MEDICOVER GENETICS

GENETIC TESTING FOR DONORS-RECIPIENTS

80 GENES TESTED

GENE	DISORDER	SPECIFICATIONS
ABCC8	Hyperinsulinemic Hypoglycemia, Familial, 1 • Diabetes Mellitus, Permanent Neonatal 3, with or without Neurologic Features	
ABCD1	Adrenoleukodystrophy • Adrenomyeloneuropathy, Adult	◆
AIRE	Autoimmune Polyendocrinopathy Syndrome, Type I, with or without Reversible Metaphyseal Dysplasia	
AR	Androgen Insensitivity • Hypospadias 1 • Spinal and Bulbar Muscular Atrophy of Kennedy	◆
ARSA	Metachromatic Leukodystrophy	
ASPA	Canavan Disease	
ATM	Ataxia Telangiectasia	
ATP7A	Menkes Disease • Occipital Horn Syndrome • Spinal Muscular Atrophy, Distal, 3	◆
ATP7B	Wilson Disease	
ATRX	Intellectual Disability-Hypotonic Facies Syndrome	◆
BBS2	Bardet-Biedl Syndrome 2	
BLM	Bloom Syndrome	
BTK	Agammaglobulinemia 1 • Isolated Growth Hormone Deficiency, Type III, with Agammaglobulinemia	◆
CERKL	Retinitis Pigmentosa 26	
CFTR	Cystic Fibrosis	
CHM	Choroideremia	◆
CLRN1	Usher Syndrome, Type 3A	
COL4A5	Alport Syndrome 1	◆
CYBB	Chronic Ganulomatous Disease • Immunodeficiency 34, Mycobacteriosis	◆
CYP21A2	Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency • Hyperandrogenism, Non-Classic type, due to 21-Hydroxylase Deficiency	
CYP27A1	Cerebrotendinous Xanthomatosis	
DHCR7	Smith-Lemli-Opitz Syndrome	
DKC1	Dyskeratosis Congenita	◆
DLD	Dihydrolipoamide Dehydrogenase Deficiency	
DMD	Duchenne Muscular Dystrophy • Becker Muscular Dystrophy	◆ ■
DYSF	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 2 • Miyoshi Muscular Dystrophy 1 • Myopathy, Distal, with Anterior Tibial Onset	
EDA	Ectodermal Dysplasia 1, Hypohidrotic	◆
ELP1	Dysautonomia, Familial	
EMD	Emery-Dreifuss Muscular Dystrophy 1	◆
F8	Hemophilia A	◆
F9	Hemophilia B • Thrombophilia 8 due to factor IX defect	◆
FANCA	Fanconi Anemia, Complementation Group A	
FANCC	Fanconi Anemia, Complementation Group C	
FKTN	Cardiomyopathy, Dilated, 1X • Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies), Type A, 4 • Muscular Dystrophy-Dystroglycanopathy (Congenital without Impaired Intellectual Development), Type B, 4 • Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 4	
FMR1	Fragile X Syndrome	◆ ■
G6PC	Glycogen Storage Disease Ia	
G6PD	Hemolytic Anemia, G6PD Deficient (Favism)	◆
GALT	Galactosemia	
GBA	Gaucher Disease, Perinatal Lethal • Gaucher Disease, Type I • Gaucher Disease, Type II • Gaucher Disease, Type III • Gaucher Disease Type IIIC	
GJB1	Charcot-Marie-Tooth Neuropathy, 1	◆

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

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GENE	DISORDER	SPECIFICATIONS
GJB2	Deafness, Autosomal Recessive 1A	
GLA	Fabry Disease • Fabry Disease, Cardiac Variant	◆
HBA1	Thalassemia, Alpha • Hemoglobin H Disease, Non-Deletional	■
HBA2	Thalassemia, Alpha • Hemoglobin H Disease, Deletional and Non-Deletional	■
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 • MASA Syndrome	◆
MCOLN1	Mucolipidosis IV	
MECP2	Encephalopathy, Neonatal Severe • Intellectual Developmental Disorder, Syndromic 13 • Intellectual Developmental Disorder, Lubs type	◆
MED17	Microcephaly, Postnatal Progressive, with Seizures and Brain Atrophy	
MID1	Opitz GBBB Syndrome	◆
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	
MTHFR	Homocystinuria due to MTHFR Deficiency	
MTM1	Myopathy, Centronuclear	◆
NDUFS6	Mitochondrial Complex I Deficiency, Nuclear Type 9	
NEB	Arthrogryposis Multiplex Congenita 6 • Nemaline Myopathy 2, Autosomal Recessive	
NR0B1	46 XY Sex Reversal 2, Dosage-Sensitive • Adrenal Hypoplasia, Congenital	◆
OCRL	Dent Disease 2 • Lowe Syndrome	◆
OPA3	3-Methylglutaconic Aciduria, Type III	
OTC	Ornithine Transcarbamylase Deficiency	◆
PAH	Phenylketonuria	
PCDH15	Deafness, Autosomal Recessive 23 • Usher Syndrome, Type 1D/F Digenic • Usher Syndrome, Type 1F	
PDHA1	Pyruvate Dehydrogenase E1-Alpha Deficiency	◆
PEX2	Peroxisome Biogenesis Disorder 5A (Zellweger) • Peroxisome Biogenesis Disorder 5B	
PRPS1	Arts Syndrome • Charcot-Marie-Tooth Disease, 5 • Deafness 1 • Gout, PRPS-related • Phosphoribosylpyrophosphate Synthetase Superactivity	◆
RPE65	Leber Congenital Amaurosis 2 • Retinitis Pigmentosa 20	
RS1	Retinoschisis	◆
SEPSECS	Pontocerebellar Hypoplasia Type 2D	
SLC6A8	Cerebral Creatine Deficiency Syndrome 1	◆
SMN1	Spinal Muscular Atrophy 1 • Spinal Muscular Atrophy 2 • Spinal Muscular Atrophy 3 • Spinal Muscular Atrophy 4	■
SMPD1	Niemann-Pick Disease, Type A • Niemann-Pick Disease, Type B	
TECPR2	Neuropathy, Hereditary Sensory and Autonomic, Type IX, with Developmental Delay	
TMC1	Deafness, Autosomal Recessive 7	
TMEM216	Joubert Syndrome 2 • Meckel Syndrome 2	
TYR	Albinism, Oculocutaneous, Type IA • Albinism, Oculocutaneous, Type IB	
USH2A	Retinitis Pigmentosa 39 • Usher Syndrome, Type 2A	
VPS53	Pontocerebellar Hypoplasia, Type 2E	
WAS	Neutropenia, Severe Congenital • Thrombocytopenia • Thrombocytopenia, Intermittent • Wiskott-Aldrich Syndrome	◆

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