

Pre-Pregnancy Genetic Testing Disease List

The following diseases are covered in Medcan's Pre-Pregnancy Genetic Testing.

ABCC8-Related Hyperinsulinism	Galactosemia	Metachromatic Leukodystrophy
Achondrogenesis Type 1B	Gaucher Disease	Mucopolipidosis IV
Achromatopsia	GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	Muscle-Eye-Brain Disease
Alkaptonuria	Glucose-6-Phosphate Dehydrogenase Deficiency	MYH-Associated Polyposis
Alpha-1 Antitrypsin Deficiency	Glutaric Acidemia Type 1	Niemann-Pick Disease Type A
Andermann Syndrome	Glycogen Storage Disease Type Ia	Niemann-Pick Disease Type C
ARSACS	Glycogen Storage Disease Type Ib	Nijmegen Breakage Syndrome
Aspartylglycosaminuria	Glycogen Storage Disease Type III	Northern Epilepsy
Ataxia With Vitamin E Deficiency	Glycogen Storage Disease Type V	Pendred Syndrome
Ataxia-Telangiectasia	GRACILE Syndrome	Phenylalanine Hydroxylase Deficiency
Autosomal Recessive Polycystic Kidney Disease	Hereditary Fructose Intolerance	Polyglandular Autoimmune Syndrome Type 1
Bardet-Biedl Syndrome, BBS1-Related	Hereditary Thymine-Uraciluria	Pompe Disease
Bardet-Biedl Syndrome, BBS10-Related	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	PPT1-Related Neuronal Ceroid Lipofuscinosis
Beta Thalassemia	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	Primary Hyperoxaluria Type 1
Biotinidase Deficiency	Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	Primary Hyperoxaluria Type 2
Bloom Syndrome	Hexosaminidase A Deficiency	Pycnodysostosis
Canavan Disease	HFE-Associated Hereditary Hemochromatosis	Recessive Multiple Epiphyseal Dysplasia
Carnitine Palmitoyltransferase IA Deficiency	Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	Rhizomelic Chondrodysplasia Punctata Type 1
Carnitine Palmitoyltransferase II Deficiency	Hurler Syndrome	Salla Disease
Cartilage-Hair Hypoplasia	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	Segawa Syndrome
Choroideremia	Hypophosphatasia, Autosomal Recessive	Short Chain Acyl-CoA Dehydrogenase Deficiency
CLN5-Related Neuronal Ceroid Lipofuscinosis	Inclusion Body Myopathy 2	Sickle Cell Disease
Congenital Disorder of Glycosylation Type Ia	Infantile Refsum Disease	Sjogren-Larsson Syndrome
Congenital Disorder of Glycosylation Type Ib	Isovaleric Acidemia	Smith-Lemli-Opitz Syndrome
Congenital Finnish Nephrosis	Krabbe Disease	Spinal Muscular Atrophy
Cystic Fibrosis	Leigh Syndrome, French-Canadian Type	Tay-Sachs Disease
Cystinosis	Limb-Girdle Muscular Dystrophy Type 2E	TPP1-Related Neuronal Ceroid Lipofuscinosis
Diastrophic Dysplasia	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Tyrosinemia Type I
Factor V Leiden Thrombophilia	Maple Syrup Urine Disease Type 1B	Usher Syndrome Type 1F
Factor XI Deficiency	Maple Syrup Urine Disease Type 3	Usher Syndrome Type 3
Familial Dysautonomia	Medium Chain Acyl-CoA Dehydrogenase Deficiency	Wilson Disease
Familial Mediterranean Fever		X-Linked Juvenile Retinoschisis
Fanconi Anemia Type C		
Fumarase Deficiency		