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A Novel Heterozygous INS Gene Mutation as a Cause of Permanent Neonatal Diabetes in an Egyptian Infant

Wafaa Laymoun*

Mansoura University Children's Hospital-Mansoura Faculty of Medicine-Egypt

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***Corresponding author:** Wafaa Laymoun, Mansoura University Children's Hospital-Mansoura Faculty of Medicine-Egypt

Background

Heterozygous missense mutations in the coding region of the INS gene have recently been reported to cause Permanent neonatal diabetes mellitus (PNDM). They act through disruption of the folding of the proinsulin molecule with subsequent accumulation of the misfolded protein in the endoplasmic reticulum (ER) causing ER stress and beta-cell apoptosis.

Objective

To report an Egyptian child with PNDM due to novel *INS* mutation.

Case Report

A 5 month-old boy, born to non-consanguineous parents, was evaluated for hyperglycemia. His weight was 6.5kg (10th centile) and length 63cm (5th centile). The rest of the examination, including the nervous system and fundus was normal. The fasting blood glucose was 200mg/dl and the 2-hour postprandial blood glucose was 385mg/dl and HbA1c was 9.5%. His fasting C-peptide level was 0.1ng/ml (0.9-4ng/ml) and the 2 hrs postprandial was 0.7ng/ml (4.5-16ng/ml). Direct sequencing of the INS gene revealed a novel de novo heterozygous missense mutation (p.Gly69Cys) resulting in a substitution of glycine with cysteine at position 69 and is likely to affect insulin protein folding. The infant was started on 4 units of insulin glargine /day and has been regularly followed up with satisfactory glycemic control.

Conclusion

INS gene mutations can cause isolated PMND and should be assessed in such cases for diagnosis and decision of therapy. To the best of our knowledge, it is the first time to report this mutation in *INS* gene.