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A Challenging Case of Hypoglycemia

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Case Presentation

We present a 6 year old boy who was admitted with episodes of seizure and hypoglycemia. He was born at term to consanguineous parents, with no history of neonatal hypoglycemia or hyperglycemia. His father is diabetic on oral hypoglycemic medication. He was first noted to be hypoglycaemic 9 months ago during a 3 month hospital admission when he required IV dextrose, corticosteroids and octreotide. He was discharged on Octreotide injections, corn starch and Glucagon as needed.

He had 2 further prolonged hospital admissions with hypoglycaemia during which his glucose was difficult to control and he required multiple dextrose boluses and high glucose infusion rate with high dose Octreotide injections and glucagon treatment. Hypoglycaemia screen demonstrated inappropriately high insulin and C-peptide. He was responsive to Diazoxide and once the blood glucose stabilised he was discharged on Diazoxide, Hydrochlorthizide and Glucagon IM injection as needed during hypoglycemia. Abdominal CT and MRI did not show any focal pancreatic lesion. Hyperinsulinemic genetic panel for ABCC8, GLUD1, GCK, KCNJ11 and whole exome sequencing was negative. Confusingly a urine sulfonylurea screen was positive. He had an 18 fluoro-DOPA scan at King Faisal Specialist Hospital & Research Center which showed diffuse uptake throughout the pancreas suggestive of diffuse hyperinsulinism. Further urine for sulfonyurea screen was negative, Genetic testing demonstrated a heterozygous HNF4α mutation (AD). He had a sub-total pancreatectomy and histopathology showed homogeneous lobulated pancreatic parenchyma with no focal lesion identified. Hypoglycemia has resolved following the pancreatectomy.