NIPD GENETICS

Oreana®

neonatal screening

ЛИСТА НА 106 ГЕНЕТСКИ СОСТОЈБИ И 142 ГЕНИ

ЕНДОКРИНОЛОШКИ НАРУШУВАЊА

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

ХЕМОГЛОБИНОПАТИИ

НАРУШУВАЊЕ *Ген*	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*	●		

НАРУШУВАЊА ПОВРЗАНИ СО ГУБЕЊЕ НА СЛУХ

НАРУШУВАЊЕ *Ген*	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*	●	●	

МЕТАБОЛНИ НАРУШУВАЊА

НАРУШУВАЊЕ *Ген*	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*	●		

НАРУШУВАЊЕ *Ген*	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*	●		

	AR	AD	XL
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*			●

	AR	AD	XL
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*	●		

ОСТАНАТИ – Генетски, имунодефицитни, пулмонални, мускулно-скелетни нарушувања..

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

НАРУШУВАЊЕ *Ген*	AR	AD	XL
Severe Combined Immunodeficiencies *ADA, IL7R, JAK3*	●		
IL2RG			●
T-cell Related Lymphocyte Deficiencies *PIK3CD*		●	

● AR: АВТОЗОМНО РЕЦЕСИВНО ● AD: АВТОЗОМНО ДОМИНАНТНО ● XL: ПОВРЗАНО СО Х ХРОМОЗОМ

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