Carrier screening -
Seznam vyšetřovaných onemocnění / List of examined disorders

Verze testu / Test version: V5

Název onemocnění / Disease name	Kód OMIM / OMIM number	Typ dědičnosti / Inheritance	Název genu / Gene name	Kód OMIM genu / OMIM gene number
46,XY SEX REVERSAL, DAX1-RELATED DOSAGE-SENSITIVE SEX REVERSAL; DSS	300018	XL	NR0B1	300473
ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF; ACADMD	201450	AR	ACADM	607008
ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD	201475	AR	ACADVL	609575
ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY; CAH	201910	AR	CYP21A2	613815
ADRENAL HYPOPLASIA, CONGENITAL; AHC	300200	XLR	NR0B1	300473
ADRENOLEUKODYSTROPHY	300100	XLR	ABCD1	300371
ACHROMATOPSIA 2; ACHM2	216900	AR	CNGA3	600053
ACHROMATOPSIA 3; ACHM3	262300	AR	CNGB3	605080
ALBINISM, OCULOCUTANEOUS, TYPE IA; OCA1A	203100	AR	TYR	606933
ALBINISM, OCULOCUTANEOUS, TYPE IB; OCA1B	606952	AR	TYR	606933
ALBINISM, OCULOCUTANEOUS, TYPE II; OCA2	203200	AR	OCA2	611409
ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD	613490	AR	SERPINA1	107400
ALPHA-THALASSEMIA	604131	AR	HBA1/ HBA2	141800/ 141850
ANAUXETIC DYSPLASIA 1; ANXD1	607095	AR	RMRP	157660
ANDROGEN INSENSITIVITY SYNDROME; AIS	300068	XLR	AR	313700
ANDROGEN INSENSITIVITY, PARTIAL; PAIS	312300	XLR	AR	313700
ANEMIA, NONSPHEROCYTIC HEMOLYTIC, DUE TO G6PD DEFICIENCY (Favism)	300908	XL	G6PD	305900
ANTERIOR SEGMENT DYSGENESIS 6; ASGD6	617315	AR	CYP1B1	601771
ARTHROGRYPOSIS MULTIPLEX CONGENITA 6; AMC6	619334	AR	NEB	161650
ASPARTYLGLUCOSAMINURIA; AGU	208400	AR	AGA	613228
AUTOIMMUNE POLYENDOCRINE SYNDROME, TYPE I, WITH OR WITHOUT REVERSIBLE METAPHYSEAL DYSPLASIA; APS1	240300	AR (vzácně AD)	AIRE	607358
BARDET-BIEDL SYNDROME 14; BBS14	615991	AR	CEP290	610142
BETA-THALASSEMIA	613985	AR	HBB	141900
CANAVAN DISEASE	271900	AR	ASPA	608034
CARDIOMYOPATHY, DILATED, 1X; CMD1X	611615	AR	FKTN	607440
CARDIOMYOPATHY, DILATED, 3B; CMD3B	302045	XL	DMD	300377
CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	255120	AR	CPT1A	600528
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, INFANTILE	600649	AR	CPT2	600650
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL	608836	AR	CPT2	600650
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED	255110	AR (alelické AD)	CPT2	600650
CARTILAGE-HAIR HYPOPLASIA; CHH	250250	AR	RMRP	157660
CEREBRAL CREATINE DEFICIENCY SYNDROME 1; CCDS1	300352	XLR	SLC6A8	300036
CEROID LIPOFUSCINOSIS, NEURONAL, 2; CLN2	204500	AR	TPP1	607998
CEROID LIPOFUSCINOSIS, NEURONAL, 5; CLN5	256731	AR	CLN5	608102
CITRULLINEMIA, CLASSIC	215700	AR	ASS1	603470
CONE-ROD DYSTROPHY 3; CORD3	604116	AR	ABCA4	601691
CONGENITAL ARTHROGRYPOSIS WITH ANTERIOR HORN CELL DISEASE; CAAHD	611890	AR	GLE1	603377
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ia; CDG1A	212065	AR	PMM2	601785
CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED	304100	XLR	L1CAM	308840
CYSTIC FIBROSIS; CF	219700	AR	CFTR	602421
DEAFNESS, AUTOSOMAL RECESSIVE 16; DFNB16	603720	AR	STRC	606440
DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A	220290	AR (DD)	GJB2/ GJB3/ GJB6	121011/ 603324/ 604418
DEAFNESS, AUTOSOMAL RECESSIVE 2; DFNB2	600060	AR (alelické AD)	MYO7A	276903
DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	600791	AR	SLC26A4	605646
DEAFNESS-INFERTILITY SYNDROME	611102	AR	STRC/ CATSPER2	606440/ 607249
DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY; DLDD	246900	AR	DLD	238331
EHLERS-DANLOS SYNDROME, CLASSIC-LIKE; EDSCLL	606408	AR	TNXB	600985
EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE; RDEB	226600	AR	COL7A1	120120
EPIDERMOLYSIS BULLOSA DYSTROPHICA, PRETIBIAL	131850	AR (alelické AD)	COL7A1	120120
EPIDERMOLYSIS BULLOSA PRURIGINOSA	604129	AR (alelické AD)	COL7A1	120120
FABRY DISEASE	301500	XL	GLA	300644
FAMILIAL MEDITERRANEAN FEVER; FMF	249100	AR (vzácně AD)	MEFV	608107
FRASER SYNDROME 3; FRASRS3	617667	AR	GRIP1	604597
FRUCTOSE INTOLERANCE, HEREDITARY	229600	AR	ALDOB	612724
GALACTOSEMIA I; GALAC1	230400	AR	GALT	606999
GAUCHER DISEASE, TYPE I	230800	AR	GBA	606463
GAUCHER DISEASE, TYPE II	230900	AR	GBA	606463
GAUCHER DISEASE, TYPE III	231000	AR	GBA	606463
GAUCHER DISEASE, TYPE IIIC	231005	AR	GBA	606463
GLAUCOMA 3, PRIMARY CONGENITAL, A; GLC3A	231300	AR	CYP1B1	601771
GLUTARIC ACIDEMIA I; GA1	231670	AR	GCDH	608801
GLYCOGEN STORAGE DISEASE Ia; GSD1A	232200	AR	G6PC	613742
GLYCOGEN STORAGE DISEASE II; GSD2 (Pompe disease)	232300	AR	GAA	606800
GLYCOGEN STORAGE DISEASE III; GSD3	232400	AR	AGL	610860
GLYCOGEN STORAGE DISEASE IV; GSD4	232500	AR	GBE1	607839
GM1-GANGLIOSIDOSIS, TYPE I	230500	AR	GLB1	611458
GM1-GANGLIOSIDOSIS, TYPE II	230600	AR	GLB1	611458

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GM1-GANGLIOSIDOSIS, TYPE III	230650	AR	GLB1	611458
HEMOPHILIA A	306700	XLR	F8	300841
HEMOPHILIA B	306900	XLR	F9	300746
HERMANSKY-PUDLAK SYNDROME 1; HPS1	203300	AR	HPS1	604982
HERMANSKY-PUDLAK SYNDROME 3; HPS3	614072	AR	HPS3	606118
HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY	236250	AR	MTHFR	607093
HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS	307000	XLR	L1CAM	308840
HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 1; HHF1	256450	AR (vzácně AD)	ABCC8	600509
HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 2; HHF2	601820	AR (vzácně AD)	KCNJ11	600937
HYPOSPADIAS 1, X-LINKED; HYSP1	300633	XLR	AR	313700
INFANTILE SIALIC ACID STORAGE DISEASE; ISSD	269920	AR	SLC17A5	604322
JOUBERT SYNDROME 2; JBTS2	608091	AR	TMEM216	613277
JOUBERT SYNDROME 3; JBTS3	608629	AR	AHI1	608894
JOUBERT SYNDROME 5; JBTS5	610188	AR	CEP290	610142
KANZAKI DISEASE	609242	AR	NAGA	104170
LEBER CONGENITAL AMAUROSIS 10; LCA10	611755	AR	CEP290	610142
LETHAL CONGENITAL CONTRACTURE SYNDROME 1; LCCS1	253310	AR	GLE1	603377
LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	609016	AR	HADHA	600890
MAPLE SYRUP URINE DISEASE TYPE IA, IB, II; MSUD1A, MSUD1B, MSUD2	248600	AR	BCKDHA/ BCKDHB/ DBT	608348/ 248611/ 248610
MASA SYNDROME (CRASH SYNDROME)	303350	XLR	L1CAM	308840
MECKEL SYNDROME, TYPE 2; MKS2	603194	AR	TMEM216	613277
MECKEL SYNDROME, TYPE 4; MKS4	611134	AR	CEP290	610142
METACHROMATIC LEUKODYSTROPHY; MLD	250100	AR	ARSA	607574
METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS; MDWH	250460	AR	RMRP	157660
METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	251000	AR	MMUT	609058
MITOCHONDRIAL DNA DEPLETION SYNDROME 4A (ALPERS TYPE); MTDPS4A	203700	AR	POLG	174763
MITOCHONDRIAL DNA DEPLETION SYNDROME 4B (MNGIE TYPE); MTDPS4B	613662	AR	POLG	174763
MITOCHONDRIAL RECESSIVE ATAXIA SYNDROME (INCLUDES SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS; SANDO and SPINOCEREBELLAR ATAXIA WITH EPILEPSY; SCAE); MIRAS	607459	AR	POLG	174763
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY; MTPD	609015	AR	HADHA	600890
MUCOLIPIDOSIS II ALPHA/BETA	252500	AR	GNPTAB	607840
MUCOLIPIDOSIS III ALPHA/BETA	252600	AR	GNPTAB	607840
MUCOLIPIDOSIS IV; ML4	252650	AR	MCOLN1	605248
MUCOPOLYSACCHARIDOSIS, TYPE IIIA; MPS3A	252900	AR	SGSH	605270
MUCOPOLYSACCHARIDOSIS, TYPE IVB; MPS4B	253010	AR	GLB1	611458
MUSCULAR DYSTROPHY, BECKER TYPE; BMD	300376	XLR	DMD	300377
MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD	310200	XLR	DMD	300377
MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2A	253600	AR (alelické AD)	CAPN3	114240
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES), TYPE A, 4; MDDGA4	253800	AR	FKTN	607440
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITHOUT IMPAIRED INTELLECTUAL DEVELOPMENT), TYPE B, 4; MDDGB4	613152	AR	FKTN	607440
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, 4; MDDGC4	611558	AR	FKTN	607440
MYASTHENIC SYNDROME, CONGENITAL, 4B, FAST-CHANNEL; CMS4B	616324	AR	CHRNE	100725
MYASTHENIC SYNDROME, CONGENITAL, 4C, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS4C	608931	AR	CHRNE	100725
NEMALINE MYOPATHY 2; NEM2	256030	AR	NEB	161650
NEPHROTIC SYNDROME, TYPE 1; NPHS1	256300	AR	NPHS1	602716
NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III; HSAN3 (Familial Dysautonomia)	223900	AR	ELP1	603722
NIEMANN-PICK DISEASE, TYPE A (Acid Sphingomyelinase Deficiency)	257200	AR	SMPD1	607608
NIEMANN-PICK DISEASE, TYPE B (Acid Sphingomyelinase Deficiency)	607616	AR	SMPD1	607608
OPITZ GBBB SYNDROME; GBBB	300000	XLR	MID1	300552
ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO	311250	XL	OTC	300461
PELIZAEUS-MERZBACHER DISEASE	312080	XLR	PLP1	300401
PENDRED SYNDROME; PDS	274600	AR	SLC26A4	605646
PHENYLKETONURIA; PKU	261600	AR	PAH	612349
PITUITARY HORMONE DEFICIENCY, COMBINED, 2; CPHD2	262600	AR	PROP1	601538
POLYCYSTIC KIDNEY DISEASE 4 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD4	263200	AR	PKHD1	606702
POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM; APBN	263570	AR	GBE1	607839
PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, AUTOSOMAL RECESSIVE 1; PEOB1	258450	AR	POLG	174763
PROPIONIC ACIDEMIA	606054	AR	PCCA/ PCCB	232000/ 232050
RETINITIS PIGMENTOSA 19; RP19	601718	AR	ABCA4	601691
RETINITIS PIGMENTOSA 39; RP39	613809	AR	USH2A	608400
RETINITIS PIGMENTOSA 61; RP61	614180	AR	CLRN1	606397
RETINITIS PIGMENTOSA, X-LINKED, AND SINORESPIRATORY INFECTIONS, WITH OR WITHOUT DEAFNESS	300455	XLR	RPGR	312610
RETINOSCHISIS 1, X-LINKED, JUVENILE; RS1	312700	XLR	RS1	300839
SALLA DISEASE; SD	604369	AR	SLC17A5	604322
SANDHOFF DISEASE	268800	AR	HEXB	606873
SENIOR-LOKEN SYNDROME 6; SLSN6	610189	AR	CEP290	610142
SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY; SRTD3	613091	AR	DYNC2H1	603297
SCHINDLER DISEASE, TYPE I	609241	AR	NAGA	104170
SICKLE CELL ANEMIA	603903	AR	HBB	141900
SMITH-LEMLI-OPITZ SYNDROME; SLOS	270400	AR	DHCR7	602858
SPASTIC PARAPLEGIA 2, X-LINKED; SPG2	312920	XLR	PLP1	300401
SPINAL MUSCULAR ATROPHY, TYPE I; SMA1	253300	AR	SMN1	600354
SPINAL MUSCULAR ATROPHY, TYPE II; SMA2	253550	AR	SMN1	600354
SPINAL MUSCULAR ATROPHY, TYPE III; SMA3	253400	AR	SMN1	600354
SPINAL MUSCULAR ATROPHY, TYPE IV; SMA4	271150	AR	SMN1	600354
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 7; SCAR7	609270	AR	TPP1	607998
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10; SCAR10	613728	AR	ANO10	613726

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STARGARDT DISEASE 1; STGD1	248200	AR	ABCA4	601691
TAY-SACHS DISEASE; TSD (Hexosaminidase A Deficiency)	272800	AR	HEXA	606869
TYROSINEMIA, TYPE I; TYRSN1	276700	AR	FAH	613871
USHER SYNDROME, TYPE I; USH1(B)	276900	AR	MYO7A	276903
USHER SYNDROME, TYPE IIA; USH2A	276901	AR	USH2A	608400
USHER SYNDROME, TYPE IIIA; USH3A	276902	AR	CLRN1	606397
VAS DEFERENS, CONGENITAL BILATERAL APLASIA OF; CBAVD	277180	AR	CFTR	602421
WILSON DISEASE	277900	AR	ATP7B	606882