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**DNA Origins and Current Consequences for Sephardi, Mizrahi,
and Ashkenazi Males and Females: Latest Results from Medical,
Genealogical-Familial, and National-Ethnic Research**

INTRODUCTION

Since the 1970s, the use of DNA has increasingly become accepted as part of scholarly research and professional applications in several substantive areas, with major attention to the medical, genealogical-familial, national-ethnic, and legal areas. With the exception of the legal area, these areas have had much overlap in interest and application, especially with respect to Jewish communities throughout the world. The major purpose of this article is to compare the two major ethnic groups within world Jewry, Sephardim-Mizrahim and Ashkenazim, with attention to specific nations or regions within the Sephardi-Mizrahi group. The definitions of Sephardim, Mizrahim, and Ashkenazim vary among researchers, frequently because of different interpretations of the complexities of

Jewish history. The purpose of this paper is not to discuss those differences, but to use one definition that is understandable for interpreting the findings of this paper. Because this is a study comparing the DNA of Sephardim, Mizrahim, and Ashkenazim, this study will use the definitions used by Doron Behar and others in their recent (2008) large-scale study of Sephardi-Mizrahi female DNA.

Behar and his co-researchers use the term Ashkenazi to refer to Jews who trace their ancestry, over the past one thousand years, to Central and Eastern Europe. They use the term non-Ashkenazim to refer to all other Jews. Within this much more geographically widespread and culturally diverse group of non-Ashkenazim, who have continuously lived in the Near East, the Middle East, North Africa, Iberia, or the countries to which the Iberia exiles went escaping the Inquisition, and who have shared common ritual practices, Behar and others differentiate three groups: Sephardim, Mizrahim, and “others.” The Sephardim are those Jews with an ancestry in Iberia. The Mizrahim are those Jews with an ancestry in the Near East, the Middle East, or North Africa, but without an ancestry in Iberia. The other groups included in this specific study which generally are accepted by scholars as not fitting into either the Sephardi, Mizrahi, or Ashkenazi communities include, for example, Jews from India, Yemen, and Ethiopia.¹

Before comparing different communities, however, it is important to discuss the current status of research in the different substantive areas because, as suggested, for Jewish communities, it is impossible to totally separate the national-ethnic findings from

¹ Doron M. Behar et al., “Counting the Founders: The Matrilinial Genetic Ancestry of the Jewish Diaspora,” <http://www.plosone.org/article>, accessed August 10, 2008.

the medical and the genealogical-familial findings. The use of DNA for legal purposes has played a major role in increasing interest in DNA, but it is not discussed in this paper because of its low overlap with the other three areas. Interestingly, much of the interest in legal uses of DNA came from the Innocence Project which has helped prove the innocence of wrongly imprisoned people, and which came out of the Benjamin N. Cardoso School of Law of Yeshiva University in New York.

THE STATUS OF MEDICAL DNA RESEARCH AS RELATED TO JEWISH COMMUNITIES

In the medical area, much of the DNA research has had a specific relevance to Jews. In 1992, for example, Israeli researchers Batsheva Bonne-Tamir and Avinoam Adam, in *Genetic Diversity Among Jews: Diseases and Markers at the DNA Level*, wrote that “The molecular revolution in the late 1970s, and the study of human variation at the gene level begun in the early 1980s, dramatically increased both the amount and quality of information on the human genome.” They further noted that the “distribution of genetic disorders among Jews has long been a focus of interest for physicians and scientists as well as for lay men” because of the uniqueness of Jewish genetic patterns. While there is some disagreement, a composite of estimates suggests that today about 70% of Jewish men throughout the world, both Sephardim-Mizrahim and Ashkenazim, still have a Middle-Eastern DNA pattern on the male line despite two thousand years or more in the diaspora, indicating less than one half of one percent of change per generation for about sixty or more generations. This genetic pattern of strong continuity,

but with some change because of a long time-period, is unique in world history and has caused the Jewish community to be a major focus of medical research.² Partly because a number of diseases are more prevalent among Jews than among non-Jews, much attention has been given to the so-called “Ashkenazi Diseases” or “Jewish Diseases” within various national Jewish communities in the world as well as within the nationality-based groups within Israel.

The attention to “Ashkenazi” or “Jewish” genetic diseases has had major consequences. Tay-Sachs Disease historically was one of the most devastating diseases for Ashkenazi Jews, with approximately one in twenty-five Ashkenazim being a carrier of this almost-always fatal disease. Because of the discovery of an inexpensive enzyme test, Tay-Sachs Disease was one of the first diseases to allow a large-scale genetic screening, and Jewish communities became fervent participants in testing beginning in the early 1970s. Israel became the first country to offer free genetic screening. In communities with above-average rates of ingroup marriages, special attention has been given to Tay-Sachs. For example, in the Lubavitcher community in Crown Heights, New York, a screening system known as Chevra Dor Yeshorim was instituted in 1983 whereby children were tested before they were of marriage age, and “During courtship, before a couple is engaged, Dor Yeshorim now informs them whether they are

² Batsheva Bonne-Tamir and Avinoam Adam, *Genetic Diversity Among Jews: Diseases and Markers at the DNA Level* (New York: Oxford University Press, 1992), ix; A.S. Santachiara-Benerecetti, “The Common, Near-Eastern Origin of Ashkenazi and Sephardi Jews Supported by the Y-Chromosome Similarity,” *Annals of Human Genetics* 57, 1993, January, 55-64.

genetically incompatible – that is, if both of them carry a mutation for Tay-Sachs.”³

Whereas one in sixteen Jewish people in Crown Heights was estimated to carry the Tay-Sachs mutation, the number of babies with Tay-Sachs has dropped from fifty a year to almost none in the Lubavitcher community. In the early 1970s in North America, about one hundred children per year were diagnosed with Tay-Sachs, with about 85 being Jewish, but by 2000 there were only one or two Jewish cases per year.⁴ In Israel, as well as in the Jewish diaspora, Tay-Sachs has almost been eliminated through large-scale genetic testing. Similar DNA screening programs have been developed for other recessive diseases common in Ashkenazi communities.

Research on BRCA1 and BRCA2, breast cancer, also has become a major area of research within the Jewish community. For example, Mary-Claire King, a pioneer in research on breast cancer for over twenty years published, along with others, a 2006 article on breast cancer among Jewish women in New York and in Israel.⁵ Another recent research project from Northern California found that estimates of prevalence of the BRCA1 Mutation were 0.5% for Asian-Americans, 1.3% for African-Americans, and

³ Jon Entine, *Abraham's Children: Race, Identity, and the DNA of the Chosen People* (New York: Grand Central Publishing, 2007), 280.

⁴ M.M. Kaback, “Screening and Prevention in Tay-Sachs Disease: Origin, Update, and Impact,” *Advances in Genetics* 4 (2001): 253-265. See also Brenda McBride, “Michael Kaback ‘59, Developer of a Screening Test for Tay-Sachs Disease, Is Elected to Johns Hopkins University Society of Scholars,” *News @ Haverford*, <http://www.haverford.edu/newsletter/june06/kaback.htm>, accessed August 2, 2008. This article states that “Although there is still no cure, the incidence of Tay-Sachs in Ashkenazi Jewish families has been reduced by 99 percent” and quotes Kaback as saying “This demonstrates how one can really have an impact on disease.”

⁵ Sharon Simchoni et al., “Familial Clustering of Site-Specific Risks Associated With BRCA1 and BRCA2 Mutations in the Ashkenazi Jewish Population,” *Proceedings of the National Academy of Science in the United States (PNAS)* 103, no.10 (March 7, 2006): 3770-3774.

3.5% for Hispanics, but 8.3% for Ashkenazim.⁶ In a disturbing report, the *New Scientist* reported on July 9, 2005, that Myriad Genetics of Salt Lake City, Utah, had won a ruling “covering a specific mutation in the BRCA2 gene, which increases the risk of breast cancer. The mutation is found in 1 in 100 women of Ashkenazi Jewish descent. The ruling means that doctors offering tests for BRCA2 mutations are now legally obliged to ask women if they are Ashkenazi Jews. If they say yes, doctors must pay a license fee to Myriad. No fee is due if a patient says she does not know.” A member of the European Study of Human Genetics objecting to this, saying “We believe there is something fundamentally wrong if one ethnic group can be singled out by patenting.”⁷

These findings also are interesting because of King’s early pioneering work in which she found Mexican-American women, without any necessary identity as Jews, carrying the mutation for breast cancer. This raised the question of whether some of these Mexican-American carriers were descendants of secret Jews of Spain and Portugal, dating back to Inquisition times. Entine summarizes this possibility by writing that “The Jewish disease mutations found among many Christian Hispanos practicing Jewish-like cultural traditions does suggest another intriguing conclusion: many crypto-Jews married other crypto-Jews – they were endogamous, just like their Jewish cousins.”⁸

⁶ Esther M. John et al., “Prevalence of Pathogenic BRCA1 Mutations Carriers in 5 U.S. Racial/Ethnic Groups,” *The Journal of the American Medical Association (JAMA)* 298, no.24 (December 26, 2007): 2869-2876.

⁷ *New Scientist*. “Patent Singles Out Ashkenazi Jewish Women.” *New Scientist* 2507 (July 9, 2005) 7. Also see: <http://www.newscientist.com/article.ns?id>, accessed September 10, 2008.

⁸ Entine, 192.

Some attention has also been given to a few diseases, such as Familial Mediterranean Fever, which is found among Sephardim-Mizrahim but generally not among Ashkenazim. But, almost all of the attention has been given to the “Ashkenazi” diseases. Whether these diseases and others are “Jewish” or limited only to “Ashkenazi” is still debated, as will be discussed later in this paper. Entine suggests that there are fewer genetic diseases specific to Sephardim and Mizrahim than to Ashkenazim because Sephardim probably “intermingled more with gentiles than did European Jews.” There might be some truth to this suggestion, but preliminary data suggest that Sephardim and Ashkenazim have similar rates of marriage within their respective Jewish communities. In fact, Entine also referenced studies of Jewish male genetics including the important study by Hammer and others, and writes that “Jewish males appeared to have mixed hardly at all with non-Jews after the founding of the Jewish population.”⁹

The Ashkenazi population is much “younger” than the Sephardi-Mizrahi population, with a much higher rate of ingroup marriage. Sephardi and Mizrahi diasporas go back to several major different areas of the world, beginning in 586 BCE with the exile to Babylonia, and had some, but not an overwhelming amount of, genetic interaction with other Sephardi and Mizrahi communities. But, in contrast to the Sephardim-Mizrahim, the Eastern European Ashkenazi community, with over eight million people, is largely descended from a relatively small number of “founders” (about

⁹ Entine, 276, 210; Michael F. Hammer et al., “Jewish and Middle Eastern Non-Jewish Populations Share a Common Pool of Y-chromosome Biallelic Haplotypes,” *Proceedings of the National Academy of Science of the United States of America (JNAS)* (June 6, 2000): 6769.

25,000) who experienced a tremendous population explosion within a period of only a few hundred years, from about 1300 to Ha Shoah, and almost totally married within the group. Fraikor, for example, writes about the “phenomenal growth rate” and the high fecundity. In addition, as also noted by Fraikor, there was constant migration into the region by Western European Jews.¹⁰ This movement into Eastern Europe increased the concentration even more. Hence, the “typical” Ashkenazi is much more closely related genetically to another Ashkenazi than the “typical” Sephardi-Mizrahi is related to another Sephardi-Mizrahi.¹¹ This smaller genetic pool, because of marriages among the descendants of the original “founder” population, would carry a higher risk of two recessive carriers marrying and hence producing a child with a specific disease.¹²

In addition, the “bottleneck effect” could also have increased the likelihood of a recessive gene being multiplied within a specific population. The “bottleneck effect”

¹⁰ G. Fraikor, “Tay-Sachs Disease: Genetic Drift Among the Ashkenazi Jews,” *Social Biology* 24 (1977): 117-134.

¹¹ Raphael Patai in *Tents of Jacob: The Diaspora, Yesterday and Today* (Englewood Cliffs, NJ: Prentice-Hall, Inc., 1971, p. 79), using data from Arthur Ruppin, suggests that in 1170, there were 1,400,000 (93.3%) Sephardi and Oriental Jews in the world, and only 100,000 (6.7%) Ashkenazim. But, importantly, most Ashkenazim lived in Europe, with a heavy concentration increasingly in Eastern Europe, whereas the Sephardim and Mizrahim lived in diverse countries with limited genetic interchange. By 1939, the Sephardi and Oriental Jews numbered 1,500,000 (9.1%), while the Ashkenazi Jews had made an amazing increase to 15,000,000 (90.9%). From 1700 to 1939, in only 239 years, the Ashkenazim increased fifteen times over, from 1,000,000 to 15,000,000. Several writers (e.g., Fraikor) have mentioned the phenomenal birth rate among the Ashkenazim in Eastern Europe, among the highest recorded in world history.

¹² If only one parent has a recessive gene that can cause a disease, there is a 50% chance that a child will also have the recessive gene, but a non-disease dominant gene from the other parent will keep the child from having the disease. However, the child still can pass on the recessive gene to the next generation. However, if both parents have a recessive gene that can cause a disease, a child of these parents has a 25% chance of not inheriting the recessive gene, a 50% chance of continuing to carry and pass on the recessive gene but not having the disease, and a 25% chance of having the disease.

refers to the demographic situation whereby a group's population is drastically reduced within a fairly short period of time (for example, by a massacre or a plague that kills a high percentage of a population), which, because of a possible nonrandom result in deaths, can possibly lead to less genetic variation within the group. There also is the possibility that those with the recessive gene randomly survive at a higher rate, thus increasing the percentage of carriers within the surviving population. Ashkenazim, concentrated in Eastern Europe, and frequently victims of anti-Semitic violence, have suffered more than Sephardim and Mizrahim from bottlenecks. There is also the possibility that the recessive gene entered into the Ashkenazi population from either a mutation or from an outside source after the settlement in Europe, and hence really is limited to Ashkenazim. This could have been from a random non-Jewish male input or male mutation, but it also could have been from a female founder. As discussed later in this paper in detail, the female founders of the Sephardim-Mizrahim are very different from the female founders of the Ashkenazim, and it is possible that different diseases passed through different female lines.

There also is the possibility that a so-called "Ashkenazi" gene is actually a "Jewish" gene that dates from before the division into different diasporas and is found among both Ashkenazi and Sephardi-Mizrahi communities, but is referred to today as an "Ashkenazi" gene simply because, using today's division of Jews into Ashkenazim and Sephardim-Mizrahim, it is found much more frequently among Ashkenazim because of one of the factors discussed above. There is also the question of whether the "Ashkenazi" label has been applied to what are really "Jewish" diseases simply because most of the

research has been conducted in Ashkenazi communities and/or by Ashkenazi medical researchers who applied a limited and incorrect term of “Ashkenazi” instead of the broader and correct term of “Jewish” because of their own limited knowledge of Sephardim and Mizrahim.¹³ The question of the origins of various recessive genes that cause serious diseases among Jews is not totally answered, but remains controversial, and possibly varies for different diseases.

Although most attention has been given to “Ashkenazi” diseases because these diseases generally have been more severe, there has been some attention given to “Sephardic” diseases. As mentioned earlier, Familial Mediterranean Fever is probably the best known “Sephardic” disease. It occurs mostly in people with Sephardic, Armenian, Arab, or Turkish heritage.¹⁴ In general in these groups, one in five can be a carrier and one in two hundred can have the disease, with the likelihood probably higher among Armenians than among the other groups. Characterized by fever and inflammation of the abdominal membrane, the gene causing FMF was found in 1997, and there is treatment. Another “Sephardic” disease is Machado Joseph Disease. As described by Tourtellotte, MJD is “a fatal genetic disorder of the nervous system that cripples and paralyzes while leaving the intellect intact.”¹⁵ First documented in the 1970s, MJD occurs primarily in

¹³ This possibility was discussed, for example, at an interdisciplinary workshop on Jewish Genetic Diseases and Hispanics in the southwest United States, sponsored by the New Mexico Jewish Historical Society, on August 5, 2007, in Albuquerque, New Mexico.

¹⁴ A. Livneh et al., “MEFV Mutation Analysis in Patients Suffering From Amyloidosis of Familial Mediterranean Fever,” *Amyloid* 6 (1999):1-6. This research group is at the Sheba Medical Center in Tel Hashomer, Israel.

¹⁵ Wallace W. Tourtellotte, “Machado Joseph Disease,” (2008),

people of Portuguese ancestry, although it has been found in some other groups. Tourtellotte writes that “It is suggested that the original MJD mutation may have arisen among the settlements of Sephardic Jews in northeastern Portugal...Interestingly, many of the Portuguese with MJD, both in Portugal and the United States, still bear family names traditionally attributed to the Sephardim.”¹⁶

It also is very important to recognize that BRCA1, the breast cancer mutation, is also found in Sephardic females, and the marker has been found in Mizrahim, indicating that this mutation predates the diaspora. The frequency *could* be higher among Ashkenazim because of the high rate of ingroup marriage or a possible bottleneck in the Ashkenazi community, or it could be that non-Ashkenazi Jews have been underrepresented in studies. Clearly a combination of explanations might sometimes be needed. Concerning Tay-Sachs Disease, for example, Frisch and others concluded that it was “the founder effect in a rapidly expanding population arising from a bottleneck” that provides an hypothesis for explaining the spread of Tay-Sachs in Ashkenazi individuals.¹⁷

Discussing “Jewish diseases” in general, Dardashti writes that “Further complicating matters is the fact that some who today identify as Ashkenazim may really have been Sephardim who migrated to Eastern Europe 500 years ago. Jewish

<http://www.mazornet.com/genetics/machado.htm>, accessed August 30, 2008.

¹⁶ Ibid.

¹⁷ Amos Frisch et al., “Origin and Spread of the 1278ins TATC Mutation Causing Tay-Sachs Disease in Ashkenazi Jews: Genetic Drift as a Robust and Parsimonious Hypothesis,” *Human Genetics* 114:4 (March 2004) 366-376.

genealogists and academic researchers are discovering Sephardim among Eastern European ancestors.”¹⁸ She quotes Dr. Daniel M. Laby of Harvard Medical School as saying that it is not correct to say that a gene is only Ashkenazi when it is Jewish, and that “It’s bordering on malpractice not to inform those who may be of non-Ashkenazi origin.” There are other diseases in both Ashkenazi and Sephardi-Mizrahi communities, but these discussions suggest the importance of the situation.

The continuing and growing research in so-called “Ashkenazi Diseases” or “Jewish diseases,” combined with a relatively strong degree of interest in genetics among individuals in the Jewish community, have led to commercialization in this area in which medical information has become more easily available to the general public. For example, DNA Traits of Houston, Texas, provides DNA tests for twenty-five diseases common among what they call “Ashkenazi Jews” including Tay-Sachs, BRCA1, Gaucher’s, Crohn’s, and others.

THE STATUS OF GENEALOGICAL/FAMILIAL DNA RESEARCH AS RELATED TO JEWISH COMMUNITIES

In the genealogical/familial area, there has been a large increase of interest in DNA. After gathering as much information as possible about relatives by using the traditional methods of checking paper records (censuses, immigrant records, wills, burial records, Social Security records, etc.), in effect exhausting the “paper trail,” an increasing

¹⁸ Schelly Talalay Dardashti, “How Do Sephardic Jews Figure Into the Genetic Equation?” *Forward*, August 25, 2006.

number of Jewish people are turning to DNA. Jews, perhaps more than members of many other groups, are likely to turn to DNA because “paper trails” frequently are more nonexistent. Frequent migrations, persecutions, and name changes have decreased the possibilities of paper trails in many cases. There are indications that there is more interest in DNA testing in the Jewish community than in the non-Jewish community.¹⁹ In 2002, for example, Nicholas Wade, in the *New York Times*, noted that “A new thread is being woven into the complex tapestry of Jewish history, a thread fashioned from a double twist of DNA. The DNA data suggest a particular version of Jewish history and origins that historians have not yet had time to appraise but that seems to be reconcilable in principle with the historical record, according to experts in Jewish studies.”²⁰ However, Wade also noted some of the studies discussed in this paper which suggest that while the large majority of Jewish men can trace their direct male genetic pattern to the Middle East, “most or all” Jewish communities were founded by local, possibly non-Jewish, women. We will look at this suggestion in much more detail elsewhere in this paper in the section on ethnic and nationality genetic patterns.

Kleiman, in *DNA & Tradition: The Genetic Link to the Ancient Hebrews*, correctly makes the point that “DNA testing is now establishing itself as the third, and newest, core source in the field of family history, supplementing knowledge gained from

¹⁹ Jamie Malernee. “Family Ties: South Florida Jews Use DNA Testing to Uncover Past,” *The Sun-Sentinel* (Palm Beach, Florida), February 10, 2008, 1A & 8A.

²⁰ Nicholas Wade, “In DNA, New Clues to Jewish Roots,” *New York Times*, May 14, 2002, F1 & F7.

oral and documentary records.”²¹ He has a chapter comparing Ashkenazi and Sephardi-Mizrahi DNA, and makes the point that “all persons of Eastern European Jewish descent share many markers because they are descended from a rather small number of individuals, perhaps 50,000, who were alive in the year 1500.”²² Kleiman offers a good discussion of the published articles on Jewish DNA, and is a very good source for a good layperson-oriented understanding of Jewish DNA. He includes a discussion of the “Cohen Gene,” an explanation of how DNA confirms the Middle East origins of world Jewry, female DNA, the place of the “Lost Tribes” in DNA research, differences between Ashkenazim and Sephardim, and other points.

Jeffrey Malka, in *Sephardic Genealogy: Discovering Your Sephardic Ancestors and Their World* (2002), makes an important contribution to the study of Sephardi and Mizrahi family genealogy.²³ Although Malka does not discuss DNA in his book, his website activities indicate links to medical information for Jews. Malka’s other activities also indicate another major source of research and interest in Jewish genealogical and familial research, participation in Jewish genealogical organizations. At the 2008 annual conference of the prestigious International Association of Jewish Genealogical Societies (IAJGS), for example, Malka gave a presentation on Sephardic surnames, and reported on his origination of JewishGen’s Sephardic SIG website and his own SephardicGen.com

²¹ Rabbi Yaakov Kleiman, *DNA & Tradition: The Genetic Link to the Ancient Hebrews* (Jerusalem: Devora Publishing, 2004), 154.

²² *Ibid.*, 156.

²³ Jeffrey S. Malka, *Sephardic Genealogy: Discovering Your Sephardic Ancestors and Their World* (Bergenfield, NJ: Avotaynu, Inc., 2002).

website.

Although only a small part of the total presentations at the international genealogy conference dealt with DNA, with most presentations discussing archival research, family research, etc., seven presentations did discuss DNA research. Nina Sitron, in “Genes for Genealogists: Genetics, Inheritance, and DNA Made Simple,” discussed “the exciting new ways genealogists are using DNA to enhance their research.” Stephen Morse, in “From DNA to Genetic Genealogy,” discussed how genetic knowledge could “be used for finding relatives you didn’t know you had, learning about your very distant ancestors and the route they traveled, and determining if you are a Jewish priest (Kohan).” Jon Entine, in “Genetic Gold Mine: Jewish DNA Disorders and Their Role in Refining the Story of the Jews,” discussed how “Jewish genealogy is inextricably entwined with the diaspora history of the Jewish people,” and how Jewish historical insularity “has also been a boon to DNA disease researchers.” He also discussed how “The field of Jewish genetics, focusing on uncovering the origins of diseases that are uncommonly frequent among Jews, also provides genealogists with critical narrative evidence of migrations of Ashkenazi, Sephardi and Mizrahi populations.” Bennett Greenspan, founding president of Family Tree DNA, in “The DNA of Ashkenazi Jewry by Genetic Groups,” explained how “sufficient results are [now] available from the Ashkenazi community to begin to describe the major and minor family groups that when stitched together provide the genetic mosaic of Ashkenazi males worldwide.” Stanley Diamond, in “Medical and Genetic Family History: The Role of Jewish Genealogists,” discussed how family genealogists are or could be the repositories of vital medical and genetic history, and how

“previously unimagined opportunities” to combine medical and genetic data with family trees can “enhance the health of the people in our families.”

Michael Hammer and Syd Mandelbaum, in “The DNA Shoah Project: A Progress Report,” gave additional information on “genealogists seeking information on family members displaced by the Holocaust.” Herbert Huebscher and Saul Issroff, in “A Y-DNA Study of 50 Related Families Within a Unique Jewish Cluster,” discussed how this cluster’s progenitor probably lived two thousand to three thousand years ago, and how the most recent common male ancestor was a Levite who lived several hundred years ago. Interestingly, the presenters also noted that “Although many of the families’ earliest known origins lie in Eastern Europe, other data suggest the cluster’s MRCA [most recent common ancestor] was a Sephardic Jew.”²⁴

As the discussions so far have shown, for Jewish communities there is a strong overlap of medical research and genealogical/familial research, with DNA research having a potentially major contribution to make in both areas. As we will see next, the

²⁴ Regarding Sephardim who left Spain and Portugal as a result of the Inquisition, there is much research discussing the migrations to North Africa, the Ottoman Empire, Italy, the Netherlands, and the Americas. A small amount of research shows that some of these Sephardim went eastward and joined the Ashkenazi community in Eastern Europe. Dardashti, for example, quoted earlier in the section on diseases, wrote about the possible errors of looking at diseases as either Ashkenazi or Sephardi because some Jews “who today identify as Ashkenazim may really have been Sephardim who migrated to Eastern Europe 500 years ago” (see footnote # 18). This author has discussed this migration issue in “Lavender, Lavenda, Labender, Labenda, Lawenda, and Lavender Surnamed Individuals From Poland, Russia, Austria, Romania, and Czechoslovakia in the 1930 United States Census: From Spain, to Eastern Europe, to America?” The study shows that some Polish Jews named Lavenda or Lavender have an oral history of being descendants of Spanish Jewish exiles who went to Amsterdam, and then later went east to a thriving Jewish community in Poland instead of west to the Americas. In the early 1600s, before conditions deteriorated in Poland, such a move probably made sense to some people who preferred that to crossing an unknown ocean to a largely unknown land. In Spanish, la venda can mean “the bandage” or, in shortened form, “the blindfold,” either of which could have been a coded name for a secret Jew (report available from lavender@aol.com).

ethnic/nationality area of research also is strongly interconnected with these two areas.

THE STATUS OF ETHNIC/NATIONALITY DNA RESEARCH AS RELATED TO JEWISH COMMUNITIES

Having seen the overlaps among the different areas of research regarding DNA, let us now turn to DNA research findings on ethnic/nationality identities in the Jewish communities of the world. We will focus on Sephardi and Mizrahi communities, as compared to or contrasted with Ashkenazi communities, in different parts of the world. The DNA results on Sephardi, Mizrahi, and Ashkenazi males, including comparisons among the groups, are very basic and straightforward. The DNA research findings on Sephardi, Mizrahi, and Ashkenazi females is more complex, largely because of the fact that the different mutation rates between females and males cause methodological problems, but also because of suggestions that the female founders of world Jewish communities had a very different genetic heritage from the male founders.

(1) Male DNA: Similarities Among Sephardim-Mizrahim and Ashkenazim

Geneticists are very thankful for genetic mutations. Genes spontaneously mutate, i.e., randomly change for no apparent reason at fairly predictable rates.²⁵ Male (Y-chromosome) genes mutate at a much faster rate than do female (mitochondrial) genes.

²⁵ Research is continuing on refining estimated mutation rates, but sufficient knowledge is available to reliably use mutation rates as an indicator of the degree of genetic relationship between two males or between two females.

Briefly put, the more mutations (differences) two males have when they compare their DNA results, the longer time period there has been since they shared a common male ancestor. Likewise, the more mutations (differences) two females have, the longer time period there has been since they shared a common female ancestor. But, because males mutate much more rapidly than females, a male, compared to a female, is generally able to go back a much shorter period of time to find a genetic “cousin” and to estimate the degree of genetic relationship between himself and another male. The female has to go much further back in time to find a “cousin” and to estimate the degree of genetic relationship between herself and another female. In fact, the period of time for females is so long that it usually is not very helpful for genealogical or family relationship purposes. *The Daughters of Eve*, authored by Brian Sykes, a professor of Molecular Medicine at Oxford University, and published to popular acclaim in England in 2001, found that women can best be divided into seven major genetic groups.²⁶ Recent research is able to refine these findings, but the male-female differences in research are still great because of the vastly different mutation rates. Knowledge of DNA is constantly being refined, but at least for a while there will continue to be a methodological problem because of the differences in mutation rates.

As discussed briefly earlier, the basically straightforward DNA results for Jewish men conclude that the vast majority of Jewish men today, throughout the world with a few exceptions, share a Middle Eastern genetic pattern, and that their closest genetic

²⁶ Brian Sykes, *The Seven Daughters of Eve: The Science That Reveals Our Genetic Ancestry* (New York: W. W. Norton & Company, 2001).

male relatives in the world are Arabs and other males from the Middle East. In “The Common, Near-Eastern Origin of Ashkenazi and Sephardi Jews Supported by Y-Chromosome Similarity,” Santachiara and others, in 1993, showed that Ashkenazi and Sephardi males strongly share a similar genetic heritage on the male chromosome, and that both had a very low admixture with non-Jews *per generation*.²⁷ In “Jewish and Middle Eastern non-Jewish Populations Share a Common Pool of Y-Chromosome Biallelic Haplotypes,” Hammer and others documented in 2000 that most male Jewish populations were not significantly different from one another at the genetic level, and that there were low levels of admixture with non-Jewish populations despite centuries living in Europe and other areas of the world. Most of the Jewish male populations were in “a relatively tight cluster that was interspersed with Middle Eastern non-Jewish populations, including Palestinians and Syrians.”²⁸ There were no significant genetic differences between Ashkenazim and Sephardim-Mizrahim.

Also in 2000, Nebel and others in “High-Resolution Y Chromosome Haplotypes of Israeli and Palestinian Arabs Reveal Geographic Substructure and Substantial Overlap With Haplotypes of Jews,” concluded that the Y chromosome distribution of Jews and

²⁷ A.S. Santachiara Benerecetti et al., “The Common, Near-Eastern Origin of Ashkenazi and Sephardi Jews Supported by Y-Chromosome Similarity,” *Annals of Human Genetics* 57 (1993): 55-64. It is important to note that the percentage is based on *generations*, so that even if the rate is very low (generally estimated by most researchers to be about one-half of one percent) per generation, the overall rate still can be large because most diasporas are at least sixty generations removed from Israel, and in a few cases, e.g., the Babylonian exile, much more. In general, only about thirty percent of Jewish men today have a non-Middle Eastern Y-chromosome.

²⁸ M.F. Hammer et al., “Jewish and Middle Eastern non-Jewish Populations Share a Common Pool of Y-Chromosome Biallelic Haplotypes,” *Proceedings of the National Academy of Science of the United States of America (JNAS)* (June 6, 2000): 6769.

Arabs was similar, although not identical, and that a large degree of genetic overlap suggested a relatively recent common ancestry.²⁹ In 2001, Nebel and others, in “The Y Chromosome Pool of Jews as Part of the Genetic Landscape of the Middle East,” concluded that Sephardic and Kurdish Jews were indistinguishable from each another, although they both differed slightly but significantly from Ashkenazi Jews. They suggested that the difference with Ashkenazim might have resulted from a “low-level gene flow from European populations and/or genetic drift during isolation.” They further concluded that Jews were more closely related genetically to Kurds, Turks, and Armenians from the northern Fertile Crescent than to their Arab neighbors.³⁰ Nebel and others also found a strong similarity between Sephardim-Mizrahim and Ashkenazim concerning the famous Cohen Gene, assumed to be found in direct male descendants of Aaron, the brother of Moses and the first high priest. They found the “Cohen Gene,” considered the most definitive Jewish genetic pattern, in 10.1% of Kurdish Jewish males, 7.6% of Ashkenazi males, and 6.4% of Sephardi males. Interestingly, they also found the “Cohen Gene” among 2.1% of Palestinian Arabs, suggesting either the shared Middle Eastern genetic heritage of Jews and Arabs, or the possibility of conversion of Jews to Arab Muslim or Arab Christian identity. Although it makes historical and sociological sense that some Jews remaining in the area of Israel over the centuries would have

²⁹ Almut Nebel et al., “High-Resolution Y Chromosome Haplotypes of Israeli and Palestinian Arabs Reveal Geographic Substructure and Substantial Overlap With Haplotypes of Jews,” *Human Genetics* 107 (2000): 630-641.

³⁰ Almut Nebel et al., “The Y Chromosome Pool of Jews as Part of the Genetic Landscape of the Middle East,” *American Journal of Human Genetics* 69 (2001):1095-1112.

converted to Islam because they lived under an Islamic system with power over them, this question is delicate for both Jews and Palestinians, and no researcher has touched it, although many references have been made to conversions under such circumstances in other countries.³¹

In November 2001, Ostrer published “A Genetic Profile of Contemporary Jewish Populations” in which he showed that earlier studies which were “single-locus studies, involving blood groups, enzymes, serum markers, immunoglobulins and human leukocyte antigen types” tended to give diverse answers as to whether Jews had significant genetic admixture with non-Jewish populations. But, Ostrer noted, more recent studies using mitochondrial (female) or Y-chromosome (male) polymorphisms give “strong evidence for both matrilineal and patrilineal transmission, and many generations of endogamy in this population.”³²

DNA studies continue to be conducted on Jewish males in different areas of the world, and refinements, adjustments, and corrections continue to be made as new information is added, but the overall conclusion is clear: the vast majority of Jewish males share a Middle Eastern DNA, this pattern is found for both Sephardim-Mizrahim and Ashkenazim, and the closest genetic relatives are non-Jews from the Middle East.

³¹ Ibid. To understand the “Cohen gene” see Michael F. Hammer et al., “Y-Chromosomes of Jewish Priests,” *Nature* 385 (January 2, 1997). Also see Yaakov Kleiman, *DNA & Tradition: The Genetic Link to the Ancient Hebrews* (Jerusalem: Devora Publishing, 2004), 179-181.

³² Harry Ostrer, “A Genetic Profile of Contemporary Jewish Populations,” *Nature Reviews Genetics* 2

(2) Female DNA: Contrasts Between Sephardim-Mizrahim and Ashkenazim

The major point that can be made about Jewish male and female DNA patterns is that there is a very different genetic history for the two genders, and that these differences are found for both Sephardim-Mizrahim and Ashkenazim. We have seen that the male founders of different Jewish diaspora groups have a similar genetic history, but there is much less similarity between female founders of various Jewish ethnic/nationality groups. There is continuing debate on this issue, and research is increasing, but the general conclusion seems to be that different Jewish ethnic/nationality groups had a small number of founding mothers, perhaps often not of Jewish origin, but, that once a Jewish community had a critical mass of members to survive, marriage to women outside of the Jewish group was strongly prohibited. One major point in this general conclusion that is still being debated is whether the founding mothers of particular Jewish ethnic/nationality communities came with Jewish men from the Middle East or Near East or whether the founding mothers came from non-Jewish communities where the Jewish men settled. The research in this area has largely been a difference of conclusions between two groups of researchers, with articles by Mark G. Thomas and Doron M. Behar showing the differences.

Mark Thomas and others, in “Founding Mothers of Jewish Communities: Geographically Separated Jewish Groups Were Independently Founded by Very Few Female Ancestors” (June 2002), aptly described their overall conclusion in their subtitle. They had analyzed mitochondrial DNA from nine geographically separated Jewish

(November 2001): 891.

groups: Askenazim, Moroccan, Iraqi, Iranian, Georgian, Bukharan, Yemeni, Ethiopian (Beta Israel), and Indian (Bene Israel). Except for the Bukharan and Indian samples, all other Jewish samples were collected in Israel. They also had data from eight non-Jewish host populations and an Israeli Arab/Palestinian population. They concluded that their results “suggest that most Jewish communities were founded by relatively few women, that the founding process was independent in different geographic areas, and that subsequent genetic input from surrounding populations was limited on the female side.”³³

The researchers conclude not only that different Jewish communities generally had different local women as founders, but that it is impossible to determine the geographic origins of the founding mothers of the different Jewish groups. They conclude that the results are too “ubiquitous” to allow any conclusion other than a general Eurasian origin.³⁴ As the explanation for their findings, the authors note that Jewish identity followed the male tribal descent in ancient Israel, that since Talmudic times (about 200 BCE to 500 CE) Jewish identity has followed the female descent, that conversions to Judaism have not been uncommon in earlier parts of Jewish history (the pre-Christian Roman Empire), and that there have been a few small group conversions to Judaism (for example, the Khazars).³⁵ Shaye Cohen, in *The Beginnings of Jewishness* (1999), also noted that in antiquity there was not a strong boundary between Jews and Gentiles, that

³³ Mark G. Thomas et al, “Founding Mothers of Jewish Communities: Geographically Separated Jewish Groups Were Independently Founded by Very Few Female Ancestors,” *American Journal of Human Genetics* 70, no. 6 (June 2002): 1411-1420.

³⁴ Ibid.

³⁵ Ibid.

even when a boundary was established by the second century BCE it could be crossed, and that “gentiles crossed it and became Jews in a variety of ways, whether by political enfranchisement, religious conversion, veneration of the Jewish God, observance of Jewish rituals, association with Jews, or other means.”³⁶ Although genetic and archaeological data is adding more information, there continues to be strong debate over the origins of the Jews in ancient Israel.³⁷

Doron Behar and others, in “The Matrilineal Ancestry of Ashkenazi Jewry: Portrait of a Recent Founder Event” (2006), conclude that about forty percent of all Ashkenazi Jews are descended from four females (founding mothers), and that these women were “likely” of Middle Eastern or Near Eastern ancestry. Behar and others, using a larger sample than that of Thomas and others, therefore state that “in contrast to Thomas et al (2002), we conclude that a significant founding event is, indeed, readily evident in the maternal history of Ashkenazi Jews.”³⁸ These researchers conclude that it is more likely that these women accompanied the men as they left the Middle East, thereby decreasing the belief that Jewish men found local women in the diaspora. The likelihood that the women came with the men from the Middle East still would not prove that the original founding mothers were Jewish, but at least it would increase that

³⁶ Shaye Cohen. *The Beginnings of Jewishness: Boundaries, Varieties, Uncertainties* (Berkeley: University of California Press, 1999).

³⁷ See, for example, William G. Dever, *Who Were the Early Israelites and Where Did They Come From?* (Grand Rapids: William B. Eerdmans Publishing Company, 2003), and Raphael Patai and Jennifer Patai, *The Myth of the Jewish Race* (Detroit: Wayne State University, 1989).

³⁸ Doron Behar et al., “The Matrilineal Ancestry of Ashkenazi Jewry: Portrait of a Recent Founder Event,” *American Journal of Human Genetics* 78, no. 3 (2006): 487-497.

probability by narrowing down the probable geographical origin to the Middle Eastern.

The findings concerning Jewish DNA have received considerable publicity in the media. Nicholas Wade, a frequent writer on Jewish DNA for the *New York Times*, reported that David Goldstein, one of the researchers with Thomas, “said that the new report did not alter his previous conclusion. The mitochondrial DNAs of a small, isolated population tend to change rapidly as some lineages fall extinct and others become more common, a process known as genetic drift. In his [Goldstein’s] view, the Technion [Behar] team has confirmed that genetic drift has played a major role in shaping Ashkenazi mitochondrial DNA. But the linkage with Middle Eastern populations is not statistically significant...” Wade goes on to conclude that Goldstein agrees that there is no question of a Middle Eastern origin for Jewish men, but that it is still “very hard” to determine the genetic ancestry of Jewish women.³⁹

The research projects, disagreements, and new contributions continue. As was discussed in the first part of this study, in April 2008, in “Counting the Founders: The Matrilinial Genetic Ancestry of the Jewish Diaspora,” Doron Behar and others made another major contribution to the DNA of Jewish females, this time specifically studying the DNA of Sephardi, Mizrahi, and “other non-Ashkenazi” females.⁴⁰ They included female samples from fourteen communities, with the indicated size of the sample for each location as follows: Iberian Exiles: Portugal (30), Turkey (123), and Bulgaria (71); Near

³⁹ Nicolas Wade, “DNA Study on Jewish Lineage: Ashkenazi Women May Have Migrated Along With Husbands,” *New York Times*, January 14, 2006.

⁴⁰ Doron M. Behar et al., “Counting the Founders: The Matrilinial Genetic Ancestry of the Jewish

and Middle East: Iran (82) and Iraq (135); North Africa: Libya (83), Tunisia (37), and Morocco (149); Indian: Mumbai (34) and Cochin (45); Caucasuses: Azerbaijan (58) and Georgia (74); Ethiopia (29); and Yemen (119). They also had other Jewish groups which were too small to analyze including Italy (9), Algeria (20), Syria (4), Uzbekistan (17), Iraq Kurdistan (12), and former Yugoslavia (1). They also had 253 non-Jews including samples of Bedouins (58), Druze (77), Palestinians (110), and Cherkess in Israel (8).

These authors noted that while the genetic ancestry of Ashkenazim has been investigated in some depth, comparative data on non-Ashkenazim is lacking. At the same time, they note that studying fourteen different communities presented an analytic challenge to population geneticists because these communities were spread over a large and diverse geographical area, and had experienced very different historical effects. As they note, their study “is intrinsically more complex than studying phylogeographically less complex cases, such as the Ashkenazi Jew.”⁴¹

A major finding of this recent study was that, unlike the Ashkenazi pattern, five of the generally larger communities in this study, those from Morocco, Iraq, Iran, and the Iberia Exile communities in Bulgaria and in Turkey, did not show the small founder effect that characterized Ashkenazim. But, a small founder effect was found for five of the smaller and more remote communities, the previously crypto-Jewish community of

Diaspora.” <http://www.plosone.org/article>.

⁴¹ Ibid., 2. The fact that the non-Ashkenazim have a more diverse genetic background for the female founders adds even more importance to the need for more research on *all* Jewish communities in the world. Unfortunately, the frequent equating of “Jewish” with “Ashkenazi” can, as has been suggested in this paper, have serious medical consequences. Research on Israeli Jews of diverse nationality backgrounds can help correct this problem, but more research also is needed outside of Israel.

Belmonte in Portugal, the Bombay (B'nei Israel) and Cochin communities in Indian, and the two Caucasian communities of Azerbaijan (Mountain Jews) and Georgia.

Regardless of whether communities had narrow or non-narrow founder effects, all of the communities had a West Eurasian genetic heritage except for three – the two Indian communities and the community from Ethiopia. These three communities probably had female founders who were from the local areas. To decide whether a community had a small or non-small founder effect, the researchers looked at the number of diverse “genetic lineages” (“founding mothers”) that were needed to account for 40% or more of the genetic heritage of the specific sample. Let’s look at the communities, beginning with those who had small founder effects.

For Belmonte, Portugal, 28 of the 30 women (93.3%) in the sample shared the same lineage, indicating a very strong founder effect. As the only community from the Inquisition which retained the secret practice of Judaism for over 400 years, this finding suggests that perhaps a major reason this community was uniquely able to retain its identity in secrecy is that nearly all of the women (and it was women who largely kept the crypto-Jewish identities alive) were descended from only one “founding” mother around 1500. In effect, much of this crypto-Jewish community was one large extended family.⁴² Among the B'nei from Bombay, India, one genetic lineage alone accounted for 41.2% of the sample, and the top three lineages accounted for 67.7%. In Cochin, India,

⁴² This author spent part of a sabbatical in 2006 in Belmonte, Portugal, asking this question. The few definitive comments he received suggested that this explanation was accepted as the main reason for the survival of the crypto-Jewish community.

the top four lineages accounted for 66.7%. In Azerbaijan, one genetic lineage accounted for 58.6%, and in Georgia, one genetic lineage accounted for 58.1%. The two North African communities also had relatively small founder effects. The top two lineages in Libya accounted for 57.9% of the sample, and these same two lineages accounted for 27.0% of the Tunisian sample. In Tunisia, the top four lineages accounted for 43.2%. The authors refer to these seven locations as having a “paucity” of founding lineages.⁴³

In contrast to the relatively small and isolated communities, and the two North African communities, which had a relatively small number of genetic lineages (“founding mothers”) in their female samples, four other locations did not show a “paucity” of founding mothers. The authors refer to these locations as having “heterogeneity” in their patterns. In the sample from Morocco, it took seven different genetic lineages to account for only 30.2% of the sample. In the two samples representing Iberian exiles from the Inquisition, the top four lineages in Bulgaria accounted for only 26.8%, and in Turkey, the top four lineages accounted for only 17.1%. The fourth “heterogeneous” location was Ethiopia where it took five lineages to account for 41.3% of the sample. As noted, the Ethiopia sample and the two Indian samples apparently had most of their inputs from local females rather than from females of Western Eurasian heritage. As the authors state, the mtDNA pool of Ethiopian Jews “reflects the rich maternal lineage variety of East Africa.”⁴⁴ It also is interesting to note that the other Iberian community, Belmonte in

⁴³ Ibid., 11.

⁴⁴ Ibid., 11.

Portugal, which did not go into exile but instead kept a secret identity in Iberia, had a very small founder effect (basically one founding mother) probably because of the isolation from other Jewish communities and the strong need for secrecy in order to survive. Those Sephardim who left Iberia and went into exile had a greater choice of partners.

In Iraq it took seven lineages to account for 60.6%, and in Iran the top seven lineages accounted for only 47.6% of the women in the sample. It took seven lineages to account for 57.2% of the Yemen sample. The authors refer to these three locations as having an “intermediate” pattern, and note that “these three communities are long-standing Diaspora communities that have historical records consistent with a founding event, but not a narrow one.”⁴⁵

These fourteen “non-Ashkenazi” communities vary in the number of founding mothers they had, and some of the communities, especially the small and isolated communities, did have a relatively small number of founding mothers. But, looking at the specific genetic lineages found within the totality of these communities, it is evident that the maternal heritage of these “non-Ashkenazi” communities is much greater than that of the Ashkenazi communities.

⁴⁵ Ibid., 11.

CONCLUSIONS

These comparisons of medical, genealogical-familial, and national-ethnic issues have shown the strong extent to which the three areas are related. We have seen that while Sephardi-Mizrahi and Ashkenazi males have a small genetic heritage, the females of both groups have a much more diverse genetic heritage. The Ashkenazi female heritage is basically homogenous because of the small number of females who “founded” the Ashkenazi community, but the various Sephardi-Mizrahi (and other non-Ashkenazi) communities have much greater diversity genetically. Debate continues on the extent to which the female mothers of both groups were from local areas or were from the Middle East and went into the diaspora with the males. As noted previously, being from the Middle East would not prove Jewish heritage but it would increase the statistical odds of a Jewish heritage. Because the Ashkenazi community has a smaller genetic base, it has serious problems with some genetic diseases. This problem is much less grave in the Sephardi-Mizrahi communities because of a larger number of founding mothers and a greater genetic diversity. A dedicated attempt to lessen these diseases is having a significant impact in some areas, and extensive research continues in other areas.

Jewish communities in general have more interest in DNA testing than non-Jewish communities for several reasons, including, especially, the concern about Jewish diseases and the disrupted family histories that have resulted from persecutions and forced exiles. A traditional concern with the concept of family and a relatively high emphasis on education and knowledge have added even more to the interest. Recognition is also increasing considering the large number of non-Jewish people in the world who

have Jewish ancestry but whose ancestors left Judaism for a variety of reasons, frequently anti-Semitic in nature. For example, a recent study by Susan M. Adams et al., “The Genetic Legacy of Religious Diversity and Intolerance: Paternal Lineages of Christians, Jews, and Muslims in the Iberian Peninsula” (*American Journal of Human Genetics* 83 [December 12, 2008]: 725-736) documents that twenty percent of non-Jewish males in contemporary Iberia come from a Jewish genetic origin. The consequences for contemporary non-Jews with Jewish genetic backgrounds are also important.