CASE DEFINITION FOR PAEDIATRIC-ONSET LEUKODYSTROPHIES

Report all children and youth less than 18 years of age (up to their 18th birthday) with a new diagnosis of a leukodystrophy defined as a genetically* determined disorder characterized by primary involvement of the white matter. Disorders characterized as leukodystrophies include, but are not limited to, the following disorders (non-exhaustive list):

- Pol-III related disorders (4H syndrome (hypomyelination, hypodontia, and hypogonadotropic hypogonadism))
- 18q minus syndrome
- X-linked adrenoleukodystrophy (X-ALD)
- Adult-onset leukodystrophy with neuroaxonal spheroids and pigmented glia (including hereditary diffuse leukoencephalopathy with spheroids, HDLS, and pigmentary type of orthochromat leukodystrophy with pigmented glia, POLD)
- Aicardi-Goutières syndrome (AGS)
- Alexander disease (AxD)
- Autosomal dominant leukodystrophy with autonomic disease (ADLD)
- Canavan disease
- Cerebrotendinous xanthomatosis (CTX)
- Chloride ion channel 2 (CIC-2) related leukoencephalopathy with intramyelinic oedema
- eIF2B related disorder (vanishing white matter disease or childhood ataxia with central nervous system hypomyelination (CACH))
- Fucosidosis
- Globoid cell leukodystrophy (Krabbe disease)
- Hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC)
- Hypomyelination with brainstem and spinal cord involvement and leg spasticity (HBSL)
- Hypomyelination with congenital cataract (HCC)
- Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL)
- Leukoencephalopathy with thalamus and brainstem involvement and high lactate (LTBL)
- Megalencephalic leukoencephalopathy with subcortical cysts (MLC)
- Metachromatrophic leukodystrophy (MLD) and its biochemical variants
- Oculodentodigital dysplasia
- Pelizaeus Merzbacher disease (PMD)
- Pelizaeus Merzbacher like-disease (PMLD)
- Peroxisomal biogenesis disorders (including Zellweger, neonatal adrenoleukodystrophy and infantile refsum)
- Polyglucosan body fisease (PGBD)
- RNAse T2 deficient leukoencephalopathy
- Sialic acid storage disorders (Salla disease, infantile sialic acid storage disease and intermediate form)
- Single enzyme deficiencies of peroxisomal fatty acid beta oxidation (including only D-bifunctional protein deficiency; sterol carrier protein X (SCPx) deficiency; peroxisomal acyl-CoA-oxidase deficiency)
- Sjögren-Larsson syndrome
- SOX10-associated PCWH: peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease

* For information on the availability of and access to genetic testing in your region, please refer to the list of study principal investigators/co-investigators at the beginning of the study protocol and contact the one who is located closest to your practice.