NBSure™

# Newborn Genetic Screening Jaundice Panel

## Detects pathogenic or likely pathogenic variants in 82 genes associated with single gene disorders presenting with a jaundice phenotype

| Disorders | Genes | Inheritance |
|---|---|---|
| Cholestasis, benign recurrent intrahepatic, 2 | *ABCB11* | AR |
| Cholestasis, progressive familial intrahepatic 2 | | AR |
| Cholestasis, progressive familial intrahepatic 3 | *ABCB4* | AR |
| Cholestasis, intrahepatic, of pregnancy, 3 | | AD/AR |
| Gallbladder disease 1 | | AD/AR |
| Adrenoleukodystrophy | *ABCD1* | XLR |
| Adrenomyeloneuropathy, adult | | XLR |
| Sitosterolemia 2 | *ABCG5* | AR |
| Sitosterolemia 1 | *ABCG8* | AR |
| Severe combined immunodeficiency due to ADA deficiency | *ADA* | AR/SMo |
| Adenosine deaminase deficiency, partial | | AR/SMo |
| Bile acid synthesis defect, congenital, 2 | *AKR1D1* | AR |
| Fructose intolerance, hereditary | *ALDOB* | AR |
| Spherocytosis, type 1 | *ANK1* | AD/AR |
| Hypercholesterolemia, familial, 2 | *APOB* | AD |
| Hypobetalipoproteinemia | | AR |
| Argininemia | *ARG1* | AR |
| Argininosuccinic aciduria | *ASL* | AR |
| Citrullinemia | *ASS1* | AR |
| Wilson disease | *ATP7B* | AR |
| Cholestasis, progressive familial intrahepatic 1 | *ATP8B1* | AR |
| Cholestasis, benign recurrent intrahepatic | | AR |
| Cholestasis, intrahepatic, of pregnancy, 1 | | AD |
| Cystic fibrosis | *CFTR* | AR |
| {Bronchiectasis with or without elevated sweat chloride 1, modifier of} | | AD |
| {Autoinflammation and autoimmunity, systemic, with immune dysregulation 1} | *COPA* | AD |
| CPT II deficiency, lethal neonatal | *CPT2* | AR |
| CPT II deficiency, infantile | | AR |
| CPT II deficiency, myopathic, stress-induced | | AD/AR |
| Cerebrotendinous xanthomatosis | *CYP27A1* | AR |
| Sclerosing cholangitis, neonatal | *DCDC2* | AR |
| Nephronophthisis 19 | | AR |
| Deafness, autosomal recessive 66 | | AR |
| Thyroid dyshormonogenesis 6 | *DUOX2* | AR |
| Elliptocytosis-1 | *EPB41* | AD/AR |
| Spherocytosis, type 5 | *EPB42* | . |
| Glutaric acidemia IIA | *ETFA* | AR |
| Glutaric acidemia IIB | *ETFB* | AR |
| Glutaric acidemia IIC | *ETFDH* | AR |
| Tyrosinemia, type I | *FAH* | AR |
| Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient | *G6PD* | XL |
| Galactose epimerase deficiency | *GALE* | AR |
| Galactosemia IV | *GALM* | AR |
| Galactosemia | *GALT* | AR |
| Gaucher disease, type I | *GBA1* | AR |
| Gaucher disease, type II | | AR |
| Gaucher disease, type III | | AR |
| Gaucher disease, type IIIC | | AR |
| Gaucher disease, perinatal lethal | | AR |
| Mitochondrial trifunctional protein deficiency 1 | *HADHA* | AR |
| LCHAD deficiency | | AR |
| HELLP syndrome, maternal, of pregnancy | | AR |
| Fatty liver, acute, of pregnancy | | AR |
| Mitochondrial trifunctional protein deficiency 2 | *HADHB* | AR |
| Thalassemias, alpha- | *HBA1* | . |
| Hemoglobin H disease, nondeletional | | . |
| Heinz body anemias, alpha- | | AD |
| Methemoglobinemia, alpha type | | AD |
| Erythrocytosis, familial, 7 | | AD |
| Thalassemia, alpha- | *HBA2* | . |
| Hemoglobin H disease, deletional and nondeletional | | . |
| Heinz body anemia | | AD |
| Erythrocytosis, familial, 7 | | AD |
| Hemochromatosis, type 1 | *HFE* | AR |
| D-bifunctional protein deficiency | *HSD17B4* | AR |
| Perrault syndrome 1 | | AR |
| Inflammatory bowel disease 25, early onset, autosomal recessive | *IL10RB* | AR |
| Nephronophthisis 2, infantile | *INVS* | AR |
| Alagille syndrome 1 | *JAG1* | AD |
| Tetralogy of Fallot | | AD |
| Charcot-Marie-Tooth disease, axonal, type 2HH | | AD |
| Wolman disease | *LIPA* | AR |
| Cholesteryl ester storage disease | | AR |
| Chediak-Higashi syndrome | *LYST* | AR |
| Methylmalonic aciduria and homocystinuria, cblC type | *MMACHC* | AR |

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| Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) | *MPV17* | AR |
| Charcot-Marie-Tooth disease, axonal, type 2EE | | AR |
| Homocystinuria-megaloblastic anemia, cblG complementation type | *MTR* | AR |
| Mevalonic aciduria | *MVK* | AR |
| Porokeratosis 3, multiple types | | AD |
| Hyper-IgD syndrome | | AR |
| Hypothyroidism, congenital nongoitrous, 5 | *NKX2-5* | AD |
| Conotruncal heart malformations, variable | | -- |
| Tetralogy of Fallot | | AD |
| Atrial septal defect 7, with or without AV conduction defects | | AD |
| Ventricular septal defect 3 | | AD |
| Hypoplastic left heart syndrome 2 | | AD |
| Alagille syndrome 2 | *NOTCH2* | AD |
| Hajdu-Cheney syndrome | | AD |
| Niemann-Pick disease, type C1 | *NPC1* | AR |
| Niemann-Pick disease, type D | | AR |
| Niemann-Pick disease, type C2 | *NPC2* | AR |
| Meckel syndrome 7 | *NPHP3* | AR |
| Nephronophthisis 3 | | AR |
| Renal-hepatic-pancreatic dysplasia 1 | | AR |
| Albinism, oculocutaneous, type II | *OCA2* | AR |
| Albinism, brown oculocutaneous | | AR |
| Ornithine transcarbamylase deficiency | *OTC* | XL |
| Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia | *PAX8* | AD |
| Peroxisome biogenesis disorder 1A (Zellweger) | *PEX1* | AR |
| Peroxisome biogenesis disorder 1B (NALD/IRD) | | AR |
| Heimler syndrome 1 | | AR |
| Neurodevelopmental disorder with epilepsy and hemochromatosis | *PIGA* | XLR |
| Multiple congenital anomalies-hypotonia-seizures syndrome 2 | | XLR |
| Polycystic kidney disease 2 | *PKD2* | AD |
| Polycystic kidney disease 4, with or without hepatic disease | *PKHD1* | AR |
| Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient | *PKLR* | AR |
| [Adenosine triphosphate, elevated, of erythrocytes] | | AD |
| Hemophagocytic lymphohistiocytosis, familial, 2 | *PRF1* | AR |
| Lymphoma, non-Hodgkin | | . |
| Aplastic anemia | | . |
| Pituitary hormone deficiency, combined, 2 | *PROP1* | AR |
| Griscelli syndrome, type 2 | *RAB27A* | AR |

| Disorders | Genes | Inheritance |
|---|---|---|
| Carnitine deficiency, systemic primary | *SLC22A5* | AR |
| Citrullinemia, adult-onset type II | *SLC25A13* | AR |
| Citrullinemia, type II, neonatal-onset | | AR |
| Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome | *SLC25A15* | AR |
| Carnitine-acylcarnitine translocase deficiency | *SLC25A20* | AR |
| Pendred syndrome | *SLC26A4* | AR |
| Deafness, autosomal recessive 4, with enlarged vestibular aqueduct | | AR |
| Glycogen storage disease Ib | *SLC37A4* | AR |
| Glycogen storage disease Ic | | AR |
| Congenital disorder of glycosylation, type IIw | | AD |
| Spherocytosis, type 4 | *SLC4A1* | AD |
| Ovalocytosis, SA type | | AD |
| Cryohydrocytosis | | AD |
| Distal renal tubular acidosis 1 | | AD |
| Distal renal tubular acidosis 4 with hemolytic anemia | | AR |
| Thyroid dyshormonogenesis 1 | *SLC5A5* | AR |
| Niemann-Pick disease, type A | *SMPD1* | AR |
| Niemann-Pick disease, type B | | AR |
| Spherocytosis, type 3 | *SPTA1* | AR |
| Elliptocytosis-2 | | AD |
| Pyropoikilocytosis | | AR |
| Spherocytosis, type 2 | *SPTB* | AD |
| Elliptocytosis-3 | | AD/AR |
| Anemia, neonatal hemolytic, fatal or near-fatal | | AD/AR |
| Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease | *STXBP2* | AR |
| Thyroid dyshormonogenesis 3 | *TG* | AR |
| Cholestasis, progressive familial intrahepatic 4 | *TJP2* | AR |
| Hypercholanemia, familial 1 | | AR |
| Thyroid dyshormonogenesis 2A | *TPO* | AR |
| Hypothyroidism, congenital, nongoitrous 4 | *TSHB* | AR |
| Hypothyroidism, congenital, nongoitrous, 1 | *TSHR* | AR |
| Hyperthyroidism, nonautoimmune | | AD |
| Hyperthyroidism, familial gestational | | AD |
| Crigler-Najjar syndrome, type I | *UGT1A1* | AR |
| Crigler-Najjar syndrome, type II | | AR |
| Hyperbilirubinemia, familial transient neonatal | | AD/AR |

Note: AD: Autosomal dominant; AR: Autosomal recessive; XL: X-linked; XLR: X-linked recessive; SMo: Somatic mosaicism

Conditions surrounded by brackets '{ }' indicate mutations that contribute to susceptibility to multifactorial disorders or to infection.
Conditions surrounded by brackets '[ ]' indicate nondiseases that mainly genetic variations that lead to apparently abnormal laboratory test values.

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