Leigh Syndrome

1. **What is Leigh Syndrome?**
   Leigh syndrome is a severe neurological disorder that usually becomes apparent in the first year of life. This condition is characterized by progressive loss of mental and movement abilities (psychomotor regression) and typically results in death within two to three years, usually due to respiratory failure. A small number of individuals do not develop symptoms until adulthood or have symptoms that worsen more slowly.

2. **What are the symptoms?**
   Symptoms of Leigh's disease usually progress rapidly. The earliest signs may be poor sucking ability, and the loss of head control and motor skills. These symptoms may be accompanied by loss of appetite, vomiting, irritability, continuous crying, and seizures. 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.

3. **What disorders are associated with Leigh Syndrome?**

   Most individuals with Leigh's syndrome have defects of mitochondrial energy production, such as deficiency of an enzyme of the mitochondrial respiratory chain complex or the pyruvate dehydrogenase complex. In most cases, Leigh's syndrome is inherited as an autosomal recessive trait. However, X-linked recessive and mitochondrial inheritance are additional modes of transmission.

4. **How is Leigh Syndrome Diagnosed?**

   The diagnosis of Leigh syndrome may be confirmed by a thorough clinical evaluation and a variety of specialized tests, particularly advanced imaging techniques. Magnetic resonance imaging (MRI) or computed tomography (CT) scans of the brain may reveal abnormal areas in certain parts of the brain (i.e., basal ganglia, brain stem, and gray matter). An MRI uses a magnetic field and radio waves to produce cross-sectional images of particular organs and bodily tissues. During CT scanning, a computer and x-rays are used to create a film showing cross-sectional images of certain tissue structures.

5. How is Leigh Syndrome treated?

The most common treatment for Leigh's disease is thiamine or Vitamin B1. Oral sodium bicarbonate or sodium citrate may also be prescribed to manage lactic acidosis. Researchers are currently testing dichloroacetate to establish its effectiveness in treating lactic acidosis. In individuals who have the X-linked form of Leigh's disease, a high-fat, low-carbohydrate diet may be recommended.

Resources:

- Genetics Home Reference: https://ghr.nlm.nih.gov/condition/leigh-syndrome
- NINDS Leigh's Disease Information Page: http://www.ninds.nih.gov/disorders/leighsdisease/leighsdisease.htm
- Genetic and Rare Diseases Information Center (GARD): https://rarediseases.info.nih.gov/diseases/6877/leigh-syndrome