Sensorineural impairment in maternally inherited diabetes mellitus and deafness (MIDD) disorder

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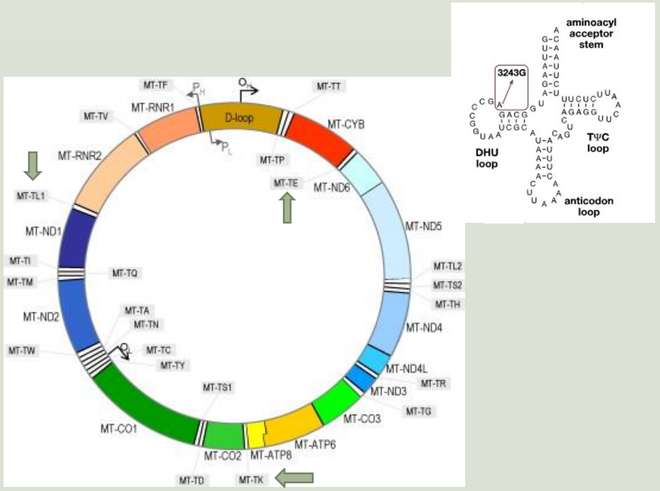
INTRODUCTION

The diagnosis of maternally inherited diabetes and deafness (MIDD) is suspected based on the presence of maternal impaired glucose tolerance or diabetes associated to hearing impairment or retinal maculopathy.

MIDD is caused usually by a mutation A>G at position 3243 of the mitochondrial DNA (figure 1).

Figure 1: The mitochondrial mutations of MIDD (1)



OBJECTIVES

Here, we report a child having a maculopathy and a hearing loss associated to MIDD. The aim of this work is to show the importance of ophthalmic imaging in the characterization and diagnosis of MIDD syndrome.

MATERIALS AND METHODS

Clinical, otologic and ophtalmic examination as well as optic multimodal imaging were conducted for a child suspected to have MIDD syndrome.

RESULTS

The case report was an eight-year-old female child from Tunisia for who a sensorineural hearing loss was diagnosed and treated by a cochlear implantation. Her diabetes was revealed by ketoacidosis. Ophthalmic examination showed an alteration of the macula with fine perifoveal deposits and a retinal pigment epithelium atrophy in the macular area. Multifocal electroretinography showed normal function of photoreceptors.

The diagnosis of MIDD was confirmed by the coexistence of diabetes mellitus, sensorineural hearing loss and a well characterized macular dystrophy. The retinal pigment epithelium impairment may be present without or precede photoreceptors dysfunction in MIDD.

CONCLUSION

Recently, it was demonstrated that MIDD may be caused by genetic changes in the MT-TL1, MT-TK, or MT-TE gene (figure 1). The clinical characteristics and severity of MIDD may be influenced by the genotype and its heterogeneity, related in particular to the heteroplasmy level of mutant mitochondrial DNA as well as the additional mitochondrial DNA mutations.

Whereas, genetic diagnosis of MIDD remains problematic and not always available, accurate diagnosis of the phenotype of the associated sensorineural impairments may be significant .

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