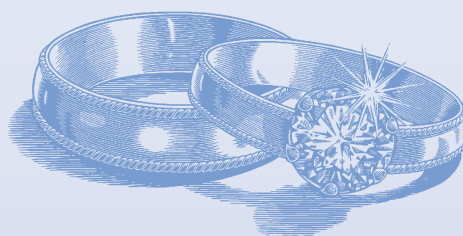


# Chicago Center for Jewish Genetic Disorders



*Mazel Tov!*



Jewish United Fund/  
Jewish Federation of  
Metropolitan Chicago

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Ben Gurion Way  
One South Franklin Street  
Chicago, Illinois 60606-4694



## Chicago Center for Jewish Genetic Disorders

*A cooperative effort of the Jewish United Fund/Jewish Federation of Metropolitan Chicago, Children's Memorial Hospital and the Illinois Jewish Genetic Disorders Committee • Supported by the Michael Reese Health Trust*

## The Chicago Board of Rabbis

*Supported by the Jewish United Fund/Jewish Federation of Metropolitan Chicago*

December 2002



Mazel Tov!

The Chicago Board of Rabbis and the Chicago Center for Jewish Genetic Disorders congratulate you on your upcoming marriage. This is a time for happiness and joy but also a time to reflect on your new life together.

Knowledge is the key to prevention of Jewish genetic disorders. Given the array of new technologies, early detection of carrier status allows couples to exercise a wide range of reproductive options. If carrier status is detected during pregnancy, options are fewer and decision-making becomes much more difficult.

We have enclosed educational materials for your review. The information sheet gives an overview of nine Jewish genetic disorders with facts about testing, frequency rates, and treatment options. The resource card provides you with a list of screening and testing locations, support agencies and helpful websites. The Action Card for Couples outlines steps you can take to help make health care decisions.

We thank you in advance for considering your responsibility to each other and to future generations. If you have any additional questions or concerns, please call the Center at (312) 357-4718, fax (312) 855-3295, email [jewishgeneticsctr@juf.org](mailto:jewishgeneticsctr@juf.org), or visit us online at [www.jewishgeneticscenter.org](http://www.jewishgeneticscenter.org).

*Rabbi Ellen W. Dreyfus*

Rabbi Ellen W. Dreyfus, President  
Chicago Board of Rabbis

*Dennis Kessler*

Dennis Kessler, Chairman  
Chicago Center for Jewish  
Genetic Disorders



# Chicago Center for Jewish Genetic Disorders

## Action Card for Couples

- 1.** Review the information about Jewish genetic disorders, screening options, and locations. Contact the Chicago Center for Jewish Genetic Disorders if you have any questions or concerns.
- 2.** Talk with your physicians about Jewish genetic disorders and the screening options. Bring the topic up with them; they may not know you are at risk for these disorders.
- 3.** Provide your partner with reinforcement. Explain how being screened is important for the future of your children.
- 4.** Follow through with these steps and get screened before you start a family.



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**The Chicago Center for Jewish Genetic Disorders**  
(312) 357-4718 • fax: (312) 855-3295  
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[www.jewishgenetics.org](http://www.jewishgenetics.org)

# Jewish Genetic Disorders Information Sheet

In almost every ethnic, racial or demographic group certain genetic diseases occur at higher frequencies among their members than in the general population. Almost all the conditions listed below are transmitted in an autosomal recessive fashion. This means that a couple can only have a child with the disorder if both parents are “carriers” of the trait for that disorder. Carriers usually have no symptoms of the condition. Carrier tests are performed on blood samples. If both parents carry a trait for the same condition, during pregnancy their fetus can be tested for the disease by sampling the placenta or the amniotic fluid. Carrier tests cannot entirely eliminate the possibility of being a carrier, but a negative test makes the chance very low.



## Ashkenazi Jewish Background

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There are several conditions that are more common in the Ashkenazi Jewish population. If one member of the couple is Jewish and the other is not, the Jewish parent can be tested first. If her/she is positive for the trait, the non-Jewish parent can then be tested. For some of these conditions, testing a non-Jewish individual is less reliable.

**Bloom syndrome** results in poor growth and poor resistance to infection. There is a high rate of cancer from which individuals usually die before age 30.

**Canavan disease** is characterized by progressive loss of white matter in the brain. Infants appear to be normal at birth but are later found to be hypotonic (floppy), fail to achieve head control and become developmentally delayed by 5-8 months. Features include an enlarged head, mental retardation, seizures and feeding difficulties. Death usually occurs within the first two decades of life.

**Familial Dysautonomia** is a disorder that results from the abnormal development of the nervous system, particularly the sensory and autonomic systems. The autonomic nervous system controls involuntary functions, such as swallowing, temperature and blood pressure regulation. Individuals with FD cannot regulate these functions. Other common manifestations are indifference to pain, inappropriate perception of heat and taste, excessive sweating, fluctuating blood pressures, gastrointestinal problems, poor speech and motor coordination.

**Fanconi anemia (Type C)** is a disorder which causes children to have anemia, short stature, learning disabilities or mental retardation. There may also be birth defects of the limbs, heart or kidneys. Risk for leukemia and early death is increased.

**Gaucher disease (Type 1)** can be very mild or severe. The absence of the enzyme glucocerebrosidase leads to the progressive build up of a fatty substance in the liver, spleen and bone marrow. Children and adults may have nosebleeds, anemia, an enlarged liver and spleen, bone pain, and easily broken bones. Because the severity is so variable and very effective treatment is available, decisions about testing are difficult.

**Mucopolysaccharidosis IV** is a disorder where children appear normal at birth but develop signs of central nervous system deterioration during the first year of life. Motor and mental retardation are usually mild to moderate, and are slowly progressive. Individuals with ML IV currently range from 1 to 30 years of age. Prognosis beyond this age and life expectancy are not known.

**Niemann-Pick disease (Type A)** is a severe neurodegenerative disorder of infancy. Affected babies exhibit an enlarged liver and spleen, poor growth and progressive physical and mental deterioration. Death usually occurs in early childhood (before age 5), due to infection.

**Tay-Sachs disease** is characterized by the onset of severe mental and developmental retardation during the first four to eight months of life. An early sign of the disease is the cherry-red spot, an unusual abnormality in the retina of the eye. The involvement of the central nervous system progresses rapidly and affected children become totally debilitated. Death usually occurs in early childhood. The Tay-Sachs gene is also more common among French Canadians from eastern Quebec and Cajuns from southern Louisiana.

**Dystonia** is an autosomal dominant disorder (an individual only needs one copy of an abnormal gene to be affected) characterized by sustained, twisting muscle spasms. With time, the frequency and duration of these spasms increase, leading to joint contracture and progressive disability. Children of an individual affected with Dystonia have a 50% chance of inheriting the gene mutation that causes the disease, but symptoms of Dystonia occur in only 30% of the individuals who inherit the gene mutation.

## Caucasian Background

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**Cystic fibrosis (CF)** is common among Caucasians, particularly those of northern European origin. It occurs with the same frequency in the Jewish and non-Jewish populations. The disease causes lung infections, difficulty breathing, and problems with bowel function, weight gain and growth. Children frequently need to be hospitalized, have physical therapy and take several medications. The average lifespan is 30 years. One in 2,500 Caucasian newborns has cystic fibrosis and 1:25 Caucasians carries the CF trait.

## Mediterranean (Greek, Italian), Sephardi Jewish, Southeast Asian, African

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**Beta-thalassemia** is a disorder that causes severe anemia in children, requiring frequent blood transfusion. Children may grow poorly, have bone deformities or fractures, and develop heart failure from their disease. Carrier frequency varies with ethnic group.<sup>i</sup>

<sup>i</sup>Sources:

The University of Chicago Hospitals, Women's Programs, Carrier Screening (Information Sheet), 2000.

*What You Should Know About... Jewish Genetic Diseases*, National Foundation for Jewish Genetic Diseases, Inc., 250 Park Avenue, Suite 1000, New York, NY, 10177

# Vital Statistics

	<b>Inheritance Pattern</b>	<b>Outlook</b>	<b>Treatment</b>	<b>Carrier Frequency</b>	<b>Carrier Screening</b>	<b>Prenatal Testing</b>
<b>Bloom Syndrome</b>	Recessive	Individuals can be functional	For symptoms	1 in 110	Yes	Yes
<b>Canavan Disease</b>	Recessive	Fatal, from early childhood through early teens	None	1 in 38	Yes	Yes
<b>Dystonia</b>	Dominant	Individuals can be functional	For symptoms	1 in 900	Yes	Yes
<b>Familial Dysautonomia</b>	Recessive	50% live to 30 years of age	For symptoms	1 in 30	Yes	Yes
<b>Fanconi Anemia (Type C)</b>	Recessive	Onset ranges from 17 months to 22 years - increased risk for cancer	For symptoms	1 in 89	Yes	Yes
<b>Gaucher Disease (Type 1)</b>	Recessive	Varies from mild to debilitating	Enzyme replacement therapy	1 in 10	Yes	Yes
<b>Mucopolipidosis IV</b>	Recessive	Developmental delays may be progressive from mild to severe	For symptoms	1 in 100	Yes	Yes
<b>Niemann - Pick Disease (Type A)</b>	Recessive	Fatal by 3-5 years of age	None	1 in 70	Yes	Yes
<b>Tay-Sachs Disease</b>	Recessive	Fatal in early childhood	None	1 in 26 to 1 in 30	Yes	Yes

## Sources:

[Jewish Genetic Disorders Program – Factbook](#), Division of Genetics, Children's Memorial Hospital, 2300 Children's Plaza, Box #59, Chicago, IL, 60614

[Carrier Testing for Genetic Conditions in Eastern European \(Ashkenazi\) Jewish People](#), Stamford Hospital, Center for Maternal-Fetal Medicine, P.O. Box 9317, Stamford, CT, 06904

[What You Should Know About...Jewish Genetic Diseases](#), National Foundation for Jewish Genetic Diseases, Inc., 250 Park Avenue, Suite 1000, New York, NY, 10177



# Chicago Center for Jewish Genetic Disorders

## Our Mission is to provide:

- ◆ Public Education and Awareness
- ◆ Professional Education
- ◆ Screening and Prevention Programs
- ◆ Information and Referral
- ◆ Advocacy



The Center is a cooperative effort of the Jewish Federation of Metropolitan Chicago, Children's Memorial Hospital and the Illinois Jewish Genetic Disorders Committee. We are supported by a generous grant from the Michael Reese Health Trust.

## Help is available...

### Chicago Area Genetic Screening and Testing Locations:

Advocate Christ Hospital and Medical Center .....	(708) 346-4395
Children's Memorial Hospital .....	(773) 880-4462
Cook County Hospital .....	(312) 633-7768
Evanston Hospital .....	(847) 570-2864
Advocate Illinois Masonic Hospital .....	(773) 296-7660
Loyola University Hospital .....	(708) 216-8167
Advocate Lutheran General Hospital .....	(847) 723-7705
Northwestern Memorial Hospital .....	(312) 926-7441
Reproductive Genetics Institute .....	(773) 472-4900
Rush Presbyterian St. Luke's Medical Center .....	(312) 942-6298
University of Chicago Medical Center .....	(773) 834-0555
University of Illinois Hospital .....	(312) 996-5807

### Chicago Area Family Support Programs:

Jewish Children's Bureau (JCB) .....	(312) 444-2090
Respite, foster care, legal advocacy, counseling, camp for children with genetic disorders and their families	
Jewish Family and Community Service (JFCS) ....	(312) 357-4800
Provides counseling and referral services	
Keshet .....	(847) 205-0274
Provides education and recreation services for children with developmental disabilities	

### Disclaimer

"This information is to be used for educational and informational purposes only. This information does not represent advice regarding medical diagnosis or treatment, referrals to health care professionals, endorsements of health care products or any other recommendations. This information should not be relied upon as a substitute for consultation with your doctor or other qualified health care professionals."

# For Information Contact...

## **Bloom Syndrome**

**Bloom Syndrome Registry**  
The New York Blood Center  
Laboratory of Molecular Genetics  
310 East 67th Street  
New York, NY 10021  
Ph: 212/570-3075 • Fax: 212/570-3195  
E-mail: jlg2003@med.cornell.edu

## **Canavan Disease**

**Canavan Foundation**  
110 Riverside Dr. #4F  
New York, NY 10024  
Ph: 212/873-4640 • Fax: 212/873-7892  
Toll free: 877/4-canavan  
E-mail: canavandisease@aol.com  
Website: www.canavanfoundation.org

## **Canavan Research Illinois**

PO Box 8194  
Rolling Meadows, IL 60067  
Ph: 847/222-0736  
Toll Free: 800/833-2194  
E-mail: canavan@canavanresearch.org  
Website: www.canavanresearch.org

## **Dysautonomia**

**Dysautonomia Foundation**  
633 Third Ave. 12<sup>th</sup> Flr.  
New York, NY 10017  
Ph: 212/949-6644 • Fax: 212/682-7625  
E-mail: dys212@aol.com  
Website: www.familialdysautonomia.org

## **FD Village FD Hope**

E-mail: info@fdvillage.org  
Website: www.fdvillage.org

## **National Dysautonomia**

**Research Foundation**  
421 W. Fourth St. Ste. #9  
Red Wing, MN 55066-2555  
Ph: 651/267-0525 • Fax: 651/267-0524  
E-mail: ndrnf@ndrnf.org  
Website: www.ndrnf.org

## **Dystonia**

**Dystonia Medical Research Foundation**  
One East Wacker Dr. Ste. #2430  
Chicago, IL 60601-1905  
Ph: 312/755-0198 • Fax: 312/803-0138  
E-mail: dystonia@dystonia-foundation.org  
Website: www.dystonia-foundation.org

## **Bachman-Strauss Dystonia & Parkinson Foundation**

Mount Sinai Medical Center  
One Gustave Levy Place, Box 1490  
New York, NY 10029  
Ph: 212/241-5614 • Fax: 212/987-0662  
E-mail: bachmann.strauss@mssm.edu  
Website: www.dystonia-parkinsons.org

## **Fanconi Anemia**

**Fanconi Anemia Research Fund, Inc.**  
1801 Willamette St. Ste. #200  
Eugene, OR 97401  
Ph: 541/687-4658 • Fax: 541/687-0548  
E-mail: info@fanconi.org  
Website: www.fanconi.org

## **Gaucher Disease**

**National Gaucher Foundation**  
5410 Edson Ln. Ste. #260  
Rockville, MD 20852-3130  
Ph: 301/816-1515 • Fax: 301/816-1516  
Toll Free: 800/428-2437  
E-mail: ngf@gaucherdisease.org  
Website: www.gaucherdisease.org

## **Mucopolidosis IV**

**ML4 Foundation**  
719 E. 17<sup>th</sup> St.  
Brooklyn, NY 11230  
Ph: 718/434-5067 • Fax: 718/859-7371  
E-mail: ml4www@aol.com  
Website: www.ml4.org

## **Niemann-Pick Disease**

**National Niemann-Pick Foundation**  
PO Box 49 415 Madison Ave.  
Ft. Atkinson, WI 53538  
Ph: 920/563-0930 • Toll Free: 877/cure-npc  
Fax: 920/563-0931  
E-mail: nnpdf@idcnet.com  
Website: www.nnpdf.org

## **Tay-Sachs Disease**

**National Tay-Sachs and Allied Disease Association**  
2001 Beacon St. Ste. #204  
Brighton, MA 02135  
Ph: 617/277-4463 • Toll Free: 800/906-8723  
Fax: 617/277-0134  
E-mail: ntsad-boston@worldnet.att.net  
Website: www.ntsad.org

## **Late-Onset Tay-Sachs Foundation**

1303 Paper Mill Rd.  
Erdenheim, PA 19038  
Ph: 215/836-9426 • Toll Free: 800/672-2022  
E-mail: mpf@bellatlantic.net  
Website: www.lotsf.org

## **Other Helpful Websites:**

Canavan Research Foundation.....www.canavan.org  
The Genetic Alliance .....www.geneticalliance.org  
Genetic and Rare Diseases Information Center .....www.genome.gov **or**  
www.nhgri.nih.gov/Info\_center/  
Jewish Genetic Disorders Program.....www.jewishgenetics.org  
March of Dimes Birth Defects Foundation .....www.modimes.org  
MUMS National Parent to Parent .....www.netnet.net/mums/  
National Organization for Rare Disorders, Inc. (NORD) .....www.rarediseases.org  
Tay-Sachs and Allied Diseases .....www.tay-sachs.org  
United Leukodystrophy Foundation .....www.ulf.org

# Jewish Genetic Disorders

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Genetic disorders often occur more frequently in defined populations. The following disorders are most commonly found in people of Ashkenazi Jewish descent (from Eastern or Central European lineage) but may also be found in other groups

Jewish genetic disorders include conditions that directly result from the abnormal genes (Mendelian disorders), and disorders that result from the combination of specific genes (disease predisposition genes) plus a range of other unknown factors.

## Mendelian Disorders

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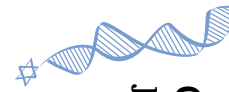
- **Bloom Syndrome**
- **Canavan Disease**
- **Dystonia**
- **Familial Dysautonomia**
- **Fanconi Anemia (Type C)**
- **Gaucher Disease (Type 1)**
- **Mucopolidosis IV**
- **Niemann-Pick Disease (Type A)**
- **Tay-Sachs Disease**

## Disease Predisposition Genes

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- **Breast Cancer (BRCA 1 and BRCA 2)**
- **Familial Colon Cancer**

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## Our Mission

To provide information to the Jewish community to help its members understand the scope of Jewish genetic disorders;

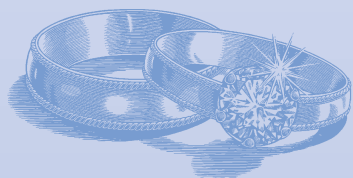
To educate physicians, other healthcare professionals and clergy;

To provide a vehicle for population-based services including education, screening, testing and genetic counseling;

To act as a clearinghouse for the full range of services required by individuals and their families who are affected by Jewish genetic disorders, including diagnostic work, treatment, counseling, support services and advocacy;

To develop a national and international presence that allows the Center to update and exchange information on the most current research and new treatment modalities;

To interface with other organizations focused on the clinical and bioethical issues that concern our community.



## Our Story

Over the last 30 years, screening tests for carriers have been developed for many of the identified Jewish genetic disorders. Unfortunately, community-based education efforts have not paralleled scientific advances.

Data collected by the International Tay-Sachs Disease Quality Control Program show that fewer than 900 people per year in the Chicago area are screened for Tay-Sachs disease, a substantial decline from figures recorded in the early 1980's. This screening rate of 3 per 1,000 Jewish people per year is less than half the screening rates in Los Angeles and New York. It is believed that this discrepancy is largely attributable to the lack of on-going education within the Chicago area.

The Center is a critical new effort to provide public and professional education and to empower community members to seek out information and prevention strategies.

With its current set of sponsors, the Chicago Center for Jewish Genetic Disorders represents the blending of science with religious, cultural and historical sensitivity and awareness. The Center seeks to become the voice that translates laboratory discoveries into accessible information for the Jewish community at risk as well as for physicians and other health care providers.

# Chicago Center for Jewish Genetic Disorders

### *Executive Committee:*

#### **Chairman**

Dennis Kessler

Joel Carp, ACSW

Joel Charrow, M.D.

Benn Greenspan, Ph.D.

Pat Yuzawa-Rubin

#### **Center Director**

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

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