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Rare Cases of Permanent Neonatal Diabetes Mellitus

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Introduction

Neonatal Diabetes mellitus is a rare form of monogenic diabetes with frequency between 1:20,000-100,000 live births. It presents in the first 6 months of life and 50% of cases have Permanent neonatal diabetes mellitus (PNDM)

Aim

To report two children with PNDM diagnosed in the first few days of life due to different genetic causes.

Case 1: A term boy with birth weight of 1.78kg developed diabetes on day two of life, He was the second child to consanguineous parents and mother had recurrent abortions due to CMV infection. He has a triangular face, normchromic normocytic anemia with low reticulocyte count and hypocellular bone marrow, congenital heart disease and exocrine pancreatic insufficiency confirmed by very low fecal elastase. Genetic testing identified a heterozygous GATA4 whole-gene deletion.

Case 2: A term boy with birth weight of 2.1 kg, developed hyperglycaemia on day 1 of life. He was born to consanguineous parents with family history of neonatal diabetes. He was dysmorphic with coarse facial features, bulging eyes and jaundice,. Investigations revealed direct hyperbilirubinemia, congenital hypothyroidism and Glucauma. Genetic testing of the previous siblings confirmed the presence of GLIS3 gene mutations.

Both children were treated initially with intravenous insulin then maintained on long acting subcutaneous insulin.

Conclusion

Despite the rarity of Neonatal Diabetes this diagnosis should be considered infants with persistent hyperglycemia requiring insulin therapy. All infants with PNDM need genetic evaluation.