

PANEL NMG6
LEUCODISTROFIAS

ABCD1	ADRENOLEUCODISTROFIA LIGADA AL CROMOSOMA X, FORMA CEREBRAL/ ADRENOMIELONEUROPATÍA
ARSA	LEUCODISTROFIA METACROMÁTICA
GALC	ENFERMEDAD DE KRABBE
PSAP	ENFERMEDAD DE KRABBE Y LEUCODISTROFIA METACROMÁTICA ATÍPICA
ASPA	CANAVAN DISEASE
GFAP	ALEXANDER DISEASE
MLC1	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS
HEPACAM	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 2A
RNASET2	LEUKOENCEPHALOPATHY, CYSTIC, WITHOUT MEGALENCEPHALY
EIF2B1	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
EIF2B2	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
EIF2B3	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
EIF2B4	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
EIF2B5	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER
PLP1	LEUKODYSTROPHY, HYPOMYELINATING, 1: PELIZAEUS-MERZBACHER DISEASE
GJC2	LEUKODYSTROPHY, HYPOMYELINATING, 2
AIMP1	LEUKODYSTROPHY, HYPOMYELINATING, 3
HSPD1	LEUKODYSTROPHY, HYPOMYELINATING, 4
FAM126A	LEUKODYSTROPHY, HYPOMYELINATING, 5
LMNB1	LEUKODYSTROPHY, ADULT-ONSET, AUTOSOMAL DOMINANT
POLR3A	LEUKODYSTROPHY, HYPOMYELINATING, 7, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM
POLR3B	LEUKODYSTROPHY, HYPOMYELINATING, 8, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM
TREX1	AICARDI-GOUTIERES SYNDROME 1, DOMINANT AND RECESSIVE
RNASEH2B	AICARDI-GOUTIERES SYNDROME 2
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3
RNASEH2A	AICARDI-GOUTIERES SYNDROME 4
SAMHD1	AICARDI-GOUTIERES SYNDROME 5
ADAR1	AICARDI-GOUTIERES SYNDROME 6
C19ORF12	NEURODEGENERACIÓN CON ACUMULACIÓN CEREBRAL DE HIERRO POR MUTACIÓN EN C19ORF12
PANK2	NEURODEGENERACIÓN CON ACUMULACIÓN DE HIERRO EN EL CEREBRO TIPO 1, FORMA CLÁSICA
PLA2G6	DISTROFIA NEUROAXONAL INFANTIL: NEURODEGENERACIÓN CON ACUMULACIÓN DE HIERRO EN EL CEREBRO TIPO 2B
FTL	NEURODEGENERACIÓN CON ACUMULACIÓN DE HIERRO EN EL CEREBRO TIPO 3
CP	ACERULOPLASMINEMIA
COASY	NEURODEGENERACION CON ACUMULO CEREBRAL DE HIERRO 6 NBIA6
SOX10	SÍNDROME DE WAARDENBURG-SHAH, VARIANTE NEUROLÓGICA/
CTC1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS; CRMCC
FA2H	DISTROFIA NEUROAXONAL INFANTIL
DARS	HYPOMYELINATION WITH BRAIN STEM AND SPINAL CORD INVOLVEMENT AND LEG SPASTICITY.
CLCN2	LEUCOENCEFALOPATIA CON ATAXIA