# Permanent neonatal diabetes mellitus due to a G32S heterozygous mutation in the insulin gene

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**Introduction and objective**: Permanent neonatal diabetes mellitus (PNDM) is a rare form of monogenic diabetes with onset less than 6 months of age. The diabetes mellitus is associated with partial or complete insulin deficiency. PNDM usually occurred atypical and a lot of the patients are treated with insulin injection at the diagnosis. However, we will find a part of them failed to have a succeed glucose control, and even worse leading to a poor clinical outcome. Recent studies have concerned the disease ultimately results from a combination of genetic predisposition and a number of largely unknown environmental factors [1]. The mode of inheritance of PNDM is autosomal dominant for mutation of KCNJ11, autosomal dominant or autosomal recessive for mutation of ABCC8 and INS, and autosomal recessive for mutation of GCK and PDX1 [2, 3, 4]. Herein, we report one case of PNDM.

**Case presentation:** We present a case reported a child diagnosed with PNDM resulting from a mutation in the insulin (INS) gene, leading to severe hyperglycemia and ketoacidosis from 5 month of life. We present a case reported a child diagnosed with PNDM resulting from a mutation in the insulin (INS) gene, leading to severe hyperglycemia and ketoacidosis from 5 month of life. This infant girl was admitted to our hospital with a four-day history of cough, fever and dyspnea. Her routine laboratory analyses revealed normal serum potassium, high levels of sodium, and extremely high glucose levels (381.6 mg/dl), and the blood gases developed metabolic acidosis. Insulin therapy was started with 0.1U/kg/h, and gradually adjusted the insulin dose by the patients glucose levels.

**Results:** Molecular analyses for KCNJ11, ABCC8, GCK, GLIS3 did not show any mutation. We sequenced the INS gene in the proband and her patents, identified the heterozygous missense mutation G32S (c.94G>A, p.Gly32Ser) in the child with diabetes. It has been hypothesized that the mutations in the INS gene disrupt the folding of the proinsulin molecule and result in a bad situation causing endoplasmic reticulum stress and pancreatic beta cell apoptosis.

**Conclusion:** Subjects with this form of diabetes will need lifelong insulin therapy. In conclusion, insulin gene mutations appear to be an important cause of neonatal diabetes.

**Reference**

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