Test Leaflet

Our test menu and service offering

Panels
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Whole Exome Tests

Variant Specific Testing
Single Gene 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

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.

All of our panels include both Sequence and Deletion/ Duplication (CNV) Analysis.

Cardiology Panels	Test Code	Genes
Ehlers-Danlos Syndrome Panel	CA0101	32
Hereditary Hemorrhagic Telangiectasia (HHT) Panel	CA0201	6
Long QT Syndrome (LQTS) Panel	CA0301	16
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel	CA0401	21
Noonan Syndrome Panel	CA0501	35
Pulmonary Artery Hypertension (PAH) Panel	CA0601	23
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel	CA0701	10
Marfan Syndrome Panel	CA0801	34
Brugada Syndrome Panel	CA0901	7
Aorta Panel	CA1001	52
Hyperlipidemia Panel	CA1101	18
Cardiomyopathy Panel	CA1201	177
Comprehensive Cardiology Panel	CA1301	217
Liddle Syndrome Panel	CA1401	2
Congenital Structural Heart Disease Panel	CA1501	114
Arrhythmia Panel	CA1601	58
Hyperlipidemia Core Panel	CA1701	4
Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel	CA1801	33
Hypertrophic Cardiomyopathy (HCM) Panel	CA1901	47
Atrial Fibrillation Panel	CA2001	19
Short QT Syndrome (SQTS) Panel	CA2101	5
Dilated Cardiomyopathy (DCM) Panel	CA2201	86
Heterotaxy and Situs Inversus Panel	CA2301	34

Dermatology Panels	Test Code	Genes
Adams-Oliver Syndrome Panel	MA1601	8
Ehlers-Danlos Syndrome Panel	CA0101	32
Hereditary Acrodermatitis Enteropathica Panel	DE0101	2
Progeria and Progeroid Syndromes Panel	DE0201	17
Epidermolysis Bullosa Panel	DE0301	26
Ectodermal Dysplasia Panel	DE0401	25
Cutis Laxa Panel	DE0501	10
Ichthyosis +B:BPanel	DE0601	39
Pachyonychia Congenita Panel	DE0701	7
Albinism Panel	DE0801	23
Palmoplantar Keratoderma Panel	DE0901	26
Waardenburg Syndrome Panel	EA0101	7
Hermansky-Pudlak Syndrome Panel	HE1101	23
Dyskeratosis Congenita Panel	IM0401	12
Neurofibromatosis Panel	MA1501	9
Hereditary Melanoma and Skin Cancer Panel	ON0501	19
Xeroderma Pigmentosum Panel	ON0601	9
Tuberous Sclerosis Panel	ON1401	2

Ear-Nose-Throat Panels	Test Code	Genes
Alport Syndrome Panel	KI1101	6
Branchio-Oto-Renal (BOR) Syndrome Panel	KI1501	4
Comprehensive Hearing Loss and Deafness Panel	EA0501	202
Hereditary Hemorrhagic Telangiectasia (HHT) Panel	CA0201	6
Non-Syndromic Hearing Loss Panel	EA0201	101
Pendred Syndrome Panel	EA0301	3
Stickler Syndrome Panel	OP1501	8
Syndromic Hearing Loss Panel	EA0401	101
Usher Syndrome Panel	OP1101	21
Waardenburg Syndrome Panel	EA0101	7

Endocrinology Panels	Test Code	Genes
Abnormal Genitalia/ Disorders of Sex Development Panel	EN0201	62
Comprehensive Monogenic Diabetes Panel	EN0401	30
Congenital Adrenal Hyperplasia Panel	EN0801	12
Glucocorticoid Deficiency Panel	EN0501	7
Hyperlipidemia Panel	CA1101	18
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	ME0601	50
Hypomagnesemia Panel	ME0501	19
Hypothyroidism and Resistance to Thyroid Hormone Panel	EN0701	22
Hypothyroidism and Resistance to Thyroid Hormone Panel	EN0701	22
Kallmann Syndrome Panel	EN0301	17
MODY Panel	EN0601	15
Monogenic Obesity Panel	KI1701	41
Premature Ovarian Failure Panel	EN0901	16

Gastroenterology Panels	Test Code	Genes
Congenital Diarrhea Panel	GA0401	29
Cholestasis Panel	GA0501	47
Congenital Hepatic Fibrosis Panel	GA0101	52
Gastrointestinal Atresia Panel	MA2801	15
Hirschsprung Disease Panel	MA1801	15
Pancreatitis Panel	GA0301	9
Polycystic Liver Disease Panel	GA0201	6

Hematology Panels	Test Code	Genes
Anemia Panel	HE0401	88
Bleeding Disorder/Coagulopathy Panel	HE1301	71
Bloom Syndrome Panel	HE0201	1
Bone Marrow Failure Syndrome Panel	HE0801	135
Coagulation Factor Deficiency Panel	HE0501	16
Comprehensive Hematology and Hereditary Cancer Panel	HE1401	348
Comprehensive Hematology Panel	HE0101	253
Congenital Neutropenia Panel	IM0501	28
Diamond-Blackfan Anemia Panel	HE0901	14
Dyskeratosis Congenita Panel	IM0401	12
Fanconi Anemia Panel	HE0301	24
Hemophagocytic Lymphohistiocytosis Panel	HE1001	15

Hematology Panels	Test Code	Genes
Hereditary Leukemia Panel	ON0101	41
Hermansky-Pudlak Syndrome Panel	HE1101	23
Platelet Function Disorder Panel	HE0701	23
Red Blood Cell Membrane Disorder Panel	HE1201	7
Thrombocytopenia Panel	HE0601	37

Hereditary Cancer Panels	Test Code	Genes
Comprehensive Hematology and Hereditary Cancer Panel	HE1401	348
Comprehensive Hereditary Cancer Panel	ON1001	154
Hereditary Breast and Gynecological Cancer Panel	ON1801	28
Hereditary Breast Cancer High Risk Panel	ON1901	8
Hereditary Cancer High Risk Panel	ON2101	28
Hereditary Colorectal Cancer Panel	ON0201	21
Hereditary Endocrine Cancer Panel	ON0701	22
Hereditary Gastrointestinal Cancer Panel	ON1601	42
Hereditary Leukemia Panel	ON0101	41
Hereditary Lung Cancer Panel	ON0401	5
Hereditary Melanoma and Skin Cancer Panel	ON0501	19
Hereditary Pancreatic Cancer Core Panel	ON2001	5
Hereditary Pancreatic Cancer Panel	ON0301	22
Hereditary Paraganglioma-Pheochromocytoma Panel	ON1201	11
Hereditary Pediatric Cancer Panel	ON0801	71
Hereditary Renal Cancer Panel	ON1501	26
Hereditary Retinoblastoma Panel	ON1701	1
Neurofibromatosis Panel	MA1501	9
Tuberous Sclerosis Panel	ON1401	2
Xeroderma Pigmentosum Panel	ON0601	9

Immunology Panels	Test Code	Genes
Autoinflammatory Syndrome Panel	IM0201	33
Bone Marrow Failure Syndrome Panel	HE0801	135
Chronic Granulomatous Disease Panel	IM0601	8
Complement System Disorder Panel	IM0701	80
Congenital Neutropenia Panel	IM0501	28
Dyskeratosis Congenita Panel	IM0401	12
Hemophagocytic Lymphohistiocytosis Panel	HE1001	15
Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel	IM0801	337
Primary Immunodeficiency Panel	IM0301	298
Severe Combined Immunodeficiency Panel	IM0101	80

Malformations Panels	Test Code	Genes
3-M Syndrome / Primordial Dwarfism Panel	MA2401	24
Adams-Oliver Syndrome Panel	MA1601	8
Amelogenesis Imperfecta and Dentinogenesis Imperfecta Panel	MA3601	16
Arthrogryposes Panel	MA0501	78
Brachydactyly / Syndactyly Panel	MA1201	21
Cerebral Cavernous Malformation Panel	MA1001	4

Malformations Panels	Test Code	Genes
Chondrodysplasia Punctata Panel	MA2701	9
Cleft Lip/Palate and Associated Syndromes Panel	MA3701	22
Comprehensive Growth Disorders / Skeletal Dysplasias and Disorders Panel	MA4301	374
Comprehensive Short Stature Syndrome Panel	MA2101	98
Comprehensive Skeletal Dysplasias and Disorders Panel	MA3301	251
Cornelia de Lange Syndrome Panel	MA3801	6
Craniosynostosis Panel	MA2901	38
Exostosis and Related Disorders Panel	MA3901	3
Facial Dysostosis and Related Disorders Panel	MA0201	27
Gastrointestinal Atresia Panel	MA2801	15
Heterotaxy and Situs Inversus Panel	CA2301	34
Hirschsprung Disease Panel	MA1801	15
Holoprosencephaly Panel	MA0601	12
Kabuki Syndrome Panel	MA0901	9
Limb Malformations Panel	MA4001	50
Lissencephaly Panel	MA0101	24
Lymphatic Malformations and Related Disorders Panel	MA4101	12
Macrocephaly / Overgrowth Syndrome Panel	MA1401	48
Meier-Gorlin Syndrome Panel	MA0801	6
Metaphyseal Dysplasia Panel	MA2501	12
Microcephaly and Pontocerebellar Hypoplasia Panel	MA0701	78
Micromelic Dysplasia Panel	MA1901	27
Neurofibromatosis Panel	MA1501	9
Neuronal Migration Disorder Panel	MA2601	59
Osteogenesis Imperfecta Panel	MA3001	30
Osteopetrosis and Dense Bone Dysplasia Panel	MA2001	25
Polymicrogyria Panel	MA0401	20
Seckel Syndrome Panel	MA0301	6
Septo-Optic Dysplasia Panel	MA2201	4
Short Rib Dysplasia / Asphyxiating Thoracic Dysplasia Panel	MA1101	18
Skeletal Dysplasia with Abnormal Mineralization Panel	MA1301	34
Skeletal Dysplasias Core Panel	MA3501	113
Spondylometaphyseal / Spondyloepi-(meta)-physeal Dysplasia Panel	MA1701	30
Vascular Malformations Panel	MA4201	14

Metabolic Disorders Panels	Test Code	Genes
Aicardi-Goutières Syndrome Panel	ME1201	7
Coenzyme q10 Deficiency Panel	ME0801	15
Comprehensive Metabolism Panel	ME0701	439
Congenital and Familial Lipodystrophy Panel	ME1001	12
Congenital Disorders of Glycosylation Panel	ME1901	48
Congenital Mono- and Disaccharide Disorders Panel	ME2301	9

Metabolic Disorders Panels	Test Code	Genes
Creatine Metabolism Deficiency Panel	ME1301	3
Cystinuria Panel	ME1801	2
Fatty Acid Oxidation Syndrome Panel	ME1701	26
Glycogen Storage Disorder Panel	ME0301	29
Hereditary Hemochromatosis Panel	ME1101	5
Homocystinuria Core Panel	ME2201	4
Hyperammonemia and Urea Cycle Disorder Panel	ME1601	49
Hyperphenylalaninemia Panel	ME2001	6
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	ME0601	50
Hypomagnesemia Panel	ME0501	19
Lysosomal Disorders and Mucopolysaccharidosis Panel	ME1501	102
Metabolic Liver Failure Panel	ME2501	16
Metabolic Myopathy and Rhabdomyolysis Panel	ME1401	53
Mitochondrial DNA Depletion Syndrome Panel	ME0201	25
Monogenic Obesity Panel	KI1701	41
Nephrolithiasis Panel	KI2201	35
Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel	ME2601	9
Organic Acidemia/Aciduria & Cobalamin Deficiency Panel	ME0901	54
Periodic Paralysis Panel	ME2101	4
Peroxisomal Disorders Panel	ME0401	27
Porphyria Panel	ME0101	9
Purine and Pyrimidine Metabolism Disorders Panel	ME2401	21
Tyrosinemia Panel	ME2701	3

Nephrology Panels	Test Code	Genes
Alport Syndrome Panel	KI1101	6
Bardet-Biedl Syndrome Panel	KI0201	27
Bartter Syndrome Panel	KI0601	10
Branchio-Oto-Renal (BOR) Syndrome Panel	KI1501	4
Ciliopathy Panel	KI0701	107
Cystic Kidney Disease Panel	KI0901	41
Diabetes Insipidus Panel	KI1801	3
Hemolytic Uremic Syndrome Panel	KI0101	9
Hypomagnesemia Panel	ME0501	19
Hypophosphatemic Rickets Panel	KI1301	13
Joubert Syndrome Panel	KI1001	36
Liddle Syndrome Panel	CA1401	2
Meckel Syndrome Panel	KI1601	13
Monogenic Obesity Panel	KI1701	41
Nephrolithiasis Panel	KI2201	35
Nephronophthisis Panel	KI1901	20
Nephrotic Syndrome Panel	KI0401	44
Polycystic Kidney Disease Panel	KI2101	12
Primary Ciliary Dyskinesia Panel	KI1201	41
Primary Hyperoxaluria Panel	KI0801	3
Pseudohypoaldosteronism Panel	KI1401	10
Renal Malformation Panel	KI2001	27
Renal Tubular Acidosis Panel	KI0301	5
Senior-Loken Syndrome Panel	KI0501	9

Neurology Panels	Test Code	Genes
Autism Spectrum Disorders Panel	NE0101	38
Amyotrophic Lateral Sclerosis Panel	NE2201	35
Ataxia Panel	NE2101	173
Beyond Paediatric Epilepsy Panel – for Europe and Middle East	NE2401	379
Cerebral Cavernous Malformation Panel	MA1001	4
Charcot-Marie-Tooth Neuropathy Panel	NE1301	105
Coenzyme q10 Deficiency Panel	ME0801	15
Collagen Type VI-Related Disorders Panel	NE1401	6
Comprehensive Epilepsy Panel	NE1001	379
Comprehensive Muscular Dystrophy / Myopathy Panel	NE0701	88
Congenital Myasthenic Syndromes Panel	NE1701	21
Creatine Metabolism Deficiency Panel	ME1301	3
Dementia Panel	NE2301	21
Dystonia Panel	NE2501	31
Emery-Dreifuss Muscular Dystrophy Panel	NE0301	6
Epileptic Encephalopathy Panel	NE0401	166
Holoprosencephaly Panel	MA0601	12
Idiopathic Generalized and Focal Epilepsy Panel	NE1101	35
Leukodystrophy and Leukoencephalopathy Panel	NE2001	81
LGMD and Congenital Muscular Dystrophy Panel	NE0801	56
Lissencephaly Panel	MA0101	24
Macrocephaly / Overgrowth Syndrome Panel	MA1401	48
Metabolic Epilepsy Panel	NE1601	47
Metabolic Myopathy and Rhabdomyolysis Panel	ME1401	53
Microcephaly and Pontocerebellar Hypoplasia Panel	MA0701	78
Migraine Panel	NE1201	10
NCL and Progressive Myoclonic Epilepsy Panel	NE1901	31
Nemaline Myopathy Panel	NE0201	13
Neuro-Ophthalmology Panel	OP1301	50
Neuronal Migration Disorder Panel	MA2601	59
Parkinson Disease Panel	NE1501	25
Periodic Paralysis Panel	ME2101	4
Polymicrogyria Panel	MA0401	20
Porphyria Panel	ME0101	9
Septo-Optic Dysplasia Panel	MA2201	4
Spastic Paraplegia Panel	NE0501	75
Spinal Muscular Atrophy Panel	NE1801	30
Tuberous Sclerosis Panel	ON1401	2
X-linked Intellectual Disability Panel	NE0601	106

Ophthalmology Panels	Test Code	Genes
Achromatopsia Panel	OP0501	8
Albinism Panel	DE0801	23
Bardet-Biedl Syndrome Panel	KI0201	27
Cataract Panel	OP0201	76

Ophthalmology Panels	Test Code	Genes
Cone Rod Dystrophy Panel	OP0401	44
Congenital Stationary Night Blindness Panel	OP1201	20
Corneal Dystrophy Panel	OP1601	29
Ectopia Lentis Panel	OP1801	14
Flecked Retina Disorders Panel	OP1401	12
Glaucoma Panel	OP1001	19
Hereditary Retinoblastoma Panel	ON1701	1
Joubert Syndrome Panel	KI1001	36
Leber Congenital Amaurosis Panel	OP1701	28
Macular Dystrophy Panel	OP0101	28
Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel	OP0601	61
Neuro-Ophthalmology Panel	OP1301	50
Optic Atrophy Panel	OP0301	31
Retinal Dystrophy Panel	OP0801	285
Retinitis Pigmentosa Panel	OP0901	116
Senior-Loken Syndrome Panel	KI0501	9
Septo-Optic Dysplasia Panel	MA2201	4
Stickler Syndrome Panel	OP1501	8
Usher Syndrome Panel	OP1101	21
Vitreoretinopathy Panel	OP0701	24

Pulmonology Panels	Test Code	Genes
Bronchiectasis Panel	PU0201	22
Central Hypoventilation and Apnea Panel	PU0401	15
Comprehensive Pulmonology Panel	PU0701	77
Cystic Fibrosis Panel	PU0601	1
Cystic Lung Disease Panel	PU0101	8
Hermansky-Pudlak Syndrome Panel	HE1101	23
Interstitial Lung Disease Panel	PU0301	30
Neonatal Respiratory Distress - Surfactant Dysfunction Panel	PU0501	5
Primary Ciliary Dyskinesia Panel	KI1201	41
Pulmonary Artery Hypertension (PAH) Panel	CA0601	23

Whole Exome tests

Whole Exome Sequencing	Test Code	Analyses
Whole Exome Plus	WE0301	Index patient +parents
Whole Exome Family Plus	WE0401	Index patient +parents

Variant Specific Testing

Blueprint Genetics offers two Variant Specific Testing products: Familial Variant Testing and Targeted Variant Testing. 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 larification of variant classification for variants reported by another laboratory.

Customization of Panels

All our over 220 Sequencing Panels can be customized by adding adding any of our over 4000 clinically relevant genes or by removing genes from the selected panel.

Expand to Exome

If previous genetic testing results are inconclusive, Expand to Exome allows you to expand from a Single Gene or Panel order to Exome Plus or Exome Family Plus. Available for orders placed after April 2018.

WES Re-evaluation and Re-analysis Services

WES Re-evaluation service reviews all available scientific information in an attempt to re-classify 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

Nucleus Connecting Clinicians

This professional network allows you to connect with clinicians from all over the world based on rare matching variants (a match is made on the exact position, or +/- 1 or +/- 2 base pairs). Connecting Clinicians provides the opportunity to share phenotypes in rare cases of VUS (variant of unknown significance) or Likely Pathogenic variants.

- After receiving the results through our online ordering portal, Nucleus, a connection is suggested when a variant is classified as a VUS or Likely Pathogenic.

Contact details:

Customer Support

The team will help you in 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

blueprintgenetics.com