Child with mutation in GATA 6 gene – case report

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BACKGROUND

The GATA family of zinc finger transcription factors including GATA4 and GATA6 are known to play an important role in the development of the pancreas.

There is evidence of the essential function of GATA6 in the development of the human pancreas.

AIM

The aim of this case-report study is to present a patient with GATA6 mutation treated in Clinic of Pediatric, Diabetology and Endocrinology, Medical University of Gdansk.

CASE REPORT

- Child was born prematurely in 36th week of pregnancy
- Birth weight of 1520 gram, with breathing problems supported by mechanical ventilation
- Echocardiography, detected tetralogy of Fallot
- From third day of life, hyperglycemia was noticed and intravenous insulin infusion started
- Because of meconium obstruction in the newborn, cystic fibrosis was suspected
- In neonatal screening, congenital hypothyroidism was diagnosed and thyroxin was prescribed
- Genetic test did not confirm CF (mutation on one allel CFTR gene – F508del) and Hirschsprung disease was suspected. In genetic test, reciprocal translocation between chromosome 1 and 7, and pericentric inversion of chromosome 9 was found

- Child received thyroid hormons, insulin, pancreatic enzymes, antispasmodic agents, ursodeoxycholic acid, rectal ingots and vitamins. In first month of life, insulin was injected intravenously and when child reached a weight of approximately 2,5 kilogram, insulin pump therapy was started with insulin dose of 0,8 U/kg/day.

On the basis of clinical picture, mutation in GATA 6 was suspected. In genetic laboratory of Medical University of Łódź, genetic tests were performed and GATA 6 mutation was detected.
Metabolic control of diabetes is poor.

Treatment	
L-thyroxin	50 ug
insulin	0,8 U/kg/d
vitamins	+
Pancreatic enzymes	Before meals
Antispasmodic agents	+

The child was qualified to cardiosurgical correction of cardiac heart defect which was performed with good outcomes.

CONCLUSION

Persistent neonatal diabetes with exocrine insufficiency of pancreas and heart defect in patient were caused by GATA6 gene mutation.

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55ESPE 281--P2

Diabetes

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DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS 10-12 SEPTEMBER

PosterSessionOnline.com