MELAS in Central Australia, a case report

Aim

Mitochondrial diabetes is thought to account for 1-3% of all cases of diabetes, with prevalence varying according to ethnicity. Maternally inherited diabetes and deafness (MIDD) and mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS) are caused by a mutation of mitochondrial DNA. The most prevalent mutation (~80%) is m.3243A>G affecting the tRNALeu(UUR) gene (MTTL1). Cases of MIDD evolving into MELAS have been reported, suggesting a spectrum of presentation for the same mutation.

Methods

We discuss a rare occurrence of MELAS/MIDD overlap in an 18-year-old First Nations female living in Central Australia.

Results

AL is an 18-year-old female who has lived with diabetes since age 10 and diagnosed with MELAS at age 15, having a mutation of m.3243A>G, heteroplasmy 59.3%. She has comorbidities of mild intellectual disability, seizures, emotional instability and significant social dyscrasia.

This case has added challenges due to a number of social complexities which has made genetic testing of other family members difficult. Interestingly, her mother shares the same gene mutation and lives with severe Diabetes of the Exocrine Pancreas (DEP) with mildly positive anti-GAD antibodies and hearing impairment. It is unclear if this is independent or related to the mutation; she certainly does not have MELAS. Two of AL’s maternal aunts, a maternal uncle, and her maternal grandmother also have a severe phenotype of diabetes. Her maternal grandmother also has severe deafness. Apart from her mother, no other relatives have undergone genetic testing.

Conclusion

Early identification of MIDD and MELAS is important for appropriate management, as they can have significant health implications. An overlap syndrome can be seen in individuals who may initially present with features consistent with MIDD and only later develop clinical features associated with MELAS, particularly under conditions of stress.