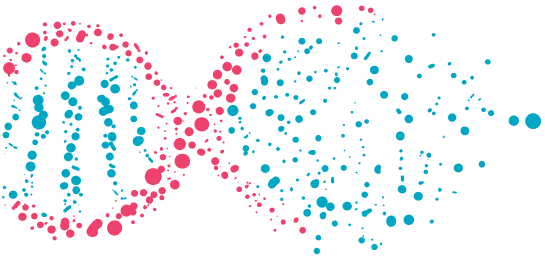

NEWBORN SCREENING

DISEASE LIST

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
17-beta-hydroxysteroid dehydrogenase X deficiency (*HSD17B10*)	Metabolic	PM
3-beta-hydroxysteroid dehydrogenase, type II, deficiency (*HSD3B2*)	Endocrine	PM
3-Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD) (*HADH*)	Metabolic	DM
3-methylcrotonyl-CoA carboxylase deficiency (*MCCC1, MCCC2*)	Metabolic	DM
3-methylglutaconic aciduria, type I (*AUH*)	Metabolic	O
3-phosphoglycerate dehydrogenase deficiency (*PHGDH*)	Metabolic	DM
46XY sex reversal 1 (*SRY*)	Endocrine	EM
Abetalipoproteinemia (*MTTP*)	Metabolic	DM
Achromatopsia (*CNGA3, CNGB3*)	Neurologic	O
Acrodermatitis enteropathica (*SLC39A4*)	Metabolic	PM
Acute infantile liver failure (*TRMU*)	Metabolic	EM
Acyl-CoA Dehydrogenase, Medium Chain, Deficiency of (*ACADM*)	Metabolic	DM
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (*CYP11B1*)	Endocrine	PM
Adrenocorticotropic hormone deficiency (*TBX19*)	Endocrine	PM
Adrenoleukodystrophy (*ABCD1*)	Neurologic	EM
Alagille syndrome (*JAG1*, *NOTCH2*)	Syndromic	EM
Alpha-Methylacetoacetic Aciduria/ß-Ketothiolase deficiency (*ACAT1*)	Metabolic	DM
Alport Syndrome (*COL4A3, COL4A4, COL4A5*)	Neurologic	EM
Alström Syndrome (*ALMS1*)	Syndromic	PM
Andermann syndrome (Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum) (*SLC12A6*)	Neurologic	EM
Anemia, with or without neutropenia and/or platelet abnormalities (*GATA1*)	Hematologic	EM
Apparent mineralocorticoid excess (*HSD11B2*)	Endocrine	PM
Argininemia (*ARG1*)	Metabolic	DM
Argininosuccinic aciduria (*ASL*)	Metabolic	DM
Arrhythmia (Brugada Syndrome 3, Timothy syndrome) (*CACNA1C*)	Cardiovascular	EM
ARVC and related phenotypes (*DSP*)	Cardiovascular	EM
Aspartylglucosaminuria (*AGA*)	Metabolic	EM
Ataxia with isolated vitamin E deficiency (*TTPA*)	Neurologic	DM
Atopic & eosinophilic disease, Hyper-IgE recurrent infection syndrome (*STAT3*)	Immunologic	PM
Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) (*AIRE*)	Immunologic	EM
Autosomal Recessive Polycystic Kidney Disease (*PKHD1*)	Urogenital	EM
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (*SACS*)	Neurologic	EM
Barth Syndrome (*TAFAZZIN*)	Metabolic	EM
Bartter syndrome type 4A (recessive) (*BSND*)	Urogenital	PM
Bernard-Soulier Syndrome (*GP1BB, GP9*)	Hematologic	PM
Biotinidase Deficiency (*BTD*)	Metabolic	PM
Bleeding disorder, platelet-type (*GP6, P2RY12*)	Hematologic	PM
Bloom syndrome (*BLM*)	Syndromic	EM
Branchiootorenal spectrum disorder (BORSD) (*EYA1, SIX1*)	Syndromic	PM
Brugada syndrome (*SCN5A*)	Cardiovascular	PM
Butyrylcholinesterase deficiency (*BCHE*)	Metabolic	O

PM=preventive management; EM=early management; DM=dietary management; O=Others

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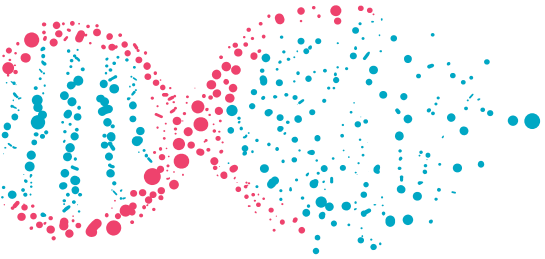

NEWBORN SCREENING

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
Canavan Disease (*ASPA*)	Neurologic	EM/supportive care
Cantu syndrome (*ABCC9*)	Musculoskeletal	EM
Carbamoylphosphate synthetase I deficiency (*CPS1*)	Metabolic	DM
Carnitine deficiency, systemic primary (Carnitine uptake defect/carnitine transport defect) (*SLC22A5*)	Metabolic	PM
Carnitine-Acylcarnitine Translocase Deficiency (*SLC25A20*)	Metabolic	DM
Carpenter Syndrome (*RAB23*)	Musculoskeletal	EM
Catecholaminergic polymorphic ventricular tachycardia (*CASQ2*)	Cardiovascular	PM
Cartilage-hair dysplasia/Anauxetic dysplasia (*RMRP*)	Syndromic	EM
CDG1b (*MPI*)	Metabolic	EM
Central hypothyroidism and testicular enlargement (*IGSF1*)	Endocrine	PM
Cerebral creatine deficiency syndrome (*GAMT, GATM*)	Metabolic	PM
Cerebrotendinous xanthomatosis (*CYP27A1*)	Metabolic	EM
Choreoathetosis, hypothyroidism, and neonatal respiratory distress (*NKX2-1*)	Endocrine	PM
Chronic granulomatous disease (*CYBA, CYBB*)	Immunologic	O
Chudley-McCullough Syndrome (*GPSM2*)	Neurologic	PM
Ciliary dyskinesia, primary (*CFAP298, CCDC39, CCDC40, CCDC65, CCNO, DNAAF1, DNAAF5, DNAH11, DNAH5, DNAI1, DNAJB13, DNAAF4, DNAAF11, DRC1, GAS8, MCIDAS, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10*)	Pulmonary	EM
Citrullinemia (*ASS1, SLC25A13*)	Metabolic	DM
Cockayne syndrome/Xeroderma Pigmentosum (DeSanctis-Cacchione)/UV-Sensitive syndrome (*ERCC6*)	Syndromic	EM
Combined malonic and methylmalonic aciduria (*ACSF3*)	Metabolic	O
Combined oxidative phosphorylation deficiency 3 (*TSFM*)	Metabolic	EM
Congenital amegakaryocytic thrombocytopenia (*MPL*)	Hematologic	EM
Congenital bile acid synthesis defect (*AKR1D1, HSD3B7*)	Metabolic	PM
Congenital nephrotic syndrome (*NPHS1, NPHS2*)	Urogenital	EM
Congenital neutropenia (*ELANE, HAX1*)	Hematologic	EM
Corticosterone methyloxidase deficiency (*CYP11B2*)	Endocrine	PM
CPT deficiency, hepatic, type IA (*CPT1A*)	Metabolic	DM
Crigler-Najjar syndrome, types 1 and 2 / Gilbert Syndrome (*UGT1A1*)	Metabolic	EM
Cutis Laxa, AD 1 / Supravalvar aortic stenosis (*ELN*)	Cardiovascular	EM
Cystic Fibrosis (*CFTR*)	Syndromic	EM
Cystinosis (*CTNS*)	Metabolic	EM
Deafness (*ACTG1, CDH23, CLDN14, ESRRB, FGF3, GIPC3, GJB2, GJB6, ILDR1, KCNQ4, LOXHD1, LRTOMT, MARVELD2, MYO15A, MYO6, MYO7A, OTOA, OTOF, OTOG, OTOGL, POU3F4, PTPRQ, PJVK, S1PR2, SLITRK6, SMPX, STRC, TBC1D24, TECTA, TMC1, TMIE, TMPRSS3, TRIOBP*)	Neurologic	PM
Diabetes mellitus, permanent neonatal (*GCK, INS*)	Endocrine	PM
Diamond-Blackfan anemia (*RPL11, RPL5, RPS19, RPS24, RPS26, RPS29*)	Hematologic	EM
Distal renal tubular acidosis and other SLC4A1-related disorders (*SLC4A1*)	Urogenital	EM
Dystonia 9, GLUT1 deficiency syndromes 1 and 2, autosomal recessive (*SLC2A1*)	Neurologic	DM
Dystonia, dopa-responsive, due to sepiapterin reductase deficiency (*SPR*)	Metabolic	PM
Early infantile epileptic encephalopathy (*SCN2A, SCN8A, KCNQ2*)	Neurologic	EM
Ehlers-Danlos Syndrome Type IV - Vascular Type (*COL3A1*)	Musculoskeletal	PM
Ethylmalonic encephalopathy (*ETHE1*)	Metabolic	EM
Fabry Disease (*GLA*)	Metabolic	PM

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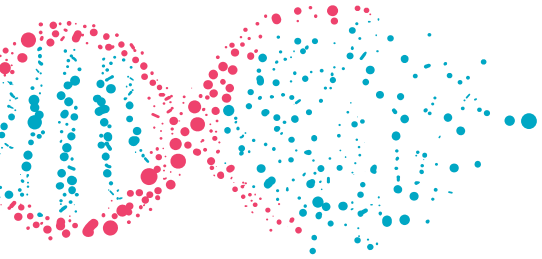

NEWBORN SCREENING

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
Factor V deficiency (*F5*)	Hematologic	PM
Factor VII deficiency (*F7*)	Hematologic	PM
Factor X deficiency (*F10*)	Hematologic	PM
Factor XI deficiency, autosomal recessive (*F11*)	Hematologic	PM
Factor XIIIA deficiency (*F13A1*)	Hematologic	EM
Factor XIIIB deficiency (*F13B*)	Hematologic	EM
Familial Adenomatous Polyposis (*APC*)	Cancer	PM
Familial dysalbuminemic hyperthyroxinemia (*ALB*)	Metabolic	PM
Familial dysautonomia (*ELP1*)	Neurologic	EM
Familial Hypercholesterolemia (*APOB, LDLR*)	Metabolic	PM
Familial infantile convulsions with paroxysmal choreoathetosis (*PRRT2*)	Neurologic	EM
Familial Mediterranean fever, AD/AR, Classic mutations associated with renal failure (*MEFV*)	Immunologic	PM
Fanconi Anemia (*FANCA*, *FANCB*, *FANCC*, *FANCD2*, *FANCG*, *FANCI*)	Hematologic	EM
Folate malabsorption, hereditary (*SLC46A1*)	Metabolic	DM
Fructose bisphosphatase deficiency (*FBP1*)	Metabolic	DM
Fructose intolerance (*ALDOB*)	Metabolic	DM
Fukuyama congenital muscular dystrophy/Limb girdle muscular dystrophy 2M/Walker-Warburg syndrome/DCM phenotype (childhood onset) (*FKTN*)	Musculoskeletal	EM
Galactokinase Deficiency with Cataracts (*GALK1*)	Metabolic	DM
Galactose epimerase deficiency (*GALE*)	Metabolic	DM
Galactosemia (*GALT*)	Metabolic	DM
Gaucher Disease, Type I (*GBA1*)	Metabolic	PM
Generalized thyrotropin-releasing hormone resistance (*TRHR*)	Endocrine	PM
Gitelman syndrome (*SLC12A3*)	Urogenital	PM
Glanzmann thrombasthenia (*ITGA2B, ITGB3*)	Hematologic	PM
Glutaric acidemia type II (*ETFA, ETFB, ETFDH*)	Metabolic	PM
Glutaricaciduria type I (*GCDH*)	Metabolic	DM
Glutathione Synthetase Deficiency (*GSS*)	Metabolic	PM
Glycogen Storage Diseases (*AGL, G6PC1, GAA, GBE1, GYS2, LAMP2, PHKA2, PHKB, PYGL, SLC37A4*)	Metabolic	DM / EM
Growth hormone deficiency, isolated, type IA (recessive) (*GH1*)	Endocrine	EM
Gyrate Atrophy of the Choroid and Retina (*OAT*)	Metabolic	DM
Hemoglobin SC disease (including beta thalassemia and Sickle cell disease) (*HBB*)	Hematologic	PM
Hemolytic anemia due to G6PD deficiency (*G6PD*)	Hematologic	PM
Hemophagocytic lymphohistiocytosis, familial, 3 (*UNC13D*)	Immunologic	PM
Hemophilia (*F8, F9*)	Hematologic	PM
Hereditary Paraganglioma-Pheochromocytoma Syndrome 4 (*SDHB*)	Cancer	PM
Hereditary spastic paraplegia type 11 (*SPG11*)	Neurologic	EM
Hermansky-Pudlak syndrome (*HPS1, HPS3, HPS4*)	Syndromic	EM
Hexosaminidase A deficiency (including Tay-Sachs disease) (*HEXA*)	Metabolic	O
HMG-CoA lyase deficiency/3-Hydroxy-3-methyglutaric aciduria (*HMGCL*)	Metabolic	DM
HMG-CoA synthase-2 deficiency (*HMGCS2*)	Metabolic	DM
Holocarboxylase synthetase deficiency (*HLCS*)	Metabolic	PM
Homocystinuria (*CBS, MTRR, MTR*)	Metabolic	DM / PM

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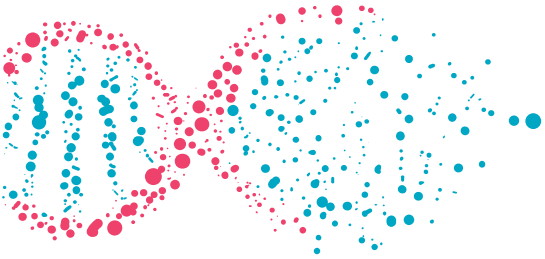

NEWBORN SCREENING

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
Hyperaldosteronism, familial type III (*KCNJ5*)	Endocrine	PM
Hyper-IgE recurrent infection syndrome, autosomal recessive (*DOCK8*)	Syndromic	PM
Hyperinsulinemic hypoglycemia (*ABCC8, KCNJ11*)	Endocrine	PM
Hyperinsulinism-hyperammonemia syndrome (*GLUD1*)	Endocrine	DM
Hypermethioninemia due to adenosine kinase deficiency (*ADK*)	Metabolic	DM
Hyperornithinemia-Hyperammonemia-Homocitrullinemia Syndrome (*SLC25A15*)	Metabolic	DM
Hyperparathyroidism (*CASR, CDC73*)	Endocrine	PM
Hyperphenylalaninemia, BH4 deficient (*GCH1, PCBD1, PTS, QDPR*)	Metabolic	PM / DM
Hypophosphatasia (*ALPL*)	Musculoskeletal	O
Hypothyroidism (*PAX8, THRA, TSHB, TSHR*)	Endocrine	PM
Hypouricemia renal, type 2 (*SLC2A9*)	Urogenital	EM
Immunodeficiency (*CARD11, CD3D, CD3E, CD40LG, CORO1A, DNMT3B, GATA2, NFKB2, PIK3CD*)	Immunologic	PM
Interleukin-2 receptor, alpha chain, deficiency of (*IL2RA*)	Immunologic	PM
Intrinsic factor deficiency (*CBLIF*)	Hematologic	PM
Isovaleric acidemia (*IVD*)	Metabolic	DM
Jervell and Lange-Nielsen syndrome (*KCNQ1*)	Cardiovascular	PM
Joubert syndrome/Meckel-Gruber syndrome/BBS13 (*MKS1*)	Syndromic	EM
Juvenile polyposis / hereditary hemorrhagic telangiectasia syndrome (*SMAD4*)	Cancer	EM
LAMA2-Related Muscular Dystrophy (*LAMA2*)	Musculoskeletal	EM
LCHAD deficiency (*HADHA*)	Metabolic	DM
Leigh syndrome French-Canadian type (*LRPPRC*)	Metabolic	EM
Li-Fraumeni syndrome (*TP53*)	Cancer	EM
Lipoamide dehydrogenase deficiency/Maple Syrup Urine Disease type III (*DLD*)	Metabolic	EM
Lipoid adrenal hyperplasia (*STAR*)	Endocrine	PM
Lipoprotein lipase deficiency (*LPL*)	Metabolic	DM
Liver Phosphorylase Kinase Deficiency / Glycogen Storage Disease IXC (GSD9C) (*PHKG2*)	Metabolic	DM
Loeys-Dietz Syndrome 5 (*TGFB3*)	Musculoskeletal	EM
Long QT Syndrome (*CALM1, CALM2, KCNH2*)	Cardiovascular	PM
Lymphoproliferative syndrome 1 (*ITK*)	Immunologic	EM
Lysinuric protein intolerance (*SLC7A7*)	Metabolic	PM
Malignant hyperthermia susceptibility (*CACNA1S*, *RYR1*)	Neurologic	PM
Malonyl-CoA decarboxylase deficiency (*MLYCD*)	Metabolic	DM
Maple syrup urine disease (*BCKDHA, BCKDHB, DBT*)	Metabolic	DM
Marfan syndrome (*FBN1*)	Musculoskeletal	EM
McArdle Disease (*PYGM*)	Metabolic	PM
Menkes Disease (*ATP7A*)	Metabolic	EM
Metachromatic leukodystrophy (*ARSA*)	Metabolic	PM
Methionine adenosyltransferase I/III deficiency (*MAT1A*)	Metabolic	DM
Methylmalonic aciduria (*LMBRD1, MMAA, MMAB, MMACHC, MMADHC, MMUT*)	Metabolic	DM
Methylmalonyl-CoA epimerase deficiency (*MCEE*)	Metabolic	DM
Mitochondrial complex I deficiency (*NDUFS6*)	Metabolic	O
MODY (*HNF1A, HNF4A*)	Endocrine	PM
Muckle-Wells syndrome / CINCA syndrome / Familial cold-induced inflammatory syndrome 1 (*NLRP3*)	Immunologic	PM

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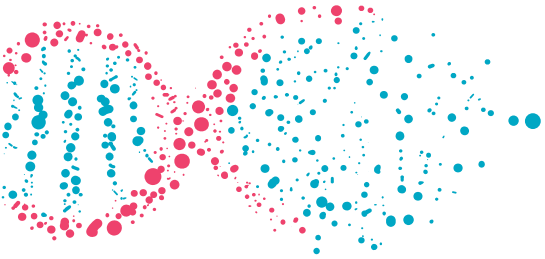

NEWBORN SCREENING

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
Mucolipidosis type IV (*MCOLN1*)	Metabolic	EM
Mucopolysaccharidosis (*ARSB, GALNS, IDS, IDUA*)	Metabolic	EM
Muenke syndrome (*FGFR3*)	Musculoskeletal	EM
Multiple endocrine neoplasia 1 (*MEN1*)	Cancer	PM
Multiple endocrine neoplasia II (*RET*)	Cancer	PM
MYH9-related disorders (MYH9RD) (*MYH9*)	Hematologic	EM
Myopathy due to CPTII Deficiency (*CPT2*)	Metabolic	DM
N-acetylglutamate synthase deficiency (*NAGS*)	Metabolic	DM
Nephrogenic diabetes insipidus, type II (*AQP2*)	Urogenital	PM
Nephrogenic syndrome of inappropriate antidiuresis X-linked (*AVPR2*)	Endocrine	PM
Neurodegeneration due to cerebral folate transport deficiency (*FOLR1*)	Neurologic	PM
Neurofibromatosis, type 1 (*NF1*)	Syndromic	EM
neuronal ceroid lipofuscinosis 1 (*PPT1*)	Metabolic	EM
Nieman-Pick disease (*NPC1, NPC2, SMPD1*)	Metabolic	EM
Noonan syndrome (*PTPN11, RIT1*)	Syndromic	EM
Osteogenesis imperfecta (*COL1A1, COL1A2*)	Musculoskeletal	PM
Osteopetrosis 1 (*TCIRG1*)	Musculoskeletal	PM
OTC deficiency (*OTC*)	Metabolic	DM
Pancreatic agenesis 1 (*PDX1*)	Syndromic	PM
Pendred Syndrome (*SLC26A4*)	Syndromic	PM
Persistent truncus arteriosus / Conotruncal heart defects (*NKX2-6*)	Cardiovascular	EM
Peutz-Jeghers Syndrome (*STK11*)	Cancer	PM
Phenylketonuria (*PAH*)	Metabolic	DM
Pheochromocytoma susceptibility (*MAX*)	Cancer	PM
Pituitary adenoma, growth hormone-secreting / Pituitary adenoma, ACTH-secreting / Pituitary adenoma, prolactin-secreting (*AIP*)	Cancer	PM
Pituitary hormone deficiency (*LHX3, POU1F1, PROP1*)	Endocrine	PM
PMM2-congenital disorders of glycosylation (*PMM2*)	Metabolic	EM
Polyposis, juvenile intestinal (*BMPR1A*)	Cancer	PM
Primary Congenital Glaucoma (*CYP1B1*)	Neurologic	PM
Primary hyperoxaluria (*AGXT, GRHPR, HOGA1*)	Metabolic	PM / DM
Propionicacidemia (*PCCA, PCCB*)	Metabolic	EM
Prothrombin Deficiency, congenital (*F2*)	Hematologic	EM
Pseudohypoaldosteronism (*SCNN1A, SCNN1B*)	Endocrine	PM
PTEN hamartoma tumor syndrome (*PTEN*)	Cancer	PM
Pyridoxamine 5'-phosphate oxidase deficiency (*PNPO*)	Metabolic	PM
Pyridoxine-dependent epilepsy (*ALDH7A1*)	Neurologic	DM
Quebec Platelet Disorder (*PLAU*)	Hematologic	PM
Renal tubular acidosis with deafness (*ATP6V1B1*)	Urogenital	PM
Reticular Dysgenesis (*AK2*)	Immunologic	PM
Retinoblastoma (*RB1*)	Cancer	PM
SCID, AR, T-negative/B-positive type (*JAK3*)	Immunologic	PM
Segawa syndrome, recessive (*TH*)	Neurologic	PM

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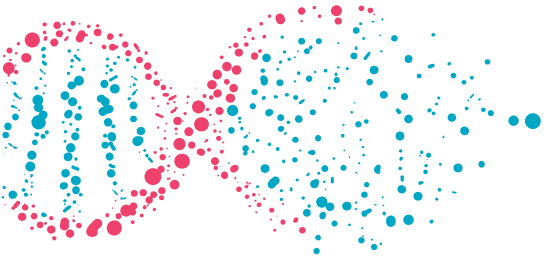

NEWBORN SCREENING

FUTURE HEALTH LABORATORIES

DISEASE	CATEGORY	INTERVENTION
Selective T-cell defect (*ZAP70*)	Immunologic	PM
SESAME syndrome (*KCNJ10*)	Syndromic	EM
Severe Combined Immunodeficiency (*ADA, DCLRE1C, IL2RG, IL7R, PTPRC, RAG1, RAG2*)	Immunologic	PM
Sialic acid storage disease (*SLC17A5*)	Metabolic	EM
Sitosterolemia (*ABCG5*)	Metabolic	DM
SLC26A2-related disorders (previously known as DTD and DTDST) (*SLC26A2*)	Musculoskeletal	EM
Smith-Lemli-Opitz Syndrome (*DHCR7*)	Syndromic	EM
Spherocytosis (*ANK1, EPB42*)	Hematologic	O
Stickler syndrome (*COL2A1, COL9A1, COL11A1*)	Syndromic	PM
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) (*SLC19A3*)	Metabolic	PM
Thiamine-responsive megaloblastic anemia syndrome (*SLC19A2*)	Metabolic	EM
Thrombophilia (*PROC, PROS1*)	Hematologic	PM
Thyroid dyshormonogenesis (*DUOX2, DUOXA2, IYD, SLC5A5, TG, TPO*)	Endocrine	PM
Transcobalamin II deficiency (*TCN2*)	Hematologic	PM
Treacher Collins syndrome (*POLR1D, TCOF1*)	Syndromic	PM
Trifunctional protein deficiency (*HADHB*)	Metabolic	DM
Tuberous sclerosis (*TSC1, TSC2*)	Syndromic	EM
Tyrosinemia (*FAH, HPD, TAT*)	Metabolic	EM / DM
Usher syndrome (*ADGRV1, CIB2, CLRN1, PCDH15, USH1C, USH1G, USH2A, WHRN*)	Syndromic	PM
Vitamin D-dependent rickets type I (*CYP27B1*)	Metabolic	PM
Vitamin K-dependent coagulation defect (*GGCX*)	Hematologic	PM
VLCAD deficiency (*ACADVL*)	Metabolic	DM
von Hippel-Lindau syndrome (*VHL*)	Cancer	PM
Waardenburg syndrome (*EDN3, MITF, PAX3, SNAI2, SOX10*)	Syndromic	PM
Wilms Tumor (*WT1*)	Cancer	PM
Wilson Disease (*ATP7B*)	Metabolic	DM
Wolman disease (infantile onset) - Cholesterol ester storage disease (*LIPA*)	Metabolic	PM

DISCLAIMER: myNewborn is a genetic screening test. The diseases included in this list are related to mutations in different genes that may not be included in the test. Furthermore, the genes included may be associated with other diseases that are not the main objective of the study. For the purpose of this list, the predominant gene-disease association has been used. The test only includes the study of diseases of genetic origin, therefore a negative result does not rule out the development of other non-genetic diseases that may appear during childhood, such as autoimmune hypothyroidism.

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