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Pelizaeus-Merzbacher Disease

National Cancer Institute

Source

National Cancer Institute. *Pelizaeus-Merzbacher Disease*. NCI Thesaurus. Code C75487.

An X-linked inherited disorder caused by mutations in the PLP1 gene on chromosome X. The signs and symptoms are the result of defective myelination of the central nervous system and include nystagmus, hypotonia, tremor, ataxia, spastic quadriparesis, and diffuse leukoencephalopathy.