Central European Annals of Clinical Research

Endoped Abstract

Hypoglycemia in an Infant: Case Report

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Submitted: 26 October 2020, accepted: 14 November 2020, published: 17 November 2020

Keywords: diazoxide; hypoglycemia; infant; jerks

How to cite: Domșa, D.-N.; Chiriță-Emandi, A.; Bugi, A.-M.; Mărginean, O.; Mărăzan, M. Hypoglycemia in an Infant: Case Report. *Cent. Eur. Ann. Clin. Res.* **2020**, *2*(1), 49; doi:10.35995/ceacr2010049.

Background and Aims

Hypoglycemia in infants may be attributed to several causes, such as: hyperinsulinism, glycogen storage disorders, fatty acid disorders, metabolic defects and hormonal deficiencies [1].

The aim of this paper is to highlight the importance of early diagnosis of hypoglycemia in order to prevent motor and cognitive impairment.

Material and Method

In this study, we present the case of a 13-month-old male patient. Clinical, biological and imaging data were evaluated.

Results

The 13-month-old male infant had the following traits: normal weight and length for age; non-significant family history; full-term birth; large for gestational age (LGA); presents at four months of age; jerks spasms of the upper limbs; unremarkable EEG; no abnormal structural changes showed by cranial sonography. In the following months of his life, delayed motor development was observed. The investigations showed a low level of plasma pyruvate, hypoglycemia (21 mg/dL), HbA1c 3.97%, insulin levels (4.76 μUI/mL), cortisol, thyroid hormones and total cholesterol values in the normal range, with inconsistently elevated levels of triglycerides. The suspicion of an inborn error of metabolism was raised at the age of 11 months and genetic testing (exome sequencing) was performed [2]. The genetic test showed a variant (variant c.87C > A; p. (Asp29Glu)) with unknown significance in the ABCC8 gene [3,4]. A diversified diet was established, six meals/day with constant carbohydrate content, without an improvement in glycemic values. Although this is a non-actionable genotype finding, correlated with clinical and biological data, we decided to initiate therapy

with diazoxide [4,5]. The starting dose of diazoxide was 7.5 mg/kgc/day/t.i.d, associated with a diversified diet consisting of six meals/day. After 24 h of treatment, a normalization of glycemic values was observed (average glycemia/24 h = 93 mg/dL). After hospital discharge, a recurrence of hypoglycemic episodes was noted. The dose of diazoxide was increased to 10 mg/kg/day/t.i.d, and there was thus an improvement of the glycemic values.

Conclusions

In some situations, a genomic result is not enough to confirm the diagnosis, however, in correlation with clinical and other biological data, it can lead to an accurate diagnosis.

Funding: This research received no external funding.

Conflicts of Interest: The authors declare no conflict of interest.

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