CENTRUM LÉKAŘSKÉ GENETIKY s.r.o.
Novohradská 68/1806, 370 08 České Budějovice

Seznam vyšetřovaných genů/onemocnění v rámci CLG Carrier Screen panelu

Seznam vyšetřovaných genů/onemocnění
CLG Carrier Screen panel, design v3, verze 3.1

Fenotyp	Fenotyp MIM kód	Dědičnost	Gen	Gen MIM kód
Acyl-CoA dehydrogenase, medium chain, deficiency	201450	AR	*ACADM*	607008
Acyl-CoA dehydrogenase, short-chain, deficiency	201470	AR	*ACADS*	606885
Acyl-CoA dehydrogenase, very long-chain, deficiency	201475	AR	*ACADVL*	609575
Usher syndrome, type 2C	605472	AR, DD	*ADGRV1*	602851
Glycogen storage disease III	232400	AR	*AGL*	610860
Fructose intolerance, hereditary	229600	AR	*ALDOB*	612724
Hypophosphatasia	146300	AD, AR	*ALPL*	171760
Androgen insensitivity	300068	XLR	*AR*	313700
Argininemia	207800	AR	*ARG1*	608313
Metachromatic leukodystrophy	250100	AR	*ARSA*	607574
Argininosuccinic aciduria	207900	AR	*ASL*	608310
Canavan disease	271900	AR	*ASPA*	608034
Citrullinemia	215700	AR	*ASS1*	603470
Ataxia-telangiectasia	208900	AR	*ATM*	607585
Wilson disease	277900	AR	*ATP7B*	606882
Maple syrup urine disease, type Ia	248600	AR	*BCKDHA*	608348
Maple syrup urine disease, type Ib	248600	AR	*BCKDHB*	248611
Homocystinuria due to cystathionine beta-synthase deficiency	236200	AR	*CBS*	613381
Usher syndrome, type 1	601067	AR, DR	*CDH23*	605516
Cystic fibrosis	219700	AR	*CFTR*	602421
Congenital bilateral absence of vas deferens	277180	AR	*CFTR*	602421
Ceroid lipofuscinosis, neuronal, 3	204200	AR	*CLN3*	607042
Achromatopsia 3	262300	AR	*CNGB3*	605080
Alport syndrome 1	301050	XLD	*COL4A5*	303630
Carnitine Palmitoyltransferase I Deficiency, hepatic, type IA	255120	AR	*CPT1A*	600528
Carnitine Palmitoyltransferase II Deficiency, lethal neonatal	608836	AR	*CPT2*	600650
Carnitine Palmitoyltransferase II Deficiency, infantile	600649	AR	*CPT2*	600650
Cystinosis, nephropathic	219800	AR	*CTNS*	606272
Cystinosis, late-onset juvenile or adolescent nephropathic	219900	AR	*CTNS*	606272
Adrenal hyperplasia, congenital, due to 17-Alpha-hydroxylase Deficiency	202110	AR	*CYP17A1*	609300
Maple syrup urine disease, type II	248600	AR	*DBT*	248610
Smith-Lemli-Opitz syndrome	270400	AR	*DHCR7*	602858
Prothrombin (G20210A)		AD	*F2*	176930
Factor V (G1691A)		AD	*F5*	612309
Tyrosinemia, type I	276700	AR	*FAH*	613871
FSHR c.2039G>A (p.Ser680Asn)		AD	*FSHR*	136435
Glycogen storage disease Ia	232200	AR	*G6PC1*	613742
Glycogen storage disease II	232300	AR	*GAA*	606800
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
Glutaric acidemia	231670	AR	*GCDH*	608801
Deafness, autosomal recessive 1A	220290	AR, DD	*GJB2*	121011
Fabry disease	301500	XL	*GLA*	300644
GM1-gangliosidosis, type I	230500	AR	*GLB1*	611458
GM1-gangliosidosis, type II	230600	AR	*GLB1*	611458

GM1-gangliosidosis, type III	230650	AR	*GLB1*	611458
Glycine encephalopathy 1	605899	AR	*GLDC*	238300
Mucolipidosis II alpha/beta	252500	AR	*GNPTAB*	607840
Mucolipidosis III alpha/beta	252600	AR	*GNPTAB*	607840
Long-chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency	609016	AR	*HADHA*	600890
Mitochondrial trifunctional protein deficiency 1	609015	AR	*HADHA*	600890
Thalassemia, beta	613985	AR	*HBB*	141900
Sickle cell anemia	603903	AR	*HBB*	141900
Tay-Sachs disease	272800	AR	*HEXA*	606869
Myasthenic syndrome, congenital, 4A, slow-channel	605809	AD, AR	*CHRNE*	100725
Myasthenic syndrome, congenital, 4B, fast-channel	616324	AR	*CHRNE*	100725
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	608931	AR	*CHRNE*	100725
Mucopolysaccharidosis I	607014/15/16	AR	*IDUA*	252800
Severe combined immunodeficiency, X-linked	300400	XLR	*IL2RG*	308380
Isovaleric acidemia	243500	AR	*IVD*	607036
3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200	AR	*MCCC1*	609010
3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210	AR	*MCCC2*	609014
Familial Mediterranean fever	249100, 134610	AR, AD	*MEFV*	608107
Myopathy, centronuclear, X-linked	310400	XLR	*MTM1*	300415
Usher syndrome, type 1B	276900	AR	*MYO7A*	276903
Deafness, autosomal recessive 2	600060	AR	*MYO7A*	276903
Nijmegen breakage syndrome	251260	AR	*NBN*	602667
Niemann-Pick disease, type C1, D	257220	AR	*NPC1*	607623
Niemann-pick disease, type C2	607625	AR	*NPC2*	601015
Optic atrophy 1	165500	AD	*OPA1*	605290
Ornithine transcarbamylase deficiency	311250	XL	*OTC*	300461
Phenylketonuria	261600	AR	*PAH*	612349
Usher syndrome, type 1	601067	AR, DR	*PCDH15*	605514
Peroxisome biogenesis disorder 1A (Zellweger)	214100	AR	*PEX1*	602136
Peroxisome biogenesis disorder 6A (Zellweger)	614870	AR	*PEX10*	602859
Peroxisome biogenesis disorder 3A (Zellweger)	614859	AR	*PEX12*	601758
Peroxisome biogenesis disorder 11A (Zellweger)	614883	AR	*PEX13*	601789
Peroxisome biogenesis disorder 13A (Zellweger)	614887	AR	*PEX14*	601791
Peroxisome biogenesis disorder 8A (Zellweger)	614876	AR	*PEX16*	603360
Peroxisome biogenesis disorder 5A (Zellweger)	614866	AR	*PEX2*	170993
Peroxisome biogenesis disorder 9B	614879	AR	*PEX7*	601757
Polycystic kidney disease 1	173900	AD	*PKD1*	601313
Polycystic kidney disease 2	613095	AD	*PKD2*	173910
Polycystic kidney disease 4, with or without hepatic disease	263200	AR	*PKHD1*	606702
Congenital disorder of glycosylation, type Ia	212065	AR	*PMM2*	601785
Disordered steroidogenesis due to cytochrome P450 oxidoreductase	613571	AR	*POR*	124015
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	616326	AR	*RAPSN*	601592
Alpha-1-antitrypsin deficiency	613490	AR	*SERPINA1*	107400
Mucopolysaccharidosis type IIIA	252900	AR	*SGSH*	605270
Carnitine deficiency	212140	AR	*SLC22A5*	603377
Carnitine-acylcarnitine translocase deficiency	212138	AR	*SLC25A20*	613698
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791	AR	*SLC26A4*	605646
Pendred syndrome	274600	AR	*SLC26A4*	605646
Spinal muscular atrophy-1	253300	AR	*SMN1*	600354
Spinal muscular atrophy, type III, modifier	253400	AR	*SMN2*	601627
Niemann-Pick disease, type A	257200	AR	*SMPD1*	607608
Niemann-Pick disease, type B	607616	AR	*SMPD1*	607608

Ichthyosis, congenital, autosomal recessive 1	242300	AR	*TGM1*	190195
Spinocerebellar ataxia, autosomal recessive 7	609270	AR	*TPP1*	607998
Ceroid lipofuscinosis, neuronal, 2	204500	AR	*TPP1*	607998
Usher syndrome, type 1C	276904	AR	*USH1C*	605242
Usher syndrome, type 2A	276901	AR	*USH2A*	608400
Retinitis pigmentosa 39	613809	AR	*USH2A*	608400

Vysvětlivky: **AR**-autozomálně recesivní; **AD**-autozomálně dominantní; **DD**-digenní dominantní; **DR**-digenní recesivní; **XL**-X vázané; **XLR**-X vázané recesivní; **XLD**-vázané dominantní