Alexander Disease

1. What is Alexander Disease?

Alexander disease is a rare genetic disorder that primarily affects the nervous system. It is a type of **leukodystrophy** (white matter degenerative disease), which means that the affected individual is unable to maintain myelin - the white, fatty tissue layer that protects the neurons of the brain and spinal cord and speeds up electrical messages between cells.

2. What happens to people with Alexander Disease?

Because Alexander disease is a **demyelinating** (loss of myelin) disorder, there will be various cognitive and motor problems. Specific symptoms depend largely on the time of onset, but can include mild to severe motor and/or intellectual disabilities, speech abnormalities, seizure activity, and poor coordination.

3. What are the long-term effects of having Alexander Disease?

The loss of myelin is generally permanent meaning that the neurologic damage will lead to a variety of physical and cognitive problems. As such, most individuals will go on to have mild to severe intellectual and physical disabilities.

4. What are some of the main symptoms of Alexander Disease?

Symptoms depend on the time of disease onset.

- **Infantile (0-4 Years Old)**
  - Ataxia (Difficulty coordinating movements)
  - Spasticity (Stiffness and poor control of arms and legs)
  - Mental Retardation
  - Delayed Physical Development
- **Juvenile (4-18 Years Old)**
  - Speech Abnormalities
  - Difficulty Swallowing
  - Ataxia (Poor coordination)
- **Adult (18+ Years Old)**
  - Highly variable symptoms, but generally milder and more survivable.

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5. What causes Alexander Disease?

Alexander Disease is caused by a mutation in the ***GFAP*** gene.

6. Is there a cure?

There is currently **no cure**, but symptom management, along with the support of appropriate specialists can help slow down developmental regression. This can include specialists in neurology, physical medicine, speech and language, and any other therapists that may be of help.