Consent for Genetic Testing: Ashkenazi Jewish Screening

By signing below, I hereby authorize the staff of Dr._____ to obtain a blood sample for genetic testing from me [or specify patient for whom you are a legally authorized representative and for whom you are consenting: _____].

The Ashkenazi Jewish Screening tests that will be done are described below, except for those tests, if any, that I indicate I do not want performed.

The staff of Dr._____ has counseled me regarding this testing, and has explained the testing to me. I understand the following:

- The testing is done on a small sample of blood.

- **Intended Benefits/Purpose of the test**

_√ To identify if I carry a gene alteration
__ To help make a medical diagnosis
__ To clarify implications of a previous medical or genetic test result

- I have read and understand the general descriptions of each specific disease/condition that is listed below for which I am being tested.

- If this DNA testing identifies a known genetic mutation, I understand that I could either be a carrier or may be affected with that condition. I understand that further genetic counseling is available to me when the results of my testing are available, and the staff of Dr._____ is available to give me this counseling or to give me a referral so that I can get counseling elsewhere. I understand that I may wish to consider further genetic testing if the results are positive.

- I understand that when DNA testing does not show a change or mutation in a gene, the chance that I might be a carrier or affected is reduced, but not eliminated.

- No test(s) will be performed on my sample other than the one(s) listed below, and any unused portion of my sample will be destroyed within 60 days of receipt by the laboratory.

- I understand that in some families, DNA testing of family members may reveal that biologic parents are different than those people who have been thought to be parents before the DNA testing was done. I understand that DNA testing might also reveal other previously unknown information about my family relationships.

- The office of Dr. _____ does not guarantee that this screening will be covered by your insurance. I understand it is entirely my responsibility either to pay the charges myself and/or to contact my insurance carrier to determine if this testing will be covered.

- The results of my testing will be kept within the offices of Dr._____ and the test results will be disclosed and used only for the regular operations of Dr._____; and as required by law; and otherwise will be given only to me, unless I give permission for my results to be disclosed to someone else. The results will not be given to my insurance company unless I specifically consent to it.

Disease	Characteristics	Treatment	Carrier Frequency	Detection Rate
Tay-Sachs Disease	Neurological condition with death usually between the ages of 2 and 5	None	1/25	Enzyme-98% Gene testing-94%
Canavan Disease	Mental retardation, large head, weak muscles, and death in early childhood	None	1/40	96%

Niemann-Pick Disease, Type A	Growth failure, enlargement of liver and spleen, and progressive deterioration of the brain and central nervous system, leading to death by age 3	None	1/90	>95%
Familial Dysautonomia	Affects neurological function, including decreased pain sensation, no tears, seizures, and digestive problems. Death may occur in infancy or early adulthood.	None	1/50	99%
Cystic fibrosis	Affects the lungs and digestive system. Average life span is 30 years.	Alleviate symptoms	1/29	>95%
Fanconi Anemia	Severe anemia, heart, kidney and limb problems and an increased risk for cancer.	Treat symptoms	1/89	99%
Bloom Syndrome	Poor growth, deficient immune system, sensitivity to sunlight, and a high rate of cancer. Average life span is approximately 30 years	None	1/100	97%
Mucolipidosis Type IV	Profound psychomotor retardation and visual impairment.	None	1/122	96%
Glycogen Storage Disease Type 1a	Hypoglycemia episodes, leading to a severely enlarged liver, poor growth, bleeding disorders and death.	Strict diet and continuous glucose feeding	1/71	99%
Maple Syrup Urine Disease	Mental retardation, physical disabilities, coma and death	Life long dietary restriction of branched-chain amino acids	1/81	99%
Gaucher Disease	Causes and enlarged spleen and bone and joint problems. May be mild.	Cerezyme administration	1/15	95%
Familial hyperinsulinism	Low blood sugar resulting in seizures, poor tone, poor feeding and trouble breathing	Glucose infusions and meds to decrease insulin	1/100	90%
Lipoamide dehydrogenase deficiency (E3)	Can be mild with fatigue to severe neurological involvement, and possibly death	Dietary supplements	1/100	>95%
Nemaline myopathy	Neuromuscular disorder with progressive onset of muscle weakness, difficulty in feeding and respiration, delayed milestones and possible death	None	1/120	95%
Usher Syndrome Type I and III	Profound hearing loss and progressive blindness	None	Type I-1/165 Type III-1/95	Type I-75% Type III-95%

** **A NEGATIVE RESULT DOES NOT ELIMINATE THE CHANCE TO HAVE AN AFFECTED CHILD.**

I understand the above information and have had all my questions answered.

_____ I wish to have the complete panel of Ashkenazi Jewish Screening

_____ I wish to have the Ashkenazi Screening with the exception of _____

_____ I chose <u>not</u> to have the Ashkenazi Jewish Screening

Signature:_____ Print Name:_____

Date:_____ Witness:_____