

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

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The authors have no conflicts of interest to declare

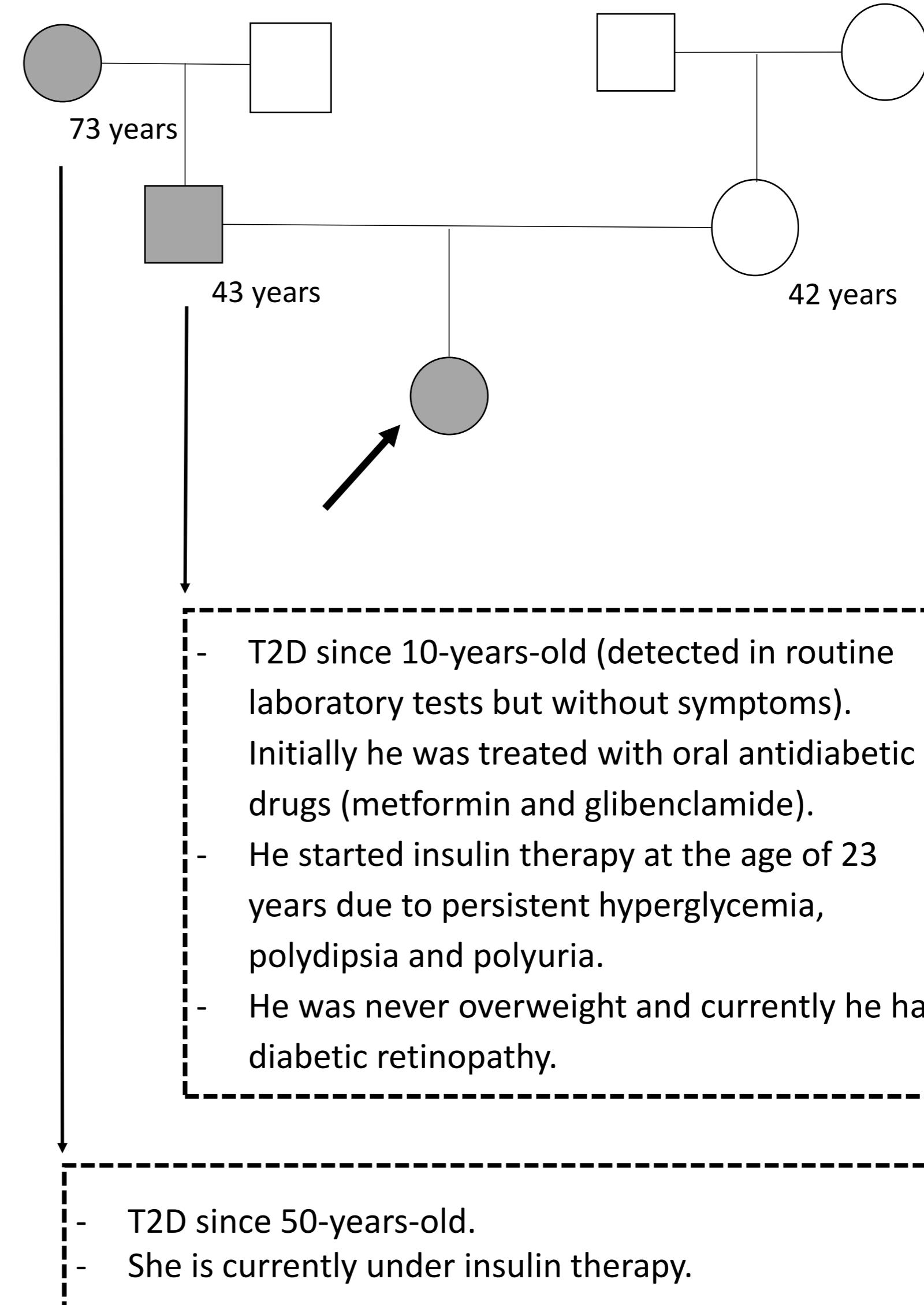


Case Report

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



11 years

- 1-month evolution of mild symptoms: polydipsia, polyuria, sporadic abdominal pain and nocturia;
- She was admitted to the emergency room due to postprandial hyperglycemia measured on her father's glucometer;
- Glucose 350 mg/dL; HbA1c 12.0%; no ketosis or acidosis
Normal lipid profile and thyroid function
Insulin 28.5 uUI/mL (reference value: 6-27)
C-peptide 2.22 ng/mL (reference value: 0.8-6.0)
- Negative auto-immunity:
Islet Cell Cytoplasmic Ab (ICA), Insulin AutoAb (IAA), Glutamic Acid Decarboxylase Ab (GADA), Zinc transporter AutoAb, Endomysial Ab (EMA), HLADQ2/DQ8, Thyroglobulin Ab (TGab), Thyroid Peroxidase Ab (TPO)

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

Currently 9 months later

- Assymptomatic
- HbA1c 8.1%

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

- ✓ Multiple daily insulin injections therapy (4 times/day)
- ✓ Total Daily Insulin 0.6 U/kg/day
- ✓ Metformin 500 mg twice a day

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.

