Premium Genetics
qendra e testimit gjenetik

Baby's Health
DNA neonatal screening

GENETIC CONDITIONS

ENDOCRINE

DISEASE Gene	AR	AD	XL
Congenital Adrenal Hyperplasia CYP11B1	●	●	
CYP17A1, HSD3B2, POR, STAR	●		
Congenital Hypothyroidism PAX8, THRA		●	
SLC5A5, TG, TPO, TSHB	●		
TSHR	●	●	
Pendred Syndrome SLC26A4	●		

HEAMOGLOBIN

DISEASE Gene	AR	AD	XL
Beta-Thalassemia HBB	●		
S, Beta-Thalassemia (Sickle Cell Beta-Thalassemia) HBB	●		
S,C Disease (Sickle Cell Disease) HBB	●		
S,S Disease (Sickle Cell Disease, Sickle Cell Anemia) HBB	●		

HEARING LOSS

DISEASE Gene	AR	AD	XL
Non-Syndromic Hearing Loss			
CDH23, MYO15A, OTOF, TMIE, TMPRSS3, TPRN, TRIOBP	●		
GJB2, GJB6, TECTA	●	●	
Syndromic Hearing Loss			
Jervell and Lange-Nielsen Syndrome KCNE1, KCNQ1	●		
Pendred Syndrome SLC26A4	●		
Shah-Waardenburg Syndrome SOX10		●	
Usher Syndrome Type 1C USH1C	●		
Usher Syndrome 1G USH1G	●		
Usher Syndrome Type 2A USH2A	●		
Usher Syndrome IID DFNB31	●		
Waardenburg Syndrome PAX3	●	●	

METABOLIC

DISEASE Gene	AR	AD	XL
2-Methyl-3-Hydroxybutyric Aciduria HSD17B10		●	●
2,4 Dienoyl-CoA Reductase Deficiency (NADKD1) NADK2	●		
3-Methylglutaconic Aciduria Type I AUH	●		
ß-Ketothiolase Deficiency ACAT1	●		
Argininemia ARG1	●		
Benign Hyperphenylalaninemia PAH	●		
Biotinidase Deficiency BTD	●		
Carnitine Palmitoyltransferase Type I Deficiency CPT1A	●		
Carnitine Uptake Defect/Carnitine Transport Defect SLC22A5	●		
Cerebrotendinous Xanthomatosis CYP27A1, LHX3	●		
Citrullinemia, Type II SLC25A13	●		
Classic Phenylketonuria PAH	●		
Congenital Disorder of Glycosylation 1b MPI	●		
Crigler-Najjar Syndrome UGT1A1	●		
Fabry Disease GLA			●
Galactokinase Deficiency GALK1	●		
Glutaric Acidemia Type I GCDH	●		

DISEASE Gene	AR	AD	XL
2-Methylbutyrylglycinuria ACADSB	●		
3-Methylcrotonyl-CoA Carboxylase Deficiency MCCC1, MCCC2	●		
3-Phosphoglycerate Dehydrogenase Deficiency PHGDH	●		
Abetalipoproteinemia MTTP	●		
Argininosuccinic Aciduria ASL	●		
Biopterin Defect In Cofactor Biosynthesis GCH1	●	●	
Carnitine Acylcarnitine Translocase Deficiency SLC25A20	●		
Carnitine Palmitoyltransferase Type II Deficiency CPT2	●		
Cerebral Creatine Deficiency Syndrome GAMT, GATM	●		
Citrullinemia, Type I ASS1	●		
Classic Galactosemia GALT	●		
Combined Pituitary Hormone Deficiency PROP1	●		
Corticosterone Methyloxidase Deficiency CYP11B2	●		
Cystinosis CTNS	●		
Galactoepimerase Deficiency GALE	●		
Glucose-6-Phosphate Dehydrogenase Deficiency G6PD			●
Glutaric Acidemia Type II ETFA, ETFB, ETFDH	●		

Glycogen Storage Disease Type 0 GYS2	●		
Glycogen Storage Disease Type Ib SLC37A4	●		
Glycogen Storage Disease IIIa AGL	●		
Hereditary Fructose Intolerance ALDOB	●		
Holocarboxylase Synthase Deficiency HLCS	●		
Hypercholesterolemia LDLR	●	●	
Hypophosphatasia ALPL	●	●	
Isovaleric Acidemia IVD	●		
Lipoprotein Lipase Deficiency (LPL) LPL	●		
Lysinuric Protein Intolerance SLC7A7	●		
Malonic Acidemia MLYCD	●		
Maple Syrup Urine Disease Type III DLD	●		
Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency HADH	●		
Methylmalonic Acidemia with Homocystinuria ABCD4, LMBRD1, MMACHC, MMADHC	●		
HCFC1			●
Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) MUT	●		
Methylmalonyl-CoA Epimerase Deficiency MCEE	●		
Mucopolysaccharidosis Type II (Hunter Syndrome) IDS			●
Nephrogenic Diabetes Insipidus Type II AQP2	●	●	
Niemann-Pick Disease Type C1 NPC1	●		
Ornithine Translocase Deficiency; Triple H Syndrome SLC25A15	●		
Primary Hyperoxaluria Type II GRHPR	●		
Propionic Acidemia PCCA, PCCB	●		
Transient Infantile Liver Failure TRMU	●		
Tyrosine Hydroxylase Deficiency TH	●		
Tyrosinemia, Type II TAT	●		
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) ACADVL	●		
X-Linked Adrenoleukodystrophy ABCD1			●

Glycogen Storage Disease Ia G6PC	●		
Glycogen Storage Disease Type II (Pompe) GAA	●		
Glycogen Storage Disease VI PYGL	●		
HMG-CoA Lyase Deficiency HMGCL	●		
Homocystinuria CBS	●		
Hypermethioninemia AHCY, GNMT	●		
MAT1A	●	●	
Isobutyrylglycinuria ACAD8	●		
Krabbe Disease GALC	●		
Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD Deficiency) HADHA	●		
Lysosomal Acid Lipase Deficiency LIPA	●		
Maple Syrup Urine Disease BCKDHA, BCKDHB, DBT	●		
Medium-Chain Acyl-CoA Dehydrogenase Deficiency ACADM	●		
Metachromatic Leukodystrophy ARSA	●		
Methylmalonic Acidemia (Cobalamin Disorders) MMAA, MMAB	●		
Methylmalonic Aciduria and Homocystinuria MTR, MTRR	●		
Mucopolysaccharidosis Type 1 IDUA	●		
N-Acetylglutamate Synthase Deficiency NAGS	●		
Niemann-Pick Disease Type A/B SMPD1	●		
Ornithine Transcarbamylase Deficiency OTC			●
Primary Hyperoxaluria Type I AGXT	●		
Primary Hyperoxaluria Type III HOGA1	●	●	
Short-Chain Acyl-CoA Dehydrogenase Deficiency ACADS	●		
Trifunctional Protein Deficiency HADHA, HADHB	●		
Tyrosinemia, Type I FAH	●		
Tyrosinemia, Type III HPD	●	●	
Wilson Disease ATP7B	●		

OTHER – Genetic, Immunodeficiency, Pulmonary, Musculoskeletal

DISEASE Gene	AR	AD	XL
Cystic Fibrosis CFTR	●		
Spinal Muscular Atrophy due to homozygous deletion of exon 7 & 8 in SMN1 SMN1, SMN2	●		

DISEASE Gene	AR	AD	XL
Severe Combined Immunodeficiencies ADA, IL7R, JAK3	●		
IL2RG			●
T-cell Related Lymphocyte Deficiencies PIK3CD		●	

● AR: AUTOSOMAL RECESSIVE ● AD: AUTOSOMAL DOMINANT ● XL: X-LINKED

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