#Doenças Genéticas#

Através da técnica de DPI é possível detectarmos diversas doenças genéticas. O Genesis Genetics Institute foi o primeiro a realizar esse teste e é mundialmente conhecido por sua habilidade em testar as desordens genéticas mais raras. As famílias afetadas por praticamente qualquer desordem genética conhecida podem reduzir o risco de terem filhos afetados realizando os testes necessários.

Lista de Desordens Genéticas Detectáveis:

Aarskog (X-FGD1)
Achondroplasia (FGFR3)
Actin-Nemalin Myopathy (ACTA1)
Adenomatous Polyposis Coli (FAP-APC)
Adrenoleukodystrophy (ABCD1)
Agammaglobulinemia-Bruton (BTK)
Alagille Syndrome (JAG1)
Aldolase A deficiency (ALDOA)
Alpha Thalassemia (HBA1)
Alpha Thalassemia/Mental Retard (ATRX)
Alpha-1-Antitrypsin Deficiency (AAT)Alport Syndrome (COL4A5)
ALS: Amyotrophic Lateral Sclerosis 1, (SOD1)
Alzheimer Disease 3 (PSEN1)
Amegakaryocytic Thrombocytopenia, Congenital (CAMT)
Amyloidosis I-Transthyretin (TTR)
Angioedema, Hereditary (C1NH)
Ankylosing spondylitis (Susceptibility to, HLA-B27)
Antithrombin Deficiency (SERPINC1)
Apert Syndrome (FGFR2)
Ataxia Telangiectasia (ATM)
Basal Cell (Gorlin) Synd (PTCH)
Beta Thalassemia (HBB)
Bloom Syndrome (BLM)
Brachydactyly-Type C (GDF5)
Breast Cancer (BRCA1 & 2)
CACH-Ataxia (EIF2B4)
CADASIL (Notch3)
Canavan Disease (ASPA)
Cardiomyopathy, Barth Type Dilated (TAZ)
Cardiomyopathy, Dilated Hypertrophic (MYH7)
Dilated Hypertrophic Cardiomyopathy MYH7
Carnitine-AcylCarn Translocase (SLC25A20)
Ceroid-Lipofuscinoses-Batten Disease (PPT1)
Ceroid-Lipofuscinoses-Finish Type (CLN5)
Ceroid-Lipofuscinoses-Juvenile Type (CLN3)
Charcot Marie Tooth 1A (PMP22)
Charcot Marie Tooth Neuropathy - 2E, (NF-L, NEFL)
Charcot-Marie-Tooth neuropathy 1B (MPZ)
Cherubism (SH3BP2)
Choroideremia (CHM)
Chronic Granulomatous Disease (CYBB)
Citrullinemia (ASS)
Cleidocranial Dysplasia (RUNX2)
Cockayne syndrome type B (CSB; ERCC6)
Colon Cancer (HNPCC; MSH2)
Congenital Adrenal Hyperplasia (CYP21A2)
Congenital Disorder Glycosylation, 1a - CDG-1a (PMM2)

Lista de Desordens Genéticas Detectáveis:

Congenital Disorder Glycosylation, 1c - CDG-1c (ALG6)
Congenital Disorder Glycosylation, 1e - CDG-1e (DPM1)
Congenital Disorder Glycosylation, 1g - CDG-1g (ALG12)
Congenital Erythropoietic Porphyria (UROS)
Cosman-Cyclic Neutropenia (ELA2)
Crigler Najjar (UGT1A1)
Crouzon Syndrome (FGFR2)
Cystic Fibrosis (CFTR)
Cystinosis (CTNS)
Darier Disease (ATP2A2)
Deafness, Recessive - (GJB2 Connexin 26)
Deafness, Recessive - (GJB6 Connexin 30)
Deafness, Recessive (DFBN1)
Denys-Drash Wilms Tumor (Wt1)
Desmin Storage Myopathy (DES)
Diamond Blackfan (DBA-RPS19)
Diamond Blackfan (DBA2) Not RPS19
Duchenne muscular dystrophy (DMD)
Dystonia (TOR1A)
Dystrophia Myotonica-1 (DMPK) CTGrpt
Dystrophia Myotonica-2 (DM2; PROMM) CCTGrpt
Ectodermal Dysplasia I EDA1
Ehlers-Danlos COL3A1
Emery-Dreifuss X-Linked Muscular Dystrophy
Emery-Dryfuss AutoDom Muscular Dystrophy (LMNA)
Epidermolysis Bullosa (KRT5)
Epidermolysis Bullosa Simplex KRT14
Epidermolysis Bullosa/Pyloric Atresia - ITGB4
Epidermolysis Dystrophic Bullosa-COL7A1
Epidermolytic Hyperkeratosis (KRT10)
Fabry (GLA)
Facioscapulohumeral Dystrophy (FSHD)
Factor 13 Deficiency (F13A1)
Familial Dysautonomia (IKBKAP)
Familial Exudative Vitreoretinopathy FZD4
Fanconi Anemia A (FANCA)
Fanconi Anemia C (FANCC)
Fanconi Anemia F (FANC F)
Fanconi Anemia J (FANCJ, BRIP1)
Fanconia Anemia G (FANCG)
Fragile X (FMR1)
Friedreich Ataxia I (FRDA)
Galactosemia (GALT)
Gastric Cancer, Cadherin-E-1 (CDH1)
Gaucher Disease (GBA)
Genotyping-Molecular Signature-Fingerprinting
Glutaric Acidemia 2A (ETFA)
Glycine Encephalopathy GLDC 80% (NKH)
Glycogen Storage Disease I, Von Girke - GSD1a (G6PC)
Glycogen Storage Disease 2, Pompe - GSD2 (GAA)
GM1 Gangliosidosis, Morquio (GLB1)
Hallervorden-Spatz-Pantothenate (PANK2)
Hemophilia A (Factor 8)
Hemophilia B (Factor 9)
Hereditary Hemmorrhagic Telangietasia Type 1 (HHT1)

Lista de Desordens Genéticas Detectáveis:

Histiocytosis, Hemophagocytic Lympho- (HLH; PRF1)
HLA DRBeta1 Class II MHC (HLA DRB1*)
HLA-Histocompatability, Transplantation Matching (HLA)
Hunter syndrome (IDS)
Huntington Disease (HD)
Hurler Syndrome (MPSI-IDUA)
Hydrocephalus:X-Linked L1CAM
Hyper IgM (CD40-ligand; TNFSF5)
Hypokalemic periodic paralysis (SCN4A-HYPP)
Hypophosphatasia (ALPL)
Hypophosphatemic VitD RicketsIcthyosis, X-Steroid Sulf Def
Icthyosis.Congenital, Harlequin (ABCA12)
Incontinentia Pigmenti (NEMO)
KELL Antigen (KEL)
Kennedy-Spinal bulbar (AR)
Krabbe (GALC)
Leber Retinal Congenital Amaurosis-I (GUCY2D)
Leber Retinal Congenital Amaurosis-X (CEP290)
Lesch-Nyhan (HPRT1)
Leukemia, Acute Lymphocytic, Transplantation (ALL)
Leukemia, Acute Myelogenous, Transplantation (AML)
Leukemia, Chronic Myelogenous, Transplantation (CML)
Leukocyte Adhesion Deficiency (ITGB2)
Li-Fraumeni Syndrome (Tp53)
Long-Chain-AcylCoA Dehydrogenase (LCHAD:HADHA)
Lymphedema-Hereditary (FOXC2)
Lymphoproliferative Disorder, X-linked (SH2D1A)
Machado-Joseph Spinocerebellar Ataxia-3 (SCA3)
Macular Dystr-Best Vitelliform (VMD2)
Maple Syurp Urine Dz E1-Beta (BCKDHB)
Marfan Syndrome (FBN1)
Meckel-Gruber Syndrome-3 (MKS3)
Menkes (ATP7A)
Merosin-deficient congenital muscular dystrophy type 1A (MDC1A)
Metachromatic Leukodystrophy (ARSA)Methylcobalamin G Deficiency (MTR)
Methylmalonic Acidemia (MUT)
Mitochondrial Myopathy-Complex I (NDUFS4)
Mucolipidosis 2, I Cell (GNPTAB)
Multiple Endocrine Neoplasia 1 (MEN1)
Multiple Endocrine Neoplasia 2 MEN2 (RET)
Multiple Extostoses (EXT1)
Multiple Extostoses (EXT2)
Myasthenia Gravis (CHRNE)
Myotubular Myopathy X-Linked (MTM)
NEMO immunodeficiency (IKBKG)
Nephrosis - Finnish (NPHS1)
Neurofibromatosis 1 (Nf1)
Neurofibromatosis 2 (Nf2)
Niemann Pick - Type A (SMPD1)
Niemann Pick - Type C (NPC1)
NonKetotic Hyperglycinemia (GLDC)N
oonan (PTPN11)Norrie (NDP)
Occulocutaneous Albinism II- (OCA2)
Occulocutaneous Albinism I, OCA1 (TYR)
Ocular Albinism-X Linked (GPR143)

Lista de Desordens Genéticas Detectáveis:

Oculodentodigital Dysplasia (GJA1)
Optic Atrophy 1 (OPA1)
Ornithine transcarbamylase deficiency (OTC)
Osteogenesis Imper II/IV & Chondrodysplasias(COL1A2)
Osteogenesis Imperfecta I (COL1A1)
Osteopetrosis (CLCN7)
Osteopetrosis (TCIRG1;APT6)
Pachyonychia Congenita (KRT6A)
Pachyonychia Congenita (KRT16A)
Pancreatitis, Chronic Calcific (PRSS1)
Paraganglioma-Nonchromaffin (SDHB)
Pelizaeus-Merzbacher, X-linked (PLP1)
Periventricular Heteropia (FLNA)
Persistent Hyperinsulinemic Hypoglycemia of Infancy (ABCC8)
Pfeiffer Syndrome (FGFR2)
Phenylketonuria PKU (PAH)
Pheochromocytoma (SDHB)
Polycystic Kidney Disease (PKD1)
Polycystic Kidney Disease (PKD2)
Polycystic Kidney Disease, Recessive (PKHD1)
Pompe, Glycogen Storage Disease 2, GSD2 (GAA)
Propionic Acidemia (PCCA)
Pseudohypoparathyroidism 1a (GNAS1)
Retinitis Pigmentosa (RHO)
Retinitis Pigmentosa adRP10 (IMPDH1)
Retinitis Pigmentosa X-linked (RPGR)
Retinoblastoma 1 (Rb1)
Retinoschisis, (Rs1)
Rhesus blood group D (RHD)
Rhizomelic Chondrodysplasia Punctata (RCDP1)
Sacral Agenesis (HLXB9)
Sanfilippo A (MPSIIIA)
Sanfillipo B (MPSIIIB) (NAGLU)
Sathre-Chotzen Craniosynostosis (TWIST)
SCIDX1 (IL2RG)
Severe Comb Immunodef (SCID)
Shwachman-Diamond Syndrome (SBDS)
Sickle Cell (HBB)
Smith-Lemli-Opitz (SLOS)
Sorsby Fundus Dystrophy (TIMP3)
Spinal muscular atrophy SMA (SMN1)
Spinocerebellar Ataxia-1, SCA1 (ATNX1)
Spinocerebellar ataxia-2, SCA2 (ATXN2)
Spinocerebellar Ataxia-3, Machado-Joseph (SCA3)
Spinocerebellar Ataxia-7 (ATXN7)
Spondyloepiphyseal dysplasia, congenital (SEDc)
Steroid Sulfatase Deficiency (STS)
Stomach-Ovarian-Endometrial Cancer (CDH1)
Supravalvular Aortic Stenosis (ELN)
Surfactant-Pulmonary B (SFTPB)
Tay-Sachs (HEXA)
Torsion dystonia (DYT1)
Treacher Collins (TCOF1)
Transplantation-BoneMarrow-StemCell (HLA locus)
Tuberous Sclerosis 1 (TSC1)

Lista de Desordens Genéticas Detectáveis:

Tuberous Sclerosis 2 (TSC2)
VanderWoude-Popliteal Pterygium (IRF6)
Von Hippel-Lindau Disease (VHL)
Waardenburg Syndrome Type II (MITF)
Waardenburg Syndrome-I/III (PAX3)
West Syndrome (ARX)
Wilms Tumor (Wt1)
Wiskott-Aldrich Syndrome (WAS)
Wolman Lipase A (LIPA)
Zellweger Peroxisome Disease (PEX1)