Pelizaeus-Merzbacher disease

Leukodystrophies are a group of disorders that involve the destruction of myelin. Myelin is a fatty covering that is used by the body to insulate nerve fibres and helps to promote rapid transmission of nerve impulses through the nervous system. If there is a problem with the myelin, as is the case in leukodystrophies, nerve impulses can be disrupted, leading to an impairment of the nervous system.

Diagnosis

Diagnosis will depend on the type of leukodystrophy, and often will need a number of specialists to be involved, including neurologists, and geneticists. The diagnostic methods vary between types, some of these include:

- Physical examination
- Blood testing
- Urine testing
- Genetic testing
- MRI
- Nerve conduction testing

Signs and Symptoms

- Vary by disease (see below)

Treatment

No specific therapy is currently available to treat these disorders. Management is generally supportive and includes a multitude of medical participants including physicians, occupational therapists, physiotherapists and many more.

A type specific to section 24 of the NDIS Act

Pelizaeus-Merzbacher disease is an inherited condition involving the nervous system. It is divided into two forms classic and connatal (present from birth).

Pelizaeus-Merzbacher disease is caused by a mutation in the PLP1 gene which is responsible for making proteolipid protein 1 and another protein called DM20.

Both these proteins are found within the cell membrane of the nerve cells, and both these proteins are found within the cell membrane of the nerve cells, and make up the majority of myelin.

In Pelizaeus-Merzbacher disease the accumulation of excess protein leads to swelling and breakdown of nerve fibres.

Frequency

The prevalence of Pelizaeus-Merzbacher disease is approximately 1 case in 200,000 to 500,000. The condition affects mainly males, and rarely females.

Signs and symptoms

- Difficulty with feeding
- Poor weight gain
- Developmental delay
- High-pitched breathing (stridor)
- Nystagmus
- Progressive speech difficulty
- Problems with balance (ataxia)
- Weak muscles (hypotonia)
- Seizures
- Contractures that affect movement (people with Pelizaeus-Merzbacher disease connatal form are never able to walk)

References

Pelizaeus-Merzbacher Disease (2019). Retrieved from https://ghr.nlm.nih.gov/condition/pelizaeus-merzbacher-disease#statistics

Pelizaeus-Merzbacher Disease (2019). Retrieved from https://rarediseases.info.nih.gov/diseases/4265/pelizaeus-merzbacher-disease