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Subthalamic nuclei involvement inLeigh disease with cytochrome c oxidase deficiency

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A 22 month-old girl born to consanguineous parents presented with generalized hypotonia, nystagmus and loss of developmental milestones. Laboratory values revealed elevated blood lactate at rest (45 mg/dL; normal, 8-22 mg/dL).

Cranial MRI showed T2 signal prolongation involving bilateral subthalamic nuclei (Fig. 1, arrow). Symmetric areas of T2 hyperintensities were also found the the medulla above the pyramidal decussation and in dentate nuclei (Fig. 2, arrow). Diagnosis of Leigh syndrome was confirmed by deficient cytochrome c oxidase (COX) activity in biopsied muscle.

Involvement of the subthalamic nuclei with sparing of basal ganglia is considered as an distinctive MRI hallmark of Leigh syndrome with COX deficiency (1, 2).

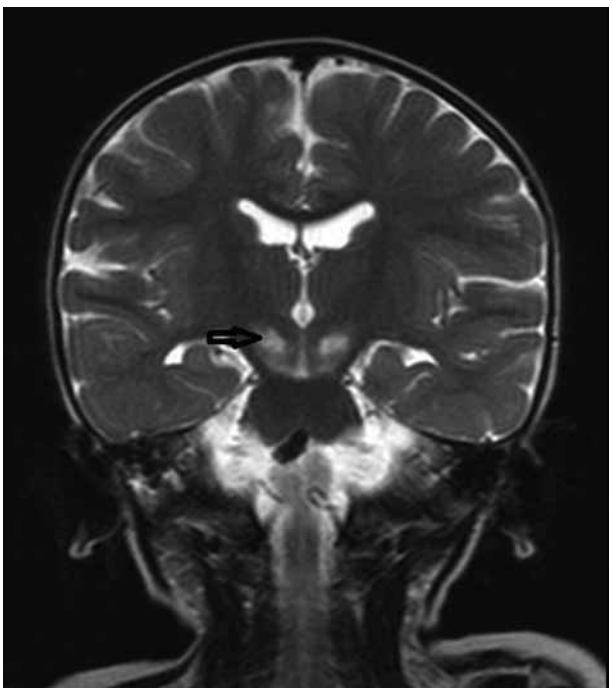

Fig. 1

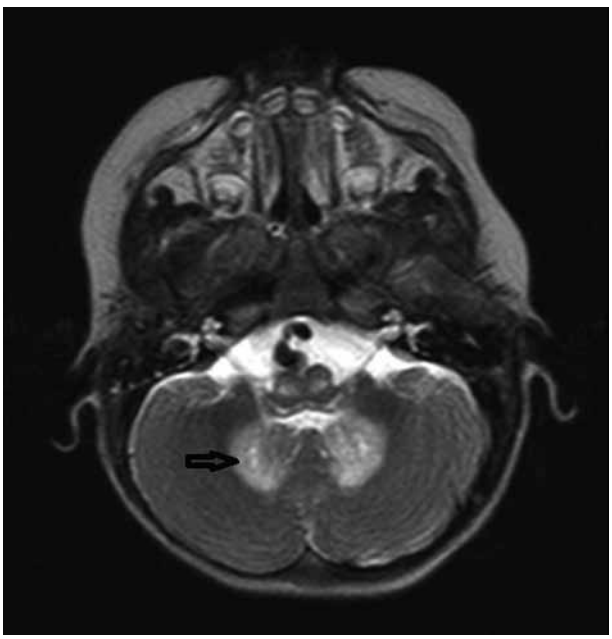

Fig. 2

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