Leigh syndrome caused by mutations in the flavoprotein (Fp) subunit of succinate dehydrogenase (SDHA).

Abstract:
Detailed clinical, neuroradiological, histological, biochemical, and genetic investigations were undertaken in a child suffering from Leigh syndrome. The clinical symptoms started at age five months and led to a severe progressive neurodegenerative disorder causing epilepsy, psychomotor retardation, and tetraspasticity. Biochemical measurement of skeletal muscle showed a severe decrease in mitochondrial complex II. Sequencing of SDHA revealed compound heterozygosity for a nonsense mutation in exon 4 (W119X) and a missense mutation in exon 3 (A83V), both absent in normal controls. In six additional patients--five with Leigh or Leigh-like syndrome and one with neuropathy and ataxia associated with isolated deficiency of complex II--mutations in SDHA were not detected, indicating genetic heterogeneity.