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NEONATAL DIABETES MELLITUS DUE TO A RARE MUTATION IN KCNJ11 GENE

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Neonatal diabetes is a heterogeneous group of rare monogenic disorders with an incidence of about 1 in 100,000 live births presenting with in first six months of life with polyuria, dehydration and ketoacidosis. We hereby present a case of neonatal diabetes mellitus due to a very rare mutation (reported first time from Indian population) affecting the KCNJ11 gene encoding for KIR6.2 subunit of K_{ATP} channels resulting in inhibition of insulin release and hyperglycemia leading to permanent neonatal diabetes for which sulfonylurea are the preferred treatment instead of insulin injection as endogenous insulin synthesis is not affected unlike other causes of permanent neonatal diabetes mellitus affecting insulin synthesis for which insulin is the only treatment.

Neonatal diabetes mellitus (NDM) is defined as insulin-requiring persistent hyperglycemia occurring in first 6 months of life. [1] It is categorized in to permanent neonatal diabetes and transient neonatal diabetes. [2] Heterozygous activating mutations in the KCNJ11 and ABCC8 genes which encode the Kir 6.2 and SUR1 subunits of the ATP-sensitive potassium (K_{ATP}) channels that control insulin secretion are the commonest and a rarer cause of permanent neonatal diabetes respectively. [3] It is vital to recognize K_{ATP} channel mutation as most of these patients don't require insulin injections and achieve better glycemic control with sulphonylureas as in current case.

We describe one such of case neonatal diabetes mellitus due to a rare KCNJ11 mutation.

References

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