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A post-mortem diagnosis of infantile-onset Pompe disease in an Iranian family

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Background

Pompe disease, also called glycogen storage disease type II, is a kind of lysosomal storage disease causes glycogen storage in body cells specially in heart, liver and muscle. The most important features of disease are HCM (hypertrophic cardiomyopathy), respiratory distress and failure to thrive (1). The frequency of Pompe disease is between 1/9000 and 1/40,000. Pompe disease has autosomal recessive inheritance and caused by mutations in GAA gene that codes alpha-glucosidase (α-1,4-glucosidase). The role of this enzyme is degradation of glycogen in lysosome (2). In the present study, we are reporting a case of infantile-onset Pompe disease in an 18 months infant.

Materials and Methods

The case is an 18 months infant with the symptoms of macroglossia and heart disorders. He was clinically diagnosed for Pompe disease. Metabolic enzyme testing was positive for him but negative for his parents. Sometimes later the infant died. The parents' marriage was not consanguineous. The parents blood specimens were collected and DNA extraction was done. Whole exome sequencing (WES) using Illumina HiSeq4000 platform was performed.

Results

The c.896T>C (p. Leu299Pro) and c.2015G>A (p. Arg672Gln) mutations in GAA gene were found in WES analysis in mother and father, respectively. According to American College of Medical Genetics (ACMG), the variants were pathogenic. Both parents were career and the deceased infant was compound heterozygous for GAA gene.

Conclusion

In this case, WES as a powerful diagnostic tool, helped us to confirm the clinical diagnosis of Pompe disease. PND (prenatal diagnosis) for future pregnancies and genetic counseling for other members of family were suggested.

References

• 1. Wang RY. A Newborn Screening, Presymptomatically Identified Infant With Late-Onset Pompe Disease: Case Report, Parental Experience, and Recommendations. Int J Neonatal Screen. 2020;6(1):22.

• 2. Adadi N, Sahli M, Egea G, Ratbi I, Taoudi M, Zniber L, et al. Post-mortem diagnosis of Pompe disease by exome sequencing in a Moroccan family: a case report. J Med Case Rep. 2018;12(1):322.