Academic Journal of
Pediatrics & Neonatology
ISSN 2474-7521

Juniper Publishers
key to the Researchers

Proceeding
Volume 3 Suppl 1 – March 2017
DOI: 10.19080/AJPN.2017.03.555650

Acad J Ped Neonatol

Thiamine-Responsive Megaloblastic Anemia In An Egyptian Infant: Thiamine Therapy Results In Complete Withdrawal Of Insulin

Yomna Shaban*

Faculty of Medicine, University of Cairo, Egypt

Submission: March 05, 2017**; Published:** March 28, 2017

***Corresponding author:** Yomna Shaban, Faculty of Medicine, University of Cairo, Egypt

Introduction

Thiamine-responsive megaloblastic anemia (TRMA) is a rare condition of anaemia, monogenic diabetes, sensorineural deafness and occasionally cardiac defects. The prevalence of TRMA is unknown but what is known is that fewer than 80 cases have been reported so far most of whom were of Middle or Far-Eastern consanguineous origins.

Objective

To report an Egyptian child with TRMA, in whom the thiamine therapy resulted in complete withdrawal of insulin.

Case Report

A 6-month old Egyptian boy presented with megaloblastic anemia, thrombocytopenia and borderline white cell count. He was subsequently discovered to have diabetes mellitus and sensorineural deafness. The child also had a large atrial septal defect requiring catheter closure. Previous sibling died of the same condition. Thiamine replacement therapy normalized his blood picture and serum glucose levels. Direct sequencing of the *SLC19A2* gene revealed a homozygous nonsense mutation in exon 4 (C 1160 G & gt; A P Trp387* (p.W387*). The patient is currently off insulin for over 3 years with normoglycemia. However he experienced occasional hyperglycemia when missing thiamine doses and when used steroids for his asthma.

Conclusion

TRMA should be considered in patients with the combination of megaloblastic anemia and early onset diabetes and treatment should instituted even without a genetic diagnosis. To our knowledge, only few patients with TRMA became off insulin with thiamine only.