Hyperglycemia
Mody: a diagnosis to remember

Ricardo Monteiro[1], Ana Laura Fitas[2], Marta Amado[3], Rosa Pina[2], Lurdes Lopes[2]

1 – Pediatric Department, Centro Hospitalar de Leiria
2 – Pediatric Endocrinology Unit, Hospital Dona Estefânia, Centro Hospitalar Lisboa-Central
3 – Pediatric Department, Centro Hospitalar do Algarve

Background

The detection of hiperglycemia on occasional evaluation raises the diagnosis of diabetes mellitus (DM). Maturity Onset Diabetes of the Young (MODY), namely glucokinase deficiency, should be considered in cases of non-progressive hyperglycemia associated with a positive family history.

We describe two unrelated cases of asymptomatic hyperglycemia where glucokinase mutations were detected.

Case Report

	Case 1	Case 2
Motive	Impaired Fasting Glucose	Occasional Hyperglycemia
Family History		
	Father – impaired fasting glucose and hepatic steatosis **Paternal Grandparents** – T2DM	**Mother** – gestational diabetes; currently T2DM **maternal grandmother and great-grandfather** – T2DM
Presentation		
	6 years old, Female **3 years old** – hyperglycemia (195 mg/dL) was detected Evaluation was repeated – fasting glycemia 110-128mg/dL **6 years old –** OGTT (fasting - 122mg/dL; 120' - 144mg/dL) Referred to our center; Clinical observation was negative	**5 years old, Male** **3 years old** – gastroenteritis , hyperglycemia (169mg/dL) Repeated home monitoring: fasting (104-117mg/dL); postprandial (120-160mg/dL) Referred to our center; Clinical observation was negative
Laboratory evaluation		
	Fasting Glycemia 121mg/dL **A1cHb** 6,6% **Insulin** 4,4ng/ml **HOMA** 1,2 Positive **Anti-GAD antibody** (5,4U/mL – N<0,9) Undetectable **ICA and IAA autoantibodies**	**OGTT** – Fasting 109mg/dL; 120' 162mg/dL **A1cHb** 6,3% **Insulin** 1,6ng/ml **HOMA** 0,3 Negative **Autoantibodies**

Diagnostic hypothesis: MODY

Genetic Study

GCK Gene Mutation **MODY2** New Mutation Heterozygous variant c.1149_1166del18	**GCK Gene Mutation** **MODY2** Heterozygous Mutation c.544G>C

In both cases, only dietetic measures have been used so far

Conclusion

In patients with asymptomatic hyperglycemia, MODY diagnosis should be considered as it has both prognostic and therapeutic implications. The genetic characterization of the subtype is essential.

References

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