

# The Invitae Test Catalog



## CARDIOLOGY

### ARRHYTHMIA AND CARDIOMYOPATHY

Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel

### ARRHYTHMIA

Invitae Arrhythmia Comprehensive Panel  
Invitae Arrhythmogenic Cardiomyopathy Panel  
Invitae Brugada Syndrome Panel  
Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel  
Invitae Long QT Syndrome Panel  
Invitae Short QT Syndrome Panel

### CARDIOMYOPATHY

Invitae Cardiomyopathy Comprehensive Panel  
Invitae Arrhythmogenic Cardiomyopathy Panel  
Invitae Dilated Cardiomyopathy Panel  
Invitae Hereditary Hemochromatosis Panel  
Invitae Hypertrophic Cardiomyopathy Panel  
Invitae Left Ventricular Noncompaction Panel  
Invitae RASopathies Comprehensive Panel  
Invitae Transthyretin Amyloidosis Test

### CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

Invitae Cardiomyopathy and Skeletal Muscle Disease Panel

### AORTOPATHY AND CONNECTIVE TISSUE DISORDERS

Invitae Aortopathy Comprehensive Panel  
Invitae Ehlers-Danlos Syndrome Panel  
Invitae Loeys-Dietz Syndrome Panel  
Invitae Marfan Syndrome Test

### FAMILIAL HYPERCHOLESTEROLEMIA

Invitae Familial Hypercholesterolemia Panel

### PULMONARY HYPERTENSION

Invitae Pulmonary Arterial Hypertension Panel  
Invitae Hereditary Hemorrhagic Telangiectasia Panel  
Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test

### CONGENITAL HEART DISEASE

Invitae Congenital Heart Disease Panel  
Invitae CHARGE Syndrome Test  
Invitae Holt-Oram Syndrome Test  
Invitae RASopathies Comprehensive Panel  
Invitae Sotos Syndrome Test



## DERMATOLOGY

### DERMATOLOGY-RELATED CANCER SYNDROMES

Invitae Basal Cell Nevus Syndrome Panel  
Invitae Birt-Hogg-Dubé Syndrome Test  
Invitae Lynch Syndrome Panel  
Invitae Melanoma Panel  
Invitae Melanoma-Pancreatic Cancer Syndrome Panel  
Invitae Neurofibromatosis Type 2 Test  
Invitae Schwannomatosis Test  
Invitae Tuberous Sclerosis Complex Panel

### SKIN DISORDERS

Invitae Cardio-Facio-Cutaneous Syndrome Panel  
Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel  
Invitae Legius Syndrome Test  
Invitae Noonan Syndrome with Multiple Lentigines Panel  
Invitae PTEN-Related Disorders Test  
Invitae TP63-Related Disorders Test  
Invitae van der Woude Syndrome Panel



## HEMATOLOGY

### BONE MARROW FAILURE SYNDROMES

Invitae Bone Marrow Failure Syndromes Panel  
Invitae Congenital Amegakaryocytic Thrombocytopenia Test  
Invitae Diamond-Blackfan Anemia Panel  
Invitae Dyskeratosis Congenita Panel  
Invitae ELANE-Related Neutropenia Test  
Invitae Fanconi Anemia Panel  
Invitae GATA1-Related X-Linked Cytopenia Test  
Invitae GATA2 Deficiency Test  
Invitae WAS-Related Disorders Test

### HEREDITARY HEMOCHROMATOSIS

Invitae Hereditary Hemochromatosis Panel

### HEREDITARY THROMBOPHILIA

Invitae Hereditary Thrombophilia Panel  
Invitae Antithrombin III Deficiency Test  
Invitae Protein C Deficiency Test  
Invitae Protein S Deficiency Test



## CROSS-CANCER

- Invitae Multi-Cancer Panel
- Invitae Common Hereditary Cancers Panel

## STAT TURNAROUND TIME

- Invitae Breast Cancer STAT Panel
- Invitae BRCA1 and BRCA2 STAT Panel

## BREAST CANCER

- Invitae Breast Cancer Guidelines-Based Panel
- Invitae Breast Cancer Panel

## BREAST AND GYNECOLOGIC CANCERS

- Invitae Breast and Gyn Cancers Guidelines-Based Panel
- Invitae Breast and Gyn Cancers Panel

## COLORECTAL CANCER

- Invitae Colorectal Cancer Guidelines-Based Panel
- Invitae Colorectal Cancer Panel

## ADDITIONAL ORGAN SYSTEMS

- Invitae Gastric Cancer Panel
- Invitae Hereditary Paraganglioma-Pheochromocytoma Panel
- Invitae Hyperparathyroidism Panel
- Invitae Melanoma Panel
- Invitae Myelodysplastic Syndrome/Leukemia Panel
- Invitae Nervous System/Brain Cancer Panel
- Invitae Pancreatic Cancer Panel
- Invitae Prostate Cancer Panel
- Invitae Renal/Urinary Tract Cancers Panel
- Invitae Sarcoma Panel
- Invitae Thyroid Cancer Panel

## PEDIATRIC ONCOLOGY

- Invitae Pediatric Solid Tumors Panel
- Invitae Pediatric Hematologic Malignancies Panel
- Invitae Pediatric Nervous System/Brain Tumors Panel

## INDIVIDUAL HEREDITARY CANCER CONDITIONS

- Invitae Ataxia-Telangiectasia Test
- Invitae BAP1 Hereditary Cancer Predisposition Syndrome Test
- Invitae Basal Cell Nevus Syndrome Test
- Invitae Birt-Hogg-Dubé Syndrome Test
- Invitae Bloom Syndrome Test
- Invitae Carney Complex Test
- Invitae CASR-Related Conditions Test
- Invitae CDC73-Related Conditions Test
- Invitae Chronic Pancreatitis Panel
- Invitae Constitutional Mismatch Repair-Deficiency Panel
- Invitae Costello Syndrome Test
- Invitae DICER1 Syndrome Test
- Invitae Familial Acute Myeloid Leukemia with Mutated CEBPA Test
- Invitae Familial Gastrointestinal Stromal Tumor Syndrome Panel
- Invitae Familial Neuroblastoma Panel
- Invitae Familial Platelet Disorder with Propensity to Myeloid Malignancy Test
- Invitae Fanconi Anemia Panel
- Invitae GATA2 Deficiency Test
- Invitae Hereditary Breast and Ovarian Cancer Syndrome Panel
- Invitae Hereditary Diffuse Gastric Cancer Syndrome Test
- Invitae Hereditary Leiomyomatosis and Renal Cell Cancer Test
- Invitae Hereditary Papillary Renal Cell Carcinoma Test
- Invitae Hereditary Polyposis Panel
- Invitae Juvenile Polyposis Syndrome Panel
- Invitae Li-Fraumeni Syndrome Test
- Invitae Lynch Syndrome Panel
- Invitae Melanoma-Pancreatic Cancer Syndrome Panel
- Invitae Multiple Endocrine Neoplasia Type 1 Test
- Invitae Multiple Endocrine Neoplasia Type 2 Test
- Invitae Neurofibromatosis Type 1 Test
- Invitae Neurofibromatosis Type 2 Test
- Invitae Nijmegen Breakage Syndrome Test
- Invitae Oligodontia-Colorectal Cancer Syndrome Test
- Invitae Perlman Syndrome Test
- Invitae Peutz-Jeghers Syndrome Test
- Invitae PTEN-Related Disorders Test
- Invitae RECQL4-Related Disorders Test
- Invitae Retinoblastoma Test
- Invitae Rhabdoid Tumor Predisposition Syndrome Panel
- Invitae Schwannomatosis Test
- Invitae Simpson-Golabi-Behmel Syndrome Test
- Invitae Small Cell Carcinoma of the Ovary Hypercalcemic Type Test
- Invitae Tuberous Sclerosis Complex Panel
- Invitae von Hippel-Lindau Syndrome Test
- Invitae Weaver Syndrome Test
- Invitae Werner Syndrome Test
- Invitae Wilms Tumor Panel
- Invitae WT1-Related Disorders Test



### PRIMARY IMMUNODEFICIENCY

Invitae Primary Immunodeficiency Panel

### ANTIBODY DEFICIENCIES

Invitae Agammaglobulinemia Panel

Invitae Common Variable Immunodeficiency Panel

Invitae Hyper IgE Syndrome Panel

Invitae Hyper IgM Syndrome Panel

### AUTOINFLAMMATORY SYNDROMES

Invitae Autoinflammatory Syndromes Panel

Invitae Familial Cold Autoinflammatory Syndrome Panel

Invitae Familial Mediterranean Fever Test

Invitae Monogenic Inflammatory Bowel Disease Panel

Invitae Periodic Fever Syndromes Panel

### COMBINED T/B CELL DEFICIENCIES

Invitae Comprehensive Severe Combined Immunodeficiency (SCID) and Combined Immunodeficiency (CID) Panel

Invitae Comprehensive Severe Combined Immunodeficiency (SCID) Panel

Invitae T-B-NK- Severe Combined Immunodeficiency (SCID) Panel

Invitae T-B-NK+ Severe Combined Immunodeficiency (SCID) Panel

Invitae T-B+NK- Severe Combined Immunodeficiency (SCID) Panel

Invitae T-B+NK+ Severe Combined Immunodeficiency (SCID) Panel

Invitae X-Linked Severe Combined Immunodeficiency (SCID) Test

Invitae Radiation-Sensitive Severe Combined Immunodeficiency (SCID) Panel

Invitae Combined Immunodeficiency (CID) Panel

Invitae Syndromic Combined Immunodeficiency (CID) Panel

### DISORDERS OF INTRINSIC AND INNATE IMMUNITY

Invitae Chronic Mucocutaneous Candidiasis Panel

Invitae Epidermodysplasia Verruciformis Panel

Invitae Herpes Simplex Encephalitis Panel

Invitae Mendelian Susceptibility to Mycobacterial Disease Panel

### IMMUNE DYSREGULATION

Invitae Monogenic Autoimmunity Panel

Invitae Autoimmune Lymphoproliferative Disorders (ALPS) Panel

Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Panel

### PHAGOCYtic DEFECTS

Invitae Phagocyte Defects Panel

### WELL-DEFINED SYNDROMES

Invitae Dyskeratosis Congenita Panel

Invitae Immunodeficiency, Centromeric Instability, Facial Anomalies Syndrome Panel



### METABOLIC DISORDERS NEWBORN SCREENING CONFIRMATION

Invitae Metabolic Disorders Newborn Screening Confirmation Panel

Invitae Lysosomal Storage Disorders Newborn Screening Panel

Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test

### PANELS BY ANALYTE

Invitae Low C0 Test

Invitae Elevated C0/(C16+C18) Test

Invitae Elevated C3 Panel

Invitae Elevated C3-DC Test

Invitae Elevated C4 Panel

Invitae Elevated C4-OH Test

Invitae Elevated C4 & C5 Panel

Invitae Elevated C5 Panel

Invitae Elevated C5-DC Test

Invitae Elevated C5-OH Panel

Invitae Elevated C6, C8 & C10 Test

Invitae Elevated C14 & C14:1 Test

Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel

Invitae Elevated C16, C16:1, C18, & C18:1 Panel

Invitae Elevated Arginine Test

Invitae Elevated Citrulline Panel

Invitae Low Citrulline Panel

Invitae Elevated Glycine Panel

Invitae Elevated Leucine Panel

Invitae Elevated Methionine Panel

Invitae Elevated Phenylalanine Panel

Invitae Elevated Proline Panel

Invitae Elevated Succinylacetone Test

Invitae Elevated Tyrosine Panel

### AMINOACIDOPATHIES

Invitae Alkaptonuria Test

Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel

Invitae Cystinuria Panel

Invitae Disorders of Serine Biosynthesis Panel

Invitae Glycine Encephalopathy Panel

Invitae Homocystinuria Panel

Invitae Hyperphenylalaninemia Panel

Invitae Hyperprolinemia Panel

Invitae Maple Syrup Urine Disease Panel

Invitae Tyrosinemia Panel

*More metabolic disorders & newborn screening tests on next page.*



## CARBOHYDRATE DISORDERS

- Invitae Galactosemia Panel
- Invitae Glucose-6-Phosphate Dehydrogenase Deficiency Test
- Invitae Glucose Transporter Type 1 Deficiency Syndrome Test
- Invitae Comprehensive Glycogen Storage Disease Panel
- Invitae Liver Glycogen Storage Disease Panel
- Invitae Muscle Glycogen Storage Disease Panel
- Invitae Hereditary Fructose Intolerance Test
- Invitae Rare Carbohydrate Disorders Panel

## CEREBROTENDINOUS XANTHOMATOSIS

- Invitae Cerebrotendinous Xanthomatosis Test

## CONGENITAL DISORDERS OF GLYCOSYLATION

- Invitae Congenital Disorders of Glycosylation Panel

## CREATINE BIOSYNTHESIS AND TRANSPORT DISORDERS

- Invitae Cerebral Creatine Deficiency Panel

## CYSTIC FIBROSIS

- Invitae Cystic Fibrosis Newborn Screening Confirmation Test

## FATTY ACID OXIDATION DEFECTS

- Invitae Fatty Acid Oxidation Defects Panel
- Invitae Ketogenesis Disorders Panel
- Invitae Ketolysis Disorders Panel
- Invitae Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) Test
- Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel
- Invitae Very Long Chain Acyl-CoA Dehydrogenase Deficiency Test

## LYSOSOMAL STORAGE DISORDERS

- Invitae Comprehensive Lysosomal Storage Disorders Panel
- Invitae Cystinosis Test
- Invitae Farber Lipogranulomatosis Test
- Invitae Fabry Disease Test
- Invitae GM2 Gangliosidosis Panel
- Invitae Krabbe Disease Test
- Invitae Lysosomal Acid Lipase Deficiency Test
- Invitae Metachromatic Leukodystrophy Panel
- Invitae Mucopolipidosis Panel
- Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel
- Invitae Mucopolysaccharidosis Type I (MPS I) Test
- Invitae Mucopolysaccharidosis Type II (MPS II) Test
- Invitae Mucopolysaccharidosis Type III (MPS III) Panel
- Invitae Mucopolysaccharidosis Type IV (MPS IV) Panel
- Invitae Multiple Sulfatase Deficiency Test
- Invitae Comprehensive Neuronal Ceroid Lipofuscinoses Panel
- Invitae Niemann-Pick Disease Types A and B Test
- Invitae Niemann-Pick Disease Type C Panel
- Invitae Oligosaccharidoses Panel
- Invitae Pompe Disease Test
- Invitae Prosaposin Deficiency Test
- Invitae Sandhoff Disease Test
- Invitae Tay-Sachs Disease Test

## METAL TRANSPORT DISORDERS

- Invitae ATP7A-Related Disorders Test
- Invitae Copper Metabolism Disorders Panel
- Invitae Wilson Disease Test

## NEUROTRANSMITTER DISORDERS

- Invitae Neurotransmitter Disorders Panel
- Invitae Hereditary Hyperekplexia Panel

## ORGANIC ACIDEMIAS

- Invitae Organic Acidemias Panel
- Invitae 2-Hydroxyglutaric Aciduria Panel
- Invitae 3-Methylcrotonyl CoA Carboxylase Panel
- Invitae 3-Methylglutaconic Aciduria Panel
- Invitae Barth Syndrome Test
- Invitae Biotinidase Deficiency Test
- Invitae Canavan Disease Test
- Invitae Glutaric Acidemia Type I Test
- Invitae Combined Methylmalonic Acidemia and Homocystinuria Panel
- Invitae Methylmalonic Acidemia Panel
- Invitae Multiple Acyl-CoA Dehydrogenase (MAD) Deficiency Panel
- Invitae Multiple Carboxylase Deficiency Panel
- Invitae Propionic Acidemia Panel

## PEROXISOMAL DISORDERS

- Invitae Adult Refsum Disease Panel
- Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Panel
- Invitae X-linked Adrenoleukodystrophy Test
- Invitae Zellweger Spectrum Disorder Panel

## PORPHYRIAS

- Invitae Acute Hepatic Porphyrins Panel

## PURINE METABOLISM DISORDERS

- Invitae Purine Metabolism Disorders Panel
- Invitae Lesch-Nyhan Syndrome Test

## PYRUVATE METABOLISM AND TRICARBOXYLIC ACID CYCLE DEFECTS

- Invitae 2-Ketoglutarate Dehydrogenase Deficiency Panel
- Invitae Citrate Transporter Deficiency Test
- Invitae Dihydroipoamide Dehydrogenase Deficiency Test
- Invitae Fumarase Deficiency Test
- Invitae Pyruvate Carboxylase Deficiency Test
- Invitae Pyruvate Dehydrogenase Deficiency Panel

## TREATABLE DISORDERS

- Invitae Treatable Neurometabolic Disorders Panel
- Invitae Biotin-Thiamine-Responsive Basal Ganglia Disease (BTBGD) Test

## UREA CYCLE DISORDERS

- Invitae Urea Cycle Disorders Panel
- Invitae Arginase Deficiency Test
- Invitae Ornithine Transcarbamylase (OTC) Deficiency Test



## MOVEMENT DISORDERS

- Invitae Dystonia Comprehensive Panel
- Invitae Hereditary Parkinson's Disease & Parkinsonism Panel

## NEURODEGENERATIVE DISORDERS

- Invitae Combined Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel
- Invitae Amyotrophic Lateral Sclerosis Panel
- Invitae Frontotemporal Dementia Panel
- Invitae Hereditary Alzheimer's Disease Panel
- Invitae Hereditary Parkinson's Disease & Parkinsonism Panel
- Invitae Hereditary Prion Disease Test

## NEUROMUSCULAR DISORDERS

- Invitae Comprehensive Neuromuscular Disorders Panel
- Invitae Congenital Myasthenic Syndrome Panel
- Invitae Malignant Hyperthermia Susceptibility Panel
- Invitae Comprehensive Muscular Dystrophy Panel
- Invitae Congenital Muscular Dystrophy Panel
- Invitae Dystroglycanopathy Panel
- Invitae Dystrophinopathies Test
- Invitae Emery-Dreifuss Muscular Dystrophy Panel
- Invitae Limb-Girdle Muscular Dystrophy Panel
- Invitae Comprehensive Myopathy Panel
- Invitae Congenital Myopathy Panel
- Invitae Autophagic Vacuolar Myopathy Panel
- Invitae Central Core Disease Test
- Invitae Centronuclear Myopathy Panel
- Invitae Congenital Fiber-Type Disproportion Panel
- Invitae Distal Myopathy Panel
- Invitae Inclusion Body Myopathy Panel
- Invitae Multiminicore Disease Panel
- Invitae Myofibrillar Myopathy Panel
- Invitae Nemaline Myopathy Panel
- Invitae Periodic Paralysis Panel
- Invitae Type VI Collagenopathy Panel
- Invitae Myotonia and Paramyotonia Congenita Panel
- Invitae Spinal Muscular Atrophy Panel

## NEUROPATHIES AND RELATED DISORDERS

- Invitae Comprehensive Neuropathies Panel
- Invitae Charcot-Marie-Tooth Disease Comprehensive Panel
- Invitae Charcot-Marie-Tooth Disease Autosomal Dominant Panel
- Invitae Charcot-Marie-Tooth Disease Autosomal Recessive Panel
- Invitae Charcot-Marie-Tooth Disease X-linked Panel
- Invitae Hereditary Sensory and Autonomic Neuropathy Panel
- Invitae Familial Dysautonomia Test
- Invitae Hereditary Motor Neuropathies Panel
- Invitae Spinal Muscular Atrophy Panel
- Invitae Small Fiber Neuropathy Test
- Invitae Riboflavin Transporter Deficiency Neuronopathy Panel
- Invitae Hereditary Spastic Paraplegia Comprehensive Panel
- Invitae Hereditary Spastic Paraplegia Autosomal Dominant Panel
- Invitae Hereditary Spastic Paraplegia Autosomal Recessive Panel
- Invitae Hereditary Spastic Paraplegia X-linked Panel

## CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

- Invitae Cardiomyopathy and Skeletal Muscle Disease Panel

## EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES

- Invitae Epilepsy Panel
- Invitae Alternating Hemiplegia of Childhood Panel
- Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel
- Invitae Cerebral Cavernous Malformations Panel
- Invitae CHARGE Syndrome Test
- Invitae Early Infantile Epileptic Encephalopathy Panel
- Invitae Holoprosencephaly Panel
- Invitae Neurodegeneration with Brain Iron Accumulation Panel
- Invitae Neuronal Ceroid Lipofuscinoses Panel
- Invitae Rett/Angelman and Related Disorders Panel
- Invitae Tuberous Sclerosis Complex Panel



## EYE DISORDERS

- Invitae Aniridia Test
- Invitae Axenfeld-Rieger Panel
- Invitae Bardet-Biedl Syndrome Panel
- Invitae CHARGE Syndrome Test
- Invitae Choroideremia Test
- Invitae Congenital Cataracts Panel
- Invitae Duane-Radial Ray Syndrome Test
- Invitae Early-Onset Glaucoma Panel
- Invitae Leber Congenital Amaurosis Panel
- Invitae Microphthalmia/Anophthalmia Disorders Panel
- Invitae Oculo-Facio-Cardio-Dental Syndrome Test
- Invitae Retinoblastoma Test
- Invitae Senior-Loken Syndrome Panel



**CILIOPATHIES**

- Invitae Ciliopathies Panel
- Invitae Skeletal Ciliopathies Panel
- Invitae Primary Ciliary Dyskinesia Panel
- Invitae Bardet-Biedl Syndrome Panel
- Invitae Joubert and Meckel-Gruber Syndromes Panel
- Invitae Nephronophthisis Panel
- Invitae Oral-Facial-Digital Syndrome Type 1
- Invitae Polycystic Kidney Disease Type 2 Panel
- Invitae Senior-Loken Syndrome Panel

**CONGENITAL HEART DEFECTS**

- Invitae Congenital Heart Defects and Heterotaxy Panel

**CYSTIC FIBROSIS AND CHRONIC PANCREATITIS**

- Invitae Cystic Fibrosis Test
- Invitae Chronic Pancreatitis Panel

**DEVELOPMENTAL DISORDERS**

- Invitae Alagille Syndrome Panel
- Invitae Alpha Thalassemia X-linked Intellectual Disability Test
- Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel
- Invitae Branchiootorenal Spectrum Disorders Panel
- Invitae Carpenter Syndrome Panel
- Invitae CASR-Related Conditions Test
- Invitae CHARGE Syndrome Test
- Invitae CHOPS Syndrome Test
- Invitae Coffin-Lowry Syndrome Test
- Invitae Cohen Syndrome Test
- Invitae Cornelia de Lange Syndrome Panel
- Invitae Glass Syndrome Test
- Invitae GLI3-Related Disorders Test
- Invitae HPRT1-Related Disorders Test
- Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel
- Invitae Kabuki Syndrome Panel
- Invitae KAT6B-Related Disorders Test
- Invitae KBG Syndrome Test
- Invitae MED12-Related Disorders Test
- Invitae Oculo-Facio-Cardio-Dental Syndrome Test
- Invitae PTEN-Related Disorders Test
- Invitae Renpenning Syndrome Test
- Invitae Rubinstein-Taybi Syndrome Panel
- Invitae Simpson-Golabi-Behmel Syndrome Test
- Invitae Smith-Lemli-Opitz Syndrome Test
- Invitae Sotos Syndrome Test
- Invitae van der Woude Syndrome Panel
- Invitae von Hippel-Lindau Syndrome Test
- Invitae WAGR Syndrome Panel
- Invitae Weaver Syndrome Test

**DISORDERS OF SEX DEVELOPMENT/ENDOCRINOLOGY**

- Invitae Disorders of Male Sex Development Panel
- Invitae Disorders of Female Sex Development Test
- Invitae Androgen Insensitivity Panel
- Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Panel

**EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES**

- Invitae Epilepsy Panel
- Invitae Alternating Hemiplegia of Childhood Panel
- Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Panel
- Invitae Cerebral Cavernous Malformations Panel
- Invitae CHARGE Syndrome Test
- Invitae Early Infantile Epileptic Encephalopathy Panel
- Invitae Holoprosencephaly Panel
- Invitae Neurodegeneration with Brain Iron Accumulation Panel
- Invitae Rett and Angelman Syndromes and Related Disorders Panel
- Invitae Tuberous Sclerosis Complex Panel

**EYE DISORDERS**

- Invitae Aniridia Test
- Invitae Axenfeld-Rieger Panel
- Invitae Bardet-Biedl Syndrome Panel
- Invitae CHARGE Syndrome Test
- Invitae Choroideremia Test
- Invitae Congenital Cataracts Panel
- Invitae Duane-Radial Ray Syndrome Test
- Invitae Early-Onset Glaucoma Panel
- Invitae Microphthalmia/Anophthalmia Disorders Panel
- Invitae Leber Congenital Amaurosis Panel
- Invitae Oculo-Facio-Cardio-Dental Syndrome Test
- Invitae Retinoblastoma Test
- Invitae Senior-Loken Syndrome Panel

**OVERGROWTH SYNDROMES**

- Invitae Overgrowth and Macrocephaly Syndromes Panel
- Invitae Perlman Syndrome Test
- Invitae Proteus Syndrome Test
- Invitae PTEN-Related Disorders Test
- Invitae Simpson-Golabi-Behmel Syndrome Test
- Invitae Sotos Syndrome Test
- Invitae Weaver Syndrome Test

**RASOPATHIES (NOONAN SPECTRUM DISORDERS)**

- Invitae RASopathies Comprehensive Panel
- Invitae Cardio-Facio-Cutaneous Syndrome Panel
- Invitae Costello Syndrome Test
- Invitae Legius Syndrome Test
- Invitae Neurofibromatosis Type 1 Test
- Invitae Noonan Syndrome Panel
- Invitae Noonan Syndrome with Multiple Lentigines Panel

*More pediatric genetics tests on next page.*



**SKELETAL DISORDERS**

- Invitae Antley-Bixler syndrome Test
- Invitae ARSE-Related Chondrodysplasia Punctata Test
- Invitae Campomelic Dysplasia Test
- Invitae Craniosynostosis Panel
- Invitae Duane-Radial Ray Syndrome Test
- Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel
- Invitae FGFR3-Related Disorders Test
- Invitae Hereditary Multiple Osteochondromas Panel
- Invitae Holt-Oram Syndrome Test
- Invitae NSDHL-Related Disorders Test
- Invitae Osteogenesis Imperfecta Panel
- Invitae Skeletal Ciliopathies Panel
- Invitae Thrombocytopenia Absent Radius Syndrome Test
- Invitae Townes-Brocks Syndrome Test
- Invitae Treacher-Collins Syndrome Test
- Invitae Trichorhinophalangeal Syndrome Panel
- Invitae Ulnar-Mammary Syndrome Test

**SKIN DISORDERS**

- Invitae Cardio-Facio-Cutaneous Syndrome Panel
- Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel
- Invitae Legius Syndrome Test
- Invitae Noonan Syndrome with Multiple Lentigines Panel
- Invitae PTEN-Related Disorders Test
- Invitae TP63-Related Disorders Test
- Invitae van der Woude Syndrome Panel

**PEDIATRIC ONCOLOGY**

- Invitae Pediatric Solid Tumors Panel
- Invitae Pediatric Hematologic Malignancies Panel
- Invitae Pediatric Nervous System/Brain Tumors Panel

- Invitae Boosted Exome Trio
- Invitae Boosted Exome Duo
- Invitae Boosted Exome Proband Only