

ACCESSION NO.	DATE / /
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PATIENT INFORMATION

PATIENT LAST NAME	PATIENT FIRST NAME	
DOB / /	BIOLOGICAL GENDER <input type="checkbox"/> M <input type="checkbox"/> F	ETHNICITY
TELEPHONE	EMAIL	
ADDRESS	CITY/STATE/ZIP	
BIOLOGICAL MOTHER LAST NAME	BIOLOGICAL FATHER LAST NAME	
BIOLOGICAL MOTHER FIRST NAME	BIOLOGICAL FATHER FIRST NAME	
BIOLOGICAL MOTHER DATE OF BIRTH / /	BIOLOGICAL FATHER DATE OF BIRTH / /	

ORDERING PROVIDER INFORMATION

PROVIDER SIGNATURE OF CONSENT (REQUIRED): I certify that this patient (and/or their legal guardian, as necessary) has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained a signed informed consent from this patient or their legal guardian for this testing in accordance with applicable laws and regulations, including N.Y. Civil Rights Law Section 79-L, and will retain this consent in the patient's medical record.

SIGNATURE _____ DATE / /

CLINICAL INDICATION (PLEASE FILL OUT ADDITIONAL INDICATIONS ON BACK)

SPECIMEN TYPE:
 Patient: Peripheral Blood Saliva Other: _____ Date of Collection: / /
 Biological mother: Peripheral Blood Saliva Other: _____ Date of Collection: / /
 Biological father: Peripheral Blood Saliva Other: _____ Date of Collection: / /
Parental samples will be used as needed in follow-up to patient testing
Please submit separate signed general consent form for each sample submitted (including parents)
For all prenatal specimens: please use Prenatal Requisition with supplemental Phenotype forms completed

BILLING INFORMATION <input type="checkbox"/> Bill Clinic <input type="checkbox"/> Bill Insurance Below <input type="checkbox"/> Self Pay		
POLICYHOLDER LAST NAME	POLICYHOLDER FIRST NAME	POLICYHOLDER DOB / /
INSURANCE CARRIER	INSURANCE ID	GROUP NO.
BILLING ADDRESS		
OTHER HEALTH COVERAGE (IDENTIFY)		
SELF-PAY: <input type="checkbox"/> Credit Card <input type="checkbox"/> Check		
Make Checks Payable to: Mount Sinai Genomics Inc., P.O. Box 21312, New York, NY 10087-1312		

PATIENT CLINICAL STATUS	PURPOSE OF STUDY
<input type="checkbox"/> Affected	<input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier Testing
<input type="checkbox"/> Unknown (no screening/evaluation)	<input type="checkbox"/> Research Study <input type="checkbox"/> Clinical Study
<input type="checkbox"/> Unaffected (all screening/evaluations(s) normal)	<input type="checkbox"/> Familial Follow-up (family variant)
	<input type="checkbox"/> Other _____

PLEASE COMPLETE ALL CLINICAL QUESTIONS ON THE BACK PAGES

ASSIGNMENT AND RELEASE: I hereby authorize my insurance benefits be paid directly to the provider and I understand that I am financially responsible for uncovered services. I also authorize the release of any information required to process the claim. Billing inquiries, please call 800-298-6570, Option 3.

SIGNATURE _____ DATE / /

LABORATORY TEST(S) ORDERED (SEE FOLLOWING PAGES FOR GENE LISTS)

HEARING AND VISION LOSS

- COMPREHENSIVE HEARING AND VISION LOSS PANEL (308 genes) includes subpanels listed below.
 - COMPREHENSIVE VISION LOSS PANEL (250 genes) includes subpanels listed below.
 - ALBINISM, HERMANSKY-PUDLAK SYNDROME, & WAARDENBURG SYNDROME PANEL (18 genes)
 - DEVELOPMENTAL EYE PANEL (21 genes)
 - RETINAL DISEASE PANEL (154 genes)
 - STICKLER & CATARACT PANEL (41 genes)
 - COMPREHENSIVE HEARING LOSS PANEL (92 genes) includes subpanels listed below.
 - BRANCHIO-OTO-RENAL SYNDROME PANEL (3 genes)
 - CONNEXIN 26 / CONNEXIN 30 DEL / DUP HEARING LOSS PANEL (GJB2/ GJB6)
 - OTOANCORIN NGS AND DEL / DUP (OTOA)
 - STEREOCILIN DEL / DUP (STRC)
 - USHER SYNDROME PANEL (11 genes)
 - ZELLWEGER SYNDROME PANEL (9 genes)
 - Add on ULTRA-HIGH RESOLUTION HEARING LOSS DEL / DUP ARRAY if panel is negative or inconclusive
 - Run simultaneous to panel

SKELETAL

- ACHONDROPLASIA (FGFR3)
 - FGFR3 Hotspot Panel reflex to sequencing if negative
 - FGFR3 Full Gene Sequencing
- CRANIOSYNOSTOSIS (8 genes)
- HYPOPHOSPHATASIA (ALPL)
- LIMB DEFECTS PANEL (8 genes)
- ROBERTS SYNDROME (ESCO2)

NEUROMUSCULAR

- DUCHENNE MUSCULAR DYSTROPHY (NGS and DEL / DUP)
- SPINAL MUSCULAR ATROPHY (SMN1/SMN2 - DEL / DUP)

CARDIOVASCULAR

- COMPREHENSIVE CARDIOVASCULAR PANEL (241 genes) includes subpanels listed below.
 - COMPREHENSIVE ARRHYTHMIAS PANEL (54 genes) includes subpanels listed below.
 - ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC) subpanel (8 genes)
 - BRUGADA SYNDROME (20 genes)
 - CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT) subpanel (8 genes)
 - LONG/SHORT QT SYNDROME (LSQT) subpanel (19 genes)
 - COMPREHENSIVE CARDIOMYOPATHY PANEL (190 genes), includes subpanels below and Comprehensive arrhythmias panel.
 - DILATED CARDIOMYOPATHY (57 genes)
 - HYPERTROPHIC CARDIOMYOPATHY (HCM) (40 genes)
 - LEFT VENTRICULAR NON-COMPACTION PANEL (LVNC) (20 genes)
 - METABOLIC CARDIOMYOPATHIES PANEL (24 genes)
 - AORTOPATHIES PANEL (33 genes)
 - CONGENITAL HEART DISEASE PANEL (44 genes)
 - FAMILIAL HYPERCHOLESTEROLEMIA PANEL (4 genes)
 - HEREDITARY HEMORRHAGIC TELANGIECTASIA PANEL (5 genes)
 - NOONAN SPECTRUM DISORDERS PANEL (19 genes)
 - PULMONARY HYPERTENSION PANEL (10 genes)
 - Add on ULTRA-HIGH RESOLUTION CARDIOVASCULAR DEL/DUP ARRAY if panel is negative or inconclusive
 - Run simultaneous to panel

NEURODEVELOPMENTAL

- COMPREHENSIVE EPILEPSY AND AUTISM PANEL (401 genes) includes subpanels listed below.
 - COMPREHENSIVE EPILEPSY PANEL (226 genes) includes subpanels listed below.
 - FOCAL, GENERALIZED, AND MYOCLONIC EPILEPSY PANEL (52 genes)
 - INFANTILE EPILEPSY PANEL (58 genes)
 - MIGRAINE PANEL (7 genes)
 - NEURONAL CEROID LIPOFUSCINOSES PANEL (9 genes)
 - NEURONAL MIGRATION PANEL (22 genes)
 - SYNDROMIC EPILEPSY AND INTELLECTUAL DISABILITY PANEL (93 genes)
 - COMPREHENSIVE AUTISM PANEL (228 genes) includes subpanels listed below.
 - FRAGILE X SYNDROME (FMR1) Full Gene Sequencing CGG Repeat
 - STAT AUTISM PANEL (30 genes)
 - MICROCEPHALY PANEL (78 genes)
 - Add on EPILEPSY PHARMACOGENETIC PANEL (8 genes)
 - Add on ULTRA-HIGH RESOLUTION NEURODEVELOPMENTAL DEL / DUP ARRAY if panel is negative or inconclusive
 - Run simultaneous to panel
 - Add on Chromosome Microarray (aCGH 180K +SNP)

IMMUNODEFICIENCY

- COMPREHENSIVE IMMUNODEFICIENCY PANEL (250 genes) includes subpanels listed below.
 - PRIMARY IMMUNODEFICIENCY PANEL (206 GENES)
 - INFLAMMATORY BOWEL DISEASE PANEL (59 GENES)
 - SEVERE COMBINED IMMUNODEFICIENCY PANEL (26 GENES)
 - Add on ULTRA-HIGH RESOLUTION IMMUNODEFICIENCY DEL/DUP ARRAY if panel is negative or inconclusive
 - Run simultaneous to panel
 - METABOLIC**
 - PORPHYRIA
 - Acute Porphyria Panel (AIP, HCP & VP)
 - ACUTE INTERMITTENT PORPHYRIA (AIP)
 - HEREDITARY COPROPORPHYRIA (HCP)
 - VARIEGATE PORPHYRIA (VP)
 - CONGENITAL ERYTHROPOIETIC PORPHYRIA (CEP)
 - ERYTHROPOIETIC PROTOPORPHYRIA (EPP)
 - PEPT2 (SLC15A2) Genotyping for Acute Porphyria Patients
 - PORPHYRIA CUTANEA TARDA (PCT)
 - METABOLIC DISORDERS SINGLE GENE DIAGNOSTIC TESTING:
 - AMINOACIDOPATHIES AND UREA CYCLE DISORDERS: gene _____
 - CHOLESTEROL: gene _____
 - DISORDERS OF CARBOHYDRATE METABOLISM: gene _____
 - FATTY ACID OXIDATION DISORDERS: gene _____
 - LYSOSOMAL STORAGE DISORDERS AND OTHER DISORDERS: gene _____
 - ORGANIC ACIDEMIAS : gene _____
 - PEROXISOMAL STORAGE DISEASES: gene _____
- Please specify gene to be analyzed in space provided; see back pages for available genes

SINGLE GENE DIAGNOSTIC TESTING

- SINGLE GENE DIAGNOSTIC TESTING
 See back pages for available genes
 Please note: any gene included on a panel may be ordered individually

OTHER

- TARGETED TESTING: gene _____ variant _____
 proband _____
- Familial follow-up to proband SEMA4 lab number: _____
- DNA extraction and Hold _____

*Targeted genotyping only.

PATIENT NAME	DATE OF BIRTH	PHYSICIAN NAME	PRACTICE NAME
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PHENOTYPE

Detailed medical records, clinical summary, pictures and family history must be attached.

PEDIGREE

Yes (Provide Details Below)

No

Unknown

- Yes (Provide Details Below)
- Alcohol Withdrawal
- Drug/Toxin-Induced
- Head Injury
- Known Environmental Risk Factors
- List Drugs Used (If Known)
- List Toxins Exposed If Known
- Maternal teratogenic exposure
- Metabolic Or Electrolyte Imbalance
- Systemic Infection
- Triggered by sleep deprivation
- Triggered by stress
- Other: _____
- No
- Unknown

- BEHAVIORAL FINDINGS**
- Yes (Provide Details Below)
 - Abnormal aggressive, impulsive or violent behavior
 - Attention deficit hyperactivity disorder
 - Autism
 - Autistic behavior
 - Hyperactivity
 - Obsessive-compulsive behavior
 - Psychiatric Abnormalities
 - Psychosis
 - Short attention span
 - Specific learning disability
 - Stereotypy
 - Other: _____
 - No
 - Unknown

BRAIN MALFORMATIONS/ABNORMAL IMAGING: *PLEASE ATTACH NEUROIMAGING STUDIES IF AVAILABLE.

- Yes (Provide Details Below)
- Abnormal corpus callosum morphology
- Abnormal morphology of the cerebellar cortex
- Abnormality of head blood vessel
- Abnormality of the basal ganglia
- Abnormality of the cerebral cortex
- Agenesis of corpus callosum
- Aplasia/Hypoplasia of the corpus callosum
- Brain atrophy
- Calcification of the small brain vessels
- Cerebellar atrophy
- Cerebellar hypoplasia
- Cerebral atrophy
- Cortical dysplasia
- Frontotemporal cerebral atrophy
- Hemimegalencephaly
- Heterotopia
- Holoprosencephaly
- Hydrocephalus
- Hypoplasia of the brainstem
- Hypoplasia of the corpus callosum
- Lissencephaly
- Molar tooth sign on MRI
- Pachygyria
- Polymicrogyria
- Ventriculomegaly
- Other: _____
- No
- Unknown

- CARDIOVASCULAR**
- Yes (Provide Details Below)
 - Abnormal morphology of the great vessels
 - Angioedema
 - Aortic aneurysm
 - Aortic root aneurysm
 - Aortic valve stenosis
 - Arrhythmia
 - Atrial cardiomyopathy
 - Atrial fibrillation
 - Atrial septal defect
 - Bradycardia
 - Cardiac arrest
 - Coarctation of aorta
 - Complete heart block with broad QRS complexes
 - Congenital malformation of the great arteries
 - Congestive heart failure
 - Dilated cardiomyopathy
 - Hypertension
 - Hypertrophic cardiomyopathy
 - Hypotension
 - Left ventricular hypertrophy
 - Left ventricular noncompaction
 - Lymphedema
 - Mitral regurgitation
 - Mitral valve prolapse
 - Myocardial infarction
 - Prolonged QT interval
 - Pulmonary arterial hypertension
 - Pulmonic stenosis
 - Shortened QT interval
 - Sudden cardiac death
 - Tetralogy of Fallot
 - Vasculitis
 - Ventricular fibrillation
 - Ventricular septal defect

- CARDIOVASCULAR (continued):**
- Ventricular tachycardia
 - Other: _____
 - No
 - Unknown

- CRANIOFACIAL DYSMORPHISM**
- Yes (Provide Details Below)
 - Head
 - Craniosynostosis
 - Frontal bossing
 - Macrocephaly
 - Microcephaly
 - Sloping forehead
 - White forelock
 - Face
 - Coarse facial features
 - Eyes
 - Aniridia
 - Bilateral microphthalmos
 - Blepharospasm
 - Blue sclerae
 - Cataract
 - Coloboma
 - Downslanted palpebral fissures
 - Ectopia lentis
 - Epicanthus
 - Heterochromia
 - Hypertelorism
 - Lisch nodules
 - Microphthalmia
 - Nystagmus
 - Ptosis
 - Strabismus
 - Unilateral microphthalmos
 - Vivid blue eyes

- Ears
- Ear abnormalities
- Ear tags
- Low-set ears
- Posteriorly rotated ears
- Nose
- Depressed nasal bridge
- Prominent nasal bridge
- Mouth
- Cleft lip
- Cleft palate
- High palate
- Long philtrum
- Macrotia
- Micrognathia
- Robin Sequence

- Neck
- Branchial arch abnormality
- Cystic hygroma
- Short neck
- Webbed neck
- Other: _____
- No
- Unknown

- GASTROINTESTINAL**
- Yes (Provide Details Below)
 - Abdominal pain
 - Aganglionic megacolon
 - Chronic diarrhea
 - Cirrhosis
 - Constipation
 - Diarrhea
 - Elevated hepatic transaminase
 - Gastritis
 - Gastroesophageal reflux
 - Gastroschisis
 - Hepatic failure
 - Hepatic fibrosis
 - Hepatomegaly
 - Inflammation of the large intestine
 - Malabsorption
 - Pyloric stenosis
 - Secretory diarrhea
 - Vomiting
 - Other: _____
 - No
 - Unknown

- GENITOURINARY**
- Yes (Provide Details Below)
 - Abnormal renal morphology
 - Abnormality of the urinary system
 - Cryptorchidism
 - Hydronephrosis
 - Micropenis
 - Nephronopththisis
 - Renal abnormality
 - Renal agenesis
 - Renal cyst
 - Renal tubular dysfunction
 - Other: _____
 - No
 - Unknown

- GROWTH:**
- Yes (Provide Details Below)

- ICD-10 CODES**
- 299.0 Autism, Current Infantile Or Childhood
 - F84.0 Autistic Disorder
 - G40 Epilepsy And Recurrent Seizures
 - H54.7 (Unspecified Visual Loss)
 - H90.5 (Unspecified Sensorineural Hearing Loss)
 - Q02 (Microcephaly)
 - Q04.9 (Congenital Malformation Of Brain, Unspecified)
 - Other: _____

- FAMILY HISTORY (PLEASE INCLUDE PEDIGREE)**
- Yes (Please Indicate Any Family Relatives With Clinical History Of Disease _____)
 - Cognitive impairment
 - Global developmental delay
 - Spontaneous abortion
 - Stillbirth
 - Other: _____
 - No
 - Unknown

- HISTORY OF CONSAQUINITY**
- Yes *Please Submit Pedigree If Available.*
 - Paternal Ancestry: _____
 - Maternal Ancestry: _____
 - No
 - Unknown

- HISTORY OF PREVIOUS TESTING (PLEASE ATTACH DETAILS)**
- Yes (Provide Details Below)
 - Chromosomal Microarray
 - Fragile X Testing
 - Karyotype
 - Sequencing Studies
 - Other: _____
 - No
 - Unknown

- AGE OF ONSET:**
- Adult onset
 - Childhood onset
 - Congenital onset
 - Infantile onset
 - Neonatal onset
 - Young adult onset
 - Other: _____

- PERINATAL OR PRENATAL HISTORY**
- Yes (Provide Details Below)
 - Hydrocephalus
 - Intrauterine growth retardation
 - Macrocephaly at birth
 - Oligohydramnios
 - Polyhydramnios
 - Preeclampsia
 - Premature birth
 - Seizures
 - Other: _____
 - No
 - Unknown

- OTHER FACTORS**
- Yes (Provide Details Below)

PATIENT NAME	DATE OF BIRTH	PHYSICIAN NAME	PRACTICE NAME
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Please submit separate signed general consent form for each sample submitted (including parents)

Informed Consent for Genetic Testing

I, _____, hereby request genetic testing, which may include molecular, cytogenetic and/or biochemical analyses, for

Myself

My child _____

I have received verbal and written information (please see sema4.com/testcatalog for test-specific information sheet) from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I/my child am about to undergo.

I understand that specimen(s), such as a peripheral blood, saliva, cheek swab, dried blood spot, skin biopsy, amniotic fluid, chorionic villi and/or urine sample, will be taken from me/my child. I understand that the samples will be used for determining if I/my child have a genetic disease, are carriers of a genetic disease, or are more likely to develop a genetic disease or condition.

The nature of the genetic test, _____, has been explained to me and the accuracy of the test and its risks and limitations have been detailed. I understand that infrequent errors may occur, even though the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small. The likelihood of this occurring has been estimated to be less than 1%. I understand that a negative result reduces, but does not eliminate, the possibility that I/my child carry a mutation(s) in the gene(s) analyzed or in other gene(s) that are not included in the test.

I understand that no test will be performed on my sample other than the one(s) authorized by this consent and my doctor.

De-identified research

Sema4 may also give the de-identified information to its research partners and may submit this de-identified information to research databases for use in scientific and medical research, including scientific databases that are maintained by the federal government, such as a database kept by the National Institutes of Health ("NIH") (an agency of the federal government that funds research). Researchers have to apply to the NIH to see the information in the database. Any information that could directly identify you (such your name or address) will not be provided to a scientific database.

If I prefer not to have any of my/my child's de-identified health information used in research consistent with this consent, I may initial here _____ or request this by contacting Sema4, including by emailing privacy@sema4.com.

Permission to contact

I understand that Sema4 may wish to contact me/my child in the future, including for the following reasons: research purposes, the provision of general information about research findings, and/or the provision of information about the results of tests on my/my child's sample(s). I understand that I may notify Sema4 to opt out of such future contact, including by emailing privacy@sema4.com.

I understand that this testing may yield results that are of unknown clinical significance and that parental or other relative's specimens may also be tested to determine whether a specific finding was inherited. In addition, incidental findings that are not related to the primary diagnosis may be identified in me/my child.

An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have stated and this test may detect non-paternity.

The results of my/my child's test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a geneticist. I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf.

I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.

Sample storage

By initialing here, I agree that Sema4 may store, de-identify, and use my/my child's sample and information to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and that I/my child will receive no compensation in connection with such research. If I do not initial here, my/my child's sample will be destroyed at the end of the testing process or not more than 60 days after collection. I understand that I may withdraw this consent by contacting Sema4 (including by emailing privacy@sema4.com).

Initials

Signature of person being tested (or guardian)

Date

Rev.10/4/2018

PANEL TABLE

CARDIOVASCULAR	Genes
COMPREHENSIVE CARDIOVASCULAR PANEL Number of markers: 241	A2ML1, ABCC9, ACADVL, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, ADAMTS2, AGK, AGL, AKAP9, ALG1, ALG12, ALMS1, ALPK3, ANK2, ANKRD1, APOB, ARSB, ATP7A, BAG3, BCOR, BMPR1B, BMPR2, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV1, CAV3, CAVIN4, CBL, CBS, CHD7, CHRM2, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COX10, COX15, CPT2, CRELD1, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAH5, DOLK, DSC2, DSP, DTNA, EFEMP2, EIF2AK4, ELAC2, ELN, EMD, ENG, EYA4, FBN1, FBN2, FHL1, FKBP14, FKBP, FKTN, FLNA, FLNC, FOXH1, GAA, GATA4, GATA6, GATAD1, GBA, GBE1, GDF1, GDF2, GJA1, GJA5, GLA, GLB1, GPC3, GPD1L, GUSB, HADHA, HAND1, HCN4, HEXB, HRAS, IDUA, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LEFTY2, LMNA, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MED13L, MEIS2, MFAP5, MIB1, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF6, NDUFS3, NDUFS4, NDUFS5, NDUFS8, NEBL, NEXN, NKX2-5, NOTCH1, NPPA, NRAS, OBSCN, PCCA, PCCB, PDLIM3, PET100, PGM1, PHYH, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PPP1CB, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCD1, SDHA, SDHAF1, SGCD, SGGH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLC39A13, SLMAP, SMAD3, SMAD4, SMAD6, SMAD9, SNTA1, SOS1, SOS2, SPRED1, SURF1, TAZ, TBX1, TBX5, TCFAP, TGFBR2, TGFBR3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNN3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3
COMPREHENSIVE CARDIOMYOPATHY PANEL Number of markers: 190	ABCC9, ACADVL, ACTA2, ACTC1, ACTN2, AGK, AGL, AKAP9, ALG1, ALG12, ALMS1, ALPK3, ANK2, ANKRD1, ARSB, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CAVIN4, CBL, CBS, CHRM2, COL3A1, COL5A1, COL5A2, COX10, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSP, DTNA, ELAC2, EMD, EYA4, FBN1, FBN2, FHL1, FKBP, FKTN, FLNA, FLNC, GAA, GATA4, GATA6, GATAD1, GBA, GBE1, GJA5, GLA, GLB1, GPD1L, GUSB, HADHA, HCN4, HEXB, HRAS, IDUA, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MED12, MIB1, MLYCD, MTO1, MUT, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF6, NDUFS3, NDUFS4, NDUFS5, NDUFS8, NEBL, NEXN, NKX2-5, NOTCH1, NPPA, NRAS, OBSCN, PCCA, PCCB, PDLIM3, PET100, PGM1, PHYH, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCD1, SDHA, SDHAF1, SGCD, SGGH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SURF1, TAZ, TBX1, TBX5, TCFAP, TGFBR2, TGFBR3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNN3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL
COMPREHENSIVE ARRHYTHMIAS PANEL Number of markers: 54 ARRYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY PANEL Number of markers: 8 BRUGADA SYNDROME PANEL Number of markers: 20 CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA PANEL Number of markers: 8 LONG/SHORT QT SYNDROME PANEL Number of markers: 19	ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, TRPM4 ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN
DILATED CARDIOMYOPATHY PANEL Number of markers: 57 HYPERTROPHIC CARDIOMYOPATHY PANEL Number of markers: 40 LEFT VENTRICULAR NONCOMPACTION PANEL Number of markers: 20 AORTOPATHIES PANEL Number of markers: 33 CONGENITAL HEART DISEASE PANEL Number of markers: 44 FAMILIAL HYPERCHOLESTEROLEMIA PANEL Number of markers: 4 HEREDITARY HEMORRHAGIC TELANGIECTASIA PANEL Number of markers: 5 METABOLIC CARDIOMYOPATHIES PANEL Number of markers: 24 NOONAN SPECTRUM DISORDERS PANEL Number of markers: 19 PULMONARY HYPERTENSION PANEL Number of markers: 10	ABCC9, ACTC1, ACTN2, AKAP9, ANK2, ANKRD1, BAG3, CACNA1C, CAV3, CAVIN4, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSP, ELAC2, EMD, EYA4, FKTN, FLNC, GATAD1, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, OBSCN, PKP2, PLN, PRDM16, RAF1, RBM20, RAS, SCD, SLC22A5, TNNC1, TNN3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL ACTC1, ACTN2, AGL, ANKRD1, BAG3, CACNA1C, CAV3, CPT2, CSRP3, DES, DMD, FHL1, FLNC, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, TCFAP, TNNC1, TNN3, TNNT2, TPM1, TTR, VCL ACTC1, DSP, DTNA, HCN4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH7, PLEKHM2, PLN, PRDM16, RYR2, SCN5A, TAZ, TNN3, TNNT2, TPM1, VCL ACTA2, ADAMTS2, ATP7A, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FKBP14, FLNA, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SLC39A13, SMAD3, SMAD4, SMAD6, TGFBR2, TGFBR3, TGFBR1, TGFBR2 A2ML1, ACTC1, ACVR2B, ALMS1, BCOR, BRAF, CBL, CHD7, CRELD1, DNAH5, ELN, FOXH1, GATA4, GATA6, GDF1, GJA1, GPC3, HAND1, HRAS, JAG1, KRAS, LEFTY2, MAP2K1, MAP2K2, MED13L, MEIS2, MYH6, NKX2-5, NKX2-6, NODAL, NOTCH1, NR2F2, NRAS, NSD1, PTPN11, RAF1, RIT1, SHOC2, SMAD6, SOS1, TBX1, TBX5, ZFPM2, ZIC3 APOB, LDLR, LDLRAP1, PCSK9 ACVRL1, ENG, GDF2, RASA1, SMAD4 ACADVL, AGL, ALG1, ALG12, ARSB, CPT2, DOLK, FKRP, GAA, GBA, GBE1, GLA, GLB1, GUSB, HADHA, HEXB, IDUA, LAMP2, MTO1, PCCA, PCCB, SLC22A5, SLC25A20, TAZ A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1 ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNA5, KCNK3, SMAD9
HEARING AND VISION LOSS	Genes
COMPREHENSIVE HEARING AND VISION LOSS PANEL Number of markers: 308	ABCA4, ABHD12, ACTG1, ADAM9, ADGRV1, AGK, AH1, AIFM1, AIPL1, ALMS1, AP3B1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BLOC1S6, BMP4, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1D, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCDC50, CDH23, CDH3, CDHR1, CEACAM16, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLDN14, CLN3, CLN5, CLN6, CLRN1, CNGA1, CNGB3, CNGB3, CNM1A, CNM1B, COCH, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP11B1, CYP4V2, DHDDS, DNJAC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, ESPN, ESRRB, EYA1, EYA4, EYS, FAM161A, FLVCR1, FOXC1, FOXC2, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FYCO1, FZD4, GCNT2, GIPC3, GJA8, GJB2, GJB6, GNA11, GNA12, GNPTG, GPR143, GPR179, GRSMD2, GRHL2, GRP1, GRK1, GRM6, GRXC1, GSDME, GUC1A1, GUC1A1B, GUCY2D, HARS, HCCS, HESX1, HGF, HPS1, HPS2, HPS4, HPS5, HPS6, HSF4, IFT140, ILDR1, IMPDH1, IMPG2, INPPE5, IQCB1, JAG1, KARS, KCNJ13, KCNQ1, KCNQ4, KCNV2, KIF11, KIF7, KLHL7, LCA5, LHFP15, LOXDH1, LRAT, LRIT3, LRP5, LRTO, LYST, LZTFL1, MAK, MARVELD2, MERTK, MFRP, MFSDB, MITF, MKKS, MKS1, MSRB3, MT-RNR1, MTPP, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, MYOC, NDP, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTOA, OTOF, OTOG, OTOG1, OTX2, P2RX2, PANK2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PGK1, PHYH, PITPNM3, PITX2, PITX3, PJK, PLA2G5, POU3F4, POU4F3, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PTPRO, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RDX, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI1, RPI1L, RP2, RPE65, RPRG, RPRG1P1, RPRG1P1L, RS1, SAG, SDCCAG8, SEMA4A, SERPINC6, SIX1, SIX5, SLC24A1, SLC26A4, SLC45A2, SMOG1, SMPX, SNRNP200, SOX10, SOX2, SPATA7, STRA6, STRC, TBC1D24, TCTN1, TCTN2, TCTN3, TDRD7, TECTA, TGFBI, TIMM8A, TIMP3, TMC1, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TMIE, TMPRSS3, TOPORS, TPP1, TRXN1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423
COMPREHENSIVE HEARING LOSS PANEL Number of markers: 92	ACTG1, ADGRV1, AIFM1, CACNA1D, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLRN1, COCH, COL11A2, DNJAC5, EDN3, EDNRB, ESPN, ESRRB, EYA1, EYA4, GIPC3, GJB2, GJB6, GRSMD2, GRHL2, GRXC1, GSDME, HARS, HGF, ILDR1, KARS, KCNQ1, KCNQ4, LHFP15, LOXDH1, LRTO, MARVELD2, MITF, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1, OTOA, OTOF, OTOG, OTOG1, P2RX2, PAX3, PCDH15, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PHYH, PJK, POU3F4, POU4F3, PRPS1, PTPRO, RDX, SERPINC6, SIX1, SIX5, SLC26A4, SMPX, SOX10, STRC, TBC1D24, TECTA, TIMM8A, TMC1, TMEM126A, TMIE, TMPRSS3, TRPN, TROBP, TSPEAR, USH1C, USH1G, USH2A, WFS1, WHRN
CONNEXIN 26 / CONNEXIN 30 DEL / DUP HEARING LOSS PANEL Number of markers: 2 BRANCHIO-OTO-RENAL SYNDROME PANEL Number of markers: 3 USHER SYNDROME PANEL Number of markers: 11 ZELLWEGER SYNDROME PANEL Number of markers: 9	GJB2, GJB6 EYA1, SIX1, SIX5 ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7
COMPREHENSIVE VISION LOSS NGS PANEL Number of markers: 250	ABCA4, ABHD12, ADAM9, ADGRV1, AGK, AH1, AIPL1, ALMS1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BLOC1S6, BMP4, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1D, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLN3, CLN5, CLN6, CLRN1, CNGA1, CNGB3, CNM1A, CNM1B, COCH, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP11B1, CYP4V2, DHDDS, DNJAC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, EYA1, EYS, FAM161A, FLVCR1, FOXC1, FOXC2, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FYCO1, FZD4, GCNT2, GJA8, GNA11, GNA12, GNPTG, GPR143, GPR179, GRP1, GRK1, GRM6, GUC1A1, GUC1A1B, GUCY2D, HARS, HCCS, HESX1, HPS1, HPS2, HPS4, HPS5, HPS6, HSF4, IFT140, IMPDH1, IMPG2, INPPE5, IQCB1, JAG1, KARS, KCNJ13, KCNV2, KIF11, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP5, LYST, LZTFL1, MAK, MERTK, MFRP, MFSDB, MITF, MKKS, MKS1, MTPP, MYH9, MYO7A, MYOC, NDP, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, PANK2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PGK1, PHYH, PITPNM3, PITX2, PITX3, PLA2G5, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PTPRO, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI1, RPI1L, RP2, RPE65, RPRG, RPRG1P1, RPRG1P1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SMOG1, SNRNP200, SOX10, SOX2, SPATA7, STRA6, TCTN1, TCTN2, TCTN3, TDRD7, TGFBI, TIMM8A, TIMP3, TMC1, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, WDR19, WHRN, ZNF423
ALBINISM / HERMANSKY-PUDLAK / WAARDENBURG PANEL Number of markers: 18 DEVELOPMENTAL EYE PANEL Number of markers: 21	AP3B1, BLOC1S6, CACNA1F, EDN3, EDNRB, GPR143, HPS1, HPS2, HPS4, HPS5, HPS6, MITF, OCA2, PAX3, SLC45A2, SOX10, TYR, TYRP1 BCOR, BMP4, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GRP1, HCCS, KIF11, MFRP, NDP, OTX2, PAX6, PITX3, PLA2G5, PXDN, SMOG1, SOX2, STRA6, VSX2
RETINAL DISEASE PANEL Number of markers: 154	ABCA4, ABHD12, ADAM9, ADGRV1, AH1, AIPL1, ALMS1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C2ORF71, C5ORF42, C8ORF37, CA4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLRN1, CNGA1, CNGB3, CNM1A, CNM1B, COCH, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GNA12, GNPTG, GPR179, GRK1, GRM6, GUC1A1, GUC1A1B, GUCY2D, HARS, IFT140, IMPDH1, IMPG2, INPPE5, IQCB1, JAG1, KARS, KCNJ13, KCNV2, KIF7, KLHL7, LCA5, LRAT, LRIT3, LZTFL1, MAK, MERTK, MFRP, MKKS, MKS1, MYO7A, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, OFD1, OPA1, OPN1SW, PANK2, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PHYH, PITPNM3, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PTPRO, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI1, RPI1L, RP2, RPE65, RPRG, RPRG1P1, RPRG1P1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SMOG1, SNRNP200, SOX10, SOX2, SPATA7, STRA6, TCTN1, TCTN2, TCTN3, TDRD7, TGFBI, TIMM8A, TIMP3, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TTC21B, TTC8, TULP1, USH1C, USH1G, USH2A, WDR19, WHRN, ZNF423
STICKLER AND CATARACT PANEL Number of markers: 41	ABHD12, AGK, BCOR, BEST1, CAPN5, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, ERCC6, EYA1, FYCO1, GCNT2, GJA8, GNPTG, HSF4, JAG1, KCNJ13, LRP5, MYH9, NDP, OAT, OPA3, OTX2, PAX6, PEX7, PHYH, PITX3, PXDN, RLBP1, TDRD7, VCAN, VSX2, WFS1 * Only copy number variants and targeted genotyping reported in these genes

PANEL TABLE (continued)

NEURODEVELOPMENTAL	Genes
	ABAT, ABCD1, ACSL4, ACY1, ADGRG1, ADGRV1, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ALDH7A1, ALG13, AMT, ANK3, ANKRD11, AP1S2, AP4B1, APAE1, AP4M1, AP4S1, ARFGEF2, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATP7A, ATP8A2, ATR, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, C4B, CACNA1A, CACNA1C, CACNA1E, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D1A, CCDC22, CCDC38C, CCM2, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTCF, CTNBB1, CTSD, CUL3, CUL4B, CYP27A1, D2HGDH, DCX, DDHD2, DDX3X, DEAF1, DEPCDC5, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNAJC5, DNMI1, DNMT3A, DOCK7, DPYD, DYNC1H1, DYRK1A, EBP, EEF1A2, EHM1, EHM2, EIF2S3, ELPA, EMB2, EPMA2, EZH2, FGD1, FGF3, FKBP, FLNA, FLVCR2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GATN2, GATN3, GCSH, GDI1, GK, GLDC, GLI2, GLI3, GNAO1, GNB1, GNS, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HCCS, HCN1, HCN4, HDAC8, HGSNAT, HIP1, HNRNP1U, HPT1, HRAS, HSD17B10, HTRA1, HUWE1, IDS, IL1RAPL1, IQSEC2, ITPA, KANSL1, KAT6A, KAT6B, KCNMA1, KCNMA2, KCNBI1, KCNBJ1, KCNJI1, KCNMA1, KCNQA2, KCNQ3, KCN1T1, KCTD13, KCTD7, KDM5C, KDM6A, KIF1A, KIF1BP, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LAS1L, LBR, LGI1, LIAS, LINS1, LRP2, MAGEL2, MAGI2, MAN1B1, MAOA, MAP2K1, MAP2K2, MAPK10, MBD5, MBTPS2, MCHP1, MECP2, MED12, MED13L, MED23, MECP2, MFS08, MID1, MTHFR, MTOR, MYT1L, NAA10, NAGLU, NDE1, NDP, NDUFA1, NECAP1, NEXMIF, NFX, NFX, NLRCL1, NLS, NIPBL, NLGN4X, NOTCH3, NPRL2, NPRL3, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCR1, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PCDH10, PDHA1, PGK1, PHF6, PHF8, PIGA, PIGB, PIGL, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POGZ, POLG, POMGNT1, POMT1, POMT2, PORCN, PPT1, PQBP1, PRICK1E1, PRICKLE2, PRPS1, PRRT2, PTCH1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAF1, RAI1, RARS2, RBM10, RELN, RIT1, RNASEH2A, RNASEH2B, RNASEH2C, ROGD1, RPL10, RPS6KA3, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINC1, SETBP1, SETD2, SETD5, SGGSH, SHANK2, SHANK3, SHOC2, SIK1, SLC12A5, SLC13A5, SLC16A2, SLC19A3, SLC1A2, SLC25A1, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A4, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMS, SNAP25, SNAP29, SOS1, SOX11, SOX5, SPRED1, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TAF1, TBC1D24, TBL1XR1, TBR1, TBX1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TPP1, TRAPPC9, TREX1, TRIO, TRPM6, TSC1, TSC2, TSPAN7, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUSC3, UBE2A, UBE3A, UNC80, UPF3B, USP9X, VANGL1, VPS13A, VPS13B, WAC, WDR45, WDR62, WDR81, WWOX, YWHAE, ZC4H2, ZDHHC9, ZEB2, ZMYND11, ZNF407, ZNF711
COMPREHENSIVE EPILEPSY AND AUTISM PANEL Number of markers: 401	
	ABAT, ADSL, ADGRG1, ADGRV1, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP1S2, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, BCKDK, CACNA1A, CACNA1E, CACNA2D2, CACNB4, CASK, CASR, CCDC38C, CCM2, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTCF, DCK7, DYRK1A, EEF1A2, EHF1C, EHM1, EHM2, EPMA2, FGD1, FGF3, FKBP, FLNA, FLVCR2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GLI2, GNAO1, GNB1, GOSR2, GPC3, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, HCN1, HCN4, HIP1, HNRNP1U, HSD17B10, HTRA1, IQSEC2, ITPA, KANSL1, KCNMA1, KCNQA2, KCNQ3, KCN1T1, KCTD13, KCTD7, KDM5C, KDM6A, KIF1A, KIF1BP, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LBR, LGI1, LIAS, MAGI2, MAPK10, MBD5, MCHP1, MECP2, MECP2, MFS08, MTHFR, MTOR, NDE1, NDUFA1, NECAP1, NLRCL1, NIPBL, NOTCH3, NPRL2, NPRL3, NRXN1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PIGA, PIGB, PIGL, PIGV, PLA2G6, PLCP1, PLP1, PNKP, PNPO, POGZ, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICK1E1, PRICKLE2, PRRT2, PTEN, RAB39B, RAI1, RARS2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGD1, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCTN5A, SCN8A, SCN9A, SERPINC1, SETBP1, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SNAP29, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TBC1D24, TBX1, TCF4, TPP1, TREX1, TRPM6, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, UBE3A, VANGL1, VPS13A, VPS13B, WDR62, WWOX, ZEB2
COMPREHENSIVE EPILEPSY PANEL Number of markers: 226	
	ABAT, ADSL, ADGRG1, ADGRV1, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP1S2, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, BCKDK, CACNA1A, CACNA1E, CACNA2D2, CACNB4, CASK, CASR, CCDC38C, CCM2, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTCF, DCK7, DYRK1A, EEF1A2, EHF1C, EHM1, EHM2, EPMA2, FGD1, FGF3, FKBP, FLNA, FLVCR2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GLI2, GNAO1, GNB1, GOSR2, GPC3, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, HCN1, HCN4, HIP1, HNRNP1U, HSD17B10, HTRA1, IQSEC2, ITPA, KANSL1, KCNMA1, KCNQA2, KCNQ3, KCN1T1, KCTD13, KCTD7, KDM5C, KDM6A, KIF1A, KIF1BP, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LBR, LGI1, LIAS, MAGI2, MAPK10, MBD5, MCHP1, MECP2, MECP2, MFS08, MTHFR, MTOR, NDE1, NDUFA1, NECAP1, NLRCL1, NIPBL, NOTCH3, NPRL2, NPRL3, NRXN1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PIGA, PIGB, PIGL, PIGV, PLA2G6, PLCP1, PLP1, PNKP, PNPO, POGZ, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICK1E1, PRICKLE2, PRRT2, PTEN, RAB39B, RAI1, RARS2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGD1, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCTN5A, SCN8A, SCN9A, SERPINC1, SETBP1, SHANK3, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SNAP29, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TBC1D24, TBX1, TCF4, TPP1, TREX1, TRPM6, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, UBE3A, VANGL1, VPS13A, VPS13B, WDR62, WWOX, ZEB2
FOCAL, GENERALIZED, AND MYOCLONIC EPILEPSY PANEL Number of markers: 52	
	ACY1, ADGRV1, ALDH5A1, ALDH7A1, BCKDK, CACNA1A, CACNA2D2, CACNB4, CASR, CCM2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CNTNAP2, CPA6, CSTB, DEPCDC5, EHF1C, EPMA2, FGD1, FGF3, FKBP, FLNA, FLVCR2, FMR1, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GNAO1, GNB1, GPHN, GRIN2B, HCN1, HIP1, ITPA, KCNQA2, KCNBI1, KCNBJ1, KCNJI1, LIAS, MAGI2, MAPK10, MECP2, MTHFR, NECAP1, PCDH19, PIGA, PIGB, PLCB1, PNKP, PNPO, RARS2, RNASEH2A, ROGD1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC35A2, SLC6A8, SPTAN1, ST3GAL3, STX1B, STXBP1, SZT2, TBC1D24, WWOX
INFANTILE EPILEPSY PANEL Number of markers: 58	
	ABAT, ADSL, ALG13, AMT, ARHGEF9, ARX, CDKL5, CHD2, DNMI1, DCK7, DYRK1A, EEF1A2, GABRA1, GABRB2, GCSH, GLDC, GNAO1, GNB1, GPHN, GRIN2B, HCN1, HIP1, ITPA, KCNQA2, KCNBI1, KCNBJ1, KCNJI1, LIAS, MAGI2, MAPK10, MECP2, MTHFR, NECAP1, PCDH19, PIGA, PIGB, PLCB1, PNKP, PNPO, RARS2, RNASEH2A, ROGD1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC35A2, SLC6A8, SPTAN1, ST3GAL3, STX1B, STXBP1, SZT2, TBC1D24, WWOX
MIGRAINE PANEL Number of markers: 7	ATP1A2, CACNA1A, NOTCH3, POLG, PRRT2, SCN1A, SLC2A1
NEURONAL CEROID LIPOFUSCINOSES PANEL Number of markers: 9	CLN3, CLN5, CLN6, CLN8, CTSD, DNAJC5, MFS08, PPT1, TPP1
NEURONAL MIGRATION PANEL Number of markers: 22	ADGRG1, COL18A1, COL4A1, EMX2, FGF3, FKBP, FLNA, LARGE1, PAFAH1B1, PAX6, POMGNT1, POMT1, POMT2, PQBP1, RELN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1
SYNDROMIC EPILEPSY AND INTELLECTUAL DISABILITY PANEL Number of markers: 93	AP1S2, ARFGEF2, ASPM, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, CASK, CCDC38C, CNTNAP2, CREBBP, CUL4B, DCX, DYRK1A, EEF1A2, EHM1, FGD1, FLVCR2, FOXG1, GAMT, GATM, GLI2, GPC3, GRIA3, GRIN1, HCN4, HNRNP1U, HSD17B10, IQSEC2, KANSL1, KCNMA1, KCNQA2, KCNBJ1, KCNJI1, KCNMA1, KCNQA2, KCNQ3, KFN1BP, KMT2D, LBR, LGI1, MBD5, MCHP1, MECP2, MECP2, MFS08, MTHFR, MTOR, NDE1, NDUFA1, NECAP1, NLRCL1, NIPBL, NRXN1, OPHN1, PAK3, PANK2, PHF6, PIGV, PLA2G6, PLP1, POLG, PQBP1, RAB39B, RAI1, RNASEH2B, RNASEH2C, ROGD1, SAMHD1, SCN1A, SCN5A, SETBP1, SHANK3, SMC1A, SMC3, SMS, SNAP25, SNAP29, SPTAN1, ST3GAL3, SLC19A3, SLC25A19, SLC2A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SNAP29, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, TBX1, TCF4, TREX1, TSC1, TSC2, UBE3A, VANGL1, VPS13A, VPS13B, WDR62, ZEB2
COMPREHENSIVE AUTISM SPECTRUM DISORDER PANEL Number of markers: 228	ABCD1, ACSL4, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ANK3, ANKRD11, AP1S2, AP4B1, APAE1, AP4M1, AP4S1, ARFGEF2, ARHGEF9, ARX, ASPM, ATP13A2, ATP7A, ATP8A2, ATR, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, C4B, CACNA1A, CASK, CBL, CC2D1A, CCDC22, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CLCN4, CNTNAP2, CREBBP, CTCF, CTNBB1, CUL3, CYP27A1, D2HGDH, DDHD2, DDX3X, DEAF1, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNMT3A, DPYD, DYNC1H1, DYRK1A, EBP, EHM1, EIF2S3, ELPA, EZH2, HD01, FMRI, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GATA2B, GDI1, GK, GLI3, GNS, GPC3, GRIA3, GRIK2, GRIN2B, GRIP1, HCCS, HDAC8, HGSNAT, HPT1, HRAS, HUWE1, IDS, IL1RAPL1, KAT6A, KAT6B, KCTD13, KDM5C, KDM6A, KIF1A, KIRREL3, KMT2A, KRAS, L1CAM, L2HGDH, LAMC3, LAMP2, LAS1L, LINS1, LRP2, MAGEL2, MAN1B1, MAOA, MAP2K1, MAP2K2, MBD5, MBTPS2, MECP2, MED12, MED13L, MED23, MECP2, MID1, MYT1L, NAA10, NAGLU, NDP, NEXMIF, NFX, NFX, NIPBL, NLGN4X, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCR1, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDHA1, PGK1, PHF6, PHF8, PIGB, PIGL, PIGV, PLA2G6, PLCP1, PLP1, PNKP, PNPO, POGZ, PORCN, PQBP1, PRPS1, PTCH1, PTC1, PTC1A, PTC1B, PTC1D1, PTC1D2, PTC1D3, PTC1D4, PTC1D5, PTC1D6, PTC1D7, PTC1D8, PTC1D9, PTC1D10, PTC1D11, PTC1D12, PTC1D13, PTC1D14, PTC1D15, PTC1D16, PTC1D17, PTC1D18, PTC1D19, PTC1D20, PTC1D21, PTC1D22, PTC1D23, PTC1D24, PTC1D25, PTC1D26, PTC1D27, PTC1D28, PTC1D29, PTC1D30, PTC1D31, PTC1D32, PTC1D33, PTC1D34, PTC1D35, PTC1D36, PTC1D37, PTC1D38, PTC1D39, PTC1D40, PTC1D41, PTC1D42, PTC1D43, PTC1D44, PTC1D45, PTC1D46, PTC1D47, PTC1D48, PTC1D49, PTC1D50, PTC1D51, PTC1D52, PTC1D53, PTC1D54, PTC1D55, PTC1D56, PTC1D57, PTC1D58, PTC1D59, PTC1D60, PTC1D61, PTC1D62, PTC1D63, PTC1D64, PTC1D65, PTC1D66, PTC1D67, PTC1D68, PTC1D69, PTC1D70, PTC1D71, PTC1D72, PTC1D73, PTC1D74, PTC1D75, PTC1D76, PTC1D77, PTC1D78, PTC1D79, PTC1D80, PTC1D81, PTC1D82, PTC1D83, PTC1D84, PTC1D85, PTC1D86, PTC1D87, PTC1D88, PTC1D89, PTC1D90, PTC1D91, PTC1D92, PTC1D93, PTC1D94, PTC1D95, PTC1D96, PTC1D97, PTC1D98, PTC1D99, PTC1D100, PTC1D101, PTC1D102, PTC1D103, PTC1D104, PTC1D105, PTC1D106, PTC1D107, PTC1D108, PTC1D109, PTC1D110, PTC1D111, PTC1D112, PTC1D113, PTC1D114, PTC1D115, PTC1D116, PTC1D117, PTC1D118, PTC1D119, PTC1D120, PTC1D121, PTC1D122, PTC1D123, PTC1D124, PTC1D125, PTC1D126, PTC1D127, PTC1D128, PTC1D129, PTC1D130, PTC1D131, PTC1D132, PTC1D133, PTC1D134, PTC1D135, PTC1D136, PTC1D137, PTC1D138, PTC1D139, PTC1D140, PTC1D141, PTC1D142, PTC1D143, PTC1D144, PTC1D145, PTC1D146, PTC1D147, PTC1D148, PTC1D149, PTC1D150, PTC1D151, PTC1D152, PTC1D153, PTC1D154, PTC1D155, PTC1D156, PTC1D157, PTC1D158, PTC1D159, PTC1D160, PTC1D161, PTC1D162, PTC1D163, PTC1D164, PTC1D165, PTC1D166, PTC1D167, PTC1D168, PTC1D169, PTC1D170, PTC1D171, PTC1D172, PTC1D173, PTC1D174, PTC1D175, PTC1D176, PTC1D177, PTC1D178, PTC1D179, PTC1D180, PTC1D181, PTC1D182, PTC1D183, PTC1D184, PTC1D185, PTC1D186, PTC1D187, PTC1D188, PTC1D189, PTC1D190, PTC1D191, PTC1D192, PTC1D193, PTC1D194, PTC1D195, PTC1D196, PTC1D197, PTC1D198, PTC1D199, PTC1D200, PTC1D201, PTC1D202, PTC1D203, PTC1D204, PTC1D205, PTC1D206, PTC1D207, PTC1D208, PTC1D209, PTC1D210, PTC1D211, PTC1D212, PTC1D213, PTC1D214, PTC1D215, PTC1D216, PTC1D217, PTC1D218, PTC1D219, PTC1D220, PTC1D221, PTC1D222, PTC1D223, PTC1D224, PTC1D225, PTC1D226, PTC1D227, PTC1D228, PTC1D229, PTC1D230, PTC1D231, PTC1D232, PTC1D233, PTC1D234, PTC1D235, PTC1D236, PTC1D237, PTC1D238, PTC1D239, PTC1D240, PTC1D241, PTC1D242, PTC1D243, PTC1D244, PTC1D245, PTC1D246, PTC1D247, PTC1D248, PTC1D249, PTC1D250, PTC1D251, PTC1D252, PTC1D253, PTC1D254, PTC1D255, PTC1D256, PTC1D257, PTC1D258, PTC1D259, PTC1D260, PTC1D261, PTC1D262, PTC1D263, PTC1D264, PTC1D265, PTC1D266, PTC1D267, PTC1D268, PTC1D269, PTC1D270, PTC1D271, PTC1D272, PTC1D273, PTC1D274, PTC1D275, PTC1D276, PTC1D277, PTC1D278, PTC1D279, PTC1D280, PTC1D281, PTC1D282, PTC1D283, PTC1D284, PTC1D285, PTC1D286, PTC1D287, PTC1D288, PTC1D289, 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PANEL TABLE (continued)

METABOLIC DISORDERS SINGLE GENE DIAGNOSTIC TESTING	Genes
AMINOACIDOPATHIES AND UREA CYCLE DISORDERS	ACAT1, ADSL, ALDH7A1, AMT, ARG1, ASL, ASPA, ASS1, BCKDHA, BCKDHB, BTD, CBS, CPS1, DBT, DLD, DPYD, FAH, GABRG2, GCDH, GCH1, GCSH, GLDC, HMGCL, HPD, IVD, MAT1A, MMACHC, MTHFR, MUT, NAGS, OTC, PAH, PCBD1, PCCA, PCCB, PHGDH, PTS, QDPR, SLC25A13, SPR, TAT
CHOLESTEROL	CYP27A1, DHCR7, EBP
DISORDERS OF CARBOHYDRATE METABOLISM	AGL, ALDOB, FBP1, G6PC, GALE, GALK1, GALT, GBE1, GLUD1, GYS2, LAMP2, NHLRC1, PFKM, PHKB, PYGL, PYGM, SLC37A4
FATTY ACID OXIDATION DISORDERS	ABAT, ACAD9, ACADM, ACADVL, ACAT1, CPS1, CPT1A, CPT2, ETF, ETFB, ETFDH, ETHE1, HADH, HADHA, HADHB, HMGCL, IVD, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, SLC22A5, SLC25A20
LYSOSOMAL STORAGE DISORDERS AND OTHER DISORDERS	AGA, ARSA, ARSB, BTD, CLN3, CLN5, CLN6, CLN8, CTNS, CTSD, CTSK, DNAJC5, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GBA, GLA, GLB1, GNE, GNPTAB, GNPTG, GNS, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LIPA, MAN2B1, MCOLN1, MFS08, NAGLU, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMP1, TPP1
ORGANIC ACIDEMIAS	ACAT1, AGK, BCKDHA, BCKDHB, BTD, GCDH, HLCS, HMGCL, HSD17B10, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTRR, MUT, OPA3, PCCA, PCCB, SLC19A3, TAZ
PEROXISOMAL STORAGE DISEASES	ABCD1, ACOX1, AGPS, AGXT, HSD17B4, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PH1H
ADDITIONAL GENES AVAILABLE FOR DIAGNOSTIC TESTING	Genes ABCB11, ABCC8, ACSF3, ADAMTS2, AIRE, AKR1D1, ALDH3A2, ALG6, ALPL, ANK1, APOL**, AQP2, ASNS, ATP6V1B1, ATP7B, AVPR2, BCS1L, BLM, BSND, CAPN3, CD3D, CD3E, CHIT1**, CHRNE, CIITA, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, CPLANE1, CYBA, CYBB, CYP11B1, CYP11B2, CYP17A1, CYP19A1, DCLRE1C, DNAH5, DNAI1, DNAI2, DUOX2, DUOX2, DYSF, EDA, EFN1, EIF2B5, ELP1, EMD, EPB42, ESCO2, EVC, FANCA, FANCC, FANCG, FBNI, GFM1, GJB1, GLE1, GP1BA, GP9, GRHR, GSS, HAX1, HJV, HMGCS2, HOGA1, HSD3B2, HSD3B7, HYL1, IGF1, IL2RG, IL7R, IVD, JAK3, LAMA3, LAMB3, LAMC2, LDLRAP1, LHX3, LIFR, LPL, LRPPRC, MEFV, MESP2, MLC1, MPI, MPL, MPV17, MTM1, MTR, NDRG1, NDUFAF5, NDUFS6, NEB, NPHS1, NPHS2, NTRK1, PAX8, PCARE, PDHB, PMM2, POU1F1, PROP1, PTRC, PUS1, RAG1, RAG2, RAPSN, RMRP, RTEL1, SACS, SEPS2, SGCA, SGC8, SGC9, SLC12A3, SLC12A6, SLC25A15, SLC26A2, SLC35A3, SLC394A, SLC4A11, SLC5A5, SLC7A7, SMARCAL1, SMN1, STAR, TCIRG1, TECPR2, TFR2, TGM1, THRA, TPO, TRHR, TRMU, TSFM, TSHB, TSHR, TTPA, TYMP, UGT1A1, VPS45, VRK1, WNT10A, WT1 ** Only targeted genotyping reported in these genes

GENES AVAILABLE ON DIAGNOSTIC TEST PANELS

A2ML1	BBS1	CEP63	D2HGDH	FKRP	GY2	KCN5A	MED17	NOTCH3	POGZ	RTEL1	SOS1	TRIO
ABAT	BBS10	CERKL	DBT	FKTN	HADH	KCNB1	MED23	NP1	POLE	RVR2	SOS2	TRIOBP
ABC44	BBS12	CFB	DCLRE1B	FLNA	HADHA	KCND3	ME2C	NP2	POLG	SACS	SOX10	TRMU
ABC811	BBS2	CFD	DCLRE1C	FLNC	HADHB	KCNE1	MEFV	NPH1	POMGNT1	SAG	SOX11	TRNT1
ABCC8	BBS4	CFH	DCX	FLVCR1	HAND1	KCNE2	MEIS2	NPH3	POMT1	SALL1	SOX2	TRPM1
ABCC9	BBS5	CFHR1	DHD2	FLVCR2	HARS	KCNE3	MERTK	NPH4	POMT2	SALL2	SOX5	TRPM4
ABCD1	BBS7	CFHR3	DDX3X	FMR1	HAX1	KCNE5	MESP2	NPH5	POR	SAMHD1	SP110	TRPM6
ABHD12	BBS9	CFI	DEAF1	FOLR1	HBA1	KCNH2	MFAP5	NPHS2	PORCN	SATB2	SPATA7	TSC1
ACAD9	BCKDHA	CFP	DEPDC5	FOXC1	HBA2	KCNJ1	MFRP	NPPA	POU1F1	SBD5	SPNK5	TSC2
ACADM	BCKDHB	CFTR	DES	FOXE3	HBB	KCNJ10	MFSDB	NPR2	POU3F4	SCARB2	SPN	TSFM
ACADVL	BCKDK	CHD2	DGKE	FOXG1	HCCS	KCNJ11	MI1	NPR3	POU4F3	SCN10A	SPRED1	TSBH
ACAT1	BCL11A	CHD7	DHCR7	FOXH1	HCN1	KCNJ13	MID1	NR2E3	PPOX	SCN1A	SPTAN1	TSHR
ACD	BCOR	CHD8	DHDDS	FOXN1	HCN4	KCNJ2	MITF	NR2F2	PPP1CB	SCN1B	ST3GAL3	TSPAN12
ACOX1	BCSL	CHIT1	DIABLO	FOXP1	HDAC8	KCNJ5	MKKS	NRAS	PPT1	SCN2A	ST3GAL5	TSPAN7
ACPF5	BEST1	CHM	DIAPH1	FOXP2	HESX1	KCNJ8	MKS1	NRL	PQB1	SCN2B	STAMPB	TSPEAR
ACSF3	BLM	CHRM2	DIS3L2	FOXP3	HEXA	KCNK3	MCL1	NRXN1	PRCD	SCN3	STAR	TT21B
ACSL4	BLOC1S6	CHRNA2	DISP1	FRAS1	HEXB	KCNMA1	MLYCD	NRXN3	PRDM16	SCN3B	STAT1	TT37
ACTA2	BMP4	CHRNA4	DKC1	FREM1	HGF	KCNQ1	MMAA	NSD1	PRF1	SCN4B	STAT3	TTCA7
ACTB	BMPR1B	CHRNA7	DLG	FREM2	HGSNAT	KCNQ2	MMAB	NSDHL	PRICKLE1	SCN5A	STAT5B	TTCS
ACTC1	BMPR2	CHRN2	DLG3	FRMD7	HIP1	KCNQ3	KCNQ3	NSUN2	PRICKLE2	SCN6	STIL	TTN
ACTG1	BRAF	CHRNAE	DLL1	FSCN2	HJV	KCNQ4	KCNQ4	MMADHC	PRKAG2	SCN9A	STIM1	TPA
ACTN2	BRWD3	CHST14	DMD	FTSJ1	HLCS	KCN7	KCN7	MPI	PRKCD	SCD1	STK4	TTR
ACVR2B	BSND	CIB2	DNAH5	FYCO1	HMB5	KCNV2	KCNV2	MPL	PRKDC	SDCCAG8	STR4	TUBA1A
ACVRL1	BTB	CITTA	DNAI1	FZD4	HMGCL	KCTD13	MPV17	OBSCN	PRKG1	SDHA	STRC	TUBA8
ACY1	BTX	CLCN2	DNAI2	G6PC	HMGCS2	KDM5C	MRE11	OCA2	PROM1	SDHAF1	STX11	TUBB
ADA	C12ORF57	CLCN4	DNAJC5	G6PC3	HNRNP1	KDM6A	MSRB3	OCLR	PROP1	SEMA4A	STX1B	TUBB2A
ADA2	C1QA	CLDN14	DNM1	G6PD	HOGA1	KIF1A	HOGA1	OFD1	PRPF3	SEPS6CS	STXB1	TUBB2B
ADAM17	C1QB	CLN3	DNMT3A	GAA	HOXD13	KIF11	KIF11	OPA1	PRPF31	SERPIN6	STXB2	TUBB3
ADAM9	C1QC	CLN5	DNMT3B	GABRA1	HPD	KIF1A	MTHFR	OPA3	PRPF6	SERPING1	SUBF1	TUBG1
ADAMTS2	C1S	CLN6	DOCK2	GABRB2	HPRT1	KIF1BP	MTM1	OPN1	PRPF8	SERPIN1	SURF1	TULP1
ADAR	C2	CLN8	DOCK7	GABRB3	HPS1	KIF2A	MT01	OPN1SW	PRPH2	SETBP1	SYN1	TUSC3
ADGRG1	C3	CLPB	DOCK8	GABRD	HPS3	KIF7	MTOR	OPRM1	PRPS1	SETD2	SYNGAP1	TWIST1
ADGRV1	C4A	CLRN1	DOLK	GABRG2	HPS4	KIRREL3	MTR	ORAI1	PRR2	SETD5	SYP	TXNRD2
ADNP	C4B	CNGA1	DPYD	GALC	HPS5	KLHL7	MTRR	OTC	PSAP	SGCA	SZT2	TYK2
ADSL	C5	CNGA3	DSC2	GALF	HPS6	KMT2A	MTPP	OTOA	PTCH1	SGCB	TAF1	TYMP
AF2	C6	CNGB1	DSG2	GALK1	HRAS	KMT2D	MUT	OTOF	PTCHD1	SGCD	TAP1	TYR
AGA	C7	CNGB3	DSP	GALNS	HSD17B10	KNL1	MVK	OTOG	PTEN	SGCG	TAP2	TYRP1
AGK	C8A	CN11M4	DTNA	GALT	HSD17B4	KRAS	MYBPC3	OTGL	PTPN11	SGSH	TAPBP	UBE2A
AGL	C8B	CNTNAP2	DUOX2	GAMT	HSD3B2	KRIT1	MYD88	OTX2	PTPRC	SH2D1A	TAT	UBE3A
AGO1	C8ORF37	COCH	DUOX2	GAS1	HSD3B7	LYN1	MYH11	P2RX2	PTRP7	SHANK2	TAZ	UBIAD1
AGPS	C9	COL11A1	DYNC1H1	GATA2	HSF4	L2HGDH	MYH14	PAC51	PTS	SHANK3	TBC1D24	UGT1A1
AGXT	CA4	COL11A2	DYRK1A	GATA3	HTR2A	LAMA3	MYH6	PAFAH1B1	PURA	SHH	TLX1XR1	UGT2B15
AHI1	CA8	COL18A1	DYF	GATA4	HTRA1	LAMA4	MYH7	PAH	PUS1	SHOC2	TBR1	UNC13D
AICDA	CABP4	COL1A1	EBP	GATA6	HUWE1	LAMB3	MYH9	PAK3	PXDN	SIK1	TBX1	UNC80
AIFM1	CACNA1A	COL1A2	EDA	GATAD1	HYAL1	LAMB2	MYL2	PANX2	PYGL	SIX1	TBX5	UNG
AIP1	CACNA1C	COL27A1	EDN3	GATAD2B	HYLS1	LAMB3	MYL3	PAK3	PYGM	SIX3	TCAP	UPF3B
AIRE	CACNA1D	COL2A1	EDN3	GATM	ICD5	LAMP2	MYLK	PAX6	QDPR	SIX5	TCF12	UROD
AK2	CACNA1F	COL3A1	EEF1A2	GBA	IDS	LAMTOR2	MYLK2	PAX8	RAB18	SKI	TCF20	UROS
AKAP9	CACNA1H	COL4A1	EFEMP1	GBE1	IDUA	LARGE1	MYO15A	PCARE	RAB23	SKIV2L	TCF4	USB1
AKR1D1	CACNA2D1	COL4A3	EFEMP2	GCHD	IFIH1	LAS1L	MYO3A	PCBD1	RAB27A	SLC12A3	TCR61	USH1C
ALAS2	CACNA2D2	COL4A4	EFHC1	GCH1	IFNGR1	LBR	MYO5B	PCCB	RAB28	SLC12A5	TGN2	USH1G
ALDH3A2	CACNA2D4	COL4A5	EFNB1	GCH2	IFNGR2	LC45	MYO6	PCCB	RAB39B	SLC12A6	TC1N1	USH2A
ALDH5A1	CACNB2	COL5A1	EHMT1	GCSH	IFT140	LCK	MYO7A	PCDH15	RAB39A	SLC13A5	TC1N2	USP9X
ALDH7A1	CACNB4	COL5A2	EIF2AK4	GDF1	IGHM	LDB3	MYOC	PCDH19	RAB39A2	SLC16A2	TC1N3	VANGL1
ALDOB	CALM1	COL7A1	EIF2B5	GDF2	IGLL1	LDLR	MYO10	PCNT	RAC2	SLC17A5	TDGF1	VCAN
ALG1	CALM2	COL9A1	EIF2S3	GDI1	IGSF1	LDLRAP1	MYO22	PCSK9	RAD21	SLC19A3	TDRD7	VCL
ALG2	CALM3	COL9A2	ELAC2	GF1	IKBK1	LEFTY2	MYN	PCD10	RAF1	SLC1A2	TECP2	VKORC1
ALG3	CAPN3	CAPN3	ELANE	GF1	IKBK1	LEFTY2	MYN	PDE6A	RAG1	SLC22A5	TECTA	VDLDR
ALG6	CAPN5	COPB	ELN	GIPC3	IL10	LHFPL5	NAA10	PDE6B	RAG2	SLC24A1	TERC	VPS13A
ALMS1	CARD11	CORO1A	ELOVL4	GJA1	IL10RA	LHX3	NAAGL	PDE6C	RAI1	SLC25A1	TERT	VPS13B
ALPK3	CARD14	COX10	ELP1	GJA5	IL10RB	LIAS	NAGS	PDE6G	RANRFP	SLC25A13	TFR2	VPS45
ALPL	CARD9	COX15	ELP4	GJA8	IL12B	LIFR	NBN	PDE6H	RAPSN	SLC25A15	TG	VRK1
AMT	CASK	CPA6	EMD	GJB1	IL12RB1	LIG1	NCF1	PDHA1	RARS2	SLC25A19	TGFB2	VSX2
ANK1	CASP10	CPPLANE1	EMX2	GJB2	IL17F	LIG4	NCF2	PDHB	RASA1	SLC25A20	TGFB3	WAC
ANK2	CASP8	CPOX	ENG	GJB6	IL17RA	LIMS1	NCF4	PDLIM3	RASA2	SLC25A22	TGFB1	WAS
ANK3	CASQ2	CP51	EP300	GK	IL17RC	LIPA	NDE1	PET100	RAX2	SLC26A2	TGFB2	WDR19
ANKRD1	CASR	CPT1A	EPB42	GLA	IL18	LMBR1	NDP	PEX1	RB1	SLC26A4	TGFB2	WDR19
ANKRD11	CAV1	CPT2	EPCAM	GLB1	IL18RAP	LMBRD1	NDRG1	PEX10	RBBP8	SLC2A1	TGFB1	WDR62
AP1S2	CAV3	CR2	EPG5	GLDC	IL1RAPL1	LMBNA	NDUFA1	PEX14	RBCY1	SLC2A10	TGM1	WDR81
AP3B1	CAVIN4	CRB1	EPM2A	GLE1	IL1RN	LOXHD1	NDUFA10	PEX16	RBM10	SLC35A2	TH	WFS1
AP4B1	CREBBP	CBL	ERCC6	GLI2	IL21	LPIN2	NDUFA12	PEX19	RBM20	SLC35A3	THOC2	WHRN
AP4E1	CBS	CRELD1	ERCC8	GLI3	IL21R	LPL	NDUFA2	PEX2	RBP3	SLC35C1	THRA	WIP1
AP4M1	CC2D1A	CRX	ESCO2	GLUD1	IL23R	LRAT	NDUFA9	PEX5	RBPA	SLC37A4	TIMM8A	WNT10A
AP4S1	CC2D2A	GRYAA	ESPN	GNAO1	IL2RA	LRBA	NDUFA2	PEX6	RD3	SLC39A13	TIMP3	WNT7A
APOB	CCDC22	CRYAB	ESRRB	GNA11	IL2RG	LRT3	NDUFA5	PEX7	RDH12	SLC39A4	TIMP2	WRAP53
APOL1	CCDC39	CRYBB1	ETFA	GNA12	IL36RN	LRP2	NDUFA6	PFKM	RDH5	SLC45A2	TLR3	WT1
AQP2	CCDC40	CRYBB3	ETFB	GNB1	IL7R	LRP5	NDUFS3	PGK1	RDX	SLC4A1	TMC1	WWOX
ARFGF2	CCDC50	CSF2RA	ETFDH	GNE	ILDR1	LRRPCC	NDUFS4	PGM1	RELN	SLC4A10	TMEM126A	XIAP
ARG1	CCDC38C	CSF3R	ETHE1	GNPTAB	ILK	LRRCSA	NDUFS6	PGM3	RET	SLC4A11	TMEM1173	YWHAE
ARHGFE9	CCM2	CSRP3	EVC	GNPTG	IMPDH1	LRTOMT	NDUFS7	PHF6	RFK5	SLC5A5	TMEM216	ZAP70
ARID1A	CD19	CSTB	EYA1	GNS	IMP2	LYST	NDUFS8	PHF9	RFKANK	SLC6A1	TMEM231	ZBTB24
ARID1B	CD247	CTC1	EYA4	GOSR2	INO80	LZTF1	NEB	PHGDH	RFKAP	SLC6A4	TMEM237	ZC4H2
ARL13B	CD27	CTCF	EYS	GP1BA	INPP5E	LZTR1	NEBL	PHK6	RGR	SLC6A8	TMEM43	ZDHHC9
ARL6	CD3D	CTLA4	EZH2	GP9	INS	MAGEL2	NECAP1	PHYH	RG59	SLC7A7	TMEM67	ZEB2
ARSA	CD3E	CTNNA3	F11	GPC3	IQCB1	MAG1	NEXMIF	PIGA	RG59BP	SLC9A3	TMEM70	ZFPM2
ARSB	CD3G	CTN1B1	F9	GPD1L	IQSEC2	MAGT1	NEXN	PIGN	RHO	SLC9A6	TMIE	ZIC2
ARX	CD40	CTNS	FADD	GPIN	IRAK4	MAK	NF1	PIGO	RHOH	SLC01B1	TMLHE	ZIC3
ASL	CD40LG	CTPS1	FAH	GPR143	IRF7	MAL71	NFAT5	PIGV	RIMS1	SLMAP	TMPO	ZMYND11
ASNS	CD46	CTSC	FAM161A	GPR179	IRF8	MAN1B1	NFIX	PIK3CD	RIT1	SMAD3	TMPRSS3	ZNF407
ASPA	CD59	CTSD	FANCA	IRGM	IRGM	MAN2B1	NFKB1	PIK3R1	RLBP1	SMAD4	TNFAIP3	ZNF423
ASPM	CD79A	CTSK	FANCC	IRG1	IRG1	MAOA	NFKB2	PITPNM3	RMRP	SMAD6	TNFRSF13B	ZNF711
ASS1	CD79B	CUL3	FANCG	IRHPR	ITGAM	MAP2K1	NFKBIA	PITX2	RNASEH2A	SMAD9	TNFRSF13C	ZRS
ATM	CD81	CUL4B	FAS	IRIA3	ITGB2	MAP2K2	NHEJ1	PITX3	RNASEH2B	SMARCA2	TNFRSF1A	
ATP13A2	CD8A	CXCR4	FASLG	GRIK2	ITK	MAP3K14	NHLRC1	PJVK	RNASEH2C	SMARCA4	TNFSF12	
ATP1A2	CDH23	CYBA	FBN1	GRIK4	ITPA	MAPK10	HNP2	PKHD1	RNU4ATAC	SMARCC1	TNNC1	
ATP2A2	CDH3	CYBB	FBN2	GRIN1	IVD	MARVELD2	NHS	PKP2	ROGD1	SMARCB1	TNNS3	
ATP6AP2	CDHR1	CYP11B1	FBN3	GRIN2A	IVD	MASP2	NIN	PLA2G5	ROR2	SMARCE1	TNNT2	
ATP6V0A2	CDK5RAP2	CYP11B2	FCN1	GRIN2B	JAG1	MAT1A	NPBL	PLA2G6	ROR2	SMC1A	TOPORS	
ATP6V1B1	CDKL5	CYP17A1	FECH	GRIP1	JAGN1	MAT2A	NKX2-5	PLC81	RORC	SMC3	TPM1	
ATP7A	CDKN1C	CYP19A1	FERMT2	GRK1	JAK1	MBD5	NKX2-6	PLC62	RP1	SMN1	TPMT	
ATP7B	CDON	CYP11B1	FERMT3	GRM6	JAK3	MBTPS2	NLGN4X	PLEKHM2	RP1L1	SMN2	TPO	
ATP8A2	CEACAM16	CYP21A2	FGD1	GRXCR1	JPH2	MCCC1	NLRCA	PLN	RP2	SMOC1	TPP1	
ATR	CEBPE	CYP27A1	FGF8	GSDME	JUP	MCCC2	NLRP2	PLD1	RPE65	SMPD1	TPRN	
ATRIP	CENPJ	CYP2B6	FGFR1	GSS	KANSL1	MCCE	NLRP3	PLP1	RPRG	SMPX	TRAPPC9	
ATRX	CEP135	CYP22B19	FGFR2	GUCY1A	KARS	MCOLN1	NMNAT1	PLM2	RPGRP1	SMS	TRDN	
AUTS2	CEP152	CYP2C9	FGFR3	GUCY1B	KAT6A	MCPH1	NO2D	PMS2	RPGRP1L	SNAP25	TREX1	
AVPR2	CEP164	CYP2D6	FGH	GUCY2C	KAT6B	MCCP2	NODAL	PNKP	RP10	SNAP29	TRHR	
B2M	CEP290	CYP3A5	FHL1	GUCY2D	KCNA1	MED12	NOP10	PNP	RPS6KA3	SNRNP200	TRIM22	
BAG3	CEP41	CYP4V2	FKBP14	GUSB	KCNA2	MED13L	NOTCH1	PNPO	RS1	SNTA1	TRIM32	