Spectrum of mitochondrial diseases in a tertiary referral centre in Hong Kong

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Background

- Mitochondrial diseases refer to diseases of mitochondrial respiratory chain caused by mutations in:
  - Mitochondrial DNA
  - Nuclear DNA
- Prevalence of common mtDNA mutation:
  - 1 in 5,000 (Schueler et al 2004)
- Pathogenic mtDNA mutation in general population:
  - At least 1 / 200 (Schulz et al 2000)
- 80 - 95% without detectable pathogenic mtDNA mutation (presumed to harbour mutations in nuclear-encoded mitochondrial gene) (Yong et al 2018)
- A wide variety of presentations and phenotypes are well recognized.
- There are lack of diagnostic facilities in Hong Kong and Chinese data.

Objective

- To analyze spectrum of mitochondrial diseases in Hong Kong
- To assess usefulness of various mitochondrial diagnostic tests

Method

- Study period: 1985 to 2009
- Hong Kong West Cluster Mitochondrial disease database
- Retrospective medical record review
- Patients actively managed and followed up:
  - Modified Adult Criteria (MAC) (Barnier et al 2002):
    - Diffuse mitochondrial disorder
  - Mitochondrial Disease Criteria (MDC) – General Criteria (West & Smeitink 2003)
    - Diffuse mitochondrial disorder
  - Probable mitochondrial disorder (Total score >= 7)
  - n=22 [male 14, female 8] including 1 patient with MELAS with incomplete biochemical data

Results

- Age of presentation
- Clinical phenotypes

Limitation and Discussion

- 82% specific mitochondrial phenotypes: underestimation of non-specific syndrome
- Over-diagnosis of Leigh diseases with non-respiratory chain defects eg PDH deficiency
- Longitudinal analysis of individual patients
- Proper documentation of hyperferritinaemia
- Use of MDC for selection of patients for muscle biopsy: low scores in organ-specific presentation
- Further enzymology and molecular investigations in progress: will expand spectrum and improve MDC / MAC scores
- Commencement of L-arginine therapy in MELAS despite low MDC score

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