Zellweger Syndrome

Zellweger syndrome is a rare disorder that is present at birth (*congenital*). It is characterized by the reduction or absence of cell structures that rid the body of toxic substances (*peroxisomes*) in the cells of the liver, kidneys, and brain. Zellweger syndrome is one of a group of genetic disorders called *peroxisomal diseases.* These affect brain development and the growth of the myelin sheath. That is the fatty covering, which acts as an insulator on nerve fibers in the brain.

SYMPTOMS

The most common features of Zellweger syndrome include:

- Enlarged liver.
- High levels of iron and copper in the blood.
- Vision disturbances.

Some affected infants may show prenatal growth failure. Symptoms at birth may include lack of muscle tone and an inability to move. Other symptoms may include:

- Unusual facial characteristics.
- Mental retardation.
- Seizures.
- Inability to suck and/or swallow.
- Jaundice.
- Gastrointestinal bleeding.

TREATMENT

There is no cure for this syndrome. Nor is there a standard course of treatment. Infections should be guarded against to prevent such complications as pneumonia and respiratory distress. Other treatment is symptomatic and supportive.

WHAT WILL HAPPEN

The prognosis for patients with this syndrome is poor. Death usually occurs within 6 months after onset. It may be caused by respiratory distress, gastrointestinal bleeding or liver failure.

RESEARCH BEING DONE

The NINDS supports research on genetic disorders. This includes leukodystrophies such as Zellweger syndrome. The goals of this research are to increase scientific understanding of these disorders, and to find ways to prevent, treat, and cure them.

Research is continually being done to learn more about this problem.

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