ACCU REFERENCE MEDICAL LAB
CAP ACCREDITED
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INFORMED CONSENT FOR CARRIER SCREENING

PATIENT NAME	DATE OF BIRTH
SAMPLE TYPE	BLOOD (EDTA)

I hereby consent to have my testing performed by ACCU Reference Medical Lab

☐ **Carrier Screening Tests - (CARR)**

☐ **Cystic Fibrosis – (CFXZ)**

Cystic Fibrosis (CF) is an autosomal recessive genetic disorder caused by two pathogenic CF variants or mutations in the CFTR gene (MIM# 602421). CF is a clinically variable multi-system disorder that may result in pulmonary and gastrointestinal / pancreatic disease, and male infertility. Genetic testing for detection of sixty pathogenic variants (located in the CFTR gene) reported to cause Cystic Fibrosis. This test is performed on the individual's DNA, using multiplex PCR followed by bead hybridization. The assay identifies 60 CF pathogenic variants including twenty-three pathogenic variants recommended for carrier screening by ACOG and ACMG.

☐ **Spinal muscular atrophy – (SMA)**

Spinal muscular atrophy refers to a group of autosomal recessive neuromuscular disorders characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Spinal Muscular Atrophy (SMA) is caused by deletions of both SMN1 (MIM# 600354) alleles in about 95% of the cases. About 5% of the cases are caused by a point pathogenic variant on one SMN1 allele and heterozygous deletion on the other SMN1 allele. This combination is not identified by the usual SMN1 gene testing. SMN2 (MIM# 601627), its paralogous gene, is a genetic modifier of the disease phenotype, and its copy number is correlated with SMA severity. The assay uses amplification of the SMN1 and SMN2 genes followed by fragment analysis and capillary electrophoresis. The assay can detect Exon 7 copy number states of 0, 1, 2 or >=3 for both SMN1 and SMN2, SMN1 to SMN2 Exon 7 gene conversion, and detect haplotype variants associated with duplication of SMA silent (2+0) carrier status.

☐ **Fragile X Syndrome Panel – (FX)**

Fragile X Syndrome is a genetic disease characterized by intellectual disability, behavioral and learning challenges, and various physical characteristics. Fragile X syndrome (FXS) is caused by mutation in the FMR1 gene (309550). In 95-99% cases, FXS is caused by a trinucleotide (CGG)n repeat expansion (MIM#309550.0004) of greater than 200 repeats. 1-5% cases may be associated with alterations in FMR1 gene such as deletions/duplication/single nucleotide variants. The assay determines the number of CGG triplet repeats in the 5′UTR of the FMR1 gene. The assay allows the detection of all kinds of alleles (normal, intermediate, premutant and mutant alleles) and quantification of expansions up to 200 CGG repeats. This assay is performed on the principle of fragment analysis and capillary electrophoresis.

I understand that a biologic specimen (blood) will be obtained from me and/or members of my family.
I understand the primary physical risks associated with most genetic tests are small, particularly for those tests that require only a blood sample.

I understand that this biological specimen will be used to attempt to determine if I am a carrier of a pathogenic variant(s) and if I and members of my family are at increased risk of being affected by the disease.

It has been explained to me and I understand that:

- Participation in genetic testing is completely voluntary. Genetic counselling is recommended prior to and following genetic testing. Further testing or additional physician consultations may be required.
- Results from genetic testing can be positive, negative, or inconclusive. Further testing may be needed to confirm the diagnosis. I understand I will be given the opportunity to talk with my physician or a genetic counselor about these results.
  - — A positive result may confirm whether a person is affected with, a carrier of or at risk for developing a genetic condition.
  - — A negative result does not exclude the possibility of being affected with or being a carrier of a genetic condition. Genetic conditions may have multiple causes, some of which may not be completely known or tested for.
  - — An inconclusive report may occur due to limitation of laboratory method/poor sample quality etc.

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- Currently, it is not standard practice for the laboratory to report likely pathogenic variants, and variants of uncertain significance.
- Although genetic results are usually accurate, several sources of error are possible, including clinical misdiagnosis of a condition, inaccurate information provided regarding family relationship, sample mislabeling or contamination, transfusion, bone marrow transplant. This assay does not detect certain types of genetic alterations (Limitations of the assay).
- If a genetic variant is identified, insurance rates, the ability to obtain disability and life insurance, and employability could be affected. The genetic information Nondiscrimination Act of 2008 extends some protection against genetic discrimination (genome.gov/10002328).
- Because of the complexity of genetic testing and the important implications of the test results, results will be reported only through a physician, genetic counselor, or other identified health care provider. The results are confidential to the extent allowed by law. They will only be released to other medical professionals and or those parties entitled to them by state and local laws with my written consent or as otherwise allowed by law.
- I understand that Accu Reference Medical Laboratory is not a specimen banking facility, and my sample will not be available after 60 days or for future clinical studies. I understand that my specimen will only be used for the genetic testing as authorized by my consent and that my sample will not be used in any identifiable fashion for research purposes without my consent.

Signatures

My signature below acknowledges my voluntary participation in this test. I understand that the genetic analysis performed by ACCU Reference Medical Laboratory is specific only for this disease/s and in no way guarantees my health, the health of an unborn child, or the health of other family members.

Patient/Guardian Signature	Date *(mm-dd-yyyy)*
Patient/Guardian Printed Name *(Last, First, Middle)*	DOB *(mm-dd-yyyy)*

I indicate my desire to opt out of participation in anonymized research studies using my sample by writing

my initials here ..... .

All samples from clients will be disposed of 60 days after testing is complete and will not be used for research or quality assurance.

Provider's or Counselor's Statement:

I have explained genetic testing (including the risks, benefits, and alternatives) to this individual. I have addressed the limitations outlined above, and I have answered this person's questions to the best of my ability.

Board Certified Physician or Genetic Counselor Signature

Date *(mm-dd-yyyy)*

Board Certified Physician or Genetic Councelor Printed Name
(Last, First, Middle)