarian ancestors. This group also exhibits high diversity and lack of a dominant modal haplotype, indicative of multiple founders or genetic drift.

### *Haplogroup E3b Among Jews*

An examination of recent DNA studies clarifies the probable origins and history of Haplogroup E3b among Jewish populations. One important study by Cruciani explores and refines the origins and distribution patterns not only of E3b, but of the entire E haplogroup (Cruciani et al. 2004).

Researchers discovered that various branches and sub-branches of haplogroup E had very different evolutionary histories and distinct migration patterns (Cruciani et al. 2004). Two branches, E1 and E2, are found predominately in Africa. The third branch, E3, is further divided into E3a and E3b. Haplogroup E3b can be further broken down into a number of sub-clades, including E-M78, E-M81, E-M123, E-M281, and E-V6. If an individual does not fall into any of these sub-clades but still has the defining mutations for E3b, he is then in the ancestral group, E-M35\* (Cruciani et al. 2004).

Although E3b arose in East Africa approximately 25,000 years ago, certain sub-clades appear to have been present in Europe and Asia for thousands of years (Cruciani et al. 2004). For example, although E-M78 occurs in about 30-20% of north and east African populations, it also occurs in 4.7% of French, 11.2% of Central Italians and 2.6% of Polish samples (Cruciani et al. 2004). It is

particularly high in the Balkans, with some population having a frequency of 25% or more (Cruciani et al. 2004).

It appears that E-M78 migrated from the Middle East to Europe during the Neolithic period. Once it reached the Balkans, a distinctive cluster formed which Cruciani (2004) refers to as the “alpha cluster.” The majority of European E-M78 appears to have originated from this cluster.

However, another cluster of E-M78, known as the “delta cluster,” appears to have migrated to Europe from North Africa or the Middle East with a distinctive haplotype already formed (Cruciani et al. 2004). It is found in low frequency among Spanish, French, Basque and Italian groups (Cruciani et al. 2004). In North Africa, it is also prevalent among Moroccan Arab, Berber and Egyptian groups. Among Middle Eastern groups, it is found in Turkish, Druze Arab and Palestinian populations (Cruciani et al. 2004). This cluster is distinguishable from the Balkan form by distinctive STR haplotype differences.

In a study that presented frequencies of haplogroups J and E among various groups, including both Ashkenazi and Sephardic populations, researchers found 14 out of 77 Ashkenazim (18.2%) were E3b, while 12 out of 40 Sephardim were E3b (30%). (Semino et al. 2004). Ashkenazim were also reported to have a frequency of 5.2% of E-M78, while Sephardim had 12.5%. Yet the prevalence of this sub-clade among Jews continues to remain unresolved. It is possible that Ashkenazi E-M78 is the result of multiple sources. Only further testing of E-M78 among Sephardic and Ashkenazi groups will determine which of Cruciani’s clusters Jewish groups belong to and whether Ashkenazi and Sephardic groups share similar E-M78 ancestry. However, the fact that Behar (2004b, Supplementary Material) found E-M78 to be much more prevalent among eastern versus western Ashkenazim (10 out of 12 results) argues in favor of admixture with Greek, Italian, Balkan or Eastern European populations. It is also possible that the origin of this sub-clade among Ashkenazim is attributable to Khazarian ancestors.

The higher frequency of E-M78 among Sephardic groups may be the result of pronounced genetic drift, or more likely, gene flow from North African and Spanish populations. The likelihood of European and North African gene flow is further supported by the fact that another sub-clade, E-M81, occurs only among Sephardim (Semino et al. 2004). It is also found in very high percentages among North Africans. Its frequency among the Sephardim at 5% is comparable to that seen in Spanish populations, again suggesting possible gene flow from Spanish and Berber populations into Sephardic groups.

Behar (2004b) deemed sub-clade E-M35\* a “major founding lineage” among Ashkenazim. But according to Semino (2004), E-M35\* only occurs among 1.3% of Ashkenazim and among 2.5% of Sephardim. Behar, on the other hand, reports finding E-35 at a frequency of 7.1% among Eastern European Ashkenazim, versus 19.1% among Ashkenazim in the west. Not only do Behar’s figures

contrast sharply with that found by Semino, but Behar also apparently discovered a significant difference in the frequency of this sub-group between eastern and western Jews. The discrepancy between Behar and Semino's results may be attributable to Behar including sub-clade E-M123 results within his larger E-M35 category. The fact that E-M123 does not appear separately as part of Behar's data suggests that he did, in fact, combine these sub-clades into a single category.

In fact, the best candidate for possible E3b Israelite ancestry among Jews is E-M123. This sub-clade occurs in almost the same proportions (approximately 10-12%) among both Ashkenazim and Sephardim (Semino et al. 2004). According to Cruciani (2004), E-M123 probably originated in the Middle East, since it is found in a large majority of the populations from that area, and then back migrated to Ethiopia. He further notes that this sub-clade may have been spread to Europe during the Neolithic agricultural expansion out of the Middle East. However, because E-M123 is also found in low percentages (1-3%) in many southern European and Balkan populations, its origin among Jewish groups remains uncertain (Semino et al. 2004). Yet the fact that both Sephardim and Ashkenazim possess this sub-clade in similar high frequency supports an Israelite/Middle Eastern origin.

As for E-M35\*, Semino (2004) did not find this group in either the Lebanese or Iraqi samples. Nor did Cruciani (2004) find it in any of his Middle Eastern samples. It is present, however, in East and North African samples; for example, it occurs in about 7.9% of Berber tribesmen from north-central Morocco (Semino et al. 2004). It also occurs in 2.7% of Andalusians in Spain, 5.5% of Sardinians and 1.5% Italian populations (Semino et al. 2004). It appears that the most likely explanation for Jewish E-M35\* is that it represents gene flow from North African populations into Spain, Italy, and Sardinia, and hence, gene flow from these European populations into Jewish groups.

### *Haplogroup J2 Among Jews*

Haplogroup J2 among Jews has been erroneously interpreted in the past as exclusively "Israelite" or "Middle Eastern" in origin. Among Ashkenazim, J2 occurs among 23.2% of the population, while Sephardim have 28.6% (Semino et al. 2004). While these percentages are nearly identical to Iraqi (22.4%) and Lebanese (25%) groups, they are also comparable to Greek (20.6%), Georgian (26.7%), Albanian (19.6%), Italian (20-29%), and to a lesser extent, French Basque (13.6%) populations (Semino et al. 2004).

Although J2 is a close cousin to J1, it is characterized by the M172 mutation, while J1 is characterized by the M267 mutation. These two branches of haplogroup J formed in neighboring but different regions of the world. The ancestral J group (J\*) is very rare and has only been observed in small numbers in the Balkans, Crete, Greece, and Oman (Di Giacomo et al. 2004). A

recent DNA study on Turkish populations also discovered a very low frequency of J\* (Cinnioglu et al. 2004).

One of the first DNA studies exploring haplogroup J among Jewish groups found the following:

*The investigation of the genetic relationship among three Jewish communities revealed that Kurdish and Sephardic Jews were indistinguishable from one another, whereas both differed slightly, yet significantly, from Ashkenazi Jews. The differences among Ashkenazim may be a result of low-level gene flow from European populations and /or genetic drift during isolation...Jews were found to be more closely related to groups in the north of the Fertile Crescent (Kurds, Turks, and Armenians) than to their Arab neighbors. (Nebel et al. 2001)*

According to the researchers, J1 originated in the southern part of the Middle East while J2 originated in the northern part (Nebel et al. 2001). Because Jewish populations possess approximately twice as much J2 as they do J1, their ancestry more closely matches that of Turkish and Transcaucasian populations. This may indicate that some of the genetic ancestry of the ancient Israelites may have closely resembled groups living in the Caucasus and the northern Levant rather than groups from the southern Levant. Additionally, it may also indicate that there were multiple waves of J1 migrating northward into the Middle East, some after the Jewish Diaspora. This is supported by the findings of Di Giacomo (2004) regarding a secondary expansion of haplogroup J1 out of the southern Levant and North Africa with Arabian tribes.

According to Di Giacomo's (2004) study, the high diversity of haplogroup J2 in Turkish and southern European populations suggests that this branch of haplogroup J originated around the Aegean, not the Middle East. Additionally, it appears that much of J2 was confined to the coastal Mediterranean areas, indicating that maritime trade, rather than earlier Neolithic agricultural expansions, may have helped spread J2 throughout the Mediterranean world.

This conclusion, however, contradicts an earlier study in which the researchers argued that certain elements of Neolithic material culture – painted pottery and figurines in particular – emanating out of the northern Levant and Anatolia during the Neolithic could be correlated with the distribution of certain Y haplogroups, including haplogroup J (Underhill and King 2002). In fact, the authors of that study concluded that the "Eu 9 (Haplogroup J2) haplogroup is the best genetic predictor of the appearance of Neolithic painted pottery and figurines at various European sites," first spreading from the regions of Anatolia and the Levant into the Balkans, Greece and the Danube basin, then subsequently into the rest of Europe.

Di Giacomo's (2004) study emphasized that J2 is "Mediterranean" or "Aegean" rather than "Semitic" in character. It is found predominately in northern Mediterranean and Turkish populations, differentiating the Aegean area from the Middle East in its haplogroup J results.

Going further, the researchers maintained that certain sub-clades of J2 appear to have originated well after the beginning of the Neolithic revolution and around the Aegean, spreading out to the rest of Europe during the expansion of the Greek world.

It is this final idea – that much of J2 is European in origin rather than Middle Eastern – that complicates the interpretation of Jewish J2 results. Sub-clade J-M102\* originated in the southern part of the Balkans and is generally absent in Middle Eastern populations (Semino et al. 2004). Ashkenazim have a 1.2% frequency of J-M102 and Sephardim have 2.4%. These results argue in favor of European gene flow into the Jewish community.

Three other sub-clades appear in Jewish populations and invite further examination of their origins. Sub-clade J-M92\* appears only in Ashkenazi populations at a frequency of 4.9%. The fact that it is absent in Sephardim indicates that the origin of this group among Ashkenazim may be attributed to European gene flow. While J-M92\* appears in small percentages among Iraqi (1.3%) and Lebanese (2.5%) groups, it occurs in higher frequencies and is much more diversified in Turkish, Balkan and Italian populations (Semino et al. 2004).

Sub-clade J-M67\* presents an equally complex picture among Jewish populations. Ashkenazi Jews have 4.9% and Sephardim have 2.4% (Semino et al. 2004). Again, J-67\* is present among populations in the northern Levant (Iraqis have 4.5% and Lebanese have 2.5%), but frequency and variance is significantly greater in Europe and Turkey than in the Middle East (Semino et al. 2004). Thus, whether Jews obtained their J-M67\* ancestry from Israelite, European, or a mixture of ancestors remains unknown at this point in time.

Semino (2004) reports the following regarding the origins of J-M67\* and J-M92\*:

*...J-M67\* and J-M92 could have arrived in Europe from Anatolia via the Bosphorus isthmus, as well as by seafaring Neolithic populations who reached southern Italy. J-M67\* and J-M92 could represent, at least in part, the Y-chromosome component that King and Underhill (2002) found to correlate with the distribution, from Anatolia toward Europe, of archaeological painted pottery and anthropomorphic figurines .*

Thus, Semino has expertly merged the findings of both Di Giacomo and King/Underhill regarding the origin and expansions of J2 (Neolithic versus Post-Neolithic Aegean/Greek) into a cohesive interpretation regarding the multiple migrations of J2 throughout the Mediterranean world.

The final sub-clade of J2 found among Jews is J-M172\*. While 12.2% of Ashkenazim are in this sub-clade, Sephardim have a frequency nearly twice as high (Semino et al. 2004). This sub-clade appears in high percentages among Lebanese and Iraqi populations (20% and 10.2%, respectively) and its presence in this region can probably be attributed to J-M172\* migrations out of Anatolia



into the northern areas of the Levant (Semino et al. 2004). J-M172\* is also found in a number of European populations, particularly among French Basque and Italian groups. Thus, its origin among Jewish populations remains unclear, though its absence among Spanish populations, but presence in Sephardic groups, supports the theory that at least some of Jewish J-M172\* may be of Israelite origin. Behar (2004b) also acknowledges that J-M172\* among the Ashkenazim may have originated with multiple ancestral sources.

### **European Admixture Among the Ashkenazi**

Although there has been strenuous opposition to intermarriage with non-Jews since biblical times, including biblical prohibitions, bans, warnings, rules and laws- law is one thing, practice often another.

It should be stressed that it was not only the Jewish communities that opposed such intermarriage. According to author Raphael Patai, the Christian authorities in Europe outlawed not only "Christian-Jewish sexual relations but also all kinds of social contact between members of the two religions, and backed up their injunctions with generally severe penalties, including the death penalty, imposed on both the Jewish and Christian partners to the crime. However, the very frequency and repetitiousness of the promulgation of such laws are ... indications of their ineffectiveness" (Patai 1989, p. 105). Unfortunately, we do not have an accurate picture of the frequency of such sexual contact between Jews and Christians, since only those relatively few cases which led to criminal prosecution are known. However, Patai believes the number was significantly higher than that reported by the authorities.

Such prohibitions did not prevent such sexual contact among Christians and Jews; nor did it prevent Christians from converting to Judaism, individually and in groups, though it was probably much more common for Jews to convert or simply leave the Jewish community, given the significant oppression they faced in Europe. The word "proselyte" originally designated a Greek person who had converted to Judaism, indicating that conversion among Greek populations must have been common enough at one time to have led to the creation of this descriptive word.

Frankly, the fact that Jews have substantial European ancestry is obvious to most onlookers – many Jews *look* like Europeans. The question for DNA researchers was: How much of that European appearance actually translates into European genetic ancestry?

Patai (1989, pp. 16-17) argues that the Jews had never lived in sufficient reproductive isolation to have developed distinctive genetic features. Rather, he states that "all the available evidence indicates that throughout their history the Jews continually received an inflow of genes from neighboring populations as a result of proselytism, intermarriage, rape, the birth of illegitimate children fathered by Gentiles, and so on." In addition, the ancient Israelites themselves were

formed from a heterogeneous mix of tribal and ethnic groups, both Semite and non-Semitic in origin. Thus, heterogeneity was there from the very beginning.

Behar (2004b) argues for an extremely low admixture rate of 8.1% to 11.4% among the Y chromosome results. He further reduces this figure to an unlikely 5% if the Jewish Dutch results are excluded due to suspected high admixture rates. However, Behar's own reported R1b (R-P25), R1a (R-M17) and I (I-P19) haplogroup frequencies indicate that these groups comprise approximately one-quarter to one-third of the Ashkenazi Y chromosomes. Furthermore, Behar acknowledges that these haplogroups are probably indicative of European admixture with Ashkenazi populations.

According to the findings of Behar (2004b, Supplementary Material), R1b comprises 44 out of 442 results, or nearly 10% of Ashkenazi results. Additionally, Behar (2004b) reports that the highly-admixed Dutch Jews have 26.1% R1b results. Haplogroup I (I-P19) comprises 18 out of 442 results, or approximately 4% of the Ashkenazi results. Thus, haplogroups R1b and I among Ashkenazi Jews comprise almost 15% of the DNA results.

Patai (1989, p. 41) provides an example of the cumulative effects of admixture within the Ashkenazi population:

*Let us assume that there was a Jewish community somewhere in the Rhineland which in the year [CE] 800 numbered 100 souls, and that it maintained the same number until [CE] 1600. If, in this community, one case of interbreeding occurred once every ten years, then, after 100 years, there were in it 95 per cent Jewish and 5 per cent Gentile genes; after 200 years, the ratio was 90.5 to 9.5; after 400 years, 82 to 18; and after 800 years, 67.1 to 32.9. In other words, after 800 years about one-third of the genes of the community would be of Gentile origin.*

There are clearly some problems with Patai's hypothetical scenario. It is unlikely, for instance, that the Ashkenazi population size remained completely static during an eight hundred year period. However, it is clear that the Jewish population grew very slowly during this time period and that the huge Ashkenazi population explosion did not happen until after 1300 CE. Ashkenazi population size remained much reduced for generations due to a history of dispersal, genetic bottlenecks and a high rate of endogamy. Further, it is unlikely that there was a constant rate of gene flow from European groups into the Ashkenazi population. Rather, such introgression probably occurred at an irregular rate, with occasional large groups like the Khazars integrating into the Jewish community and adding their genetic legacy to the already diverse gene pool of the Ashkenazim.

Patai's ultimate conclusion regarding admixture is particularly intriguing given the lack of DNA data available when he wrote his book. He relied heavily on other genetic data, including blood

groups, fingerprint patterns, and genetic diseases, to reach his conclusions. Despite these limitations, Patai (1989, p. 294) concluded that while Jewish populations retain evidence of their Mediterranean and Middle Eastern origins, they have clearly experienced extensive admixture with their European neighbors. He cites various authors, including Cavalli-Sforza and Carmelli, who estimate such admixture rates to be approximately 40% for Ashkenazi Jews.

## **Jewish mtDNA Results**

### *A Few Founding Mothers*

Jewish maternally inherited mitochondrial DNA (mtDNA) results are examined in depth in only two published DNA studies. In the first study, researchers examined nine different Jewish groups and compared their mtDNA to eight non-Jewish groups as well as an Israeli Arab/Palestinian population (Thomas et al. 2002).

Thomas discovered a common characteristic to almost all Jewish mtDNA – the high frequency of particular mtDNA haplotypes within the Jewish populations. In addition, Jewish mtDNA results displayed significantly lower diversity than any non-Jewish population tested as part of the study, yet was also characterized by greater differentiation between the Jewish groups as well as their hosts.

These unusual results suggested to the researchers that an extreme female-specific founder effect had occurred in the genetic histories of most Jewish populations. The founder effects had, in fact, been so severe that mtDNA frequencies in Jewish groups differed significantly from those seen in any of the non-Jewish populations.

As to the origin of these maternal founders, Thomas (2002) acknowledged that “in many cases, it is not possible to infer the geographic origin of the founding mtDNAs within the different Jewish groups with any confidence.” One thing, however, was clear to the researchers: the Jewish groups formed independently from each other around a small group of maternal founders. In other words, many of the Jewish groups did not share the same female ancestors. Furthermore, it appeared to Thomas that the founding of these maternal lineages occurred “immediately after the establishment of the communities or over a longer period of time.” Since haplogroup diversity was so low, female-specific gene flow from the surrounding non-Jewish community must have been limited once the original community was established.

Finally, Thomas (2002) noted that although Ashkenazi Jews were commonly believed to have suffered a sharp founder effect, the group had a modal haplotype frequency similar to their non-Jewish host populations (9% vs. 6.9%). While this could be evidence that no such founder events had occurred in this population, it could also indicate “that present-day Ashkenazic Jews may

represent a mosaic group that is descended on the maternal side from several independent founding events.”

In the second Ashkenazi mtDNA study, Behar (2004a) attempted to answer the question of founder events among Ashkenazim posed by Thomas. Unfortunately, it could be argued that this entire study is directed at convincing the reader that “Ashkenazi populations as a whole are genetically more similar to Near Eastern non-Jewish populations than to European non-Jewish populations.”

In order to prove this, a complex analysis regarding “mismatch distributions” between Jewish and non-Jewish populations is performed. A careful reading, however, indicates that these mismatch calculations are based on a number of unfounded assumptions, including a shared common history of Pleistocene population growth between Jewish and Middle Eastern groups. However, since only a small percentage (10% - 20%) of the Jewish mtDNA is definitively stated to be of Middle Eastern origin in the study, calculations based on this assumption are questionable (Behar et al. 2004a).

Behar (2004a) attributes the obvious peculiarity of Ashkenazi mtDNA, namely reduced mtDNA diversity coupled with usually high frequencies of particular mtDNA haplotypes, to strong genetic drift rather than to independent founder events. Furthermore, Behar suggests the unusual Ashkenazi mtDNA results are due to a Jewish population bottleneck that occurred in the Near East. According to the study,

*[o]ur computer simulations confirm that the frequencies of the zero and one class of the Ashkenazi mismatch distribution are significantly elevated over that observed for the sequences sampled from Near Eastern populations. This is a strong indication of a recent population bottleneck and further simulations suggest the data best fit a 200-fold reduction in size approximately 150 generations ago.*

Behar (2004a) acknowledges that the rationale for such a bottleneck can be sustained only if supported by two major assumptions: “the Ashkenazim have not admixed with European host populations and that the mutation rate is  $1.2 \times 10^{-3}$  per sequence per generation.” However, postulating no admixture between Jewish and non-Jewish European host populations is both historically and scientifically untenable, particularly in light of Behar’s own Y chromosome results indicating extensive admixture.

A close inspection of Jewish mtDNA results refutes any argument for lack of maternal admixture with European populations. According to Behar (2004a), only four mtDNA groups account for approximately 70% of Ashkenazi mtDNA results. These haplogroups are K (32%), H (21%), N1b (10%) and J1 (7%). However, Behar indicates the origins of three out the four groups (H, K and J) are unknown. More importantly, he acknowledges that certain other haplogroups among the

Ashkenazi – V and U5 in particular – appear to be of European origin, thereby negating altogether the assumption of no admixture. Finally, the slow mutational changes that occur within mtDNA are unlikely to be strongly influenced by population isolation and genetic drift occurring over a very short time span, as is the case with the Jewish Diaspora. Thus, there is a much greater probability that independent founder events occurring during the Jewish Diaspora rather than genetic drift are the cause of Jewish mtDNA variability and lower haplogroup diversity. However, it is also possible that both factors had an effect on Jewish mtDNA.

The origin of Jewish mtDNA Haplogroup K is unclear at this time. The most common haplotypes, as distinguished by HVR1 mutations, are as follows: 223T-224C-234T-311C (33%); 224T-234C-311C (24%); 093C-224C-311C (19%); and 224C-311C (16%). The first two haplotypes are almost completely restricted to Ashkenazi populations, perhaps an indicator of pronounced genetic drift (Behar et al. 2004a). Shen (2004) found that the majority of Ashkenazi K lineages also shared transitions at nucleotide positions 11470 and 11914, which are specific to clade K1a. Except for the Ashkenazi, this particular K1a motif has only been reported in one Palestinian, one Romanian, one Czech, and one Basque (Shen, et al. 2004). Because of their near absence in non-Jewish populations, the most common Ashkenazi K1a haplotypes can be used as indicators of Ashkenazi ancestry.

Behar (2004a) noted that mtDNA haplogroup N1b exhibits a significant lack of haplotype diversity, indicating a probable common ancestral origin for this group. Additionally, Ashkenazim results display only a single transition from the putative ancestral HVR1 haplotype (145A-176G-223T) which Behar (2004a) reports is almost completely restricted to Middle Eastern populations. The inference that N1b is of Israelite origin is further supported by the fact that this group appears to be spread throughout eastern and western Ashkenazim at almost equal frequencies (Behar et al. 2004a, Supplementary Material).

Behar (2004a) does state that certain other haplogroups – L2, pre-HV, U7, M1, and U1b- which appear at very low frequencies among Ashkenazim, may have either a Middle Eastern, African, or Mediterranean origin. Unfortunately, this does little to clarify the probable origins of these groups among Ashkenazim.

The haplogroups that comprise the remaining 30% of Ashkenazim mtDNA including the following: J (J\*, J1, J2), T (T\*, T1-T5), HV1, U6 (U6a\*, U6a1, U6b), HV\*, W, X, I, M\*, U4, U1a/U1b, U2/U2e, U3, R (R\*, R1, R2). Behar (2004a) lists their provenance as unknown. However, a close examination of mtDNA haplogroups J1 and J2, which comprise 7% of Ashkenazi results, reveal that they are common only among Eastern Ashkenazim (Behar et al. 2004a, Supplementary Material).

Therefore, Ashkenazi mtDNA J can probably be attributed to Eastern European admixture. In fact,

Shen (2004) notes that Ashkenazi J1 and T2b haplotypes have exact HVS1 matches with European groups, suggesting admixture.

Although it may initially appear that Ashkenazi mtDNA groups such as HV\* and HV1 are Middle Eastern/Israelite in origin, the fact that both mtDNA groups are found almost exclusively among Eastern European Jews points to admixture as a more likely source of this ancestry. On the other hand, pre-HV1 and L2a are found in low frequency among both eastern and western groups and are more likely to be of Israelite origin (Behar et al. 2004, Supplementary Material).

Haplogroup U among Ashkenazim comprises 32 out of 565 results, with U7 comprising 8 out of the 32 results (Behar et al. 2004, Supplementary Material). In a study on mtDNA in the Volga-Ural region, researchers found U7 to be typical of Middle Eastern populations, including Jordan, Kuwait, Iran and Saudi Arabia (Bermisheva et al. 2002). This lends support to Behar's theory that U7 among Ashkenazi Jews is of probable Middle Eastern origin. Shen (2004), however, is less certain about its origins, stating that "it is difficult to assess whether Haplogroups U7 and HV, as well as HVS-I haplotypes of the Ashkenazi K2, I, W, and U2 lineage, represent the original gene pool of the Jewish founders or are due to admixture with European populations."

U2 among Ashkenazim appears to be of European origin, since the common haplotype resembles that seen in European populations (HVR1 motif 051G, 129C, 189C) (Behar et al. 2004a, Supplementary Material). Although Behar (2004a, Supplementary Material) suggested that Ashkenazi U1b was "Middle Eastern, African, or Mediterranean" in origin, this sub-clade is found at a low frequency only among Polish and Russian Jews; thus, European admixture is probably the source of this group among Ashkenazim. U3 among the Ashkenazi (2 out of 32) could be a genetic inheritance from Khazarian ancestors, given that the highest diversity of this subgroup is found in the Caucasus (Ossetia, Georgia, Armenia) and in Turkey (Bermisheva et al. 2002).

U4 is also probably European (1 out of 32), though the distribution of U5 is more complex, given that it occurs not only in European groups, but also in the Middle East and Central Asia. The fact that Behar (2004a) identifies Ashkenazi U5 as European in origin may indicate that the Jewish haplotypes more closely resemble those seen in Eastern European populations.

Bermisheva (2002) also explored haplogroup T, noting certain HVR1 haplotypes that are common among Finno-Ugric and Udmurt populations of the Ural region (126, 294; 126, 294, 296, 304; T1: 126, 294, 163, 186, 189). Ashkenazi T1-T5 (excluding T\*) comprise 21 individuals out of 565 in Behar's (2004a) study, some of which have identical or similar haplotypes to those found in Bermisheva's samples.

*Eastern vs. Western Ashkenazim*

One important discovery made in Behar's (2004a) study is the apparent differences in mtDNA haplogroup frequency between various Ashkenazi populations, particularly between eastern and western Ashkenazim. Behar divides the various Ashkenazi populations as follows: French Jews, German Jews, Austrian Jews, Lithuanian Jews, Polish Jews, Romanian Jews, Russian Jews, and Ukrainian Jews.

One apparent difference is that eastern Ashkenazim, particularly Polish Jews, appear to have as great a diversity of mtDNA haplotypes as Middle Eastern and European populations. Thomas (2002) had noted this feature in the Ashkenazi results in his own study. Some of these haplotypes do not appear at all among the western Ashkenazim. In fact, the western Ashkenazim display a remarkably low diversity of haplogroups and haplotypes, much lower than that seen in either eastern Ashkenazim or non-Jewish European/Middle Eastern groups. Haplogroups that appear in eastern Ashkenazi, but are rare to absent in western Jewish groups, include HV\*, HV1, pre-HV1, J1, J2, U1-6, W, V, and certain sub-clades of H (Behar et al; 2004a, Supplementary Material).

This would strongly favor an independent founder hypothesis among these populations. It would appear that the Ashkenazim share a common set of founders of both European and Middle Eastern origin, while a separate group of maternal founders entered the population of eastern Ashkenazi communities sometime during the Diaspora.

The fact that some of these mtDNA groups are rare to absent in western Ashkenazi populations argues in favor of a post-Diaspora European origin. Furthermore, many scholars believe that Eastern European Jewry has its genetic basis among the western Ashkenazim; Eastern communities were founded when Jews migrated from Germany and France after the 12th-13th centuries. Certain mtDNA haplogroups shared between the two populations, for example N1b and K, indicate that the eastern Ashkenazi communities do indeed share some common mtDNA genetic history with western groups, some of probable Middle Eastern origin. Yet it also appears that eastward moving Ashkenazim absorbed a number of separate European maternal founders once they settled in Eastern Europe. This absorption would explain a number of mtDNA haplotypes that Behar identifies as European in origin and are restricted primarily to eastern Ashkenazim, in particular, U5 and V. It may also explain the high frequency of mtDNA haplogroup J, as well as a number of H sub-clades, that are not present in the western groups.

#### *Exploration of Ashkenazi mtDNA Haplogroup H*

The frequencies of mtDNA Haplogroup H sub-clades among Ashkenazim are shown in Table 5 (Pereira et al. 2005, Table 1)

#### **Table 5**

## Frequency of Haplogroup H Sub-clades in Ashkenazim

H Sub-Clade	Frequency
H1	0.051
H2	0
H3	0.44
H4	0.007
H5a	0
H6	0.028
H7	0
H13	0.028
H*	0.052
Total (All H)	0.21

Pereira (2005) also listed sub-clade frequencies for a number of European and Middle Eastern populations, thereby allowing comparison to Ashkenazi results. However, it should be noted that Pereira relied exclusively on Behar's samples for his Ashkenazi results and only subtyped 29 out of 119 H mtDNA results. Forty (40) individuals out of the 119 had the CRS (Cambridge Reference Sequence) (Behar et al. 2004a, Supplementary Material).

In regards to H1, Pereira (2005) states the following:

*H1 is almost exclusively European, with its only incursion into the Near East being a few Palestinian individuals bearing the most common haplotype. This absence of derived lineages in the Near East sample suggests that the H1 sub-clade had its origin in Europe.*

Therefore, while it appears the H1 among Ashkenazim is of probable European origin, the possibility of a Middle Eastern origin based on the Palestinian findings remains unresolved. However, given that H1 does not occur in other reported Middle Eastern groups (Gulf States, Kurds) and in only low percentages in the Caucasus, a European origin for Ashkenazi H1 seems probable (Pereira et al. 2005).



As to H3 among Ashkenazim, its provenance is almost certainly European, given that it occurs in none of the Middle Eastern groups, including Palestinians. In fact, Pereira (2005) deemed H3 “exclusively European.”

Sub-clades H4 and H13 are found in Europe, the Caucasus and the Middle East; therefore, the origins of these groups among Ashkenazim remain unresolved. The same can be stated for H\*, which began in the Middle East, but is found at its highest frequency in east-central Europe and the Balkans, as well as along the Atlantic fringes of Europe, such as Spain and Ireland (Pereira et al. 2005).

Sub-clade H6 is identified as Eastern European and Transcaucasian in origin and distribution (Pereira et al. 2005). The description is in agreement with findings from another mtDNA study which located H6 and its sub-groups almost exclusively within Slavic and Turkish groups (Loogvali et al. 2004). However, there are hints in both studies that H6 and its sub-clades may also be found in low frequencies among some western European groups, such as the French and Irish (Loogvali et al. 2004; Pereira et al. 2005). In fact, Pereira suggests that H6 may have its earliest roots in Western Europe, and Loogvali indicates the precursor mutation to H6 (16362C) is found primarily in the Balkans. Thus, it appears that Ashkenazim obtained their H6 ancestry from European maternal founders, possibly Slavic or Khazarian in origin. The argument for a Khazarian origin for this sub-clade is strengthened by the fact that the highest frequency of H6 is found among the peoples of Chuvash, Russia (Pereira et al. 2005). The Khazar language is believed to have been a Chuvash dialect of Turkish (Koestler 1976, p. 21).

In conclusion, it appears that much of Ashkenazi H can be attributed to European founding mothers, though the origin of certain sub-clades, in particular H4, H13 and H\*, remain unresolved.

### **Conclusion: Future Jewish DNA Studies**

The DNA studies have revealed a high degree of genetic interrelatedness among Ashkenazi groups, particularly among those of Eastern Europe. This common ancestry can be attributed to a small founding population, coupled with rapid population growth and a high rate of endogamy over the past 500 years. The studies also indicate a sharing of genetic ancestry between eastern and western Ashkenazim, supporting the view that some portion of Eastern European Jewry was founded by western Ashkenazim.

DNA research has also revealed significant genetic links between Sephardic and Ashkenazi Jewish populations, despite their separation for generations. With the Cohanim study, researchers found a clear genetic connection between the Jewish priests and a shared Israelite ancestor from the past. Additional genetic results suggest that the Ashkenazim can trace at least part of their ancestry to their Israelite forbearers.

But Jewish DNA presents a picture that is far more complex than just the Cohanim results. This picture is also far more diverse than what many genetic studies on Ashkenazi Jews would suggest. Instead, many of those studies have focused heavily on the Israelite DNA results, often downplaying the significant contribution of European and Khazarian ancestors. The examination of only a single component of Jewish ancestry has resulted in an incomplete and, to a certain extent, distorted presentation of the Jewish genetic picture.

Diversity was present from Jewish beginnings, when various Semitic and Mediterranean peoples came together to form the Israelites of long ago. The genetic picture was clearly enriched during the Diaspora, when Jews spread far and wide across Europe, attracting converts and intermarrying over time with their European hosts. The most recent DNA evidence indicates that from this blending of Middle Eastern and European ancestors, the diverse DNA ancestry of the Ashkenazi Jews emerged.

Although the debate over the fate of the Khazars is far from over, DNA research suggests that remnants of these mysterious people continue to exist within the genetic makeup of Ashkenazi Jews. In fact, the Levite results indicate that the Khazars became fully integrated into the Ashkenazi communities and came to play an important role within the Jewish priesthood.

The Cohanim results do not disprove the genetic contribution of the Khazars. Rather, the DNA studies indicate that Jews are not entirely Khazarian, Israelite or European in genetic makeup, but a complex and unique mixture of all these peoples.

Genetic studies of the future will hopefully clarify many of the remaining mysteries surrounding the origins and formation of the Ashkenazi communities. For instance, the origins and distribution of the most common mtDNA haplogroup among Ashkenazim – haplogroup K – remains unexplored. Additionally, tantalizing differences in the genetic makeup of western and eastern Ashkenazi populations remain to be fully investigated by DNA researchers.

In addition to the Ashkenazim, many other Jewish groups are ripe for study by genetic researchers. Examination of these groups will no doubt help illuminate their common genetic bonds as well as their differences with other Jewish populations. Groups such as the Sephardic and Mizrahi Jews await study of their own unique DNA makeup.

In conclusion, much remains to be explored regarding the DNA of various Jewish populations. Future DNA studies will undoubtedly provide a clearer picture of the various heterogeneous peoples who came together over time to form the Jewish people of today.

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## Electronic-Database Information

[www.yhrd.org](http://www.yhrd.org)

YHRD STR Database

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