ZNÁTE SVOU GENETICKOU VÝBAVU ?!
OTESTUJTE SE ...

Pro koho je SGT určený?

Test je určen pro všechny zájemce, kteří chtějí zjistit, zda je jejich genetická výbava v pořádku. Cílem testu je významně omezit genetickou zátěž především pro páry plánující rodinu.

Vyšetření DNA profilů určí, zdali partneři nesdílí takovou genetickou výbavu, která by mohla ohrozit zdraví společných potomků.

Kdy je možno očekávat výsledek testu?

Analýza uvedeného širokého spektra genetických variant probíhá metodou „sekvenování nové generace (NGS)“. Tento proces je analyticky a interpretačně náročný a výsledková zpráva je tak k dispozici do 2-3 měsíců od odběru.

Cena testu: 9 900,- Kč
Číslo účtu: 1828978329/0800,
Variabilní symbol: 81601
Zpráva pro příjmece: SGT

QR platba

U.S.G.POL s.r.o.
1828978329/0800

www.usgpol.cz

Jak se na SGT objednat? Kde a jak se provádí odběr vzorků?

- ***Odběr v Centru U.S.G. POL***

  Kontaktujte prosím telefonicky či prostřednictvím e-mailu centrum U.S.G. POL.

  Obdržíte termín (datum a hodinu) vyšetření. V ordinaci podepíšete informovaný souhlas. Následně Vám bude proveden odběr malého množství žilní krve. Před odběrem se můžete najíst.

  Jako první se provádí odběr krevního vzorku u ženy - žadatelky. V případě, že test prokáže významnou DNA abnormalitu, následuje další odběr u partnera pro komplexní vyhodnocení genetického rizika.

- ***Odběr na smluvním pracovišti***

  Odběr vzorku lze provádět i přímo u gynekologa či praktického lékaře.

V případě pozitivního nálezu je další postup řešen klinickým genetikem.

Genetická poradna a laboratoř molekulární diagnostiky U.S.G.POL uvádí do klinické praxe

SAFETY GENE TEST (SGT)

Co je Safety Gene Test (SGT)?

Jde o test na určení přenašečství genetických onemocnění, které mohou podmiňovat dědičnou zátěž jedince, příbuzných a potomků.

Test odráží nejnovější vývoj v oblasti DNA diagnostiky.

Jedná se o vyšetření genů, jejichž poruchy mohou vést k nejčastějším a nejzávažnějším genetickým onemocněním. Jde např. o cystickou fibrózu, spinální muskulární atrofii, svalové dystrofie, mentální poruchy, hluchotu, dědičné metabolické vady, jakými jsou fenylketonurie, cystinurie, glykogenózy, apod. . ..

Celkem jsou vyšetřeny **varianty genů pro víc jak 400 nemocí**.

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Seznam nemocí a vyšetřovaných genů

17-beta hydroxysteroid dehydrogenase 3 deficiency	HSD17B3
3-beta-hydroxysteroid dehydrogenase type II deficiency	HSD3B2
3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency	HMGCL
3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1
3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2
5-alpha reductase deficiency	SRD5A2
6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency	PTS
Abetalipoproteinemia	MTTP
Achalasia-addisonianism-alacrima syndrome	AAAS
Achondrogenesis, type 1B	SLC26A2
Achromatopsia, CNGA3-related	CNGA3
Achromatopsia, CNGB3-related	CNGB3
Acrodermatitis enteropathica	SLC39A4
Acute infantile liver failure, TRMU-related	TRMU
Acyl-CoA oxidase I deficiency	ACOX1
Adrenoleukodystrophy, X-linked	ABCD1
Agenesis of the corp.callosum with peripheral neuropathy (Andermann sy)	SLC12A6
Aicardi-Goutieres syndrome, RNASEH2C-related	RNASEH2C
Aicardi-Goutieres syndrome, TREX1-related	TREX1
Aicardi-Goutires syndrome	SAMHD1
Alkaptonuria	HGD
Alpha-1-antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia	HBA1
Alpha-thalassemia	HBA2
Alpha-thalassemia intellectual disability syndrome, X-linked	ATRX
Alport syndrome, COL4A3-related	COL4A3
Alport syndrome, COL4A4-related	COL4A4
Alport syndrome, X-linked	COL4A5
Alström syndrome	ALMS1
Androgen insensitivity syndrome, X-linked	AR
Argininemia	ARG1
Argininosuccinate lyase deficiency	ASL
Aromatase deficiency	CYP19A1
Arts syndrome, X-linked	PRPS1
Asparagine synthetase deficiency	ASNS
Aspartylglucosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia	ATM
Ataxia-telangiectasia-like disorder 1	MRE11
Autism spectrum, epilepsy, and arthrogryposis	SLC35A3
Autoimmune polyendocrinopathy syndrome, type I	AIRE
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	SACS
Bardet-Biedl syndrome 1	BBS1
Bardet-Biedl syndrome 10	BBS10
Bardet-Biedl syndrome 11	TRIM32
Bardet-Biedl syndrome 12	BBS12
Bardet-Biedl syndrome 2	BBS2
Bardet-Biedl syndrome 4	BBS4
Bardet-Biedl syndrome 6	MKKS
Bardet-Biedl syndrome 9	BBS9
Bare lymphocyte syndrome, CIITA-related	CIITA
Bartter syndrome, type 4a	BSND
Bernard-Soulier syndrome, type A2	GP1BA
Bernard-Soulier syndrome, type B	GP1BB
Bernard-Soulier syndrome, type C	GP9
Beta-hemoglobinopathies	HBB
Beta-ketothiolase deficiency	ACAT1
Beta-ureidopropionase deficiency	UPB1
Bilateral frontoparietal polymicrogyria	ADGRG1
Biotinidase deficiency	BTD
Bloom syndrome	BLM
Canavan disease	ASPA
Carbamoyl phosphate synthetase I deficiency	CPS1
Carnitine deficiency	SLC22A5
Carnitine palmitoyltransferase IA deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency	CPT2
Carnitine-acylcarnitine translocase deficiency	SLC25A20
Carpenter syndrome	RAB23
Cartilage-hair hypoplasia	RMRP
Catecholaminergic polymorphic ventricular tachycardia	CASQ2
Cerebrotendinous xanthomatosis	CYP27A1
Ceroid lipofuscinosis, neuronal, 1	PPT1
Ceroid lipofuscinosis, neuronal, 10 (CLN10 disease)	CTSD
Ceroid lipofuscinosis, neuronal, 2	TPP1
Ceroid lipofuscinosis, neuronal, 3	CLN3
Ceroid lipofuscinosis, neuronal, 5	CLN5
Ceroid lipofuscinosis, neuronal, 6	CLN6
Ceroid lipofuscinosis, neuronal, 7	MFSD8
Ceroid lipofuscinosis, neuronal, 8 (a.k.a. Northern epilepsy)	CLN8
Charcot-Marie-Tooth disease type 4D	NDRG1
Charcot-Marie-Tooth disease with deafness, X-linked	GJB1
Chediak-Higashi syndrome	LYST
Choreo-acanthocytosis	VPS13A
Choroideremia, X-linked	CHM
Chronic granulomatous disease, CYBA-related	CYBA
Chronic granulomatous disease, X-linked	CYBB
Ciliary dyskinesia, primary 1	DNAI1
Ciliary dyskinesia, primary 3	DNAH5
Ciliary dyskinesia, primary 9	DNAI2
Ciliary dyskinesia, primary, 16	DNAL1
Ciliopathies, RPGRIP1L-related	RPGRIP1L
Citrullinemia, type 1	ASS1
Citrullinemia, type II	SLC25A13
Cockayne syndrome, type A	ECC8
Cockayne syndrome, type B	ERCC6
Cohen syndrome	VPS13B
Combined malonic and methylmalonic aciduria	ACSF3
Combined oxidative phosphorylation deficiency 1	GFM1
Combined oxidative phosphorylation deficiency 3	TSFM
Combined pituitary hormone deficiency 2	PROP1
Congenital adrenal hyperplasia, 11-beta-hydroxylase-deficient	CYP11B1
Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency	CYP17A1
Congenital adrenal hyperplasia, 21-hydroxylase-deficient	CYP21A2
Congenital adrenal hypoplasia, X-linked	NROB1
Congenital amegakaryocytic thrombocytopenia	MPL
Congenital chloride diarrhea	SLC26A3
Congenital disorder of glycosylation, type 1A, PMM2-related	PMM2
Congenital disorder of glycosylation, type 1B	MPI
Congenital disorder of glycosylation, type 1C	ALG6
Congenital Finnish nephrosis	NPHS1
Congenital hyperinsulinism, KCNJ11-related	KCNJ11
Congenital hypothyroidism	TPO
Congenital hypothyroidism	TSHB
Congenital insensitivity to pain with anhidrosis (CIPA)	NTRK1
Congenital myasthenic syndrome, CHRNE-related	CHRNE
Congenital myasthenic syndrome, DOK7-related	DOK7
Congenital myasthenic syndrome, RAPSN-related	RAPSN
Congenital neutropenia, HAX1-related	HAX1
Congenital neutropenia, VPS45-related	VPS45
Corneal dystrophy and perceptive deafness	SLC4A11
Corticosterone methyloxidase deficiency	CYP11B2
Costeff syndrome (3-methylglutaconic aciduria, type 3)	OPA3
Creatine transporter defect (cerebral creatine deficiency sy 1, X-linked)	SLC6A8
Crigler-Najjar syndrome	UGT1A1
Cystic fibrosis	CFTR
Cystinosis	CTNS
Cystinuria, type A	SLC3A1
Cystinuria, type B	SLC7A9
Cytochrome P450 oxidoreductase deficiency	POR
Cytochrome-c oxidase deficiency	PET100
D-bifunctional protein deficiency	HSD17B4
Deafness, autosomal dominant 36, autosomal recessive 7	TMC1
Deafness, autosomal recessive 16	STRC
Deafness, autosomal recessive 77	LOXHD1
Deafness, autosomal recessive, 3	MYO15A
Desbuquois dysplasia 1	CANT1
Dihydrolipoamide dehydrogenase deficiency	DLD
Dihydropyrimidine dehydrogenase deficiency	DPYD
Dopa-responsive dystonia	GCH1
Duchenne/Becker muscular dystrophy	DMD
Dysautonomia, familial (IKBKAP or ELP1)	IKBKAP
Dyskeratosis congenita, RTEL1-related	RTEL1
Dyskeratosis congenita, X-linked	DKC1
Dystrophic epidermolysis bullosa, COL7A1-related	COL7A1
Ehlers-Danlos syndrome, type VIIC	ADAMTS2
Ellis-van Creveld syndrome, EVC-related	EVC
Ellis-van Creveld syndrome, EVC2-related	EVC2
Emery-Dreifuss muscular dystrophy 1, X-linked	EMD
Enhanced S-cone syndrome	NR2E3
Erythrokeratodermia variabilis et progressiva	GJB3
Escobar syndrome	CHRNG
Ethylmalonic encephalopathy	ETHE1
Fabry disease	GLA
Factor XI deficiency	F11
Familial dilated cardiomyopathy	TTN
Familial hypercholesterolemia, LDLR-related	LDLR
Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1
Familial hyperinsulinism, ABCC8-related	ABCC8
Familial Mediterranean fever	MEFV
Familial nephrogenic diabetes insipidus, AQP2-related	AQP2
Fanconi anemia, group A	FANCA
Fanconi anemia, group C	FANCC
Fanconi anemia, group G	FANCG
Fanconi anemia, group J	BRIP1
Fibrochondrogenesis, type 2	COL11A2
Fumarase deficiency	FH
Galactokinase deficiency (galactosemia, type II)	GALK1
Galactose epimerase deficiency	GALE
Galactosemia	GALT
Gaucher disease	GBA
Geroderma osteodysplastica	GORAB
Gitelman syndrome	SLC12A3
Glanzmann thrombasthenia	ITGB3
Glucose-6-phosphate dehydrogenase deficiency	G6PD
Glutaric acidemia, type 1	GCDH
Glutaric acidemia, type 2A	ETFA
Glutaric acidemia, type 2B	ETFB
Glutaric acidemia, type 2C	ETFDH
Glycine encephalopathy, AMT-related	AMT
Glycine encephalopathy, GLDC-related	GLDC
Glycogen storage disease, type IA	G6PC
Glycogen storage disease, type IB	SLC37A4
Glycogen storage disease, type II (Pompe disease)	GAA
Glycogen storage disease, type III (Cori/Forbes)	AGL
Glycogen storage disease, type IV	GBE1
Glycogen storage disease, type V (McArdle disease)	PYGM
Glycogen storage disease, type VII	PFKM
GM3 synthase deficiency	ST3GAL5
GRACILE syndrome	BCS1L
Grebe syndrome	GDF5
Guanidinoacetate methyltransferase deficiency	GAMT
Harlequin ichthyosis	ABCA12
Heme oxygenase-1 deficiency	HMOX1
Hemochromatosis, type 1	HFE
Hemochromatosis, type 2A	HFE2
Hemochromatosis, type 3, TFR2-related	TFR2
Hemophilia A	F8
Hemophilia B	F9
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17
Hereditary fructose intolerance	ALDOB
Hereditary spastic paraparesis, type 49	TECPR2
Herlitz junctional epidermolysis bullosa, LAMA3-related	LAMA3
Herlitz junctional epidermolysis bullosa, LAMB3-related	LAMB3
Herlitz junctional epidermolysis bullosa, LAMC2-related	LAMC2
Hermansky-Pudlak syndrome 1	HPS1
Hermansky-Pudlak syndrome 3	HPS3
Hermansky-Pudlak syndrome 4	HPS4
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria due to deficiency of MTHFR	MTHFR
Homocystinuria, CBS-related	CBS
Homocystinuria, type cblE	MTRR
Hydatidiform mole, recurrent	NLRP7
Hydrolethalus syndrome	HYLS1
Hypermethioninemia	MAT1A
Hyperomithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15
Hyperoxaluria, primary, type 1	AGXT
Hyperphosphatemic familial tumoral calcinosis	GALNT3
Hypohidrotic ectodermal dysplasia	EDAR
Hypohidrotic ectodermal dysplasia, X-linked	EDA
Hypophosphatasia, ALPL-related	ALPL
Hypothyroidism, congenital, nongoitrous, 1	TSHR
Inclusion body myopathy 2	GNE
Infantile neuroaxonal dystrophy 1	PLA2G6
Isolated growth hormone deficiency, type IA/II	GH1
Isolated growth hormone deficiency, type IB	GHRHR
Isolated growth hormone deficiency, type III, X-linked	BTK
Isovaleric acidemia	IVD
Joubert syndrome 2/Meckel syndrome 2	TMEM216
Juvenile nephronophthisis	NPHP1
Juvenile retinoschisis, X-linked	RS1
Krabbe disease	GALC
LAMA2-related muscular dystrophy	LAMA2
Lamellar ichthyosis, type 1	TGM1
Leber congenital amaurosis 1	GUCY2D
Leber congenital amaurosis 2	RPE65
Leber congenital amaurosis 8	CRB1
Leber congenital amaurosis, type CEP290	CEP290
Leber congenital amaurosis, type LCA5	LCA5
Leber congenital amaurosis, type RDH12	RDH12
Leigh syndrome	SURF1
Leigh syndrome, French-Canadian type	LRPPRC
Lethal congenital contracture syndrome 1	GLE1
Leukoencephalopathy with vanishing white matter	EIF2B5
Leydig cell hypoplasia	LHCGR
Limb-girdle muscular dystrophy, type 2A	CAPN3
Limb-girdle muscular dystrophy, type 2B	DYSF
Limb-girdle muscular dystrophy, type 2C	SGCG
Limb-girdle muscular dystrophy, type 2D	SGCA
Limb-girdle muscular dystrophy, type 2E	SGCB
Limb-girdle muscular dystrophy, type 2F	SGCD
Limb-girdle muscular dystrophy, type 2I	FKRP
Lipoid congenital adrenal hyperplasia	STAR
Lipoprotein lipase deficiency	LPL
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA
Lowe syndrome, X-linked	OCRL
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency	LIPA
Malonyl-CoA decarboxylase deficiency	MLYCD
Maple syrup urine disease, type 1A	BCKDHA
Maple syrup urine disease, type 1B	BCKDHB
Maple syrup urine disease, type 2	DBT
Meckel-Gruber syndrome, type 1	MKS1
Medium chain acyl-CoA dehydrogenase deficiency	ACADM
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1
Megaloblastic anemia syndrome	SLC19A2
Menkes syndrome, X-linked	ATP7A
Mental retardation, autosomal recessive 3	CC2D1A
Mental retardation, enteropathy, deafness, per neuropathy, ichthyosis, andkeratoderma (MEDNIK)	AP1S1
Metachromatic leukodystrophy, ARSA-related	ARSA
Metachromatic leukodystrophy, PSAP-related	PSAP
Methylmalonic aciduria and homocystinuria, type cblC	MMACHC
Methylmalonic aciduria and homocystinuria, type cblD	MMADHC
Methylmalonic aciduria, MMAA-related	MMAA
Methylmalonic aciduria, MMAB-related	MMAB
Methylmalonic aciduria, type mut(0)	MUT
Microcephaly, postnatal progressive, with seizures and brain atrophy	MED17
Microphthalmia/Anophthalmia, VSX2-related	VSX2
MIRAGE syndrome	SAMD9
Mitochondrial complex I deficiency	NDUFS4
Mitochondrial complex I deficiency, ACAD9-related	ACAD9
Mitochondrial complex I deficiency, NDUFAF5-related	NDUFAF5
Mitochondrial complex I deficiency, NDUFS6-related	NDUFS6
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	SUCLA2
Mitochondrial myopathy and sideroblastic anemia (MLASA1)	PUS1
Molybdenum cofactor deficiency	MOCS1
Mucolipidosis II/IIIA	GNPTAB
Mucolipidosis III gamma	GNPTG
Mucolipidosis, type IV	MCOLN1
Mucopolysaccharidosis, type I (Hurler syndrome)	IDUA
Mucopolysaccharidosis, type II (Hunter syndrome)	IDS
Mucopolysaccharidosis, type IIIA (Sanfilippo A)	SGSH
Mucopolysaccharidosis, type IIIB (Sanfilippo B)	NAGLU
Mucopolysaccharidosis, type IIIC (Sanfilippo C)	HGSNAT
Mucopolysaccharidosis, type IIID (Sanfilippo D)	GNS
Mucopolysaccharidosis, type IVA	GALNS
Mucopolysaccharidosis, type IVB / GM1 gangliosidosis	GLB1
Mucopolysaccharidosis, type VI (Maroteaux-Lamy)	ARSB
Mucopolysaccharidosis, type VII	GUSB
Mulibrey nanism syndrome	TRIM37
Multiple congenital anomalies-hypotonia-seizures syndrome 1	PIGN
Multiple sulfatase deficiency	SUMF1
Muscle-eye-brain disease, POMGNT1-related	POMGNT1
Myasthenic syndrome, congenital	PREPL
Myoneurogastrointestinal encephalopathy (MNGIE)	TYMP
Myotubular myopathy, X-linked	MTM1
N-acetylglutamate synthase deficiency	NAGS
Nemaline myopathy, NEB-related	NEB
Niemann-Pick disease, type C1/D	NPC1
Niemann-Pick disease, type C2	NPC2
Niemann-Pick disease, types A/B	SMPD1
Nijmegen breakage syndrome	NBN
Non-syndromic hearing loss (a.k.a. connexin 26)	GJB2
Non-syndromic hearing loss (a.k.a. connexin 30)	GJB6
Oculocutaneous albinism, type 1	TYR
Oculocutaneous albinism, type 3	TYRP1
Oculocutaneous albinism, type 4	SLC45A2
Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome	WNT10A
Omenn syndrome, RAG1-related	RAG1
Omenn syndrome, RAG2-related	RAG2
Ornithine aminotransferase deficiency	OAT
Ornithine transcarbamylase deficiency	OTC
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1
Pantothenate kinase-associated neurodegeneration	PANK2
Papillon-Lefevre syndrome	CTSC
Pendred syndrome	SLC26A4
Peroxisome biogenesis disorder 1A (Zellweger)	PEX1
Peroxisome biogenesis disorder 3A (Zellweger)	PEX12
Peroxisome biogenesis disorder 4A (Zellweger)	PEX6
Peroxisome biogenesis disorder 5A (Zellweger)	PEX2
Peroxisome biogenesis disorder 6A (Zellweger)	PEX10
Persistent Müllerian duct syndrome, type 1	AMH
Persistent Müllerian duct syndrome, type 2	AMHR2
Phenylketonuria	PAH
Phosphoglycerate dehydrogenase deficiency	PHGDH
POLG-related disorders	POLG
Polycystic kidney disease, autosomal recessive	PKHD1
Pontocerebellar hypoplasia	TSEN54
Pontocerebellar hypoplasia, type 1 and 6, RARS2-related	RARS2
Pontocerebellar hypoplasia, type 1A	VRK1
Pontocerebellar hypoplasia, type 1B	EXOSC3
Pontocerebellar hypoplasia, type 2D	SEPSEC5
Pontocerebellar hypoplasia, type 2E	VPS53
Primary congenital glaucoma	CYP1B1
Primary hyperoxaluria, type 2	GRHPR
Primary hyperoxaluria, type 3	HOGA1
Progressive familial intrahepatic cholestasis, type 1	ATP8B1
Progressive familial intrahepatic cholestasis, type 2	ABCB11
Progressive familial intrahepatic cholestasis, type 3	ABCB4
Progressive pseudorheumatoid dysplasia	WISP3
Prolidase deficiency	PEPD
Propionic acidemia, PCCA-related	PCCA
Propionic acidemia, PCCB-related	PCCB
Prothrombin deficiency	F2
Pseudocholinesterase deficiency	BCHE
Pseudoxanthoma elasticum	ABCC6
Pycnodysostosis	CTSK
Pyridoxal 5'-phosphate-dependent epilepsy	PNPO
Pyridoxine-dependent epilepsy	ALDH7A1
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase deficiency, PDHB-related	PDHB
Pyruvate dehydrogenase deficiency, X-linked	PDHA1
Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1
Retinal dystrophies, RLBP1-associated	RLBP1
Retinitis pigmentosa 25	EYS
Retinitis pigmentosa 26	CERKL
Retinitis pigmentosa 28	FAM161A
Retinitis pigmentosa 59	DHDDS
RETT syndrome	MECP2
Rhizomelic chondrodysplasia punctata, type 1	PEX7
Rhizomelic chondrodysplasia punctata, type 3	AGPS
Roberts syndrome	ESCO2
Salla disease	SLC17A5
Sandhoff disease	HEXB
Schimke immunoosseous dysplasia	SMARCAL1
Segawa syndrome, TH-related	TH
Severe combined immunodeficiency, ADA-related	ADA
Severe combined immunodeficiency, type athabaskan	DCLRE1C
Severe combined immunodeficiency, X-linked	IL2RG
Short chain acyl-CoA dehydrogenase deficiency	ACADS
Short/branched chain acyl-CoA dehydrogenase deficiency	ACADSB
Shwachman-Diamond syndrome S	BDS
Sialidosis	NEU1
Sjögren-Larsson syndrome	ALDH3A2
Smith-Lemli-Opitz syndrome	DHCR7
Spastic paraplegia type 15	ZFYVE26
Spinal muscular atrophy	SMN1 SC
Spondylothoracic dysostosis, MESP2-related	MESP2
Stargardt disease, type 1	ABCA4
Steroid-resistant nephrotic syndrome	NPHS2
Stuve-Wiedemann syndrome	LIFR
Tay-Sachs disease	HEXA
Tricho-hepato-enteric syndrome	TTC37
Trifunctional protein deficiency	HADHB
Tyrosinemia, type 3	HPD
Tyrosinemia, type I	FAH
Tyrosinemia, type II	TAT
Usher syndrome, type 1B	MYO7A
Usher syndrome, type 1C	USH1C
Usher syndrome, type 1D	CDH23
Usher syndrome, type 1F	PCDH15
Usher syndrome, type 2A	USH2A
Usher syndrome, type	3 CLRN1
Very long chain acyl-CoA dehydrogenase deficiency	ACADVL
Vitamin D – dependent rickets, type 1A	CYP27B1
Von Willebrand disease	VWF
Walker-Warburg syndrome, FKTN-related	FKTN
Werner syndrome	WRN
Wilson disease	ATP7B
Wiskott-Aldrich syndrome, X-linked	WAS
Wolcott-Rallison syndrome	EIF2AK3
Woolly hair/hypotrichosis syndrome	LIPH
Xeroderma pigmentosum group A	XPA
Xeroderma pigmentosum group C	XPC
Xeroderma pigmentosum group E	DDB2
Xeroderma pigmentosum variant	POLH
Xeroderma pigmentosum, group B	ERCC3
Xeroderma pigmentosum, group D	ERCC2
Xeroderma pigmentosum, group F	ERCC4
Xeroderma pigmentosum, group G	ERCC5