Extended Abstract | Acute and Chronic Disease Reports | 2019 | Vol.3 No.1

Rare Diseases Congress 2019: Clinical case of congenital hyperinsulinism in infant born by mother with type 2 diabetes - N B Belykh - Omsk State Medical University

N B Belykh

Omsk State Medical University, Russia

Relevance: Congenital hyperinsulinism (CHI) is a rare hereditary disease characterized by insulin hyper secretion and severe persistent hypoglycaemia in children.

Aim: The aim of the study is to present a clinical case of CHI in a child born of mother with type 2 diabetes.

Patients & **Methods:** Analysis of the clinical case and medical documentation. Congenital hyperinsulinism (CHI) is that the leading explanation for persistent hypoglycaemia in infants, causing neurodevelopmental and cognitive delays in up to 25–50% of affected children. Early recognition and appropriate treatment are important to stop such adverse outcomes. CHI is caused by genetic defects that cause disease through changes in channels expression/activity, transcription factors expression and disturbed enzyme activities. All these defects cause the common finding of recurrent episodes of hyperinsulinemic hypoglycaemia thanks to an inappropriate secretion of insulin by the pancreatic beta-cells. Channels expression activity transcription factors expression and disturbed enzyme activities which comprise two subunits of KATP-HI, the beta-cell plasma dependent membrane ATP-potassium channel. Most pathogenic variants in these 2 genes are During his hospital stay, the GIR was gradually increased to 16 mg/kg/min recessive and cause loss of function; however, some cases of dominantly inherited inactivating mutations have been reported. In the past, clinical phenotype of patients However, on DOL 21, he developed hypoglycaemic episodes (BG below 50 mg/dL) after IV dextrose was weaned off and despite consuming 26 cal/oz. of formula with dominant variants was different from those with recessive variants but the biochemical phenotype was not different. Polycose supplement was added to his feeds with the goal of maintaining plasma glucose levels on the brink of 70 mg/dL when titrating IV dextrose. Extensive workup for CHI was initiated on DOL 25, and therefore the results revealed an elevated insulin level of 16.3mica/mL coincident with a plasma glucose level of 48 mg/dL and beta hydroxybutyrate level of but 100memo/L. This variant has been previously reported during a family where it segregated with several affected individuals with CHI. It is rare in population databases Congenital hyperinsulinism (CHI) is the leading cause of persistent hypoglycaemia in infants. The infants of diabetic mothers (IDMs) very frequently present with neonatal hypoglycaemia associated to transient hyperinsulinism however the incidence of CHI in IDMs is unknown and a special clinical phenotype for HNF4A-associated CHI. IDMs typically present with transient hyperinsulinism lasting no quite 2–3 days. Since being an IDM doesn't exclude CHI, this diagnosis should be considered because the mostly likely ethology if neonatal hypoglycaemia persists longer than the described time-frame and genetic testing for CHI confirmation is very suggested.

Results: The girl from the 3rd pregnancy proceeding against the background of type 2 diabetes, 3 preterm births (35-36 weeks), weigh is 3410 g. After birth, the glycaemia was 0.1 moll/l and then stabilized (5.0-4.3 moll/l). In the first year of life glycaemia was in the range of 3.0-4.0 moll/l, the neurodevelopment corresponded to the age. At 11 months of age, the level of insulin was 17.4? E/ml. At the age of one year on the background of a long hungry pause glycaemia was 1.6 moll/l; the child became lethargic, convulsions were noted. The girl was urgently hospitalized in the hospital with suspected CHI. The diagnosis was confirmed in the National Research Centre for Endocrinology, where during the examination the glycaemia was 2.7 moll/l, insulin-3.78 ?E/ml, C-peptide-0.731 ng/ml. On the background of dioxide intake in a dose of 5.6 mg/kg/day after a hungry period of 11.5 hours, glycaemia-2.9 moll/l, ketonemia-1.1 moll/l, insulin-1.56?E/ml. To clarify the variant of the disease is carried out molecular genetic study. The child was prescribed dioxide therapy (5.6 mg/kg/day), against which persistent glycaemia and adequate insulin suppression are achieved. The girl is currently under the supervision of a paediatrician and endocrinologist at the place of residence. The tolerability of the therapy is satisfactory. Glycaemia rates correspond to the norm; the child does not lag behind his peers in neurodevelopment.

Conclusion: The disease manifested itself as a hypoglycaemic state on the 2nd day, but later, due to the absence of signs of hypoglycaemia, the condition was regarded as transient. The manifestation at the age of one year required an in-depth examination of the child, during which the CHI was diagnosed.

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This work is partly presented at 9th World Congress on Rare Diseases and Orphan Drugs June on 17-18, 2019 held at Berlin, Germany