Krabbe Leukodystrophy

Other names that may be used for this condition are:

- Galactocerebrosidase Deficiency
- Galactosylceramide Lipidosis
- Globoid Cell Leukodystrophy
- Krabbe Disease
- Sphingolipidosis, Krabbe Type

Krabbe Leukodystrophy is a rare inborn error of metabolism (IEM) that is the result of a deficiency of the galactosylceramidase (GALC) enzyme. Krabbe belongs to a group known as the Lysosomal Storage Diseases. This enzyme is involved in the breakdown of galactosylceramide which is a fat found in the myelin, kidney, and certain cells in the small intestine and colon. GALC enzyme deficiency leads to abnormal myelin formation in the central and peripheral nervous system. The genetic defect has been located to chromosome 14 (14q31). Although there are some common mutations, there are over 70 different mutations identified, which can make a genetic diagnosis difficult.

Krabbe Disease is a Leukodystrophy; "Leuko" refers to the myelin or the white matter in the brain and "dystrophy" refers to abnormal growth or formation. The disorder affects the myelin. This is the protective layer that surrounds certain nerve cells. Nerve cells have a cell body and an axon. The axon connects with the next nerve cell and is covered in myelin. Myelin ensures messages are passed quickly between nerve cells – damage to the myelin means the messages are sent slowly. If the GALC enzyme is deficient, it causes a build-up of fats that have not been digested and affects the growth of the myelin sheath. Krabbe Leukodystrophy is a progressive disorder. The symptoms become worse with time.

This information has been kindly checked by Dr E. Wassmer (Birmingham) and is correct as of 06/08/13. This is a short internet summary only. For a full summary and further more detailed information please contact CLIMB by email, letter or telephone.

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