THE STORY OF A HETEROZYGOUS HNF1A MUTATION

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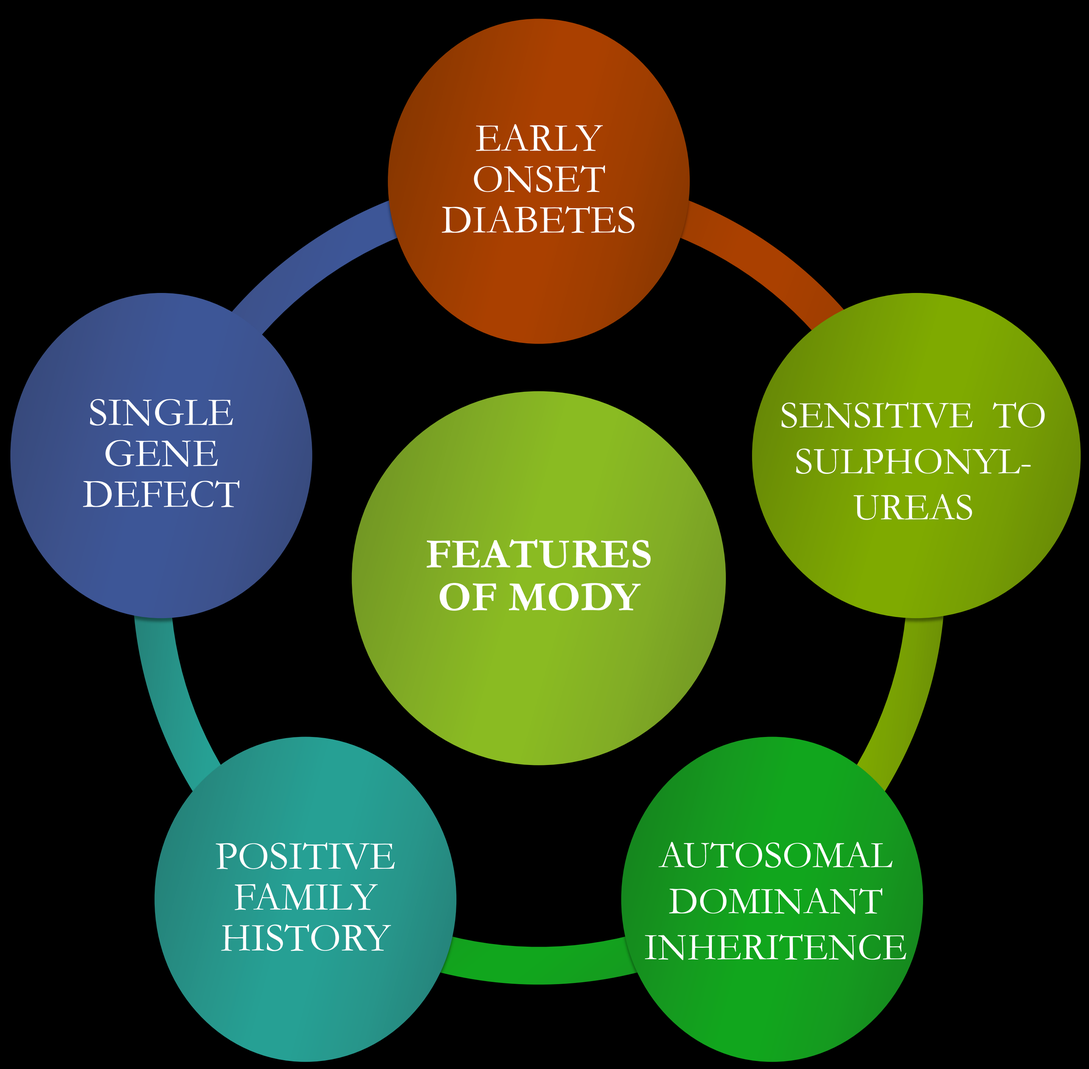
BACKGROUND



OBJECTIVE & HYPOTHESIS

We report a 15 year old girl with a HNF1A mutation who presented with MODY without a positive family history.

HNF1A-MODY is often misdiagnosed as type 1 or type 2 diabetes.

Genetic confirmation of MODY in patients treated with insulin helps in making changes in the treatment modalitiy.

We tested our patient for HNF1A mutation as she showed features of not being insulin dependent.

- Not developing ketoacidosis in the absence of insulin
- Good glycaemic control on a small dose of insulin
- Detectable C-peptide reflecting intrinsic insulin secretion

METHOD / CASE STUDY

A 15 year old girl presented with fainting episodes and feeling thirsty. There was no family history of diabetes.

Investigations & Results Part I

Blood glucose at initial presentation	12mmol/L
OGTT fasting blood glucose	6.7mmol/L
OGTT blood glucose at 120 minutes	14.9mmol/L
Blood glucose	7 -11 mmol/L
HbA1c	64mmol/mol

She was commenced on MDI with Levemir as basal and Novorapid as bolus at 0.25units/kg/day.

2 months later…Investigations & Results Part II

Islet Cell Antibody	Negative
GAD antibody	Negative
C Peptide	522pmol/L
Blood glucose	4-8mmol/l
Heterozygous HNF1A mutation	Positive

Insulin was stopped and she was started on sulphonylureas.

CONCLUSION

The molecular diagnosis of MODY is important to

- Classify the diabetes
- Predict prognosis
- Screen asymptomatic family members

The diagnosis, in this case, began with clinical suspicion.

We recommend that genetic testing of MODY should be considered for carefully selected individuals even when there is no family history of diabetes.

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