The Clinical Variability of Maternally Inherited Diabetes and Deafness Is Associated with the Degree of Heteroplasmy in Blood Leukocytes

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Résumé en anglais	**Context:** Maternally inherited diabetes and deafness (MIDD) is a rare form of diabetes with a matrilineal transmission, sensorineural hearing loss, and macular pattern dystrophy due to an A to G transition at position 3243 of mitochondrial DNA (mtDNA) (m.3243A>G). The phenotypic heterogeneity of MIDD may be the consequence of different levels of mutated mtDNA among mitochondria in a given tissue. **Objective:** The aim of the present study was thus to ascertain the correlation between the severity of the phenotype in patients with MIDD and the level of heteroplasmy in the blood leukocytes. **Participants:** The GEDIAM prospective multicenter register was initiated in 1995. Eighty-nine Europid patients from this register, with MIDD and the mtDNA 3243A>G mutation, were included. Patients with MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes) or with mitochondrial diabetes related to other mutations or to deletions of mtDNA were excluded. **Results:** A significant negative correlation was found between levels of heteroplasmy and age of the patients at the time of sampling for molecular analysis, age at the diagnosis of diabetes, and body mass index. After adjustment for age at sampling for molecular study and gender, the correlation between heteroplasmy levels and age at the diagnosis of diabetes was no more significant. The two other correlations remained significant. A significant positive correlation between levels of heteroplasmy and HbA1c was also found and remained significant after adjustment for age at molecular sampling and gender. **Conclusions:** These results support the hypothesis that heteroplasmy levels are at least one of the determinants of the severity of the phenotype in MIDD. Heteroplasmy levels are at least one of the determinants of the severity of the phenotype of maternally inherited diabetes and deafness.
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