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Objective and hypotheses

In this study we investigated patients with onset of neonatal diabetes (ND) mellitus during the first 9 months of life.

Methods

We studied 48 probands with permanent or transient ND diagnosed within the first 6 months of life (n=27) and with permanent ND diagnosed between 6 and 9 months of age (n=21).

KCNJ11, *INS* and *ABCC8* genes were sequenced in all patients. For those who were negative in the initial screening, we also tested for 6q24 and used tNGS of all known ND genes in any child diagnosed in the first 6 months of life.

Conclusions

1. Every child with diabetes mellitus onset before 9 months should undergo genetic testing for ND.
2. tNGS technology increased the number of patients with a confirmed genetic etiology of ND.

References

E. Globa, N. Zelinska, D.J.G. Mackay, Karen I. Temple, et al. Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. *J. Pediatr Endocrinol Metab.* – 2015. – Nov; 28 (11-12). – P. 1279-86.

Results

Due to the Ukraine Pediatric Diabetes Register the number of children with DM1 0-18 y.o. in 2015 was 8388 (a prevalence of 1 in 907), with DM2 – 36 (1 in 211 519), with ND - 52 reported cases (1 in 146 436).

We determined the genetic etiology in 28 of 48 (58,3%) probands diagnosed with diabetes before 9 months: in 88.9% of those diagnosed before 6 months and in 19% diagnosed between 6-9 months. K_{ATP} channel mutations were the commonest cause of ND accounting for 50% of cases. All of these patients transferred from insulin to sulfonylureas (SU). After 1 year of SU treatment all had a HbA1c level <6.5%, p=0.01.

Fig 1. Genetic causes of ND in Ukraine

