



Guidelines-focused panels:

The Fundamental and Fundamental Plus panels are designed to maximize clinical benefit within current payer policy.

Fundamental panel

Disease	Gene	Guidelines
Cystic fibrosis	<i>CFTR</i>	ACOG, ACMG
Spinal muscular atrophy*	<i>SMN1</i>	ACOG, ACMG
Fragile X syndrome*	<i>FMR1</i>	ACOG,‡ ACMG

*Analyzed using custom assay

‡Recommended when there is a family history of intellectual disability or fragile X syndrome

Scan here to
learn more



Fundamental Plus panel

Disease	Gene	Guidelines
Alpha thalassemia*	<i>HBA1/ HBA2</i>	ACOG, ACMG
Bloom syndrome	<i>BLM</i>	ACOG, ACMG
Canavan disease	<i>ASPA</i>	ACOG, ACMG
Cystic fibrosis	<i>CFTR</i>	ACOG, ACMG
Familial dysautonomia	<i>ELP1</i>	ACOG, ACMG
Fanconi anemia, <i>FANCC</i> -related	<i>FANCC</i>	ACOG, ACMG
Gaucher disease*	<i>GBA</i>	ACOG, ACMG
Beta globin-related hemoglobinopathy (including beta thalassemia and sickle cell disease)	<i>HBB</i>	ACOG, ACMG
Hexosaminidase A deficiency (including Tay-Sachs disease)	<i>HEXA</i>	ACOG, ACMG
Mucopolidosis IV	<i>MCOLN1</i>	ACOG, ACMG
Niemann-Pick disease, <i>SMPD1</i> -related	<i>SMPD1</i>	ACOG, ACMG
Spinal muscular atrophy*	<i>SMN1</i>	ACOG, ACMG
Dystrophinopathy† (including Duchenne/Becker muscular dystrophy)	<i>DMD</i>	ACMG
Fragile X syndrome*†	<i>FMR1</i>	ACOG, ACMG

ACOG:

Indicates disease listed in American College of Obstetricians and Gynecologists guidelines

ACMG:

Indicates disease listed in American College of Medical Genetics and Genomics guidelines

*Analyzed using custom assay

†Indicates X-linked disorders

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