Abstract: Approximately 1-2% of diabetes mellitus cases are caused by single gene mutations with autosomal dominant inheritance. These cases are classified as monogenic diabetes. Diabetes diagnosed within the first 6 months of life is defined as neonatal diabetes and belongs to the group of monogenic diabetes. Permanent neonatal diabetes has been studied intensively, leading to the unraveling of the genetic mutations leading to this disease phenotype. Elucidation of the molecular mechanism of permanent neonatal diabetes led to a spectacular change in the treatment of these children who can be switched from insulin to sulphonylurea treatment.

Keywords: neonatal diabetes, KCJN11, ABCC8, insulin gene, sulphonylurea treatment