Hospital Sant Pau 2020 − Servei de Genètica
Malaltia de vas petit + Cavernomatosis (n=64 + n=3)

Gen	Chr	OMIM
ABCD1	chrX	Adrenomyeloneuropathy, adult, 300100, X−linked recessive Adrenoleukodystrophy, 300100, X−linked recessive
AIMP1	chr4	Leukodystrophy, hypomyelinating, 3, 260600, AR
ALDH3A2	chr17	Sjogren−Larsson syndrome, 270200, AR
APP	chr21	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714, AD Alzheimer disease 1, familial, 104300, AD
ARSA	chr22	Metachromatic leukodystrophy, 250100, AR
ASPA	chr17	Canavan disease, 271900, AR
CCM2	chr7	Cerebral cavernous malformations−2, 603284, AD
COL4A1	chr13	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773, AD Brain small vessel disease with or without ocular anomalies, 175780, AD {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000, AD Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564, AD
COL4A2	chr13	Brain small vessel disease 2, 614483, AD {Hemorrhage, intracerebral, susceptibility to}, 614519
CSF1R	chr5	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476, AR Leukoencephalopathy, diffuse hereditary, with spheroids, 221820, AD
CST3	chr20	Cerebral amyloid angiopathy, 105150, AD {Macular degeneration, age−related, 11}, 611953
CTC1	chr17	Cerebroretinal microangiopathy with calcifications and cysts, 612199, AR
CTSA	chr20	Galactosialidosis, 256540, AR
CYP27A1	chr2	Cerebrotendinous xanthomatosis, 213700, AR
DARS2	chr1	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105, AR
EGR2	chr10	Dejerine−Sottas disease, 145900, AR, AD Hypomyelinating neuropathy, congenital, 1, 605253, AR, AD Charcot−Marie−Tooth disease, type 1D, 607678, AD
EIF2B1	chr12	Leukoencephalopathy with vanishing white matter, 603896, AR
EIF2B2	chr14	Ovarioleukodystrophy, 603896, AR Leukoencephalopathy with vanishing white matter, 603896, AR
EIF2B3	chr1	Leukoencephalopathy with vanishing white matter, 603896, AR
EIF2B4	chr2	Leukoencephalopathy with vanishing white matter, 603896, AR Ovarioleukodystrophy, 603896, AR
EIF2B5	chr3	Leukoencephalopathy with vanishing white matter, 603896, AR Ovarioleukodystrophy, 603896, AR
FAM126A	chr7	Leukodystrophy, hypomyelinating, 5, 610532, AR
FMR1	chrX	Premature ovarian failure 1, 311360, X−linked Fragile X tremor/ataxia syndrome, 300623, X−linked dominant Fragile X syndrome, 300624, X−linked dominant
GALC	chr14	Krabbe disease, 245200, AR
GBE1	chr3	Polyglucosan body disease, adult form, 263570, AR Glycogen storage disease IV, 232500, AR
GFAP	chr17	Alexander disease, 203450, AD
GJC2	chr1	Spastic paraplegia 44, autosomal recessive, 613206, AR Lymphatic malformation 3, 613480, AD Leukodystrophy, hypomyelinating, 2, 608804, AR
GLA	chrX	Fabry disease, 301500, X−linked Fabry disease, cardiac variant, 301500, X−linked
GSN	chr9	Amyloidosis, Finnish type, 105120, AD
HEPACAM	chr11	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925, AR Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926, AD

HSPD1	chr2	Spastic paraplegia 13, autosomal dominant, 605280, AD Leukodystrophy, hypomyelinating, 4, 612233, AR
HTRA1	chr10	{Macular degeneration, age−related, neovascular type}, 610149 {Macular degeneration, age−related, 7}, 610149 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779, AD CARASIL syndrome, 600142, AR
ITM2B	chr13	Dementia, familial British, 176500, AD ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079, AD Dementia, familial Danish, 117300, AD
KRIT1	chr7	Cavernous malformations of CNS and retina, 116860, AD Cerebral cavernous malformations−1, 116860, AD Hyperkeratotic cutaneous capillary−venous malformations associated with cerebral capillary malformations, 116860, AD
LAMB1	chr7	Lissencephaly 5, 615191, AR
LMNB1	chr5	Leukodystrophy, adult−onset, autosomal dominant, 169500, AD
MLC1	chr22	Megalencephalic leukoencephalopathy with subcortical cysts, 604004, AR
MPZ	chr1	Charcot−Marie−Tooth disease, type 2J, 607736, AD Charcot−Marie−Tooth disease, type 1B, 118200, AD Dejerine−Sottas disease, 145900, AR, AD Hypomyelinating neuropathy, congenital, 2, 618184, AD Charcot−Marie−Tooth disease, dominant intermediate D, 607791, AD Roussy−Levy syndrome, 180800, AD Charcot−Marie−Tooth disease, type 2I, 607677, AD
MTHFR	chr1	{Vascular disease, susceptibility to} {Schizophrenia, susceptibility to}, 181500, AD Homocystinuria due to MTHFR deficiency, 236250, AR {Neural tube defects, susceptibility to}, 601634, AR {Thromboembolism, susceptibility to}, 188050, AD
NOTCH3	chr19	?Myofibromatosis, infantile 2, 615293, AD Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310, AD Lateral meningocele syndrome, 130720, AD
PDCD10	chr3	Cerebral cavernous malformations 3, 603285
PLP1	chrX	Pelizaeus−Merzbacher disease, 312080, X−linked recessive Spastic paraplegia 2, X−linked, 312920, X−linked recessive
POLG	chr15	Progressive external ophthalmoplegia, autosomal dominant 1, 157640, AD Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662, AR Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459, AR Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700, AR Progressive external ophthalmoplegia, autosomal recessive 1, 258450, AR
POLR3A	chr10	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694, AR Wiedemann−Rautenstrauch syndrome, 264090, AR
POLR3B	chr12	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381, AR
PROC	chr2	Thrombophilia due to protein C deficiency, autosomal dominant, 176860, AD Thrombophilia due to protein C deficiency, autosomal recessive, 612304, AR
PROS1	chr3	Thrombophilia due to protein S deficiency, autosomal recessive, 614514, AR Thrombophilia due to protein S deficiency, autosomal dominant, 612336, AD
PSAP	chr10	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722, AR Combined SAP deficiency, 611721, AR Metachromatic leukodystrophy due to SAP−b deficiency, 249900, AR
PSEN1	chr14	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822, AD Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822, AD ?Acne inversa, familial, 3, 613737, AD Alzheimer disease, type 3, 607822, AD Dementia, frontotemporal, 600274, AD Pick disease, 172700, AD Cardiomyopathy, dilated, 1U, 613694, AD
PSEN2	chr1	Alzheimer disease−4, 606889, AD Cardiomyopathy, dilated, 1V, 613697, AD
RNASEH2A	chr19	Aicardi−Goutieres syndrome 4, 610333, AR

RNASEH2B	chr13	Aicardi−Goutieres syndrome 2, 610181, AR
RNASEH2C	chr11	Aicardi−Goutieres syndrome 3, 610329, AR
RNASET2	chr6	Leukoencephalopathy, cystic, without megalencephaly, 612951, AR
RPIA	chr2	Ribose 5−phosphate isomerase deficiency, 608611, AR
SAMHD1	chr20	?Chilblain lupus 2, 614415, AD Aicardi−Goutieres syndrome 5, 612952, AR
SCP2	chr1	?Leukoencephalopathy with dystonia and motor neuropathy, 613724, AR
SDHAF1	chr19	Mitochondrial complex II deficiency, 252011, AR
SLC16A2	chrX	Allan−Herndon−Dudley syndrome, 300523, X−linked
SLC17A5	chr6	Sialic acid storage disorder, infantile, 269920, AR Salla disease, 604369, AR
SOX10	chr22	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584, AD PCWH syndrome, 609136, AD Waardenburg syndrome, type 4C, 613266, AD
SUOX	chr12	Sulfite oxidase deficiency, 272300, AR
TREX1	chr3	{Systemic lupus erythematosus, susceptibility to}, 152700, AD Vasculopathy, retinal, with cerebral leukodystrophy, 192315, AD Aicardi−Goutieres syndrome 1, dominant and recessive, 225750, AR, AD Chilblain lupus, 610448, AD
TTR	chr18	Amyloidosis, hereditary, transthyretin−related, 105210, AD [Dystransthyretinemic hyperthyroxinemia], 145680, AD Carpal tunnel syndrome, familial, 115430, AD
TUBB4A	chr19	Leukodystrophy, hypomyelinating, 6, 612438, AD Dystonia 4, torsion, autosomal dominant, 128101, AD
TYMP	chr22	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041, AR
WRN	chr8	Werner syndrome, 277700, AR