

# Neonatal diabetes as a first symptom of IPEX syndrom

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## BACKGROUND

- ❖ Immunodysregulation polyendocrinopathy enteropathy x-linked syndrome (IPEX) is characterized by systemic autoimmunity, typically beginning in the first year of life.
- ❖ Presentation is most commonly the clinical triad of diarrhea, dermatitis, and endocrinopathy.

## CASE REPORT

- ❖ Male patient from 1st pregnancy, born with body weight 3840 g, Apgar 10 points
- ❖ In 13th day of life vomiting and tachypnoe were noted and in laboratory tests hyperglycemia 653mg/dl and ketoacidosis were reported. Patient was diagnosed with diabetes and treatment with insulin pump was started. Antibodies typical for diabetes type 1 were negative.
- ❖ In 9th month of life boy was hospitalized in Clinic of Pediatrics, Diabetology and Endocrinology in Gdansk, mutation in KCNJ 11 was excluded and autoimmune thyroiditis was diagnosed and L-tyroxin treatment was started.
- ❖ In 12th month of life patient was diagnosed with nephrotic syndrome resistant to steroids. Patient had also periodical skin lesions and diarrhea.
- ❖ According to clinical presentation IPEX syndrome was suspected, T regulatory cells level was normal, sample for genetic test for FOXP3 mutations was taken.
- ❖ After 1 month patient started seizures, in MRI scans there were not seen any changes, but anti-neuronal antibodies (ABA) were highly positive.
- ❖ Also anti-tissue transglutaminase antibodies were positive and gluten free diet was started.
- ❖ In molecular tests performer in Department of Clicial Genetic in Łódź mutation in FOXP3 was found.
- ❖ Patient had started immunosupresion and had bone marrow transpalantation performed in Medical University in Wroclaw at the age of 2 years.

## CONCLUSION

Neonatal diabetes in boys can be first symptom of IPEX syndrome

