INVITAE

INVITAE DIAGNOSTIC TESTING RESULTS

Patient name:	Albana Kjemali	**Sample type:**	Buccal Swab	**Report date:**	12/06/2021
DOB:	11/13/2013	**Sample collection date:**	11/18/2021	**Invitae #:**	RQ2851745
Sex assigned at birth:	Female	**Sample accession date:**	11/19/2021	**Clinical team:**	Moroz Mariia
Gender:		**MRN:**	AK111313		

Reason for testing

Diagnostic test for a personal history of disease

Test performed

Sequence analysis and deletion/duplication testing of the 853 genes listed in the Genes Analyzed section.
Multiple panels/genes ordered: see Methods for complete list.

RESULT: INCREASED RISK

One Increased Risk Allele identified in ABCG8.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
ABCG8	c.55G>C (p.Asp19His)	heterozygous	Increased Risk Allele
BSCL2	c.1010G>A (p.Gly337Glu)	heterozygous	Uncertain Significance
CLPB	c.1807C>T (p.Arg603Cys)	heterozygous	Uncertain Significance
COQ4	c.397G>T (p.Val133Leu)	heterozygous	Uncertain Significance
CPS1	c.1021T>G (p.Leu341Val)	heterozygous	Uncertain Significance
CSGALNACT1	c.998A>G (p.Glu333Gly)	heterozygous	Uncertain Significance
DHTKD1	c.847A>G (p.Met283Val)	heterozygous	Uncertain Significance
SPR	c.706G>A (p.Val236Met)	heterozygous	Uncertain Significance
TTPA	c.616G>A (p.Val206Ile)	heterozygous	Uncertain Significance
CHIT1	c.1049_1072dup (p.Trp358*)	heterozygous	Benign (reportable variant)
CHIT1	c.304G>A (p.Gly102Ser)	heterozygous	Benign (reportable variant)
GALC	c.1685T>C (p.Ile562Thr)	heterozygous	Benign (Pseudodeficiency allele)
GALC	c.742G>A (p.Asp248Asn)	heterozygous	Benign (Pseudodeficiency allele)

About this test

This diagnostic test evaluates 853 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

Laboratory Director: Kristin Gibson, PhD, FACMG, NYCQ, CGMB, ErCLG
Invitae 1400 16th Street, San Francisco, CA 94103 · clientservices@invitae.com · 800.436.3037

Next steps

- This is a medically important result that should be discussed with a healthcare provider, such as a genetic counselor, to learn more about this result and the appropriate next steps for further evaluation, treatment and/or management. This result should be interpreted within the context of additional laboratory results, family history and clinical findings.

- Consider sharing this result with relatives as they may also be at risk. Details on our Family Variant Testing program can be found at **www.invitae.com/family**.

- Testing of up to two family members for the Variant(s) of Uncertain Significance (VUS) identified in BSCL2 and DHTKD1 is available at no additional cost. Please consider this individual's clinical features and availability of informative family members to test before ordering VUS resolution testing. More details on our VUS Resolution Program, including required documentation, can be found at **www.invitae.com/family**.

- One or more variants were identified that are not known to cause disease. See the CHIT1 and GALC variant(s) in the Variant Details section for more information.

- Register your test at **www.invitae.com/patients** to download a digital copy of your results. You can also access educational resources about how your results can help inform your health.

Clinical summary

An Increased Risk Allele, c.55G>C (p.Asp19His), was identified in ABCG8.

- The ABCG8 gene is associated with autosomal recessive sitosterolemia (MedGen UID: 87466). In addition, the c.55G>C (p.Asp19His) variant has been associated with increased risk of developing gallstones (PMID: 22898925).
- This variant is associated with reduced serum phytosterol levels and confers increased susceptibility to developing gallstones (PMID: 11893785, 17632509, 21039838, 21274884, 22898925). This variant is present in approximately 9% of the population, including multiple homozygous individuals, and confers an approximately 2-fold increased risk of developing gallstones (PMID: 22898925). This variant has not been associated with autosomal recessive sitosterolemia.
- Biological relatives have a chance of being at an increased risk for disease and should consider testing.

A Variant of Uncertain Significance, c.1010G>A (p.Gly337Glu), was identified in BSCL2.

- The BSCL2 gene is associated with a spectrum of autosomal dominant neurological conditions, including Charcot-Marie-Tooth disease type 2 (CMT2) (PMID: 23142943), also known as distal hereditary motor neuropathy type 5 (HMN5) (MedGen UID: 318838), and spastic paraplegia 17 (SPG17), also known as Silver syndrome (MedGen UID: 419034). It is also associated with a spectrum of autosomal recessive conditions including congenital generalized lipodystrophy, type 2 (CGL2) (MedGen UID: 318593) and progressive encephalopathy with or without lipodystrophy (PELD) (MedGen UID: 863137).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- This variant qualifies for complimentary family studies as part of our VUS Resolution Program. Familial VUS testing is recommended if informative family members are available and are likely to provide additional evidence for future variant reclassification. Details on our VUS Resolution Program can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.1807C>T (p.Arg603Cys), was identified in CLPB.

- The CLPB gene is associated with autosomal recessive 3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia (MEGCANN) (MedGen UID: 907853).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.397G>T (p.Val133Leu), was identified in COQ4.

- The COQ4 gene is associated with autosomal recessive primary coenzyme Q10 deficiency (MedGen UID: 833081).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.1021T>G (p.Leu341Val), was identified in CPS1.

- The CPS1 gene is associated with autosomal recessive carbamoyl phosphate synthetase I (CPS1) deficiency (MedGen UID: 199727).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.998A>G (p.Glu333Gly), was identified in CSGALNACT1.

- The CSGALNACT1 gene is associated with an autosomal recessive skeletal dysplasia (PMID: 27599773, 31325655).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.847A>G (p.Met283Val), was identified in DHTKD1.

- The DHTKD1 gene is associated with autosomal recessive 2-aminoadipic 2-oxoadipic aciduria (AMOXAD) (MedGen UID: 395350), a biochemical phenotype which may or may not result in a clinical condition. The DHTKD1 gene is also associated with autosomal dominant Charcot-Marie-Tooth disease type 2Q (CMT2Q) (MedGen UID: 767280). Additionally, the DHTKD1 gene has preliminary evidence supporting a correlation with autosomal recessive steroid resistant nephrotic syndrome (PMID: 29127259).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- This variant qualifies for complimentary family studies as part of our VUS Resolution Program. Familial VUS testing is recommended if informative family members are available and are likely to provide additional evidence for future variant reclassification. Details on our VUS Resolution Program can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.706G>A (p.Val236Met), was identified in SPR.

- The SPR gene is associated with autosomal recessive sepiapterin reductase deficiency (MedGen UID: 120642). Additionally, the SPR gene has preliminary evidence supporting a correlation with autosomal dominant dopa-responsive dystonia (PMID: 15241655).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.616G>A (p.Val206Ile), was identified in TTPA.

- The TTPA gene is associated with autosomal recessive ataxia with vitamin E deficiency (AVED) (MedGen UID: 341248).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

Variant details

ABCG8, Exon 1, c.55G>C (p.Asp19His), heterozygous, Increased Risk Allele

- This sequence change replaces aspartic acid, which is acidic and polar, with histidine, which is basic and polar, at codon 19 of the ABCG8 protein (p.Asp19His).
- This variant is present in population databases (rs11887534, gnomAD 10%), including at least one homozygous and/or hemizygous individual.
- Population-based case-control studies have shown that this variant is associated with reduced serum phytosterol levels and confers susceptibility to gallstone disease (PMID: 11893785, 17632509, 21039838, 21274884, 22898925). In a large meta-analysis with 4,381 cases and 3,765 controls (PMID: 22898925), individuals carrying this variant had an increased overall risk of gallstone disease (2.07, 95% CI: 1.65-2.60).
- ClinVar contains an entry for this variant (Variation ID: 4975).
- Algorithms developed to predict the effect of missense changes on protein structure and function are either unavailable or do not agree on the potential impact of this missense change (SIFT: "Deleterious"; PolyPhen-2: "Possibly Damaging"; Align-GVGD: "Class C0").
- Experimental studies have shown that this missense causes a gain of ABCG8 protein function in vitro, contrary to the loss of ABCG8 protein function associated with sitosterolemia (PMID: 22898925).
- In summary, this is a common variant that is associated with an increased risk for developing disease. For these reasons, this variant has been classified as an Increased Risk Allele.

BSCL2, Exon 10, c.1010G>A (p.Gly337Glu), heterozygous, Uncertain Significance

- This sequence change replaces glycine, which is neutral and non-polar, with glutamic acid, which is acidic and polar, at codon 337 of the BSCL2 protein (p.Gly337Glu).

- This variant is present in population databases (rs767463971, gnomAD 0.008%).
- This variant has not been reported in the literature in individuals affected with BSCL2-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 567989).
- Algorithms developed to predict the effect of missense changes on protein structure and function (SIFT, PolyPhen-2, Align-GVGD) all suggest that this variant is likely to be tolerated.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

CLPB, Exon 16, c.1807C>T (p.Arg603Cys), heterozygous, Uncertain Significance

- This sequence change replaces arginine, which is basic and polar, with cysteine, which is neutral and slightly polar, at codon 603 of the CLPB protein (p.Arg603Cys).
- This variant is present in population databases (rs186989806, gnomAD 0.002%).
- This variant has not been reported in the literature in individuals affected with CLPB-related conditions.
- Algorithms developed to predict the effect of missense changes on protein structure and function are either unavailable or do not agree on the potential impact of this missense change (SIFT: "Deleterious"; PolyPhen-2: "Probably Damaging"; Align-GVGD: "Class C0").
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

COQ4, Exon 4, c.397G>T (p.Val133Leu), heterozygous, Uncertain Significance

- This sequence change replaces valine, which is neutral and non-polar, with leucine, which is neutral and non-polar, at codon 133 of the COQ4 protein (p.Val133Leu).
- This variant is not present in population databases (gnomAD no frequency).
- This variant has not been reported in the literature in individuals affected with COQ4-related conditions.
- Algorithms developed to predict the effect of missense changes on protein structure and function (SIFT, PolyPhen-2, Align-GVGD) all suggest that this variant is likely to be tolerated.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

CPS1, Exon 10, c.1021T>G (p.Leu341Val), heterozygous, Uncertain Significance

- This sequence change replaces leucine, which is neutral and non-polar, with valine, which is neutral and non-polar, at codon 341 of the CPS1 protein (p.Leu341Val).
- This variant is present in population databases (rs138424013, gnomAD 0.06%).
- This variant has not been reported in the literature in individuals affected with CPS1-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 334019).
- Algorithms developed to predict the effect of missense changes on protein structure and function (SIFT, PolyPhen-2, Align-GVGD) all suggest that this variant is likely to be tolerated.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

CSGALNACT1, Exon 7, c.998A>G (p.Glu333Gly), heterozygous, Uncertain Significance

- This sequence change replaces glutamic acid, which is acidic and polar, with glycine, which is neutral and non-polar, at codon 333 of the CSGALNACT1 protein (p.Glu333Gly).
- This variant is present in population databases (rs146205646, gnomAD 0.2%).
- This variant has not been reported in the literature in individuals affected with CSGALNACT1-related conditions.
- Algorithms developed to predict the effect of missense changes on protein structure and function are either unavailable or do not agree on the potential impact of this missense change (SIFT: "Tolerated"; PolyPhen-2: "Probably Damaging"; Align-GVGD: "Class C0").

- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

DHTKD1, Exon 5, c.847A>G (p.Met283Val), heterozygous, Uncertain Significance

- This sequence change replaces methionine, which is neutral and non-polar, with valine, which is neutral and non-polar, at codon 283 of the DHTKD1 protein (p.Met283Val).
- This variant is present in population databases (rs145337285, gnomAD 0.1%).
- This variant has not been reported in the literature in individuals affected with DHTKD1-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 1053636).
- Algorithms developed to predict the effect of missense changes on protein structure and function output the following: SIFT: "Deleterious"; PolyPhen-2: "Benign"; Align-GVGD: "Class C0". The valine amino acid residue is found in multiple mammalian species, which suggests that this missense change does not adversely affect protein function.
- Algorithms developed to predict the effect of sequence changes on RNA splicing suggest that this variant may create or strengthen a splice site.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

SPR, Exon 3, c.706G>A (p.Val236Met), heterozygous, Uncertain Significance

- This sequence change replaces valine, which is neutral and non-polar, with methionine, which is neutral and non-polar, at codon 236 of the SPR protein (p.Val236Met).
- This variant is present in population databases (rs371904378, gnomAD 0.02%).
- This variant has not been reported in the literature in individuals affected with SPR-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 660797).
- Algorithms developed to predict the effect of missense changes on protein structure and function output the following: SIFT: "Tolerated"; PolyPhen-2: "Benign"; Align-GVGD: "Class C0". The methionine amino acid residue is found in multiple mammalian species, which suggests that this missense change does not adversely affect protein function.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

TTPA, Exon 4, c.616G>A (p.Val206Ile), heterozygous, Uncertain Significance

- This sequence change replaces valine, which is neutral and non-polar, with isoleucine, which is neutral and non-polar, at codon 206 of the TTPA protein (p.Val206Ile).
- This variant is present in population databases (rs554118281, gnomAD 0.008%).
- This variant has not been reported in the literature in individuals affected with TTPA-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 363560).
- Algorithms developed to predict the effect of missense changes on protein structure and function are either unavailable or do not agree on the potential impact of this missense change (SIFT: "Deleterious"; PolyPhen-2: "Benign"; Align-GVGD: "Class C25").
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

CHIT1, Exon 10, c.1049_1072dup (p.Trp358*), heterozygous, Benign (reportable variant)

- Chitotriosidase enzymatic activity is a prognostic and therapeutic biomarker for certain lysosomal storage disorders, such as Gaucher and Niemann-Pick disease A/B/C (PMID: 8750610, 15669690, 17869233).
- This variant is present at very high frequency in population databases (57% in the East Asian population in gnomAD)
- An increasing body of evidence exists for its utility as biomarker for other, non-lysosomal, disorders associated with inflammation and macrophage activation, including sarcoidosis (PMID: 24594143, 31906975), interstitial lung disease (PMID: 31092718, 17631992), and neuroinflammatory or neurodegenerative disorders (PMID: 22014002, 25563799).
- ClinVar contains an entry for this variant (Variation ID: 294920).

- The c.1049_1072dup variant has been shown to lead to aberrant mRNA and result in an inactive human chitotriosidase enzyme (PMID: 7592832, 9748235).
- While chitotriosidase deficiency is not associated with any human disease, the presence of this variant makes chitotriosidase enzymatic activity an unreliable biomarker. For these reasons, this variant is classified as a Benign Reportable Variant.

CHIT1, Exon 4, c.304G>A (p.Gly102Ser), heterozygous, Benign (reportable variant)

- Chitotriosidase enzymatic activity is a prognostic and therapeutic biomarker for certain lysosomal storage disorders, such as Gaucher and Niemann-Pick disease A/B/C (PMID: 8750610, 15669690, 17869233).
- This variant is present at very high frequency in population databases (37% in gnomAD)
- An increasing body of evidence exists for its utility as biomarker for other, non-lysosomal, disorders associated with inflammation and macrophage activation, including sarcoidosis (PMID: 24594143, 31906975), interstitial lung disease (PMID: 31092718, 17631992), and neuroinflammatory or neurodegenerative disorders (PMID: 22014002, 25563799).
- ClinVar contains an entry for this variant (Variation ID: 9526).
- This missense variant has been shown to cause a 30-50% reduction in CHIT1 enzymatic activity both in vivo and in vitro (PMID: 19725875, 24060732).
- While chitotriosidase deficiency is not associated with any human disease, the presence of this variant makes chitotriosidase enzymatic activity an unreliable biomarker. For these reasons, this variant is classified as a Benign Reportable Variant.

GALC, Exon 15, c.1685T>C (p.Ile562Thr), heterozygous, Benign (Pseudodeficiency allele)

- This sequence change replaces isoleucine, which is neutral and non-polar, with threonine, which is neutral and polar, at codon 562 of the GALC protein (p.Ile562Thr).
- This variant is present in population databases (rs398607, gnomAD 61%).
- This variant is a known pseudodeficiency allele and individuals with this variant can exhibit low galactocerebrosidase activity during enzyme analysis. On its own, this variant mildly reduces enzyme activity. However, it has been shown to further reduce GALC enzyme activity when it is located on the same chromosome (in cis) with pathogenic GALC variants (PMID: 26795590, 26865610, 27126738, 27638593). Individuals with pseudodeficiency alleles may exhibit false positive results on related biochemical tests, but pseudodeficiency alleles are not known to cause disease. Although pseudodeficiency alleles do not cause disease, other carrier relatives may have abnormal enzyme testing.
- This variant is also known as p.Ile546Thr or p.I546T.
- ClinVar contains an entry for this variant (Variation ID: 92497).
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is not expected to disrupt GALC protein function.
- For these reasons, this variant has been classified as a Benign pseudodeficiency allele.

GALC, Exon 7, c.742G>A (p.Asp248Asn), heterozygous, Benign (Pseudodeficiency allele)

- This sequence change replaces aspartic acid, which is acidic and polar, with asparagine, which is neutral and polar, at codon 248 of the GALC protein (p.Asp248Asn).
- This variant is present in population databases (rs34362748, gnomAD 16%).
- This variant is a known pseudodeficiency allele (PMID: 24388568, 26795590, 27638593), and individuals with this variant can exhibit low galactocerebrosidase activity during enzyme analysis. Individuals with pseudodeficiency alleles may exhibit false positive results on related biochemical tests, but pseudodeficiency alleles are not known to cause disease. Although pseudodeficiency alleles do not cause disease, other carrier relatives may have abnormal enzyme testing.
- This variant is also known as p.D232N.
- ClinVar contains an entry for this variant (Variation ID: 92509).
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is not expected to disrupt GALC protein function.
- For these reasons, this variant has been classified as a Benign pseudodeficiency allele.

Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. Results are negative unless otherwise indicated in the report. Benign and Likely Benign variants are not included in this report but are available upon request. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details.

GENE	TRANSCRIPT
A4GALT	NM_017436.5
AAAS	NM_015665.5
AARS2	NM_020745.3
AASS	NM_005763.3
ABAT	NM_020686.5
ABCB7	NM_004299.4
ABCC8	NM_000352.4
ABCD1	NM_000033.3
ABCD3	NM_002858.3
ABCD4	NM_005050.3
ABCG5	NM_022436.2
ABCG8	NM_022437.2
ACACA	NM_198839.2
ACAD8	NM_014384.2
ACAD9	NM_014049.4
ACADM	NM_000016.5
ACADS	NM_000017.3
ACADSB	NM_001609.3
ACADVL	NM_000018.3
ACAT1	NM_000019.3
ACBD5	NM_145698.4
ACO2	NM_001098.2
ACOX1	NM_004035.6
ACOX2	NM_003500.3
ACSF3	NM_174917.4
ADA	NM_000022.2
ADAR	NM_001111.4
ADK	NM_001123.3
ADSL	NM_000026.2
AFG3L2	NM_006796.2
AGA	NM_000027.3
AGK	NM_018238.3
AGL	NM_000642.2
AGXT	NM_000030.2
AHCY	NM_000687.3

GENE	TRANSCRIPT
AIFM1	NM_004208.3
AK2	NM_001625.3
AKT2	NM_001626.5
ALAD	NM_000031.5
ALAS2	NM_000032.4
ALDH18A1	NM_002860.3
ALDH3A2	NM_000382.2
ALDH4A1	NM_003748.3
ALDH5A1	NM_001080.3
ALDH6A1	NM_005589.3
ALDH7A1	NM_001182.4
ALDOA	NM_000034.3
ALDOB	NM_000035.3
ALG1	NM_019109.4
ALG11	NM_001004127.2
ALG12	NM_024105.3
ALG13	NM_001099922.2
ALG14	NM_144988.3
ALG2	NM_033087.3
ALG3	NM_005787.5
ALG6	NM_013339.3
ALG8	NM_024079.4
ALG9	NM_024740.2
ALPL	NM_000478.5
AMACR	NM_014324.5
AMN*	NM_030943.3
AMPD1	NM_000036.2
AMT	NM_000481.3
AP1S1	NM_001283.3
AP4M1	NM_004722.3
APOPT1	NM_032374.4
APPL1	NM_012096.2
APRT	NM_000485.2
APTX	NM_175073.2
ARCN1	NM_001655.4

GENE	TRANSCRIPT
ARG1	NM_000045.3
ARHGEF9	NM_015185.2;NM_001173479.1
ARSA	NM_000487.5
ARSB	NM_000046.3
ASAH1	NM_177924.3
ASL	NM_000048.3
ASNS	NM_133436.3
ASPA	NM_000049.2
ASS1	NM_000050.4
ATAD1	NM_001321967.1
ATIC	NM_004044.6
ATP13A2	NM_022089.3
ATP5A1	NM_001001937.1
ATP5D	NM_001001975.1
ATP5E	NM_006886.3
ATP6AP1	NM_001183.5
ATP6AP2	NM_005765.2
ATP6V0A2	NM_012463.3
ATP6V1A	NM_001690.3
ATP6V1E1	NM_001696.3
ATP7A	NM_000052.6
ATP7B	NM_000053.3
ATPAF2	NM_145691.3
AUH	NM_001698.2
B3GALNT2	NM_152490.4
B3GALT6	NM_080605.3
B3GAT3	NM_012200.3
B3GLCT	NM_194318.3
B4GALNT1	NM_001478.4
B4GALT1	NM_001497.3
B4GALT7	NM_007255.2
B4GAT1	NM_006876.2
BAG3	NM_004281.3
BCAP31	NM_001139441.1
BCAT2	NM_001190.3

GENE	TRANSCRIPT
BCKDHA	NM_000709.3
BCKDHB	NM_183050.2
BCKDK	NM_005881.3
BCS1L	NM_004328.4
BLK	NM_001715.2
BOLA3	NM_212552.2
BSCL2	NM_032667.6
BSND	NM_057176.2
BTD	NM_000060.3
C12orf65	NM_152269.4
C19orf12	NM_001031726.3
C19orf70	NM_205767.2
C1GALT1C1	NM_001011551.2
C1QBP	NM_001212.3
CA5A	NM_001739.1
CACNA1C*	NM_000719.6;NM_001129840.1
CACNA1D	NM_000720.3
CAD	NM_004341.4
CANT1	NM_138793.3
CARS2	NM_024537.3
CASR	NM_000388.3
CBS	NM_000071.2
CCDC115	NM_032357.3
CD320	NM_016579.3
CDKN1C	NM_000076.2
CEP89	NM_032816.4
CFTR*	NM_000492.3
CHAT	NM_020549.4
CHCHD10	NM_213720.2
CHIT1	NM_003465.2
CHST14	NM_130468.3
CHST3	NM_004273.4
CHST6	NM_021615.4
CHSY1	NM_014918.4
CLCN5	NM_000084.4
CLCNKB*	NM_000085.4
CLDN16	NM_006580.3
CLDN19	NM_148960.2
CLN3	NM_001042432.1

GENE	TRANSCRIPT
CLN5	NM_006493.2
CLN6	NM_017882.2
CLN8	NM_018941.3
CLPB	NM_030813.5
CLPP	NM_006012.2
CLPX	NM_006660.4
CNNM2	NM_017649.4
COA3	NM_001040431.2
COA5	NM_001008215.2
COA6	NM_001012985.2
COA7	NM_023077.2
COASY	NM_025233.6
COG1	NM_018714.2
COG2	NM_007357.2
COG4	NM_015386.2
COG5	NM_006348.3
COG6	NM_020751.2
COG7	NM_153603.3
COG8	NM_032382.4
COPA	NM_004371.3
COPB2	NM_004766.2
COQ2	NM_015697.7
COQ4	NM_016035.4
COQ6	NM_182476.2
COQ7	NM_016138.4
COQ8A	NM_020247.4
COQ8B	NM_024876.3
COQ9	NM_020312.3
COX10*	NM_001303.3
COX14	NM_032901.3
COX15	NM_004376.6
COX20	NM_198076.5
COX4I2	NM_032609.2
COX6A1	NM_004373.3
COX6B1	NM_001863.4
COX7B	NM_001866.2
COX8A	NM_004074.2
CP	NM_000096.3
CPOX	NM_000097.5

GENE	TRANSCRIPT
CPS1	NM_001875.4
CPT1A	NM_001876.3
CPT2	NM_000098.2
CRAT	NM_000755.3
CSGALNACT1	NM_001130518.1
CTNNB1	NM_001904.3
CTNS	NM_004937.2
CTSA	NM_000308.3
CTSD	NM_001909.4
CTSF	NM_003793.3
CTSK	NM_000396.3
CUBN	NM_001081.3
CYC1	NM_001916.4
CYCS	NM_018947.5
CYP27A1	NM_000784.3
CYP27B1	NM_000785.3
CYP2R1	NM_024514.4
CYP7B1	NM_004820.3
D2HGDH	NM_152783.4
DARS2	NM_018122.4
DBH	NM_000787.3
DBT	NM_001918.3
DCAF17	NM_025000.3
DDC*	NM_000790.3
DDOST	NM_005216.4
DES	NM_001927.3
DGUOK	NM_080916.2
DHCR7	NM_001360.2
DHDDS	NM_024887.3
DHFR	NM_000791.3
DHTKD1	NM_018706.6
DLAT	NM_001931.4
DLD	NM_000108.4
DMP1	NM_004407.3
DMXL2*	NM_001174116.1
DNA2	NM_001080449.2
DNAJC12	NM_021800.2
DNAJC19	NM_145261.3
DNAJC5	NM_025219.2

GENE	TRANSCRIPT
DNM1L	NM_012062.4
DOLK	NM_014908.3
DPAGT1	NM_001382.3
DPM1	NM_003859.1
DPM2	NM_003863.3
DPM3	NM_153741.1
DSE	NM_013352.3
EARS2	NM_001083614.1
ECHS1	NM_004092.3
EGF	NM_001963.5
EIF2AK3	NM_004836.6
ELAC2	NM_018127.6
ENO3	NM_053013.3
ENPP1	NM_006208.2
EOGT	NM_173654.2
ETFA	NM_000126.3
ETFB	NM_001985.2
ETFDH	NM_004453.3
ETHE1	NM_014297.3
EXT1	NM_000127.2
EXT2	NM_207122.1
EXTL3	NM_001440.3
FA2H	NM_024306.4
FAH*	NM_000137.2
FAM111A	NM_022074.3
FAM20C	NM_020223.3
FARS2	NM_006567.3
FASTKD2	NM_014929.3
FBP1	NM_000507.3
FBXL4	NM_012160.4
FDX2	NM_001031734.3
FECH	NM_000140.3
FGF23	NM_020638.2
FGFR1	NM_023110.2
FH*	NM_000143.3
FKRP	NM_024301.4
FKTN	NM_001079802.1
FLAD1	NM_025207.4
FOLR1	NM_016725.2
FOXP3	NM_014009.3
FOXRED1	NM_017547.3
FTCD	NM_006657.2
FTL	NM_000146.3
FUCA1	NM_000147.4
FUK	NM_145059.2
FUT8	NM_178155.2
FXYD2	NM_001680.4
G6PC	NM_000151.3
G6PC3	NM_138387.3
G6PD	NM_001042351.2
GAA	NM_000152.3
GABBR2	NM_005458.7
GABRA1	NM_000806.5
GABRA2	NM_001330690.1
GABRB1	NM_000812.3
GABRB3	NM_000814.5
GABRG2	NM_000816.3
GAD1	NM_000817.2
GALC*	NM_000153.3
GALE	NM_000403.3
GALK1	NM_000154.1
GALM*	NM_138801.2
GALNS	NM_000512.4
GALNT3	NM_004482.3
GALT	NM_000155.3
GAMT	NM_000156.5
GANAB	NM_198335.3
GARS	NM_002047.2
GATA1	NM_002049.3
GATA4	NM_002052.3
GATA6	NM_005257.5
GATM	NM_001482.2
GBE1	NM_000158.3
GCDH	NM_000159.3
GCGR	NM_000160.4
GCH1	NM_000161.2
GCK	NM_000162.3
GCLC	NM_001498.3
GDAP1	NM_018972.2
GFER	NM_005262.2
GFM1	NM_024996.5
GFM2	NM_032380.4
GFPT1	NM_001244710.1
GH1*	NM_000515.4
GHR*	NM_000163.4
GIF	NM_005142.2
GJA1	NM_000165.4
GLA	NM_000169.2
GLB1	NM_000404.2
GLDC	NM_000170.2
GLIS3	NM_152629.3
GLRA1	NM_000171.3
GLRB	NM_000824.4
GLRX5	NM_016417.2
GLUD1	NM_005271.4
GLUL	NM_002065.6
GLYCTK	NM_145262.3
GM2A	NM_000405.4
GMPPA	NM_205847.2
GMPPB	NM_021971.2
GNAS	NM_000516.5
GNE	NM_001128227.2
GNMT	NM_018960.5
GNPTAB	NM_024312.4
GNPTG	NM_032520.4
GNS	NM_002076.3
GORAB	NM_152281.2
GOSR2	NM_004287.3
GOT2	NM_002080.3
GPAA1	NM_003801.3
GPC3*	NM_004484.3
GPHN	NM_020806.4
GRHPR	NM_012203.1
GRIN2B	NM_000834.3
GRIN2D	NM_000836.2
GRN	NM_002087.3
GSS	NM_000178.2

GENE	TRANSCRIPT
GTPBP2	NM_019096.4
GTPBP3	NM_133644.3
GUSB	NM_000181.3
GYG1	NM_004130.3
GYG2	NM_003918.2
GYS1	NM_002103.4
GYS2	NM_021957.3
HADH	NM_005327.4
HADHA	NM_000182.4
HADHB	NM_000183.2
HARS2	NM_012208.3
HCCS	NM_005333.4
HCFC1	NM_005334.2
HESX1	NM_003865.2
HEXA	NM_000520.4
HEXB	NM_000521.3
HGD	NM_000187.3
HGSNAT	NM_152419.2
HIBCH	NM_014362.3
HK1	NM_000188.2
HLCS	NM_000411.6
HMBS	NM_000190.3
HMGCL	NM_000191.2
HMGCS2	NM_005518.3
HNF1A	NM_000545.5
HNF1B	NM_000458.3
HNF4A	NM_175914.4
HOGA1	NM_138413.3
HPD	NM_002150.2
HPRT1	NM_000194.2
HRAS	NM_005343.2
HSD17B10	NM_004493.2
HSD17B4	NM_000414.3
HSD3B7	NM_025193.3
HSPD1	NM_002156.4
HTRA2	NM_013247.4
HYAL1	NM_153281.1
IARS2	NM_018060.3
IBA57	NM_001010867.3

GENE	TRANSCRIPT
IDH2	NM_002168.3
IDH3B	NM_006899.4
IDS*	NM_000202.6
IDUA	NM_000203.4
IER3IP1	NM_016097.4
IFIH1	NM_022168.3
IMPDH1	NM_000883.3
INS	NM_000207.2
INSR	NM_000208.3
ISCA1	NM_030940.3
ISCA2	NM_194279.3
ISCU	NM_213595.3
ISPD	NM_001101426.3
IVD	NM_002225.3
JAGN1	NM_032492.3
KARS	NM_001130089.1
KCNA1	NM_000217.2
KCNJ10	NM_002241.4
KCNJ11	NM_000525.3
KCTD7	NM_153033.4
KDM6A	NM_021140.3
KIF1A	NM_004321.6
KLF11	NM_003597.4
KMT2D	NM_003482.3
L2HGDH	NM_024884.2
LAMP2	NM_002294.2
LARGE1	NM_004737.4
LARS	NM_020117.10
LARS2	NM_015340.3
LDHA	NM_005566.3
LFNG	NM_001040167.1
LHX3	NM_014564.4
LIAS	NM_006859.3
LIPA	NM_000235.3
LIPT1	NM_145199.2
LIPT2	NM_001144869.2
LMBRD1	NM_018368.3
LONP1	NM_004793.3
LPIN1	NM_145693.2

GENE	TRANSCRIPT
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LYRM7	NM_181705.3
MAGT1	NM_032121.5
MAN1B1	NM_016219.4
MAN2B1	NM_000528.3
MANBA	NM_005908.3
MAOA	NM_000240.3
MARS2	NM_138395.3
MAT1A	NM_000429.2
MBTPS1	NM_003791.3
MCCC1	NM_020166.4
MCCC2	NM_022132.4
MCEE	NM_032601.3
MCOLN1	NM_020533.2
MECR	NM_016011.3
MFF*	NM_020194.5
MFN2	NM_014874.3
MFSD8	NM_152778.2
MGAT2	NM_002408.3
MGME1	NM_052865.3
MICU1	NM_006077.3
MIPEP	NM_005932.3
MLYCD	NM_012213.2
MMAA	NM_172250.2
MMAB	NM_052845.3
MMACHC	NM_015506.2
MMADHC	NM_015702.2
MNX1	NM_005515.3
MOCOS	NM_017947.2
MOCS1	NM_001358530.2
MOCS2A	NM_176806.3
MOCS2B	NM_004531.4
MOCS3	NM_014484.4
MOGS	NM_006302.2
MPC1	NM_016098.3
MPDU1	NM_004870.3
MPI	NM_002435.2
MPV17	NM_002437.4

GENE	TRANSCRIPT
MRPL12	NM_002949.3
MRPL3	NM_007208.3
MRPL40	NM_003776.3
MRPL44	NM_022915.3
MRPS14	NM_022100.2
MRPS16	NM_016065.3
MRPS2	NM_016034.4
MRPS22	NM_020191.2
MRPS23	NM_016070.3
MRPS34	NM_001300900.1
MRPS7	NM_015971.3
MSMO1	NM_006745.4
MSTO1*	NM_018116.3
MTFMT	NM_139242.3
MTHFD1	NM_005956.3
MTHFR*	NM_005957.4
MTO1	NM_012123.3
MTPAP	NM_018109.3
MTR	NM_000254.2
MTRR	NM_002454.2
MUT	NM_000255.3
NADK2	NM_001085411.2
NAGA	NM_000262.2
NAGLU	NM_000263.3
NAGS	NM_153006.2
NANS	NM_018946.3
NARS2	NM_024678.5
NAXE	NM_144772.2
NDUFA1	NM_004541.3
NDUFA10	NM_004544.3
NDUFA11	NM_175614.4
NDUFA12	NM_018838.4
NDUFA13	NM_015965.6
NDUFA2	NM_002488.4
NDUFA4	NM_002489.3
NDUFA6	NM_002490.4
NDUFA9	NM_005002.4
NDUFAF1	NM_016013.3
NDUFAF2	NM_174889.4

GENE	TRANSCRIPT
NDUFAF3	NM_199069.1
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NDUFAF5	NM_024120.4
NDUFAF6	NM_152416.3
NDUFAF7	NM_001083946.1
NDUFB11*	NM_019056.6
NDUFB3	NM_002491.2
NDUFB8	NM_005004.3
NDUFB9	NM_005005.2
NDUFS1	NM_005006.6
NDUFS2	NM_004550.4
NDUFS3	NM_004551.2
NDUFS4	NM_002495.3
NDUFS6	NM_004553.4
NDUFS7	NM_024407.4
NDUFS8	NM_002496.3
NDUFV1	NM_007103.3
NDUFV2	NM_021074.4
NEU1	NM_000434.3
NEUROD1	NM_002500.4
NEUROG3	NM_020999.3
NFS1	NM_021100.4
NFU1	NM_001002755.2
NGLY1	NM_018297.3
NKX2-2	NM_002509.3
NNT	NM_012343.3
NPC1	NM_000271.4
NPC2	NM_006432.3
NR0B1	NM_000475.4
NR2F1	NM_005654.5
NR3C1	NM_001018077.1
NSD1	NM_022455.4
NSUN3	NM_022072.3
NT5C3A	NM_016489.12
NUBPL	NM_025152.2
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NUS1	NM_138459.3
OAT*	NM_000274.3
OCRL	NM_000276.3

GENE	TRANSCRIPT
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OPA3	NM_025136.3
OPLAH	NM_017570.4
OTC	NM_000531.5
OTX2	NM_172337.2
OXCT1	NM_000436.3
PAH	NM_000277.1
PANK2	NM_153638.2
PAPSS2*	NM_001015880.1
PARS2	NM_152268.3
PAX4	NM_006193.2
PC	NM_000920.3
PCBD1	NM_000281.3
PCCA	NM_000282.3
PCCB	NM_000532.4
PCK1	NM_002591.3
PCK2	NM_004563.3
PCSK1	NM_000439.4
PDHA1	NM_000284.3
PDHB	NM_000925.3
PDHX	NM_003477.2
PDK3	NM_001142386.2
PDP1	NM_018444.3
PDSS1*	NM_014317.4
PDSS2	NM_020381.3
PDX1	NM_000209.3
PET100	NM_001171155.1
PEX1	NM_000466.2
PEX10	NM_153818.1
PEX11B	NM_003846.2
PEX12	NM_000286.2
PEX13	NM_002618.3
PEX14	NM_004565.2
PEX16	NM_004813.2
PEX19	NM_002857.3
PEX2	NM_000318.2
PEX26	NM_017929.5

GENE	TRANSCRIPT
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PEX6	NM_000287.3
PEX7	NM_000288.3
PFKM	NM_000289.5
PGAM2	NM_000290.3
PGAP1	NM_024989.3
PGAP2	NM_001256240.1
PGAP3	NM_033419.4
PGK1	NM_000291.3
PGM1*	NM_002633.2
PGM3	NM_001199917.1
PHEX	NM_000444.5
PHGDH	NM_006623.3
PHKA1	NM_002637.3
PHKA2	NM_000292.2
PHKB	NM_000293.2;NM_001031835.2
PHKG2	NM_000294.2
PHYH	NM_006214.3
PIGA	NM_002641.3
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PIGC	NM_002642.3
PIGG	NM_001127178.2
PIGL	NM_004278.3
PIGM	NM_145167.2
PIGN	NM_176787.4
PIGO	NM_032634.3
PIGP	NM_153681.2
PIGQ	NM_004204.3
PIGT	NM_015937.5
PIGU	NM_080476.4
PIGV	NM_017837.3
PIGW	NM_178517.3
PIGY	NM_001042616.2
PINK1	NM_032409.2
PITRM1	NM_001242309.1
PLA2G6	NM_003560.2
PMM2	NM_000303.2
PMPCA	NM_015160.2
PMPCB	NM_004279.2
PNKD	NM_015488.4
PNP	NM_000270.3
PNPLA8	NM_015723.4
PNPO	NM_018129.3
PNPT1	NM_033109.4
POFUT1	NM_015352.1
POGLUT1	NM_152305.2
POLG	NM_002693.2
POLG2	NM_007215.3
POMC	NM_001035256.2
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POMGNT2	NM_032806.5
POMK	NM_032237.4
POMT1	NM_007171.3
POMT2	NM_013382.5
POP1	NM_015029.2
PPA2	NM_176869.2
PPARG	NM_015869.4
PPM1K	NM_152542.4
PPOX	NM_000309.3
PPT1	NM_000310.3
PRDX1	NM_002574.3
PREPL	NM_006036.4
PRKCSH	NM_002743.3
PRODH*	NM_016335.4
PROP1	NM_006261.4
PROSC	NM_007198.3
PRPS1	NM_002764.3
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PSAT1	NM_058179.3
PSPH*	NM_004577.3
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PTS	NM_000317.2
PUS1	NM_025215.5
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QARS	NM_005051.2
QDPR	NM_000320.2
QRSL1	NM_018292.4
RANBP2*	NM_006267.4
RAPSN	NM_005055.4
RARS*	NM_002887.3
RARS2	NM_020320.3
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REEP1	NM_022912.2
REPS1	NM_001286611.1
RFT1	NM_052859.3
RFX6	NM_173560.3
RMND1	NM_017909.3
RNASEH1	NM_002936.4
RNASEH2A	NM_006397.2
RNASEH2B	NM_024570.3
RNASEH2C	NM_032193.3
RPN2	NM_002951.4
RPS6KA3	NM_004586.2
RRM2B	NM_015713.4
RXYLT1	NM_014254.2
SACS	NM_014363.5
SAMHD1	NM_015474.3
SAR1B*	NM_001033503.2
SARS2	NM_017827.3
SCN1A	NM_001165963.1
SCN4A	NM_000334.4
SCN8A	NM_014191.3;NM_001330260.1
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SCO2	NM_005138.2
SCP2	NM_002979.4
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SDHAF1	NM_001042631.2
SDHB	NM_003000.2
SDHC*	NM_003001.3
SDHD	NM_003002.3
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SEC23B	NM_006363.4
SEC24D	NM_014822.3
SEC63	NM_007214.4
SERAC1	NM_032861.3

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SIRT1	NM_012238.4
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SLC12A3	NM_000339.2
SLC13A3	NM_022829.5
SLC13A5	NM_177550.4
SLC16A1	NM_003051.3
SLC17A5	NM_012434.4
SLC18A2	NM_003054.4
SLC19A1	NM_194255.2
SLC19A2	NM_006996.2
SLC19A3	NM_025243.3
SLC1A2	NM_004171.3
SLC1A3	NM_004172.4
SLC1A4	NM_003038.4
SLC22A5	NM_003060.3
SLC25A1	NM_005984.4
SLC25A12	NM_003705.4
SLC25A13	NM_014251.2
SLC25A15	NM_014252.3
SLC25A19	NM_021734.4
SLC25A20	NM_000387.5
SLC25A21	NM_030631.3
SLC25A22	NM_024698.5
SLC25A26*	NM_001164796.1
SLC25A3	NM_005888.3
SLC25A32	NM_030780.4
SLC25A38	NM_017875.2
SLC25A4	NM_001151.3
SLC25A42	NM_178526.4
SLC25A46	NM_138773.2
SLC26A2	NM_000112.3
SLC2A1	NM_006516.2
SLC2A2	NM_000340.1
SLC30A10	NM_018713.2
SLC33A1	NM_004733.3
SLC34A1	NM_003052.4
SLC34A3	NM_080877.2
SLC35A1	NM_006416.4
SLC35A2	NM_001042498.2
SLC35A3	NM_012243.2
SLC35C1	NM_018389.4
SLC35D1	NM_015139.2
SLC37A4	NM_001164277.1
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SLC39A8	NM_022154.5
SLC3A1	NM_000341.3
SLC46A1	NM_080669.5
SLC52A1	NM_017986.3
SLC52A2	NM_024531.4
SLC52A3	NM_033409.3
SLC5A1	NM_000343.3
SLC6A1	NM_003042.3
SLC6A19	NM_001003841.2
SLC6A3	NM_001044.4
SLC6A5	NM_004211.3
SLC6A8	NM_005629.3
SLC6A9	NM_201649.3
SLC7A13	NM_138817.2
SLC7A7	NM_001126106.2
SLC7A9	NM_014270.4
SLC9A7	NM_001257291.1
SMPD1	NM_000543.4
SOX2	NM_003106.3
SOX3	NM_005634.2
SPAST	NM_014946.3
SPG7	NM_003119.3
SPR	NM_003124.4
SQSTM1	NM_003900.4
SRD5A3	NM_024592.4
SSR3	NM_001308197.1
SSR4	NM_001204526.1
ST3GAL3	NM_006279.3
ST3GAL5	NM_003896.3
STAT2	NM_005419.3
STT3A	NM_001278503.1
STT3B	NM_178862.2
STXBP1	NM_003165.3
SUCLA2	NM_003850.2
SUCLG1	NM_003849.3
SUCLG2	NM_001177599.1
SUGCT	NM_024728.2
SUMF1	NM_182760.3
SUOX	NM_000456.2
SURF1	NM_003172.3
TACO1	NM_016360.3
TANGO2	NM_152906.6
TARS2	NM_025150.4
TAT	NM_000353.2
TAZ	NM_000116.4
TBX19	NM_005149.2
TCN1	NM_001062.3
TCN2	NM_000355.3
TFAM	NM_003201.2
TGDS	NM_014305.3
TH	NM_199292.2
THAP11	NM_020457.2
TIMM50	NM_001001563.3
TIMM8A	NM_004085.3
TIMMDC1	NM_016589.3
TK2	NM_004614.4
TMEM126A	NM_032273.3
TMEM126B	NM_018480.4
TMEM165	NM_018475.4
TMEM199	NM_152464.2
TMEM70	NM_017866.5
TOP1MT	NM_052963.2
TOP3A	NM_004618.4
TPI1	NM_000365.5
TPK1	NM_022445.3
TPP1	NM_000391.3
TRAK1	NM_001042646.2
TRAP1	NM_016292.2
TRAPPC11	NM_021942.5

GENE	TRANSCRIPT
TRAPPC12	NM_001321102.1
TRAPPC2	NM_001011658.3
TRAPPC6B	NM_001079537.1
TRAPPC9	NM_031466.7
TREX1	NM_033629.4
TRIP11	NM_004239.4
TRIT1	NM_017646.5
TRMT10A	NM_152292.4
TRMT10C	NM_017819.3
TRMT5	NM_020810.3
TRMU	NM_018006.4
TRNT1	NM_182916.2
TRPM6	NM_017662.4
TSFM*	NM_001172696.1
TTC19	NM_017775.3
TTPA	NM_000370.3
TUFM	NM_003321.4
TUSC3	NM_006765.3
TWNK	NM_021830.4
TXN2	NM_012473.3
TYMP	NM_001953.4
UCP2	NM_003355.2
UMOD	NM_003361.3
UMPS	NM_000373.3
UQCC2	NM_032340.3
UQCC3	NM_001085372.2
UQCRB	NM_006294.4
UQCRC2	NM_003366.3
UQCRQ	NM_014402.4
UROD	NM_000374.4
UROS	NM_000375.2
VARS2	NM_001167734.1
VDR	NM_001017535.1
VMA21	NM_001017980.3
VPS13B	NM_017890.4
VPS33A	NM_022916.4
WARS2	NM_015836.3
WDR45	NM_007075.3
WFS1	NM_006005.3

GENE	TRANSCRIPT
XDH	NM_000379.3
XPNPEP3	NM_022098.3
XYLT1	NM_022166.3
XYLT2	NM_022167.3
YARS2	NM_001040436.2
YME1L1	NM_139312.2
ZFP57	NM_001109809.2
ZNF143	NM_003442.5

Methods

- Complete list of tests performed: Invitae Neurodegeneration with Brain Iron Accumulation Panel, Invitae Metabolic Newborn Screening Confirmation Panel, Invitae Elevated C14 and C14:1 (VLCAD deficiency) Test, Invitae Elevated Arginine (Arginase deficiency) Panel, Invitae Elevated Citrulline (Citrullinemia) Panel, Invitae Elevated Leucine (MSUD) Panel, Invitae Elevated Glycine Panel (including Glycine Encephalopathy), Invitae Elevated Tyrosine (Tyrosinemia) Panel, Invitae Alkaptonuria Test, Invitae Methylmalonic Acidemia and Homocystinuria Panel, Invitae Cystinuria Panel, Invitae Elevated Phenylalanine (Hyperphenylalaninemia) Panel, Invitae Elevated Proline (Hyperprolinemia) Panel, Invitae Galactosemia Panel, Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test, Invitae Congenital Disorders of Glycosylation Panel, Invitae Comprehensive Glycogen Storage Disease Panel, Invitae Hereditary Fructose Intolerance Test, Invitae Rare Carbohydrate Disorders Panel, Invitae Cerebrotendinous Xanthomatosis Test, Add-on Sitosterolemia Genes, Invitae Cerebral Creatine Deficiency Panel, Invitae Fatty Acid Oxidation Defects Panel, Invitae Elevated C6, C8 and C10 (MCAD deficiency) Test, Invitae Ketolysis Disorders Panel, Invitae Comprehensive Lysosomal Storage Disorders Panel, Add-on Chitotriosidase Deficiency Gene, Add-on Preliminary Evidence Gene, Add-on Adult-onset Neuronal Ceroid Lipofuscinoses Genes, Invitae Cystinosis Test, Invitae Wilson Disease Test, Invitae Mucopolysaccharidoses Plus (MPS+) Panel, Invitae Organic Acidemias Panel, Invitae 3-Methylcrotonyl-CoA Carboxylase Panel, Invitae Biotinidase Deficiency Test, Invitae Elevated C5-DC (Glutaric Aciduria Type I) Test, Invitae Elevated C4 and C5 (Multiple Acyl-CoA Dehydrogenase deficiency) Panel, Invitae Propionic Acidemia Panel, Invitae Copper Metabolism Disorders Panel, Invitae Neurotransmitter Disorders Panel, Invitae Hereditary Hyperekplexia Panel, Invitae Adult Refsum Disease Panel, Invitae Elevated Very Long Chain Fatty Acids Panel (including X-ALD), Invitae Zellweger Spectrum Disorder Panel, Invitae Urea Cycle Disorders Panel, Add-on Hyperammonemia Genes, Add-on Hereditary Orotic Aciduria Gene, Invitae Purine Metabolism Disorders Panel, Invitae Cystic Fibrosis Newborn Screening Confirmation Test, Invitae Treatable Neurometabolic Disorders Panel, Invitae Ornithine Transcarbamylase (OTC) Deficiency Test , Add-on Hereditary Orotic Aciduria Gene, Add-on Low Citrulline Genes, Invitae Acute Hepatic Porphyrias Panel, Invitae Primary Hyperoxaluria Panel, Invitae Alpha-1 Antitrypsin Deficiency Test, Invitae Monogenic Diabetes Panel, Invitae Comprehensive Porphyrias Panel, Invitae X-Linked Hypophosphatemia Test, Invitae Hypophosphatemia Panel, Invitae Nuclear Mitochondrial Disorders Panel, Invitae Supplemental Metabolic Newborn Screening Panel, Invitae Pyruvate Metabolism and Related Disorders Panel, Invitae Comprehensive Neurometabolic Disorders Panel, Invitae Hypoglycemia Panel
- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with ≥50x depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated below. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 10bp of flanking intronic sequence (20bp for BRCA1/2), and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. For some genes only targeted loci are analyzed (indicated in the table above). Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For PMS2 exons 12-15, the reference genome has been modified to force all sequence reads derived from PMS2 and the PMS2CL pseudogene to align to PMS2, and variant calling algorithms are modified to support an expectation of 4 alleles. If a rare SNP or indel variant is identified by this method, both PMS2 and the PMS2CL pseudogene are amplified by long-range PCR and the location of the variant is determined by Pacific Biosciences (PacBio) SMRT sequencing of the relevant exon in both long-range amplicons. If a CNV is identified, MLPA or MLPA-seq is run to confirm the variant. If confirmed, both PMS2 and PMS2CL are amplified by long-range PCR, and the identity of the fixed differences between PMS2 and PMS2CL are sequenced by PacBio from the long-range amplicon to disambiguate the location of the CNV. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778). For C9orf72 repeat expansion testing, hexanucleotide repeat units are detected by repeat-primed PCR (RP-PCR) with fluorescently labeled primers followed by capillary electrophoresis. Interpretation Reference Ranges: Benign (Normal Range): <25 repeat units, Uncertain: 25-30 repeat units, Pathogenic (Full Mutation): >=31 repeat units. A second round of RP-PCR utilizing a non-overlapping set of primers is used to confirm the initial call in the case of suspected allele sizes of 22 or more repeats. For RNA analysis of the genes indicated in the Genes Analyzed table, complementary DNA is synthesized by reverse transcription from RNA derived from a blood specimen and enriched for specific gene sequences using capture hybridization. After high-throughput sequencing using Illumina technology, the output reads are aligned to a reference sequence (genome build GRCh37; custom derivative of the RefSeq transcriptome) to identify the locations of exon junctions through the detection of split reads. The relative usage of exon junctions in a test specimen is assessed quantitatively and compared to the usage seen in control specimens. Abnormal exon junction usage is evaluated as evidence in the Sherloc variant interpretation framework. If an abnormal splicing

pattern is predicted based on a DNA variant outside the typical reportable range, as described above, the presence of the variant is confirmed by targeted DNA sequencing. RNA sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2094793). Technical component of Fibroblast cell-culturing and gDNA extraction from skin punch biopsy is performed by Invitae Corporation (5 Technology Drive, Irvine CA 92618, #05D1052995).

- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at **http://www.ncbi.nlm.nih.gov/pubmed**.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (**http://exac.broadinstitute.org**), gnomAD (**http://gnomad.broadinstitute.org**), and dbSNP (**http://ncbi.nlm.nih.gov/SNP**).
- A MedGen ID is a unique identifier referring to an article in MedGen, NCBI's centralized database of information about genetic disorders and phenotypes. Search by MedGen ID at **http://www.ncbi.nlm.nih.gov/medgen**. An OMIM number is a unique identifier referring to a comprehensive entry in Online Mendelian Inheritance of Man (OMIM). Search by OMIM number at **http://omim.org/**.
- Invitae uses information from individuals undergoing testing to inform variant interpretation. If "Invitae" is cited as a reference in the variant details this may refer to the individual in this requisition and/or historical internal observations.

Limitations

Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination. Invitae's RNA analysis is not designed for use as a stand-alone diagnostic method and cannot determine absolute RNA levels. Results from the RNA analysis may not be informative for interpreting copy number gains. GALM: Deletion/duplication analysis is not offered for exons 5-7. PAPSS2: Sequencing analysis is not offered for exon 3. GH1: Deletion/duplication and sequencing analysis is not offered for exon 1. SLC25A26: Deletion/duplication analysis is not offered for exon 5. CLCNKB: Deletion/duplication analysis is not offered for this gene. FH: Sequencing analysis for exons 9 includes only cds +/- 10 bp. MTHFR: The NM_005957.4:c.665C>T (p.Ala222Val) (aka 677C>T) and c.1286A>C (p.Glu429Ala) (aka 1298A>C) variants are not reported in our primary report. GPC3: Sequencing analysis for exons 3 includes only cds +/- 10 bp. MSTO1: Deletion/duplication analysis is not offered for exons 1-7, 10, 12-14 and sequencing analysis is not offered for exons 1-7, 10, 13-14. PIGB: Deletion/duplication analysis is not offered for exon 9. RARS: Deletion/duplication analysis is not offered for exon 14. Sequencing analysis for exons 14 includes only cds +/- 10 bp. GALC: Deletion/duplication analysis is not offered for exon 6. OAT: Deletion/duplication analysis is not offered for exon 2. PGM1: Deletion/duplication analysis is not offered for exon 11. SDHC: Sequencing analysis for exons 2, 6 includes only cds +/- 10 bp. FAH: Deletion/duplication analysis is not offered for exon 14. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. PRODH: Deletion/duplication analysis is not offered for exons 8, 12. PDSS1: Deletion/duplication analysis is not offered for exon 2. NDUFB11: Deletion/duplication and sequencing analysis is not offered for exon 1. TSFM: Sequencing analysis is not offered for exon 5. PSPH: Deletion/duplication and sequencing analysis is not offered for exons 4-5. CACNA1C: Deletion/duplication and sequencing analysis is not offered for exons 44-45. AMN: Deletion/duplication analysis is not offered for exon 1. DMXL2: Deletion/duplication analysis is not offered for exon 2. MFF: Deletion/duplication analysis is not offered for exon 3. COX10: Deletion/duplication and sequencing analysis is not offered for exon 6. RANBP2: Deletion/duplication and sequencing analysis is not offered for exons 1-11, 15-29. SERPINA1: Deletion/duplication analysis is not offered for exon 3. DDC: Deletion/duplication analysis is not offered for exons 10-11. SAR1B: Deletion/duplication analysis is not offered for exon 5. CFTR: Sequencing analysis for exons 7 includes only cds +/- 10 bp. SDHA: Deletion/duplication analysis is not offered for this gene and sequencing analysis is not offered for exon 14. Sequencing analysis for exons 6-8 includes only cds +/- 10 bp. IDS: Detection of complex rearrangements not offered (PMID: 7633410, 20301451).

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Invitae. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

Genes analyzed

A4GALT, AAAS, AARS2, AASS, ABAT, ABCB7, ABCC8, ABCD1, ABCD3, ABCD4, ABCG5, ABCG8, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACO2, ACOX1, ACOX2, ACSF3, ADA, ADAR, ADK, ADSL, AFG3L2, AGA, AGK, AGL, AGXT, AHCY, AIFM1, AK2, AKT2, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, AMACR, AMN*, AMPD1, AMT, AP1S1, AP4M1, APOPT1, APPL1, APRT, APTX, ARCN1, ARG1, ARHGEF9, ARSA, ARSB, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATIC, ATP13A2, ATP5A1, ATP5D, ATP5E, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, ATP7B, ATPAF2, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BAG3, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BLK, BOLA3, BSCL2, BSND, BTD, C12orf65, C19orf12, C19orf70, C1GALT1C1, C1QBP, CA5A, CACNA1C*, CACNA1D, CAD, CANT1, CARS2, CASR, CBS, CCDC115, CD320, CDKN1C, CEP89, CFTR*, CHAT, CHCHD10, CHIT1, CHST14, CHST3, CHST6, CHSY1, CLCN5, CLCNKB*, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CLPX, CNNM2, COA3, COA5, COA6, COA7, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10*, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, COX8A, CP, CPOX, CPS1, CPT1A, CPT2, CRAT, CSGALNACT1, CTNNB1, CTNS, CTSA, CTSD, CTSF, CTSK, CUBN, CYC1, CYCS, CYP27A1, CYP27B1, CYP2R1, CYP7B1, D2HGDH, DARS2, DBH, DBT, DCAF17, DDC*, DDOST, DES, DGUOK, DHCR7, DHDDS, DHFR, DHTKD1, DLAT, DLD, DMP1, DMXL2*, DNA2, DNAJC12, DNAJC19, DNAJC5, DNM1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EARS2, ECHS1, EGF, EIF2AK3, ELAC2, ENO3, ENPP1, EOGT, ETFA, ETFB, ETFDH, ETHE1, EXT1, EXT2, EXTL3, FA2H, FAH*, FAM111A, FAM20C, FARS2, FASTKD2, FBP1, FBXL4, FDX2, FECH, FGF23, FGFR1, FH*, FKRP, FKTN, FLAD1, FOLR1, FOXP3, FOXRED1, FTCD, FTL, FUCA1, FUK, FUT8, FXYD2, G6PC, G6PC3, G6PD, GAA, GABBR2, GABRA1, GABRA2, GABRB1, GABRB3, GABRG2, GAD1, GALC*, GALE, GALK1, GALM*, GALNS, GALNT3, GALT, GAMT, GANAB, GARS, GATA1, GATA4, GATA6, GATM, GBE1, GCDH, GCGR, GCH1, GCK, GCLC, GDAP1, GFER, GFM1, GFM2, GFPT1, GH1*, GHR*, GIF, GJA1, GLA, GLB1, GLDC, GLIS3, GLRA1, GLRB, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAS, GNE, GNMT, GNPTAB, GNPTG, GNS, GORAB, GOSR2, GOT2, GPAA1, GPC3*, GPHN, GRHPR, GRIN2B, GRIN2D, GRN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYG2, GYS1, GYS2, HADH, HADHA, HADHB, HARS2, HCCS, HCFC1, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HOGA1, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HSD3B7, HSPD1, HTRA2, HYAL1, IARS2, IBA57, IDH2, IDH3B, IDS*, IDUA, IER3IP1, IFIH1, IMPDH1, INS, INSR, ISCA1, ISCA2, ISCU, ISPD, IVD, JAGN1, KARS, KCNA1, KCNJ10, KCNJ11, KCTD7, KDM6A, KIF1A, KLF11, KMT2D, L2HGDH, LAMP2, LARGE1, LARS, LARS2, LDHA, LFNG, LHX3, LIAS, LIPA, LIPT1, LIPT2, LMBRD1, LONP1, LPIN1, LRPPRC, LYRM4, LYRM7, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MARS2, MAT1A, MBTPS1, MCCC1, MCCC2, MCEE, MCOLN1, MECR, MFF*, MFN2, MFSD8, MGAT2, MGME1, MICU1, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MNX1, MOCOS, MOCS1, MOCS2A, MOCS2B, MOCS3, MOGS, MPC1, MPDU1, MPI, MPV17, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34,

MRPS7, MSMO1, MSTO1*, MTFMT, MTHFD1, MTHFR*, MTO1, MTPAP, MTR, MTRR, MUT, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11*, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFS1, NFU1, NGLY1, NKX2-2, NNT, NPC1, NPC2, NR0B1, NR2F1, NR3C1, NSD1, NSUN3, NT5C3A, NUBPL, NUP62, NUS1, OAT*, OCRL, OGDH, OGT, OPA1, OPA3, OPLAH, OTC, OTX2, OXCT1, PAH, PANK2, PAPSS2*, PARS2, PAX4, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PCSK1, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1*, PDSS2, PDX1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGAP2, PGAP3, PGK1, PGM1*, PGM3, PHEX, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIGA, PIGB*, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PINK1, PITRM1, PLA2G6, PMM2, PMPCA, PMPCB, PNKD, PNP, PNPLA8, PNPO, PNPT1, POFUT1, POGLUT1, POLG, POLG2, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POP1, PPA2, PPARG, PPM1K, PPOX, PPT1, PRDX1, PREPL, PRKCSH, PRODH*, PROP1, PROSC, PRPS1, PSAP, PSAT1, PSPH*, PTF1A, PTS, PUS1, PYGL, PYGM, QARS, QDPR, QRSL1, RANBP2*, RAPSN, RARS*, RARS2, RBCK1, REEP1, REPS1, RFT1, RFX6, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RPN2, RPS6KA3, RRM2B, RXYLT1, SACS, SAMHD1, SAR1B*, SARS2, SCN1A, SCN4A, SCN8A, SCO1, SCO2, SCP2, SDHA*, SDHAF1, SDHB, SDHC*, SDHD, SEC23A, SEC23B, SEC24D, SEC63, SERAC1, SERPINA1*, SFXN4, SGSH, SIRT1, SLC10A7, SLC12A1, SLC12A3, SLC13A3, SLC13A5, SLC16A1, SLC17A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26*, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC26A2, SLC2A1, SLC2A2, SLC30A10, SLC33A1, SLC34A1, SLC34A3, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A8, SLC3A1, SLC46A1, SLC52A1, SLC52A2, SLC52A3, SLC5A1, SLC6A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A13, SLC7A7, SLC7A9, SLC9A7, SMPD1, SOX2, SOX3, SPAST, SPG7, SPR, SQSTM1, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STAT2, STT3A, STT3B, STXBP1, SUCLA2, SUCLG1, SUCLG2, SUGCT, SUMF1, SUOX, SURF1, TACO1, TANGO2, TARS2, TAT, TAZ, TBX19, TCN1, TCN2, TFAM, TGDS, TH, THAP11, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM165, TMEM199, TMEM70, TOP1MT, TOP3A, TPI1, TPK1, TPP1, TRAK1, TRAP1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TREX1, TRIP11, TRIT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TSFM*, TTC19, TTPA, TUFM, TUSC3, TWNK, TXN2, TYMP, UCP2, UMOD, UMPS, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UROD, UROS, VARS2, VDR, VMA21, VPS13B, VPS33A, WARS2, WDR45, WFS1, XDH, XPNPEP3, XYLT1, XYLT2, YARS2, YME1L1, ZFP57, ZNF143

This report has been reviewed and approved by:

Matteo Vatta, Ph.D., FACMG
Clinical Molecular Geneticist