Integrated GENETICS
LabCorp Specialty Testing Group

# DLD, FHI, Nemaline Myopathy, Usher IF, Usher III, Joubert 2, Walker-Warburg

## SAMPLE REPORT

**Patient Name:**
**Referring Physician:**
**Specimen #:** **Client #:**
**Patient ID#:** **Case#:**

**City Hospital**
**123 City Avenue**
**Anywhere, ST 12345**

DOB: 00/00/1987
Sex:

Date Collected: 4/26/13
Date Received: 4/27/13
Lab ID #:
Hospital ID #:
Specimen Type: Peripheral blood

**Ethnicity:** Ashkenazi Jewish

**Indication:** Carrier test / No family history

| Disease | Result | Interpretation |
|---|---|---|
| Dihydrolipoamide Dehydrogenase Deficiency | Positive for one copy of the c.685G>T (p.G229C) mutation | CARRIER. Genetic counseling is recommended. See Comments. |
| Familial Hyperinsulinism | Negative | Carrier risk reduced from 1/52 (1.9%) to 1/1700 (0.1%). |
| Joubert Syndrome 2 | Negative | Carrier risk reduced from 1/92 (1.1%) to 1/9100 (0.01%). |
| Nemaline Myopathy | Negative | Carrier risk reduced from 1/168 (0.6%) to <1/3341 (0.03%). |
| Usher Syndrome Type IF | Negative | Carrier risk reduced from 1/147 (0.7%) to <1/585 (0.2%). |
| Usher Syndrome Type III | Negative | Carrier risk reduced from 1/120 (0.8%) to 1/5951 (0.02%). |
| Walker-Warburg Syndrome | Negative | Carrier risk reduced from 1/79 (1.3%) to 1/7800 (0.01%). |

**COMMENTS:**
Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of these results, as well as recommendations for testing family members and, when applicable, this individual's partner.

This analysis does not rule out the presence of disease-causing mutations in other regions of the genes analyzed or in other genes, and does not detect germline mosaicism. This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of these conditions. Unless otherwise noted, interpretations are based on a negative family history and the absence of symptoms.

If additonal carrier testing has been ordered, results will be reported separately.

Under the direction of:

Date: 5/6/2013 17:19

(REPORT CONTINUED…)

# SAMPLE REPORT

## DLD, FHI, Nemaline Myopathy, Usher IF, Usher III, Joubert 2, Walker-Warburg

**Patient Name:** **Referring Physician:** **Specimen #:** **Patient ID#:**

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**METHOD/LIMITATIONS:**

DNA is isolated from the sample, and disease-gene specific regions are amplified by the polymerase chain reaction (PCR). PCR products are subjected to allele-specific primer extension with subsequent hybridization to a bead array and fluorescence detection, and/or are subjected to bi-directional direct DNA sequencing using capillary gel electrophoresis and fluorescence detection. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships, or contamination of a fetal sample with maternal cells.

The test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment, when available.

### Carrier Frequency and Risk Reductions for Ashkenazi Jewish* Individuals with No Family History

| Disease Name (Gene Name) | Reference Sequence: Mutations Analyzed | Carrier Frequency (of mutations analyzed) | Detection Rate | Detection Rate References |
|---|---|---|---|---|
| Dihydrolipoamide Dehydrogenase Deficiency (*DLD*) | NM_000108.3: c.104dupA (p.Y35X), c.685G>T (p.G229C) | 1 in 107 | >95% | Scott SA, et al. Hum Mutat. 2010; 31(11):1240-1250. |
| Familial Hyperinsulinism (*ABCC8*) | NM_000352.3: c.4160_4162delTCT (p.F1387del), c.3989-9G>A | 1 in 52 | 97% | Glaser B, et al. Genet Med. 2011; 13(10):891-894. |
| Joubert Syndrome Type 2 *(TMEM216)* | NM_001173990.1: c.218G>T (p.R73L) | 1 in 92 | 99% | Edvardson S, et al. Am J Hum Genet. 2010; 86: 93-97. |
| Nemaline Myopathy (*NEB*) | NM_004543.4: c.7432-2025_7536 +372del2502bp (p.R2478_D2512del35) | 1 in 168 | >95% | Scott SA, et al. Hum Mutat. 2010; 31(11):1240-1250. |
| Usher Syndrome Type IF (*PCDH15*) | NM_033056.3: c.733C>T (p.R245X) | 1 in 147 | >75% | Scott SA, et al. Hum Mutat. 2010; 31(11):1240-1250. |
| Usher Syndrome Type III (*CLRN1 aka USH3A*) | NM_174878.2: c.144T>G (p.N48K) | 1 in 120 | 98% | Ness SL, et al. J Med Genet. 2003; 40:767-772. Fields RR, et al. Am J Hum Genet. 2002; 71:607-617. |
| Walker-Warburg Syndrome *(FKTN)* | NM_001079802.1: c.1167dupA (p.F390fs) | 1 in 79** | 99% | Manzini MC, et al. Hum Mutat. 2008; 29(11): E231-E241. Chang W, et al. Prenat Diagn. 2009; 29:560-569. |

* The carrier frequency and detection rate in the non-Ashkenazi Jewish population are expected to be low.

** Based on analysis of >2000 Ashkenazi Jewish carrier screening results

Under the direction of:

Date: 5/6/2013 17:19

Molecular Testing Performed at Esoterix Genetic Laboratories, LLC 3400 Computer Drive Westborough, MA 01581 1-800-255-7357