

# **CONSUMER'S RESPONSE**

## **CARRIER TESTING IN THE ASHKENAZI JEWISH POPULATION**

**Elisa Engel Ross**

**President, Genetic Disease Foundation**

**New York, New York**

# CARRIER SCREENING IN THE ASHKENAZI JEWISH POPULATION

---

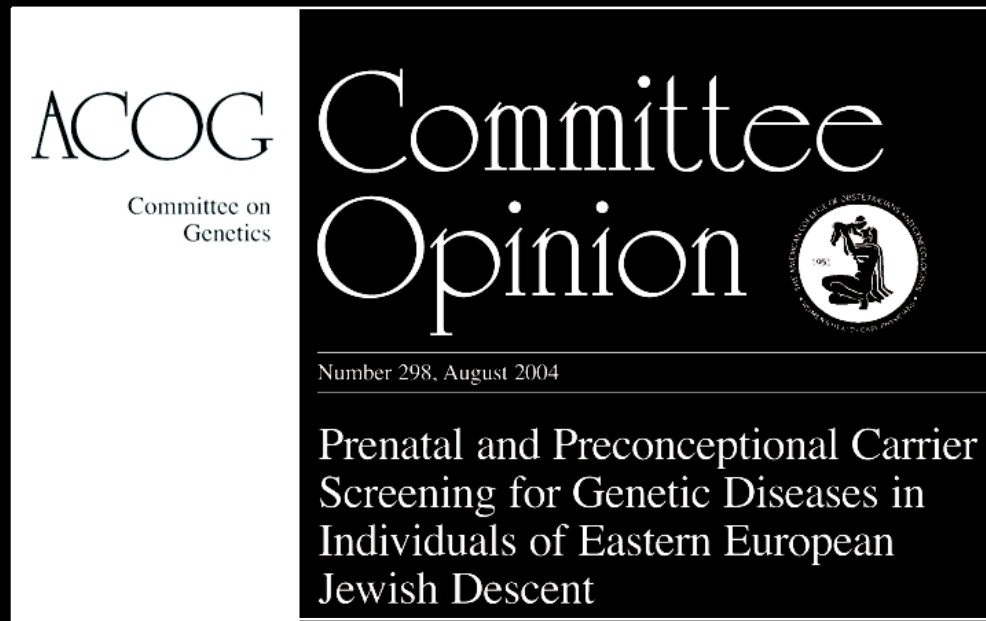
- Current Test Recommendations & Availability of Expanded Disease Tests
  - Counseling & Patient Choice for Test Selection
  - Acceptance Rates Among Ashkenazi Jews
-

# ACOG GUIDELINES FOR GENETIC SCREENING OF ASHKENAZI JEWISH PATIENTS

---

- **1976:** “Prevention of Tay-Sachs Disease: Carrier Identification and Genetic Counseling”. Tech Bulletin 34
  - Updated 1987
- **1991:** “Screening for Tay-Sachs Disease”. Cmte Opin 93
  - Updated 1995: Cmte Opin 162
  - Updated 2005: Cmte Opin 318
- **1998:** “Screening for Canavan Disease”. Cmte Opin 212
- **2001:** “Preconception and Prenatal Carrier Screening for Cystic Fibrosis”

# Obstet Gynecol 104:425, 2004: Cmte Opin 298



- **Recommended Testing - 4 Diseases:**

Tay-Sachs & Canavan Diseases, Cystic Fibrosis, & Familial Dysautonomia

- **Optional Testing - 5 Diseases:**

Bloom Syndrome, Fanconi Anemia Group C, Gaucher Disease Type 1, Mucopolysaccharidosis IV, & Niemann-Pick Type A Disease

# ACMG RECOMMENDATIONS - 2008

## Carrier Screening in Individuals of Ashkenazi Jewish Descent

Gross et al., *Genet Med* 10:54-56, 2008

---

## Technical Standards and Guidelines for Reproductive Screening in the Ashkenazi Jewish Population

Monaghan et al., *Genet Med* 10:54-57-72, 2008

### Recommended Testing for 9 Genetic Diseases

Bloom Syndrome

Canavan Disease

Cystic Fibrosis

Familial Dysautonomia

Fanconi Anemia Group C

Gaucher Disease Type 1

Mucopolysaccharidosis IV

Tay-Sach Disease

Niemann-Pick Type A Disease

“Others To Be Considered”

# GENETIC DISEASE FOUNDATION AWARENESS/ADVOCACY PROGRAM - 2005



**GENETIC  
DISEASE  
FOUNDATION**

**2005**

**Additional Diseases Tested**  
Glycogen Storage Type IA  
Maple Syrup Urine Disease

**ONE TEST. ELEVEN ANSWERS.**



**We can't cure genetic diseases. We can prevent them.**  
*Ask your doctor about genetic testing now!*

There are over 6000 genetic disorders that can be passed down through generations. The Genetic Disease Foundation has identified eleven life-threatening genetic diseases which are more likely to affect the children of Jewish descent; however, they can also affect the children of other origins. If you or your partner have any Jewish ancestry, consider screening for these diseases which can be fatal or severely debilitating.

- Tay-Sachs
- Familial Dysautonomia (FD)
- Canavan Disease
- Niemann-Pick
- Gaucher Disease (type I)
- Cystic Fibrosis (CF)
- Fanconi Anemia (type c)
- Bloom Syndrome
- Mucopolysaccharidosis type IV
- Maple Syrup Urine Disease
- Glycogen Storage (type Ia)

The good news is that it's rare for two prospective parents to both have the same gene that can cause one of these eleven diseases.

There's even better news: your doctor can perform a simple blood test that can determine if you both have the same genetic disorder or susceptibility.

As advances occur regularly, it is imperative to ask your doctor, genetic counselor, or geneticist about these and other genetic testing options.

*Be smart. Be sure. Test before.*



**GENETIC DISEASE  
FOUNDATION**

212-659-6704  
[www.geneticdiseasefoundation.org](http://www.geneticdiseasefoundation.org)

# GENETIC DISEASE FOUNDATION AWARENESS/ADVOCACY PROGRAM - 2007



**GENETIC  
DISEASE  
FOUNDATION**

**2007**

**Additional Diseases Tested**

**Familial Hyperinsulinemia**

**Nemaline Myopathy**

**Lipoamide Dehydrogenase Deficiency**

**Usher I**

**Usher III**

## ONE TEST. SIXTEEN ANSWERS



**We can't cure genetic diseases. We can prevent them.**  
*Ask your doctor about genetic testing now!*

There are over 6000 genetic disorders that can be passed down through generations. The Genetic Disease Foundation has identified eleven life-threatening genetic diseases which are more likely to affect the children of Jewish descent; however they can also affect the children of other origins. If you or your partner have any Jewish ancestry, consider screening for these diseases which can be fatal or severely debilitating.

- Tay-Sachs
- Fanconi Dysautonomia (FD)
- Canavan Disease
- Niemann-Pick
- Gaucher Disease (type 1)
- Cystic Fibrosis (CF)
- Falcot Anemia (type c)
- Bicon Spinaure
- Mucopolidosis type IV
- Maple Syrup Urine Disease
- Glycogen Storage (type II)

The good news is that it's rare for two prospective parents to both have the same gene that can cause one of these eleven diseases.

There's even better news: your doctor can perform a simple blood test that can determine if you both have the same genetic disorder or susceptibility.

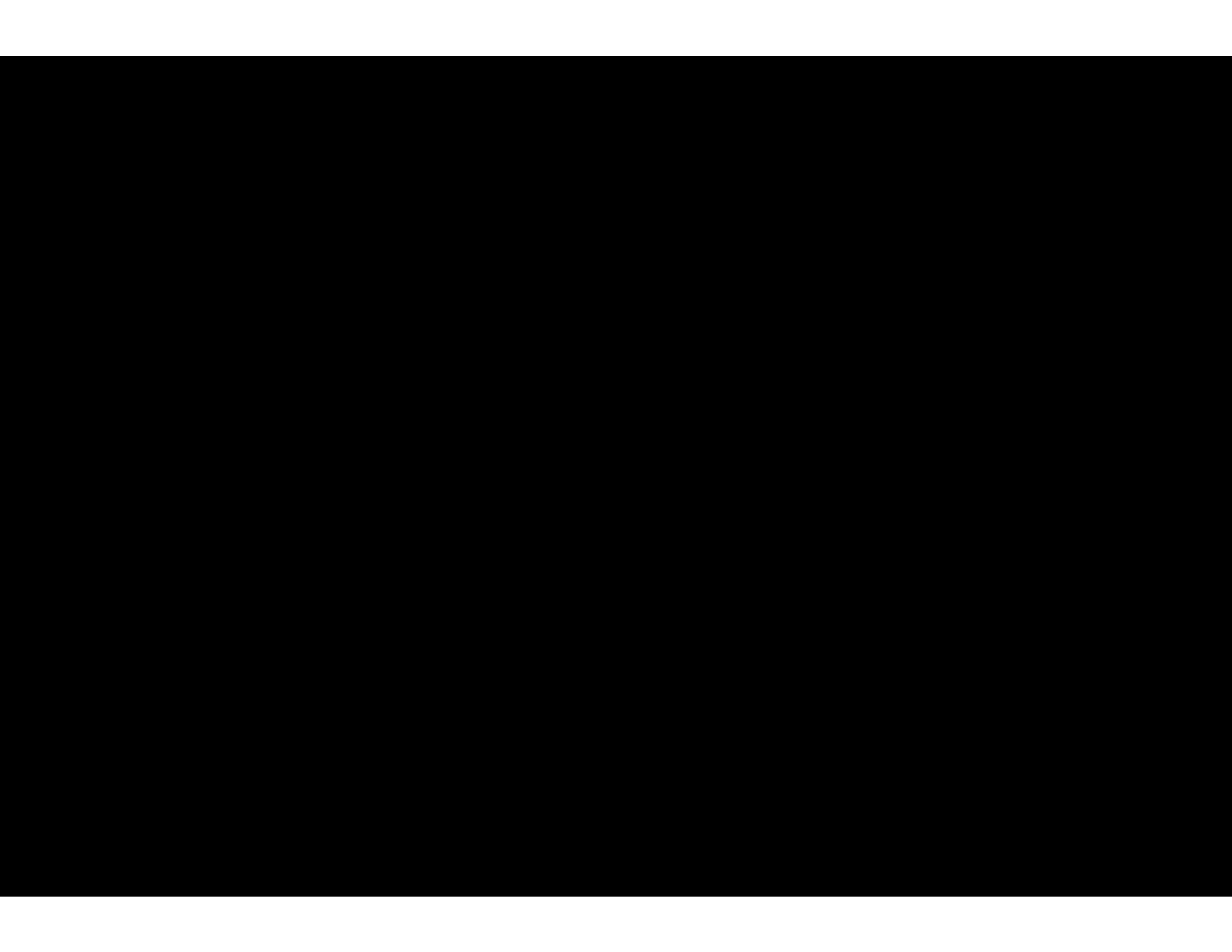
As advances occur regularly, it is imperative to ask your doctor, genetic counselor, or geneticist about these and other genetic testing options.

*Be smart. Be sure. Test before.*

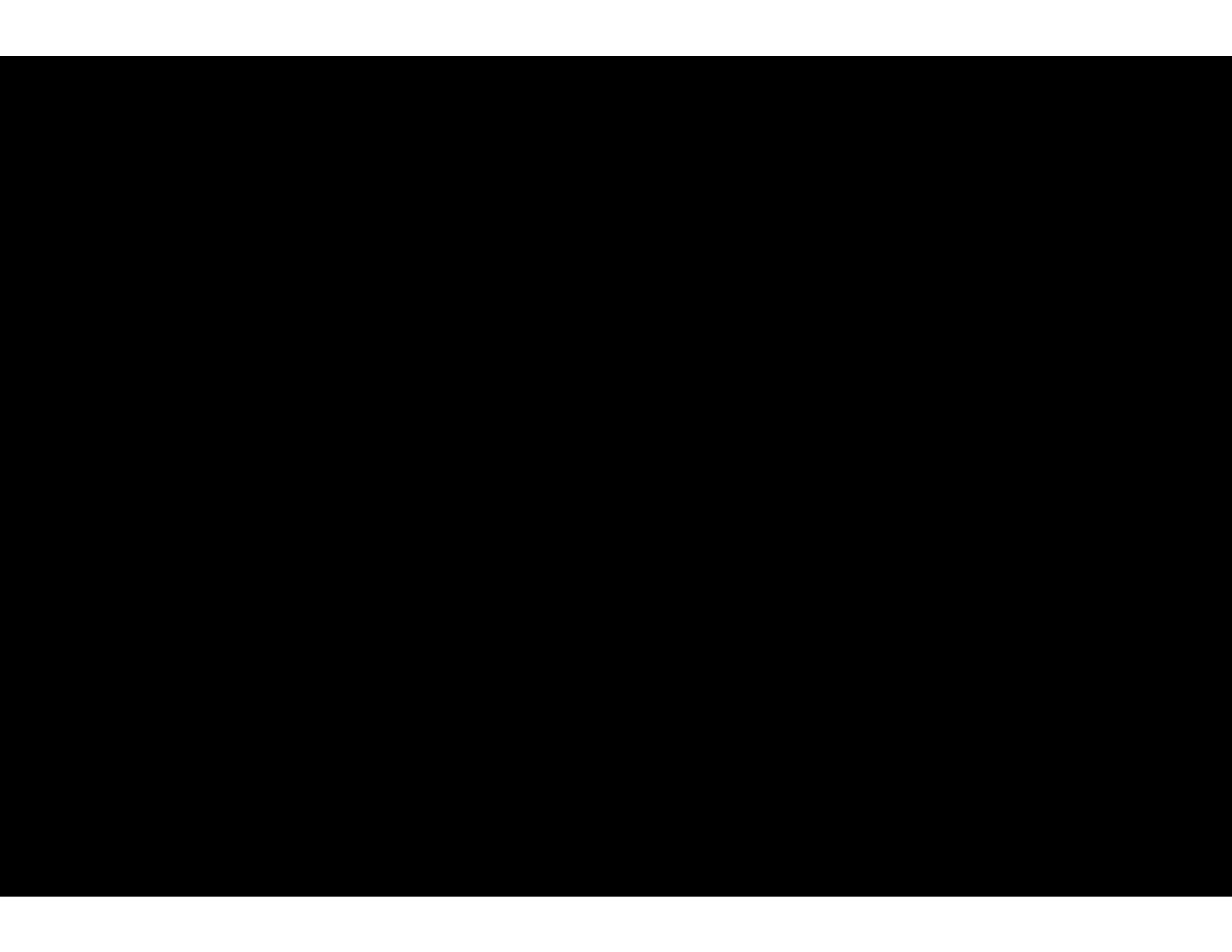


**GENETIC DISEASE  
FOUNDATION**

212-659-6704  
www.geneticdiseasefoundation.org







# JEWISH POPULATION STATISTICS

Country/ Metropolitan Area	Jewish Population	% World Jewish Population	
World	13,155,000	-	
Israel	5,393,400	40.9	
USA	5,275,000	40.1	
<b>New York Area</b>	<b>2,051,000</b>	<b>25.6</b>	
Los Angeles	668,000	5.1	20.0
SE Florida	527,500	4.0	
Chicago	270,500	2.6	
Boston	235,000	1.8	
San Francisco	227,800	1.7	
Washington DC	216,300	1.6	
Philadelphia	206,100	1.6	
Atlanta	119,800	0.9	
Baltimore	91,400	0.7	

# ASHKENAZI JEWISH PRENATAL CARRIER SCREENING PROGRAM

## *Mount Sinai School of Medicine*

Year Initiated	Cumulative # of Tests	Disease	Method
1973	1	Tay-Sachs Disease	Enzyme (DNA, 1990)
1992	3	Gaucher Disease	DNA
		Cystic Fibrosis	DNA
1995	4	Canavan Disease	DNA
1996	5	Niemann-Pick Disease	DNA
1999	7	Bloom Syndrome	DNA
		Fanconi Anemia Group C	DNA
2001	8	Familial Dysautonomia	DNA
2002	9	Mucopolysaccharidosis IV	DNA
2004	10	Maple Syrup Urine Disease	DNA
2005	11	Glycogen Storage Type 1a	DNA
2007	16	Familial Hyperinsulinemia, Lipoamide Dehydrogenase Deficiency, Netherton Neuropathy, Usher Syndrome Types 1 & 3	DNA DNA DNA