Background: Leigh syndrome (LS) is caused by mutations in one of more than 30 genes; most of which are associated with the mitochondrial respiratory chain (MRC). Aim: To identify the genetic cause of disease in a patient with an overall clinical picture of Leigh syndrome.

Patient & Methods: A girl with clinically suspected diagnosis of LS was first hospitalized at 2 years because of febrile seizures and partial left paresis, with subsequent left-sided arm and leg weakness, severe ataxia and worsening dystonia at 6 years. Brain MRI showed symmetrical atrophy of basal ganglia and thalamus, decreased signal intensity in the caudate heads, putamens, and centrum semiovale.

Conclusion: Whole exome sequencing (WES) identified a novel compound heterozygous mutation in the NDUFA10 gene, which encodes the ND5 subunit of complex I, associated with Leigh syndrome. This finding highlights the importance of WES in the diagnosis of mitochondrial disorders.

Whole exome sequencing identifies novel compound heterozygous mutations in NDUFA10 in affected siblings with a mitochondrial phenotype

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Background: In leucencephalopathy, mutation in gene DARS2 is associated with this syndrome. The gene is located on the long arm of chromosome 1 (1q 25.1), the disease is associated with deficiency of mitochondrial aspartyl - RNA synthetase.

Case report: A 1-year-old boy with static and psychomotor delay was admitted to the hospital with dysarthria, feeding difficulties, and hypertonia at 6 months of age. An examination revealed decreased muscle tone, hypertonia and hyperreflexia. The karyotype analysis showed no evidence of a genetic disorder.

Conclusion: In our case, we were able to find a single mutation and we developed a clinical diagnosis.