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Genetická laboratoř, Laboratoř reprodukční genetiky

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Rozšířený screening přenašečství recesivních monogenních onemocnění

SEZNAM VYŠETŘENÝCH ONEMOCNĚNÍ

Carrier screening & Reproduction disorders Genetika Plzeň, V2, část A – Carrier screening

Název onemocnění	Kód OMIM	Typ dědičnosti	Název genu	Kód OMIM genu
ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF; ACADMD	201450	AR	ACADM	607008
ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD	201475	AR	ACAVLD	609575
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
ALKAPTONURIA; AKU	203500	AR	HGD	607474
ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD	613490	AR	SERPINA1	107400
ALPHA-THALASSEMIA	604131	AR	HBA1	141800
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
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 13; BBS13	615900	AR	MKS1	609883
BETA-THALASSEMIA	613985	AR	HBB	141900
BLOOM SYNDROME; BLM	210900	AR	RECQL3	604610
CANAVAN DISEASE	271900	AR	ASPA	608034
CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	255120	AR	CPT1A	600528
CARTILAGE-HAIR HYPOPLASIA; CHH	250250	AR	RMRP	157660
CEROID LIPOFUSCINOSIS, NEURONAL, 1; CLN1	256730	AR	PPT1	600722
CEROID LIPOFUSCINOSIS, NEURONAL, 2; CLN2	204500	AR	TPP1	607998
CEROID LIPOFUSCINOSIS, NEURONAL, 5; CLN5	256731	AR	CLN5	608102
CITRULLINEMIA, CLASSIC	215700	AR	ASS1	603470
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
CYSTINOSIS, ADULT NONNEPHROPATHIC	219750	AR	CTNS	606272
CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE	219900	AR	CTNS	606272
CYSTINOSIS, NEPHROPATHIC; CTNS	219800	AR	CTNS	606272
DEAFNESS, AUTOSOMAL RECESSIVE 16; DFNB16	603720	AR	STRC	606440
DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A	220290	AR (DD)	GJB2	121011
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	606440
DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY; DLDD	246900	AR	DLD	238331
FABRY DISEASE	301500	XL	GLA	300644
FAMILIAL MEDITERRANEAN FEVER; FMF	249100	AR (vzácně AD)	MEFV	608107
FANCONI ANEMIA, COMPLEMENTATION GROUP C; FANCC	227645	AR	FANCC	613899
FRUCTOSE INTOLERANCE, HEREDITARY	229600	AR	ALDOB	612724
GALACTOSEMIA	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
GITELMAN SYNDROME; GTLMNS	263800	AR	SLC12A3	600968
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
GM1-GANGLIOSIDOSIS, TYPE I	230500	AR	GLB1	611458
GM1-GANGLIOSIDOSIS, TYPE II	230600	AR	GLB1	611458
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
HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	236200	AR	CBS	613381
HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY	236250	AR	MTHFR	607093
HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS	307000	XLR	L1CAM	308840
HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS	307000	XLR	L1CAM	308840
HYDROLETHALUS SYNDROME 1; HLS1	236680	AR	HYLS1	610693
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 28; JBTS28	617121	AR	MKS1	609883
LETHAL CONGENITAL CONTRACTURE SYNDROME 1; LCCS1	253310	AR	GLE1	603377

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Carrier screening & Reproduction disorders Genetika Plzeň, V2, část A – Carrier screening

LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	609016	AR	HADHA	600890
MAPLE SYRUP URINE DISEASE; MSUD Type IA	248600	AR	BCKDHA	608348
MAPLE SYRUP URINE DISEASE; MSUD Type IB	248600	AR	BCKDHB	248611
MAPLE SYRUP URINE DISEASE; MSUD Type II	248600	AR	DBT	248610
MASA SYNDROME (CRASH SYNDROME)	303350	XLR	L1CAM	308840
MECKEL SYNDROME, TYPE 1; MKS1	249000	AR	MKS1	609883
MECKEL SYNDROME, TYPE 2; MKS2	603194	AR	TMEM216	613277
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	POLG1	174763
MITOCHONDRIAL DNA DEPLETION SYNDROME 4B (MNGIE TYPE); MTDPS4B	613662	AR	POLG1	174763
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY; MTPD	609015	AR	HADHA	600890
MUCOLIPIDOSIS II ALPHA/BETA	252500	AR	GNTPTAB	607840
MUCOLIPIDOSIS III ALPHA/BETA	252600	AR	GNTPTAB	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, DUCHENNE TYPE; DMD	310200	XLR	DMD	300377
MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2A	253600	AR (alelické AD)	CAPN3	114240
MYASTHENIC SYNDROME, CONGENITAL, 4A, SLOW-CHANNEL; CMS4A	605809	AR (vzácně AD)	CHRNE	100725
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
NEPHROTIC SYNDROME, TYPE 1; NPHS1	256300	AR	NPHS1	602716
NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III; HSAN3 (Familial Dysautonomia)	223900	AR	IKBKAP	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
NIJMEGEN BREAKAGE SYNDROME; NBS	251260	AR	NBN	602667
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
PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, AUTOSOMAL RECESSIVE 1; PEOB1	258450	AR	POLG1	174763
PROPIONIC ACIDEMIA	606054	AR	PCCB	232050
RETINITIS PIGMENTOSA 28; RP28	606068	AR	FAM161A	613596
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
SALLA DISEASE; SD	604369	AR	SLC17A5	604322
SANDHOFF DISEASE	268800	AR	HEXB	606873
SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS; SANDO	607459	AR	POLG1	174763
SICKLE CELL ANEMIA	603903	AR	HBB	141900
SMITH-LEMLI-OPITZ SYNDROME; SLOS	270400	AR	DHCR7	602858
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	235400	AR	SMN1	600354
SPINAL MUSCULAR ATROPHY, TYPE IV; SMA4	271150	AR	SMN1	600354
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 7; SCAR7	609270	AR	TPP1	607998
STARGARDT DISEASE 1; STGD1	248200	AR	CNGB3	605080
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
XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A; XPA	278700	AR	XPA	611153

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