Complex phenotypes lead to challenging diagnoses

Whole Exome Sequencing (WES) analyzes thousands of genes simultaneously to investigate the molecular basis of genetic disorders, shortening the diagnostic odyssey often associated with single-gene tests or multi-gene panels.

**WES is recommended for patients with:**

- Heterogeneous phenotypes, such as:
  - Intellectual disability
  - Epilepsy
  - Bone and connective tissue disorders
  - Complex dysmorphic features
  - Developmental delay
  - Muscular dystrophy
  - Immunodeficiency
  - Neuropathy
  - Deafness
  - Retinitis pigmentosa
  - Cardiomyopathy
  - Metabolic disorders

- Atypical or complex findings that may represent a new or rare syndrome
- A suspected genetic disorder where previous testing has been uninformative

**Flexible Test Options Tailored to Clinical Indication and Family History**

<table>
<thead>
<tr>
<th>Features</th>
<th>Methodology</th>
<th>Coverage*</th>
<th>Genomic Regions Covered</th>
<th>Read Depth</th>
<th>Turn-around Time**</th>
<th>Deletion/Duplication via aCGH (&gt;25kb for copy number gain and &gt;200kb for copy number loss)</th>
<th>Mitochondrial genome (&gt;1000x read depth)</th>
<th>Prenatal Testing**</th>
<th>Available as Singleton (solo) or Trio***</th>
<th>Availability of Research Report</th>
<th>Availability of Raw Data****</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Agilent SureSelect Human All Exon V6 capture kit (~60mb)</td>
<td>97-98% of targeted bases covered at minimum 10x</td>
<td>Exons, deep intronic mutations, 10bp exon/intron boundaries</td>
<td>100x average read depth</td>
<td>4-6 weeks</td>
<td>Available as an add-on</td>
<td>Available as an add-on</td>
<td>Available with rapid turn-around time</td>
<td>Yes</td>
<td>Available on request</td>
<td>Yes</td>
</tr>
</tbody>
</table>

* Exomes do not target all exons within the human genome and ~2.3% of the targeted exons may not be well covered due to various technical reasons (Choi M, et al. (2009) Proc Natl Acad Sci. Yang Y, et al. (2013) N Engl J Med). These missing regions are not backfilled by Sanger; however, we will provide coverage statistics about targeted genes. ** Rapid turn-around time of 2-3 weeks for prenatal testing is available at an additional cost. *** The chance of identifying a causative mutation is maximized when exome testing is performed for both the index patient and parents or other first-degree relatives. This trio approach improves the diagnostic rate by facilitating variant analysis. **** .fastq and .bam files are available for 30 days after report delivery and .vcf files are available indefinitely.

Visit www.lifelabsgenetics.com to learn more about exome sequencing.
Contact us Ask.Genetics@LifeLabs.com | 1-844-GENEHELP (1-844-363-4357)
Whole Exome Sequencing

Ordering instructions

1. Download the hereditary testing requisition form under the Healthcare Providers tab at LifeLabsGenetics.com

2. The ordering checklist on page 1 can be used to ensure you’ve completed all the necessary pages of the requisition.

3. In the Whole Exome Sequencing (WES) section on page 2 of the requisition form, indicate:
   - Gold or Platinum testing
   - The number of samples
   - Any additional analyses
   - A request for raw data

4. Both pages of the Informed Consent are required to be signed.

Whole Exome Sequencing analyzes the coding regions of >20,000 genes and might identify findings outside of the primary reason for genetic testing.

For all testing, we strongly encourage you to provide the patient’s in-depth medical history to ensure the most comprehensive analysis and interpretation of the genetic testing results. This can be done through any of the following three ways:

- Writing in the Relevant Medical and Family History section on page 1
- Completing the checkboxes on page 5
- Providing the patient’s hospital charts

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