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Homozygosity for Proopiomelanocortin (Pomc) Mutation in a Palestininan Child

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Introduction

Congenital Pro opiomelanocortin deficiency (POMC) is a rare autosomal recessive disorder characterized by adrenal insufficiency, early onset obesity, hyperphagia and altered skin & hair pigmentation. POMC is a complex propeptide encoding a range of melanocortin peptides released by tissue-specific proteolytic processing. These peptides have important roles in a range of functions such as skin pigmentation, control of adrenal growth and functions. In the central nervous system, POMC is most highly expressed in the arcuate nucleus of the hypothalamus, and POMC-expressing neurons are critically involved in the control of appetite and energy balance.

Objective

To describe a Palestinian family with congenital POMC deficiency due to a novel homozygosity mutation in the POMC gene.

Case Report

A Palestinian infant, born to consanguineous parents, presented with early onset obesity, hyperphagia, adrenal insufficiency, and red hair. She had very low ACTH level with undetectable cortisol pre and post ACTH stimulation. Congenital POMC deficiency syndrome was suspected. This diagnosis was confirmed by direct sequencing of the POMC gene which revealed a homozygous c.296delG (p.G99AfsX59) mutation in exon 3 in the child, while her parents were heterozygous for the same mutation. This mutation has been previous reported as a disease causing mutation in a compound heterozygous status.

Conclusion

To our knowledge, this is the first description of this disease in a Palestinian family. This genetic diagnosis will allow accurate genetic counseling and early therapeutic interventions.