

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

Disorders	Genes	Inheritance
Thyroid dysharmonogenesis 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		AR
LCHAD deficiency	HADHA	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, cbIC type	MMACHC	AR

Disorders	Genes	Inheritance	
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	MPV17	AR	
Charcot-Marie-Tooth disease, axonal, type 2EE		AR	
Homocystinuria-megaloblastic anemia, cbIG 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 dysmorphogenesis 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 dysmorphogenesis 3	TG	AR
Cholestasis, progressive familial intrahepatic 4	TJP2	AR
Hypercholanemia, familial 1		AR
Thyroid dysmorphogenesis 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

'Bronchiectasis with or without elevated sweat chloride 1, modifier of' and 'Autoinflammation and autoimmunity, systemic, with immune dysregulation 1' indicate mutations that contribute to susceptibility to multifactorial disorders or to susceptibility to infection. 'Adenosine triphosphate, elevated, of erythrocytes' is nondiseases, mainly genetic variations that lead to apparently abnormal laboratory test values.

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