Comprehensive testing for informed decision-making

Published professional society guidelines address carrier testing for some of the most common genetic disorders, and set the minimum standards for clinical practice. But your practice goes beyond the minimum for your patients, and so do we. The Preparent Exon panel fulfills many couples’ desire to maximize the information available to them—before, during, and after a pregnancy—and empowers them to make informed choices for their family. Positive results on this test are highly accurate, but just as importantly, negative results are reassuring.

Combined technology for higher sensitivity

Preparent Exon combines full exon sequencing and select copy number variant (CNV) analysis for a complete evaluation of your patient's carrier status. This test design results in clinical sensitivities that are, on average, 95% or higher in the general population. Exon sequencing detects small changes within the coding region of each gene. CNV analysis looks for large extra or missing pieces of select genes in which this type of variation, otherwise missed by exon sequencing alone, is a common cause of disease.

Full exon sequencing and select copy number variant analysis

Reporting of clinically actionable variants, both published and novel

Accurate and clear results

Carefully selected disorders for results that matter

The hereditary disorders included in the Preparent Exon panel are individually rare but collectively common. Disorders tested include those with published recommendations, such as cystic fibrosis, spinal muscular atrophy, hemoglobinopathies, and Ashkenazi Jewish disorders, as well as many more recessive conditions for which your patients are at risk. X-linked disorders are also tested, such as fragile X syndrome and Duchenne/Becker muscular dystrophy. Each disorder is categorized by key clinical impact areas.

CLINICAL IMPACT AREAS

- Life Expectancy: Decreased life expectancy and increased childhood mortality
- Quality of Life: Severe impact on quality of life; typically no effect on life expectancy
- Treatment Benefits: Early medical intervention can increase life expectancy or reduce symptoms
- Intellectual Ability: Associated with varying degrees of intellectual disability

fast facts:

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<th>PREPARENT EXON PANEL</th>
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<td>150+ genetic disorders</td>
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REFERENCES

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