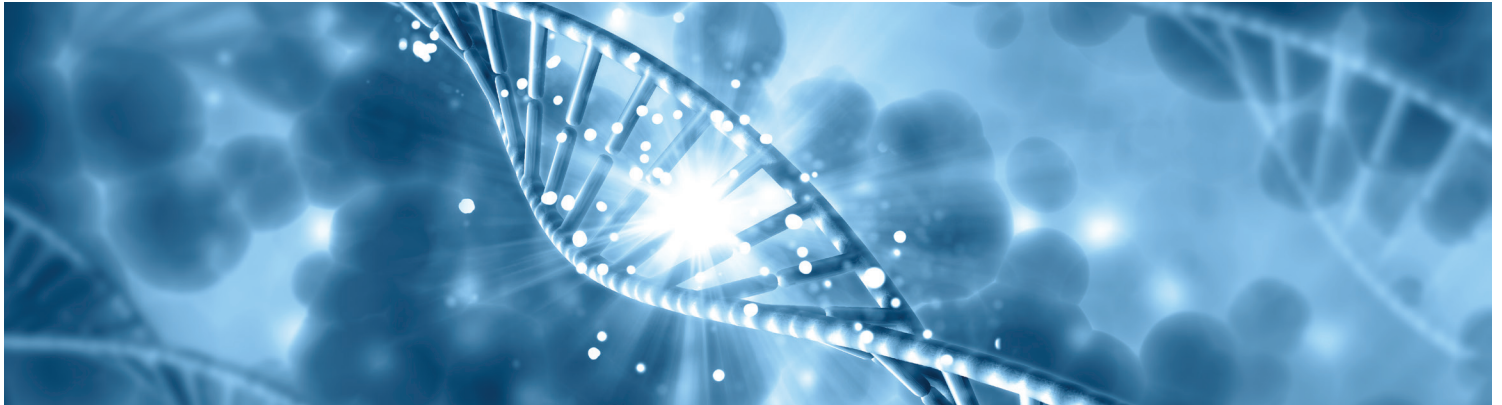


Exon Level Deletion and Duplication Analysis



BACKGROUND

Exon level deletion and duplication analysis is a new addition to the genetic testing portfolio at DDC Clinic Molecular Diagnostics Laboratory, capable of detecting intragenic Copy Number Variations (CNV) to increase detection rates.

The assay is designed to cover the whole genome (25,980 genes), with an increased focus on over 7,000 clinically relevant genes. This technology is more sensitive than traditional methods for detecting small deletions and duplications.

Using a high resolution chromosomal microarray platform consisting of approximately 6.85 million probes, we are now able to detect single-exon deletion and duplication events across the human genome.

BENEFITS

Exon level deletion and duplication analysis provides the sensitivity and flexibility required to improve and complement the detection of important genetic variants.

A recent study shows that for certain disease groups, up to 15.6% of genetic defects are caused by small (exon-level) deletions or duplications, which cannot be detected by sequencing analysis or traditional deletion and duplication analysis techniques.

Exon level deletion and duplication analysis can assist in diagnosing patients and increase detection rates, providing answers to patients and families and helping to improve patient outcomes.

HOW TO ORDER

Deletion and duplication analysis of any gene is available using highly sensitive microarray technology. We now offer reflex, concurrent, or standalone testing for the following:

- ANY clinically relevant gene if previous testing has not detected a mutation or if a gene of interest has a high proportion of known disease causing deletions or duplications
- Panel testing alongside established Next Generation Sequencing (NGS) Panels to increase detection and optimize turnaround time

Please refer to our website (www.DDCCliniclab.org) for additional information.

Specimen

Requires 3-5 ml whole blood or cord blood in a lavender top EDTA tube (2-3 ml for infants and children under two). Sample should be refrigerated until shipped. Ship by overnight delivery or courier, protecting sample from temperature extremes.

Alternate Specimens: please contact lab to coordinate.

Turn Around Time

2-4 weeks

Price

Single gene: standalone \$750, with NGS \$650
2-10 genes: standalone \$1250, with NGS \$1,050
10+ genes: standalone \$1750, with NGS \$1,450