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Acta Medica Iranica
2009;47(4) : 236-240

Leigh syndrome: Clinical and paraclinical study

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Abstract:

During two years study about mitochondrial disease (Sep 1999-Agu 2001), 15 cases of Leigh syndrome (LS) were diagnosed, that consisted of 11 boys and 4 girls aged between 6 to 156 (mean: 40.5) months. Most of the patients (46.7%) became symptomatic between 1-5 years of age. Triggering factors were reported in 66.6% of the patients and 40% of them became symptomatic after infections. The most frequent presenting symptoms of the patients were somnolence and lethargy (40%), developmental regression (20%) and seizure (13.3%). The most common neurologic findings were developmental regression or arrest (93.3%), seizure (93.3%) abnormal tone (86.7%) and abnormal movements (53.3%). Blood lactate increased in 93.3% and blood ammonia elevated in 26.7% of the cases. Symmetric striatal necrosis (100%) and caudate nucleus involvement (73.3%) were the most frequent neuroimaging findings of the patients.

Keywords:

Leigh syndrome ، Mitochondrial disease ، Dystonia ، Encephalopathy ، Lethargy ، Blood ammonia ، Blood lactate

TUMS ID: 1107

Full Text HTML Full Text PDF 188 KB

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