



Need to talk? Call us **0845 2412173** Monday to Friday
9am to 5pm



Supporting those affected by
Inherited Metabolic Disorders

Pelizaeus Merzbacher Disease

This disorder affects the central nervous system due to abnormalities of the brain's white matter. There is a lack of fatty coverings (myelin sheaths) that cover the nerve fibres in the brain. It is caused by an alteration of the gene that controls the production of proteolipid protein, which is the main protein present within myelin in the brain. There is considerable confusion about the use of the term Pelizaeus Merzbacher disease in the literature. It is currently deemed desirable to reserve the designation Pelizaeus-Merzbacher disease for the disorder due to abnormality of proteolipid protein, a disorder inherited as an X-linked recessive trait. Classically this presents in late infancy though some cases have presented in the neonatal period, or in early infancy, and hence been termed the connatal form. There is a separate autosomal recessive disorder that can mimic the connatal form, though the majority of connatal cases are X-linked. Similarly an autosomal recessive disorder has been described with mutations in the GJA12 gene that is said to be "Pelizaeus-Merzbacher-like". An adult-onset disorder that resembles multiple sclerosis and follows autosomal dominant inheritance has also been called "late-onset Pelizaeus Merzbacher disease" but is clearly a separate entity due to mutations in an entirely different gene.

Synonyms

Alternative names for this condition are:

- PMD
- Pelizaeus Merzbacher Brain Sclerosis
- Sclerosis, Diffuse Familial Brain
- Sudanophilic Leukodystrophy, Pelizaeus Merzbacher Type

Further information about this condition is available from Climb.



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