

Список из 102 моногенных заболеваний плода

ГЕНЫ	ДОМИНАНТНЫЕ МОНОГЕННЫЕ ЗАБОЛЕВАНИЯ	ГЕНЫ	ДОМИНАНТНЫЕ МОНОГЕННЫЕ ЗАБОЛЕВАНИЯ
BRAF	Cardiofaciocutaneous syndrome 1, LEOPARD syndrome 3, Noonan syndrome	NOTCH2	Alagille syndrome 2
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	NRAS	Noonan syndrome
CHD7	CHARGE syndrome	NSD1	Sotos syndrome 1
COL1A1	Osteogenesis imperfecta	OTC	Ornithine transcarbamylase deficiency
COL1A2	Osteogenesis imperfecta	PAFAH1B1	Lissencephaly 1
COL3A1	Ehlers-Danlos syndrome	POLR1C	Treacher Collins syndrome
CREBBP	Rubinstein-Taybi syndrome	PTPN11	LEOPARD syndrome
EFNB1	Craniofrontonasal dysplasia	RAD21	Cornelia de Lange syndrome 4
EP300	Rubinstein-Taybi syndrome	RAF1	Noonan syndrome
FBN1	Marfan syndrome	SCN1A	Dravet syndrome
FGFR1	Pfeiffer syndrome	SHH	Holoprosencephaly type 3
FGFR2	Crouzon syndrome	SHOC2	Noonan-like syndrome with loose anagen hair, Noonan syndrome
FGFR3	Hypochondroplasia	SMC1A	Cornelia de Lange syndrome 2
FOXG1	Congenital Rett-like syndrome	SMC3	Cornelia de Lange syndrome 3
G6PD	Glucose-6-phosphate dehydrogenase deficiency	SOS1	Noonan syndrome
HDAC8	Cornelia de Lange syndrome 5	STXBP1	Early infantile epileptic encephalopathy
HRAS	Costello syndrome, Noonan syndrome	TBX5	Holt-Oram syndrome
JAG1	Alagille syndrome	TCF4	Pitt-Hopkins syndrome
KAT6B	Genitopatellar syndrome	TCOF1	Treacher Collins syndrome
KMT2D	Kabuki syndrome	TSC1	Tuberous sclerosis 1
KRAS	Noonan syndrome	TSC2	Tuberous sclerosis 2
MAP2K1	Cardiofaciocutaneous syndrome 3, Noonan syndrome	TUBA1A	Lissencephaly
MAP2K2	Cardiofaciocutaneous syndrome 4, Noonan syndrome	TUBB2B	Tubulinopathy with polymicrogyria/lissencephaly
MECP2	Rett syndrome	TWIST1	Saethre-Chotzen syndrome
NIPBL	Cornelia de Lange syndrome 1	ZIC2	Nonsyndromic holoprosencephaly

СПЕЦИФИЧНОСТЬ 99,0%

ЧУВСТВИТЕЛЬНОСТЬ 99,8%

ГЕНЫ	РЕЦЕССИВНЫЕ МОНОГЕННЫЕ ЗАБОЛЕВАНИЯ
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency
ALMS1	Alström syndrome
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis type VI
ASPA	Canavan disease
ATP7B	Wilson disease
BBS2	Bardet–Biedl syndrome
CC2D2A	Meckel/Joubert syndrome
CEP290	Meckel/Joubert syndrome
CFTR	Cystic fibrosis
DHCR7	Smith-Lemli-Opitz syndrome
DYNC2H1	Short-rib thoracic dysplasia (ciliopathy)
EVC	Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome
GAA	Pompe disease (Glycogen storage disease II)
GALC	Krabbe disease
GALT	Galactosemia
GBA1	Gaucher disease
GJB2	Congenital deafness
GLB1	GM1 gangliosidosis
GNPTAB	Mucopolipidosis II/III
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HBB	Beta thalassemia
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease

ГЕНЫ	РЕЦЕССИВНЫЕ МОНОГЕННЫЕ ЗАБОЛЕВАНИЯ
HGSNAT	Mucopolysaccharidosis type IIIC
IDUA	Mucopolysaccharidosis type I
IFT140	Jeune syndrome
IVD	Isovaleric acidemia
LAMA2	Congenital muscular dystrophy MDC1A
MEFV	Familial mediterranean fever
MMAA	Methylmalonic aciduria, cblA type
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMUT	Methylmalonic aciduria, mut(0) type
NAGLU	Mucopolysaccharidosis type IIIB
NPC1	Niemann–Pick disease type C 1
NPC2	Niemann–Pick disease type C 2
PAH	Phenylketonuria
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PEX1	Peroxisome biogenesis disorder type 1A (Zellweger)
PKHD1	Autosomal recessive polycystic kidney disease
PMM2	Congenital disorder of glycosylation type 1A
POMT1	Walker-Warburg syndrome
RPGRIP1L	Meckel/Joubert syndrome
SERPINA1	Alpha 1 antitrypsin deficiency
SLC26A2	Diastrophic dysplasia/achondrogenesis IB
SLC3A1	Cystinuria
SLC7A9	Cystinuria
SMPD1	Niemann–Pick disease type A/B
TMEM67	Jouberts syndrome

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