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## Medical genetics of Jews

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The **medical genetics of Jews** is the study, screening, and treatment of [genetic disorders](#) more common in particular [Jewish](#) populations than in the population as a whole.<sup>[1]</sup> The genetics of [Ashkenazi Jews](#) have been particularly well-studied, resulting in the discovery of many genetic disorders associated with this [ethnic group](#).

In contrast, the medical genetics of [Sephardic Jews](#) and [Mizrahi Jews](#) are more complicated, since they are more genetically diverse and consequently no genetic disorders are more common in these groups as a whole; instead, they tend to have the genetic diseases common in their various countries of origin.<sup>[1][2]</sup>

Several organizations, such as [Dor Yeshorim](#),<sup>[3]</sup> offer [screening](#) for Ashkenazi genetic diseases, and these screening programs have had a significant impact, in particular by reducing the number of cases of [Tay–Sachs disease](#).<sup>[4]</sup>

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Genetics of Jewish populations [\[edit\]](#)

Different ethnic groups tend to suffer from different rates of hereditary diseases, with some being more common, and some less common. Hereditary diseases, particularly [hemophilia](#), were recognized early in [Jewish history](#), even being described in the [Talmud](#).<sup>[5]</sup> However, the scientific study of hereditary disease in Jewish populations was initially hindered by [scientific racism](#), which was based on [racial supremacism](#).<sup>[6][7]</sup>

However, modern studies on the genetics of particular ethnic groups have the tightly defined purpose of avoiding the birth of children with genetic diseases, or identifying people at particular risk of developing a disease in the future.<sup>[6]</sup> Consequently, the Jewish community has been very supportive of modern genetic testing programs; this high level of cooperation has raised concerns that conclusions may lead to stigmatization of the Jewish community.<sup>[5]</sup>

However, most populations contain hundreds of **alleles** that could potentially cause disease and most people are **heterozygotes** for one or two **recessive** alleles that would be lethal in a **homozygote**.<sup>[8]</sup> Although the overall frequency of disease-causing alleles does not vary much between populations, the practice of consanguineous marriage (marriage between second cousins or closer relatives) is common in some Jewish communities, which produces a small increase in the number of children with congenital defects.<sup>[9]</sup>

According to Daphna Birenbaum Carmeli at the [University of Haifa](#), Jewish populations have been studied thoroughly because:<sup>[10]</sup>

- Jewish populations, and particularly the large Ashkenazi Jewish population, are ideal for such research studies, because they exhibit a high degree of **endogamy**, and at the same time are a large group.
- Jewish populations are overwhelmingly urban and are concentrated near biomedical centers where such research has been carried out.

The result is a form of [ascertainment bias](#). This has sometimes created an impression that Jews are more susceptible to genetic disease than other populations. Carmeli writes, "Jews are over-represented in human genetic literature, particularly in mutation-related contexts."<sup>[10]</sup>

This set of advantages have led to Ashkenazi Jews in particular being used in many genetic studies, not just in the study of genetic diseases. For example, a series of publications on Ashkenazi centenarians established their longevity was strongly inherited and associated with lower rates of age-related diseases.<sup>[11]</sup> This "healthy aging" phenotype may be due to higher levels of [telomerase](#) in these individuals.<sup>[12]</sup> It may also be due to the well established correlation between high intelligence and longevity.<sup>[13][14]</sup>

Ashkenazi diseases [\[edit\]](#)

Today's 10 million Ashkenazi Jews descend from a population of only 350 individuals who lived about 600–800 years ago. <sup>[15]</sup> That population derived from both Europe and the Middle East. There is evidence that the [population bottleneck](#) may have allowed [deleterious alleles](#) to become more prevalent in the population due to [genetic drift](#).<sup>[16]</sup> As a result, this group has been particularly intensively studied, so many mutations have been identified as common in Ashkenazis.<sup>[17]</sup> Of these diseases, many also occur in other Jewish groups and in non-Jewish populations, although the specific mutation which causes the disease may vary between populations. For example, two different mutations in the [glucocerebrosidase](#) gene causes [Gaucher's disease](#) in Ashkenazis, which is their most common genetic disease, but only one of these mutations is found in non-Jewish groups.<sup>[4]</sup> A few diseases are unique to this group; for example, [familial dysautonomia](#) is almost unknown in other populations.<sup>[4]</sup>

Genetic disorders common in Ashkenazi Jews<sup>[1]</sup>

Disease	Mode of inheritance	Gene	Carrier frequency
<a href="#">Favism</a>	X-linked	<a href="#">G6PD</a>	
<a href="#">Bloom syndrome</a>	Autosomal recessive	<a href="#">BLM</a>	1/100
<a href="#">Breast cancer</a> and <a href="#">ovarian cancer</a>	Autosomal dominant	<a href="#">BRCA1</a> or <a href="#">BRCA2</a>	1/100 and 1/75, respectively
<a href="#">Canavan disease</a>	Autosomal recessive	<a href="#">ASPA</a>	1/60
<a href="#">Congenital deafness</a>	Autosomal recessive	<a href="#">GJB2</a> or <a href="#">GJB6</a>	1/25
<a href="#">Cystic fibrosis</a>	Autosomal recessive	<a href="#">CFTR</a>	1/25
<a href="#">Haemophilia C</a>	Autosomal recessive	<a href="#">F11</a>	1/12
<a href="#">Familial dysautonomia</a>	Autosomal recessive	<a href="#">IKBKAP</a>	1/30
<a href="#">Familial hypercholesterolemia</a>	Autosomal dominant	<a href="#">LDLR</a>	1/69
<a href="#">Familial hyperinsulinism</a>	Autosomal recessive	<a href="#">ABCC8</a>	1/125–1/160
<a href="#">Fanconi anemia C</a>	Autosomal recessive	<a href="#">FACC</a>	1/100
<a href="#">Gaucher disease</a>	Autosomal recessive	<a href="#">GBA</a>	1/7–1/18
<a href="#">Glycogen Storage Disease type 1a</a>	Autosomal recessive	<a href="#">G6PC</a>	1/71
<a href="#">Mucopolidosis IV</a>	Autosomal recessive	<a href="#">MCOLN1</a>	1/110
<a href="#">Niemann–Pick (type A)</a>	Autosomal recessive	<a href="#">SMPD1</a>	1/90
<a href="#">Nonclassical 21 OHase deficiency</a>	Autosomal recessive	<a href="#">CPY21</a>	1/6
<a href="#">Parkinson's disease</a>	Autosomal dominant	<a href="#">LRRK2</a>	1/42 <sup>[18]</sup>
<a href="#">Tay–Sachs</a>	Autosomal recessive	<a href="#">HEXA</a>	1/25–1/30
<a href="#">Torsion dystonia</a>	Autosomal dominant	<a href="#">DYT1</a>	1/4000
<a href="#">Usher syndrome</a>	Autosomal recessive	<a href="#">PCDH15</a>	1/72

Tay–Sachs disease [\[edit\]](#)

[Tay–Sachs disease](#), which can present as a fatal illness of children that causes mental deterioration prior to death, was historically extremely common among Ashkenazi Jews,<sup>[19]</sup> with lower levels of the disease in some Pennsylvania Dutch, southern Louisiana Cajun, and eastern Quebec French Canadian populations.<sup>[20]</sup> Since the 1970s, however, proactive genetic testing has been quite effective in eliminating Tay–Sachs from the Ashkenazi Jewish population.<sup>[21]</sup>

Lipid transport diseases [\[edit\]](#)

[Gaucher's disease](#), in which [lipids](#) accumulate in inappropriate locations, occurs most frequently among Ashkenazi Jews;<sup>[22]</sup> the mutation is carried by roughly one in every 15 Ashkenazi Jews, compared to one in 100 of the general American population.<sup>[23]</sup> Gaucher's disease can cause [brain damage](#) and [seizures](#), but these effects are not usually present in the form manifested among Ashkenazi Jews; while sufferers still bruise easily, and it can still potentially rupture the [spleen](#), it generally has only a minor impact on life expectancy.

Ashkenazi Jews are also highly affected by other [lysosomal storage diseases](#), particularly in the form of [lipid storage disorders](#). Compared to other ethnic groups, they more frequently act as carriers of [mucopolidosis](#)<sup>[24]</sup> and [Niemann–Pick disease](#),<sup>[25]</sup> the latter of which can prove fatal.

The occurrence of several lysosomal storage disorders in the same population suggests the alleles responsible might have conferred some [selective advantage](#) in the past.<sup>[26]</sup> This would be similar to the [hemoglobin](#) allele which is responsible for [sickle-cell disease](#), but solely in people with two copies; those with just one copy of the allele have a [sickle cell trait](#) and gain partial immunity to [malaria](#) as a result. This effect is called [heterozygote advantage](#).<sup>[27]</sup>

Familial dysautonomia [\[edit\]](#)

[Familial dysautonomia](#) (Riley–Day syndrome), which causes [vomiting](#), speech problems, an inability to [cry](#), and false [sensory perception](#), is almost exclusive to Ashkenazi Jews;<sup>[28]</sup> Ashkenazi Jews are almost 100 times more likely to carry the disease than anyone else.<sup>[29]</sup>

Other Ashkenazi diseases and disorders [\[edit\]](#)

Diseases inherited in an [autosomal recessive](#) pattern often occur in [endogamous](#) populations. Among Ashkenazi Jews, a higher

incidence of specific [genetic disorders](#) and [hereditary diseases](#) have been verified, including:

- [Colorectal cancer](#) due to [hereditary nonpolyposis colorectal cancer](#)<sup>[30]</sup>
- [Congenital adrenal hyperplasia](#) (nonclassical form) <sup>[31]</sup>
- [Congenital insensitivity to pain with anhidrosis](#)<sup>[32]</sup>
- [Crohn's disease](#) (the *NOD2/CARD15* locus appears to be implicated) <sup>[33]</sup>
- [Joubert syndrome](#) type 2 is disproportionately frequent among people of Jewish descent; this has been attributed to the resistance to intermarriage of this population.<sup>[34]</sup>
- [Kaposi's sarcoma](#)<sup>[35]</sup>
- [Maple syrup urine disease](#) <sup>[36]</sup>
- [Mucopolipidosis IV](#) <sup>[37]</sup>
- [Myeloproliferative Neoplasms](#) including [Polycythemia Vera](#) and [Essential Thrombocythemia](#) <sup>[38]</sup>
- [Nonsyndromic hearing loss and deafness, DFNB1 \(connexin 26\)](#) <sup>[39]</sup>
- [Parkinson's disease](#) (*G2019S/LRRK2* mutation;<sup>[40]</sup> The *LRRK2* mutation on the main haplotype, shared by Ashkenazi Jews, North Africans, and Europeans, initially arose in the Near East at least 4000 years ago. Because of a founder effect, the ancestors of present-day Ashkenazi Jews may have kept the low-frequency *G2019S* mutation through the different diasporas, whereas Near Eastern daughter populations lost the mutation. The mutation might then have been "reintroduced by recurrent gene flow from Ashkenazi populations to other Jewish, European, and North African populations. The present-day frequency of the mutation in control populations (0.05% in Europeans, 0.5% in North-African Arabs and 1% in Ashkenazi Jews) may support this scenario".)<sup>[41][42]</sup>
- [Pemphigus vulgaris](#)<sup>[43]</sup>
- [Schizophrenia](#) (*DNST3* gene variation)<sup>[44]</sup>
- [Von Gierke disease](#)<sup>[45]</sup>
- [Zellweger syndrome](#)<sup>[46]</sup>

### Non-Ashkenazi disorders [\[edit\]](#)

In contrast to the Ashkenazi population, [Sephardic](#) and [Mizrahi Jews](#) are much more divergent groups, with ancestors from Spain, Portugal, Morocco, Tunisia, Algeria, Italy, Libya, the Balkans, Iran, Iraq, India, and Yemen, with specific genetic disorders found in each regional group, or even in specific subpopulations in these regions.<sup>[1]</sup>

Genetic disorders common in Sephardic and Mizrahi Jews <sup>[1]</sup>				
Disease	Mode of inheritance	Gene or enzyme	Carrier frequency	Populations
<a href="#">Oculocutaneous albinism</a>	Autosomal recessive	<a href="#">TYR</a>	1/30	Morocco
<a href="#">Ataxia telangiectasia</a>	Autosomal recessive	<a href="#">ATM</a>	1/80	Morocco, Tunisia
<a href="#">Creutzfeldt–Jakob disease</a>	Autosomal dominant	<a href="#">PRNP</a>	1/24,000	Libya
<a href="#">Cerebrotendinous xanthomatosis</a>	Autosomal recessive	<a href="#">CYP27A1</a>	1/70	Morocco
<a href="#">Cystinuria</a>	Autosomal recessive	<a href="#">SLC7A9</a>	1/25	Libya
<a href="#">Familial Mediterranean fever</a>	Autosomal recessive	<a href="#">MEFV</a>	1/5–7	All MENA (Middle Eastern and North African countries).
<a href="#">Glycogen storage disease III</a>	Autosomal recessive	<a href="#">AGL</a>	1/35	Morocco, North Africa
<a href="#">Limb girdle muscular dystrophy</a>	Autosomal recessive	<a href="#">DYSF</a>	1/10	Libya
<a href="#">Tay–Sachs</a>	Autosomal recessive	<a href="#">HEXA</a>	1/110	Morocco
<a href="#">11-β-hydroxylase deficiency</a>	Autosomal recessive	<a href="#">CYP11B1</a>	1/30–1/128	Morocco

Genetic disorders common in Mizrahi Jews <sup>[1]</sup>				
Disease	Mode of inheritance	Gene or enzyme	Carrier frequency	Populations
<a href="#">Beta-thalassemia</a>	Autosomal recessive	<a href="#">HBB</a>	1/6	Iran, Iraq, Kurdistan
<a href="#">Factor VII deficiency</a>	Autosomal recessive	<a href="#">F7</a>	1/40	Iran
<a href="#">Familial Mediterranean fever</a>	Autosomal recessive, but heterozygous carriers also can show clinical manifestations.	<a href="#">MEFV</a>	1/5–1/7	Iraq, Iran, Armenia, North African Jews, Ashkenazi <sup>[47]</sup>

Glucose-6-phosphate dehydrogenase deficiency	X-linked	G6PD	1/4	Iraq, esp. Kurdistan, Syria and all MENA countries. Female heterozygotes can also show clinical symptoms due to lyonization (X-inactivation) especially during pregnancy. <sup>[48]</sup>
Inclusion body myopathy	Autosomal recessive	GNE	1/12	Iran
Metachromatic leukodystrophy	Autosomal recessive	ARSA	1/50	Yemen
Oculopharyngeal muscular dystrophy	Autosomal, recessive or dominant	PABPN1	1/7	Bukhara
Phenylketonuria	Autosomal recessive	PAH	1/35	Yemen

## Genetic testing in Jewish populations <sup>[edit]</sup>

One of the first [genetic testing](#) programs to identify [heterozygote](#) carriers of a genetic disorder was a program aimed at eliminating Tay–Sachs disease. This program began in 1970, and over one million people have now been screened for the mutation.<sup>[49]</sup> Identifying carriers and counseling couples on reproductive options have had a large impact on the incidence of the disease, with a decrease from 40–50 per year worldwide to only four or five per year.<sup>[4]</sup> Screening programs now test for several genetic disorders in Jews, although these focus on the Ashkenazi Jews, since other Jewish groups cannot be given a single set of tests for a common set of disorders.<sup>[2]</sup> In the USA, these screening programs have been widely accepted by the Ashkenazi community, and have greatly reduced the frequency of the disorders.<sup>[50]</sup>

Prenatal testing for several genetic diseases is offered as commercial panels for Ashkenazi couples by both [CIGNA](#) and [Quest Diagnostics](#). The CIGNA panel is available for testing for parental/preconception screening or following chorionic villus sampling or amniocentesis and tests for Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia, Gaucher disease, mucopolidosis IV, Neimann-Pick disease type A, Tay-Sachs disease, and torsion dystonia. The Quest panel is for parental/preconception testing and tests for Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Neimann-Pick disease types A and B, and Tay-Sachs disease.

The official recommendations of the [American College of Obstetricians and Gynecologists](#) is that Ashkenazi individuals be offered screening for Tay-Sachs disease, Canavan disease, cystic fibrosis, and familial dysautonomia as part of routine obstetrical care.<sup>[51]</sup>

In the [orthodox community](#), an organization called [Dor Yeshorim](#) carries out anonymous genetic screening of couples before marriage to reduce the risk of children with genetic diseases being born.<sup>[52]</sup> The program educates young people on medical genetics and screens school-aged children for any disease genes. These results are then entered into an anonymous database, identified only by a unique ID number given to the person who was tested. If two people are considering getting married, they call the organization and tell them their ID numbers. The organization then tells them if they are genetically compatible. It is not divulged if one member is a carrier, so as to protect the carrier and his or her family from stigmatization.<sup>[52]</sup> However, this program has been criticized for exerting social pressure on people to be tested, and for screening for a broad range of recessive genes, including disorders such as Gaucher disease.<sup>[3]</sup>

## See also <sup>[edit]</sup>

- Ethnicity and health
- Finnish heritage disease
- Genetic studies on Jews
- Jewish genealogy
- Jewish medical ethics



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## External links <sup>[[edit](#)]</sup>

- [Chicago Center for Jewish Genetic Disorders](#) <sup>🔗</sup>
- [Ashkenazi Jews](#) <sup>🔗</sup> – The Hebrew University of Jerusalem
- [Jewish Diseases Genetic Testing](#) <sup>🔗</sup> – The Center for Medical Genetics
- [Ashkenazi Jewish Genetic Diseases](#) <sup>🔗</sup> – Victor Center for Jewish Genetic Diseases
- [Ashkenazi Jewish Diseases](#) <sup>🔗</sup> – Tufts Medical Center
- [Jewish Genetic Disease Consortium](#) <sup>🔗</sup>
- [Center for Jewish Genetic Diseases](#) <sup>🔗</sup> – Mount Sinai Medical Center
- [Mendelian disorders among Jews](#) <sup>📄</sup> – Israeli National Genetic Database
- [Studies Show Jews' Genetic Similarity](#) <sup>🔗</sup> by *The New York Times*

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