WEB TABLE I CLINICAL FEATURES AND AGE OF PRESENTATION OF LYSOSOMAL STORAGE DISORDERS IN CHILDREN WITH NEUROREGRESSION (N=309)

Name of disorder	*No. (%)*	*Most common clinical signs*	*Age of presentation*
Defects in degradation of glycolipids	155 (50.2)		
GM1 gangliosidosis	43 (13.9)	Cherry red spot, coarse facial features, mongolian spots, developmental delay, mild to moderate hepatomegaly/ hepatosplenomegaly	7 m-3.5 y
GM2 gangliosidosis	40 (12.94)	Cherry red spot, Convulsions, myoclonic jerks	10 m- 3.3 y
Tay-Sachs disease			
Sandhoff disease	31 (10.03)	Cherry red spot, Convulsions, myoclonic jerks, abdominal distension	9 m- 2.5 y
Niemann-pick disease A/B(NPD-A/B)	35 (11.3)	Cherry red spot, developmental delay, moderate to severe hepatomegaly/ hepatosplenomegaly	5 m- 9 y
Late Juvenile form of NPD-A/B	2 (0.64)	Mild to moderate hepatomegaly/ hepatosplenomegaly	11 y- 13 y
Gaucher disease	4 (1.3)	Severe hepatosplenomegaly, developmental delay, thrombocytopenia	4.5 m- 2.5 y
Defects in mucopolysaccharide (MPS) degradation	67 (21.7)		
Hurler disease (MPS-I)	20 (6.5)	Coarse facial features, corneal clouding, skeletal abnormality	9 m- 9 y
Hunter disease (MPS-II)	19 (6.1)	Coarse facial features, skeletal	6 m- 6 y
Sanfilippo disease A & B(MPS-IIIA & IIIB)	23 (7.5)	abnormality Coarse features, behavioral abnormality, hypertrichosis	1 y- 10 y
Late Juvenile to Adult onset form of MPS-IIIA & IIIB	4 (1.3)	Coarse features, behavioral abnormality	11 y- 17 y
Morateaux-Lamy disease(MPS-VI)	1 (0.32)	Skeletal dysplasia, mild cerebral atrophic changes	6.5 m
Defects in sulphatides degradation	54 (17.5)		
Metachromatic Leukodystrophy (MLD)	34 (11)	Leukodystrophy, convulsions, spasticity	7 m-10 y
Late Juvenile form	3	Hypotonia, leukodystrophy	15 y – 18 y
Krabbe disease	15 (4.8)	Leukodystrophy, convulsions, spasticity, hypotonia	4 m- 3 y
Late Juvenile form	2 (0.6)	Spasticity, hypotinia, leukodystrophy	13 y-15 y
Defects in protein degradation	23 (7.44)		
Neuronal ceroid lipofuscinoses-1 (NCL1)	7 (2.26)	Cerebral & cerebellar atrophy, seizures, visual impairment	3 m- 2 y
Neuronal ceroid lipofuscinoses-2 (NCL2)	13 (4.2)	Cerebral and cerebellar atrophy, seizures, myoclonic jerks	1.1 y- 8 y
Late Juvenile to Adult onset form	3 (0.97)	Cerebral and cerebellar atrophy, seizures, myoclonic jerks	15 y- 20 y
Defects in lysosomal transporters	6 (1.94)		
Mucolipidosis-II/III(ML II/III)	3 (0.97)	Multiple skeletal abnormalities, coarse face	2 y-5 y
Sialic acid storage disorder	3 (0.97)	Coarse features, spasticity	1.5 y- 9 y
Defects in lysosomal trafficking proteins	3 (0.97)		
Niemann-pick disease type-C (NPC)	3 (0.97)	Vertical supranuclear gaze palsy, hepatomegaly/ hepatosplenomegaly	1.9 y – 3 y
Defects in glycoprotein degradation	1 (0.32)		
Fucosidosis	1 (0.32)	Developmental delay, Dysmorphic features, Intellectual disability	13 y