	FEMALE	MALE
Account Number: LCA-BN	**SAMPLE REPORT, F-630217**	**SAMPLE REPORT, M-630217**
Ordering Physician:	11/11/2001	12/12/2002
Date Reported: 08/21/2019 15:12 (Local)	00000000110	00000000120

SAMPLE REPORT, F-630217

Specimen ID: 00000000110
Accession ID: A00055030
Sample Type: Blood
Date Collected: 08/05/2019 12:00 (Local)
Date Received: 08/06/2019
Date Entered: 08/06/2019

Gender: Female
DOB: 11/11/2001
Ethnicity: Unknown
Indication: Carrier testing

SAMPLE REPORT, M-630217

Specimen ID: 00000000120
Accession ID: A00055031
Sample Type: Blood
Date Collected: 08/05/2017 12:00 (Local)
Date Received: 08/06/2019
Date Entered: 08/06/2019

Gender: Male
DOB: 12/12/2002
Ethnicity: Unknown
Indication: Carrier testing

Account Number: LCA-BN
Ordering:
Referring:

ID:
NPI:
Lab ID: MNEGA

Date Reported: 08/21/2019 15:12 (Local)

SUMMARY: POSITIVE RESULTS: CARRIER(S) IDENTIFIED

POSITIVE RESULTS: CARRIER(S) IDENTIFIED

DISORDER *(GENE)*	RESULTS - SAMPLE REPORT, F-630217	RESULTS - SAMPLE REPORT, M-630217	INTERPRETATION
Alpha-mannosidosis (*MAN2B1*) NMID: NM_000528	NEGATIVE	POSITIVE Heterozygous for c.1383C>A (p.Tyr461Ter), pathogenic Chr19:12768296 (GRCh37)	This result reduces, but does not eliminate, the risk for an affected pregnancy. **Residual risk for affected pregnancy:** 1 in 56,000
Neuronal ceroid-lipofuscinosis (*TPP1*) NMID: NM_000391	POSITIVE Heterozygous for c.1266G>C (p.Gln422His), pathogenic Chr11:6636673 (GRCh37)	NEGATIVE	This result reduces, but does not eliminate, the risk for an affected pregnancy. **Residual risk for affected pregnancy:** 1 in 60,000

NEGATIVE RESULTS: NOT AT INCREASED REPRODUCTIVE RISK

DISORDER *(GENE)*	RESULTS - SAMPLE REPORT, F-630217	RESULTS - SAMPLE REPORT, M-630217	INTERPRETATION
Cystic fibrosis (*CFTR*) NMID: NM_000492	NEGATIVE	NEGATIVE	This result reduces, but does not eliminate, the risk for an affected pregnancy.

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Spinal muscular atrophy (*SMN1*) NMID: NM_000344	NEGATIVE 3 (or more) copies of SMN1	NEGATIVE 3 (or more) copies of SMN1	This result reduces, but does not eliminate, the risk for an affected pregnancy.
Fragile X syndrome (*FMR1*) NMID: NM_002024	NEGATIVE: 29 and 36 repeats	Males are not tested for X-linked disorders.	This result reduces, but does not eliminate, the risk for an affected pregnancy.
ALL OTHER DISORDERS	NEGATIVE	NEGATIVE	This result reduces, but does not eliminate, the risk for an affected pregnancy. For a complete list of residual risks for all genes on this panel, visit www.integratedgenetics.com.

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of positive results, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Integrated Genetics Genetic Counselors please visit www.integratedgenetics.com/genetic-counselingor call (855) GC-CALLS (855-422-2557).

ADDITIONAL CLINICAL INFORMATION

Alpha-mannosidosis: Alpha-mannosidosis is an autosomal recessive lysosomal storage disorder with variable severity and age at onset. Signs and symptoms may include progressive neurological deterioration, intellectual disability, skeletal and facial abnormalities, immunodeficiency, and hearing impairment. Bone marrow transplant may be an option for some individuals. Treatment is otherwise supportive. (Malm, PMID:18651971).

Neuronal ceroid-lipofuscinosis: The neuronal ceroid-lipofuscinoses (NCLs) are a group of lysosomal storage disorders with variable severity and age at onset, ranging from infantile through adult. At least twelve genes with different inheritance patterns cause the NCL. Several autosomal recessive genes are included in this analysis. Signs and symptoms may include progressive intellectual and motor deterioration, seizures, speech loss, progressive vision loss, and a reduced life expectancy. Enzyme replacement therapy may be available for individuals affected with TPP1-associated NCL, also known as CLN2. Treatment is otherwise supportive. (Sun, PMID: 30740407).

COMMENTS

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder(s) tested. References and additional information about the disorders tested are available at **www.integratedgenetics.com**.

The standard of care for Tay-Sachs disease carrier detection in all ethnic groups is enzyme (hexosaminidase A) analysis. For maximum sensitivity and specificity, enzyme analysis should be performed in addition to DNA variant analysis (Schneider, PMID19876898). If Tay-Sachs enzyme analysis was ordered, results are reported separately.

The standard of care for determining carrier status for sickle cell disease and other hemoglobinopathies is to combine information from clinical assessment, complete blood count, hemoglobin electrophoresis, and DNA testing (Traeger-Synodinos, PMID25052315). If hemoglobin electrophoresis was ordered, results are reported separately.

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METHODS/LIMITATIONS

Single Nucleotide Polymorphism and Small Indel Sequencing Assessment: Genomic regions of interest are selected using a custom capture reagent for target enrichment (Twist Bioscience) and sequenced via the Illumina® next generation sequencing platform. Sequencing reads are aligned with the human genome reference GRCh37/hg19 build. Regions of interest include all exons and intron/exon junctions (+/- 20 nucleotides) for each gene analyzed. A minimum of 99% of bases are covered at >15X. Analytical sensitivity is estimated to be >99% for single nucleotide variants, >97% for insertions/deletions less than six base pairs, and >95% for insertions/deletions between six and fifteen base pairs. Uncovered regions with known pathogenic variants are sequenced in a targeted manner (List based on ClinVar Database: July 2019 release). All reported variants are confirmed by a second method.

Copy Number Variant Assessment: Next Generation Sequencing is performed and the data are assessed with Illumina's DRAGEN (Dynamic Read Analysis for GENomics) Bio-IT Platform. Genes listed in ClinVar with pathogenic deletions less than 10 exons in size are padded with additional intronic probes to allow single exon resolution CNV detection (List based on ClinVar Deletion Database: January 2019 release; see list below). For other genes, large deletions (>10 exons) can be detected. The c.1263_1317del55 variant in *GBA* is assessed by targeted PCR and gel electrophoresis. The resolution of this analysis can vary depending on region-specific features. Reported variants are confirmed by a second method. Analytical sensitivity is estimated to be >95%. Padded genes: *ABCA12, ABCD1, ACADM, ACOX1, ADAMTS2, ADGRV1, AGL, AGPAT2, AGXT, AHI1, AIRE, ALDOB, ALMS1, AP3B1, ARL6, ARSA, ARSB, ATM, ATP7A, ATRX, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHB, BLM, BRIP1, CAPN3, CBS, CDH23, CFTR, CLCN5, CLN3, CLN5, CLN8, CNTNAP2, COL4A5, CP, CPT1A, CTNS, CYBB, DBT, DCLRE1C, DHCR7, DMD, DOCK8, DOK7, DYSF, EIF2B5, ELP1, EMD, ERCC4, ETHE1, EYS, FA2H, FAM126A, FANCA, FANCC, FANCD2, FANCI, FKRP, FKTN, GAA, GALC, GALNS, GALT, GBE1, GLDC, GNE, GNPTAB, GUSB, HBB, HEXA, HEXB, HINT1, HJV, HPD, HSD17B4, IDS, IFT140, IL7R, ITPA, KCTD7, L1CAM, LAMA2, LAMP2, MCOLN1, MEGF8, MKKS, MKS1, MLC1, MMAB, MTM1, NBN, NCF2, NDUFAF2, NDUFS6, NEB, NPHP1, NR0B1, NTRK1, OAT, OCRL, OTC, PAH, PANK2, PCCA, PCDH15, PDHX, PEX1, PEX6, PHKA1, PHKA2, PHKB, PKHD1, PLA2G6, PMM2, POLH, POMGNT1, RAPSN, RDH12, RPGRIP1, RPS6KA3, SGCD, SGCG, SLC25A20, SLC26A4, SLC2A10, SLC35A3, SLC7A7, SPG11, STRC, STX11, SYNE4, TAZ, TMEM231, TMEM237, TMEM38B, TMEM70, TRIM32, USH2A, VLDLR, VPS13B, VRK1, WRN.*

Congenital Adrenal Hyperplasia: This analysis will detect most large rearrangements/deletions/duplications within the *CYP21A2* gene, as well as the presence of seven of the most common pathogenic variants in the gene: 1) c.518T>A (p.Ile173Asn), Chr6:32007203 (GRCh37); 2) c.713T>A (p.Val238Glu); Chr6:32007587 (GRCh37); 3) c.719T>A (p.Met240Lys); Chr6:32007593 (GRCh37); 4) c.923dup (p.Leu308Phefs); Chr6:32007966 (GRCh37); 5) c.293-13C/A>G; Chr6:32006858 (GRCh37); 6) c.332_339delGAGACTAC (p.Gly111Valfs); Chr6:32006910-32006917 (GRCh37) 7) c.-113G>A; Chr6:32006087 (GRCh37). Other point mutations and small indels and reciprocal changes between *CYP21A2* and *CYP21A1P* are not detected by this analysis. The analytical sensitivity of this assay is estimated to be >99%.

Alpha thalassemia: Variants included in the analysis of the alpha-globin (*HBA*) gene cluster 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 >99% for the targeted variants.

Spinal Muscular Atrophy: This analysis will detect the copy number of exon 7 of the *SMN1* gene. When no copies of *SMN1* exon 7 are detected, *SMN2* exon 7 copy number is assessed and reported. This test is unable to differentiate between two copies of the *SMN1* gene on one allele (in cis) versus two copies of the gene on different alleles (in trans). When two copies of *SMN1* exon 7 are detected, the NGS data are assessed for the presence of the c.*3+80T>G "silent carrier" variant. This analysis does not test for any other variants that may be present in other regions of the *SMN1* gene. Therefore, normal results reduce, but do not eliminate the risk of this patient being a carrier of SMA. Post-test carrier risk reductions for individuals with no family history are shown in the table below.

Population	Pre-test carrier risk	Post-test risk of being a carrier with 2 copies**		Post-test risk of being a carrier with 3 copies
		POSITIVE for the c.*3+80T>G SNP[a]	NEGATIVE for the c.*3+80T>G SNP	
African American	1 in 72	1 in 34	1 in 375	1 in 4200
Ashkenazi Jewish	1 in 67	High risk	1 in 918	1 in 5400
Asian	1 in 59	High risk	1 in 907	1 in 5600
Caucasian	1 in 47	1 in 29	1 in 921	1 in 5600
Hispanic	1 in 68	1 in 140	1 in 906	1 in 5400
Mixed or other ethnic backgrounds	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.			

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** includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay

[a]Luo et al., PMID 23788250

Fragile X Syndrome: Repeat-primed PCR is used to detect the number of CGG repeats on each allele of the *FMR1* gene. The reportable range is 5-200 repeats. Alleles with expansions above 200 repeats are reported as >200. In females, excluding prenatal specimens, alleles between 55 and 90 repeats are assessed by a PCR assay to determine the number and position of AGG interruptions within the CGG repeats (analysis performed at Esoterix Genetic Laboratories, LLC, 3400 Computer Drive Westborough MA 01581, (800)255-7357, Bernice Allitto, PhD., Laboratory Director). Interpretation of repeat expansion results is based on the following ranges: Negative: < 45 repeats; intermediate: 45-54 repeats; premutation: 55-200 repeats; full mutation: >200 repeats. The analytical sensitivity of this assay for the detection of expanded alleles in the *FMR1* gene is estimated to be >99%. Repeat numbers are typically ±1 for alleles containing up to 60 repeats, ±3 for alleles containing 61-119 repeats, and ±10 for alleles with >119 repeats. Low levels of mosaicism (<5%) and *FMR1* variants unrelated to trinucleotide expansion are not detected by this assay.

Reported Variants and Risk Revisions: Pathogenic and likely pathogenic variants are reported after confirmation by an appropriate technology. Variants in *GJB2, GJB6,* and *OPA3* that act in a dominant fashion are not reported. *NEB* variants occurring in exons 82-105 may not be reliably detected by this analysis and are not reported. Nondeletion variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, http://www.hgvs.org/). Variants of uncertain significance, likely benign, and benign variants are not reported. Variant classification is consistent with ACMG standards and guidelines (Richards, PMID:25741868). Detailed variant classification information is available upon request. When provided, carrier rates and detection rates are derived from gnomAD and ClinVar. For unknown or mixed ethnicities, the ethnic background with the most conservative risk estimate is used. For a complete list of residual risks for all genes on this panel, visit www.integratedgenetics.com.

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations, including rearrangements, gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants. 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.

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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*.

Acute infantile liver failure (3 genes). Autosomal recessive: *LARS, 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, X-linked (1 gene). 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.

Autoimmune polyglandular syndrome type 1 (1 gene). Autosomal recessive: *AIRE*.

Autosomal recessive congenital ichthyosis (ARCI) (12 genes). Autosomal recessive: *ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, TGM1*.

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) (1 gene). Autosomal recessive: *SACS*.

Axonal neuropathy with neuromyotonia, autosomal recessive (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: *TAZ*. Males are not tested for X-linked disorders.

Bartter syndrome (5 genes). Autosomal recessive: *BSND, CLCNKA, CLCNKB, KCNJ1, SLC12A1*.

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 (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*.

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Carnitine-acylcarnitine translocase deficiency (1 gene). Autosomal recessive: *SLC25A20*.

Carpenter syndrome (2 genes). Autosomal recessive: *MEGF8, 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*.

Chronic granulomatous disease (5 genes). Autosomal recessive: *CYBA, NCF1, 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*.

Congenital adrenal hyperplasia (6 genes). Autosomal recessive: *CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR*. Fusion *CYP11B1* genes will not be reported;

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 disorder of deglycosylation (1 gene). Autosomal recessive: *NGLY1*.

Congenital disorders of glycosylation type 1 (4 genes). Autosomal recessive: *ALG1, ALG6, MPI, PMM2*.

Congenital generalized lipodystrophy (2 genes). Autosomal recessive: *AGPAT2, CAVIN1*

Congenital insensitivity to pain with anhidrosis (1 gene). Autosomal recessive: *NTRK1*.

Congenital myasthenic syndrome (5 genes). Autosomal recessive: *CHAT, COLQ, DOK7, GFPT1, RAPSN*.

Corneal dystrophy and perceptive deafness (1 gene). Autosomal recessive: *SLC4A11*.

Costeff optic atrophy syndrome, autosomal recessive (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.

Deafness and hearing loss, nonsyndromic (7 genes). Autosomal recessive: *GJB2, GJB6, LOXHD1, OTOF, STRC, SYNE4*. Only recessively inherited variants will be reported for *GJB2* and *GJB6*; 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.

Dihydrolipoamide dehydrogenase deficiency (1 gene). Autosomal recessive: *DLD*.

Dihydropyrimidine dehydrogenase deficiency (1 gene). Autosomal recessive: *DPYD*.

Distal spinal muscular atrophy, autosomal recessive (1 gene). Autosomal recessive: *PLEKHG5*.

Donnai-Barrow syndrome (1 gene). Autosomal recessive: *LRP2*.

Dystrophinopathies, including Duchenne and Becker muscular dystrophy and X-linked cardiomyopathy (1 gene). X-Linked: *DMD*. Males are not tested for X-linked disorders.

Early infantile epileptic encephalopathy (2 genes). Autosomal recessive: *CAD, ITPA*.

Ehlers-Danlos syndrome type VIIC (1 gene). Autosomal recessive: *ADAMTS2*.

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.

Familial dysautonomia (1 gene). Autosomal recessive: *ELP1*.

Familial hemophagocytic lymphohistiocytosis (4 genes). Autosomal recessive: *PRF1, STX11, STXBP2, UNC13D*.

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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: *GBA*.

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: *G6PC, 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 (10 genes). Autosomal recessive: *AP3B1, AP3D1, 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 (1 gene). Autosomal recessive: *CBS*.

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*.

Hypomyelination and congenital cataract (1 gene). Autosomal recessive: *FAM126A*.

Hypophosphatasia (1 gene). Autosomal recessive: *ALPL*.

Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome (4 genes). Autosomal recessive: *CDCA7, DNMT3B, HELLS, ZBTB24*.

Immunodysregulation, polyendocrinopathy, and enteropathy (1 gene). X-Linked: *FOXP3*. Males are not tested for X-linked disorders.

Inclusion body myopathy 2 (1 gene). Autosomal recessive: *GNE*.

Isovaleric acidemia (1 gene). Autosomal recessive: *IVD*.

Joubert syndrome and related disorders, including Meckel-Gruber syndrome (19 genes). Autosomal recessive: *AHI1, ARL13B, B9D1, B9D2, 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*.

Krabbe disease (1 gene). Autosomal recessive: *GALC*.

L1 syndrome (1 gene). X-Linked: *L1CAM*. Males are not tested for X-linked disorders.

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Leber congenital amaurosis (9 genes). Autosomal recessive: *AIPL1, LCA5, LRAT, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1*

Leigh syndrome, autosomal recessive (11 genes). Autosomal recessive: *COX15, 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, autosomal recessive (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*.

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.

Mitochondrial complex I deficiency (1 gene). Autosomal recessive: *ACAD9*.

Mitochondrial complex V deficiency (1 gene). Autosomal recessive: *TMEM70*.

Mitochondrial DNA depletion syndrome, MVP17-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*.

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 sulphatase deficiency (1 gene). Autosomal recessive: *SUMF1*.

Muscular dystrophy, LAMA2-related (1 gene). Autosomal recessive: *LAMA2*.

Myotubular myopathy (1 gene). X-Linked: *MTM1*. Males are not tested for X-linked disorders.

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*.

Omenn syndrome (3 genes). Autosomal recessive: *DCLRE1C, RAG1, RAG2*.

Ornithine transcarbamylase deficiency (1 gene). X-Linked: *OTC*. Males are not tested for X-linked disorders.

Ornithine translocase deficiency (1 gene). Autosomal recessive: *SLC25A15*.

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	FEMALE	MALE
Account Number: LCA-BN	**SAMPLE REPORT, F-630217**	**SAMPLE REPORT, M-630217**
Ordering Physician:	11/11/2001	12/12/2002
Date Reported: 08/21/2019 15:12 (Local)	00000000110	00000000120

Osteogenesis imperfecta, autosomal recessive (9 genes). Autosomal recessive: *BMP1, CRTAP, FKBP10, P3H1, PLOD2, PPIB, SERPINF1, TMEM38B, WNT1*

Osteopetrosis, autosomal recessive (3 genes). Autosomal recessive: *OSTM1, TCIRG1, TNFSF11*.

Pantothenate kinase-associated neurodegeneration (1 gene). Autosomal recessive: *PANK2*.

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*.

Polycystic kidney disease, autosomal recessive (1 gene). Autosomal recessive: *PKHD1*.

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 carnitine deficiency (1 gene). Autosomal recessive: *SLC22A5*.

Primary congenital glaucoma (1 gene). Autosomal recessive: *CYP1B1*.

Primary hyperoxaluria (3 genes). Autosomal recessive: *AGXT, GRHPR, HOGA1*.

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*.

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 dehydrogenase deficiency (5 genes). Autosomal recessive: *DLAT, PDHB, PDHX, PDP1*. X-Linked: *PDHA1*. Males are not tested for X-linked disorders.

Renal tubular acidosis and deafness (2 genes). Autosomal recessive: *ATP6V0A4, ATP6V1B1*.

Retinitis pigmentosa (11 genes). Autosomal recessive: *CERKL, 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*.

Sandhoff disease (1 gene). Autosomal recessive: *HEXB*.

SELENON-related disorders (1 gene). Autosomal recessive: *SELENON*.

Severe combined immunodeficiency (SCID) (25 genes). Autosomal recessive: *AK2, CD247, CD3D, CD3E, CD3G, CD8A, CORO1A, DOCK8, FOXN1, IKBKB, IL2RA, IL7R, JAK3, LCK, LIG4, MALT1, MTHFD1, NHEJ1, PGM3, PNP, PRKDC, PTPRC, 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, autosomal recessive (1 gene). Autosomal recessive: *HAX1*.

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*.

Spondylothoracic dysostosis (1 gene). Autosomal recessive: *MESP2*.

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*.

Tay-Sachs disease (1 gene). Autosomal recessive: *HEXA*.

Tetrahydrobiopterin deficiency (3 genes). Autosomal recessive: *PCBD1, PTS, QDPR*.

Trichohepatoenteric syndrome (2 genes). Autosomal recessive: *SKIV2L, TTC37*.

Trifunctional protein deficiency (1 gene). Autosomal recessive: *HADHB*.

Triple A syndrome (1 gene). Autosomal recessive: *AAAS*.

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		FEMALE	MALE
Account Number:	LCA-BN	**SAMPLE REPORT, F-630217**	**SAMPLE REPORT, M-630217**
Ordering Physician:		11/11/2001	12/12/2002
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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) (9 genes). Autosomal recessive: *ADGRV1, CDH23, CIB2, CLRN1, PCDH15, USH1C, USH1G, USH2A, WHRN*.

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (1 gene). Autosomal recessive: *ACADVL*.

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*.

X-linked syndromic mental retardation (1 gene). X-Linked: *NONO*. 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*.

This test was developed and its performance characteristics determined by Medical Neurogenetics, LLC. It has not been cleared or approved by the Food and Drug Administration.

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