P2-487

Congenital hyperinsulinism in Ukraine

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Abstract

Congenital hyperinsulinism (CHI) has not been studied in the Ukraine.

Objective and hypotheses

We investigated the genetic aetiology and treatment of patients with CHI.

Methods

Routine clinical and laboratory investigations were performed in children with hypoglycemia.
Genetic testing was undertaken for 7 patients with CHI (77,8%) from 9 families. *KCNJ11, ABCC8, HNF4A* genes were sequenced in all patients. For those who were negative in the initial screening, were also tested for *GLUD1* gene, as well as next generation sequencing of all known CHI genes was performed. ^{18}F-DOPA PET-CT (and ^{68}Ga-DOTA PET-CT) scans were performed in selected cases.

Results

In 7 patients hypoglycaemia (glucose 0,8 [0,5; 1,2] mmol/l) with detectable insulin (43,1 [1,2; 45,9] mIU/l) and/or C-peptide (6,9 [1,1; 9,9] ng/ml) confirmed CHI. The median age at diagnosis was 55,4 [1,0; 330] days and the median birth weight was 4078 [2850; 5200] g. The incidence of CHI in the Ukraine was calculated at 1 in 258,650 births.

Table 1. Genetic causes of CHI.

Patient	Gene	Protein change	Inheritance		Treatment	Histological form
			Mother	Father		
1	*ABCC8*	p.Q444H		p.Q444H	Surgical	Focal
2	*ABCC8*	p.?		c.4415-13G>A	Surgical	Focal
3	*ABCC8*	p.Q444H p.Q923X	p.Q923X	p.Q444H	Medication	
4	*ABCC8*	p.R1437X		p.R1437X	Surgical	Focal
5	Not found				Surgical	Atypical
6	*KCNJ11*	p.F333S		p.F333S	Surgical	Focal
7	*ABCC8*	p.R1251X p.Y1287X	p.Y1287X	p.R1251X	Surgical	Diffuse

Postoperative complications included transient fasting hyperglycemia (1), cicatricial hernia development after convulsions (1), persistent subclinical exocrine insufficiency (1), and relapse of CHI (in patient with atypical form).

Conclusions

Children with hypoglycemia and unsuppressed insulin and C-peptide levels should undergo genetic and eventual PET CT scan for characterization of the type of CHI. Further studies to identify novel CHI genes are required.

ESPE 2015
487--P2
Hypoglycaemia
Globa Eugenia
DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology