PREVALENCE OF DEAFNESS IN CHILDREN WITH MITOCHONDRIAL DNA DISEASE

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Background: An estimated 840 children are born with sensori-neural hearing loss (SNHL) each year in the UK and in some a causative genetic defect is identified. Many children 'acquire' a hearing deficit during childhood, which may be a consequence of an inherited mitochondrial DNA (mtDNA) mutation. The association between SNHL and mitochondrial disease is well established in adults, although the correlation between genotype and phenotype remains unclear.

Aims: To determine the prevalence of SNHL in children with mtDNA disease; and to assess correlation between mutation load and severity of SNHL.

Methods: Retrospective analysis of a validated mitochondrial disease rating scale and audiology in 34 patients aged < 30yrs with disease presenting in childhood, attending a specialist clinic was performed. Heteroplasmy level in urine was determined for each patient.

Results: 11 patients had SNHL (6 female, 5 male). Patients harbouring the m.3243A>G mutation formed the largest cohort (8/11 patients), with the highest prevalence in patients harbouring the same mutation (8/21 patients). The earliest age of onset of SNHL was in the m.3243A>G group (2 years) but this did not predict severity or progression of hearing loss. Heteroplasmy levels did not predict onset or severity of hearing loss.

Conclusions: Several genotypes recognised in mitochondrial disease are associated with SNHL in childhood. Both onset in infancy, and a gradual progression to deafness in later childhood, is recognised. In those children with recognised SNHL, heteroplasmy level does not predict the severity. Formal audiology is advised in all children harbouring pathogenic mtDNA mutations.