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Case Report
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# Leighs Disease: A Rare Case Report

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**Abstract**

Leighs disease is a rare neurological disease of childhood characterized by degeneration of nervous system. It majorly occurs in infants due to mitochondrial DNA deficiency. This condition is characterized by progressive loss of mental and movement abilities and typically results in death within two or three years.

**Keywords:** Leighs disease; Infants; Mitochondrial enzyme; Pyruvate dehydrogenase; Hypotonia; Impairment

**Abbreviations:** DNA: Deoxyribonucleic Acid; Inj: Injection, HR: Heart Rate; RR: Respiratory Rate; CNS: Central Nervous System; Hb: Hemoglobin; WBC: White Blood Cells; Plt: Platelets; CRP: C Reactive Protein; CSF: Cerebrospinal Fluid; CT: Computed Tomography; MRI: Magnetic Resonance Imaging; SOL: Space Occupying Lesion; BD: Twice in a day; TID: Three times in a Day; OD: Once in a Day; NADH: Nicotinamide Adenine Dinucleotide; CoQ: Coenzyme Q ; SURF-1: Surfeit Locus Protein 1; ATP: Adenosine Tri Phosphate

## Introduction

Leigh's disease is the rare progressive disorder of the childhood. Leighs disease is also known as subacute necrotizing encephalopathy. Leigh syndrome was named after Denis Leigh, who first described a unique neuropathology in an infant who had died of the disease; it is now known to affect approximately 1 per 40,000 live births. It is characterized by the degeneration of the central nervous system. The symptoms of Leigh syndrome usually begin between the ages of three months and two years, but some patients do not exhibit signs and symptoms until several years later. Symptoms may include loss of previously acquired motor skills, loss of appetite, vomiting, irritability, and/or seizure activity. As Leigh syndrome progresses, symptoms may also include generalized weakness, lack of muscle tone (hypotonia), and episodes of lactic acidosis, which may lead to impairment of respiratory and kidney function [1].

## Case Report

A 8 months old male child was admitted in the hospital with the complaints of fever and breathlessness since 3 days and had history of convulsions 1 episode in the form of tonic posturing of all four limbs and up rolling of eyes and was treated by giving inj. phenytoin. Patient was admitted in the hospital for 21 days. On initial examination patient was drowsy.HR-162/min, RR-36/min, CNS examination showed increased power in both lower limbs and bilateral extensor plantar reflexes. Milestones of the patient were not appropriate according to the age of the patient. The above clinical findings were highly suggestive of a neurodegenerative disorder and the patient was further investigated. Heamogramrevealed hb-8.1g/dl, WBC-6100c/cumm, Plt-4.18lakh/cumm. CRP was found to be positive. Gram and ZN staining of the CSF showed no organism and pus cells. Serum lactate, Liver and renal function tests were found to be normal. CT brain shows Bilateral symmetrical hypodensity both caudate and ganglionic regions. MRI shows Bilateral symmetrical T2W, Flair hyperintensecoreas with subtle diffusion restriction in basal ganglia, corona radiate. No evidence of hemorrhage or SOL in brain. Mild dilation of lateral ventricles, prominent extra cerebral spaces. Possibility of Leighs disease may be considered. Based on laboratory investigations the patients was diagnosed with leighs disease on 10th day of admission. From Day1-10 patient was treated with INJ. Ceftraixone 320mg BD, Inj. Amikacin 50mg BD, Inj. Phenytoin 20mg BD, Inj. Levetiracetam 33mg BD, Inj. Vancomycin 100mg TID. From Day 11-21 therapy was Inj. Cefotraixone 320mg BD, Inj.Leviteracetam 33mg BD, T.Carnitine ½ tab, T.Riboflavin 1 tab, T.Biotin 1tab, T.CoenzymeQ 1tab, T.Pyridoxine ½ tab, T.Folic acid ½ tab oral once in a day. On day-21 patient was discharged with therapy T.Carnitine ½ tab, T.Riboflavin 1 tab, T.Biotin 1tab, T.CoenzymeQ 1tab, T.Pyridoxine ½ tab, T.Folic acid ½ tab oral once in a day, Syp. Levetiracetam 1.5ml oral OD, Syp.PCT 5ml oral SOS [2].

## Discussion

Leighs disease is a rare nervous system disorder caused by mutations in mitochondrial DNA or deficiencies of an enzymes called pyruvate deoxygenase, mitochondrial respiratory chain emzyme andother nDNA-based enzyme deficiencies (i.e., NADH-CoQ and cytochrome C oxidase) have also been implicated as a cause of some cases of autosomal recessive Leigh syndrome. It was first reported in 1951 by Denis Leigh, a British neuropathologist, in a 7-month-old infant that progressed rapidly and resulted in death over a 6-week period. These specific enzyme deficiencies have been linked to several different gene mutations of the SURF1 gene located on chromosome 9 causes Leigh syndrome associated with cytochrome C oxidase deficiency. All of these different genetic defects seem to have a common effect on the central nervous system, resulting in progressive neurological deterioration. These enzyme deficinies are caused by mutations in different disease genes. These mutations may be inherited as an autosomal recessive trait, an X-linked recessive trait, or as a mutation found within the DNA of mitochondria. Women who are carriers of an X-linked disorder have a 50 percent risk of transmitting the carrier condition to their daughters, and a 50 percent risk of transmitting the disease to their sons. The mtDNA from the father is carried by sperm cells. However, during the process of fertilization, the father's mtDNA is lost. As a result, all human mtDNA comes from the mother. An affected mother will pass the traits to all of her children, but only the daughters will pass the mutation(s) onto the next generation [3,4].

There is no cure for leighs disease. Specific therapy for mitochondrial disorders for children is not available. symptomatic therapy is given to improve production of ATP and to lower lactate levels. Improvement of neurological symptoms in some patients is achieved by giving thiamine, a cofactor of pyruvate dehydrogenase. ATP Production was improved in some patients treated with Riboflavin. Rapid clinical and biochemical improvement was observed in patients with acute central respiratory failure with the use of intravenous soya bean oil (ketogenic emulsion). Ketogenic diet has been found to improve the outcome in those with a deficiency of pyruvate dehydrogenase. Coenzyme Q and Carnitine have also been found to be effective in reducing symptoms. This patient was treated with T. Carnitine ½ tab, T. Riboflavin 1 tab, T. Biotin 1tab, T. CoenzymeQ 1tab, T. Pyridoxine ½ tab.

## Conclusion

The diagnosis of Leigh's disease should be considered in appropriate clinical and laboratory settings whenever symmetrical hypodensities are encountered in the putamina and midbrain on CT and further investigated with MRI. Our experience suggested that bilateral symmetric T2 prolongation involving multiple brainstem nuclei/ structures associated with basal ganglia abnormalities in a child with neurological problems should prompt the clinician to consider and other laboratory investigations such as CSF lactate and respiratory chain enzymes activities are required to confirm leigh's syndrome in a child.

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