Difficulties in hypothyroidism and diabetes treatment in patient with GATA6 gene mutation –case report

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Introduction

Patients with GATA6 gene mutations have broad spectrum of clinical presentation, but most of them have pancreatic agenesis or hypotrophy, exocrine pancreatic insufficiency, insulin-treated neonatal diabetes and cardiac malformations. Some of them have significant neurocognitive deficits, hypopituitarism, hypothyroidism, gut abnormalities, biliary atresia, gallbladder agenesis. There are about over 50 cases described worldwide

Patient's characteristic

The 5.5 year old female patient with mutation R493X in GATA6 gene is followed up in our Pediatric Diabetology Outpatient Clinic since she was diagnosed with diabetes in second week of life

Clinical phenotype

- Pancreatic hypotrophy,
- insulin dependent diabetes since second week of life,
- exocrine pancreatic insufficiency required enzyme supplementation, meconium ileus, constipations
- tetralogy of Fallot requiring surgery in second year of life, corection DORV and PS; PI (+++)
- severe hypothyroidism, since she was born
- psychomotor delay
- Left hemiparesis

Anthropometry and development

Age (years)	37 Hbd	0,5	1	2,16	2,9	4	5,5
Length/ Height (cm)	50 +0,5SD	62 z-score -3,38	72 z-score- 2,58	89 z-score 0,07	96 Z-score 0,46	99 Z-score -1,48	101 z-score -2,72
Weight (kg)	1,5 <-3SD	5,5 Z-score - 3,5	8,7 z-score -1,77	13,5 z-score -0,17	15,6 z-score 0,64	13,8 z-score -1,6	16,9 Z-score -1,18
Psychomotor development	In range	Left hemiparesis	Left hemiparesis psychomotor delay	Sits without support, left hemiparesis	Sits without support, left hemiparesis	Left hemiparesis walks without support	Left hemiparesis walks without support

Diabetes and thyroid treatment

Age	15 days	0,5	1	2,16	2,9	4	5,5
DDI j/kg	0,75; infusion pump iv	1 CSII	0,7 CSII	0,6 CSII	0,5 CSII	0,44 MDI	0,5 MDI
Sample daily glucose profile mmol/l	3,1-17,3	3,1-13,9	11,1-22,2	2,8-19,4	2,7-25	2,7-17,8	3,3-21,7
HbA1c %	-	7	11,1	9,3	10	8,7	9,6
L-tyroxine ug/kg	8	7,3	5	3,2	3,2; 2 weeks during hospitalization 4,8	3,4	6
TSH uU/ml	4	7,3	0,012	429,4	>1000 3 weeks after hospitalization 1,29	30,86	0,6
fT4 pmol/l	14,2	13,08	7,48	<5,15	<5,15; 3 weeks after hospitalization 22,1	12,75	14,13

Other laboratory tests

antibodies p/TGA negative
APTT 44 elevated ; INR 1,05 and PT 93- in range
Serum osmolality 287 mOsm/kg H20 in range
IGF-1 15,47 ng/ml below limit
ACTH 34,1 pg/ml in range
Prolaktyna 1821,76 mU/l (with TSH 1800) -elevated
cortisol 371 nmol/l -in range
C-peptyd 0,1 ng/ml -below limit
25OHD 18,8 ng/ml below limit

Diagnostic Imaging

Abdominal ultrasoud examination
- Hepatomegaly; 117mm;
- Pancreas was revealed only fragmentarily
- Gas retention in the intestines; retention of fecal masses in rectum

Thyroid ultrasound examination
Thyroid was not revealed in typical places
Lung scintigraphy
Left pulmonary field - no perfusion
Right pulmonary field -weaker perfusion in upper lung lobe
MRI of hypophisis -mother didn't agree to the examination

Conclusions

Difficulties in treatment in the patient could result from
- ✓ clinical presentation of GATA6 mutation
- ➢ problems with cooperation with patient's parents.

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