GENE	PHENOTYPE	PHENOTYPE MIM NUMBER	INHERITANCE	CPT CODE
AARS1	Charcot-Marie-Tooth Neuropathy, axonal, type 2N (CMT2N)	613287	AD	81479
AARS2	Leukoencephalopathy, progressive, with ovarian failure (LKENP)	615889	AR	81479
DARS1	Hypomyelination with brainstem and spinal cord involvement and leg spasticity (HBSL)	615281	AR	81479
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation (LBSL)	611105	AR	81479
EARS2	Combined oxidative phosphorylation deficiency, 12 (COXPD12)	614924	AR	81479
FAM126A	Leukodystrophy, hypomyelinating, 5 (HLD5)	610532	AR	81479
GJC2	Leukodystrophy, hypomyelinating, 2 (HLD2)	608804	AR	81479
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A (MLC2A)	613925	AR	81479
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation (MLC2B)	613926	AD	81479
LMNB1	Leukodystrophy, adult-onset (ADLD)	169500	AD	81479
MARS2	Combined oxidative phosphorylation deficiency, 25 (COXPD25)	616430	AR	81479
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts (MLC1)	604004	AR	81479
PLP1	Pelizaeus-Merzbacher disease (PMD)	312080	XL	81405
PLP1	Spastic paraplegia 2 (SPG2)	312920	XL	81405
POLR1C	Leukodystrophy, hypomyelinating, 11 (HLD11)	616494	AR	81479
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism (HLD7)	607694	AR	81479
POLR3B	Leukodystrophy, hypomyelinating, 8 with or without oligodontia and/or hypogonadotropic hypogonadism (HLD8)	614381	AR	81479
RARS1	Leukodystrophy, hypomyelinating, 9 (HLD9)	616140	AR	81479
SLC16A2	Allen-Herndon-Dudley Syndrome (AHDS)	300523	XL	81405
TUBB4A	Leukodystrophy, hypomyelinating, 6 (HLD6)	612438	AD	81479

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