Clinical and genetic features of permanent neonatal diabetes mellitus

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Abstract

Neonatal diabetes mellitus (NDM) is one of the most unusual and exceptional type of diabetes that occurs in infants before the age of 6 months. Transient neonatal diabetes mellitus (TNDM) and permanent neonatal diabetes mellitus (PNDM) are identified clinically. The study conducted was a retrospective cohort study by selecting eight children with neonatal diabetes mellitus between March 2009 and February 2012. The study was presented to King Abdul Aziz University Hospital, in Jeddah, Saudi Arabia. Mutational analysis was performed retrospectively to identify phenotype and genotype characteristics. All patients had NDM and the first symptoms were observed during 1 to 17 weeks of birth, with five males and three females. None of them showed dysmorphic features, seizures, or developmental delay. The timespan of symptoms reported by parents before diagnosis ranged from 3 to 10 days with mean duration of 5.6 days. In two patients (25 %), genetic studies revealed positive mutations, with one patient depicting KCNJ11 mutation and the other had an INS mutation additional screening for ABCC8 and FOXP3 mutations were negative. All patients showed permanent NDM and no transient NDM or the remission at any stage of the disease was observed. Neonatal diabetes is a rare medical condition which needs to be differentiated from transient neonatal hyperglycemia. The medical practitioners should look for molecular basis of neonatal diabetes in order to treat it as it will lead to proper treatment with an appropriate therapy.

Keywords

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