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NERVOUS SYSTEM

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with the Nervous System.

Clinical Conditions | Diseases | Syndromes 46

- Amyotrophic Lateral Sclerosis (ALS)
- Ataxia
- Autism Spectrum Disorders
- Bethlem Myopathy
- Cerebral Cavernous Malformation
- Cerebral Creatinine Deficiency Syndrome
- Charcot-Marie-Tooth
- Coenzyme Q10 Deficiency
- Congenital Muscular Dystrophy
- Congenital Myasthenic Syndrome
- Dementia
- Dystonia
- Emery-Dreifuss Muscular Dystrophy
- Epilepsy
- Epileptic Encephalopathy
- Focal Epilepsy
- Holoprosencephaly
- Idiopathic Generalized Epilepsy
- Leukodystrophy
- Leukoencephalopathy
- Limb-girdle Muscular Dystrophies
- Lissencephaly
- Macrocephaly
- Metabolic Epilepsy
- Metabolic Myopathies
- Microcephaly
- MIgrane
- Muscular Dystrophy
- Muscular Myopathy
- Nemaline Myopathy
- Neuronal Ceroid Lipofuscinoses (NCL)
- Neuronal Migration Disorder
- Overgrowth Syndromes
- Parkinson's Disease
- Periodic Paralysis
- Polymicrogyria
- Pontocerebellar Hypoplasia
- Porphyria
- Progressive Myoclonic Epilepsy
- Rhabdomyolysis
- Septo-Optic Dysplasia
- Spastic Paraplegia
- Spinal Muscular Atrophy
- Tuberous Sclerosis
- Ullrich Congenital Muscular Dystrophy
- X-Linked Intellectual Disability

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HEMATOLOGICAL SYSTEM

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with the Hematological System.

Clinical Conditions | Diseases | Syndromes 18

- Anemia
- Bleeding Disorders
- Bone Marrow Failure Syndromes
- Coagulation Factor Deficiencies
- Coagulopathies
- Congenital Neutropenia
- Cytopenia
- Diamond-Blackfan Anemia
- Dyskeratosis Congenita
- Fanconi Anemia
- Hemophagocytic Lymphohistiocytosis
- Herediatary Lukemia
- Hermansky-Pudlak Syndrome
- Immunohematological Disorders
- Inherited Hematological Diseases
- Platelet Function Disorders
- Red Blood Cell Membrane Disorders
- Thrombocytopenia

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IMMUNE SYSTEM

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with the Immune System.

Clinical Conditions | Diseases | Syndromes 11

- Autoinflammatory Syndromes
- Bone Marrow Failure Syndromes
- Chronic Granulomatous Disease
- Complement System Disorders
- Immunohematological Disorders
- Congenital Neutropenia
- Dyskeratosis Congenita
- Hemophagocytic Lymphohistiocytosis
- Primary Immunodeficiencies
- Primary Ciliary Dyskinesia
- Severe Combined Immunodeficiency

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URINARY SYSTEM

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with the Urinary System

Clinical Conditions | Diseases | Syndromes 21

Alport Syndrome
Bardet-Biedl Syndrome
Bartter Syndrome
Branchiootorenal Syndrome
Ciliopathies
Cystic Kidney Disease
Diabetes Insipidus
Familial Hypomagnesemia with Secondary Hypocalcemia
Hemolytic Uremic Syndrome
Hypophospatemic Rickets
Joubert Syndrome
Liddle Syndrome
Meckel Syndrome
Nephrolithiasis
Nephrotic Syndrome
Polycystic Kidney
Primary Ciliary Dyskinesia
Primary Hyperoxaluria
Renal Malformations
Renal Tubular Acidosis
Senior Loken Syndrome

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GASTROINTESTINAL SYSTEM

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with the Gastrointestinal System.

Clinical Conditions | Diseases | Syndromes 7

Cholestasis
Congenital Diahrea
Congenital Hepatic Fibrosis
Gastrointestinal Atresia
Hirschprung's Disease
Pancreatitis
Polycystic Liver Disease

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ONCOLOGY

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with several Oncological Conditions.

Clinical Conditions | Diseases | Syndromes 19

- Breast Cancer
- Colorectal Cancers
- Endocrine Cancers
- Gastrointestinal Cancers
- Gynaecological Cancers
- Hepatic Cancers
- Leukemia
- Lung Cancers
- Melanoma
- Neurofibromatosis
- Ophthalmic cancers
- Pancreatic Cancers
- Paraganglioma
- Paediatric Cancers
- Pheochromocytoma
- Renal Cancers
- Skin Cancers
- Tuberous Sclerosis
- Xeroderma Pumentosum

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EAR , NOSE & THROAT (ENT)

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with the Ear, Nose, and Throat.(ENT)

Clinical Conditions | Diseases | Syndromes 10

Alport Syndrome
Branchio-Oto-Renal Syndrome
Deafness
Hearing Loss
Hereditary Haemorrhagic Telangiectasia
Non Syndromic Hearing Loss
Pandred Syndrome
Stickler Syndrome
Usher Syndrome
Waardenburg Syndrome

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OPHTHALMOLOGY

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with Ophthalmology.

Clinical Conditions | Diseases | Syndromes 23

- Achromatopsia
- Albinism
- Anophthalmia
- Anterior Segment Dysgenesis
- Bardet Biedl Syndrome
- Cataract
- Conditional Stationary Night Blindness
- Cone Rod Dystrophies
- Corneal Dystrophies
- Ectopia Lentis
- Flecked Retina Disorders
- Glaucoma
- Joubert Syndrome
- Leber Congenital Amaurosis
- Macular Dystrophy
- Microphthalmia
- Optic Atrophy
- Retinal Dystrophy
- Retinitis Pigmentosa
- Septo Optic Dysplasia
- Stickler Syndrome
- Usher Syndrome
- Viteroretinopathy

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ORTHODONTOLOGY

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with Orthodontolgy.

Clinical Conditions | Diseases | Syndromes 6

Anodontia
Ameliogenesis Imperfecta
Cleft Lip and Palate
Dentinogenesis Imperfecta
Dentin Dysplasia
Hypodontia

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ENDOCRINE SYSTEM

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with the nervous system.

Clinical Conditions | Diseases | Syndromes 14

Congenital Adrenal Hyperplasia
Disorders of Sex Development
Glucocorticoid Deficiency
Hyperinsulinism
Hyperlipidaemia
Hyperparathyroidism
Hypoglycaemia
Hypomagnesaemia
Hypothyroidism
Kallmann Syndrome
Maturity-Onset Diabetes of the Young (mody)
Monogenic Diabetes
Primary Congenital Hypothyroidism
Premature Ovarian Failure

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CARDIOVASCULAR SYSTEM

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with the Cardiovascular System.

Clinical Conditions | Diseases | Syndromes 23

Aortic diseases
Heart arrhythmia
Arrhythmogenic right ventricular dysplasia / cardiomyopathy
Atrial fibrillation
Brugada syndrome
Cardiomyopathies
Catecholaminergic polymorphic ventricular tachycardia
Congentital heart defects
Dilated cardiomyopathy
Ehlers-Danlos Syndrome
Hereditary haemorrhagic telangiectasia
Heterotaxy syndrome
Hyperlipidaemia
Hypertrophic cardiomyopathy
Left ventricular noncompaction
Liddle's syndrome
Long QT syndrome
Marfan syndrome
Noonan syndrome
Pulmonary arterial hypertension
Short QT syndrome

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MITOCHONDRIAL DISORDERS

Our unique system based approach allows us to analyze **over 90 clinically actionable genes** in addition to more robustly associated genes with Mitochondrial Disorders.

Clinical Conditions | Diseases | Syndromes 36

Metabolic Disorders
- Metabolic Myopathy
- Rhabdomyolysis

Cardiology
- Cardiomyopathy
- Hypertrophic Cardiomyopathy

Mitochondrial Disorders
- Mitochondrial Depletion Syndrome
- Mitochondrial Diseases

Pulmonology
- Inherited Pulmonary Diseases

Nephrology
- Nephrotic Syndrome

Endocrinology
- Monogenic Diabetes
- Maturity-onset Diabetes of the Young (MODY)

Ear, Nose & Throat
- Deafness
- Hearing Loss
- Non-Syndromic Hearing Loss
- Syndromic Hearing Loss

Neurology
- Autism Spectrum Disorders
- Ataxia
- Charcot-Marie-Tooth Disease
- Dementia
- Dystonia
- Epilepsy
- Epileptic Encephalopathies
- Myopathy
- Muscular Dystrophies
- Leukodystrophy
- Leukoencephalopathy
- Metabolic Epilepsy
- Metabolic Myopathy
- Migraine
- Neuro-Ophthalmological Disorders
- Parkinson Disease
- Rhabdomyolysis

Ophthalmology
- Cataract
- Neuro-Ophthalmological Disorders
- Optic Atrophy
- Retinal Dystrophies
- Retinitis Pigmentosa

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OBSTETRICS & GYNAECOLOGY

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with Obstetrics and Gynaecology conditions

Clinical Conditions | Diseases | Syndromes 5

Diagnostic and Screening
All conditions affecting womens' health (System Based)
Carrier Screening
New-born Screening
Prenatal Genetic Testing (NIPT)
Reproductive Health

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DERMATOLOGY

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with Dermatology.

Clinical Conditions | Diseases | Syndromes 18

Acrodermatitis Enteropathica
Adams-Oliver Syndrome
Albinism
Cutis Laxa
Dyskeratosis Congenita
Ectodermal Dysplasias
Ehlers–Danlos Syndrome
Epidermolysis Bullosa
Hermansky-Pudlak Syndrome
Hutchinson-Gilford Progeria Syndrome
Ichthyosis
Neurofibromatosis
Pachyonychia Congenita
Palmoplantar Keratoderma
Skin Cancer
Tuberous Sclerosis
Waardenburg Syndrome
Xeroderma Pigmentosum

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PULMONOLOGY

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes with Pulmonology.

Clinical Conditions | Diseases | Syndromes 8

Bronchiectasis
Cystic Lung Diseases
Hermansky-Pudlak Syndrome
Idiopathic Congenital Central Hypoventilation Syndrome
Interstitial Lung Diseases
Primary Ciliary Dyskinesia
Pulmonary Artery Hypertension
Respiratory Distress Syndrome

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MALFORMATIOND

Our unique system based approach allows us to analyze over **90 clinically actionable genes** in addition to more robustly associated genes Malformations.

Clinical Conditions | Diseases | Syndromes 48

- Adams Oliver Syndrome
- Amelogenesis Imperfecta
- Arthrogryposes
- Asphyxiating Thoracic Dysplasia
- Brachydactyly
- Cerebral Cavernous Malformations
- Chondrodysplasia Punctata
- Cleft Lip
- Clift Palate
- Cornelia De Lange Syndrome
- Craniosynostosis
- Dense Bone Dysplasia
- Dentogenesis Imperfecta
- Facial Dysostosis
- Gastrointestinal Atresias
- Growth Disorders
- Hereditary Multiple Exostosis
- Heterotaxy
- Hirschsprung Disease
- Holoprosencephaly
- Hypophosphatasia
- Kabuki Syndrome
- Limb Malformations
- Lissencephaly
- Lymphatic Malformations
- Macrocephaly
- Meir-Gorlin Syndrome
- Metaphyseal Dysplasia
- Microcephaly
- Micromelic Dysplasia
- Neurofibromatosis
- Neuronal Migration Disorders
- Osteogenesis Imperfecta
- Osteoperosis
- Overgrowth Syndrome
- Polymicrogyria
- Pontocerebellar Hypoplasia
- Primordial Dwarfism
- Seckel Syndromr
- Septo-Optic Dysplasia
- Short Rib Dysplasia
- Short Stature Syndromes
- Situs Inversus
- Skeletal Dysplasias
- Spondylometaphyseal Dysplasia
- Syndactyly
- Three M Syndrome
- Vascular Malformations