DIAGNOSTIC GENETIC TESTING REQUISITION

50000

SPECIMENS: 1428 Madison Ave., Rm AB2-25, New York, NY 10029
MAIL: One Gustave L. Levy Place, Box 1497, New York, NY 10029-6574
Phone: **800-298-6470** / Fax: 646-859-6870
Tax ID# 47-5349024/ CLIA# 33D2097541

ACCESSION NO.	DATE / /

PATIENT INFORMATION

PATIENT LAST NAME	PATIENT FIRST NAME	
DOB / /	BIOLOGICAL GENDER ☐M ☐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 / /	

BILLING INFORMATION ■Bill Clinic ■Bill Insurance Below ■Self Pay

POLICYHOLDER LAST NAME	POLICYHOLDER FIRST NAME	POLICYHOLDER DOB / /
INSURANCE CARRIER	INSURANCE ID	GROUP NO.

BILLING ADDRESS

OTHER HEALTH COVERAGE (IDENTIFY)

SELF-PAY: ☐ Credit Card ☐ Check

Make Checks Payable to: Mount Sinai Genomics Inc., P.O. Box 21312, New York, NY 10087-1312

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 / /

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

PATIENT CLINICAL STATUS
☐ Affected
☐ Unknown (no screening/evaluation)
☐ Unaffected (all screening/evaluations(s) normal)

PURPOSE OF STUDY
☐ Diagnostic ☐ Carrier Testing
☐ Research Study ☐ Clinical Study
☐ Familial Follow-up (family variant)_____
☐ Other_____

PLEASE COMPLETE ALL CLINICAL QUESTIONS ON THE BACK PAGES

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 (240 genes) includes subpanels listed below
  - ☐ COMPREHENSIVE ARRHYTHMIAS PANEL (54 genes) includes subpanels 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 (43 genes)
  - ☐ FAMILIAL HYPERCHOLESTEROLEMIA PANEL (4 genes)
  - ☐ HEREDITARY HEMORRHAGIC TELANGIECTASIA PANEL (5 genes)
  - ☐ METABOLIC CARDIOMYOPATHIES PANEL (24 genes)
  - ☐ NOONAN SPECTRUM DISORDERS PANEL (18 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 (10 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

PHENOTYPE

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

PEDIGREE

ICD-10 CODES

- ☐ 299.0 Autism, Current Infantile Or Childhood HP:0000717
- ☐ F84.0 Autistic Disorder HP:0000717
- ☐ G40 Epilepsy And Recurrent Seizures HP:0001250
- ☐ H54.7 (Unspecified Visual Loss) HP:0000505
- ☐ H90.5 (Unspecified Sensorineural Hearing Loss) HP:0000407
- ☐ Q02 (Microcephaly) HP:0000252
- ☐ 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 HP:0100543
  - ☐ Global developmental delay HP:0001263
  - ☐ Spontaneous abortion HP:0005268
  - ☐ Stillbirth HP:0003826
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

HISTORY OF CONSANGUINITY

- ☐ 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 HP:0003581
- ☐ Childhood onset HP:0011463
- ☐ Congenital onset HP:0003577
- ☐ Infantile onset HP:0003593
- ☐ Neonatal onset HP:0003623
- ☐ Young adult onset HP:0011462
- ☐ Other: _____

PERINATAL OR PRENATAL HISTORY

- ☐ Yes (Provide Details Below)
  - ☐ Hydrocephalus HP:0000238
  - ☐ Intrauterine growth retardation HP:0001511
  - ☐ Macrocephaly at birth HP:0004488
  - ☐ Oligohydramnios HP:0001562
  - ☐ Polyhydramnios HP:0001561
  - ☐ Preeclampsia HP:0100602
  - ☐ Premature birth HP:0001622
  - ☐ Seizures HP:0001250
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

OTHER FACTORS

- ☐ 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 HP:0011438
  - ☐ Metabolic Or Electrolyte Imbalance
  - ☐ Systemic Infection
  - ☐ Triggered by sleep deprivation HP:0025222
  - ☐ Triggered by stress HP:0025226
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

BEHAVIORAL FINDINGS

- ☐ Yes (Provide Details Below)
  - ☐ Abnormal aggressive, impulsive or violent behavior HP:0006919
  - ☐ Attention deficit hyperactivity disorder HP:0007018
  - ☐ Autism HP:0000717
  - ☐ Autistic behavior HP:0000729
  - ☐ Hyperactivity HP:0000752
  - ☐ Obsessive-compulsive behavior HP:0000722
  - ☐ Psychiatric Abnormalities
  - ☐ Psychosis HP:0000709
  - ☐ Short attention span HP:0000736
  - ☐ Specific learning disability HP:0001328
  - ☐ Stereotypy HP:0000733
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

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

- ☐ Yes (Provide Details Below)
  - ☐ Cerebellar atrophy HP:0001272
  - ☐ Cerebellar hypoplasia HP:0001321
  - ☐ Cerebral atrophy HP:0002059
  - ☐ Hypoplasia of the brainstem HP:0002365
  - ☐ Pachygyria HP:0001302
  - ☐ Ventriculomegaly HP:0002119
    - ☐ Abnormal corpus callosum morphology HP:0001273
    - ☐ Abnormal morphology of the cerebellar cortex HP:0031422
    - ☐ Abnormality of head blood vessel HP:3000036
    - ☐ Abnormality of the basal ganglia HP:0002134
    - ☐ Abnormality of the cerebral cortex HP:0002538
    - ☐ Agenesis of corpus callosum HP:0001274
    - ☐ Aplasia/Hypoplasia of the corpus callosum HP:0007370
    - ☐ Brain atrophy HP:0012444
    - ☐ Calcification of the small brain vessels HP:0002504
    - ☐ Cortical dysplasia HP:0002539
    - ☐ Frontotemporal cerebral atrophy HP:0006892
    - ☐ Hemimegalencephaly HP:0007206
    - ☐ Heterotopia HP:0002282
    - ☐ Holoprosencephaly HP:0001360
    - ☐ Hydrocephalus HP:0000238
    - ☐ Hypoplasia of the corpus callosum HP:0002079
    - ☐ Lissencephaly HP:0001339
    - ☐ Molar tooth sign on MRI HP:0002419
    - ☐ Polymicrogyria HP:0002126
    - ☐ Other: _____
- ☐ No
- ☐ Unknown

CARDIOVASCULAR

- ☐ Yes (Provide Details Below)
  - ☐ Abnormal morphology of the great vessels HP:0030962
  - ☐ Angioedema HP:0100665
  - ☐ Aortic aneurysm HP:0004942
  - ☐ Aortic root aneurysm HP:0002616
  - ☐ Aortic valve stenosis HP:0001650
  - ☐ Arrhythmia HP:0011675
  - ☐ Atrial cardiomyopathy HP:0200127
  - ☐ Atrial fibrillation HP:0005110
  - ☐ Atrial septal defect HP:0001631
  - ☐ Bradycardia HP:0001662
  - ☐ Cardiac arrest HP:0001695
  - ☐ Coarctation of aorta HP:0001680
  - ☐ Complete heart block with broad QRS complexes HP:0005170
  - ☐ Congenital malformation of the great arteries HP:0011603
  - ☐ Congestive heart failure HP:0001635
  - ☐ Dilated cardiomyopathy HP:0001644
  - ☐ Hypertension HP:0000822
  - ☐ Hypertrophic cardiomyopathy HP:0001639
  - ☐ Hypotension HP:0002615
  - ☐ Left ventricular hypertrophy HP:0001712
  - ☐ Left ventricular noncompaction HP:0030682
  - ☐ Lymphedema HP:0001004
  - ☐ Mitral regurgitation HP:0001653
  - ☐ Mitral valve prolapse HP:0001634
  - ☐ Myocardial infarction HP:0001658
  - ☐ Prolonged QT interval HP:0001657
  - ☐ Pulmonary arterial hypertension HP:0002092
  - ☐ Pulmonic stenosis HP:0001642
  - ☐ Shortened QT interval HP:0012232
  - ☐ Sudden cardiac death HP:0001645
  - ☐ Tetralogy of Fallot HP:0001636
  - ☐ Vasculitis HP:0002633

CARDIOVASCULAR (continued):

- ☐ Ventricular fibrillation HP:0001663
- ☐ Ventricular septal defect HP:0001629
- ☐ Ventricular tachycardia HP:0004756
- ☐ Other: _____
- ☐ No
- ☐ Unknown

CRANIOFACIAL DYSMORPHISM

- ☐ Yes (Provide Details Below)
  - **Head**
  - ☐ Craniosynostosis HP:0001363
  - ☐ Frontal bossing HP:0002007
  - ☐ Macrocephaly HP:0000256
  - ☐ Microcephaly HP:0000252
  - ☐ Sloping forehead HP:0000340
  - ☐ White forelock HP:0002211
  - **Face**
  - ☐ Coarse facial features HP:0000280
  - **Eyes**
  - ☐ Aniridia HP:0000526
  - ☐ Bilateral microphthalmos HP:0007633
  - ☐ Blepharospasm HP:0000643
  - ☐ Blue sclerae HP:0000592
  - ☐ Cataract HP:0000518
  - ☐ Coloboma HP:0000589
  - ☐ Downslanted palpebral fissures HP:0000494
  - ☐ Ectopia lentis HP:0001083
  - ☐ Epicanthus HP:0000286
  - ☐ Heterochromia HP:0001100
  - ☐ Hypertelorism HP:0000316
  - ☐ Lisch nodules HP:0009737
  - ☐ Microphthalmia HP:0000568
  - ☐ Nystagmus HP:0000639
  - ☐ Ptosis HP:0000508
  - ☐ Strabismus HP:0000486
  - ☐ Unilateral microphthalmos HP:0011480
  - ☐ Vivid blue eyes HP:0000635
  - **Ears**
  - ☐ Ear abnormalities HP:0000377
  - ☐ Ear tags HP:0000384
  - ☐ Low-set ears HP:0000369
  - ☐ Posteriorly rotated ears HP:0000358
  - **Nose**
  - ☐ Depressed nasal bridge HP:0005280
  - ☐ Prominent nasal bridge HP:0000426
  - **Mouth**
  - ☐ Cleft lip HP:0410030
  - ☐ Cleft palate HP:0000175
  - ☐ High palate HP:0000218
  - ☐ Long philtrum HP:0000343
  - ☐ Macrotia HP:0000400
  - ☐ Micrognathia HP:0000347
  - ☐ Robin Sequence HP:0000201
  - **Neck**
  - ☐ Branchial arch abnormality HP:0009794
  - ☐ Cystic hygroma HP:0000476
  - ☐ Short neck HP:0000470
  - ☐ Webbed neck HP:0000465
  - **Other**
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

GASTROINTESTINAL

- ☐ Yes (Provide Details Below)
  - ☐ Abdominal pain HP:0002027
  - ☐ Aganglionic megacolon HP:0002251
  - ☐ Chronic diarrhea HP:0002028
  - ☐ Cirrhosis HP:0001394
  - ☐ Constipation HP:0002019
  - ☐ Diarrhea HP:0002014
  - ☐ Elevated hepatic transaminase HP:0002910
  - ☐ Gastritis HP:0005263
  - ☐ Gastroesophageal reflux HP:0002020
  - ☐ Gastroschisis HP:0001543
  - ☐ Hepatic failure HP:0001399
  - ☐ Hepatic fibrosis HP:0001395
  - ☐ Hepatomegaly HP:0002240
  - ☐ Inflammation of the large intestine HP:0002037
  - ☐ Malabsorption HP:0002024
  - ☐ Pyloric stenosis HP:0002021
  - ☐ Secretory diarrhea HP:0005208
  - ☐ Vomiting HP:0002013
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

GENITOURINARY

- ☐ Yes (Provide Details Below)
  - ☐ Abnormal renal morphology HP:0012210
  - ☐ Abnormality of the urinary system HP:0000079
  - ☐ Cryptorchidism HP:0000028
  - ☐ Hydronephrosis HP:0000126
  - ☐ Micropenis HP:0000054
  - ☐ Nephronophthisis HP:0000090
  - ☐ Renal abnormality HP:0000077
  - ☐ Renal agenesis HP:0000104
  - ☐ Renal cyst HP:0000107
  - ☐ Renal tubular dysfunction HP:0000124
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

PATIENT NAME	DATE OF BIRTH	PHYSICIAN NAME	PRACTICE NAME

PHENOTYPE

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

GROWTH:
- ☐ Yes (Provide Details Below)
  - ☐ Failure to thrive HP:0001508
  - ☐ Growth delay HP:0001510
  - ☐ Overgrowth HP:0001548
  - ☐ Short stature HP:0004322
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

HEARING AND VISION LOSS

LATERALITY:
- ☐ Bilateral HP:0012832
  - ☐ Bilateral conductive hearing impairment HP:0008513
  - ☐ Bilateral sensorineural hearing impairment HP:0008619
- ☐ Unilateral HP:0012833
  - ☐ Unilateral conductive hearing impairment HP:0040119
  - ☐ Unilateral deafness HP:0009900

PROGRESSION:
- ☐ Fluctuating
- ☐ Progressive hearing impairment HP:0001730
- ☐ Stable
- ☐ Unknown

HEARING LOSS:
- ☐ Yes (Provide Details Below)
  - ☐ Absent ABR w/ cochlear microphonic HP:0004463
  - ☐ Auditory Neuropathy/Dys-Synchrony
  - ☐ Balance problems HP:0002141
  - ☐ Conductive hearing impairment HP:0000405
  - ☐ Enlarged vestibular aqueduct HP:0011387
  - ☐ Mixed hearing impairment HP:0000410
  - ☐ Mondini dysplasia HP:0000376
  - ☐ Present Otoacoustic Emissions
  - ☐ Sensorineural hearing impairment HP:0000407
  - ☐ Stapes fixation HP:0000381
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

SEVERITY (PTA): *PLEASE ATTACH AUDIOGRAM IF AVAILABLE.*

Left Ear:
- ☐ Mild hearing impairment (15-30Db) HP:0012712
- ☐ Moderate hearing impairment (31-50Db) HP:0012713
- ☐ Moderately-Severe (51-70Db)
- ☐ Severe hearing impairment (71-90Db) HP:0012714
- ☐ Profound hearing impairment (>90Db) HP:0012715

Right Ear:
- ☐ Mild hearing impairment (15-30Db) HP:0012712
- ☐ Moderate hearing impairment (31-50Db) HP:0012713
- ☐ Moderately-Severe (51-70Db)
- ☐ Severe hearing impairment (71-90Db) HP:0012714
- ☐ Profound hearing impairment (>90Db) HP:0012715

VISION LOSS
- ☐ Yes (Provide Details Below)
  - ☐ Abnormal ERG HP:0000512
  - ☐ Achromatopsia HP:0011516
  - ☐ Color blindness HP:0007641
  - ☐ Cystoid macular edema HP:0011505
  - ☐ Delayed pupillary response HP:0030211
  - ☐ Glaucoma HP:0000501
  - ☐ Keratoconus HP:0000563
  - ☐ Macular degeneration HP:0000608
  - ☐ Myopia HP:0000545
  - ☐ Night Blindness HP:0007642
  - ☐ Ophthalmoplegia HP:0000602
  - ☐ Optic atrophy HP:0000648
  - ☐ Photophobia HP:0000613
  - ☐ Retinal degeneration HP:0000546
  - ☐ Retinal Detachment HP:0000541
  - ☐ Rod-cone dystrophy HP:0000510
  - ☐ Tunnel vision HP:0007994
  - ☐ Visual impairment HP:0000505
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

IMMUNE

AUTOIMMUNE
- ☐ Yes (Provide Details Below)
  - ☐ Autoimmune hemolytic anemia HP:0001890
  - ☐ Fatigue HP:0012378
  - ☐ Fever HP:0001945
  - ☐ Joint pain
  - ☐ Keratoconjunctivitis sicca HP:0001097
  - ☐ Skin rash HP:0000988
  - ☐ Systemic lupus erythematosus HP:0002725
  - ☐ Xerostomia HP:0000217
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

IMMUNODEFICIENCY
- ☐ Yes (Provide Details Below)
  - ☐ Absence of CD8-positive T cells HP:0005422
  - ☐ Chronic bronchitis HP:0004469
  - ☐ Impaired T cell function HP:0005435
  - ☐ Otitis media HP:0000388
  - ☐ Pneumonia HP:0002090
  - ☐ Recurrent infections HP:0002719
  - ☐ Recurrent opportunistic infections HP:0005390
  - ☐ Severe combined immunodeficiency HP:0004430
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

INTELLECTUAL DISABILITY
- ☐ Yes (Provide Details Below)
  - ☐ Intellectual disability HP:0001249
  - ☐ Intellectual disability, mild HP:0001256
  - ☐ Intellectual disability, moderate HP:0002342
  - ☐ Intellectual disability, severe HP:0010864
- ☐ No
- ☐ Unknown
- ☐ Cognitive Details (Provide Iq Score If Known) _____

METABOLIC
- ☐ Yes (Provide Details Below)
  - ☐ Decreased plasma carnitine HP:0003234
  - ☐ Feeding difficulties HP:0011968
  - ☐ Hyperalaninemia HP:0003348
  - ☐ Hypercholesterolemia HP:0003124
  - ☐ Hypoglycemia HP:0001943
  - ☐ Increased CSF lactate HP:0002490
  - ☐ Increased serum pyruvate HP:0003542
  - ☐ Ketosis HP:0001946
  - ☐ Lactic acidosis HP:0003128
  - ☐ Obesity HP:0001513
  - ☐ Organic aciduria HP:0001992
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

MUSCULAR
- ☐ Yes (Provide Details Below)
  - ☐ Abnormal levels of creatine kinase in blood HP:0040081
  - ☐ Areflexia HP:0001284
  - ☐ Babinski sign HP:0003487
  - ☐ Distal amyotrophy HP:0003693
  - ☐ Distal muscle weakness HP:0002460
  - ☐ Dysarthria HP:0001260
  - ☐ Dysphagia HP:0002015
  - ☐ Foot dorsiflexor weakness HP:0009027
  - ☐ Hyporeflexia HP:0001265
  - ☐ Lower limb muscle weakness HP:0007340
  - ☐ Muscular dystrophy HP:0003560
  - ☐ Reduced tendon reflexes HP:0001315
  - ☐ Upper limb muscle weakness HP:0003484
    - ☐ Abnormality of movement HP:0100022
    - ☐ Elevated serum creatine phosphokinase HP:0003236
    - ☐ Flexion contracture HP:0001371
    - ☐ Generalized hypotonia HP:0001290
    - ☐ Hyperreflexia HP:0001347
    - ☐ Hypertonia HP:0001276
    - ☐ Joint hypermobility HP:0001382
    - ☐ Muscle weakness HP:0001324
    - ☐ Muscular hypotonia HP:0001252
    - ☐ Other: _____
- ☐ No
- ☐ Unknown

NEUROLOGICAL CONDITIONS
- ☐ Yes (Provide Details Below)
  - ☐ Abnormal nerve conduction velocity HP:0040129
  - ☐ Ataxia HP:0001251
  - ☐ Bulbar signs HP:0002483
  - ☐ Cerebral hypomyelination HP:0006808
  - ☐ Chorea HP:0002072
  - ☐ CNS hypomyelination HP:0003429
  - ☐ Congenital peripheral neuropathy HP:0006903
  - ☐ Distal sensory impairment HP:0002936
  - ☐ Dystonia HP:0001332
  - ☐ Facial palsy HP:0010628
  - ☐ Headache HP:0002315
  - ☐ Migraine HP:0002076
  - ☐ Motor axonal neuropathy HP:0007002
  - ☐ Motor polyneuropathy HP:0007178
  - ☐ Parkinsonism HP:0001300
  - ☐ Peripheral hypomyelination HP:0007182
  - ☐ Peripheral neuropathy HP:0009830
  - ☐ Pes cavus HP:0001761
  - ☐ Pressure Palsy
  - ☐ Recurrent paroxysmal headache HP:0002331
  - ☐ Sensory neuropathy HP:0000763
  - ☐ Sleep apnea HP:0010535
  - ☐ Spasticity HP:0001257
  - ☐ Stroke HP:0001297
  - ☐ Stroke-like episode HP:0002401
  - ☐ Sudden episodic apnea HP:0002882
  - ☐ Tremor HP:0001337
  - ☐ Upper motor neuron dysfunction HP:0002493
  - ☐ Vocal cord paresis HP:0001604
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

NEUROLOGICAL DEVELOPMENT
- ☐ Yes (Provide Details Below)
  - ☐ Absent speech HP:0001344
  - ☐ Delayed fine motor development HP:0010862
  - ☐ Delayed gross motor development HP:0002194
  - ☐ Delayed speech and language development HP:0000750
  - ☐ Developmental regression HP:0002376
  - ☐ Global developmental delay HP:0001263
  - ☐ Specific learning disability HP:0001328
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

ONCOLOGY
- ☐ Yes (Provide Details Below)
  - ☐ Adenomatous colonic polyposis HP:0005227
  - ☐ Breast carcinoma HP:0003002
  - ☐ Colorectal polyposis HP:0200063
  - ☐ Leukemia HP:0001909
  - ☐ Mucinous colorectal carcinoma HP:0031497
  - ☐ Myelofibrosis HP:0011974
  - ☐ Neoplasm of the lung HP:0100526
  - ☐ Neoplasm of the skin HP:0008069
  - ☐ Paraganglioma HP:0002668
  - ☐ Pheochromocytoma HP:0002666
  - ☐ Retinoblastoma HP:0009919
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

PULMONARY
- ☐ Yes (Provide Details Below)
  - ☐ Ciliary dyskinesia HP:0012265
  - ☐ Situs inversus totalis HP:0001696
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

SEIZURES
- ☐ Yes (Provide Details Below)
  - ☐ Absence seizures HP:0002121
    - ☐ Typical absence seizures HP:0011147
    - ☐ Atypical absence seizures HP:0007270
  - ☐ Atonic seizures HP:0010819
  - ☐ EEG abnormality HP:0002353
  - ☐ Epileptic encephalopathy HP:0200134
  - ☐ Epileptic spasms HP:0011097
  - ☐ Febrile seizures HP:0002373
  - ☐ Focal autonomic seizures HP:0011154
  - ☐ Focal clonic seizures HP:0002266
  - ☐ Focal motor seizures HP:0011153
    - ☐ With Psychic Symptoms
  - ☐ Focal myoclonic seizures HP:0011166
  - ☐ Focal seizures HP:0007359
  - ☐ Generalized clonic seizures HP:0011169
  - ☐ Generalized myoclonic seizures HP:0002123
  - ☐ Generalized seizures HP:0002197
  - ☐ Generalized tonic-clonic seizures HP:0002069
  - ☐ Infantile encephalopathy HP:0007105
  - ☐ Infantile spasms HP:0012469
  - ☐ Segmental myoclonic seizures HP:0025191
  - ☐ Specify Syndrome Or Differential Diagnosis (If Known) _____
  - ☐ Status epilepticus HP:0002133
  - ☐ Syndromic-Related Epilepsy
  - ☐ Other Seizure-Related Causes Or Complaints Reported
    - ☐ Syncope HP:0001279
    - ☐ Arrhythmia HP:0011675
    - ☐ Migraine HP:0002076
    - ☐ Vertigo HP:0002321
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

SKELETAL
- ☐ Yes (Provide Details Below)
  - ☐ Brachydactyly HP:0001156
  - ☐ Osteopenia HP:0000938
  - ☐ Osteoporosis HP:0000939
  - ☐ Pectus excavatum HP:0000767
  - ☐ Platyspondyly HP:0000926
  - ☐ Polydactyly HP:0010442
  - ☐ Rhizomelia HP:0008905
  - ☐ Scoliosis HP:0002650
  - ☐ Short ribs HP:0000773
  - ☐ Syndactyly HP:0001159
  - ☐ Talipes equinovarus HP:0001762
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

SKIN
- ☐ Yes (Provide Details Below)
  - ☐ Axillary freckling HP:0000997
  - ☐ Fragile skin HP:0001030
  - ☐ Inguinal freckling HP:0030052
  - ☐ Jaundice HP:0000952
  - ☐ Neurofibromas HP:0001067
  - ☐ Soft skin HP:0000977
  - ☐ Xanthomatosis HP:0000991
  - ☐ Other: _____
- ☐ No
- ☐ Unknown

PATIENT NAME	DATE OF BIRTH	PHYSICIAN NAME	PRACTICE NAME

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(s) that have been ordered in connection with this consent 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 me and my healthcare provider. I have reviewed the test order made in connection with this consent, and I hereby give consent to have my specimen tested as set forth in the order.

De-identified research

Sema4 may de-identify and use all data and information generated and received in connection with this test to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and I will receive no compensation in connection with such research. Data and information are “de-identified” by removing any information that could be used to identify a specific person, such as a name, email address, or date of birth. Sema4 may also give the de-identified data and information to its research partners and may submit it 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.

If I do not want to have any of my de-identified data and information used in research consistent with this consent, I may initial here_____, or I may withdraw this consent 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 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.

_____ _____

Signature of person being tested (or guardian) Date

Rev.04/17/2020

PANEL TABLE

CARDIOVASCULAR	Genes
COMPREHENSIVE CARDIOVASCULAR PANEL *Number of genes: 240*	*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, DSG2, DSP, DTNA, EFEMP2, EIF2AK4, ELAC2, ELN, EMD, ENG, EYA4, FBN1, FBN2, FHL1, FKBP14, FKRP, 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, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LEFTY2, LMNA, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MED13L, MEIS2, MFAP5, MIB1, MLYCD, MTO1, MUT, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF6, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NODAL, NOTCH1, NPPA, NR2F2, NRAS, NSD1, OBSCN, PCCA, PCCB, PCSK9, 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, SCO1, SDHA, SDHAF1, SGCD, SGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLC39A13, SLMAP, SMAD3, SMAD4, SMAD6, SMAD9, SNTA1, SOS1, SOS2, SPRED1, SURF1, TAZ, TBX1, TBX5, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3*
COMPREHENSIVE CARDIOMYOPATHY PANEL *Number of genes: 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, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FBN1, FBN2, FHL1, FKRP, 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, NDUFS7, 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, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO1, SDHA, SDHAF1, SGCD, SGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SURF1, TAZ, TBX1, TBX5, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL*
COMPREHENSIVE ARRHYTHMIAS PANEL *Number of genes: 54*	*ABCC9, ACTN2, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CTNNA3, DES, DSC2, DSG2, DSP, EMD, GJA5, GPD1L, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LDB3, LMNA, NKX2-5, NPPA, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLMAP, SNTA1, TGFB3, TMEM43, TRDN, TRPM4*
ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY PANEL *Number of genes: 8*	*DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43*
BRUGADA SYNDROME PANEL *Number of genes: 20*	*ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SLMAP, TRPM4*
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA PANEL *Number of genes: 8*	*ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN*
LONG/SHORT QT SYNDROME PANEL *Number of genes: 19*	*AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN*
DILATED CARDIOMYOPATHY PANEL *Number of genes: 57*	*ABCC9, ACTC1, ACTN2, AKAP9, ANK2, ANKRD1, BAG3, CACNA1C, CAV3, CAVIN4, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, 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, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL*
HYPERTROPHIC CARDIOMYOPATHY PANEL *Number of genes: 40*	*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, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL*
LEFT VENTRICULAR NONCOMPACTION PANEL *Number of genes: 20*	*ACTC1, DSP, DTNA, HCN4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH7, PLEKHM2, PLN, PRDM16, RYR2, SCN5A, TAZ, TNNI3, TNNT2, TPM1, VCL*
AORTOPATHIES PANEL *Number of genes: 33*	*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, TGFB2, TGFB3, TGFBR1, TGFBR2*
CONGENITAL HEART DISEASE PANEL *Number of genes: 43*	*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*
FAMILIAL HYPERCHOLESTEROLEMIA PANEL *Number of genes: 4*	*APOB, LDLR, LDLRAP1, PCSK9*
HEREDITARY HEMORRHAGIC TELANGIECTASIA PANEL *Number of genes: 5*	*ACVRL1, ENG, GDF2, RASA1, SMAD4*
METABOLIC CARDIOMYOPATHIES PANEL *Number of genes: 24*	*ACADVL, AGL, ALG1, ALG12, ARSB, CPT2, DOLK, FKRP, GAA, GBA, GBE1, GLA, GLB1, GUSB, HADHA, HEXB, IDUA, LAMP2, MTO1, PCCA, PCCB, SLC22A5, SLC25A20, TAZ*
NOONAN SPECTRUM DISORDERS PANEL *Number of genes: 18*	*BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1*
PULMONARY HYPERTENSION PANEL *Number of genes: 10*	*ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNA5, KCNK3, SMAD9*

HEARING AND VISION LOSS	Genes
COMPREHENSIVE HEARING AND VISION LOSS PANEL *Number of genes: 308*	*ABCA4, ABHD12, ACTG1, ADAM9, ADGRV1, AGK, AHI1, 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, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COCH, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP1B1, CYP4V2, DHDDS, DIABLO, DIAPH1, DNAJC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, ESPN, ESRRB, EYA1, EYA4, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FYCO1, FZD4, GCNT2, GIPC3, GJA8, GJB2, GJB6, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GPSM2, GRHL2, GRIP1, GRK1, GRM6, GRXCR1, GSDME, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HESX1, HGF, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, ILDR1, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KARS, KCNJ13, KCNQ1, KCNQ4, KCNV2, KIF11, KIF7, KLHL7, LCA5, LHFPL5, LOXHD1, LRAT, LRIT3, LRP5, LRTOMT, LYST, LZTFL1, MAK, MARVELD2, MERTK, MFRP, MFSD8, MITF, MKKS, MKS1, MSRB3, MT-RNR1, MTTP, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PGK1, PHYH, PITPNM3, PITX2, PITX3, PJVK, PLA2G5, POU3F4, POU4F3, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PTPRQ, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RDX, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SERPINB6, SIX1, SIX5, SLC24A1, SLC26A4, SLC45A2, SMOC1, 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, TPRN, TREX1, TRIM32, TRIOBP, TRPM1, TSPAN12, TSPEAR, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423*
COMPREHENSIVE HEARING LOSS PANEL *Number of genes: 92*	*ACTG1, ADGRV1, AIFM1, CACNA1D, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLRN1, COCH, COL11A2, DIABLO, DIAPH1, EDN3, EDNRB, ESPN, ESRRB, EYA1, EYA4, GIPC3, GJB2, GJB6, GPSM2, GRHL2, GRXCR1, GSDME, HARS, HGF, ILDR1, KARS, KCNQ1, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MITF, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PHYH, PJVK, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SIX5, SLC26A4, SMPX, SOX10, STRC*, TBC1D24, TECTA, TIMM8A, TMC1, TMEM126A, TMIE, TMPRSS3, TPRN, TRIOBP, TSPEAR, USH1C, USH1G, USH2A, WFS1, WHRN*
CONNEXIN 26 / CONNEXIN 30 DEL / DUP HEARING LOSS PANEL *Number of genes: 2*	*GJB2, GJB6*
BRANCHIO-OTO-RENAL SYNDROME PANEL *Number of genes: 3*	*EYA1, SIX1, SIX5*
USHER SYNDROME PANEL *Number of genes: 11*	*ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN*
ZELLWEGER SYNDROME PANEL *Number of genes: 9*	*PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7*
COMPREHENSIVE VISION LOSS NGS PANEL *Number of genes: 250*	*ABCA4, ABHD12, ADAM9, ADGRV1, AGK, AHI1, AIPL1, ALMS1, AP3B1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BLOC1S6, BMP4, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP1B1, CYP4V2, DHDDS, DNAJC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, EYA1, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FYCO1, FZD4, GCNT2, GJA8, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GRIP1, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HESX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KCNJ13, KCNV2, KIF11, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP5, LYST, LZTFL1, MAK, MERTK, MFRP, MFSD8, MITF, MKKS, MKS1, MTTP, 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, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SMOC1, SNRNP200, SOX10, SOX2, SPATA7, STRA6, TCTN1, TCTN2, TCTN3, TDRD7, TGFBI, TIMM8A, TIMP3, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423*
ALBINISM / HERMANSKY-PUDLAK / WAARDENBURG PANEL *Number of genes: 18*	*AP3B1, BLOC1S6, CACNA1F, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, MITF, OCA2, PAX3, SLC45A2, SOX10, TYR, TYRP1*
DEVELOPMENTAL EYE PANEL *Number of genes: 21*	*BCOR, BMP4, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GRIP1, HCCS, KIF11, MFRP, NDP, OTX2, PAX6, PITX3, PLA2G5, PXDN, SMOC1, SOX2, STRA6, VSX2*
RETINAL DISEASE PANEL *Number of genes: 154*	*ABCA4, ABHD12, ADAM9, ADGRV1, AHI1, 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, CNGA3, CNGB1, CNGB3, CNNM4, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GNAT2, GPR179, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, IFT140, IMPDH1, IMPG2, INPP5E, IQCB1, 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, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, SAG, SDCCAG8, SEMA4A, SLC24A1, SNRNP200, SPATA7, TCTN1, TCTN2, TCTN3, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TTC21B, TTC8, TULP1, USH1C, USH1G, USH2A, WDR19, WHRN, ZNF423*
STICKLER AND CATARACT PANEL *Number of genes: 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, PEX16, 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
COMPREHENSIVE EPILEPSY AND AUTISM PANEL *Number of genes: 401*	*ABAT, ABCD1, ACSL4, ACY1, ADGRG1, ADGRV1, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ALDH7A1, ALG13, AMT, ANK3, ANKRD11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARFGEF2, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATP7A, ATP8A2, ATR, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, CA8, CACNA1A, CACNA1C, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D1A, CCDC22, CCDC88C, CCM2, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTCF, CTNNB1, CTSD, CUL3, CUL4B, CYP27A1, D2HGDH, DCX, DDHD2, DDX3X, DEAF1, DEPDC5, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNAJC5, DNM1, DNMT3A, DOCK7, DPYD, DYNC1H1, DYRK1A, EBP, EEF1A2, EFHC1, EHMT1, EIF2S3, ELP4, EMX2, EPM2A, EZH2, FGD1, FGFR3, FKRP, FLNA, FLVCR2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATAD2B, GATM, GCSH, GDI1, GK, GLDC, GLI2, GLI3, GNAO1, GNB1, GNS, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HCCS, HCN1, HCN4, HDAC8, HGSNAT, HIP1, HNRNPU, HPRT1, HRAS, HSD17B10, HTRA1, HUWE1, IDS, IL1RAPL1, IQSEC2, ITPA, KANSL1, KAT6A, KAT6B, KCNA1, KCNA2, KCNB1, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, 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, MCPH1, MECP2, MED12, MED13L, MED23, MEF2C, MFSD8, MID1, MTHFR, MTOR, MYT1L, NAA10, NAGLU, NDE1, NDP, NDUFA1, NECAP1, NEXMIF, NF1, NFIX, NHLRC1, NHS, NIPBL, NLGN4X, NOTCH3, NPRL2, NPRL3, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCRL, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PDHA1, PGK1, PHF6, PHF8, PIGA, PIGN, PIGO, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POGZ, POLG, POMGNT1, POMT1, POMT2, PORCN, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRPS1, PRRT2, PTCH1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAF1, RAI1, RARS2, RBM10, RELN, RIT1, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPL10, RPS6KA3, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, SETBP1, SETD2, SETD5, SGSH, 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 PANEL *Number of genes: 226*	*ABAT, ACY1, ADGRG1, ADGRV1, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP1S2, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, BCKDK, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CCDC88C, CCM2, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTSD, CUL4B, DCX, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EMX2, EPM2A, FGD1, FGFR3, FKRP, 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, HNRNPU, HSD17B10, HTRA1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF1BP, KMT2D, KRIT1, LARGE1, LBR, LGI1, LIAS, MAGI2, MAPK10, MBD5, MCPH1, MECP2, MEF2C, MFSD8, MTHFR, MTOR, NDE1, NDUFA1, NECAP1, NHLRC1, NIPBL, NOTCH3, NPRL2, NPRL3, NRXN1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PHF6, PIGA, PIGO, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRRT2, PTEN, RAB39B, RAI1, RARS2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, 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 genes: 52*	*ACY1, ADGRV1, ALDH5A1, ALDH7A1, BCKDK, CACNA1H, CACNA2D2, CACNB4, CASR, CCM2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CNTNAP2, CPA6, CSTB, DEPDC5, EFHC1, EPM2A, FOLR1, GABRB3, GABRD, GABRG2, GNB1, GOSR2, GRIN2A, HTRA1, KCNJ11, KCNMA1, KCNQ3, KCTD7, KRIT1, NDE1, NDUFA1, NHLRC1, NPRL2, NPRL3, PDCD10, PRICKLE1, PRICKLE2, PTEN, RARS2, SCARB2, SCN1B, SCN3A, SCN9A, SERPINI1, SLC6A1, SNAP25, TRPM6*
INFANTILE EPILEPSY PANEL *Number of genes: 58*	*ABAT, ADSL, ALG13, AMT, ARHGEF9, ARX, CDKL5, CHD2, DNM1, DOCK7, DYRK1A, EEF1A2, GABRA1, GABRB2, GCSH, GLDC, GNAO1, GNB1, GPHN, GRIN2B, HCN1, HIP1, ITPA, KCNA2, KCNB1, KCNQ2, KCNT1, LIAS, MAGI2, MAPK10, MECP2, MTHFR, NECAP1, PCDH19, PIGA, PIGO, PLCB1, PNKP, PNPO, RARS2, RNASEH2A, ROGDI, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC35A2, SLC6A8, SPTAN1, ST3GAL3, STX1B, STXBP1, SZT2, TBC1D24, WWOX*
MIGRAINE PANEL *Number of genes: 7*	*ATP1A2, CACNA1A, NOTCH3, POLG, PRRT2, SCN1A, SLC2A1*
NEURONAL CEROID LIPOFUSCINOSES PANEL *Number of genes: 9*	*CLN3, CLN5, CLN6, CLN8, CTSD, DNAJC5, MFSD8, PPT1, TPP1*
NEURONAL MIGRATION PANEL *Number of genes: 22*	*ADGRG1, C0L18A1, COL4A1, EMX2, FGFR3, FKRP, FLNA, LARGE1, PAFAH1B1, PAX6, POMGNT1, POMT1, POMT2, PQBP1, RELN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1*
SYNDROMIC EPILEPSY AND INTELLECTUAL DISABILITY PANEL *Number of genes: 93*	*AP1S2, ARFGEF2, ASPM, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, CASK, CCDC88C, CNTNAP2, CREBBP, CUL4B, DCX, DYRK1A, EEF1A2, EHMT1, FGD1, FLVCR2, FOXG1, GAMT, GATM, GLI2, GPC3, GRIA3, GRIN1, HCN4, HNRNPU, HSD17B10, IQSEC2, KANSL1, KCNA1, KCNA2, KCNJ1, KCNJ10, KDM5C, KIF1BP, KMT2D, LBR, LGI1, MBD5, MCPH1, MECP2, MEF2C, MTOR, NIPBL, NRXN1, OPHN1, PAK3, PANK2, PHF6, PIGV, PLA2G6, PLP1, POLG, PQBP1, RAB39B, RAI1, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCN1A, SCN5A, SETBP1, SHANK3, SLC12A5, SLC19A3, SLC25A19, SLC2A1, SLC4A10, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SNAP29, ST3GAL5, SYN1, SYNGAP1, SYP, TBX1, TCF4, TREX1, TSC1, TSC2, UBE3A, VANGL1, VPS13A, VPS13B, WDR62, ZEB2*
COMPREHENSIVE AUTISM SPECTRUM DISORDER PANEL *Number of genes: 228*	*ABCD1, ACSL4, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ANK3, ANKRD11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARID1A, ARID1B, ARX, ATP13A2, ATP7A, ATP8A2, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, CA8, CACNA1C, CASK, CBL, CC2D1A, CCDC22, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CLCN4, CNTNAP2, CREBBP, CTCF, CTNNB1, CUL3, CYP27A1, D2HGDH, DDHD2, DDX3X, DEAF1, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNMT3A, DPYD, DYNC1H1, DYRK1A, EBP, EHMT1, EIF2S3, ELP4, EZH2, FGD1, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GATAD2B, GDI1, GK, GLI3, GNS, GPC3, GRIA3, GRIK2, GRIN2B, GRIP1, HCCS, HDAC8, HGSNAT, HPRT1, 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, MEF2C, MID1, MYT1L, NAA10, NAGLU, NDP, NEXMIF, NF1, NFIX, NHS, NIPBL, NLGN4X, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCRL, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PCDH19, PDHA1, PGK1, PHF6, PHF8, PIGN, PNKP, POGZ, PORCN, PQBP1, PRPS1, PTCH1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAF1, RAI1, RARS2, RBM10, RIT1, RPL10, RPS6KA3, SATB2, SCN1A, SCN2A, SETBP1, SETD2, SETD5, SGSH, SHANK2, SHANK3, SHOC2, SLC16A2, SLC25A1, SLC6A1, SLC6A4, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SOS1, SOX11, SOX5, SPRED1, STXBP1, SYN1, SYNGAP1, TAF1, TBL1XR1, TBR1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TRAPPC9, TRIO, TSC1, TSC2, TSPAN7, TUBA1A, TUSC3, UBE2A, UBE3A, UNC80, UPF3B, USP9X, VPS13B, WAC, WDR45, WDR81, YWHAE, ZC4H2, ZDHHC9, ZEB2, ZMYND11, ZNF407, ZNF711*
STAT AUTISM SPECTRUM DISORDER PANEL *Number of genes: 30*	*AHI1, AP1S2, ARX, CACNA1C, CDKL5, CNTNAP2, DHCR7, FMR1, GPC3, GRIA3, IL1RAPL1, KDM5C, MECP2, NLGN4X, NRXN1, NSD1, OPHN1, OTC, PCDH19, PTCHD1, PTEN, RAB39B, SHANK2, SHANK3, SLC6A8, SLC9A6, TSC1, TSC2, UBE3A, UPF3B*
EPILEPSY PHARMACOGENETIC PANEL *Number of genes: 10*	*COMT**, CYP2B6*, CYP2C19*, CYP2C9*, CYP2D6*, GRIK4**, HTR2A**, OPRM1**, POLG, UGT2B15***
MICROCEPHALY PANEL *Number of genes: 78*	*ACTB, ACTG1, ADGRG1, ARFGEF2, ARX, ASPM, ATR, ATRIP, CASK, CDK5RAP2, CDKL5, CDON, CENPJ, CEP135, CEP152, CEP63, CREBBP, DCX, DHCR7, DISP1, DLL1, DYNC1H1, EP300, ERCC6, ERCC8, FGF8, FKRP, FKTN, FOXG1, FOXH1, GAS1, GLI2, HDAC8, KIF11, KIF2A, KNL1, LARGE1, MCPH1, MECP2, MED17, MRE11, NBN, NDE1, NHEJ1, NIN, NIPBL, NODAL, PAFAH1B1, PCNT, PHGDH, PNKP, POMGNT1, POMT1, POMT2, PTCH1, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RBBP8, RELN, RNU4ATAC, SHH, SIX3, SLC25A19, SLC9A6, SMC1A, SMC3, STAMBP, STIL, TDGF1, TGIF1, TUBA1A, TUBG1, UBE3A, VLDLR, WDR62, ZIC2*

*Only copy number variants and targeted genotyping reported in these genes ** Only targeted genotyping reported in these genes.

PORPHYRIA	Genes
Acute Intermittent Porphyria (AIP)	*HMBS*
Acute Porphyrias Panel (AIP, HCP, & VP) *Number of genes: 3*	*HMBS, CPOX, PPOX*
Congenital Erythropoietic Porphyria (CEP) *Number of genes: 2*	*UROS, ALAS2 (exon 11)*
Erythropoietic Protoporphyria Panel	*FECH, ALAS2 (Exon 11)*
PEPT2 (SLC15A2) Genotyping for Acute Porphyria Patients	*SLC15A2*
Hereditary Coproporphyria (HCP)	*CPOX*
Porphyria Cutanea Tarda (PCT)	*UROD*
Variegate Porphyria (VP)	*PPOX*

SKELETAL	Genes
CRANIOSYNOSTOSIS PANEL *Number of genes: 8*	*ENFB1**, FGFR 1**, FGFR2**, FGFR3**, MSX2**, POR**, RAB23**, TWIST1***
LIMB DEFECTS PANEL *Number of genes: 8*	*GLI3, HOXD13, ROR2, SALL1, SALL4, TBX5, WNT7A, ZRS*

**Only targeted genotyping reported in these genes

IMMUNODEFICIENCY	Genes
COMPREHENSIVE IMMUNODEFICIENCY PANEL *Number of genes: 250*	*ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, B2M, BTK, C1QA, C1QB, C1QC, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCDC39, CCDC40, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD59, CD79A, CD79B, CD81, CD8A, CEBPE, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DGKE, DKC1, DNMT3B, DOCK2, DOCK8, DUOX2, ELANE, EPCAM, EPG5, FADD, FAS, FASLG, FCN1, FERMT1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA2, GATA3, GFI1, GUCY2C, HAX1, HPS1, HPS4, HPS6, ICOS, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL18, IL18RAP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF7, IRF8, IRGM, ISG15, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, LAMTOR2, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYST, MAGT1, MALT1, MAP3K14, MASP2, MEFV, MRE11, MVK, MYD88, MY05B, NBN, NCF1, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, NRAS, ORAI1, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, PRF1, PRKCD, PRKDC, PTEN, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RET, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SBDS, SERPING1, SH2D1A, SKIV2L, SLC35C1, SLC37A4, SLC9A3, SMARCAL1, SP110, SPINK5, STAT1, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCN2, TERC, TERT, TINF2, TLR3, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF12, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNG, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24*
PRIMARY IMMUNODEFICIENCY PANEL *Number of genes: 206*	*ACD, ACP5, ADA, ADA2, ADAR, AICDA, AIRE, AK2, AP3B1, B2M, BTK, C1QA, C1QB, C1QC, C1S, C2, C4A, C5, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD59, CD79A, CD79B, CD8A, CEBPE, CFD, CFH, CFI, CFP, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DGKE, DKC1, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, FADD, FAS, FASLG, FCN1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA2, GATA3, GFI1, HAX1, ICOS, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IL10, IL10RA, IL12RB1, IL17RA, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF7, IRF8, ISG15, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, LAMTOR2, LCK, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MASP2, MEFV, MVK, MYD88, NBN, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, NRAS, ORAI1, PGM3, PIK3CD, PIK3R1, PLCG2, PNP, POLE, PRF1, PRKCD, PRKDC, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SH2D1A, SKIV2L, SLC35C1, SLC37A4, SMARCAL1, SP110, SPINK5, STAT1, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCN2, TERC, TERT, TINF2, TLR3, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF12, TREX1, TRNT1, TTC7A, TYK2, UNC13D, UNG, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24*
INFLAMMATORY BOWEL DISEASE PANEL *Number of genes: 59*	*ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL17RA, IL21, IL23R, IL2RA, IL2RG, IL7R, ITGB2, LIG4, LRBA, LYST, MEFV, MVK, NCF2, NCF4, NHEJ1, NLRC4, NOD2, ORAI1, PIK3CD, PIK3R1, PLCG2, PNP, PRF1, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RTEL1, SH2D1A, SLC37A4, STAT1, STAT3, STX11, STXBP2, TTC7A, WAS, XIAP, ZAP70*
SEVERE COMBINED IMMUNODEFICIENCY PANEL *Number of genes: 26*	*ADA, AK2, CD247, CD3D, CD3E, CORO1A, DCLRE1C, DOCK8, FOXN1, IL2RG, IL7R, JAK3, LIG4, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RMRP, STAT5B, STIM1, TBX1, ZAP70*

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, ETFA, 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, MFSD8, NAGLU, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, 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, PHYH*

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, DUOXA2, DYSF, EDA, EFNB1, EIF2B5, ELP1, EMD, EPB42, ESCO2, EVC, FANCA, FANCC, FANCG, FBN1, GFM1, GJB1, GLE1, GP1BA, GP9, GRHPR, GSS, HAX1, HJV, HMGCS2, HOGA1, HSD3B2, HSD3B7, HYLS1, IGSF1, IL2RG, IL7R, IYD, 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, PTPRC, PUS1, RAG1, RAG2, RAPSN, RMRP, RTEL1, SACS, SEPSECS, SGCA, SGCB, SGCG, SLC12A3, SLC12A6, SLC25A15, SLC26A2, SLC35A3, SLC39A4, 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

ABAT
ABCA4
ABCB11
ABCC8
ABCC9
ABCD1
ABHD12
ACAD9
ACADM
ACADVL
ACAT1
ACD
ACOX1
ACP5
ACSF3
ACSL4
ACTA2
ACTB
ACTC1
ACTG1
ACTN2
ACVR2B
ACVRL1
ACY1
ADA
ADA2
ADAM17
ADAM9
ADAMTS2
ADAR
ADGRG1
ADGRV1
ADNP
ADSL
AFF2
AGA
AGK
AGL
AGO1
AGPS
AGXT
AHI1
AICDA
AIFM1
AIPL1
AIRE
AK2
AKAP9
AKR1D1
ALAS2
ALDH3A2
ALDH5A1
ALDH7A1
ALDOB
ALG1
ALG12
ALG13
ALG6
ALMS1
ALPK3
ALPL
AMT
ANK1
ANK2
ANK3
ANKRD1
ANKRD11
AP1S2
AP3B1
AP4B1
AP4E1
AP4M1
AP4S1
APOB
APOL1
AQP2
ARFGEF2
ARG1
ARHGEF9
ARID1A
ARID1B
ARL13B
ARL6
ARSA
ARSB
ARX
ASL
ASNS
ASPA
ASPM
ASS1
ATM
ATP13A2
ATP1A2
ATP2A2
ATP6AP2
ATP6V0A2
ATP6V1B1
ATP7A
ATP7B
ATP8A2
ATR
ATRIP
ATRX
AUTS2
AVPR2
B2M
BAG3
BBS1
BBS10
BBS12
BBS2
BBS4
BBS5
BBS7
BBS9
BCKDHA
BCKDHB
BCKDK
BCL11A
BCOR
BCS1L
BEST1
BLM
BLOC1S6
BMP4
BMPR1B
BMPR2
BRAF
BRWD3
BSND
BTD
BTK
C120RF57
C1QA
C1QB
C1QC
C1S
C2
C3
C4A
C4B
C5
C6
C7
C8A
C8B
C8ORF37
C9
CA4
CA8
CABP4
CACNA1A
CACNA1C
CACNA1D
CACNA1F
CACNA1H
CACNA2D1
CACNA2D2
CACNA2D4
CACNB2
CACNB4
CALM1
CALM2
CALM3
CAPN3
CAPN5
CARD11
CARD14
CARD9
CASK
CASP10
CASP8
CASQ2
CASR
CAV1
CAV3
CAVIN4
CBL
CBS
CC2D1A
CC2D2A
CCDC22
CCDC39
CCDC40
CCDC50
CCDC88C
CCM2
CD19
CD247
CD27
CD3D
CD3E
CD3G
CD40
CD40LG
CD46
CD59
CD79A
CD79B
CD81
CD8A
CDH23
CDH3
CDHR1
CDK5RAP2
CDKL5
CDKN1C
CDON
CEACAM16
CEBPE
CENPJ
CEP135
CEP152
CEP164
CEP290
CEP41
CEP63
CERKL
CFB
CFD
CFH
CFHR1
CFHR3
CFI
CFP
CFTR
CHD2
CHD7
CHD8
CHIT1
CHM
CHRM2
CHRNA2
CHRNA4
CHRNA7
CHRNB2
CHRNE
CHST14
CIB2
CIITA
CLCN2
CLCN4
CLDN14
CLN3
CLN5
CLN6
CLN8
CLPB
CLRN1
CNGA1
CNGA3
CNGB1
CNGB3
CNNM4
CNTNAP2
COCH
COL11A1
COL11A2
COL18A1
COL1A1
COL1A2
COL27A1
COL2A1
COL3A1
COL4A1
COL4A3
COL4A4
COL4A5
COL5A1
COL5A2
COL7A1
COL9A1
COL9A2
COMT
COPA
CORO1A
COX10
COX15
CPA6
CPLANE1
CPOX
CPS1
CPT1A
CPT2
CR2
CRB1
CREBBP
CRELD1
CRX
CRYAA
CRYAB
CRYBB1
CRYBB3
CSF2RA
CSF3R
CSRP3
CSTB
CTC1
CTCF
CTLA4
CTNNA3
CTNNB1
CTNS
CTPS1
CTSC
CTSD
CTSK
CUL3
CUL4B
CXCR4
CYBA
CYBB
CYP11B1
CYP11B2
CYP17A1
CYP19A1
CYP1B1
CYP21A2
CYP27A1
CYP2B6
CYP2C19
CYP2C9
CYP2D6
CYP3A5
CYP4V2
D2HGDH
DBT
DCLRE1B
DCLRE1C
DCX
DDHD2
DDX3X
DEAF1
DEPDC5
DES
DGKE
DHCR7
DHDDS
DIABLO
DIAPH1
DIS3L2
DISP1
DKC1
DLD
DLG3
DLL1
DMD
DNAH5
DNAI1
DNAI2
DNAJC5
DNM1
DNMT3A
DNMT3B
DOCK2
DOCK7
DOCK8
DOLK
DPYD
DSC2
DSG2
DSP
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