Case of family neonatal diabetes with \textit{KCNJ11} gene mutation: dynamics monitoring

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Background: Neonatal diabetes is a rare pathology occurring in around 1 in every 200,000 - 400,000 live births. The most common cause of permanent neonatal diabetes (PNDM) is heterozygous activating mutations in the \textit{KCNJ11} gene encoding the pore-forming Kir6.2 subunit of the pancreatic beta cell KATP channel.

\section*{CLINICAL CASE}

\textbf{Method:} We studied a family (mother and child) with PNDM diagnosed within the first 6 months of life. Carbohydrate metabolism was studied by iPro-2 monitoring, HbA1c, C-peptide and insulin levels during 8 months of SU therapy. The \textit{KCNJ11} gene was sequenced by Sanger.

\textbf{Objective and hypotheses:} To determine the dynamic of carbohydrate metabolism in family transferred from insulin to sulphonylureas (SU).

\textbf{Results:} The mutation in \textit{KCNJ11}, R201H was identified in the child and the mother at the age of 2 months and 28 years. Insulin has been canceled. At the beginning of treatment, the child’s daily dose of SU was divided into 6 doses (0.27 mg / kg / day) with each feeding, but at the age of 10 months, the frequency of taking the drug was 4 doses / day (0.17 mg / kg / day). After 8 months of SU treatment, an improvement in glycemic control was observed (HbA1c level decreased 5.15% versus 13.9%). The level of C-peptide increased from 0.09 ng/ml to 0.5 ng/ml after 8 months of treatment of SU. Daily monitoring of glycemia showed a noticeable decrease in fluctuations in glycemia and improved glycemic control (from 13.8 [2.6-26.6] mmol / l before treatment with SU to 6.0 [3.3-10.2] mmol / l - after). After 5 years of monitoring, the child grew and developed according to age, taking SU twice a day (1 mg / s -0.05 mg / kg / day). HbA1c level - 5.9%, C-peptide 0.41 ng / ml. The average rates of glycemic fluctuations per day were [4.8 [8.6-3.8] mmol / l]). But after 6 years the dose of the drug increased again to 1.5 mg / day of glibenclamide.

\section*{Conclusion:} with the manifestation of diabetes mellitus during the first 6 months of life, the patient after genetic testing shows the pathogenetic treatment of SU. The daily dose of SU in a child over the course of 5 years of observation decreased on average by 40% from the initial dose due to the stabilization of carbohydrate metabolism. However, a decrease in the level of C-peptide by 20% from the initial one was also noted. However, when trying to reduce the dose of the drug, the compensation for carbohydrate metabolism worsened (HbA1c = 10.29%) and the dose again became the same (1.5 mg / day). Further observation required.