

Pelizaeus-Merzbacher disease (PMD) is a rare X-linked genetic disorder affecting the central nervous system that is associated with abnormalities of the white matter of the brain and spinal cord. It is one of the leukodystrophies in which disease is due to abnormal development of one or more components (predominantly fats or proteins) that make up the white matter (myelin sheath) of the brain. The myelin sheath is the protective covering of the nerve and nerves cannot function normally without it. In PMD, many areas of the central nervous system may be affected, including the deep portions of the cerebrum (subcortical), cerebellum, brain stem and spinal cord. Signs may include the impaired ability to coordinate movement (ataxia), involuntary muscle spasms (spasticity) that result in slow, stiff movements of the legs, delays in reaching developmental milestones, late onset loss of motor abilities, and progressive deterioration of intellectual function. The neurologic signs of PMD are usually slowly progressive.