Novel mutation in the HNF4-alpha gene and reclassification of diabetes in a family?

**Case Report**

**Past Medical History**
- Female
- First child of non-consanguineous parents
- Born at full term by vaginal delivery
- Uneventful pregnancy without diagnosis of gestational diabetes
- Apgar score of 10/10 at 1st and 5th minute
- Weight 3760g (p90)
- Uneventful neonatal period
- Adequate growth and development
- Weight between p85-97 since the age of 5 years - overweight
- No previous hospitalizations or surgery
- Father and grandmother with Diabetes

**Family History**

**Presumptive diagnosis of Maturity-Onset Diabetes of the Young (MODY)**

- Multiple daily insulin injections therapy (4 times/day)
- Total Daily Insulin 0.5 U/kg/day
- Metformin 500 mg twice a day

**Genetic study was requested** → The variant c.602A>C (p.His201Pro) in the HNF4-alpha gene was found in heterozygosity

Subsequently, a genetic study was also performed on the father, and the same variant was found.

- Assymptomatic
- Hba1c 8.1%

**CONCLUSIONS**

- The authors decided to present this case since this genetic variant is not described in the literature.
- The diagnosis of this adolescent also allowed the reclassification of the father’s diagnosis of diabetes.
- A correct classification of diabetes is important because it can predict the clinical course of the disease, clinical orientation and pharmacological treatment.

The authors have no conflicts of interest to declare.

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