Consent for Genetic Testing: Fragile X Syndrome, Spinal Muscular Atrophy, and Cystic Fibrosis 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 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 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 <u>does not guarantee</u> 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 and the test results will be disclosed and used only for the regular operations 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	Inheritance	Carrier Frequency	Detection Rate
Fragile X syndrome	From mild learning disabilities to severe mental retardation and autism	None	X-linked	1 in 150 * -> 1/260	>99%
Spinal Muscular Atrophy (SMA)	Loss of muscle control, including swallowing, head and neck control, walking and crawling, typically beginning before 2 years of age.	None	Recessive	1 in 40	94%
Cystic Fibrosis	Affects the lungs and digestive system. Average life span is 30 years.	Alleviate symptoms	Recessive	1/29	Caucasian: 93% Ash.Jewish: 97% Af. Amer: 81% Hispanic: 78%

** **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 Fragile X, SMA and Cystic Fibrosis Screening

_____ I wish to only have screening for _____

_____ I chose <u>not</u> to have Fragile X, SMA and Cystic Fibrosis Screening

Signature:_____ Print Name:_____

Date:_____ Witness:_____

Genetic testing refusal

I understand the above information, have had all my questions answered, and decline to have this testing.

Signature:_____ Print Name:_____

Date:_____Witness:_____