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# Hereditary Disease Testing

Bridge the gap with  
diagnostic accuracy

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# Genetic Testing Tailored To Your Patients

- **Single Genes and Known/Familial Variants** for patients with monogenic diseases or specific variants
- **Fixed Panels** for patients with less heterogeneous or more clearly identifiable conditions
- **Progressive Panels** for patients with a range of clinical indications, all within the same clinical area, and the ability to reflex to a large data set
- **Expanded Panels and Whole Exome Sequencing** for patients with highly heterogeneous phenotypes
- **Whole Genome Sequencing** for patients with undiagnosed diseases

Robust, Reliable,  
and Responsible  
Genetic Testing



Enhanced coverage of targeted bases at appropriate read depth

End-to-end bioinformatics analyses of raw data, including variant identification and interpretation

Extensive and detailed clinical reports

End-to-end support by Canadian geneticists and genetic counsellors

## Flexible Testing Options Based on Clinical Indications and Family History

	Single Genes	Fixed Panels	Progressive Panels	Expanded Panels	Whole Exome Seq	Whole Genome Seq
<b>Genes Covered</b>	1	2-200+	Custom	~6,700	>20,000	>20,000
<b>Coverage</b>	100% of targeted bases covered at >20x	~98% of targeted bases covered at >20x <sup>1</sup>	>99% of targeted bases covered at >10x	95% of targeted bases covered at >20x <sup>2</sup>	97-98% of targeted bases covered at >10x	>99% of targeted bases covered at >10x
<b>Avg. Read Depth</b>	>200x	>150x	30x	80-100x	100x	30x
<b>Turn-Around Time</b>	4-6 weeks from receipt of samples in the testing lab					
<b>Deletion/Duplication</b>	Add-on <sup>3</sup>	Add-on <sup>3,4</sup>	Included <sup>5</sup>	Add-on <sup>6</sup>	Add-on <sup>6</sup>	Included <sup>5</sup>
<b>Mitochondrial Genome</b>	Add-on <sup>7</sup>	Add-on <sup>7</sup>	Included <sup>8</sup>	Add-on <sup>7</sup>	Add-on <sup>7</sup>	Included <sup>8</sup>
<b>Prenatal Testing</b>	Available for most tests - please contact us before sending prenatal samples					
<b>Available as Singleton (solo) or Trio</b>	Proband only				Singleton and Trio	
<b>Availability of Research Report</b>	No	No	No <sup>9</sup>	No	Yes	Yes
<b>Availability of Raw Data<sup>10</sup></b>	No	No	No <sup>9</sup>	Yes	Yes	Yes

<sup>1</sup> Sanger fill-in of missing regions to achieve 100% coverage is available at additional cost; <sup>2</sup> With 100% coverage of targeted bases in >4,000 genes; <sup>3</sup> via MLPA, qPCR, or NGS data; <sup>4</sup> May be included in some panels; <sup>5</sup> via WGS NGS data; <sup>6</sup> via NGS data or aCGH (>25kb for copy number gain and >200kb for copy number loss); <sup>7</sup> via NGS with >1,000x average read depth; <sup>8</sup> Included when requested at the time of ordering; <sup>9</sup> Available after reflex to whole genome data set; <sup>10</sup> Raw data available in fastq, bam, or vcf file formats



Each  
diagnostic  
report is:

Validated by a team of renowned and experienced clinical specialists



Cross-checked against CentoMD®, an extensive and evolving database of known mutations, as well as HGMD and ClinVar



Comprehensive and includes a description of the testing method and a full explanation of results & recommendations

## Variant Reporting

- ✓ All disease-causing variants relating to the phenotype and clinical indications reported by the ordering physician are matched against reported variants in HGMD®, ClinVar, and CentoMD® (classes 1 and 2)
- ✓ All variants with minor allele frequencies of less than 1% in the ExAC database are also considered in the medical interpretation
- ✓ Class 1, 2, and 3 variants are evaluated
- ✓ All relevant inheritance patterns are considered based on the family history and clinical information submitted with the sample(s)

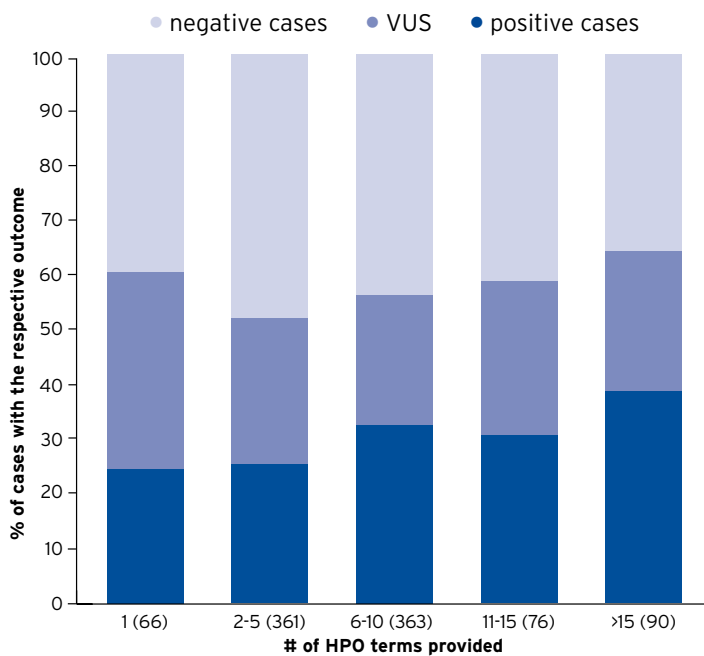
## Incidental Findings

LifeLabs Genetics and Centogene adhere to the ACMG Recommendations for Reporting of Incidental Findings.

- ✓ Patients select whether or not they would like to receive information on the 59 genes or classes of genes outlined in these recommendations, which are known to be medically actionable.
- ✓ Carrier information of the parents for identified incidental findings of the index can be requested with new consent from the parents.
- ✓ Incidental findings are not reported for fetal samples or samples from deceased persons.

## Increase Your Diagnostic Yield

### Clinical Information is Key to Success

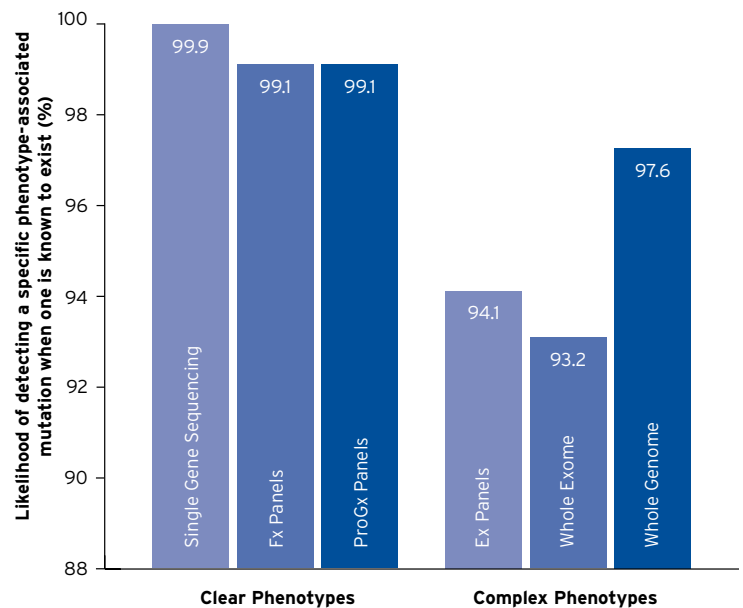


#### The art is in the interpretation

Providing as many Human Phenotype Ontology (HPO) terms as possible will increase the number of positive cases, while reducing the number of VUSes.

This figure has been adapted from Trujillano et al. 2017 EJHG 25:176-182

### Increase Detection With the Right Methodology



#### Proper methodology for the suspected phenotype

Diagnostic accuracies for complex phenotypes, where a disease-associated mutation is known to exist, are higher with advanced genetic testing methodologies.

Figure from Centogene's internal data

## Sample requirements (one of the following):

- ✓ 2 - 8 mL EDTA blood (2 mL for young children, 2 X 4 mL for all others)
- ✓ 1 filter card (10 dried blood spots)
- ✓ 1-10 µg purified DNA
- ✓ Fresh tissue (minimum 20 mg, wash with RPMI (RT), fix by snap-freezing in liquid nitrogen, ship express on dry ice)
- ✓ Tissue in paraffin block-FFPE (minimum 25 - 50 mg, fixed in 10% formalin or 4% paraformaldehyde)
- ✓ Tissue paraffin sections-FFPE (minimum 10 sections @ 5 - 10 µm, fixed in 10% formalin or 4% paraformaldehyde)

# Shorten the diagnostic odyssey with LifeLabs' extensive genetic testing portfolio

## How to order

Diagnostic genetic testing must be ordered by a physician and, given the complexity of results, should only be ordered after genetic counselling by a qualified healthcare provider.

If you have any questions, LifeLabs Genetics has a team of genetic counsellors available to support clinicians throughout the ordering process.

STEP  
**1**



Download and complete the requisition and consent forms from the LifeLabs Genetics website at: [www.lifelabsgenetics.com](http://www.lifelabsgenetics.com).

Please note that detailed clinical information, including a detailed family history and pedigree, is *mandatory* and that omitting this information may impact test results and their interpretation.

STEP  
**2**



Apply for funding from your province's Ministry of Health, if applicable. Please contact us for support.

STEP  
**3**



Collect and send sample(s) to LifeLabs or send your patient to any LifeLabs Patient Service Centre for sample collection. Prenatal and tissue samples should be sent directly to Centogene. Please contact us before sending prenatal samples.

Visit [www.lifelabsgenetics.com](http://www.lifelabsgenetics.com) to learn more about our testing options.  
Contact us at [Ask.Genetics@LifeLabs.com](mailto:Ask.Genetics@LifeLabs.com) | 1-84-GENEHELP (1-844-363-4357)

*LifeLabs Genetics maintains IQMH, ISO 15189, and CAP accreditation, allowing us to put the power of genetic testing on your side with accurate results, expert advice, and ongoing support.*

*LifeLabs Genetics has partnered with Centogene, a CAP, CLIA, and ISO accredited laboratory and global leader in the field of diagnostic testing for rare hereditary diseases, to offer high-quality genetic testing to Canadians.*