BREAKTHROUGH
GENOMICS

CATALOG OF GENETIC PANEL TESTING

2023

2 Hughes, #100, Irvine, CA 92618

www.BTGenomics.com

Table of Contents

Cardiology **3**
Hereditary Cancer **4**
Metabolic Disorders **5-6**
Neurology **6-7**
Malformations **8-9**
Ophthalmology **9-10**
Hematology **11**
Ear, Nose & Throat **12**
Gastroenterology **12**
Nephrology **12-13**
Dermatology **13-14**
Endocrinology **14-15**
Immunology **15**
Pulmonology **16**

Cardiology Panels
Aorta Panel
Arrhythmia Panel
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel
Atrial Fibrillation Panel
Brugada Syndrome Panel
Cardiomyopathy Panel
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel
Comprehensive Cardiology Panel
Congenital Structural Heart Disease Panel
Dilated Cardiomyopathy (DCM) Panel
Ehlers-Danlos Syndrome Panel
Hereditary Hemorrhagic Telangiectasia (HHT) Panel
Heterotaxy and Situs Inversus Panel
Hyperlipidemia Core Panel
Hyperlipidemia Panel
Hypertrophic Cardiomyopathy (HCM) Panel
Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel
Liddle Syndrome Panel
Long QT Syndrome (LQTS) Panel
Marfan Syndrome Panel
Noonan Syndrome Panel
Pulmonary Artery Hypertension (PAH) Panel
Short QT Syndrome (SQTS) Panel

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Hereditary Cancer panels
Comprehensive Hereditary Cancer Panel
Hereditary Breast and Gynecological Cancer Panel
Hereditary Breast Cancer High Risk Panel
Hereditary Cancer High Risk Panel
Hereditary Colorectal Cancer Panel
Hereditary Endocrine Cancer Panel
Hereditary Gastrointestinal Cancer Panel
Hereditary Leukemia Panel
Hereditary Lung Cancer Panel
Hereditary Melanoma and Skin Cancer Panel
Hereditary Pancreatic Cancer Core Panel
Hereditary Pancreatic Cancer Panel
Hereditary Paraganglioma-Pheochromocytoma Panel
Hereditary Pediatric Cancer Panel
Hereditary Renal Cancer Panel
Neurofibromatosis Panel
Tuberous Sclerosis Panel
Xeroderma Pigmentosum Panel

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Metabolic Disorders panels
Aicardi-Goutières Syndrome Panel
Coenzyme q10 Deficiency Panel
Comprehensive Metabolism Panel
Congenital and Familial Lipodystrophy Panel
Congenital Disorders of Glycosylation Panel
Congenital Mono- and Disaccharide Disorders Panel
Creatine Metabolism Deficiency Panel
Cystinuria Panel
Fatty Acid Oxidation Syndrome Panel
Glycogen Storage Disorder Panel
Hereditary Hemochromatosis Panel
Homocystinuria Core Panel
Hyperammonemia and Urea Cycle Disorder Panel
Hyperphenylalaninemia Panel
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel
Hypomagnesemia Panel
Lysosomal Disorders and Mucopolysaccharidosis Panel
Metabolic Liver Failure Panel
Metabolic Myopathy and Rhabdomyolysis Panel
Mitochondrial DNA Depletion Syndrome Panel
Monogenic Obesity Panel
Nephrolithiasis Panel
Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel
Organic Acidemia/Aciduria & Cobalamin Deficiency Panel
Periodic Paralysis Panel

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Peroxisomal Disorders Panel
Porphyria Panel
Purine and Pyrimidine Metabolism Disorders Panel
Tyrosinemia Panel

Neurology panels
Amyotrophic Lateral Sclerosis Panel
Ataxia Panel
Autism Spectrum Disorders Panel
Beyond Paediatric Epilepsy Panel – for Europe and
Cerebral Cavernous Malformation Panel
Charcot-Marie-Tooth Neuropathy Panel
Coenzyme q10 Deficiency Panel
Collagen Type VI-Related Disorders Panel
Comprehensive Epilepsy Panel
Comprehensive Muscular Dystrophy / Myopathy Panel
Congenital Myasthenic Syndromes Panel
Creatine Metabolism Deficiency Panel
Dementia Panel
Dystonia Panel
Emery-Dreifuss Muscular Dystrophy Panel
Epileptic Encephalopathy Panel
Holoprosencephaly Panel
Idiopathic Generalized and Focal Epilepsy Panel
Leukodystrophy and Leukoencephalopathy Panel

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LGMD and Congenital Muscular Dystrophy Panel
Lissencephaly Panel
Macrocephaly / Overgrowth Syndrome Panel
Metabolic Epilepsy Panel
Metabolic Myopathy and Rhabdomyolysis Panel
Microcephaly and Pontocerebellar Hypoplasia Panel
Migraine Panel
NCL and Progressive Myoclonic Epilepsy Panel
Nemaline Myopathy Panel
Neuro-Ophthalmology Panel
Neuronal Migration Disorder Panel
Parkinson Disease Panel
Periodic Paralysis Panel
Polymicrogyria Panel
Porphyria Panel
Septo-Optic Dysplasia Panel
Spastic Paraplegia Panel
Spinal Muscular Atrophy Panel
Tuberous Sclerosis Panel
X-linked Intellectual Disability Panel

Malformations panels
3-M Syndrome / Primordial Dwarfism Panel
Adams-Oliver Syndrome Panel
Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel

Arthrogryposes Panel
Brachydactyly / Syndactyly Panel
Cerebral Cavernous Malformation Panel
Chondrodysplasia Punctata Panel
Cleft Lip/Palate and Associated Syndromes Panel
Comprehensive Growth Disorders / Skeletal Dysplasias and Disorders Panel
Comprehensive Short Stature Syndrome Panel
Comprehensive Skeletal Dysplasias and Disorders Panel
Cornelia de Lange Syndrome Panel
Craniosynostosis Panel
Exostosis and Related Disorders Panel
Facial Dysostosis and Related Disorders Panel
Gastrointestinal Atresia Panel
Heterotaxy and Situs Inversus Panel
Hirschsprung Disease Panel
Holoprosencephaly Panel
Kabuki Syndrome Panel
Limb Malformations Panel
Lissencephaly Panel
Lymphatic Malformations and Related Disorders Panel
Macrocephaly / Overgrowth Syndrome Panel
Meier-Gorlin Syndrome Panel
Metaphyseal Dysplasia Panel
Microcephaly and Pontocerebellar Hypoplasia Panel
Micromelic Dysplasia Panel

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Neurofibromatosis Panel
Neuronal Migration Disorder Panel
Osteogenesis Imperfecta Panel
Osteopetrosis and Dense Bone Dysplasia Panel
Polymicrogyria Panel
Seckel Syndrome Panel
Septo-Optic Dysplasia Panel
Short Rib Dysplasia / Asphyxiating Thoracic Dysplasia Panel
Skeletal Dysplasia with Abnormal Mineralization Panel
Skeletal Dysplasias Core Panel
Spondylometaphyseal / Spondyloepi-(meta)-physeal Dysplasia Panel
Vascular Malformations Panel

Ophthalmology panels
Achromatopsia Panel
Albinism Panel
Bardet-Biedl Syndrome Panel
Cataract Panel
Cone Rod Dystrophy Panel
Congenital Stationary Night Blindness Panel
Corneal Dystrophy Panel
Ectopia Lentis Panel
Flecked Retina Disorders Panel
Glaucoma Panel
Joubert Syndrome Panel

Leber Congenital Amaurosis Panel
Macular Dystrophy Panel
Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel
Neuro-Ophthalmology Panel
Optic Atrophy Panel
Retinal Dystrophy Panel
Retinitis Pigmentosa Panel
Senior-Loken Syndrome Panel
Septo-Optic Dysplasia Panel
Stickler Syndrome Panel
Usher Syndrome Panel
Vitreoretinopathy Panel

Hematology panels
Anemia Panel
Bleeding Disorder/Coagulopathy Panel
Bloom Syndrome Panel
Bone Marrow Failure Syndrome Panel
Coagulation Factor Deficiency Panel
Comprehensive Hematology Panel
Congenital Neutropenia Panel
Diamond-Blackfan Anemia Panel
Dyskeratosis Congenita Panel
Fanconi Anemia Panel
Hemophagocytic Lymphohistiocytosis Panel

Hereditary Leukemia Panel
Hermansky-Pudlak Syndrome Panel
Platelet Function Disorder Panel
Red Blood Cell Membrane Disorder Panel
Thrombocytopenia Panel

Ear, Nose & Throat panels

Alport Syndrome Panel
Branchio-Oto Renal (BOR) Syndrome Panel
Comprehensive Hearing Loss and Deafness Panel
Hereditary Hemorrhagic Telangiectasia (HHT) Panel
Non-Syndromic Hearing Loss Panel
Pendred Syndrome Panel
Stickler Syndrome Panel
Syndromic Hearing Loss Panel
Usher Syndrome Panel
Waardenburg Syndrome Panel

Gastroenterology panels

Cholestasis Panel
Congenital Diarrhea Panel
Congenital Hepatic Fibrosis Panel
Gastrointenstinal Atresia Panel
Hirschsprung Disease Panel
Pancreatitis Panel

Polycystic Liver Disease Panel

Nephrology panels
Alport Syndrome Panel
Bardet-Biedl Syndrome Panel
Bartter Syndrome Panel
Branchio-Oto-Renal (BOR) Syndrome Panel
Ciliopathy Panel
Cystic Kidney Disease Panel
Diabetes Insipidus Panel
Hemolytic Uremic Syndrome Panel
Hypomagnesemia Panel
Hypophosphatemic Rickets Panel
Joubert Syndrome Panel
Liddle Syndrome Panel
Meckel Syndrome Panel
Monogenic Obesity Panel
Nephrolithiasis Panel
Nephronophthisis Panel
Nephrotic Syndrome Panel
Polycystic Kidney Disease Panel
Primary Ciliary Dyskinesia Panel
Primary Hyperoxaluria Panel
Pseudohypoaldosteronism Panel
Renal Malformation Panel

Renal Tubular Acidosis Panel
Senior-Loken Syndrome Panel

Dermatology panels
Adams-Oliver Syndrome Panel
Albinism Panel
Cutis Laxa Panel
Dyskeratosis Congenita Panel
Ectodermal Dysplasia Panel
Ehlers-Danlos Syndrome Panel
Epidermolysis Bullosa Panel
Hereditary Acrodermatitis Enteropathica Panel
Hereditary Melanoma and Skin Cancer Panel
Hermansky-Pudlak Syndrome Panel
Ichthyosis Panel
Neurofibromatosis Panel
Pachyonychia Congenita Panel
Palmoplantar Keratoderma Panel
Progeria and Progeroid Syndromes Panel
Tuberous Sclerosis Panel
Waardenburg Syndrome Panel
Xeroderma Pigmentosum Panel

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Endocrinology panels
Abnormal Genitalia/Disorders of Sex Development Panel
Comprehensive Monogenic Diabetes Panel
Congenital Adrenal Hyperplasia Panel
Glucocorticoid Deficiency Panel
Hyperlipidemia Panel
Hyperparathyroidism Panel
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel
Hypomagnesemia Panel
Hypothyroidism and Resistance to Thyroid Hormone Panel
Kallmann Syndrome Panel
MODY Panel
Monogenic Obesity Panel
Premature Ovarian Failure Panel

Immunology panels
Autoinflammatory Syndrome Panel
Chronic Granulomatous Disease Panel
Congenital Neutropenia Panel
Hemophagocytic Lymphohistiocytosis Panel
Primary Immunodeficiency Panel
Bone Marrow Failure Syndrome Panel
Complement System Disorder Panel
Dyskeratosis Panel
Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel

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Severe Combined Immunodeficiency Panel

Pulmonology panels
Bronchiectasis Panel
Comprehensive Pulmonology Panel
Cystic Lung Disease Panel
Interstitial Lung Disease Panel
Primary Ciliary Dyskinesia Panel
Central Hypoventilation and Apnea Panel
Cystic Fibrosis Panel
Hermansky-Pudlak Syndrome Panel
Neonatal Respiratory Distress – Surfactant Dysfunction Panel
Pulmonary Artery Hypertension (PAH) Panel

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