CHILD NEUROLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
PerkinElmer Genomics	Lantern Project	Fabry disease Pompe disease Gaucher disease Niemann-Pick Type A and B (*ASMD*) Mucopolysaccharidosis I (MPS I) and Other MPS Disorders Focused Neuromuscular Disease Panel	hypotonia, concerns for muscular dystrophy, or clinical suspicion of any of the conditions on the panels. This project includes multiple panel options for molecular and/or biochemical testing (MPS enzyme panel, Lyso-GI1, Lyso-GL3, alpha-iduronidase, and acid alpha-glucosidase).	The Lantern Project » PerkinElmer Genomics
PerkinElmer Genomics	DeCode Duchenne	Duchnenne and Becker muscular dystrophy (*DMD*)	Suspicion for Duchenne or Becker Muscular Dystrophy; carrier testing for approved family members (see application for carrier testing here: https://www.parentprojectmd.org/about-duchenne/decode-duchenne/carrier-testing/)	DeCode Duchenne » PerkinElmer Genomics
Invitae	Detect Muscular Dystrophy	Invitae Dystrophinopathies Test Invitae Limb-Girdle Muscular Dystrophy Panel Invitae Comprehensive Muscular Dystrophy Panel Invitae Comprehensive Neuromuscular Disorders Panel	individuals suspected of having a muscular dystrophy with one or more of the following: progressive muscle weakness, elevated CK levels, presumptive positive DMD from NBS, cardiac or respiratory involvement, calf hypertrophy or pseudohypertrophy, muscle biopsy showing dystrophic changes and/or immunohistochemical evidence for specific muscular dystrophy subtype, family history of muscular dystrophy* *Please note this program is not intended for carrier screening of unaffected	Detect Muscular Dystrophy
Invitae	SMA Identified	Invitae Spinal Muscular Atrophy Panel Invitae Spinal Muscular Atrophy STAT Panel Invitae SMA Carrier Screen	individuals with a suspected diagnosis of, or family history of, SMA	SMA Identified
PerkinElmer Genomics	ACDase	*ASAH1* gene testing	suspicion for ASAH1 related disorder	ASAH1 Gene Testing for ACDase » PerkinElmer Genomics
PerkinElmer Genomics	Ultragenyx MPS panel	Ultragenyx MPS panel	suspicion for MPS	Ultragenyx-Sponsored MPS Panel Testing » PerkinElmer Genomics
Invitae	Detect Lysosomal Storage Diseases	Invitae Comprehensive LSD Panel Invitae Mucopolysaccharidoses Plus (MPS+) Panel Invitae Cardiomyopathy Comprehensive Panel Invitae Comprehensive Neuromuscular Disorders Panel + a number of single genes tests for specific LSDs	individuals must be suspected of having an LSD based on at least one of the following: clinical features; suspicion of, or known diagnosis of, a specific lysosomal storage disease; family history related to LSDs; lab results suggestive of LSDs; presumptive positive NBS	Detect Lysosomal Storage Diseases
PreventionGenetics	Fabry Disease Diagnostic Testing	Fabry disease	individuals who have health issues seen more often in Fabry disease and/or have a known family history of Fabry disease	AAKP Fabry Disease
Invitae	Behind the Seizure	Invitae Epilepsy Panel	any child up to 8 years of age (96 months) who has had an unprovoked seizure	Behind the Seizure
Invitae	Alnylam Act Acute Hepatic Porphyria	Invitae Comprehensive Porphyrias Panel	individuals must be pubescent or older and meet one of the following criteria: • family history of acute hepatic porphyria • elevated urinary porphobilinogen (PBG) or aminolaevulinic acid (ALA) levels • unexplained recurrent, prolonged (>24 hours) episodes of severe, diffuse (poorly localized) abdominal pain AND at least two additional criteria (see webpage):	Alnylam Act Acute Hepatic Porphyria
Invitae	Long-Chain Fatty Acid Oxidation Disorders	Invitae Fatty Acid Oxidation Defects Panel	individuals who meet at least one of the following criteria: has a completed UltraCare Start Form for LC-FAOD, or is suspected of having or has been diagnosed with a LC-FAOD and a plasma acylcarnitine test (regardless of result) has been performed or ordered	Long-Chain Fatty Acid Oxidation Disorders
Invitae	PTC Pinpoint Neurotransmitter Disorders	Invitae Neurotransmitter Disorders Panel	individuals suspected of having, or have clinical symptoms consistent with, a neurotransmitter disorder	PTC Pinpoint Neurotransmitter Disorders
Invitae	PTC Pinpoint CP Spectrum Disorders	Invitae Cerebral Palsy Spectrum Disorders Panel	individuals with symptoms suggestive of cerebral palsy in the absence of risk factors for an acquired brain injury	PTC Pinpoint CP Spectrum Disorders
Invitae	UCD Genetic Testing Program	Invitae Hyperammonemia Panel	individuals who meet at least one of the following criteria: • a suspected diagnosis of a urea cycle disorder, OR • a family history of a urea cycle disorder	UCD Genetic Testing Program
Invitae	Leukodystrophies	The Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	symptomatic or asymptomatic individuals with a clinical diagnosis or suspicion of leukodystrophy, genetic leukoencephalopathy, and/or a family history of either	Leukodystrophies
PreventionGenetics	X-linked adrenoleukodystrophy NBS Reflex	26-gene Panel (see webpage)	individuals who meet one of the criteria below: • testing will be offered to children who screened positive for X-ALD on initial state NBS but negative for the disease after further laboratory testing • testing will be offered to a blood relative (sibling, cousin) of a PBD-ZSD positive patient who was tested at PreventionGenetics	X-linked adrenoleukodystrophy (X-ALD) newborn screening reflex test
Invitae	Think Arginine	Invitae Comprehensive Hereditary Spastic Paraplegia Panel Invitae Epilepsy Panel Invitae Cerebral Palsy Spectrum Disorders Panel Invitae Hyperammonemia Panel Invitae Elevated Arginine (Arginase deficiency) Panel Invitae Urea Cycle Disorders Panel Invitae Treatable Neurometabolic Disorders Panel	individuals 40 years or younger with minimum plasma arginine of 115 µM (record value), AND one of the following: diagnosis of HSP, spasticity, or global developmental delay	Think Arginine
PreventionGenetics	Thymidine Kinase 2 Deficiency	Thymidine kinase 2 deficiency	individuals with a suspected or clinical diagnosis of thymidine kinase 2 deficiency (TK2d)	Thymidine Kinase 2 Deficiency
PreventionGenetics	Peroxisomal biogenesis disorder-Zellweger spectrum disorder (PBD-ZS) Test Program	13-gene Panel (see webpage)	individuals with clinical symptoms suggestive of a peroxisomal disorder; patients must meet ONE of the criteria below: • diagnosed PBD-ZSD • clinical suspicion of PBD-ZSD (e.g. neurological, vision, hearing, hepatic deterioration)	Peroxisomal biogenesis disorder-Zellweger spectrum disorder (PBD-ZSD)
CARDIOLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	Alnylam Act hATTR	Invitae Transthyretin-mediate Amyloidosis Test Invite Cardiomyopathy Comprehensive Panel Invitae Comprehensive Neuropathies Panel	individuals 18 years of age and older with a suspected diagnosis or a confirmed family history of hATTR amyloidosis	Alnylam Act hATTR
Invitae	Detect Cardiomyopathy & Arrhythmia	Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	individuals suspected of having a familial cardiomyopathy or arrhythmia	Detect Cardiomyopathy and Arrhythmia
Invitae	Detect Lysosomal Storage Diseases	Invitae Comprehensive LSD Panel Invitae Mucopolysaccharidoses Plus (MPS+) Panel Invitae Cardiomyopathy Comprehensive Panel Invitae Comprehensive Neuromuscular Disorders Panel + a number of single genes tests for specific LSDs	individuals must be suspected of having an LSD based on at least one of the following: clinical features; suspicion of, or known diagnosis of, a specific lysosomal storage disease; family history related to LSDs; lab results suggestive of LSDs; presumptive positive NBS	Detect Lysosomal Storage Diseases
PreventionGenetics	Familial Chylomicronemia Syndrome	*APOA5, APOC2, GPD1, GPIHBP1, LMF1, LPL*	individuals with a clinical diagnosis of FCS who meet testing eligibility criteria: minimum of 2 consecutive fasting triglycerides levels ≥750 mg/DL or 8.4 mmol/L in the absence of secondary causes or medical conditions known to cause hypertriglyceridemia (HTG)	Familial Chylomicronemia Syndrome (FCS)
IMMUNOLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	NavigateAPDS	Invitae Primary Immunodeficiency Panel	individuals who meet any 2 or more of a list of bulleted criteria (see webpage)	NavigateAPDS
Invitae	PATH4WARD	Invitae Primary Immunodeficiency Panel	individuals with a suspicion of congenital neutropenia AND neutropenia (not drug related or secondary to a viral infection) at any point in life with ANC ≤750/uL	PATH4WARD
NEPHROLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	KIDNEYCODE	Invitae Progressive Renal Disease Panel	individuals with at least one of the following: • suspected or biopsy-confirmed diagnosis of Alport syndrome or FSGS • family member with a suspected or biopsy-confirmed diagnosis of Alport or FSGS; or • eGFR ≤90mL/min/1.73m2 and either hematuria or family history of kidney disease	KIDNEYCODE
Invitae	Hypophosphatemia	Invitae Hypophosphatemia Panel	individuals must be aged 6 months or older AND meet one of the following criteria: • has completed the UltraCare Start Form for XLH, or • has a previous diagnosis related to hypophosphatemia, or • has a family member with a confirmed XLH diagnosis, or • exhibits TWO or more of the following clinical signs and/or s symptoms (see webpage)	Hypophosphatemia

Invitae	Alnylam Act Primary Hyperoxaluria Type 1	Invitae Primary Hyperoxaluria Panel Invitae Nephrolithiasis Panel	individuals must have a family history or suspected diagnosis of primary hyperoxaluria with one or more of the following symptoms: • adult (18 years or older) with either elevated urinary oxalate OR plasma oxalate • child with one of the following: failure to thrive AND impaired kidney function, nephrolithiasis, nephrocalcinosis, elevated urinary oxalate OR plasma oxalate	Alnylam Act Primary Hyperoxaluria Type 1
OPHTHALMOLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	ID Your IRD	Invitae Inherited Retinal Disorders Panel	individuals suspected of having an inherited retinal disorder and who have experienced one or more of the following: peripheral field loss, nyctalopia, deterioration in color vision, central vision loss, photophobia, any of the following	ID Your IRD
PreventionGenetics	Early-Onset Bilateral Cataracts	66-gene Panel (see webpage)	individuals age of 18 months to 35 year, and patient has current or history of idiopathic bilateral cataracts (e.g. not known to be due to infectious causes, trauma,	Early-Onset Bilateral Cataracts
ENDOCRINOLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	Discover Dysplasias	Invitae Skeletal Disorders Panel	individuals suspected of having a skeletal dysplasia; to be eligible, patients must have at least one of the following: skeletal abnormalities suggestive of skeletal dysplasia, short stature, disproportionate growth, dysmorphic facial features, other	Discover Dysplasia
PreventionGenetics	Familial Chylomicronemia Syndrome	*APOA5, APOC2, GPD1, GPIHBP1, LMF1, LPL*	individuals with a clinical diagnosis of FCS who meet testing eligibility criteria: minimum of 2 consecutive fasting triglycerides levels ≥750 mg/DL or 8.4 mmol/L in the absence of secondary causes or medical conditions known to cause	Familial Chylomicronemia Syndrome (FCS)
Invitae	Hypoparathyroidism	The Invitae Hypoparathyroidism and Hyperparathyroidism Panel	individuals with an established diagnosis of non-surgical hypoparathyroidism or who have a first-degree relative with an established diagnosis of genetic hypoparathyroidism	Hypoparathyroidism
PreventionGenetics	Rare Genetic Disorders of Obesity	79-gene Panel (see webpage)	individuals with early onset non-syndromic obesity or individuals suspected to have a syndrome with obesity as a predominant feature	Rare Genetic Disorders of Obesity
HEMATOLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
PerkinElmer Genomics	AnemiaID	AnemiaID panel	anemia or suspicion for hereditary anemia, suspicion for any gene disorder on the panel	» PerkinElmer Genomics
MISCELLANEOUS				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
PreventionGenetics	Rare Calcification Disorders	*ENPP1* or *ABCC6*	individuals must meet eligibility criteria (available on the informed consent form)	Rare Calcification Disorders
Invitae	Amplify	The Invitae Comprehensive Deafness Panel	individuals less than 5 years of age with all of the following: • absent or highly abnormal auditory brainstem response in both ears • presence of distortion product otoacoustic emissions in ≥ 3 frequences in at least	Amplify
PreventionGenetics	Cholestasis	77-gene Panel (see webpage)	individuals must meet ONE of the criteria below: • patient is cholestatic, or has a history of cholestasis, without an identified cause • unexplained chronic liver disease	Cholestasis
ADULT NEUROLOGY				
LAB	**PROGRAM NAME**	**TEST / PANEL NAME**	**ELIGIBILITY CRITERIA**	**LINK TO TEST INFORMATION**
Invitae	Adult Neurodegenerative Disorders	Invitae Hereditary ALS, FTD, and Alzheimer Disease Panel Invitae Hereditary Parkinson Disease and Parkinsonism Panel	individuals 18 years or older with a clinical diagnosis or suspicion of one of the following conditions: ALS, Parkinson disease, FTD, Alzheimer disease with onset <65 years of age, hereditary prion disease; AND asymptomatic individuals with either a family history of early (<65 years of age) onset diagnosis of one of the conditions above, or a family member with a known disease-causing variant in one of the	Adult Neurodegenerative Disorders
Invitae	Uncovering Periodic Paralysis	Invitae Periodic Paralysis Panel	individuals 18 years of age and older; and episodic muscle weakness/paralysis attacks or episodic pain after attacks; and episodes are provoked by at least one of the common triggers for hyperkalemic or hypokalemic primary periodic paralysis	Uncovering Periodic Paralysis
Invitae	ALS Identified	Invitae ALS with C9orf72 Panel	individuals 18 years of age or older with a diagnosis of ALS or a family history of ALS	ALS Identified
Invitae	Detect Muscular Dystrophy	Invitae Dystrophinopathies Test Invitae Limb-Girdle Muscular Dystrophy Panel Invitae Comprehensive Muscular Dystrophy Panel Invitae Comprehensive Neuromuscular Disorders Panel	individuals suspected of having a muscular dystrophy with one or more of the following: progressive muscle weakness, elevated CK levels, presumptive positive DMD from NBS, cardiac or respiratory involvement, calf hypertrophy or pseudohypertrophy, muscle biopsy showing dystrophic changes and/or immunohistochemical evidence for specific muscular dystrophy subtype, family history of muscular dystrophy* *Please note this program is not intended for carrier screening of unaffected	Detect Muscular Dystrophy
PerkinElmer Genomics	Lantern Project	Fabry disease Pompe disease Gaucher disease Niemann-Pick Type A and B (*ASMD*) Mucopolysaccharidosis I (MPS I) and Other MPS Disorders Focused Neuromuscular Disease Panel	hypotonia, concerns for muscular dystrophy, or clinical suspicion of any of the conditions on the panels. This project includes multiple panel options for molecular and/or biochemical testing (MPS enzyme panel, Lyso-GI1, Lyso-GL3, alpha-iduronidase, and acid alpha-glucosidase).	The Lantern Project » PerkinElmer Genomics
PerkinElmer Genomics	DeCode Duchenne	Duchnenne and Becker muscular dystrophy (*DMD*)	Suspicion for Duchenne or Becker Muscular Dystrophy, carrier testing for approved family members (see application for carrier testing here: https://www.parentprojectmd.org/about-duchenne/decode-duchenne/carrier-	DeCode Duchenne » PerkinElmer Genomics
Invitae	Leukodystrophies	The Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	symptomatic or asymptomatic individuals with a clinical diagnosis or suspicion of leukodystrophy, genetic leukoencephalopathy, and/or a family history of either	Leukodystrophies
Invitae	Alnylam Act Acute Hepatic Porphyria	Invitae Comprehensive Porphyrias Panel	individuals must be pubescent or older and meet one of the following criteria: • family history of acute hepatic porphyria • elevated urinary porphobilinogen (PBG) or aminolaevulinic acid (ALA) levels • unexplained recurrent, prolonged (>24 hours) episodes of severe, diffuse (poorly localized) abdominal pain AND at least two additional criteria (see webpage):	Alnylam Act Acute Hepatic Porphyria
PreventionGenetics	Wilson Disease Testing Program	*ATP7B* gene testing	individuals with a clinical diagnosis of Wilson disease who meet eligibility criteria: patients cannot have had previous ATP7B genetic testing, unless previous testing only found 1 variant, or was negative; and patients must have 2 of the following	Wilson Disease
Invitae	PTC Pinpoint CP Spectrum Disorders	Invitae Cerebral Palsy Spectrum Disorders Panel	individuals with symptoms suggestive of cerebral palsy in the absence of risk factors for an acquired brain injury	PTC Pinpoint CP Spectrum Disorders
Invitae	SMA Identified	Invitae Spinal Muscular Atrophy Panel Invitae Spinal Muscular Atrophy STAT Panel Invitae SMA Carrier Screen	individuals with a suspected diagnosis of, or family history of, SMA	SMA Identified