



Proceeding

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Permanent Neonatal Diabetes Mellitus with a Mutation in the *KCNJ11* Gene on Oral Sulfonylurea Treatment

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Introduction

Neonatal diabetes mellitus is considered a rare disease, affecting 1: 500,000 births. It is diagnosed in the first six months of life, with persistent hyperglycemia requiring insulin treatment and can be either transient or permanent. There has been major progress in recent years uncovering the genetic causes of diabetes presenting in the first year of life which can aid in the long-term management of NDM.

Case Presentation

Our patient is now 2 years old; his initial presentation was at the age of 2 months with an acute illness in the form of fever and increase work of breathing. He was found to have hyperglycemia (blood glucose 27mmol/l) with clinical and biochemical evidence of ketoacidosis. He was started on an insulin infusion and then

transitioned to subcutaneous insulin (NPH/regular insulin) after DKA resolution.

He was born at 38 weeks gestation, with a birth weight of 2.3kg. His parents were consanguineous with history of type 2 diabetes in the family. Genetic testing demonstrated a de novo heterozygous missense mutation in *KCNJ11* gene, and we recognise that patients with a gene mutation in the K-ATP channel may respond to oral sulfonylureas. At age of seven months, he was transferred successfully from subcutaneous insulin to oral glibenclamide with good glycemic control and improvement in the HbA1c level.

Conclusion

Genetic testing in neonatal diabetes can alter the management and improve outcomes in these rare patients.



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