Exome testing is a well-established tool for diagnosing genetic conditions in situations where no targeted gene testing exists, or when targeted genetic testing has failed to identify the cause of disease. For many, a genetic diagnosis obviates the need for future diagnostic procedures or testing, and can inform treatment choices.

Exome testing has been shown to be successful at identifying the genetic cause of conditions when patients have multiple congenital anomalies and/or neurodevelopmental disorders. Detection rates range from about 28.8% to as high as 57.5% for this patient population.1,7 Furthermore, in those with a genetic cause identified, 20.9% to 49% of patients had a change in their management based on the exome results.1,8

Exome with CNV Evaluation from Quest Diagnostics provides a new level of diagnostic and treatment possibilities positively impacting patient care and quality of life. Our test delivers exome sequencing with a mean read depth of 126X, a validated CNV evaluation, and mitochondrial genome results with a mean read depth of 550X. The exome provides a coverage of 99.6% at 20X. These leading metrics come with the added benefit of having both mitochondrial genome sequencing and CNV analysis included.

Quest’s metrics, combined with our strong portfolio of cytogenetic and biochemical genetic tests that are often precursors to exome testing, make us a single source for all your testing needs, both routine and advanced testing.

Quest Diagnostics delivers the quality testing patients need. Our in-network status in most health plans helps minimize patients’ out-of-pocket costs. You can order with confidence through your routine ordering channels.
Standardized classification method used for variant curation and reporting

To assess the pathogenicity of genetic variants in patients, we use a rules-based, weighted process (published by our group in 2015) that is aligned with the American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) guidelines. We conduct a thorough investigation of published research, which is then integrated with data from our internal dataset, external databases, research collaborations, and clinician-provided phenotypes. Close collaboration among our genomic variant scientists, genetic counselors, and clinical laboratory directors facilitates the collection and systemic analysis of relevant information in order to provide clinically-informative results to our clients.

For more information on the variant assessment process, please visit QuestDiagnostics.com/VariantIQ.

Learn more at QuestExome.com

Quest Diagnostics offers healthcare providers access to genetic experts who can assist in understanding how to interpret your patient’s results. For questions related to the Exome with CNV Evaluation, please contact our Genomic Client Services team at 1.866.GENE.INFO (1.866.436.3463).

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics’ understanding at the time of the report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the healthcare provider’s clinical evaluation.

References

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References