Alpers Disease

Alpers disease also known as **Christensen's disease**, **Christensen-Krabbe disease**, **Progressive Sclerosing Poliodystrophy**, **Alpers Progressive Infantile Poliodystrophy**, **Alpers Syndrome**, **Alpers-Huttenlocher Syndrome** or **Neuronal Degeneration Of Childhood With Liver Disease, Progressive (PNDC)** is a rare, genetically determined disease of the brain that causes progressive degeneration of grey matter in the cerebrum. It is inherited in an autosomal recessive pattern, meaning that affected children's parents are both carriers of the abnormal gene. Because it is so infrequently diagnosed, its incidence and prevalence are difficult to estimate.

Signs and Symptoms

The first sign of the disease usually begins early in life with **convulsions**. Other symptoms are **developmental delay**, **progressive mental retardation**, **hypotonia** (low muscle tone), **spasticity** (stiffness of the limbs), **dementia**, and **liver conditions** such as jaundice and cirrhosis that can lead to liver failure. **Optic atrophy** may also occur, often causing blindness.[1]

Causes

Researchers believe that Alpers disease is caused by an underlying metabolic defect that results in high rates of programmed cell death (apoptosis). Some patients have mutations in mitochondrial DNA.

Diagnosis

Exams and Tests

Researchers suspect that Alpers' disease is sometimes misdiagnosed as childhood jaundice or liver failure, since the only method of making a definitive diagnosis is by autopsy or brain biopsy after death. The diagnosis can be strongly suspected based on **histological, radiographic, and laboratory abnormalities**. Liver biopsy specimens may show bile duct proliferation and scarring. Genetic studies (DNA sequencing) may show mutations in the gene encoding DNA polymerase. Brain abnormalities may also be evident on MRI scans or electroencephalograms.[2]

Treatment

Medications

There is no cure for Alpers' disease and no way to slow its progression. Treatment is symptomatic and supportive. Anticonvulsants may be used to treat the seizures. Valproate should not be used since it can increase the risk of liver failure.[3] Physical therapy may help to relieve spasticity and maintain or increase muscle tone.

Expected Outcome

Survival Rate

The prognosis for individuals with Alpers' disease is poor. Those with the disease usually die within their first decade of life. Continuous, unrelenting seizures often lead to death. Liver failure and cardiorespiratory failure may also occur.

History

The disease was first described by Alfons Maria Jakob[4]; more extensive details were published by Bernard Jacob Alpers, Erna Christensen, and Knud Haraldsen Krabbe.

References

1. Harding BN. Progressive neuronal degeneration of childhood with liver disease (Alpers-Huttenlocher syndrome): a personal review. J Child Neurol. 1990 Oct;5(4):273-87
2. Gordon N. Alpers syndrome: progressive neuronal degeneration of children with liver disease. Dev Med Child Neurol. 2006 Dec;48(12):1001-3.
3. Schwabe MJ, Dobyns WB, Burke B, Armstrong DL. Valproate-induced liver failure in one of two siblings with Alpers disease. Pediatr Neurol. 1997 May;16(4):337-43.
4. Alpers BJ. Diffuse progressive degeneration of gray matter of cerebrum. Arch. Neurol. Psychiat. 25:469-505, 1931.

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