ADRENOLEUKODYSTROPHIES:

Disease	Inheritance	Enzyme	Clinical Picture	MRI
Adrenoleukodystrophy "Peroxisomal"	XL-R ABCD gene	Failure of peroxisomal oxidation of VLCFA	3 Phenotypes: - Childhood cerebral adrenoleukodystrophy - Adrenomyeloneuropathy - Addison's disease	- **Posterior** - Spares U fibers - 3 zones of different intensities in T2 (inner hyper, middle Iso and outer hypo)
Zellwiger (cerebrohepatorenal syndrome) "Peroxisomal"	AR PEX1 gene	Absence of peroxisomes	- Facies: high forehead, midface hypoplasia - Hepatomegally – Renal cysts	- **Diffuse hypomyelination involving U fibers** - Gyral abnormalities (frontal microgyria , occipital pachygyriia)
Metachromatic "Lysosomal"	AR ARSA gene	Arylsulfatase	- Late infantile: gait abnormality, muscle rigidity, loss of vision, developmental delay - Juvenile: slower course - Adult: dementia – psychiatric features	- Periventricular **- Spares U fibers leading to a "butterfly pattern"**
Krabbe (Globoid cell) "Lysosomal"	AR GALC gene	Galactocerebrosidase	Peripheral neuropathy, developmental delay, optic atrophy, globoid cells	**CT: Hyperdense thalami** MRI: Periventricular
Fabry "Lysosomal"	XL-R	Galactosidase	Posterior circulation strokes - Peripheral neuropathy of hands/feet, angiokeratomas, cardiovascular disease	**Pulvinar hyperintensity**
Canavan (spongiform degeneration of white matter) "Amino-acid"		Aspartocyclase	Macrocephaly, severe mental deficits and blindness.	**Macrocephaly** **Diffuse hypomyelination involving U fibers** **Increased NAA in MRS**
Alexander "Fibrinoid"	GFAP gene		Macrocephaly, severe mental deficits Rosenthal fibers in pathology	**Macrocephaly** **Frontal predominance**