LASTNAME, FIRSTNAME

Patient ID:

Specimen ID: **000-000-0000-0**

DOB: **mm/dd/yyyy**

Age: **00**

Sex: **Female**

Account Number: **00000000**

Ordering Physician:

labcorp

Date Collected: **mm/dd/yyyy**	Date Received: **mm/dd/yyyy**	Date Reported: **mm/dd/yyyy**	Date Entered: **mm/dd/yyyy**

Specimen Type: **Whole Blood**

Ethnicity: **Not Provided**

Indication: **Carrier Test / Screening**

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SAMPLE REPORT

Summary: ■ POSITIVE

Variants Detected

Disorder *(Gene)*	Result	Interpretation
Fragile X syndrome *(FMR1)* NM_002024.6	POSITIVE: PREMUTATION CARRIER PCR repeats: 29 and 56 Premutation allele AGG interruption(s): 1 Haplotype:(CGG)25 AGG (CGG)31	Premutation carrier of fragile X syndrome. This individual may be at risk for primary ovarian insufficiency and late-onset fragile X-associated tremor/ataxia syndrome (FXTAS), and for having children with fragile X syndrome. **Risk:** AT INCREASED RISK FOR AFFECTED PREGNANCY. Risk of expansion to full mutation in offspring is <1% (Domniz, PMID:30619448). Genetic counseling is recommended.
Gaucher disease *(GBA1)* NM_001005741.3	POSITIVE: CARRIER Heterozygous for c.1226A>C (p.Asn409Thr), pathogenic, CHR1:155205634.	Predicted to be a carrier. Carriers may be at risk for Parkinsonism. **Risk:** AT INCREASED RISK FOR AFFECTED PREGNANCY. If this individual's reproductive partner is also a carrier of a pathogenic variant in the same gene, the risk for an affected fetus is 25%. Genetic counseling and reproductive partner carrier screening is recommended.

Negative Results

Disorder *(Gene)*	Result	Interpretation
Cystic fibrosis *(CFTR)* NM_000492.4	NEGATIVE	This result reduces, but does not eliminate, the risk to be a carrier. **Risk:** NOT at an increased risk for an affected pregnancy.
Spinal muscular atrophy *(SMN1)* NM_000344.4	NEGATIVE 2 copies of *SMN1*; c.*3+80T>G risk variant not present.	This result reduces, but does not eliminate, the risk to be a carrier. **Risk:** NOT at an increased risk for an affected pregnancy.
ALL OTHER DISORDERS	NEGATIVE	This result reduces, but does not eliminate the risk to be a carrier. **Risk:** This individual is NOT at an increased risk for having a pregnancy that is affected with one of the other disorders covered by this test. For partner's gene-specific risks, visit https://womenshealth.labcorp.com.

Recommendations

If the above result is positive, genetic counseling is recommended to discuss the potential clinical and/or reproductive implications, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Labcorp Genetic Counselors please visit https://womenshealth.labcorp.com/genetic-counseling or call (855) GC-CALLS (855-422-2557).

Additional Clinical Information

Fragile X syndrome is an X-linked disorder of intellectual disability with variable severity. Expansions of CGG repeat sequences in the *FMR1* gene account for 99% of variants causing fragile X syndrome. The risk of expansion from a premutation allele of 55-90 repeats to a full mutation in offspring, when transmitted by a carrier female, is reduced with increasing number of AGG interruptions in the CGG repeat sequence (Yrigollen, PMID:22498846; Nolin, PMID:25210937). Greater than 99% of males and approximately 50% of females with the full mutation are intellectually disabled. Other signs and symptoms may include delayed speech and language skills, autism, hyperactivity, developmental delay, increased susceptibility to seizures, macroorchidism in males, a long, narrow face with prominent ears, and joint laxity. Individuals with a premutation do not have fragile X syndrome, but may have an increased risk for fragile X-related disorders. Females may have fragile X-associated primary ovarian insufficiency (FXPOI), which can cause infertility or early menopause. Most males with a premutation and some females are at risk for fragile X-associated tremor and ataxia syndrome (FXTAS),

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Additional Clinical Information (Cont.)

which can affect balance and is associated with tremor and memory problems in older individuals. Treatment is supportive and focuses on educational and behavioral support and management of symptoms. (Santoro, PMID:22017584).

Gaucher disease is an autosomal recessive disorder of variable severity and age at onset, caused by pathogenic variants in the *GBA1* gene. Signs and symptoms may include hepatosplenomegaly, bone disease, cytopenia, and primary neurological involvement in some individuals. Pathogenic variants in *GBA1* may be a risk factor for Parkinsonism. Enzyme replacement therapy and substrate reduction therapy are available for many individuals. Treatment is otherwise supportive. (Sidransky, PMID:19846850; Alcalay, PMID:24756352; Pastores, PMID:20301446; Charrow, PMID:11025794).

Comments

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder(s) tested. Information about the disorder(s) tested is available at https://womenshealth.labcorp.com.

Methods/Limitations

Next-generation Sequencing (NGS): Genomic regions of interest are selected using the Twist Bioscience® hybridization capture method and sequenced via the Illumina® NGS platform. Sequencing reads are aligned to the human genome reference GRCh37/hg19 build. Regions of interest include coding exons, intron/exon junctions (typically +/- 20 nucleotides) and additional genomic regions with known significant pathogenic variants. Estimated analytical sensitivity is >99% for single nucleotide variants and insertions/deletions <45 base pairs. Regions with low NGS coverage are selected for Sanger sequencing based on analytical sensitivity and probability of pathogenic variant(s). QIAGEN CLC Genomics and in-house algorithms identify copy number variants (CNVs) by comparing normalized read depth for each target in the region of interest with a set of clinical control samples or to the median read depth across the samples within the same NGS run. The overall analytical sensitivity for CNV detection in *CFTR* and *DMD* is >99% and, based on simulation studies, the estimated analytical sensitivity for single exon deletions is >97% and for single exon duplications is >82%. For all other genes, the assay is designed to detect CNVs with genomic size >10 kb and typically involving two or more consecutive coding exons with an overall analytical sensitivity of 96.7%. Large single-exon deletions or duplications may be detected. Precise breakpoints are not reported. Confirmatory testing by orthogonal technologies may include Sanger sequencing, MLPA, gap PCR, or low coverage whole genome sequencing analysis.

If the following genes are included in this test, these analysis restrictions are applied: *F2* includes c.*97G>A variant only (also known as 20210G>A); *F5* includes the F5 Leiden c.1601G>A (p.Arg543Gln) variant only (also known as R506Q); *GJB2* analysis includes deletions involving the 5' end of *GJB6* and regulatory elements of *GJB2* that result in reduced *GJB2* expression; *HFE* includes full gene deletion and five variants: c.187C>G (p.His63Asp), c.502G>T (p.Glu168X), c.506G>A (p.Trp169X), c.845G>A (p.Cys282Tyr), c.1006+1G>A; *NEB* excludes exons 82-105; *TNXB* excludes exons 32-44. Regions not included in CNV analysis: *BBS9* exons 15-17, *CEP290* gene, *CORO1A* exon 11, *TMEM231* exons 2-3. Copy number gains cannot be detected in *PLN* and *RMRP*. Regions that may have lower analytical sensitivity due to intrinsic sequence properties: *ARX* exon 2, *RPGR* exon 13, *RPS6KA3* exon 20, *SELENON* exon1 and *SLC6A8* exon 1.

Reported variants: Pathogenic and likely pathogenic variants are reported for all tests. Benign and likely benign variants are typically not reported. Variants of uncertain significance are reported when included in the test specification. Variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, http://www.hgvs.org/). Variant classification and confirmation are consistent with ACMG standards and guidelines (Richards, PMID:25741868; Rehm, PMID:23887774). Detailed variant classification information and reevaluation are available upon request.

Alpha thalassemia: Analysis of the alpha-globin (HBA) gene cluster is performed by NGS. Positive results are confirmed by MLPA, gap PCR, or Sanger sequencing. There are two alpha-globin genes in the HBA gene cluster, *HBA1* and *HBA2*. Typically, an individual with a normal genotype has these two genes on each chromosome (alpha alpha/alpha alpha). A deletion that removes two of the genes on one of the chromosomes is described as - -/alpha alpha. Alpha-globin variants included in the analysis are the Constant Spring non-deletion variant and the following deletions: -alpha3.7, -alpha4.2, --alpha20.5, --SEA, --FIL, --THAI, --MED, and the HS-40 regulatory region. This analysis does not detect other variants in the alpha-globin genes or variants in the beta-globin gene and may not detect the co-occurrence of a deletion and a duplication. Analytical sensitivity is estimated to be >98% for the targeted variants.

Congenital Adrenal Hyperplasia: Analysis is performed by NGS. This analysis detects most large deletions within the *CYP21A2* gene, as well as the presence of seven of the most common pathogenic variants in the gene: c.518T>A (p.Ile173Asn), exon 6 mutation cluster [c.713T>A (p.Val238Glu), c.719T>A (p.Met240Lys)], c.923dup (p.Leu308Phefs), c.293-13C/A>G, c.332_339delGAGACTAC (p.Gly111Valfs), and c.-113G>A. MLPA is used to confirm all targeted variants identified by next-generation sequencing. Other variants are not detected by this analysis. Co-existence of a small variant with a large deletion may not be detected. The analytical sensitivity of this assay is estimated to be >99%.

Dystrophinopathies, including Duchenne and Becker muscular dystrophies and dilated cardiomyopathy: Analysis is performed by NGS. The overall analytical sensitivity for CNV detection in *DMD* is >99% and, based on simulation studies, the estimated analytical sensitivity for single exon deletions is >97% and for single exon duplications is >82%. In-frame and out-of-frame deletions cannot be distinguished by this analysis, which does not determine precise breakpoints in the *DMD* gene. Approximately 67% of the time a *DMD* pathogenic variant is inherited, and approximately 33% of the time the variant is *de novo* and not previously seen in the family. If a pathogenic variant is *de novo*, the risk that the mother of an affected male has germline mosaicism is 15-20%. This analysis does not detect germline mosaicism. An individual who has a negative carrier screen may have germline mosaicism and be at risk for having an affected child.

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Methods/Limitations (Cont.)

Fragile X Syndrome: PCR analysis with fluorescent primers is used to amplify the CGG repeat region of the FMR1 gene and Amelogenin loci on the X and Y chromosomes. PCR products are sized using capillary electrophoresis and GeneMapper fragment analysis software. The reportable CGG repeat ranges are: negative: <45; intermediate: 45-54; premutation: 55-200; full mutation: >200. Postnatal female samples with premutations of 55-90 repeats are reflexed to assess AGG interruptions within the CGG repeats. The predicted risk for the premutation to expand to a full mutation in the next generation is based on the size of the premutation and the number of AGG interruptions. If indicated, methylation PCR analysis using methylation-specific immunoprecipitation is performed. Relative amounts of methylated and unmethylated products are assessed to determine the methylation status: unmethylated, partially methylated, or completely methylated. The analytical sensitivity of this assay for the detection of expanded alleles in the *FMR1* gene is estimated to be >99%. Reproducibility of repeat numbers is typically ±1 for alleles containing up to 60 repeats, ±3 for alleles with 61-119 repeats, and ±10 for alleles with >119 repeats. Low levels of mosaicism and *FMR1* variants unrelated to trinucleotide expansion are not detected by this assay.

Spinal muscular atrophy: The copy number of *SMN1* exon 7 is assessed relative to internal standard reference genes by quantitative polymerase chain reaction (qPCR). A mathematical algorithm calculates 0, 1, 2 and 3 copies with statistical confidence. In fetal specimens and specimens with 0 or 1 copies, the primer and probe binding sites are sequenced to rule out variants that could interfere with copy number analysis. *SMN2* copy number is assessed by digital droplet PCR analysis relative to an internal standard reference gene in samples with no copies of *SMN1*. For carrier screening, when two copies of *SMN1* are detected, allelic discrimination qPCR targeting c.*3+80T>G in *SMN1* is performed.

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants, or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships.

References

Gregg AR, Aarabi M, Klugman S *et al*. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 23, 1793 (2021). PMID: 34285390

Disorders Tested

3-Methylcrotonyl-CoA carboxylase deficiency (2 genes). Autosomal recessive: *MCCC1*, *MCCC2*
3M syndrome (3 genes). Autosomal recessive: *CCDC8*, *CUL7*, *OBSL1*
Abetalipoproteinemia (1 gene). Autosomal recessive: *MTTP*
Achromatopsia (1 gene). Autosomal recessive: *CNGB3*
Acrodermatitis enteropathica (1 gene). Autosomal recessive: *SLC39A4*
Acute infantile liver failure (3 genes). Autosomal recessive: *LARS1*, *NBAS*, *TRMU*
Adenosine deaminase deficiency (1 gene). Autosomal recessive: *ADA*
Adrenoleukodystrophy, X-linked (1 gene). X-linked: *ABCD1*. Males are not tested for X-linked disorders.
Agammaglobulinemia, X-linked (1 gene). X-linked: *BTK*. Males are not tested for X-linked disorders.
Aicardi-Goutières syndrome (4 genes). Autosomal recessive: *RNASEH2A*, *RNASEH2B*, *RNASEH2C*, *SAMHD1*
Allan-Herndon-Dudley syndrome (1 gene). X-linked: *SLC16A2*. Males are not tested for X-linked disorders.
Alpha-mannosidosis (1 gene). Autosomal recessive: *MAN2B1*
Alpha-thalassemia (2 genes). Autosomal recessive: *HBA1/HBA2*
Alpha-thalassemia X-linked intellectual disability syndrome (1 gene). X-linked: *ATRX*. Males are not tested for X-linked disorders.
Alport syndrome (3 genes). Autosomal recessive: *COL4A3*, *COL4A4*; X-linked: *COL4A5*. Males are not tested for X-linked disorders.
Alström syndrome (1 gene). Autosomal recessive: *ALMS1*
Andermann syndrome (1 gene). Autosomal recessive: *SLC12A6*
Arginase deficiency (1 gene). Autosomal recessive: *ARG1*
Argininosuccinic aciduria (1 gene). Autosomal recessive: *ASL*
Aromatic l-amino acid decarboxylase deficiency (1 gene). Autosomal recessive: *DDC*
Arterial tortuosity syndrome (1 gene). Autosomal recessive: *SLC2A10*
Arthrogryposis, mental retardation, and seizures (AMRS) (1 gene). Autosomal recessive: *SLC35A3*
Asparagine synthetase deficiency (1 gene). Autosomal recessive: *ASNS*
Aspartylglucosaminuria (1 gene). Autosomal recessive: *AGA*
Ataxia with vitamin E deficiency (1 gene). Autosomal recessive: *TTPA*
Ataxia-telangiectasia (1 gene). Autosomal recessive: *ATM*
ATP7A-related copper transport disorders, includes Menkes syndrome (1 gene). X-linked: *ATP7A*. Males are not tested for X-linked disorders.
Atransferrinemia (1 gene). Autosomal recessive: *TF*

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Disorders Tested (Cont.)

Autoimmune polyglandular syndrome type 1 (1 gene). Autosomal recessive: *AIRE*
Autosomal recessive congenital ichthyosis (ARCI) (11 genes). Autosomal recessive: *ABCA12*, *ALOX12B*, *ALOXE3*, *CERS3*, *CYP4F22*, *LIPN*, *NIPAL4*, *PNPLA1*, *SDR9C7*, *SLC27A4*, *TGM1*
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (AR*SACS*) (1 gene). Autosomal recessive: *SACS*
Axonal neuropathy with neuromyotonia (1 gene). Autosomal recessive: *HINT1*
Bardet-Biedl syndrome (12 genes). Autosomal recessive: *ARL6*, *BBS1*, *BBS10*, *BBS12*, *BBS2*, *BBS4*, *BBS5*, *BBS7*, *BBS9*, *MKKS*, *SDCCAG8*, *TTC8*
Bare lymphocyte syndrome type II (4 genes). Autosomal recessive: *CIITA*, *RFX5*, *RFXANK*, *RFXAP*
Barth syndrome (1 gene). X-linked: *TAFAZZIN*. Males are not tested for X-linked disorders.
Bartter syndrome (3 genes). Autosomal recessive: *BSND*, *KCNJ1*, *SLC12A1*
Basal ganglia disease, biotin-thiamine-responsive (1 gene). Autosomal recessive: *SLC19A3*
Beta-hemoglobinopathies, includes sickle cell disease and beta-thalassemias (1 gene). Autosomal recessive: *HBB*
Beta-ketothiolase deficiency (1 gene). Autosomal recessive: *ACAT1*
Beta-mannosidosis (1 gene). Autosomal recessive: *MANBA*
Biotinidase deficiency, profound and partial (1 gene). Autosomal recessive: *BTD*
Bloom syndrome (1 gene). Autosomal recessive: *BLM*
Brittle cornea syndrome (2 genes). Autosomal recessive: *PRDM5*, *ZNF469*
Canavan disease (1 gene). Autosomal recessive: *ASPA*
Carbamoyl phosphate synthetase I deficiency (1 gene). Autosomal recessive: *CPS1*
Carnitine palmitoyltransferase I deficiency (1 gene). Autosomal recessive: *CPT1A*
Carnitine palmitoyltransferase II deficiency (1 gene). Autosomal recessive: *CPT2*
Carnitine-acylcarnitine translocase deficiency (1 gene). Autosomal recessive: *SLC25A20*
Carpenter syndrome (1 gene). Autosomal recessive: *RAB23*
Cartilage-hair hypoplasia (1 gene). Autosomal recessive: *RMRP*
Cerebellar hypoplasia, *VLDLR*-associated (1 gene). Autosomal recessive: *VLDLR*
Cerebral creatine deficiency syndromes (3 genes). Autosomal recessive: *GAMT*, *GATM*; X-linked: *SLC6A8*. Males are not tested for X-linked disorders.
Cerebrotendinous xanthomatosis (1 gene). Autosomal recessive: *CYP27A1*
Chediak-Higashi syndrome (1 gene). Autosomal recessive: *LYST*
Choreacanthocytosis (1 gene). Autosomal recessive: *VPS13A*
Chronic granulomatous disease (4 genes). Autosomal recessive: *CYBA*, *NCF2*, *NCF4*; X-linked: *CYBB*. Males are not tested for X-linked disorders.
Ciliopathies (2 genes). Autosomal recessive: *CEP290*, *MKS1*
Citrullinemia (2 genes). Autosomal recessive: *ASS1*, *SLC25A13*
Coats plus syndrome and dyskeratosis congenita, *CTC1*-related (1 gene). Autosomal recessive: *CTC1*
Cockayne syndrome (2 genes). Autosomal recessive: *ERCC6*, *ERCC8*
Coffin-Lowry syndrome (1 gene). X-linked: *RPS6KA3*. Males are not tested for X-linked disorders.
Cohen syndrome (1 gene). Autosomal recessive: *VPS13B*
Cold-induced sweating syndrome, includes Crisponi syndrome (2 genes). Autosomal recessive: *CLCF1*, *CRLF1*
Combined malonic and methylmalonic aciduria (1 gene). Autosomal recessive: *ACSF3*
Combined oxidative phosphorylation deficiency (2 genes). Autosomal recessive: *GFM1*, *TSFM*
Combined pituitary hormone deficiency (2 genes). Autosomal recessive: *LHX3*, *PROP1*
Congenital adrenal hyperplasia (7 genes). Autosomal recessive: *CYP11A1*, *CYP11B1*, *CYP17A1*, *CYP21A2*, *HSD3B2*, *POR*, *STAR*
Congenital adrenal hypoplasia, X-linked (1 gene). X-linked: *NR0B1*. Males are not tested for X-linked disorders.
Congenital amegakaryocytic thrombocytopenia (1 gene). Autosomal recessive: *MPL*
Congenital arthrogryposis with anterior horn cell disease (1 gene). Autosomal recessive: *GLE1*
Congenital disorder of deglycosylation (1 gene). Autosomal recessive: *NGLY1*
Congenital disorders of glycosylation (4 genes). Autosomal recessive: *ALG1*, *ALG6*, *MPI*, *PMM2*
Congenital generalized lipodystrophy (2 genes). Autosomal recessive: *AGPAT2*, *CAVIN1*
Congenital hydrocephalus 1 (1 gene). Autosomal recessive: *CCDC88C*
Congenital insensitivity to pain with anhidrosis (1 gene). Autosomal recessive: *NTRK1*
Congenital myasthenic syndrome (6 genes). Autosomal recessive: *CHAT*, *CHRNE*, *COLQ*, *DOK7*, *GFPT1*, *RAPSN*
Corneal dystrophy and perceptive deafness (1 gene). Autosomal recessive: *SLC4A11*
Costeff syndrome (1 gene). Autosomal recessive: *OPA3*
Cutis laxa (5 genes). Autosomal recessive: *ATP6V0A2*, *ATP6V1E1*, *EFEMP2*, *LTBP4*, *PYCR1*
Cystic fibrosis (1 gene). Autosomal recessive: *CFTR*
Cystinosis (1 gene). Autosomal recessive: *CTNS*
D-bifunctional protein deficiency (1 gene). Autosomal recessive: *HSD17B4*
Danon disease (1 gene). X-linked: *LAMP2*. Males are not tested for X-linked disorders.

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Disorders Tested (Cont.)

Deafness and hearing loss, nonsyndromic (5 genes). Autosomal recessive: *GJB2*, *LOXHD1*, *OTOF*, *SYNE4*; X-linked: *POU3F4*. Males are not tested for X-linked disorders.
Dent disease (2 genes). X-linked: *CLCN5*, *OCRL*. Males are not tested for X-linked disorders.
Developmental and epileptic encephalopathy (3 genes). Autosomal recessive: *CAD*, *ITPA*; X-linked: *ARX*. Males are not tested for X-linked disorders.
Dihydrolipoamide dehydrogenase deficiency (1 gene). Autosomal recessive: *DLD*
Dihydropyrimidine dehydrogenase deficiency (1 gene). Autosomal recessive: *DPYD*
Distal spinal muscular atrophy (1 gene). Autosomal recessive: *PLEKHG5*
Donnai-Barrow syndrome (1 gene). Autosomal recessive: *LRP2*
Dystrophic epidermolysis bullosa (1 gene). Autosomal recessive: *COL7A1*
Dystrophinopathies, including Duchenne and Becker muscular dystrophy and X-linked cardiomyopathy (1 gene). X-linked: *DMD*. Males are not tested for X-linked disorders.
Ehlers Danlos syndrome, *ADAMTS2*-related (1 gene). Autosomal recessive: *ADAMTS2*
Ehlers–Danlos-like syndrome (1 gene). Autosomal recessive: *TNXB*
Ellis-van Creveld syndrome (1 gene). Autosomal recessive: *EVC2*
Emery-Dreifuss muscular dystrophy (2 genes). X-linked: *EMD*, *FHL1*. Males are not tested for X-linked disorders.
Ethylmalonic encephalopathy (1 gene). Autosomal recessive: *ETHE1*
Fabry disease (1 gene). X-linked: *GLA*. Males are not tested for X-linked disorders.
Factor IX deficiency (hemophilia B) (1 gene). X-linked: *F9*. Males are not tested for X-linked disorders.
Familial dysautonomia (1 gene). Autosomal recessive: *ELP1*
Familial hemophagocytic lymphohistiocytosis (4 genes). Autosomal recessive: *PRF1*, *STX11*, *STXBP2*, *UNC13D*
Familial hyperinsulinism (1 gene). Autosomal recessive: *ABCC8*
Familial Mediterranean fever (1 gene). Autosomal recessive: *MEFV*
Fanconi anemia (10 genes). Autosomal recessive: *BRIP1*, *FANCA*, *FANCC*, *FANCD2*, *FANCE*, *FANCF*, *FANCG*, *FANCI*, *FANCL*; X-linked: *FANCB*. Males are not tested for X-linked disorders.
Fragile X syndrome (1 gene). X-linked: *FMR1*. Males are not tested for X-linked disorders.
Fraser syndrome (3 genes). Autosomal recessive: *FRAS1*, *FREM2*, *GRIP1*
Fucosidosis (1 gene). Autosomal recessive: *FUCA1*
Galactosemia (3 genes). Autosomal recessive: *GALE*, *GALK1*, *GALT*
Galactosialidosis (1 gene). Autosomal recessive: *CTSA*
Gaucher disease (1 gene). Autosomal recessive: *GBA1*
Glutaric acidemia type I (1 gene). Autosomal recessive: *GCDH*
Glutaric acidemia type II (3 genes). Autosomal recessive: *ETFA*, *ETFB*, *ETFDH*
Glutathione synthetase deficiency (1 gene). Autosomal recessive: *GSS*
Glycine encephalopathy (2 genes). Autosomal recessive: *AMT*, *GLDC*
Glycogen storage disease type I (2 genes). Autosomal recessive: *G6PC1*, *SLC37A4*
Glycogen storage disease type III (1 gene). Autosomal recessive: *AGL*
Glycogen storage disease type IV (1 gene). Autosomal recessive: *GBE1*
Glycogen storage disease type IX (4 genes). Autosomal recessive: *PHKB*, *PHKG2*; X-linked: *PHKA1*, *PHKA2*. Males are not tested for X-linked disorders.
Glycogen storage disease type V (1 gene). Autosomal recessive: *PYGM*
Glycogen storage disease type VII (1 gene). Autosomal recessive: *PFKM*
GM1 gangliosidosis and mucopolysaccharidosis type IVB (1 gene). Autosomal recessive: *GLB1*
GRACILE syndrome (1 gene). Autosomal recessive: *BCS1L*
Gyrate atrophy of choroid and retina (1 gene). Autosomal recessive: *OAT*
Hepatic venoocclusive disease with immunodeficiency (1 gene). Autosomal recessive: *SP110*
Hereditary folate malabsorption (1 gene). Autosomal recessive: *SLC46A1*
Hereditary fructose Intolerance (1 gene). Autosomal recessive: *ALDOB*
Hereditary spastic paraplegia (4 genes). Autosomal recessive: *CYP7B1*, *SPG11*, *SPG21*, *TECPR2*
Hermansky-Pudlak syndrome (9 genes). Autosomal recessive: *AP3B1*, *BLOC1S3*, *BLOC1S6*, *DTNBP1*, *HPS1*, *HPS3*, *HPS4*, *HPS5*, *HPS6*
HMG-CoA lyase deficiency (1 gene). Autosomal recessive: *HMGCL*
Holocarboxylase synthetase deficiency (1 gene). Autosomal recessive: *HLCS*
Homocystinuria (2 genes). Autosomal recessive: *CBS*, *MTRR*
HSD10 disease (1 gene). X-linked: *HSD17B10*. Males are not tested for X-linked disorders.
Hyaline fibromatosis syndrome (1 gene). Autosomal recessive: *ANTXR2*
Hydrolethalus syndrome (1 gene). Autosomal recessive: *HYLS1*
Hyper IgM syndrome, X-linked (1 gene). X-linked: *CD40LG*. Males are not tested for X-linked disorders.
Hypohidrotic ectodermal dysplasia (1 gene). X-linked: *EDA*. Males are not tested for X-linked disorders.
Hypomyelination and congenital cataract (1 gene). Autosomal recessive: *HYCC1*

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Disorders Tested (Cont.)

Hypophosphatasia (1 gene). Autosomal recessive: *ALPL*
Immunodeficiency with hyper IgM syndrome (3 genes). Autosomal recessive: *AICDA*, *CD40*, *UNG*
Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome (4 genes). Autosomal recessive: *CDCA7*, *DNMT3B*, *HELLS*, *ZBTB24*
Inclusion body myopathy 2 (1 gene). Autosomal recessive: *GNE*
Intellectual developmental disorder, *NONO*-related (1 gene). X-linked: *NONO*. Males are not tested for X-linked disorders.
IPEX syndrome (1 gene). X-linked: *FOXP3*. Males are not tested for X-linked disorders.
Isovaleric acidemia (1 gene). Autosomal recessive: *IVD*
Joubert syndrome and related disorders, including Meckel-Gruber syndrome (20 genes). Autosomal recessive: *AHI1*, *ARL13B*, *B9D1*, *B9D2*, *CC2D2A*, *CEP104*, *CPLANE1*, *INPP5E*, *KIF14*, *NPHP1*, *NPHP3*, *RPGRIP1L*, *TCTN1*, *TCTN2*, *TCTN3*, *TMEM138*, *TMEM216*, *TMEM231*, *TMEM237*, *TMEM67*
Junctional epidermolysis bullosa (3 genes). Autosomal recessive: *LAMA3*, *LAMB3*, *LAMC2*
Juvenile hereditary hemochromatosis (2 genes). Autosomal recessive: *HAMP*, *HJV*
Juvenile retinoschisis, X-linked (1 gene). X-linked: *RS1*. Males are not tested for X-linked disorders.
Krabbe disease (1 gene). Autosomal recessive: *GALC*
L1 syndrome (1 gene). X-linked: *L1CAM*. Males are not tested for X-linked disorders.
Leber congenital amaurosis (9 genes). Autosomal recessive: *AIPL1*, *LCA5*, *LRAT*, *RD3*, *RDH12*, *RPE65*, *RPGRIP1*, *SPATA7*, *TULP1*
Leigh syndrome (10 genes). Autosomal recessive: *FBXL4*, *FOXRED1*, *LRPPRC*, *NDUFAF2*, *NDUFAF5*, *NDUFS4*, *NDUFS6*, *NDUFS7*, *NDUFV1*, *SURF1*
Leukoencephalopathy with vanishing white matter (5 genes). Autosomal recessive: *EIF2B1*, *EIF2B2*, *EIF2B3*, *EIF2B4*, *EIF2B5*
Limb-girdle muscular dystrophy (12 genes). Autosomal recessive: *CAPN3*, *DYSF*, *FKRP*, *POMGNT1*, *POMT1*, *POMT2*, *SGCA*, *SGCB*, *SGCD*, *SGCG*, *TRAPPC11*, *TRIM32*
Lipoprotein lipase deficiency, familial (1 gene). Autosomal recessive: *LPL*
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency (1 gene). Autosomal recessive: *HADHA*
Lysinuric protein intolerance (1 gene). Autosomal recessive: *SLC7A7*
Lysosomal acid lipase deficiency (1 gene). Autosomal recessive: *LIPA*
Macular corneal dystrophy (1 gene). Autosomal recessive: *CHST6*
Maple syrup urine disease (3 genes). Autosomal recessive: *BCKDHA*, *BCKDHB*, *DBT*
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency (1 gene). Autosomal recessive: *ACADM*
Megalencephalic leukoencephalopathy with subcortical cysts type 1 (1 gene). Autosomal recessive: *MLC1*
Metachromatic leukodystrophy (2 genes). Autosomal recessive: *ARSA*, *PSAP*
Methylmalonic acidemia (4 genes). Autosomal recessive: *MCEE*, *MMAA*, *MMAB*, *MMUT*
Methylmalonic acidemia with homocystinuria (5 genes). Autosomal recessive: *ABCD4*, *LMBRD1*, *MMACHC*, *MMADHC*; X-linked: *HCFC1*. Males are not tested for X-linked disorders.
Mevalonate kinase deficiency (1 gene). Autosomal recessive: *MVK*
Microcephaly, postnatal progressive, with seizures and brain atrophy (1 gene). Autosomal recessive: *MED17*
Mitochondrial complex deficiency (1 gene). Autosomal recessive: *SCO2*
Mitochondrial complex I deficiency (1 gene). Autosomal recessive: *ACAD9*
Mitochondrial complex V deficiency (1 gene). Autosomal recessive: *TMEM70*
Mitochondrial DNA depletion syndrome, *MPV17*-related (1 gene). Autosomal recessive: *MPV17*
Mitochondrial DNA depletion syndrome, *TK2*-related (1 gene). Autosomal recessive: *TK2*
Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (1 gene). Autosomal recessive: *PUS1*
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease (1 gene). Autosomal recessive: *TYMP*
Mitochondrial trifunctional protein deficiency (1 gene). Autosomal recessive: *HADHB*
Molybdenum cofactor deficiency (3 genes). Autosomal recessive: *GPHN*, *MOCS1*, *MOCS2*
Mucolipidosis III gamma (1 gene). Autosomal recessive: *GNPTG*
Mucolipidosis type II and III (1 gene). Autosomal recessive: *GNPTAB*
Mucolipidosis type IV (1 gene). Autosomal recessive: *MCOLN1*
Mucopolysaccharidosis type I (1 gene). Autosomal recessive: *IDUA*
Mucopolysaccharidosis type II (1 gene). X-linked: *IDS*. Males are not tested for X-linked disorders.
Mucopolysaccharidosis type III (4 genes). Autosomal recessive: *GNS*, *HGSNAT*, *NAGLU*, *SGSH*
Mucopolysaccharidosis type IVA (1 gene). Autosomal recessive: *GALNS*
Mucopolysaccharidosis type IX (1 gene). Autosomal recessive: *HYAL1*
Mucopolysaccharidosis type VI (1 gene). Autosomal recessive: *ARSB*
Mucopolysaccharidosis type VII (1 gene). Autosomal recessive: *GUSB*
Multiple pterygium syndrome (1 gene). Autosomal recessive: *CHRNG*
Multiple sulfatase deficiency (1 gene). Autosomal recessive: *SUMF1*
Muscular dystrophy, *LAMA2*-related (1 gene). Autosomal recessive: *LAMA2*
Myotonia congenita (1 gene). Autosomal recessive: *CLCN1*

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Disorders Tested (Cont.)

Myotubular myopathy (1 gene). X-linked: *MTM1*. Males are not tested for X-linked disorders.
N-acetylglutamate synthetase deficiency (1 gene). Autosomal recessive: *NAGS*
Nemaline myopathy (1 gene). Autosomal recessive: *NEB*
Nephrogenic diabetes insipidus (1 gene). X-linked: *AVPR2*. Males are not tested for X-linked disorders.
Nephrotic syndrome (2 genes). Autosomal recessive: *NPHS1*, *NPHS2*
Neurodegeneration with brain iron accumulation disorder (7 genes). Autosomal recessive: *ATP13A2*, *C19orf12*, *COASY*, *CP*, *DCAF17*, *FA2H*, *PLA2G6*
Neuronal ceroid-lipofuscinosis (10 genes). Autosomal recessive: *CLN3*, *CLN5*, *CLN6*, *CLN8*, *CTSD*, *CTSF*, *KCTD7*, *MFSD8*, *PPT1*, *TPP1*
Niemann-Pick disease type C (2 genes). Autosomal recessive: *NPC1*, *NPC2*
Niemann-Pick disease types A and B (1 gene). Autosomal recessive: *SMPD1*
Nijmegen breakage syndrome (1 gene). Autosomal recessive: *NBN*
Oculocutaneous albinism (2 genes). Autosomal recessive: *OCA2*, *TYR*
Opitz G/BBB syndrome (1 gene). X-linked: *MID1*. Males are not tested for X-linked disorders.
Ornithine transcarbamylase deficiency (1 gene). X-linked: *OTC*. Males are not tested for X-linked disorders.
Ornithine translocase deficiency (1 gene). Autosomal recessive: *SLC25A15*
Osteogenesis imperfecta, autosomal recessive (9 genes). Autosomal recessive: *BMP1*, *CRTAP*, *FKBP10*, *P3H1*, *PLOD2*, *PPIB*, *SERPINF1*, *TMEM38B*, *WNT1*
Osteopetrosis (3 genes). Autosomal recessive: *OSTM1*, *TCIRG1*, *TNFSF11*
Pantothenate kinase-associated neurodegeneration (1 gene). Autosomal recessive: *PANK2*
Pelizaeus-Merzbacher disease (1 gene). X-linked: *PLP1*. Males are not tested for X-linked disorders.
Pendred syndrome (1 gene). Autosomal recessive: *SLC26A4*
Peroxisomal acyl-CoA oxidase deficiency (1 gene). Autosomal recessive: *ACOX1*
Phenylalanine hydroxylase deficiency, includes phenylketonuria (PKU) (1 gene). Autosomal recessive: *PAH*
Phosphoglycerate dehydrogenase deficiency (1 gene). Autosomal recessive: *PHGDH*
Pitt-Hopkins-like syndrome 1 (1 gene). Autosomal recessive: *CNTNAP2*
POLG-related disorders (1 gene). Autosomal recessive: *POLG*
Polycystic kidney disease, autosomal recessive (1 gene). Autosomal recessive: *PKHD1*
Polymicrogyria (1 gene). Autosomal recessive: *ADGRG1*
Pompe disease (1 gene). Autosomal recessive: *GAA*
Pontocerebellar hypoplasia (11 genes). Autosomal recessive: *AMPD2*, *CHMP1A*, *CLP1*, *EXOSC3*, *RARS2*, *SEPSECS*, *TSEN2*, *TSEN34*, *TSEN54*, *VPS53*, *VRK1*
Primary congenital glaucoma (1 gene). Autosomal recessive: *CYP1B1*
Primary hyperoxaluria (3 genes). Autosomal recessive: *AGXT*, *GRHPR*, *HOGA1*
Primary microcephaly (1 gene). Autosomal recessive: *MCPH1*
Progressive familial intrahepatic cholestasis (3 genes). Autosomal recessive: *ABCB11*, *ABCB4*, *ATP8B1*
Progressive pseudorheumatoid dysplasia (1 gene). Autosomal recessive: *CCN6*
Propionic acidemia (2 genes). Autosomal recessive: *PCCA*, *PCCB*
Pseudocholinesterase deficiency (1 gene). Autosomal recessive: *BCHE*
Pulmonary surfactant metabolism dysfunction (1 gene). Autosomal recessive: *ABCA3*
Pycnodysostosis (1 gene). Autosomal recessive: *CTSK*
Pyridoxal 5'-phosphate-dependent epilepsy (1 gene). Autosomal recessive: *PNPO*
Pyridoxine-dependent epilepsy (1 gene). Autosomal recessive: *ALDH7A1*
Pyruvate carboxylase deficiency (1 gene). Autosomal recessive: *PC*
Pyruvate dehydrogenase deficiency (4 genes). Autosomal recessive: *PDHB*, *PDHX*, *PDP1*; X-linked: *PDHA1*. Males are not tested for X-linked disorders.
Renal tubular acidosis with deafness (2 genes). Autosomal recessive: *ATP6V0A4*, *ATP6V1B1*
Retinitis pigmentosa (13 genes). Autosomal recessive: *CERKL*, *CNGA1*, *CNGB1*, *CWC27*, *DHDDS*, *EYS*, *FAM161A*, *IFT140*, *MAK*, *PRCD*, *RLBP1*; X-linked: *RP2*, *RPGR*. Males are not tested for X-linked disorders.
Rhizomelic chondrodysplasia punctata (3 genes). Autosomal recessive: *AGPS*, *GNPAT*, *PEX7*
Roberts syndrome (1 gene). Autosomal recessive: *ESCO2*
Sandhoff disease (1 gene). Autosomal recessive: *HEXB*
Schimke immunoosseous dysplasia (1 gene). Autosomal recessive: *SMARCAL1*
Schindler disease (1 gene). Autosomal recessive: *NAGA*
SELENON-related disorders (1 gene). Autosomal recessive: *SELENON*
Severe combined immunodeficiency (SCID) (28 genes). Autosomal recessive: *AK2*, *CD247*, *CD3D*, *CD3E*, *CD3G*, *CD8A*, *CORO1A*, *DCLRE1C*, *DOCK8*, *FOXN1*, *IKBKB*, *IL2RA*, *IL7R*, *JAK3*, *LCK*, *LIG4*, *MALT1*, *MTHFD1*, *NHEJ1*, *PGM3*, *PNP*, *PRKDC*, *PTPRC*, *RAG1*, *RAG2*, *STK4*, *TTC7A*, *ZAP70*
Severe combined Immunodeficiency (SCID), X-linked (1 gene). X-linked: *IL2RG*. Males are not tested for X-linked disorders.
Severe congenital neutropenia (2 genes). Autosomal recessive: *HAX1*, *VPS45*
Short-rib thoracic dysplasia (1 gene). Autosomal recessive: *DYNC2H1*

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Disorders Tested (Cont.)

Short/branched chain acyl-CoA dehydrogenase deficiency (1 gene). Autosomal recessive: *ACADSB*
Sialic acid storage disorders (1 gene). Autosomal recessive: *SLC17A5*
Sialidosis (1 gene). Autosomal recessive: *NEU1*
Sjogren-Larsson syndrome (1 gene). Autosomal recessive: *ALDH3A2*
Smith-Lemli-Opitz syndrome (1 gene). Autosomal recessive: *DHCR7*
Spinal muscular atrophy (1 gene). Autosomal recessive: *SMN1*
Spinocerebellar ataxia 10 (1 gene). Autosomal recessive: *ANO10*
Spondylothoracic dysostosis (1 gene). Autosomal recessive: *MESP2*
Stüve-Wiedemann syndrome (1 gene). Autosomal recessive: *LIFR*
Sulfate transporter-related osteochondrodysplasias, includes achondrogenesis type 1B, atelosteogenesis type 2, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia (1 gene). Autosomal recessive: *SLC26A2*
Sulfite oxidase deficiency (1 gene). Autosomal recessive: *SUOX*
Systemic primary carnitine deficiency (1 gene). Autosomal recessive: *SLC22A5*
Tay-Sachs disease (1 gene). Autosomal recessive: *HEXA*
Tetrahydrobiopterin deficiency (3 genes). Autosomal recessive: *PCBD1*, *PTS*, *QDPR*
Trichohepatoenteric syndrome (2 genes). Autosomal recessive: *SKIC2*, *SKIC3*
Trimethylaminuria (1 gene). Autosomal recessive: *FMO3*
Triple A syndrome (1 gene). Autosomal recessive: *AAAS*
Tyrosine hydroxylase deficiency (1 gene). Autosomal recessive: *TH*
Tyrosinemia type I (1 gene). Autosomal recessive: *FAH*
Tyrosinemia type II (1 gene). Autosomal recessive: *TAT*
Tyrosinemia type III (1 gene). Autosomal recessive: *HPD*
Usher syndrome (hearing loss and retinitis pigmentosa) (10 genes). Autosomal recessive: *ADGRV1*, *CDH23*, *CIB2*, *CLRN1*, *MYO7A*, *PCDH15*, *USH1C*, *USH1G*, *USH2A*, *WHRN*
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (1 gene). Autosomal recessive: *ACADVL*
Vitamin D-dependent rickets (1 gene). Autosomal recessive: *CYP27B1*
Walker-Warburg syndrome and other *FKTN*-related dystrophies (1 gene). Autosomal recessive: *FKTN*
Werner syndrome (1 gene). Autosomal recessive: *WRN*
Wilson disease (1 gene). Autosomal recessive: *ATP7B*
Wiskott-Aldrich syndrome (1 gene). X-linked: *WAS*. Males are not tested for X-linked disorders.
Xeroderma pigmentosum (8 genes). Autosomal recessive: *DDB2*, *ERCC2*, *ERCC3*, *ERCC4*, *ERCC5*, *POLH*, *XPA*, *XPC*
Zellweger spectrum disorder/ peroxisome biogenesis disorder (13 genes). Autosomal recessive: *PEX1*, *PEX10*, *PEX11B*, *PEX12*, *PEX13*, *PEX14*, *PEX16*, *PEX19*, *PEX2*, *PEX26*, *PEX3*, *PEX5*, *PEX6*

Performing Labs

Component Type	Performed at	Laboratory Director
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Technical component, analysis	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG
Professional component	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG

For inquiries, the physician may contact the lab at 800-255-7357

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Patient Details
LASTNAME, FIRSTNAME

Phone:
Date of Birth: **mm/dd/yyyy**
Age: **00**
Sex: **Female**
Patient ID:
Alternate Patient ID:

Physician Details
CLIENT NAME
CLIENT ADDRESS

Phone: **000000000**
Account Number: **00000000**
Physician ID:
NPI:

Specimen Details
Specimen ID: **00000000000**
Control ID:
Alternate Control Number:
Date Collected: **mm/dd/yyyy 0000 Local**
Date Received: **mm/dd/yyyy 0000 ET**
Date Entered: **mm/dd/yyyy 0000 ET**
Date Reported: **mm/dd/yyyy 0000 ET**

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