HNL Lab Medicine

Answers for Life

Personalized Genetic Carrier Screening

HNL Genomics genetic carrier screening can detect >99% of inherited pathogenic variants from the genes tested and provides your patients with important information that will help them make critical life plans. Our screening offers highly targeted single genes and gene panels, providing your patients with:

- Comprehensive screening
- Relevant and actionable results
- Billing and financial assistance

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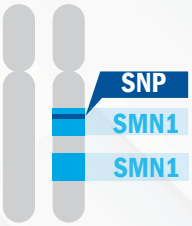
Single Disorder Carrier Screening

Carrier screening is a type of genetic test that can tell you whether your patient is carrying a DNA mutation that could cause a genetic disorder in the child. Conditions include:

Cystic Fibrosis

Cystic fibrosis carrier testing includes full gene sequencing and large deletion/duplication analysis of all 27 coding exons plus two deep intronic variants to improve detection rates among all ethnic groups. This test will detect approximately 99% of reported pathogenic variants across all ethnic groups and will provide a more accurate assessment of your patient's carrier status than a common mutation panel.

Spinal Muscular Atrophy



Spinal muscular atrophy carrier testing will identify individuals at risk of having a child with SMA. This test also assesses the risk of being a silent carrier of SMA, with two copies of SMN1 on a single chromosome and no copies of SMN1 on the other. About 1 in 50 people are carriers for SMA, regardless of ethnicity.[3] Carrier tests that do not assess silent carrier status may miss about 5 - 8% of carriers.[4]

Fragile X

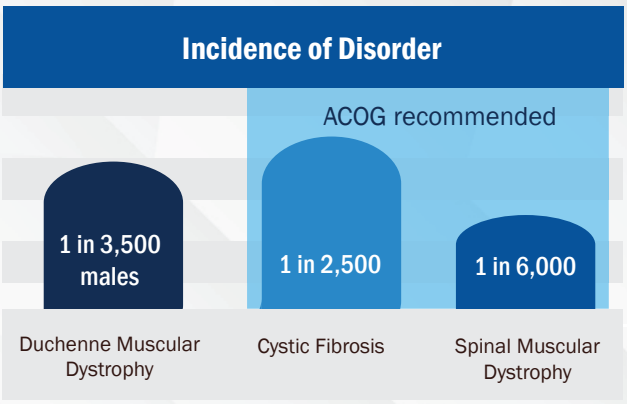
Fragile X carrier testing will detect individuals susceptible to triple repeat expansions in the FMR1 gene on the X chromosome, the cause of Fragile X syndrome in 99% of patients.

This test will identifythe following Fragile X-associated disorders:

- Women at risk of having a child with Fragile X syndrome
- Women at risk of having Fragile X associated premature ovarian failure
- Individuals at risk of having Fragile X associated tremor/ataxia syndrome

Duchenne Muscular Dystrophy

Duchenne muscular dystrophy, an X-linked disorder, is the most common muscular dystrophy in children. DMD is primarily seen in males. Females may be asymptomatic carriers or may exhibit mild symptoms of the disorder. DMD occurs in approximately 1/3500 boys and approximately 2/3 of affected boys inherit the mutation from their mothers.[5] Women who are carriers should be evaluated for clinical features every 5 years, with each pregnancy, or if they experience related symptoms.[6,7]



Common Carrier Screening Panel

A highly targeted panel that includes genes recommended or suggested for carrier screening by The American College of Obstetrics and Gynecology (ACOG) or the American College of Genetics and Genomics (ACMG).

Disorder	Gene
Alpha thalassemia	HBA1/HBA2
Beta thalassemia and sickle cell disease	HBB
Bloom syndrome	BLM
Canavan disease	ASPA
Cystic fibrosis	CFTR
Duchenne muscular dystrophy	DMD
Familial dysautonomia	ELP1 (IKBKAP)
Fanconi anemia C	FANCC
Fragile X syndrome	FMR1
Gaucher disease	GBA
Maple syrup urine disease, type 1A	BCKDHA
Mucolipidosis IV	MCOLN1
Niemann - Pick disease, type A and B	SMPD1
Phenylketonuria	PAH
Spinal muscular atrophy	SMN1/SMN2
Tay-Sachs disease	HEXA

Extended Carrier Screening Panel

The extended carrier screening panel includes all the disorders in the common carrier screening panel plus an additional 29 genes to provide a more comprehensive assessment of genetic disorder carrier status. Please see the Carrier Screening requisition form for the full list of genes on this panel.

Benefits of Carrier Screening through HNL Genomics

Information for Counseling

Early Diagnosis and Treatment

Full Gene Sequencing

Pan-Ethnic Testing

Questions?

Contact Us at 877-402-4221

Our customer service team is available to assist you.

REFERENCES

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