Pelizaeus-Merzbacher disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pelizaeus-Merzbacher disease. ORPHA:702

Pelizaeus-Merzbacher disease (PMD) is an X-linked leukodystrophy characterized by developmental delay, nystagmus, hypotonia, spasticity, and variable intellectual deficit. It is classified into three sub-forms based on the age of onset and severity: connatal, transitional, and classic PMD (see these terms).