

Carrier Screening ACT Sheet

Ashkenazi Jewish Genetic Disorders

Carrier Screening: Carrier screening preconceptionally or prenatally is recommended to identify individuals of Ashkenazi Jewish descent (one or more Jewish grandparents) who may carry one or more common mutations for a number of disorders: Tay-Sachs disease, Canavan disease, Gaucher disease, cystic fibrosis, familial dysautonomia, Niemann-Pick disease, type A, Bloom syndrome, mucopolidosis IV, and Fanconi anemia group C. Expanded panels in some laboratories may also include glycogen storage disease, maple syrup urine disease, dihydrolipoamide dehydrogenase deficiency, familial hyperinsulinism, nemaline myopathy, Joubert syndrome and Usher syndrome types 1F and III. These tests, except for TSD, are performed using mutation panels specific for individuals of Ashkenazi Jewish descent and may not identify mutations present in individuals of other ethnicities. The biochemical enzyme assay for TSD provides the highest detection rate in all populations.

Condition Description: Each of these disorders is inherited in an autosomal recessive manner. The carrier rates vary from 1/25 to 1/120 in the Ashkenazim and are much less frequent in other ethnicities. Ashkenazi Jewish individuals may be a carrier of more than one of these disorders. The onset and severity of these disorders is highly variable ranging from death in childhood to milder adult disease.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Inform individual of carrier screening result.
 - Consult with a genetic counselor or geneticist for additional testing recommendations for the reproductive partner.
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Reproductive Implications: When an individual is found to be a carrier of an Ashkenazi Jewish genetic disorder, the reproductive partner should be offered carrier screening for mutations in the same gene. In some circumstances, particularly if the partner is not Ashkenazi Jewish, a more comprehensive test may be needed. If both are carriers, the risk of the offspring having the disorder is 1 in 4 (25%), and prenatal or preimplantation testing should be offered.

Additional Information:

[Genetics Home Reference](#)

[Tay Sachs Disease](#)

[Canavan Disease](#)

[Gaucher Disease](#)

[Cystic Fibrosis](#)

[Familial Dysautonomia](#)

[Niemann-Pick Disease, Type A](#)

[Bloom Syndrome](#)

[Mucopolidosis IV](#)

[Fanconi Anemia Group C](#)

[Glycogen Storage Disease](#)

[Maple Syrup Urine Disease](#)

[National Tay Sachs and Allied Diseases Association](#)

[American College of Medical Genetics Practice Guideline](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/fabry-disease>

OMIM

<http://www.ncbi.nlm.nih.gov/omim/301500>

Genetics Home Reference: <http://ghr.nlm.nih.gov/BrowseConditions>

Tay Sachs Disease: <http://ghr.nlm.nih.gov/condition/tay-sachs-disease>

Canavan Disease: <http://ghr.nlm.nih.gov/condition/canavan-disease>

Gaucher Disease: <http://ghr.nlm.nih.gov/condition/gaucher-disease>

Cystic Fibrosis: <http://ghr.nlm.nih.gov/condition/cystic-fibrosis>

Familial Dysautonomia: <http://ghr.nlm.nih.gov/condition/familial-dysautonomia>

Niemann-Pick Disease, Type A: <http://ghr.nlm.nih.gov/condition/niemann-pick-disease>

Bloom Syndrome: <http://ghr.nlm.nih.gov/condition/bloom-syndrome>

Mucopolidosis IV: <http://ghr.nlm.nih.gov/condition/mucopolidosis-type-iv>

Fanconi Anemia Group C: <http://ghr.nlm.nih.gov/gene/FANCC>

Glycogen Storage Disease: <http://ghr.nlm.nih.gov/condition/glycogen-storage-disease-type-i>

Maple Syrup Urine Disease: <http://ghr.nlm.nih.gov/condition/maple-syrup-urine-disease>

National Tay Sachs and Allied Diseases Association

<http://www.ntsad.org/>

American College of Medical Genetics Practice Guideline

http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/ContentDisplay.cfm&ContentID=2746

Referral (local, state, regional and national):

Testing

<http://www.genetests.org>

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=genetests>

Find Genetic Services

<http://www.acmg.net/GIS/>

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