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CONGENITAL HYPERINSULINISM: FIRST CASE REPORTS FROM THE REPUBLIC OF ARMENIA.

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Abstract

Congenital hyperinsulinism is a rare disorder of insulin-producingbeta-cells affecting 1 in 50,000 newborns. Individuals with congenital hyperinsulinism have frequent episodes of hypoglycemia due to inappropritate plasma insulin levels for the blood glucose level. Some forms of congenital hyperinsulinism-resolve spontaneously and are transient, whereas others persist for longer and are considered permanent. The main risk groups for developing transienthyperinsulinaemic hypoglycemia aresmall for gestational age newborns, those subjected to hypoxic fetal distress and those born to mothers with diabetes mellitus.

At least 14 further genes have to harbor causative mutations in individuals with congenital hyperinsulinism, including the Glucokinase gene which encodes the "glucose sensor" for the beta cells. These dominantly acting mutations increase the enzymatic activity of Glucokinase gene and often arise spontaneously.

The clinical severity of congenital hyperinsulinism varies widely among patients, even among the individuals with the same genetic aetiology. Not all children with congenital hyperinsulinism experience hypoglycemia right after birth or during the first month of life (60-65%). Almost 30% of affected individuals develop hypoglycemic episodes during infancy. The main clinical symptoms are irritability, sleepiness, lethargy, hunger and tachycardia, as wellas weakness, tiredness, confusion, tachycardia and aggressive behaviours. More severe symptoms, such as seizure and coma, occur after prolonged hypoglycemia or due to a rapid drop of blood glucose level. Recurrent moderate hypoglycaemias, as well as severe hypoglycemic episodes may cause brain injury and cognitive impairment in the longterm.With early treatment and careful prevention of hypoglycemic episodes the brain damage can be prevented.

There are no previous reports of congenital hyperinsulinism from Armenia.In this manuscript we describe 3 cases of congenital hyperinsulinism from Armenia who presented with different clinical phenotypes and report the results of genetic testing. Current cases are the first reports from Armenia, and can serve as a base for increase awareness of congenital hyperinsulinism in the relevant country.

Keywords: *congenital hyperinsulinism, hypoglycemia, glucokinase mutation, diazoxide*

Introduction

Presentation of cases:

Case 1. A 5-month-old girl was referred to the Endocrinology department of "Muratsan" University Hospital with a diagnosis of epilepsy and underwent an EEG and a brain MRI scan and started on anti-

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