Our test menu and service offering

Panels
Panel Customization
Whole Exome Tests

Variant Specific Testing
Single Gene Testing
Mitochondrial DNA Testing

Blueprint Genetics

We provide comprehensive, actionable genetic diagnostics for all medical specialties.

Finding the needle in the haystack

Inherited disorders are found in approximately 5% of the world's population, or around 350 million people. Although the number of these diseases is sizeable, many are so rare that they are unknown to the majority of clinicians. Identifying them using conventional diagnostics is a process of trial and error that often yields no definitive answers.

Accessibility

The time and expense needed to provide results through traditional genetic testing methods has meant this type of diagnostic test was often used as a last resort. Today's genetic testing technology means that patients can get their results in a fraction of the time – and at a fraction of the cost.

A clear path to early treatment

Anything we can do to improve and speed up diagnosis not only allows us to plan the best and most cost-effective treatments, but also plays an important part in long-term prognosis. This is where genetic testing can truly make a difference.

Blueprint Genetics

Panels

We offer more than 220 panels covering all medical specialties. Consider ordering a panel when your patient has a known or suspected clinical diagnosis and the causative genes are well-described and available on one of our panels.

For example, panel testing is recommended for patients with a suspected diagnosis of hypertrophic cardiomyopathy, epilepsy, or retinal dystrophy.

Maximized diagnostic potential

- All of our panels include both Sequence and Deletion/Duplication (CNV) Analysis
- Clinically relevant, deep intronic variants included
- Increased capabilities in difficult-to-sequence regions
- mtDNA analysis included in panels for conditions where symptoms or findings can be caused by mtDNA mutations
- Panels can be customized by adding up to 200 genes (including the mtDNA genes)

Cardiology Panels	Genes
Aorta Panel	53
Arrhythmia Panel	62
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel	21
Atrial Fibrillation Panel	19
Brugada Syndrome Panel	7
mtDNA Cardiomyopathy Panel	217
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel	10
mtDNA Comprehensive Cardiology Panel	260
Congenital Structural Heart Disease Panel	125
mtDNA Dilated Cardiomyopathy (DCM) Panel	130
Ehlers-Danlos Syndrome Panel	41
Hereditary Hemorrhagic Telangiectasia (HHT) Panel	6
Heterotaxy and Situs Inversus Panel	34
Hyperlipidemia Core Panel	4
Hyperlipidemia Panel	20
mtDNA Hypertrophic Cardiomyopathy (HCM) Panel	92
Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel	33
Liddle Syndrome Panel	2
Long QT Syndrome (LQTS) Panel	18
Marfan Syndrome Panel	36
Noonan Syndrome Panel	36
Pulmonary Artery Hypertension (PAH) Panel	23
Short QT Syndrome (SQTS) Panel	5

Dermatology Panels	Genes
Adams-Oliver Syndrome Panel	8
Albinism Panel	26
Cutis Laxa Panel	10
Dyskeratosis Congenita Panel	15
Ectodermal Dysplasia Panel	25
Ehlers-Danlos Syndrome Panel	41
Epidermolysis Bullosa Panel	26
Hereditary Acrodermatitis Enteropathica Panel	2
Hereditary Melanoma and Skin Cancer Panel	19
Hermansky-Pudlak Syndrome Panel	23
Ichthyosis +B:BPanel	39
Neurofibromatosis Panel	9
Pachyonychia Congenita Panel	7
Palmoplantar Keratoderma Panel	26
Progeria and Progeroid Syndromes Panel	17
Tuberous Sclerosis Panel	2
Waardenburg Syndrome Panel	7
Xeroderma Pigmentosum Panel	9

Please use the test code when placing orders. We are continuously developing the gene set description and panel composition to match the latest research findings. Please find the most updated list of genes, panels and panel descriptions at blueprintgenetics.com.

Ear-Nose-Throat Panels	Genes
Alport Syndrome Panel	6
Branchio-Oto-Renal (BOR) Syndrome Panel	4
mtDNA Comprehensive Hearing Loss and Deafness Panel	288
Hereditary Hemorrhagic Telangiectasia (HHT) Panel	6
mtDNA Non-Syndromic Hearing Loss Panel	138
Pendred Syndrome Panel	3
mtDNA Resonate Program Panel (US only)	288
Stickler Syndrome Panel	8
mtDNA Syndromic Hearing Loss Panel	138
Usher Syndrome Panel	21
Waardenburg Syndrome Panel	7

Endocrinology Panels	Genes
Abnormal Genitalia / Disorders of Sex Development Panel	73
mtDNA Comprehensive Monogenic Diabetes Panel	67
Congenital Adrenal Hyperplasia Panel	12
Glucocorticoid Deficiency Panel	7
Hyperlipidemia Panel	20
Hyperparathyroidism Panel	14
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	50
Hypomagnesemia Panel	19
Hypothyroidism and Resistance to Thyroid Hormone Panel	22
Kallmann Syndrome Panel	31
mtDNA MODY Panel	54
Monogenic Obesity Panel	41
Premature Ovarian Failure Panel	16

Gastroenterology Panels	Genes
Cholestasis Panel	52
Congenital Diarrhea Panel	29
Congenital Hepatic Fibrosis Panel	52
Gastrointestinal Atresia Panel	15
Hirschsprung Disease Panel	15
Pancreatitis Panel	9
Polycystic Liver Disease Panel	6

Hematology Panels	Genes
Anemia Panel	88
Bleeding Disorder / Coagulopathy Panel	71
Bone Marrow Failure Syndrome Panel	156
Coagulation Factor Deficiency Panel	16
Comprehensive Hematology and Hereditary Cancer Panel	369
Comprehensive Hematology Panel	270
Comprehensive Immune and Cytopenia Panel	642
Congenital Neutropenia Panel	28
Diamond-Blackfan Anemia Panel	14
Dyskeratosis Congenita Panel	15
Fanconi Anemia Panel	24
Hemophagocytic Lymphohistiocytosis Panel	15

Hematology Panels	Genes
Hereditary Leukemia Panel	42
Hermansky-Pudlak Syndrome Panel	23
Platelet Function Disorder Panel	23
Red Blood Cell Membrane Disorder Panel	7
Thrombocytopenia Panel	37

Hereditary Cancer Panels	Genes
Comprehensive Hematology and Hereditary Cancer Panel	369
Comprehensive Hereditary Cancer Panel	160
Hereditary Breast and Gynecological Cancer Panel	28
Hereditary Breast Cancer High Risk Panel	8
Hereditary Cancer High Risk Panel	28
Hereditary Colorectal Cancer Panel	23
Hereditary Endocrine Cancer Panel	22
Hereditary Gastrointestinal Cancer Panel	43
Hereditary Leukemia Panel	42
Hereditary Lung Cancer Panel	5
Hereditary Melanoma and Skin Cancer Panel	19
Hereditary Pancreatic Cancer Core Panel	5
Hereditary Pancreatic Cancer Panel	22
Hereditary Paraganglioma-Pheochromocytoma Panel	11
Hereditary Pediatric Cancer Panel	71
Hereditary Renal Cancer Panel	26
Hereditary Retinoblastoma Panel	1
Neurofibromatosis Panel	9
Tuberous Sclerosis Panel	2
Xeroderma Pigmentosum Panel	9

Immunology Panels	Genes
Autoinflammatory Syndrome Panel	47
Bone Marrow Failure Syndrome Panel	156
Chronic Granulomatous Disease Panel	8
Comprehensive Immune and Cytopenia Panel (contracted only)	642
Complement System Disorder Panel	80
Congenital Neutropenia Panel	28
Dyskeratosis Congenita Panel	15
Hemophagocytic Lymphohistiocytosis Panel	15
Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel	383
Primary Immunodeficiency Panel	336
Severe Combined Immunodeficiency Panel	80

Malformations Panels	Genes
3-M Syndrome / Primordial Dwarfism Panel	24
Adams-Oliver Syndrome Panel	8
Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	16
Arthrogryposes Panel	78

Malformations Panels	Genes
Brachydactyly / Syndactyly Panel	21
Cerebral Cavernous Malformation Panel	4
Chondrodysplasia Punctata Panel	10
Cleft Lip/Palate and Associated Syndromes Panel	22
Comprehensive Growth Disorders / Skeletal Dysplasias and Disorders Panel	510
Comprehensive Short Stature Syndrome Panel	100
Comprehensive Skeletal Dysplasias and Disorders Panel	411
Cornelia de Lange Syndrome Panel	6
Craniosynostosis Panel	38
Discover Dysplasias Program Panel - for Europe and Middle East	419
Exostosis and Related Disorders Panel	3
Facial Dysostosis and Related Disorders Panel	27
Gastrointestinal Atresia Panel	15
Heterotaxy and Situs Inversus Panel	34
Hirschsprung Disease Panel	15
Holoprosencephaly Panel	12
Kabuki Syndrome Panel	9
Limb Malformations Panel	50
Lissencephaly Panel	24
Lymphatic Malformations and Related Disorders Panel	12
Macrocephaly / Overgrowth Syndrome Panel	48
Meier-Gorlin Syndrome Panel	6
Metaphyseal Dysplasia Panel	12
Microcephaly and Pontocerebellar Hypoplasia Panel	78
Micromelic Dysplasia Panel	27
Neurofibromatosis Panel	9
Neuronal Migration Disorder Panel	59
Osteogenesis Imperfecta Panel	33
Osteopetrosis and Dense Bone Dysplasia Panel	27
Polymicrogyria Panel	20
Seckel Syndrome Panel	6
Septo-Optic Dysplasia Panel	4
Short Rib Dysplasia / Asphyxiating Thoracic Dysplasia Panel	18
Skeletal Dysplasia with Abnormal Mineralization Panel	36
Skeletal Dysplasias Core Panel	113
Spondylometaphyseal / Spondyloepi-(meta)-physeal Dysplasia Panel	36
Vascular Malformations Panel	6

Metabolic Disorders Panels	Genes
Aicardi-Goutières Syndrome Panel	7
Coenzyme q10 Deficiency Panel	15

Metabolic Disorders Panels	Genes	
Comprehensive Metabolism Panel	505	mtDNA
Congenital and Familial Lipodystrophy Panel	12	
Congenital Disorders of Glycosylation Panel	48	
Congenital Mono- and Disaccharide Disorders Panel	9	
Creatine Metabolism Deficiency Panel	33	
Cystinuria Panel	2	
Fatty Acid Oxidation Syndrome Panel	26	
Glycogen Storage Disorder Panel	29	
Hereditary Hemochromatosis Panel	5	
Homocystinuria Core Panel	4	
Hyperammonemia and Urea Cycle Disorder Panel	49	
Hyperphenylalaninemia Panel	6	
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	50	
Hypomagnesemia Panel	19	
Lysosomal Disorders and Mucopolysaccharidosis Panel	102	
Metabolic Liver Failure Panel	16	
Metabolic Myopathy and Rhabdomyolysis Panel	127	mtDNA
Mitochondrial DNA Depletion Syndrome Panel	62	mtDNA
Monogenic Obesity Panel	41	
Nephrolithiasis Panel	35	
Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel	9	
Organic Acidemia/Aciduria & Cobalamin Deficiency Panel	54	
Periodic Paralysis Panel	4	
Peroxisomal Disorders Panel	27	
Porphyria Panel	9	
Purine and Pyrimidine Metabolism Disorders Panel	21	
Tyrosinemia Panel	3	

Mitochondrial disorders Panels	Genes	
Mitochondrial Genome Test	37	mtDNA
Mitochondrial DNA Depletion Syndrome Panel	62	mtDNA

Nephrology Panels	Genes
Alport Syndrome Panel	6
Bardet-Biedl Syndrome Panel	27
Bartter Syndrome Panel	10
Branchio-Oto-Renal (BOR) Syndrome Panel	4
Ciliopathy Panel	119
Cystic Kidney Disease Panel	43
Diabetes Insipidus Panel	3
Hemolytic Uremic Syndrome Panel	9
Hypomagnesemia Panel	19
Hypophosphatemic Rickets Panel	13
Joubert Syndrome Panel	36
Liddle Syndrome Panel	2
Meckel Syndrome Panel	13
Monogenic Obesity Panel	41
Nephrolithiasis Panel	45
Nephronophthisis Panel	20

Nephrology Panels	Genes
Nephrotic Syndrome Panel (mtDNA)	96
Polycystic Kidney Disease Panel	13
Primary Ciliary Dyskinesia Panel	47
Primary Hyperoxaluria Panel	3
Pseudohypoaldosteronism Panel	10
Renal Malformation Panel	27
Renal Tubular Acidosis Panel	5
Senior-Loken Syndrome Panel	9

Neurology Panels	Genes
Amyotrophic Lateral Sclerosis Panel	35
Ataxia Panel (mtDNA)	257
Autism Spectrum Disorders Panel (mtDNA)	75
Beyond Paediatric Epilepsy Panel – for Europe and Middle East (mtDNA)	511
Cerebral Cavernous Malformation Panel	4
Charcot-Marie-Tooth Neuropathy Panel (mtDNA)	153
Coenzyme q10 Deficiency Panel	15
Collagen Type VI-Related Disorders Panel	6
Comprehensive Epilepsy Panel (mtDNA)	511
Comprehensive Muscular Dystrophy / Myopathy Panel (mtDNA)	161
Congenital Myasthenic Syndromes Panel	21
Creatine Metabolism Deficiency Panel	3
Dementia Panel (mtDNA)	58
Dystonia Panel (mtDNA)	68
Emery-Dreifuss Muscular Dystrophy Panel	6
Epileptic Encephalopathy Panel (mtDNA)	203
Holoprosencephaly Panel	12
Idiopathic Generalized and Focal Epilepsy Panel	35
Leukodystrophy and Leukoencephalopathy Panel (mtDNA)	118
LGMD and Congenital Muscular Dystrophy Panel	56
Lissencephaly Panel	24
Macrocephaly / Overgrowth Syndrome Panel	48
Metabolic Epilepsy Panel (mtDNA)	84
Metabolic Myopathy and Rhabdomyolysis Panel (mtDNA)	90
Microcephaly and Pontocerebellar Hypoplasia Panel	78
Migraine Panel (mtDNA)	47
NCL and Progressive Myoclonic Epilepsy Panel	31
Nemaline Myopathy Panel	13
Neuro-Ophthalmology Panel (mtDNA)	97
Neuronal Migration Disorder Panel	59
Parkinson Disease Panel (mtDNA)	82
Periodic Paralysis Panel	4
Polymicrogyria Panel	20
Porphyria Panel	9

Neurology Panels	Genes
Septo-Optic Dysplasia Panel	4
Spastic Paraplegia Panel	75
Spinal Muscular Atrophy Panel	30
Tuberous Sclerosis Panel	2
X-linked Intellectual Disability Panel	106

Ophthalmology Panels	Genes
Achromatopsia Panel	8
Albinism Panel	26
Bardet-Biedl Syndrome Panel	27
Cataract Panel (mtDNA)	113
Cone Rod Dystrophy Panel	44
Congenital Stationary Night Blindness Panel	20
Corneal Dystrophy Panel	29
Ectopia Lentis Panel	14
Flecked Retina Disorders Panel	12
Glaucoma Panel	19
Joubert Syndrome Panel	36
Leber Congenital Amaurosis Panel	28
Macular Dystrophy Panel	28
Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel	61
My Retina Tracker Program Panel (US only) (mtDNA)	351
Neuro-Ophthalmology Panel (mtDNA)	97
Optic Atrophy Panel (mtDNA)	76
Retinal Dystrophy Panel (mtDNA)	351
Retinitis Pigmentosa Panel (mtDNA)	159
Senior-Loken Syndrome Panel	9
Septo-Optic Dysplasia Panel	4
Stickler Syndrome Panel	8
Usher Syndrome Panel	21
Vitreoretinopathy Panel	24

Pulmonology Panels	Genes
Bronchiectasis Panel	22
Central Hypoventilation and Apnea Panel	15
Comprehensive Pulmonology Panel (mtDNA)	114
Cystic Lung Disease Panel	8
Hermansky-Pudlak Syndrome Panel	23
Interstitial Lung Disease Panel	30
Neonatal Respiratory Distress - Surfactant Dysfunction Panel	5
Primary Ciliary Dyskinesia Panel	41
Pulmonary Artery Hypertension (PAH) Panel	23

Whole Exome Tests

Whole Exome Sequencing	Analyses
Whole Exome Plus	Index patient
Whole Exome Family Plus	Index patient +parents

Single Gene Testing

Our test menu includes over 3900 single gene tests. Consider ordering single gene testing when you highly suspect that your patient's symptoms are caused by a particular gene or when you want to do carrier testing for a couple where one member is known to be a carrier of a variant in a particular gene. For example, a patient is a known carrier of cystic fibrosis and their partner requests testing to determine their risk of having an affected child.

Variant Specific Testing

We offer two Variant Specific Testing products:

- Choose Familial Variant Testing when there is a known variant in a family member, and you want to do diagnostic testing in affected family members, predictive testing in unaffected family members, carrier testing in the case of autosomal recessive and X-linked disorders or segregation of variants.
- Choose Targeted Variant Testing for confirmatory analysis of research or direct-to-consumer results, testing of published founder or common variants or clarification of variant classification for variants reported by another laboratory.

Customization of Panels

All our 220+ Sequencing Panels can be customized by adding any of our 4000+ clinically relevant genes or by removing genes from the selected panel. Customized panels can be saved for later use in our ordering platform, Nucleus.

WES Re-evaluation and Re-analysis Services

WES Variant Re-evaluation service reviews all available scientific information in an attempt to reclassify variants originally classified as variants of uncertain significance (VUS) or as Likely Pathogenic (LP) free of charge.

WES Re-analysis Service allows the sequencing data from a previous exome to be re-analyzed in its entirety. The goal is to find new clinically relevant variants that may explain or contribute to your patient's diagnosis.

Nucleus:
Easy ordering platform

Place orders, follow progress, and read results with our online ordering portal, Nucleus.

Sign-in or register to Nucleus:
nucleus.blueprintgenetics.com

Contact details:

Customer Support

The team will help help you with questions concerning:

- Placing orders
- Billing
- Ordering sample collection kits
- Sample requirements

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We are continuously developing our services and offering. We may amend service descriptions from time to time by posting new versions on our website. For up-to-date information, please visit blueprintgenetics.com.

Clinical Genetics Support

Our expert team of geneticists, physicians, clinical consultants, and genetic counselors is available for:

- Discussions on selecting the optimal diagnostic tools for your patient
- Questions concerning the clinical statement
- Discussions on variant classification
- Assistance in genetic counseling of your patient and their family
- Tailor-made service options for your specific clinical needs

E-mail: genetics.support@blueprintgenetics.com

Blueprint Genetics

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