

# WHEN TYPE MODY II DIABETES SIMULATES TYPE I DIABETES

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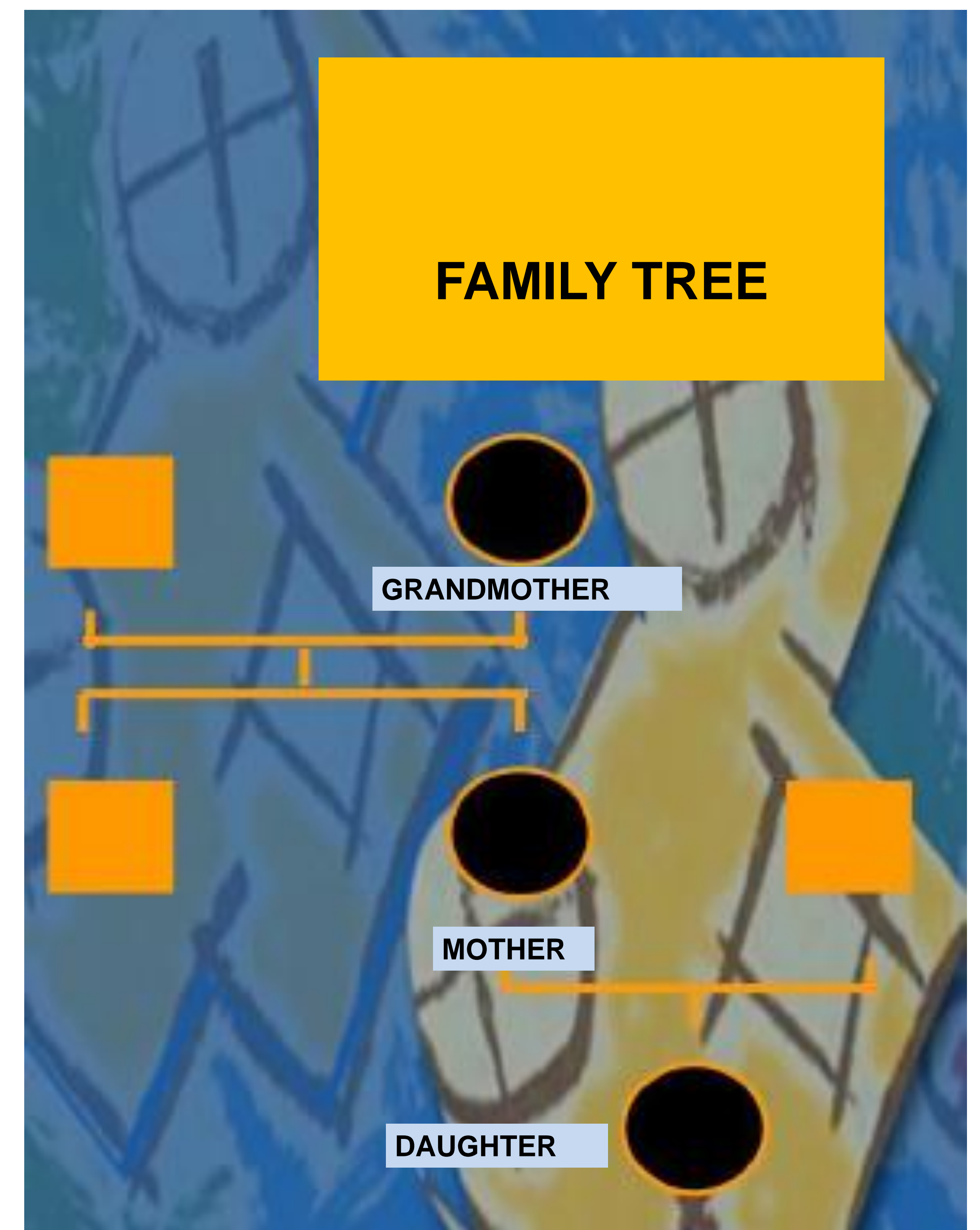


## BACKGROUND

The Monogenic diabetes in the childhood is the most frequent type after Type 1 diabetes. There have been described 8 subtypes, being the type Mody 2 diabetes the most frequent form in our population. It is characterized by mutation in heterozygosis of the gene that codifies glucokinase (7p15), formed by 10 exons. Its function is the phosphorylation of 6-glucose-phosphate. This enzyme is considered as the glycaemic sensor of the beta pancreatic cell, which increases the minimum threshold of glycemia for the release of insulin. More than 200 mutations are known, without there being correlation between the mutation and the phenotype. Its pattern is dominant autosomal inheritance. Is characterized by mild hyperglycemia especially in fastes, asymptomatic and non progressive, with little risk of developing microvascular and macrovascular complications

## CASE REPORT

A 4.5 years old female infant, was referred due to presenting polyuria, polydipsia and fasting hyperglycemia of 126-130mg/dl and 2 hours post-intake blood glucose level of 150-220mg/dl  
 She was born by normal delivery and was symmetrical IUGR, her weight and height in the 25th percentile.  
 Family History: Her mother was diagnosed with gestational diabetes, controlled by diet. One year later, she was diagnosed with Type I Diabetes requiring insulin treatment. Maternal grandmother was diagnosed with Type II Diabetes requiring at the beginning oral antidiabetics.



<b>FASTING GLUCOSE LEVEL</b>	<b>121 AND 130MG/DL</b>
<b>INSULIN LEVEL</b>	<b>3.8mUI/ML</b>
<b>C-PEPTIDE</b>	<b>0.4NG/ML</b>
<b>HEMOGLOBIN A1C LEVEL</b>	<b>6.2%</b>
<b>THYROID HORMONES</b>	<b>NORMAL</b>
<b>THYROID ANTIBODIES</b>	<b>NEGATIVE</b>
<b>NEGATIVE DIABETES ANTIBODIES</b>	<b>(ANTI-GAD, ISLET, INSULIN AUTOANTIBODIES)</b>
<b>COELIAC BLOOD TEST</b>	<b>NEGATIVE</b>
<b>URINE TEST</b>	<b>NEGATIVE MICROALBUMINURIA. NOT GLUCOSURIA.</b>
<b>HLA TYPING TEST</b>	<b>NOT COMPATIBLE WITH TYPE I DIABETES</b>
<b>MODY MOLECULAR STUDY II</b>	<b>NEGATIVE SEQUENCING OF EXONS 1 TO 10 AS WELL AS FLANKING ZONES OF THE GCK GENE.</b>

<b>OGTT</b>	<b>0'</b>	<b>120'</b>
<b>GLUCOSE (mg/dl)</b>	<b>230</b>	<b>298</b>

**MPLA STUDY(MULIPLEX LIGATION DEPENDENT PROBE AMPLIFICATION) OF GLUCOKINASE GEN SHOWS HETEROZIGOUS DELETION OF EXONS 1 TO 10 IN THE 5'UTR(p.Men1\_Gln465del;c.1\_1395+del). SAME MUTATION WAS FOUND IN MOTHER'S TOO**

## EVOLUTION

At the first year of evolution she presented with clinical signs of polyuria, polydipsia, fasting and post-intake hyperglycemia greater than 250mg/dL with HbA1C of 7.2%, following which she started treatment with a low dose of glargine insulin (3 units daily). After the MPLA study that confirmed the correct diagnosis of Type II Mody Diabetes, treatment with diet was performed and immediately her glycaemia normalized and clinical symptoms disappeared. The HbA1c in mother and her was 6.2 and 6.5 respectively.

## CONCLUSION

Type 2 Mody Diabetes is characterized by the fact that it does not present with classic symptoms of diabetes, with little risk of developing complications, so it does not usually require treatment, although it is necessary to monitorize closely all the cases. The correct and early detection of the disease through molecular studies (MPLA), will allow us to identify those patients who with the RCP technique were false negatives. Nevertheless, the fundamental goal is a clinical and personalized follow-up of the patients to improve their quality of life.