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The first case report on maturity-onset diabetes of young-11 from West Bengal, India

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ABSTRACT

Maturity-onset diabetes of young (MODY) is a clinically group of heterogeneous disorder characterized by non-insulin-dependent diabetes diagnosed at a young age (<25 years) with autosomal dominant transmission and lack of autoantibodies. This case report is on MODY in a 30-day-old baby. The patient also has an umbilical hernia and choledochal cyst. We present an instance of 30-day-old child with MODY with a family background of diabetes. While more data are needed to justify universal screening for diabetes with tests. Practitioners should be vigilant with family history of screening of diabetes. In families two or more generations of diabetes, there should be a low threshold for asymptomatic screening with a serum Hb1Ac.

Key words: Diabetes; Maturity-onset diabetes of young; Screening; Treatment; Prognosis

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INTRODUCTION

Maturity-onset diabetes of young (MODY) is caused by autosomal dominant mutations and 14 MODY subtypes have been identified at present. The correct diagnosis provides accurate genetic counseling and may help to appropriate management. However, the clinical overlap between MODY, Type 1 diabetes, and Type 2 diabetes makes it difficult to diagnose in an accurate and timely manner. The prevalence is 1.2% in neonatal diabetic population. Approximately 99% of cases of MODY result from mutations in HNF4A (MODY 1), Glucokinase (MODY 2)[1], and HNF1A (MODY 3). The presentation of MODY is heterogeneous which makes identification of these patients difficult. The clinical phenotype and progression among patients with the same underlying mutation can be variable, reflecting the effect of the environment on gene expression. The most common presentation of MODY is hyperglycemia with a family history of autosomal dominant diabetes. Other symptoms may include nocturia, gastrointestinal symptoms, and rarely, DKA. The gene responsible for MODY 11 is tyrosine kinase B-lymphocyte specific (8p23-p22); primary defect MIN6 beta cells. MODY should be clinically suspected when the following criteria are met: Age of diagnosis <25 years, non-insulin dependent, and autosomal dominant family history of diabetes. Screening Hb1Ac, blood glucose levels, and islet cell autoantibodies are the initial diagnosis of MODY; however confirmatory direct gene sequencing is required for diagnosis if the presentation and clinical history suggest MODY.

CASE REPORT

The patient is 30-day-old baby presented with refractory hypoglycemia and convulsions. He has also umbilical

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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 chr8:11420574G>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

hernia and choledochal cysts. Routine investigations indicated hyperinsulinemia. He was investigated for genomic sequencing. The findings: Gene and Transcript: BLK (NM_001715.3), variant: c.1267G>C (p.Val423Leu), location: (Exon 12) and the disorder is MODY 11 (613375).

DISCUSSION

Although the prevalence of MODY is much smaller than that of other forms of diabetes, the diagnosis of this condition is often delayed or misdiagnosed as either Type 1 diabetes mellitus or Type 2 diabetes mellitus. At present, no screening tests exist for MODY.[2] Direct gene sequencing to be used as a screening tool. Early diagnosis and appropriate management can aid in preventing potentially irreversible consequences of undiagnosed, persistent hypoglycemia. The MODY 11 is probably the first case reported from West Bengal, India. There is no proper reference at all. I am reporting the extremely rare case for further study and discovery of new treatment protocol.

CONCLUSION

We present a case of 30-day-old baby with MODY with a family history of diabetes. More data are needed to justify universal screening for diabetes. Practitioners should be vigilant with family history of screening of diabetes. In families two or more generations of diabetes, there should be a low threshold for asymptomatic screening with a serum Hb1Ac.

MODY 11 is rare of rarest variety, probably the first case from West Bengal, India. Although MODY is very difficult to diagnose, this variety of MODY is so severe that further study and treatment strategies are highly expected.

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