Leigh's Disease

Leigh's disease is a rare inherited neurometabolic disorder characterized by degeneration of the central nervous system. Leigh's disease can be caused by mutations in mitochondrial DNA or by deficiencies of an enzyme called pyruvate dehydrogenase. Symptoms (problems) of Leigh's disease usually begin between the ages of 3 months to 2 years and progress rapidly. In most children, the first signs may be poor sucking ability and loss of head control and motor (movement) skills. These symptoms may be accompanied by loss of appetite, vomiting, irritability, continuous crying, and seizures (convulsions). As the disorder progresses, symptoms may also include generalized weakness, lack of muscle tone, and episodes of lactic acidosis, which can lead to impairment of respiratory and kidney function. Heart problems may also occur. In rare cases, Leigh's disease can begin during late adolescence or early adulthood and progress more slowly.

TREATMENT

The most common treatment for Leigh's disease is thiamine or Vitamin B1. In patients who have a deficiency of pyruvate dehydrogenase enzyme complex, a high-fat, low-carbohydrate diet may be recommended. Oral sodium bicarbonate or sodium citrate may also be prescribed to manage lactic acidosis. Experimental protocols are currently using dichloroacetate to treat patients with lactic acidosis.

WHAT WILL HAPPEN

The prognosis (likely outcome) for individuals with Leigh's disease is poor. Individuals who lack mitochondrial complex IV activity and those with pyruvate dehydrogenase deficiency tend to have the worst prognosis and die within a few years. Those with partial deficiencies have a better prognosis, and may live to be 6 or 7 years of age. Some have survived to their mid-teenage years.

RESEARCH BEING DONE

The NINDS supports and encourages a broad range of basic and clinical research on neurogenetic disorders such as Leigh's disease. The goal of this research is to understand what causes these disorders and then to apply these findings to new ways to diagnose, treat, and prevent them.

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

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