

Genetic investigation of Leukodystrophy in Iran

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Leukodystrophy is group of disorders characterized by degeneration of the white matter in the brain. The leukodystrophies are caused by imperfect growth or development of the myelin sheath, the fatty covering that acts as an insulator around nerve fibers. Myelin in the CNS is produced by oligodendrocytes. When damage occurs to white matter tissue immune responses can lead to inflammation in the CNS, along with loss of myelin.

Here we are going to discuss about different type of leukodystrophy such as:

• **Adrenoleukodystrophy**

ALD is caused by mutations in ABCD1, located at Xq28 and demonstrates X-linked recessive inheritance. The gene ABCD1 encodes a peroxisomal membrane transporter which is responsible for transporting very long chain fatty acid substrate into the peroxisomes for degradation. Mutations in this gene that interfere with this process cause this syndrome.

• **Metachromatic leukodystrophy**

MLD is directly caused by a deficiency of the enzyme arylsulfatase A (ARSA) and is characterized by enzyme activity in leukocytes that is less than 10% of normal controls. However, assay of the ARSA enzyme activity alone is not sufficient for diagnosis; MLD has an autosomal recessive inheritance pattern. The inheritance probabilities per birth are as follows: If both parents are carriers: 25% (1 in 4) children will have the disease, 50% (2 in 4) children will be carriers, but unaffected, 25% (1 in 4) children will be free of MLD - unaffected child that is not a carrier

• **Hereditary CNS demyelinating disease**

- Krabbe disease (also known as globoid cell leukodystrophy or galactosylceramidolipidosis) is a rare, often fatal degenerative disorder that affects the myelin sheath of the nervous system. It is a form of sphingolipidosis, as it involves dysfunctional metabolism of sphingolipids. This condition is inherited in an autosomal recessive pattern.
- Pelizaeus-Merzbacher disease (PMD) is a rare central nervous system disorder in which coordination, motor abilities, and intellectual function are delayed to variable extents.
- Canavan disease also called Canavan-Van Bogaert-Bertrand disease, aspartoacylase deficiency or aminoacylase 2 deficiency, is an autosomal recessive degenerative disorder that causes progressive damage to nerve cells in the brain. Canavan disease is one of the most common degenerative cerebral diseases of infancy.

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- Leukoencephalopathy with vanishing white matter (VWM disease) is an autosomal recessive neurological disease. The cause of the disease are mutations in any of the 5 genes encoding subunits of the translation initiation factor EIF 2B: EIF2B1, EIF2B2, EIF2B3, EIF2B4, or EIF2B5
- Alexander disease also known as fibrinoid leukodystrophy, is a slowly progressing and fatal neurodegenerative disease. It is a very rare disorder which results from a genetic mutation and mostly affects infants and children, causing developmental delay and changes in physical characteristics
- Refsum disease is an autosomal recessive neurological disease that results from the over-accumulation of phytanic acid in cells and tissues.

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