Description
Alexander disease (also called fibrinoid leukodystrophy) is a rare but fatal neurodegenerative disorder that affects mainly infants and children. This disease is caused by defects in the gene that encode GFAP (Glial Fibrillary Acidic Protein), which is a filament protein involved in structural development of cells in the central nervous system. Alexander disease belong to a group of neurodegenerative disorders called leukodystrophies which is characterized by abnormal growth and development of myelin sheath (an insulating layer) that protects nerve fibres in the brain. The clinical hallmark of Alexander disease is abnormal production of Rosenthal fibres which are thick clumps of protein that accumulate in specific cell types in the brain called astrocytes. The National Institute of Health (NIH) has classified Alexander disease as a “rare disease” because only about 500 cases have ever been reported since the disorder was first described in 1949.

Alexander disease is divided into three forms (infantile, juvenile and adult onset). The age of onset for the infantile form is 0–2 years and is the most aggressive form of Alexander disease. It is characterized by an abnormally large brain (megalencephaly) and/or buildup of fluid in the brain (hydrocephaly). Individuals with this form of the disease also suffer from seizures, ataxia, vomiting, progressive psychomotor retardation, failure to thrive and a loss of acquired milestones. Such individuals do not survive beyond childhood. The juvenile form of Alexander disease has an onset age of 2-14 years and the affected children can survive from the early teens to the 20s-30s. This form of the disease is associated with breathing difficulties, intellectual decline, seizures, weakness in lower extremities and a decline in motor function. For the adult onset form of Alexander disease, the age of onset is generally above 12 years of age. This form of the disease develops much slower and presents with the following symptoms: double vision, slurred speech, sleep apnea, difficulties in walking, difficulties in swallowing, muscle spasms and weakness in one side of the body.

How is Alexander’s disease diagnosed?
Diagnosis for Alexander disease is based on genetic testing of an individual’s blood sample. Neuroimaging studies such as Magnetic Resonance Imaging (MRI) and Electroencephalograms (EEG) are also performed. Genetic tests are also recommended for pregnant women with a family history of Alexander disease.

Treatment
There is no cure for Alexander disease, nor is there a standard course of treatment. Current treatments for Alexander disease are symptomatic and supportive; e.g. attention to care and nutritional needs, medications for control of seizures, physical and occupational therapy when required and helping individuals cope with learning disabilities and cognitive impairments.
Prognosis
The prognosis is poor for individuals suffering from Alexander disease. In most cases, children with the infantile form of Alexander disease do not live beyond the age of six. For those with the Juvenile form of the disease, death generally occurs within 10 years following the onset of symptoms.

References


Further Information and support: