PANDA CARRIER
Popis vyšetřovaných genů a onemocnění

Gen	Onemocnění
AAAS	Achalasia-addisonianism-alacrima syndrome
ABCB11	Progressive familial intrahepatic cholestasis, type II
ABCC6	Pseudoxanthoma elasticum
ABCC8	Familial hyperinsulinemic hypoglycemia type 1
ABCD1	Adrenoleukodystrophy (X-linked)
ACADM	Medium chain Acyl-CoA dehydrogenase deficiency
ACADS	Short chain Acyl-CoA dehydrogenase deficiency
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency
ACADVL	Very long chain Acyl-CoA dehydrogenase deficiency
ACAT1	Beta-ketothiolase deficiency (Alpha-methylacetoacetic aciduria)
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ADA	Severe combined immunodeficiency due to ADA deficiency
ADAMTS2	Ehlers Danlos syndrome, type VIIC
ADAR	Aicardi-Goutieres syndrome 6
ADGRG1	Bilateral frontoparietal polymicrogyria
AGA	Aspartylglycosaminuria
AGL	Glycogen storage disease, type III (a&b)
AGPS	Rhizomelic chondrodysplasia punctata, type III
AGXT	Primary hyperoxaluria, type I
AIRE	Polyglandular autoimmune syndrome, type I (Autoimmune polyendocrinopathy syndrome type I, with or without reversible metaphyseal dysplasia)
ALDH3A2	Sjögren-Larsson syndrome
ALDH7A1	Pyridoxine-dependent epilepsy
ALDOB	Hereditary fructose intolerance
ALG6	Congenital disorder of glycosylation, type Ic
ALPL	Hypophosphatasia, autosomal recessive
AMT	Glycine encephalopathy, AMT-related
AP1S1	MEDNIK syndrome
AP3B1	Hermansky-Pudlak syndrome, type 2
AR	Androgen insensitivity syndrome
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type VI
ASL	Argininosuccinic aciduria
ASNS	Asparagine Synthetase deficiency

ASPA	Canavan disease
ASS1	Citrullinemia, type I
ATM	Ataxia-telangiectasia
ATP13A2	Kufor-Rakeb syndrome (KRS);
	Autosomal recessive spastic paraplegia-78 (SPG78)
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related
ATP7B	Wilson disease
BBS1	Bardet-Biedl syndrome 1
BBS2	Bardet-Biedl syndrome 2
BBS4	Bardet-Biedl syndrome 4
BBS7	Bardet-Biedl syndrome 7
BBS9	Bardet-Biedl syndrome 9
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BCS1L	GRACILE syndrome;
	Bjornstad syndrome;
	Leigh syndrome;
	Mitochondrial complex III deficiency, nuclear type 1
BLM	Bloom syndrome
BSND	Bartter syndrome, type IV
BTD	Biotinidase deficiency
CANT1	Desbuquois dysplasia, type I;
	Epiphyseal dysplasia, multiple, 7
CAPN3	Limb-girdle muscular dystrophy, type 2A
CBS	Homocystinuria, CBS-related
CDH23	Usher syndrome, type ID;
	Deafness, autosomal recessive 12
CEP290	Leber congenital amaurosis 10;
	Joubert syndrome 5;
	Meckel syndrome 4;
	Senior-Loken syndrome 6
CERKL	Retinitis pigmentosa 26
CFTR	Cystic fibrosis;
	Congenital bilateral absence of vas deferens
CHAT	Congenital myasthenic syndrome 6
CHM	Choroideremia
CHRNE	Congenital myasthenic syndrome 4A;
	Congenital myasthenic syndrome 4B;
	Congenital myasthenic syndrome 4C
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related

CLN6	Neuronal ceroid lipofuscinosis, CLN6-related
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related;
	Northern epilepsy
CLRN1	Usher syndrome, type IIIA
CNGA3	Achromatopsia 2, CNGA3-related
CNGB3	Achromatopsia 3, CNGB3-related
COL4A3	Alport syndrome
COL4A4	Alport syndrome 2
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa, autosomal recessive
COLQ	Congenital myasthenic syndrome 5
CPT1A	Carnitine palmitoyltransferase IA deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CRB1	Leber congenital amaurosis 8; Retinitis pigmentosa-12, autosomal recessive
CTNS	Cystinosis, atypical nephropathic;
	Cystinosis, late-onset juvenile or adolescent nephropathic;
	Cystinosis, nephropathic;
	Cystinosis, ocular nonnephropathic
CTSD	Neuronal ceroid lipofuscinosis 10
CTSF	Neuronal ceroid lipofuscinosis 13
CTSK	Pycnodysostosis
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CYP11B2	Cortisone methyl oxidase type II deficiency; Cortisone methyl oxidase type I deficiency
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Primary congenital glaucoma 3A
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type I
DBT	Maple syrup urine disease, type II
DCLRE1C	Omenn syndrome;
	Severe combined immunodeficiency, Athabascan type
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa 59
DKC1	Dyskeratosis congenita, X-linked
DLD	Dihydrolipoamide dehydrogenase deficiency
DMD	Duchenne/Becker muscular dystrophy
DNAH5	Primary ciliary dyskinesia type 3, DNAH5-related
DNAI1	Primary ciliary dyskinesia type 1, DNAI1-related
DNAI2	Primary ciliary dyskinesia type 9, DNAI2-related
DNAJC5	Neuronal ceroid lipofuscinosis type 4

DOK7	Fetal akinesia deformation sequence, DOK7-related; Congenital myasthenic syndrome, 10
DPYD	Dihydropyrimidine dehydrogenase deficiency
DYSF	Limb-girdle muscular dystrophy, type 2B; Miyoshi myopathy and distal myopathy with anterior tibial onset
EDA	Hypohidrotic ectodermal dysplasia, X-linked
EDAR	Hypohidrotic ectodermal dysplasia 10B
EMD	Emery-Dreifuss muscular dystrophy
ERCC2	Xeroderma pigmentosum
ETFA	Glutaric acidemia, Type IIA
ETFB	Glutaric acidemia, Type IIB
ETFDH	Glutaric acidemia, Type IIC
ETHE1	Ethylmalonic encephalopathy
EXOSC3	Pontocerebellar hypoplasia type 1B
EYS	Retinitis pigmentosa 25, EYS-related
F8	Hemophilia A
F9	Hemophilia B
F11	Factor XI Deficiency
FAH	Tyrosinemia, type I
FAM161A	Retinitis pigmentosa 28
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FANCG	Fanconi Anemia, complementation group G
FH	Fumarase deficiency
FKRP	Limb-girdle muscular dystrophy, type 2I; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
FKTN	Muscular dystrophy-dystroglycanopathy type 4A (Walker-Warburg syndrome); Muscular dystrophy-dystroglycanopathy type 4B; Muscular dystrophy-dystroglycanopathy type 4C; Cardiomyopathy, dilated, 1X
FMR1	Fragile X syndrome; FMR1-related primary ovarian insufficiency; Fragile X-associated tremor/ataxia syndrome
G6PC	Glycogen storage disease, type IA
G6PD	Glucose-6-phosphate dehydrogenase deficiency; Hemolytic anemia due to G6PD deficiency
GAA	Glycogen storage disease, type II
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency
GALNT3	Hyperphospatemic tumoral calcinosis, familial
GALT	Galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GBA	Gaucher disease
GBE1	Glycogen storage disease, type IV
GCDH	Glutaric acidemia, type I

GCSH	Nonketotic hyperglycinemia
GDF5	Du Pan syndrome; Chondrodysplasia, Grebe type; Brachydactyly type A1,C; Acromesomelic dysplasia, Hunter-Thompson type
GFPT1	Congenital myasthenic syndrome 12
GJB1	Charcot-Marie-Tooth disease, GJB1-related
GJB2	Nonsyndromic hearing loss, GJB2-related
GJB6	Nonsyndromic Hearing Loss, GJB6-related
GLA	Fabry disease
GLB1	GM1-gangliosidosis; Mucopolysaccharidosis type IVB
GLDC	Glycine encephalopathy, GLDC-related
GNE	Inclusion body myopathy, type II
GNPAT	Rhizomelic chondrodysplasia punctata, type II
GNPTAB	Mucolipidosis, type II alpha/beta; Mucolipidosis, type III alpha/beta
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis type IIID
GORAB	Geroderma osteodysplastica
GRHPR	Primary hyperoxaluria, type II
GRN	Neuronal ceroid lipofuscinosis type 11
GUCY2D	Leber congenital amaurosis 1; Choroidal dystrophy, central areolar 1
HADH	Familial hyperinsulinemic hypoglycemia, familial 4; 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	Long-chain 3-Hydroxyacyl-CoA dehydrogenase deficiency; Trifunctional protein deficiency
HADHB	Mitochondrial trifunctional protein deficiency
HAX1	Severe congenital neutropenia 3, autosomal recessive
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, and other hemoglobinopathies
HEPACAM	Megalencephalic Leukoencephalopathy with Subcortical Cysts, types 2A & 2B
HEXA	Tay-Sachs disease; GM2-ganlgliosidosis
HEXB	Sandhoff disease
HFE2	Hereditary hemochromatosis type 2A, HFE2-related
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, type IIIC
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaryl CoA lyase deficiency
HOGA1	Primary hyperoxaluria, type III
HPRT1	Lesch-Nyhan syndrome; HPRT-related gout
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome, type 3
HPS4	Hermansky-Pudlak syndrome, type 4
HPS5	Hermansky-Pudlak syndrome, type 5
HPS6	Hermansky-Pudlak syndrome, type 6
HSD17B3	17-beta-hydroxysteroid dehydrogenase deficiency, type III

HSD17B4	D-bifunctional protein deficiency;
	Perrault syndrome 1
HSD3B2	Congential adrenal hyperplasia due to 3-Beta-hydroxysteroid dehydrogenase deficiency, type II
IDS	Mucopolysaccharidosis, type II (Hunter syndrome)
IDUA	Mucopolysachharidosis, type I (Hurler syndrome)
IKBKAP	Familial dysautonomia (HSAN3)
IL2RG	Severe combined immunodeficiency, X-linked
ISPD	Walker-Warburg (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7
IVD	Isovaleric acidemia
KCNJ11	Familial hyperinsulinemic hypoglycemia type 2, KCNJ11-related
KCTD7	Neuronal ceroid lipofuscinosis 14 (progressive myoclonic epilepsy type 3)
L1CAM	L1 syndrome;
	MASA syndrome, CRASH syndrome
LAMA3	Herlitz junctional epidermolysis bullosa, LAMA3-related; Laryngoonychocutaneous syndrome;
	Epidermolysis bullosa, generalized atrophic benign
LAMB3	Herlitz junctional epidermolysis bullosa;
	non-Herlitz junctional epidermolysis bullosa
LAMC2	Herlitz junctional epidermolysis bullosa;
	non-Herlitz junctional epidermolysis bullosa
LARGE1	Muscular dystrophy-dystroglycanopathy, congenital with brain and eye anomalies, type 6A (Walker-Warburg); Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type 6B
LCA5	Leber congenital amaurosis 5, LCA5-related
LHCGR	Leydig cell hypoplasia; Luteinizing hormone resistance
LIFR	Stuve-Wiedemann syndrome
LIPA	Cholesteryl ester storage disease
LIPH	Woolly hair/hypotrichosis, autosomal recessive
LOXHD1	Autosomal recessive deafness 77
LPL	Lipoprotein lipase deficiency
LRPPRC	Leigh syndrome with COX deficiency (French Canadian type)
LYST	Chediak-Higashi syndrome
MAN2B1	Alpha-mannosidosis type I & II
MCCC1	3-Methylcrontonyl-CoA carboxylase 1 deficiency
MCCC2	3-Methylcrontonyl-CoA carboxylase 2 deficiency
MCOLN1	Mucolipidosis type IV
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEFV	Familial Mediterranean fever
MFSD8	Neuronal ceroid-lipofuscinosis 7, MFSD8-related;
	Macular dystrophy with central cone involvement
MKKS	Bardet-Biedl syndrome 6; McKusick-Kaufman syndrome
MKS1	Bardet-Biedl syndrome 13;

	Joubert syndrome 28;
	Meckel syndrome 1
MLC1	Megalencephalic Leukoencephalopathy with subcortical cysts, type I
MMAA	Methylmalonic aciduria, cblA type
MMAB	Methylmalonic aciduria, cblB type
MMADHC	Methylmalonic aciduria, cblD type
MPI	Congenital disorder of glycosylation, type IB
MPL	Congenital amegakaryocytic thrombocytopenia
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome 6, MPV17-related
MRE11A	Ataxia-telangiectasia-like disorder 1
MTHFR	Homocystinuria due to MTHFR deficiency, severe;
	Neural tube defects folate-sensitive
MTM1	Myotubular myopathy, MTM1-related
MTTP	Abetalipoproteinemia
MUT	Methylmalonic aciduria mut(0) type, MUT-related
MYO7A	Usher syndrome, type IB;
	Deafness, autosomal recessive 2
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NDUFAF6	Mitochondrial complex 1 deficiency, NDUFAF6-related
NEB	Nemaline myopathy 2
NPC1	Niemann-pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHS1	Nephrotic syndrome, type I
NPHS2	Nephrotic syndrome, type II
NR2E3	Enhanced S-cone syndrome
NTRK1	Congenital insensitivity to pain with anhidrosis
OPA3	3-methylglutaconic aciduria, type III
OTC	Ornithine transcarbamylase deficiency
PAH	Phenylalanine hydroxylase deficiency
PCCA	Propionic acidemia, PCCA-related
PCCB	Propionic acidemia, PCCB-related
PCDH15	Usher syndrome, type IF;
	Deafness, autosomal recessive 23
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, X-Linked
PDHB	Pyruvate dehydrogenase E1-beta deficiency, autosomal recessive
PEPD	Prolidase deficiency
PET100	Mitochondrial complex IV deficiency
PEX1	Zellweger syndrome spectrum, PEX1-related
PEX2	Zellweger syndrome spectrum, PEX2-related
PEX6	Zellweger syndrome spectrum, PEX6-related
PEX7	Rhizomelic chondrodysplasia punctata, type I;
	Peroxisome biogenesis disorder 9B
PEX10	Zellweger syndrome spectrum, PEX10- related

PEX12	Zellweger syndrome spectrum, PEX12- related
PEX26	Zellweger syndrome spectrum, PEX26- related
PFKM	Glycogen storage disease, type VII
PHGDH	3-phosphoglycerate dehydrogenase deficiency;
	Neu-Laxova syndrome 1
PKHD1	Polycystic kidney disease, autosomal recessive
PMM2	Congenital disorder of glycosylation, type IA
POLG	Mitochondrial DNA depletion syndrome type 4A (Alpers type);
	Mitochondrial DNA depletion syndrome type 4B (MNGIE type);
	Mitochondrial Recessive Ataxia Syndrome (includes SANDO and SCAE);
	Progressive external ophthalmoplegia with mitochondrial deletions autosomal recessive type 1
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type 3A;
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type 3B;
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type 3C
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1;
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1;
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2;
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2;
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2
PPT1	Neuronal ceroid lipofuscinosis 1, PPT1-related
PRPS1	Arts Syndrome; Charcot-Marie-Tooth disease, PRPS1-related;
	Gout, PRPS1-related
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency; Atypical Gaucher disease; Atypical Krabbe disease
PUS1	Mitochondrial myopathy and sideroblastic anemia 1
PYGL	Glycogen storage disease VI
PYGM	Glycogen storage disease, type V
RAB23	Carpenter syndrome
RAG1	Severe combined immunodeficiency, RAG1-related;
	Omenn syndrome;
	Combined cellular and humoral immune defects with granulomas
RAG2	Omenn syndrome;
	Severe combined immunodeficiency, Athabascan type
RAPSN	Congenital myasthenic syndrome 11, RAPSN-related;
	Fetal akinesia deformation sequence
RARS2	Pontocerebellar hypoplasia type 6
RDH12	Leber congenital amaurosis 13
RNASEH2A	Aicardi-Goutieres syndrome 4
RNASEH2B	Aicardi-Goutieres syndrome 2

RNASEH2C	Aicardi-Goutieres syndrome 3, RNASEH2C-related
RPE65	Leber congenital amaurosis 2;
	Retinitis pigmentosa 20
RS1	Juvenile retinoschisis, X-linked
RTEL1	Dyskeratosis congenita, autosomal recessive 5
SACS	Spastic ataxia of Charlevoix-Saguenay, autosomal recessive
SAMD9	Familial tumoral calcinosis, normophosphatemic
SAMHD1	Aicardi-Goutieres syndrome 5
SBDS	Shwachman-Diamond syndrome
SEPSECS	Pontocerebellar hypoplasia 2D
SERPINA1	Alpha-1 antitrypsin deficiency
SGCA	Limb-girdle muscular dystrophy, type 2D
SGCB	Limb-girdle muscular dystrophy, type 2E
SGCG	Limb-girdle muscular dystrophy, type 2C
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A)
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)
SLC17A5	Salla disease; Infantile sialic acid storage disorder
SLC22A5	Primary carnitine deficiency
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Ornithine translocase deficiency)
SLC26A2	Sulfate transporter-related osteochondrodysplasia;
	Achondrogenesis Ib;
	Atelosteogenesis II;
	Diastrophic dysplasia;
	Epiphyseal dysplasia, multiple, 4
SLC26A4	Pendred syndrome;
	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
SLC35A3	Arthrogryposis, mental retardation and seizures
SLC37A4	Glycogen storage disease, type 1b; Gycogen storage disease type 1c
SLC39A4	Acrodermatitis enteropathica
SLC4A11	Corneal dystrophy and perceptive deafness syndrome; Autosomal recessive corneal dystrophy
SLC6A8	Creatine transporter defect, SLC6A8-related (Cerebral creatine deficiency syndrome 1)
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease type A; Niemann-Pick disease type B
ST3GAL5	Amish infantile epilepsy syndrome
STAR	Congenital lipoid adrenal hyperplasia
STS	X-linked ichthyosis
SUMF1	Multiple sulphatase deficiency
TAT	Tyrosinemia type II (Richner-Hanhart syndrome)
TCIRG1	Osteopetrosis type 1, infantile malignant
TECPR2	Hereditary spastic paraparesis, type 49
TFR2	Hereditary hemochromatosis type 3, TFR2-related

TGM1	Lamellar ichthyosis type I
TH	Segawa syndrome (tyrosine hydroxylase deficiency)
TMEM216	Joubert syndrome 2; Meckel syndrome 2
TPP1	Neuronal ceroid lipofuscinosis 2, TPP1-related; Spinocerebellar ataxia type 7
TREX1	Aicardi-Goutieres syndrome type 1, TREX1-related
TRIM37	Mulibrey nanism
TSEN2	Pontocerebellar hypoplasia type 2B
TSEN34	Pontocerebellar hypoplasia type 2C
TSEN54	Pontocerebellar hypoplasia type 2A;
	pontocerebellar hypoplasia type 4;
	Pontocerebellar hypoplasia type 5
TTC8	Bardet-Biedl syndrome 8;
	Retinitis pigmentosa 51
TTN	Early onset myopathy with fatal cardiomyopathy;
	Limb-girdle muscualr dystrophy 2J;
	Salih myopathy
TTPA	Ataxia with vitamin E deficiency
UBR1	Johanson-Blizzard syndrome
UGT1A1	Crigler-Najjar syndrome, type I;
	Crigler-Najjar syndrome, type II;
	Hyperbilirubinemia, familial transient neonatal Gilbert syndrome
USH1C	Usher syndrome, type IC;
	Deafness, autosomal recessive 18A
USH2A	Usher syndrome, Type 2A;
	Retinitis pigmentosa 39
VPS13A	Choreoacanthocytosis
VPS53	Pontocerebellar hypoplasia 2E
VRK1	Pontocerebellar hypoplasia, type 1A
XPA	Xeroderma pigmentosum group A
XPC	Xeroderma pigmentosum group C
ZFYVE26	Spastic paraplegia type 15, ZFYVE26-related