Wittestofziekten genpanel v7
(168 genen)

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
AARS		Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
AARS2		Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCD1		Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ADAR1		Aicardi-Goutieres syndrome 6, 615010
		Dyschromatosis symmetrica hereditaria, 127400
AGPS		Rhizomelic chondrodysplasia punctata, type 3, 600121
AIFM1		Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
AIMP1		Leukodystrophy, hypomyelinating, 3
ALDH3A2		Sjogren-Larsson syndrome, 270200
AMT		Glycine encephalopathy, 605899
APOPT1		Mitochondrial complex IV deficiency, 220110
APP		Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714
		Alzheimer disease 1, familial, 104300
ARSA		Metachromatic leukodystrophy, 250100
ASPA		Canavan disease, 271900
ATAD3A		Harel-Yoon syndrome, 617183
ATAD3B		
ATN1		Dentatorubro-pallidoluysian atrophy, 125370
ATRN		
AUH		3-methylglutaconic aciduria, type I, 250950
BCAP31		Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA		Maple syrup urine disease, type Ia, 248600
BCKDHB		Maple syrup urine disease, type Ib, 248600
BOLA3		Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPIFA2		
CBS		Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CLCN2		Leukoencephalopathy with ataxia, 615651
		{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
		{Epilepsy, juvenile absence, susceptibility to, 2}, 607628
		{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLPP		Perrault syndrome 3, 614129
CNTNAP1		Lethal congenital contracture syndrome 7, 616286
COL4A1		?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2		Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COX6B1		Mitochondrial complex IV deficiency, 220110

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
CSF1R		Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CST3		Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953
CTC1		Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA		Galactosialidosis, 256540
CYP27A1		Cerebrotendinous xanthomatosis, 213700
D2HGDH		D-2-hydroxyglutaric aciduria, 600721
DARS		Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2		Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT		Maple syrup urine disease, type II, 248600
DHAPAT		Rhizomelic chondrodysplasia punctata, type 2, 222765
EARS2		Combined oxidative phosphorylation deficiency 12, 614924
EIF2B1		Leukoencephalopathy with vanishing white matter, 603896
EIF2B2		Leukoencephalopathy with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EIF2B3		Leukoencephalopathy with vanishing white matter, 603896
EIF2B4		Leukoencephalopathy with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EIF2B5		Leukoencephalopathy with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EPRS		
ERCC2		Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3		Trichothiodystrophy 2, photosensitive, 616390
		Xeroderma pigmentosum, group B, 610651
ERCC6		Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC8		Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
FAH		Tyrosinemia, type I, 276700
FAM126A		Leukodystrophy, hypomyelinating, 5, 610532
FBXL4		Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FOLR1		Neurodegeneration due to cerebral folate transport deficiency, 613068
FUCA1		Fucosidosis, 230000
GALC		Krabbe disease, 245200
GAN		Giant axonal neuropathy-1, 256850
GBE1		Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH		Glutaricaciduria, type I, 231670
GCSH		Glycine encephalopathy, 605899
GFAP		Alexander disease, 203450
GJA1		Atrioventricular septal defect 3, 600309
		Craniometaphyseal dysplasia, autosomal recessive, 218400

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
		Erythrokeratodermia variabilis et progressiva, 133200 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB1		Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2		Leukodystrophy, hypomyelinating, 2, 608804
		Lymphedema, hereditary, IC, 613480
		Spastic paraplegia 44, autosomal recessive, 613206
GLA		Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1		GM1-gangliosidosis, type I, 230500
		GM1-gangliosidosis, type II, 230600
		GM1-gangliosidosis, type III, 230650
		Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC		Glycine encephalopathy, 605899
GLRX5		Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GM2A		GM2-gangliosidosis, AB variant, 272750
GSN		Amyloidosis, Finnish type, 105120
GTF2H5		Trichothiodystrophy 3, photosensitive, 616395
HEPACAM		Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEXA		GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB		Sandhoff disease, infantile, juvenile, and adult forms, 268800
HMBS		Porphyria, acute intermittent, 176000
		Porphyria, acute intermittent, nonerythroid variant, 176000
HMGCL		HMG-CoA lyase deficiency, 246450
HSPD1		Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA1		CARASIL syndrome, 600142
		Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
IBA57		?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
IDH1		{Glioma, susceptibility to, somatic}, 137800
IDH2		D-2-hydroxyglutaric aciduria 2, 613657
IKBKAP		Dysautonomia, familial, 223900
ISCA2		Multiple mitochondrial dysfunctions syndrome 4, 616370
ITM2B		?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079
		Dementia, familial British, 176500 Dementia, familial Danish, 117300
KARS		?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
		Deafness, autosomal recessive 89, 613916
L2HGDH		L-2-hydroxyglutaric aciduria, 236792

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
LAMA2		Muscular dystrophy, congenital merosin-deficient, 607855
		Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMB1		Lissencephaly 5, 615191
LIAS		Hyperglycinemia, lactic acidosis, and seizures, 614462
LMBRD1		Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNB1		Leukodystrophy, adult-onset, autosomal dominant, 169500
LYRM7		Mitochondrial complex III deficiency, nuclear type 8, 615838
MLC1		Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMACHC		Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC		Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MOG		?Narcolepsy 7, 614250
MTFMT		Combined oxidative phosphorylation deficiency 15, 614947
MTHFR		Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTR		Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
		{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR		Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
NDUFA2		Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFS1		Mitochondrial complex I deficiency, 252010
NDUFS4		Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7		Leigh syndrome, 256000
NDUFS8		Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1		Mitochondrial complex I deficiency, 252010
NFU1		Multiple mitochondrial dysfunctions syndrome 1, 605711
NKX6-2		
NOTCH3		?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720
NUBPL		Mitochondrial complex I deficiency, 252010
PAHX		Refsum disease, 266500
PCCA		Propionicacidemia, 606054
PCCB		Propionicacidemia, 606054
PEX1		Heimler syndrome 1, 234580
		Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10		Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12		Peroxisome biogenesis disorder 3A (Zellweger), 614859
		Peroxisome biogenesis disorder 3B, 266510
PEX13		Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14		Peroxisome biogenesis disorder 13A (Zellweger), 614887

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
PEX16		Peroxisome biogenesis disorder 8A (Zellweger), 614876
		Peroxisome biogenesis disorder 8B, 614877
PEX19		Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2		Peroxisome biogenesis disorder 5A (Zellweger), 614866
		Peroxisome biogenesis disorder 5B, 614867
PEX26		Peroxisome biogenesis disorder 7A (Zellweger), 614872
		Peroxisome biogenesis disorder 7B, 614873
PEX3		?Peroxisome biogenesis disorder 10B, 617370
		Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5		Peroxisome biogenesis disorder 2A (Zellweger), 214110
		Peroxisome biogenesis disorder 2B, 202370
		Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6		Heimler syndrome 2, 616617
		Peroxisome biogenesis disorder 4A (Zellweger), 614862
		Peroxisome biogenesis disorder 4B, 614863
PEX7		Peroxisome biogenesis disorder 9B, 614879
		Rhizomelic chondrodysplasia punctata, type 1, 215100
PHGDH		Neu-Laxova syndrome 1, 256520
		Phosphoglycerate dehydrogenase deficiency, 601815
PHYH		Refsum disease, 266500
PLP1		Pelizaeus-Merzbacher disease, 312080
		Spastic paraplegia 2, X-linked, 312920
POLR1C		Leukodystrophy, hypomyelinating, 11, 616494
		Treacher Collins syndrome 3, 248390
POLR3A		Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B		Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POLR3D		
PSAP		Combined SAP deficiency, 611721
		Gaucher disease, atypical, 610539
		Krabbe disease, atypical, 611722
		Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1		?Phosphoserine aminotransferase deficiency, 610992
		Neu-Laxova syndrome 2, 616038
RARS		Leukodystrophy, hypomyelinating, 9, 616140
RARS2		Pontocerebellar hypoplasia, type 6, 611523
RMND1		Combined oxidative phosphorylation deficiency 11, 614922
RNASEH2A		Aicardi-Goutieres syndrome 4, 610333
RNASEH2B		Aicardi-Goutieres syndrome 2, 610181
RNASEH2C		Aicardi-Goutieres syndrome 3, 610329
RNASET2		Leukoencephalopathy, cystic, without megalencephaly, 612951
RPIA		?Ribose 5-phosphate isomerase deficiency, 608611
SAMHD1		?Chilblain lupus 2, 614415
		Aicardi-Goutieres syndrome 5, 612952
SDHA		Cardiomyopathy, dilated, 1GG, 613642
		Leigh syndrome, 256000
		Mitochondrial respiratory chain complex II deficiency, 252011
		Paragangliomas 5, 614165
SDHAF1		Mitochondrial complex II deficiency, 252011
SDHB		Cowden syndrome 2, 612359
		Gastrointestinal stromal tumor, 606764
		Paraganglioma and gastric stromal sarcoma, 606864

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		Paragangliomas 4, 115310 Pheochromocytoma, 171300
SLC16A2		Allan-Herndon-Dudley syndrome, 300523
SLC17A5		Salla disease, 604369
		Sialic acid storage disorder, infantile, 269920
SLC19A3		Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A4		Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1		Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12		Epileptic encephalopathy, early infantile, 39, 612949
SNORD118		Leukoencephalopathy, brain calcifications, and cysts, 614561
SOX10		PCWH syndrome, 609136
		Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPTAN1		Epileptic encephalopathy, early infantile, 5, 613477
SUMF1		Multiple sulfatase deficiency, 272200
SURF1		Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
TMEM106B		
TREM2		Nasu-Hakola disease, 221770
TREX1		Aicardi-Goutieres syndrome 1, dominant and recessive, 225750
		Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315
		{Systemic lupus erythematosus, susceptibility to}, 152700
TTR		Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUBB4A		Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TYMP		Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYROBP		Nasu-Hakola disease, 221770
UFM1		
VPS11		Leukodystrophy, hypomyelinating, 12, 616683

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Genes in bold are core genes
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
"No OMIM phenotype" signifies a gene without a current OMIM association
OMIM phenotype descriptions between {} signify risk factors, ? is unconfirmed data