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## SPECTRUM OF MITOCHONDRIAL DISEASES IN A TERTIARY REFERRAL CENTRE IN HONG KONG

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**Background:** Disorders of the mitochondrial respiratory chain may present with various neurologic features, including encephalopathy, myopathy and hearing loss. Non-neurologic presentations occur in over 30% of paediatric patients. We review the clinical presentations of patients with mitochondrial diseases in our centre.

**Methods:** From 1995 to 2009, restropective review of medical records revealed 22 patients with mitochondrial diseases (15 males and 7 females). The diagnosis of mitochondrial disease is either based on the clinical phenotype or the Mitochondrial Disease Criteria proposed by Morava et al as definite mitochondrial disorders. Respiratory chain enzymology was performed in Murdoch Childrens Research Institute, Australia. Genetic analysis was either performed in the former centre or locally.

**Results:** The age of presentation ranged from immediately after birth to 10 years of age. The most common clinical phenotype was Mitochondrial Encephalomyopathy, Lactic acidosis, and Stroke-like episodes (MELAS) (n=7, 32%). Only 2 carried the typical A3243 mutation. The second most common presentation was Leigh disease (LS) (n=6, 27%). Mutation in the SURF1 gene causing complex IV deficiency was found in 1 patient. A patient with Kearns-Sayre syndrome (KSS) and another with Pearson syndrome had a deletion in the mitochondrial DNA genome. One had Leber's Hereditary Optic Neuropathy (LHON). A patient who had mutation in the POLG gene presented with a spinocerebellar ataxia syndrome with intractable epilepsy. 2 patients with complex IV deficiency had a phenotype of encephalomyopathy with or without epilepsy. A patient with complex I deficiency presented with a multi-system disease including cataract, hearing impairment, global developmental delay, short statue and recurrent hypoglycaemia. 2 patients had an ill-defined phenotype. Both met the Mitochondrial Disease Criteria as definite mitochondrial disorders. One presented with a fatal cardiomyopathy and corneal opacity of neonatal onset. The other had a combination of global developmental delay with regression, intractable epilepsy, cortical visual impairment and generalized dystonia. Further biochemical and genetic analysis was in progress. Significant mortality was found among the cohort (n=7, 32%).

**Conclusion:** Mitochondrial oxidative phosphorylation defects form an important group of inborn error of metabolism causing significant morbidity and mortality in neurologically disabled children. High index of suspicion is necessary for a proper diagnosis including biochemical and genetic analysis.