

The Invitae Test Catalog



CARDIOLOGY

ARRHYTHMIA AND CARDIOMYOPATHY

Invitae Arrhythmia and Cardiomyopathy Comprehensive Test

ARRHYTHMIA

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

CARDIOMYOPATHY

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

CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

Invitae Cardiomyopathy and Skeletal Muscle Disease Test

AORTOPATHY AND CONNECTIVE TISSUE DISORDERS

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

FAMILIAL HYPERCHOLESTEROLEMIA

Invitae Familial Hypercholesterolemia Test

PULMONARY HYPERTENSION

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

CONGENITAL HEART DISEASE

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



DERMATOLOGY

DERMATOLOGY-RELATED CANCER SYNDROMES

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

SKIN DISORDERS

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



HEMATOLOGY

BONE MARROW FAILURE SYNDROMES

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

HEREDITARY HEMOCHROMATOSIS

Invitae Hereditary Hemochromatosis Test

HEREDITARY THROMBOPHILIA

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



CROSS-CANCER

- Invitae Multi-Cancer Test
- Invitae Common Hereditary Cancers Test

STAT TURNAROUND TIME

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

BREAST CANCER

- Invitae Breast Cancer Guidelines-Based Test
- Invitae Breast Cancer Test

BREAST AND GYNECOLOGIC CANCERS

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

COLORECTAL CANCER

- Invitae Colorectal Cancer Guidelines-Based Test
- Invitae Colorectal Cancer Test

ADDITIONAL ORGAN SYSTEMS

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

PEDIATRIC ONCOLOGY

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

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 Test
- Invitae Constitutional Mismatch Repair-Deficiency Test
- Invitae Costello Syndrome Test
- Invitae DICER1 Syndrome Test
- Invitae Familial Acute Myeloid Leukemia with Mutated CEBPA Test
- Invitae Familial Gastrointestinal Stromal Tumor Syndrome Test
- Invitae Familial Neuroblastoma Test
- Invitae Familial Platelet Disorder with Propensity to Myeloid Malignancy Test
- Invitae Fanconi Anemia Test
- Invitae GATA2 Deficiency Test
- Invitae Hereditary Breast and Ovarian Cancer Syndrome Test
- 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 Test
- Invitae Juvenile Polyposis Syndrome Test
- Invitae Li-Fraumeni Syndrome Test
- Invitae Lynch Syndrome Test
- Invitae Melanoma-Pancreatic Cancer Syndrome Test
- 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 Test
- Invitae Schwannomatosis Test
- Invitae Simpson-Golabi-Behmel Syndrome Test
- Invitae Small Cell Carcinoma of the Ovary Hypercalcemic Type Test
- Invitae Tuberous Sclerosis Complex Test
- Invitae von Hippel-Lindau Syndrome Test
- Invitae Weaver Syndrome Test
- Invitae Werner Syndrome Test
- Invitae Wilms Tumor Test
- Invitae WT1-Related Disorders Test



PRIMARY IMMUNODEFICIENCY

Invitae Primary Immunodeficiency Test

ANTIBODY DEFICIENCIES

Invitae Agammaglobulinemia Test

Invitae Common Variable Immunodeficiency Test

Invitae Hyper IgE Syndrome Test

Invitae Hyper IgM Syndrome Test

AUTOINFLAMMATORY SYNDROMES

Invitae Autoinflammatory Syndromes Test

Invitae Familial Cold Autoinflammatory Syndrome Test

Invitae Familial Mediterranean Fever Test

Invitae Monogenic Inflammatory Bowel Disease Test

Invitae Periodic Fever Syndromes Test

COMBINED T/B CELL DEFICIENCIES

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

Invitae Comprehensive Severe Combined Immunodeficiency (SCID) Test

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

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

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

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

Invitae X-Linked Severe Combined Immunodeficiency (SCID) Test

Invitae Radiation-Sensitive Severe Combined Immunodeficiency (SCID) Test

Invitae Combined Immunodeficiency (CID) Test

Invitae Syndromic Combined Immunodeficiency (CID) Test

DISORDERS OF INTRINSIC AND INNATE IMMUNITY

Invitae Chronic Mucocutaneous Candidiasis Test

Invitae Epidermodysplasia Verruciformis Test

Invitae Herpes Simplex Encephalitis Test

Invitae Mendelian Susceptibility to Mycobacterial Disease Test

IMMUNE DYSREGULATION

Invitae Monogenic Autoimmunity Test

Invitae Autoimmune Lymphoproliferative Disorders (ALPS) Test

Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Test

PHAGOCYTIC DEFECTS

Invitae Phagocyte Defects Test

WELL-DEFINED SYNDROMES

Invitae Dyskeratosis Congenita Test

Invitae Immunodeficiency, Centromeric Instability, Facial Anomalies Syndrome Test



METABOLIC DISORDERS NEWBORN SCREENING CONFIRMATION

Invitae Metabolic Disorders Newborn Screening Confirmation Test

Invitae Lysosomal Storage Disorders Newborn Screening Test

Invitae X-Linked Adrenoleukodystrophy Newborn Screening Confirmation Test

TESTS BY ANALYTE

Invitae Low C0 Test

Invitae Elevated C0/(C16+C18) Test

Invitae Elevated C3 Test

Invitae Elevated C3-DC Test

Invitae Elevated C4 Test

Invitae Elevated C4-DC Test

Invitae Elevated C4-OH Test

Invitae Elevated C4 & C5 Test

Invitae Elevated C5 Test

Invitae Elevated C5-DC Test

Invitae Elevated C5-OH Test

Invitae Elevated C6, C8 & C10 Test

Invitae Elevated C14 & C14:1 Test

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

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

Invitae Elevated Arginine Test

Invitae Elevated Citrulline Test

Invitae Low Citrulline Test

Invitae Elevated Glycine Test

Invitae Elevated Leucine Test

Invitae Elevated Methionine Test

Invitae Elevated Phenylalanine Test

Invitae Elevated Proline Test

Invitae Elevated Succinylacetone Test

Invitae Elevated Tyrosine Test

AMINOACIDOPATHIES

Invitae Alkaptonuria Test

Invitae Combined Methylmalonic Acidemia and Homocystinuria Test

Invitae Cystinuria Test

Invitae Disorders of Serine Biosynthesis Test

Invitae Glycine Encephalopathy Test

Invitae Homocystinuria Test

Invitae Hyperphenylalaninemia Test

Invitae Hyperprolinemia Test

Invitae Maple Syrup Urine Disease Test

Invitae Tyrosinemia Test

More metabolic disorders & newborn screening tests on next page.



CARBOHYDRATE DISORDERS

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

CEREBROTENDINOUS XANTHOMATOSIS

- Invitae Cerebrotendinous Xanthomatosis Test

CONGENITAL DISORDERS OF GLYCOSYLATION

- Invitae Congenital Disorders of Glycosylation Test

CREATINE BIOSYNTHESIS AND TRANSPORT DISORDERS

- Invitae Cerebral Creatine Deficiency Test

CYSTIC FIBROSIS

- Invitae Cystic Fibrosis Newborn Screening Confirmation Test

FATTY ACID OXIDATION DEFECTS

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

LYSOSOMAL STORAGE DISORDERS

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

NEUROTRANSMITTER DISORDERS

- Invitae Neurotransmitter Disorders Test
- Invitae Hereditary Hyperekplexia Test

ORGANIC ACIDEMIAS

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

PEROXISOMAL DISORDERS

- Invitae Adult Refsum Disease Test
- Invitae Rhizomelic Chondrodysplasia Punctata Spectrum Test
- Invitae X-linked Adrenoleukodystrophy (X-ALD) Test
- Invitae Zellweger Spectrum Disorder Test

PORPHYRIAS

- Invitae Acute Hepatic Porphyrins Test

PRIMARY HYPEROXALURIAS

- Invitae Primary Hyperoxaluria Test

PURINE METABOLISM DISORDERS

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

PYRUVATE METABOLISM AND TRICARBOXYLIC ACID CYCLE DEFECTS

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

PURINE METABOLISM DISORDERS

- Invitae Hyperammonemia Test
- Invitae Mendelian Disorders with Psychiatric Symptoms Test
- Invitae Metabolic Non-Immune Fetal Hydrops Test

More metabolic disorders & newborn screening tests on next page.



TREATABLE DISORDERS

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

UREA CYCLE DISORDERS

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

MOVEMENT DISORDERS

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

NEURODEGENERATIVE DISORDERS

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

NEUROMUSCULAR DISORDERS

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

NEUROPATHIES AND RELATED DISORDERS

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

CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

- Invitae Cardiomyopathy and Skeletal Muscle Disease Test

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES

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



EYE DISORDERS

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



CILIOPATHIES

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

CONGENITAL HEART DEFECTS

- Invitae Congenital Heart Defects and Heterotaxy Test

CYSTIC FIBROSIS AND CHRONIC PANCREATITIS

- Invitae Cystic Fibrosis Test
- Invitae Chronic Pancreatitis Test

DEVELOPMENTAL DISORDERS

- Invitae Alagille Syndrome Test
- Invitae Alpha Thalassemia X-linked Intellectual Disability Test
- Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Test
- Invitae Branchiootorenal Spectrum Disorders Test
- Invitae Carpenter Syndrome Test
- 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 Test
- Invitae Glass Syndrome Test
- Invitae GLI3-Related Disorders Test
- Invitae HPRT1-Related Disorders Test
- Invitae Isolated Gonadotropin-Releasing Hormone Deficiency Syndrome Test
- Invitae Kabuki Syndrome Test
- 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 Test
- Invitae Simpson-Golabi-Behmel Syndrome Test
- Invitae Smith-Lemli-Opitz Syndrome Test
- Invitae Sotos Syndrome Test
- Invitae van der Woude Syndrome Test
- Invitae von Hippel-Lindau Syndrome Test
- Invitae WAGR Syndrome Test
- Invitae Weaver Syndrome Test

DISORDERS OF SEX DEVELOPMENT/ENDOCRINOLOGY

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

EPILEPSY, SEIZURES, AND DEVELOPMENTAL BRAIN ABNORMALITIES

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

EYE DISORDERS

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

OVERGROWTH SYNDROMES

- Invitae Overgrowth and Macrocephaly Syndromes Test
- 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 Test
- Invitae Cardio-Facio-Cutaneous Syndrome Test
- Invitae Costello Syndrome Test
- Invitae Legius Syndrome Test
- Invitae Neurofibromatosis Type 1 Test
- Invitae Noonan Syndrome Test
- Invitae Noonan Syndrome with Multiple Lentigines Test

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 Test
- Invitae Duane-Radial Ray Syndrome Test
- Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Test
- Invitae FGFR3-Related Disorders Test
- Invitae Hereditary Multiple Osteochondromas Test
- Invitae Holt-Oram Syndrome Test
- Invitae NSDHL-Related Disorders Test
- Invitae Osteogenesis Imperfecta Test
- Invitae Skeletal Ciliopathies Test
- Invitae Thrombocytopenia Absent Radius Syndrome Test
- Invitae Townes-Brocks Syndrome Test
- Invitae Treacher-Collins Syndrome Test
- Invitae Trichorhinophalangeal Syndrome Test
- Invitae Ulnar-Mammary Syndrome Test

SKIN DISORDERS

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

PEDIATRIC ONCOLOGY

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

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