Spectrum of Mitochondrial Disorders in Children

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Aims & Objectives

To study the spectrum of mitochondrial disorders in children.

Material & Methods

The spectrum of mitochondrial disorders in children, the presentations, inheritance, diagnosis and progression was studied in 19 subjects. This study included all patients with Mitochondrial disease that were diagnosed in our hospital over a period of five years.

Results &Conclusions

Out of 19 patients 78% were males and 63% presented during infancy. 100% of the patients presented with primary developmental delay and neuro-regression. Family history was significant in 31%. Diagnosis was made from clinical profile, metabolic workup and MRI Brain findings. Muscle biopsies were done when indicated. 3 patients were diagnosed as LEIGHS, 2 as MELAS and 1 as KSS while the others remained unclassified.