PEDIATRIC GENETIC TESTING

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Pediatric genetic testing involves examining a child's DNA to identify genetic mutations or variations that may be linked to certain diseases or conditions.

More than 7000 rare diseases have been clinically defined, and a significant majority, about 75%, impact children.

Obtaining an early and precise molecular diagnosis can significantly enhance outcomes for children affected by rare diseases.

This type of testing can help diagnose genetic disorders early, guide treatment decisions, and provide valuable information for families about the risk of passing on genetic conditions to future generations. It's often recommended when there's a family history of genetic disorders or when a child shows symptoms that suggest a genetic condition.

EARLY DIAGNOSIS

TREATMENT PLANNING

FAMILY PLANNING

PREVENTIVE MEASURES

COMMON PEDIATRIC GENETIC DISORDERS

Neurodevelopmental Disorders

Congenital Heart Diseases

Metabolic Disorders

Muscular Disorders

Sensory Disorders

Cystic Fibrosis

Inborn Error of Metabolisms

Chromosomal Disorders

Rare Genetic Syndromes

INDICATIONS

- Abnormal New Born Screening Results
- Family History of Genetic Disorders
- Clinical Suspicion of a Genetic Disorder
- Abnormality detected on Prenatal Ultrasound

NGS TESTS

Clinical Exome Test

- Clinical Exome is a custom focused exome curated in-house by experts at Anderson and covers 6161 genes encompassing both nuclear and mitochondrial genes.
- This exome provides better coverage of disease associated genes including coding variants, splice variants.
- It gives very high diagnostic utility at a low cost compared to whole exome sequencing Clinical Exome Panel is enriched for disease associated genes (with strong evidence) and genes with limited but emerging evidence from OMIM, Orphanet and other sources.

Whole Exome Test

- Uniform coverage across exome region with a mean depth of >80-100X.
- More than 98% of targeted base pairs covered at ≥10x
- All protein-coding regions along with the intron-exon boundary regions of ~24383 genes
- (including autosomal recessive, dominant and X-linked) and nuclear encoded mitochondrial
- genes
- Comprehensive detection and analysis of SNVs
- Requisite quality control steps throughout the workflow from the laboratory sample
- processing till the interpretation ensures consistency, validity and accuracy of results.

Cardiology

Comprehensive Cardiology Panel **(260 genes)**
Long QT Syndrome (LQTS) Panel **(18 genes)**
Pulmonary Artery Hypertension (PAH) Panel **(23 genes)**
Congenital Structural Heart Disease Panel **(125 genes)**
Noonan Syndrome Panel **(36 genes)**

Dermatology

Ectodermal Dysplasia Panel **(25 genes)**
Ichthyosis Panel **(39 genes)**
Tuberous Sclerosis Panel **(2 genes)**
Epidermolysis Bullosa Panel **(26 genes)**
Neurofibromatosis Panel **(9 genes)**

Sensory Panel

Branchio-Oto-Renal (BOR) Syndrome Panel **(4 genes)**
Non-Syndromic Hearing Loss Panel **(138 genes)**
Stickler Syndrome Panel **(8 genes)**
Comprehensive Hearing Loss and Deafness Panel **(288 genes)**
Pendred Syndrome Panel **(3 genes)**
Waardenburg Syndrome Panel **(7 genes)**

Immunology

Bone Marrow Failure Syndrome **(156 genes)**
Comprehensive Immune and Cytopenia Panel
Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia **(383 genes)**
Primary Immunodeficiency Panel **(336 genes)**
Severe Combined Immunodeficiency Panel **(80 genes)**
Chronic Granulomatous Disease Panel **(8 genes)**

Nephrology

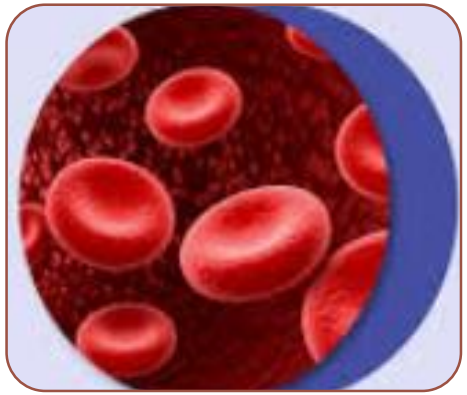
Bartter Syndrome **(10 genes)**
Ciliopathy panel **(119 genes)**
Cystic Kidney Disease **(43 genes)**
Joubert Syndrome **(36 genes)**
Primary Ciliary Dyskinesia **(47 genes)**
Renal Malformation **(27 genes)**
Polycystic Kidney Disease **(13 genes)**
Hypophosphatemic Rickets **(13 genes)**

Malformations

Cleft lip/Palate and Associated Syndromes **(22 genes)**
Comprehensive Short Stature Syndrome **(100 genes)**
Macrocephaly/Overgrowth Syndrome **(48 genes)**
Craniosynostosis **(38 genes)**
Osteogenesis Imperfecta **(33 genes)**
Comprehensive Growth Disorders/ Skeletal Dysplasias **(510 genes)**

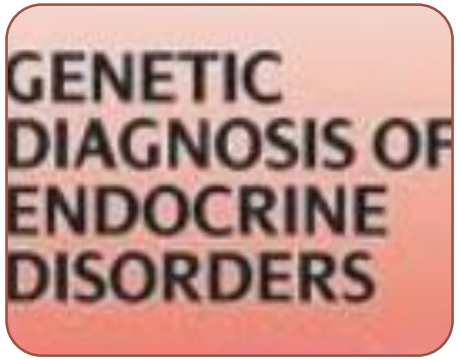
Neurology

Comprehensive Epilepsy Panel **(511 genes)**
Epileptic Encephalopathy Panel **(203 genes)**
Comprehensive Muscular Dystrophy/Myopathy Panel **(161 genes)**
Neuronal Migration Disorder Panel **(59 genes)**
Leukodystrophy and Leukoencephalopathy **(118 genes)**



Hematology

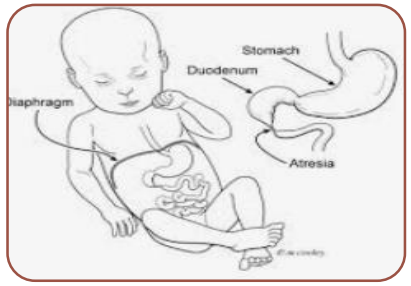
Bleeding disorder /Coagulopathy Panel **(71 genes)**
Bone Marrow Failure Syndrome **(156 genes)**
Comprehensive Immune and Cytopenia Panel
Diamond-Blackfan Anemia Panel **(14 genes)**
Hereditary Leukemia Panel **(42 genes)**
Congenital Neutropenia Panel **(28 genes)**
Fanconi Anemia Panel **(24 genes)**



Endocrinology

Abnormal Genitalia / Disorders of Sex Development Panel **(73 genes)**
Hypothyroidism and Resistance to Thyroid Hormone Panel **(22 genes)**
MODY Panel **(54 genes)**
Comprehensive Monogenic Diabetes Panel **(67 genes)**
Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel **(50 genes)**

Gastroentology



Cholestasis **(52 genes)**
Gastrointestinal Atresia **(15 genes)**
Congenital Diarrhea **(29 genes)**
Pancreatitis **(9 genes)**
Hirschsprung Disease **(15 genes)**

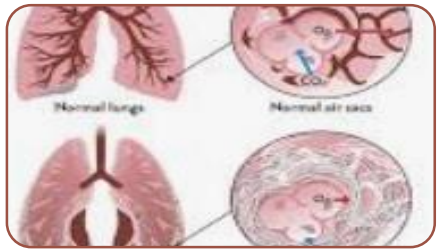
Ophthalmology

Albinism **(26 genes)**
Cataract **(113 genes)**
Bardet-Biedl Syndrome **(27 genes)**
Retinal Dystrophy **(351 genes)**
Leber Congenital Amaurosis **(28 genes)**
Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel **(61 genes)**

Metabolic Disorders

Comprehensive metabolic panel **(505 genes)**
Glycogen storage disorder **(29 genes)**
Hyperphenylalaninemia **(6 genes)**
Lysosomal disorders and Mucopolysaccharidosis **(102 genes)**
Fatty Acid Oxidation Syndrome **(26 genes)**
Peroxisomal Disorders **(27 genes)**

Pulmonology

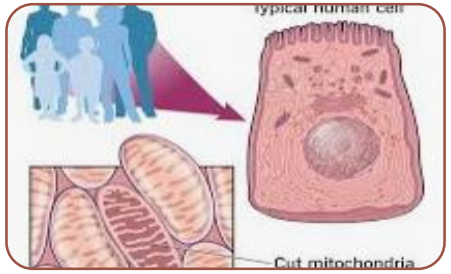


Bronchiectasis Panel **(22 genes)**
Central Hypoventilation and Apnea Panel **(15 genes)**
Interstitial Lung Disease Panel **(30 genes)**
Neonatal Respiratory Distress – Surfactant Dysfunction Panel **(5 genes)**

Hereditary Cancer

Hereditary Leukemia Panel **(42 genes)**
Hereditary Pediatric Cancer Panel **(71 genes)**
Neurofibromatosis Panel **(9 genes)**
Tuberous Sclerosis Panel **(2 genes)**

Mitochondrial Disorders

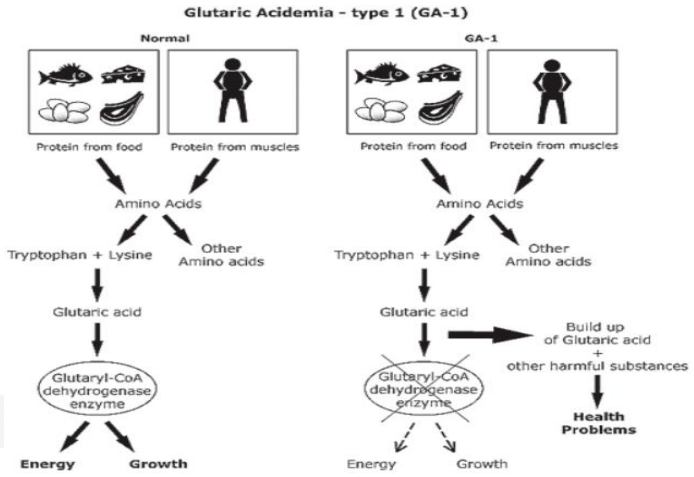


Mitochondrial DNA depletion syndrome Panel **(62 genes)**

CASE 1

Proband (Male/5 years), born to consanguineous parents, presented with chief complaints of difficulty in walking, trembling and slurry speech. He was suspected to be affected with Type 1 Glutaric aciduria.

Clinical Exome Sequencing revealed likely pathogenic homozygous missense variation in Exon 9 of the *GCDH* (p.Ala293Thr) gene associated with Glutaric aciduria, type I



CASE 2

Proband (Male/5 years) born to consanguineous parents, presented with skin lesions at birth, microcephaly, and corneal clouding. He had multiple bullae over both elbows, knees, oral cavity, and dorsum of feet, mild scarring with milia present over chest post-inflammatory hypopigmentation, normal fingernails, and total dystrophy of the right great toe. Skin immunopathology test revealed dystrophic epidermolysis bullosa. He has mitten hands and feet and an absence of speech at 6 years of age.

Whole Exome Sequencing revealed Pathogenic homozygous truncation variation in exon 110 of the *COL7A1*(p.Arg2685Ter) gene associated with Epidermolysis bullosa.

Cases	Clinical Indications	Test	Variant
1) Male/ 7 Months	focal epilepsy Abnormal EEG	Whole Exome Sequencing	***PRRT2*** **(p. Arg217GlnfsTer12) Heterozygous Dominant Likely Pathogenic**
2) Male/5 years (consanguineous)	difficulty in walking, trembling and slurry speech Suspected - Type 1 Glutaric aciduria	Clinical Exome Sequencing	***GCDH*** **(p.Ala293Thr) Homozygous Recessive Likely Pathogenic**
3) Male/5 years (consanguineous)	skin lesions at birth, microcephaly, corneal clouding, multiple bullae over both elbows, knees, oral cavity, dorsum of feet, mild scarring with milia present over chest post-inflammatory hypopigmentation, Mitten hands and feet, Absent speech Skin Immunopathology – Epidermolysis Bullosa	Whole Exome Analysis	***COL7A1*****(p.Arg2685Ter) Homozygous Recessive Pathogenic**
4) Male/ 1 Day (Non-consanguineous)	white body hair, light brownish eye color and baby pink color of genitalia Family history present Suspected - Albinism	Whole Exome Analysis	***TYR*** **(p.Arg278Ter) Homozygous Recessive Likely Pathogenic**