



**Please fill out all the highlighted fields. Failure to do so may result in delayed testing and delivery of results.**

## PATIENT INFORMATION

Sema4 will use this information to contact the patient via automatic email, SMS, and/or phone regarding payment, testing status, and online results access. By submitting this requisition, I confirm that I have obtained the patient's authorization to be contacted by Sema4 by these means (email address must be specific to patient listed on form).

PATIENT EMAIL ADDRESS <i>RECOMMENDED</i>	PATIENT MOBILE/PRIMARY NUMBER <i>REQUIRED</i>
LAST NAME <i>REQUIRED</i>	FIRST NAME <i>REQUIRED</i> MI
DATE OF BIRTH MM / DD / YYYY	BIOLOGICAL SEX <input type="checkbox"/> M <input type="checkbox"/> F <i>REQUIRED</i>
CLIENT MRN	
ADDRESS <i>REQUIRED</i>	CITY / STATE / ZIP <i>REQUIRED</i>
BIOLOGICAL MOTHER LAST NAME	BIOLOGICAL FATHER LAST NAME
BIOLOGICAL MOTHER FIRST NAME	BIOLOGICAL FATHER FIRST NAME
BIOLOGICAL MOTHER DATE OF BIRTH MM / DD / YYYY	BIOLOGICAL FATHER DATE OF BIRTH MM / DD / YYYY

## BILLING INFORMATION

Bill to:  Client/Institution  Insurance  Self Pay/No Insurance

POLICYHOLDER LAST NAME <i>REQUIRED</i>	POLICYHOLDER FIRST NAME <i>REQUIRED</i>	POLICYHOLDER DOB MM / DD / YYYY <i>REQUIRED</i>
INSURANCE CARRIER <i>REQUIRED</i>	INSURANCE ID <i>REQUIRED</i>	GROUP NO. <i>REQUIRED</i>

BILLING ADDRESS  
*REQUIRED*

SECONDARY INSURANCE  YES  NO

SECONDARY INSURANCE NAME

GROUP NO.

Pre-Authorization #: \_\_\_\_\_ Please include a copy of all insurance paperwork.

**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 800-298-6470.

SIGNATURE \_\_\_\_\_ DATE MM / DD / YYYY

## ORDERING PROVIDER INFORMATION

NAME <i>REQUIRED</i>	GENETIC COUNSELOR
ADDRESS <i>REQUIRED</i>	CLINIC / INSTITUTION <i>REQUIRED</i>
	TELEPHONE
	FAX

**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 MM / DD / YYYY

## CLINICAL INDICATION (PLEASE FILL OUT ADDITIONAL INDICATIONS ON BACK)

**SPECIMEN TYPE:**

Patient:  Peripheral Blood  Saliva  Other: \_\_\_\_\_ Date of Collection: MM / DD / YYYY

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	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\***

## 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

### SKELTAL

- 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 LIPOFUSCINOSSES 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.

**COMPLETE AND SUBMIT ALL PAGES TO LABORATORY**

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**

- 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 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 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
  - Stopping 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 diarrhoea HP:0002028
  - Cirrhosis HP:0001394
  - Constipation HP:0002019
  - Diarrhea HP:0002014
  - Elevated hepatic transaminase HP:0002910
  - Gastritis HP:0005263
  - Gastroesophageal reflux HP:0002020
  - Gastrochisis 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
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## 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:0002151
  - 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:0001322
  - 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:00031497
  - 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
- Syndrom-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:0003052
  - Jaundice HP:0000952
  - Neurofibromas HP:0001067
  - Soft skin HP:0000977
  - Xanthomatosis HP:0000991
  - Other: \_\_\_\_\_
- No
- Unknown

**COMPLETE AND SUBMIT ALL PAGES TO LABORATORY**

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](http://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](mailto: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](mailto: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

FFP0122GE0121  
Revised 01/20/2021

# PANEL TABLE

Panel Name	Genes
<b>CARDIOVASCULAR</b> COMPREHENSIVE CARDIOVASCULAR PANEL Number of markers: 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, EYAA, FBN1, FBN2, FHL1, FKBP, FKTN, FLNA, FLNC, FOXH1, GAA, GATA4, GATA6, GATA7, 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, 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, SCD1, SDHA, SDHAF1, SGCD, SGGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLC39A13, SLMAP, SMAD3, SMAD4, SMAD6, SMAD9, SNTA1, SOS1, SOS2, SPRED1, SURF1, TAZ, TBX1, TBX5, TCFAP, TGF2, TGF3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNM3, 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, DSG2, DSP, DTNA, ELAC2, EMD, EYAA, FBN1, FBN2, FHL1, FKBP, FKTN, FLNA, FLNC, GAA, GATA4, GATA6, GATA7, 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, 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, SCD1, SDHA, SDHAF1, SGCD, SGGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SURF1, TAZ, TBX1, TBX5, TCFAP, TGF2, TGF3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNM3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL
COMPREHENSIVE ARRHYTHMIAS PANEL Number of markers: 54 ARRHYTHMOGENIC 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 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: 43 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: 18 PULMONARY HYPERTENSION PANEL Number of markers: 10	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 ABCC9, ACTC1, ACTN2, AKAP9, ANK2, ANKRD1, BAG3, CACNA1C, CAV3, CAVIN4, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYAA, FKTN, FLNC, GATA7, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, OBSCN, PKP2, PLN, PRDM16, RAF1, RBM20, RYR2, SCNA5, SGCD, SLC22A5, TAZ, TCFAP, TMEM43, TNM3, 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, TNM3, TNNT2, TPM1, TTR, VCL ACTC1, DSP, DTNA, HCN4, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH7, PLEKHM2, PLN, PRDM16, RYR2, SCNA5, TAZ, TNM3, 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, TGF2, TGF3, TGFBR1, TGFBR2 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, RAS1, 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 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
<b>HEARING AND VISION LOSS</b> COMPREHENSIVE HEARING AND VISION LOSS PANEL Number of markers: 308 COMPREHENSIVE HEARING LOSS PANEL Number of markers: 92 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	Genes 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, CLN8, CLRN1, CNGA1, CNGB3, CNGB1, CNGB3, CNM4, COCH, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP11B1, CYP4V2, DHSDS, DIABLO, DIAPH1, DNAJC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, ENPM, ESRRB, EYA1, EYA4, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FRA1, FREM1, FREM2, FSD4, GNT2, GIPC3, GJA8, GJB2, GJB6, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GPSM2, GRHL2, GRIP1, GRK1, GRM6, GRXCRT1, GSDME, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HEXS1, HGF, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, ILDR1, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KARS, KCNJ13, KCNQ1, KOW4, KCNV2, KIF11, KIF7, KLHL7, LCA5, LHFP15, LHXAD1, LRAT, LRIT3, LRP5, LRTOMT, LYST, LZTF1, MAK, MARVELD2, MERTK, MFRP, MFSDB, MITF, MKKS, MKS1, MSRB3, MT-RNR1, MTFP, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, MYOC, NDR, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPB3, OPN1SW, OTOA, OTOF, OTOG, OTOLG, 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, RRGRIPI1, RRGRIPI1L, RSI, SAG, SDCCAG8, SEMA4A, SERPINB6, SIX1, SIX5, SLC24A1, SLC26A4, SLC45A2, SMOCT1, SMPX, SNRNP200, SOX10, SOX2, SPATA7, STRA6, STRC, TBC1D24, TCTN1, TCTN2, TCTN3, TDRD7, TECTA, TGFBI, TIMM8A, TIMP3, TMC1, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TMIE, TMPPRS3, TOPORS, TPP1, TPRN, TREX1, TRIM32, TRIOBP, TRPM1, TSPAN12, TSPAN12, TSPAR, TTC12B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423 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, GRXCRT1, GSDME, HARS, HGF, ILDR1, KARS, KCNQ1, KCNQ4, LHFP15, LHXAD1, LRTOMT, MARVELD2, MITF, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1, OTOA, OTOF, OTOG, OTOLG, P2RX2, PAX3, PCDH15, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PHYH, PJK, POU3F4, POU4F3, PRPS1, PTPRO, RDX, SERPINB6, SIX1, SIX5, SLC26A4, SMPX, SOX10, STRC, TBC1D24, TECTA, TIMM8A, TMC1, TMEM126A, TMIE, TMPPRS3, TPRN, TRIOBP, TSPAR, USH1C, USH1G, USH2A, WFS1, WHRN 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 ALBINISM / HERMANSKY-PUDLAK / WAARDENBURG PANEL Number of markers: 18 DEVELOPMENTAL EYE PANEL Number of markers: 21 RETINAL DISEASE PANEL Number of markers: 154 STICKLER AND CATARACT PANEL Number of markers: 41	ABCA4, ABHD12, ADAM9, ADGRV1, AH1, AIPL1, ALMS1, 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, CNGB3, CNGB1, CNGB3, CNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP11B1, CYP4V2, DHSDS, DNAJC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, EYA1, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FREM3, FSD4, GNT2, FCO11, FZD4, GCNT2, GJA8, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GRIP1, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HEXS1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KARS, KCNJ13, KCNV2, KIF11, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP5, LYST, LZTF1, MAK, MERTK, MFRP, MFSDB, MITF, MKKS, MKS1, MTFP, MYH9, MYO7A, MYOC, NDR, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPB3, OPN1SW, OTOX, OTX2, PANK2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PHYH, PITPNM3, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI1, RPI1L, RP2, RPE65, RPRG, RRGRIPI1, RRGRIPI1L, RSI, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SMOCT1, SNRNP200, SOX10, SOX2, SPATA7, STRA6, STRC, TBC1D24, TCTN1, TCTN2, TCTN3, TDRD7, TGFBI, TIMM8A, TIMP3, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423 BCOR, BMP4, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GRIP1, HCCS, KIF11, MFRP, NDR, OTX2, PAX6, PITX3, PLA2G5, PXDN, SMOCT1, SOX2, STRA6, VSX2 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, CDH3, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLRN1, CNGA1, CNGB3, CNGB1, CNGB3, CNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP11B1, CYP4V2, DHSDS, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FREM3, FSD4, GNT2, FCO11, FZD4, GCNT2, GJA8, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GRIP1, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HEXS1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KARS, KCNJ13, KCNV2, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP5, LYST, LZTF1, MAK, MERTK, MFRP, MKKS, MKS1, MYO7A, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, OFD1, OPA1, OPB3, 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, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI1, RPI1L, RP2, RPE65, RPRG, RRGRIPI1, RRGRIPI1L, RSI, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC45A2, SMOCT1, 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 ABHD12, AGK, BCOR, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRX, CRYAA, CRYBB1, CRYBB3, ERCC6, EYA1, FCO11, GCNT2, GJA8, GNPTG, HSF4, JAG1, KCNJ13, LRP5, MYH9, NDR, 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
<p>ABAT, ABCD1, ACSL4, ACY1, ADGRG1, ADGRV1, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ALDH7A1, ALG13, AMT, ANK3, ANKRD11, AP1S2, AP4B1, APAE1, AP4M1, AP4S1, ARFGF2, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6VOA2, ATP7A, ATP8A2, ATR, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, CA8, CACNA1A, CACNA1C, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D1A, CDC22, CDC38C, CDM2, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CHRNA2, CHRNA4, CHRNA7, CHRN2B, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, CPA6, CREBBP, CSTB, CTCF, CTNNB1, CTSO, CUL3, CUL4B, CYP27A1, D2HGDH, DCX, DDHD2, DDX3X, DEAF1, DEPPDC5, DHCRT7, DIS3L2, DKC1, DLG3, DMD, DNAJC5, DNMI1, DNMT3A, DOCK7, DPYD, DYNC1H1, DYRK1A, EBP, EEF1A2, EFHC1, EHM1T1, EIF2S3, ELP4, EMX2, EPMA2, EZH2, FGD1, FGF3, FKBP, 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, HNR1NP, HPR1, 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, KIF1B, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LAS1, LBR, LGI1, LIAS, LINS1, LRP2, MAGEL2, MAGI2, MAN1B1, MAOA, MAP2K1, MAP2K2, MAPK10, MBDS, MPTP2, MOPH1, MECP2, MED12, MED13L, MED23, MEFC2, MFS08, MID1, MTHFR, MTOR, MYT1L, NAA10, NAGLU, NDE1, NDP, NDUFA1, NECAP1, NEXMIF, NFIX, NFX, NHLRC1, NHR, NIPBL, NLGN4X, NOTCH3, NPRL2, NPRL3, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OGR1, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PDHA1, PGK1, PHF6, PHF8, PIGA, PIGN, PIGB, PIGL, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNP0, 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, ROGD1, RPL10, RPS6KA3, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, 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, SYNI1, SYNGAP1, SYP, SZT2, TAF1, TBC1D24, TBL1XR1, TBR1, TBX1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TRAPP3, 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</p>	<p>ABAT, ACY1, ADGRG1, ADGRV1, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP1S2, ARFGF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP2A2, ATP6AP2, ATP6VOA2, ATR, ATRX, BCKDK, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CDC21A, CDC22, CDC38C, CDM2, CDKL5, CHD2, CHD7, CHD8, CHRNA2, CHRNA4, CHRNA7, CHRN2B, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, CPA6, CREBBP, CSTB, CTCF, CTNNB1, CTSO, CUL3, CUL4B, CYP27A1, D2HGDH, DCX, DDHD2, DDX3X, DEAF1, DEPPDC5, DHCRT7, DIS3L2, DKC1, DLG3, DMD, DNAJC5, DNMI1, DNMT3A, DOCK7, DPYD, DYNC1H1, DYRK1A, EBP, EEF1A2, EFHC1, EHM1T1, EIF2S3, ELP4, EMX2, EPMA2, EZH2, FGD1, FGF3, FKBP, FLNA, FLVCR2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GLI2, GLI3, GNAO1, GNB1, GNS, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HCCS, HCN1, HCN4, HDAC8, HGSNAT, HIP1, HNR1NP, HPR1, 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, KIF1B, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LAS1, LBR, LGI1, LIAS, LINS1, LRP2, MAGEL2, MAGI2, MAN1B1, MAOA, MAP2K1, MAP2K2, MAPK10, MBDS, MPTP2, MOPH1, MECP2, MED12, MED13L, MED23, MEFC2, MFS08, MID1, MTHFR, MTOR, MYT1L, NAA10, NAGLU, NDE1, NDP, NDUFA1, NECAP1, NEXMIF, NFIX, NFX, NHLRC1, NHR, NIPBL, NLGN4X, NOTCH3, NPRL2, NPRL3, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OGR1, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PDHA1, PGK1, PHF6, PHF8, PIGA, PIGN, PIGB, PIGL, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNP0, 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, ROGD1, RPL10, RPS6KA3, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, SETBP1, SHANK3, 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, SYNI1, SYNGAP1, SYP, SZT2, TBC1D24, TBX1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TRAPP3, TRIO, TSC1, TSC2, TSPAN7, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1</p>
<p>Number of markers: 401</p>	
<p>COMPREHENSIVE EPILEPSY AND AUTISM PANEL</p>	
<p>Number of markers: 226</p>	
<p>COMPREHENSIVE EPILEPSY PANEL</p>	
<p>Number of markers: 226</p>	
<p>FOCAL, GENERALIZED, AND MYOCLONIC EPILEPSY PANEL</p>	
<p>Number of markers: 52</p>	
<p>INFANTILE EPILEPSY PANEL</p>	
<p>Number of markers: 58</p>	
<p>MIGRAINE PANEL</p>	
<p>Number of markers: 7</p>	
<p>NEURONAL CEROID LIPOFUSCINOSES PANEL</p>	
<p>Number of markers: 9</p>	
<p>NEURONAL MIGRATION PANEL</p>	
<p>Number of markers: 22</p>	
<p>SYNDROMIC EPILEPSY AND INTELLECTUAL DISABILITY PANEL</p>	
<p>Number of markers: 93</p>	
<p>COMPREHENSIVE AUTISM SPECTRUM DISORDER PANEL</p>	
<p>Number of markers: 228</p>	
<p>STAT AUTISM SPECTRUM DISORDER PANEL</p>	
<p>Number of markers: 30</p>	
<p>EPILEPSY PHARMACOGENETIC PANEL</p>	
<p>Number of markers: 10</p>	
<p>MICROCEPHALY PANEL</p>	
<p>Number of markers: 78</p>	
	<p>*Only copy number variants and targeted genotyping reported in these genes ** Only targeted genotyping reported in these genes.</p>
<p><b>PORPHYRIA</b></p>	<p><b>Genes</b></p>
<p>Acute Intermittent Porphyria (AIP)</p>	<p>HMBS</p>
<p>Acute Porphyrias Panel (AIP, HCP, &amp; VP)</p>	<p>HMBS, CPOX, PPOX</p>
<p>Number of markers: 3</p>	
<p>Congenital Erythropoietic Porphyria (CEP)</p>	<p>URO5, ALAS2 (exon 11)</p>
<p>Number of markers: 2</p>	
<p>Erythropoietic Protoporphyrin Panel</p>	<p>FECH, ALAS2 (Exon 11)</p>
<p>PEPT2 (SLC15A2) Genotyping for Acute Porphyria Patients</p>	<p>SLC15A2</p>
<p>Hereditary Coproporphyrin (HCP)</p>	<p>CPOX</p>
<p>Porphyria Cutanea Tarda (PCT)</p>	<p>UROD</p>
<p>Variegate Porphyria (VP)</p>	<p>PPOX</p>
<p><b>SKELETAL</b></p>	<p><b>Genes</b></p>
<p>CRANIOSYNOSTOSIS PANEL</p>	<p>ENFB1**, FGF1**, FGF2**, FGF3**, MSX2**, POR**, RAB23**, TWIST1**</p>
<p>Number of markers: 8</p>	
<p>LIMB DEFECTS PANEL</p>	<p>GLI3, HOXD13, ROR2, SALL1, SALL4, TBX5, WNT7A, ZRS</p>
<p>Number of markers: 8</p>	
	<p>**Only targeted genotyping reported in these genes</p>
<p><b>IMMUNODEFICIENCY</b></p>	<p><b>Genes</b></p>
<p>COMPREHENSIVE IMMUNODEFICIENCY PANEL</p>	<p>ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, B2M, BTK, C10A, C10B, C10C, 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, CE2PE, CFB, CFC, 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, EPICAM, EP65, FADD, FAS, FASLG, FCN1, FERMT1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA2, GATA3, GF1, HAX1, ICOS, IFI1, IFNGR1, IFNGR2, IGHM, IGLL1, IKKBK, IKKKB, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL18, IL18RAP, IL1RN, IL21, IL21R1, IL23R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF7, IRF8, IRF8, IRF8, ISG15, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, LAMTOR2, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYST, MAGT1, MALT1, MAP3K14, MASP2, MEV1, MRE11, MKV, MYD88, MYO5B, NBN, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRCA, NLRP12, NLRP3, NOD2, NOD10, NRAS, ORA1, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, PRF1, PRKCD, PRKDC, PTKEN, 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, SMARCA2, SP110, SPINK5, STAT1, STAT3, STAT5B, STIM1, STK4, STX11, STXB2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCN2, TERC, TERT, TINF2, TLR3, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF12, TREX1, TRIM22, TRN1, TTC37, TTC7A, TYK2, UNC13D, UNG, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24</p>
<p>Number of markers: 206</p>	
<p>PRIMARY IMMUNODEFICIENCY PANEL</p>	
<p>Number of markers: 206</p>	
<p>INFLAMMATORY BOWEL DISEASE PANEL</p>	
<p>Number of markers: 59</p>	
<p>SEVERE COMBINED IMMUNODEFICIENCY PANEL</p>	
<p>Number of markers: 26</p>	

## PANEL TABLE (continued)

METABOLIC DISORDERS SINGLE GENE DIAGNOSTIC TESTING	Genes
AMINOACIDOPATHIES AND UREA CYCLE DISORDERS	ACAT1, ADL1, 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, GLUT1, 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, MFSDB, 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
<b>ADDITIONAL GENES AVAILABLE FOR DIAGNOSTIC TESTING</b>	<b>Genes</b> 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, DYSLF, EDA, EFNB1, EIF2B5, ELP1, EMD, EPB42, ESCO2, EVC, FANCA, FANCC, FANCG, FBN1, GFM1, GJB1, GLE1, GP1BA, GP9, GRHRP, 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, PROX1, PTPRC, PUS1, RAG1, RAG2, RAPSIN, RMRP, RTEL1, SACS, SEPSCE3, SGCA, SGCB, SGGC, 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

