

GENETIC TESTING FOR DONORS-RECIPIENTS - REPRODUCTIVE PANEL

40 GENES TESTED

GENE	CONDITION	SPECIFICATIONS
ABCD1	Adrenoleukodystrophy • Adrenomyeloneuropathy, Adult	◆
AR	Androgen Insensitivity • Hypospadias 1 • Spinal and Bulbar Muscular Atrophy of Kennedy • Androgen insensitivity, partial, with or without breast cancer	◆
ATP7A	Menkes Disease • Occipital Horn Syndrome • Spinal Muscular Atrophy, Distal, 3	◆
ATRX	Intellectual Disability-Hypotonic Facies Syndrome	◆
BTK	Agammaglobulinemia, 1 • Isolated Growth Hormone Deficiency, Type III, with Agammaglobulinemia	◆
CFTR	Cystic Fibrosis	
CHM	Choroideremia	◆
COL4A5	Alport Syndrome 1	◆
CYBB	Chronic Granulomatous Disease • Immunodeficiency 34, Mycobacteriosis	◆
DKC1	Dyskeratosis Congenital	◆
DMD	Duchenne Muscular Dystrophy • Becker Muscular Dystrophy	◆■
EDA	Ectodermal Dysplasia 1, Hypohidrotic	◆
EMD	Emery-Dreifuss Muscular Dystrophy 1	◆
F8	Hemophilia A	◆
F9	Hemophilia B • Thrombophilia 8 due to Factor IX Defect	◆
FMR1	Fragile X Syndrome	◆■
G6PD	Hemolytic Anemia, G6PD Deficient (Favism)	◆
GJB1	Charcot-Marie-Tooth Neuropathy, 1	◆
GJB2	Deafness, Autosomal Recessive 1A	
GJB6	Deafness, Autosomal Recessive 1B • Deafness, Digenic GJB2/GJB6	
GLA	Fabry Disease • Fabry Disease, Cardiac Variant	◆
HBA-1	Thalassemia, Alpha • Hemoglobin H Disease, Nondeletional	■
HBA-2	Thalassemia, Alpha • Hemoglobin H Disease, Deletional and Nondeletional	■
HBB	Sickle Cell Anemia • Thalassemia, Beta	
HEXA	GM2-Gangliosidosis, Several Forms • Tay-Sachs Disease	
IDS	Mucopolysaccharidosis II	◆
IL2RG	Combined Immunodeficiency, Moderate • Severe Combined Immunodeficiency	◆
L1CAM	Hydrocephalus, Congenital, X-linked • MASA Syndrome	◆
MECP2	Encephalopathy, Neonatal Severe • Intellectual Developmental Disorder, Syndromic 13 • Intellectual Developmental Disorder, Lubs Type	◆
MID1	Opitz GBBB Syndrome	◆
MTM1	Myopathy, Centronuclear	◆
NROB1	46 XY Sex Reversal 2, Dosage-Sensitive • Adrenal Hypoplasia, Congenital	◆
OCRL	Dent Disease 2 • Lowe Syndrome	◆
OTC	Ornithine Transcarbamylase Deficiency	◆
PDHA1	Pyruvate Dehydrogenase E1-Alpha Deficiency	◆
PRPS1	Arts Syndrome • Charcot-Marie-Tooth Disease, 5 • Deafness, 1 • Gout, PRPS-related • Phosphoribosylpyrophosphate Synthetase Superactivity	◆
RS1	Retinoschisis	◆
SLC6A8	Cerebral Creatine Deficiency Syndrome 1	◆
SMN1	Spinal Muscular Atrophy 1 • Spinal Muscular Atrophy 2 • Spinal Muscular Atrophy 3 • Spinal Muscular Atrophy 4	■
WAS	Neutropenia, Severe Congenital • Thrombocytopenia • Thrombocytopenia, Intermittent • Wiskott-Aldrich Syndrome	◆

◆ X-Linked Inheritance ■ Other techniques apart from Next Generation Sequencing apply, consult the report for specifications