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Insights on Krabbe Disease

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Description

Krabbe disease is an inherited disorder in which the protective coating (myelin) of nerve cells in the brain and throughout the nervous system is destroyed. In most cases, Krabbe disease manifests itself in babies before the age of six months, and the disease usually results in death by the age of two. When it manifests in older children and adults, the disease's course can vary greatly. There is no cure for Krabbe disease, and treatment is primarily supportive. However, stem cell transplants have shown some success in infants treated before symptoms appear, as well as in some older children and adults. Krabbe disease develops when a person inherits two copies of a faulty (mutated) gene [1-3], one from each parent.

A gene acts as a blueprint for the production of proteins. If this blueprint contains an error, the protein product may not function properly. Two mutated copies of a specific gene result in little or no production of an enzyme called galactocerebrosidase in the case of Krabbe disease (GALC). Enzymes, such as GALC, are in charge of breaking down certain substances in the recycling centre of a cell (lysosome). The accumulation of certain types of fats known as galactolipids occurs in Krabbe disease due to a lack of GALC enzymes. Prior to the onset of symptoms, as well as in some older children and adults. Galactolipids are normally found in cells that produce and maintain nerve cell protective coatings (myelin). An overabundance of galactolipids, on the other hand, is toxic. Some galactolipids cause self-destruction of myelin-forming cells.

Other galactolipids are taken up by microglia, which are specialized debris-eating cells in the nervous system. The process of removing excess galactolipids converts these normally beneficial cells into abnormal, toxic cells known as globoid cells, which promote myelin-damaging inflammation. Demyelination, or the loss of myelin, prevents nerve cells from sending and receiving messages. A blood sample will be sent to a lab to be tested for GALC enzyme activity. Krabbe disease may be indicated by very low or no GALC activity levels. Although the results assist a doctor in making a diagnosis, they do not indicate how quickly the disease may progress. For example, very low GALC activity does not always indicate that the condition will progress quickly. Your doctor may order one or more imaging tests to detect myelin loss (demyelination) in affected brain regions.

To confirm a diagnosis, a blood sample may be subjected to a genetic test. There are different versions of the mutated gene that causes Krabbe disease. The specific type of mutation may provide some benefits. The initial screening test assesses the activity of the GALC enzyme [4,5]. If the enzyme activity is found to be low, additional GALC and genetic tests are performed. The use of newborn screening tests is still in its early stages. Researchers are still trying to figure out how to use these tests most effectively, how well they lead to an accurate diagnosis, and how well they predict the course of the disease. To date, research suggests that identifying Krabbe disease markers before symptoms appear may open up a new treatment window.

There is currently no treatment that can change the course of Krabbe disease in infants who have already developed symptoms. As a result, treatment focuses on symptom management and supportive care. Interventions could include any of the following: Anticonvulsant medications to control seizures, Muscle spasticity and irritability medications, Physical therapy to prevent muscle tone deterioration Nutritional assistance, such as the use of a tube to deliver fluids and nutrients directly to the stomach (gastric tube).

Hematopoietic stem cells are specialised cells that can differentiate into all types of blood cells in the body. These stem cells also give rise to microglia, which are specialized debris-eating cells that live in the nervous system. Microglia are transformed into toxic globoid cells in Krabbe disease. Donor stem cells are delivered into the recipient's bloodstream via a tube known as a central venous catheter during stem cell transplantation. The donor stem cells assist the body in producing healthy microglia capable of populating the nervous system and delivering functional GALC enzymes. This treatment may aid in the restoration of some normal myelin production and maintenance.

Conflict of Interest

None.

References

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