The First Case Report on Maturity Onset Diabetes of Young-11 (Mody-11) from West Bengal, India

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Abstract

Background: MODY is a clinically group of heterogeneous disorder characterized by non-insulin dependent diabetes diagnosed at a young age (<25 yrs) with autosomal dominant transmission & lack of autoantibodies.

Case report: This case report is on MODY in a 30 days old baby. The patient also has a umbilical hernia & choledochal cyst.

Conclusion: We present an instance of 30 days old child with MODY (1) with a family background of diabetes. While more information is expected to legitimize widespread evaluating for diabetes with tests. Practitioners ought to be cautious with family h/o screening of diabetes. In families at least two ages of diabetes, there ought to be a low edge for asymptomatic screening with a serum Hb1Ac.

Keywords: Diabetes; MODY; Screening; Treatment; Prognosis

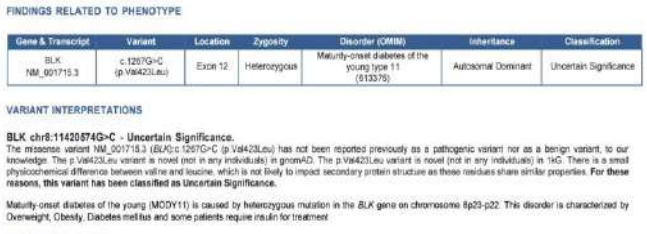
Introduction

Development beginning diabetes of young (MODY) is brought about via autosomal prevailing transformations and 14 MODY subtypes have been recognized at present. The right finding gives precise hereditary advising and may assist with appropriating management. However, the clinical cross-over between MODY, Type 1 diabetes and Type 2 diabetes makes it hard to analyze in an exact and opportune manner. The commonness is 1.2% in neonatal diabetic population. Approximately close to 100% of instances of MODY result from changes in HNF4A (MODY 1), Glucokinase (MODY 2) and HNF1A (MODY 3). The show of MODY is heterogeneous which makes distinguishing proof of these patients difficult. The clinical aggregate and movement among patients with a similar hidden transformation can be variable, reflecting the impact of the climate on quality expression. The

most normal show of MODY is hyperglycemia with a family h/o autosomal predominant diabetes. Other side effects might incorporate nocturia, GI indications and seldom DKA. The quality liable for MODY 11 is Tyrosine kinase B-lymphocyte specific(8p23-p22); primary imperfection MIN6beta cells. MODY ought to be clinically presumed when the accompanying measures are met: Age of analysis <25 yrs, non-insulin subordinate and autosomal predominant family background of diabetes. Screening Hb1Ac, blood glucose levels and islet cell autoantibodies are the underlying conclusion of MODY; however corroborative direct quality sequencing is needed for determination if the show and clinical history recommend MODY.

Case report

The patient is 30 days old baby presented with refractory hypoglycemia & convulsions. He has also umbilical hernia & choledochal cysts. Routine investigations indicated hyperinsulinemia. He was investigated for genomic sequencing. The findings: Gene & Transcript: BLK(NM_001715.3), variant: c.1267G>C (p. Val423Leu), location: Exon 12) & the disorder is MODY 11(613375).



FINDINGS RELATED TO PHENOTYPE

Gene & Transcript	Variant	Location	Zygosity	Disorder (OMIM)	Inheritance	Classification
BLK NM_001715.3	c.1267G>C (p.Val423Leu)	Exon 12	Heterozygous	Maturity-onset diabetes of the young type 11 (613375)	Autosomal Dominant	Uncertain Significance

VARIANT INTERPRETATIONS

BLK chr8:11420874G>C - Uncertain Significance.
The missense variant NM_001715.3 (*BLK*):c.1267G>C (p.Val423Leu) has not been reported previously as a pathogenic variant nor as a benign variant, to our knowledge. The p.Val423Leu variant is novel (not in any individuals) in gnomAD. The p.Val423Leu variant is novel (not in any individuals) in 1kG. There is a small physicochemical difference between valine and leucine, which is not likely to impact secondary protein structure as these residues share similar properties. **For these reasons, this variant has been classified as Uncertain Significance.**

Maturity-onset diabetes of the young (MODY11) is caused by heterozygous mutation in the *BLK* gene on chromosome 8p23-p22. This disorder is characterized by Overweight, Obesity, Diabetes mellitus and some patients require insulin for treatment

Discussion

Albeit the pervasiveness of MODY is a lot more modest than that of different types of diabetes, the finding of this condition is regularly postponed or misdiagnosed as either T1DM or T2DM. Currently, no evaluating tests exist for MODY. Direct quality sequencing as a screening device. Early determination and suitable administration can help in preventing potentially irreversible consequences of undiscovered, diligent hypoglycemia. The MODY 11 is presumably the principal case detailed from West Bengal, India. There is no legitimate reference at all. I am announcing the very uncommon case for additional review and revelation of new treatment convention.

Conclusion

We present an instance of 30 days old child with MODY with a family background of diabetes. While more information are expected to legitimize widespread evaluating for diabetes with tests. Practitioners ought to be watchful with family h/o screening of diabetes. In families at least two ages of diabetes, there ought to be a low edge for asymptomatic screening with a serum Hb1Ac.

MODY 11 is uncommon of most extraordinary variety, probably the primary case from West Bengal, India. Although MODY is undeniably challenging to analyze yet this assortment of MODY is extremely serious that further review and treatment systems are exceptionally anticipated.

The case report is approved by Institutional Ethics Committee.

References

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