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, mucolipidosis 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.

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
  - Mucolipidosis 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

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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
- Mucolipidosis IV: http://ghr.nlm.nih.gov/condition/mucolipidosis-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/