Infantile Refsum Disease

Infantile Refsum disease is a genetic disorder affecting the peroxisomes. It is characterized by:

- Impairment in the proper breakdown of phytanic acid (a substance found in food).
- The gathering of phytanic acid in blood plasma and tissue.

This disease affects the growth of the myelin sheath. This is the fatty covering, which helps the nerve to work properly on nerve fibers in the brain. This disease is part of the group called the peroxisomal biogenesis disorders that cause a leukodystrophy. This disorder begins in early infancy.

SYMPTOMS

Symptoms and signs include:

- Developmental delay – motor and language.
- Seizures.
- Visual problems (rapid, involuntary jerky eye movements).
- Hearing problems.
- Decreased muscle tone (*hypotonia*).
- Impaired muscle coordination.
- Scaly skin.
- Failure to grow normally.
- Enlargement of the liver.
- A diminished amount of cholesterol in the blood.
- Mild abnormalities in the form and structure of the face.

TREATMENT

- Treatment is supportive. Physical, occupational and speech therapy are helpful to slow the progression of the disease.
- Dietary treatment and plasmapheresis have been helpful for the adult form, but have not been shown to be helpful for the infantile form. Cow's milk products should be avoided to decrease the amount of phytanic acid.

PROGNOSIS

Treatments are do not cure this disease. The disease gets worse over time (*progressive*) with half of the patients dying by the third decade.

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