10. GLUTS GENETIC DEFECTS AS CAUSES OF DIABETES

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Introduction. Diabetes mellitus (DM) is a chronic disease marked by insulin secretion or action defects. The cause of diabetes can be genetic defects of glucose transporters – GLUT, the most characteristic being the GLUT2 defect, associated with Fanconi-Bickel syndrome (FBS). Until now approximately 144 cases of FBS with 70 different variants of the SLC2A2 gene have been reported.

Aim of study. The purpose of the study is to elucidate and describe the consequence GLUT dysfunction and the occurrence of diabetes.

Methods and materials. PubMed, Hinari, GoogleScholar. Published between 2018-2023. Key-words: „GLUT", „Diabetes".

Results. Deficiency or absence of GLUT-encoding genes has been intensively studied in mice, then making the tangent to deficits in humans. In humans, SLC2A2 gene defects are the cause of FBS. Those with this condition display a phenotype mirroring that of mice lacking the SLC2A2 gene. Mainly expressed in tissues that play important roles in glucose homeostasis: renal tubular cells, enterocytes, pancreatic β-cells, hepatocytes. Dysfunction and decreased GLUT2 expression lead to dysglycemia (fasting hypoglycemia, postprandial hyperglycemia, glucose intolerance), hepatomegaly, galactose intolerance. The cause of DM in these patients would be the GLUT2 defect in: 1) pancreas - insulin secretion deregulation; 2) liver - disturbances in glucose homeostasis, accumulation of fat and glycogen in the liver, problems in glucose synthesis.

Conclusion. DM is a chronic multifactorial disease that requires a vast study to identify the primordial cause of the disease. The primordial cause can be hidden in the human genetic code, and the treatment is closely related to it. Future GLUTs studies will be required for a better understanding of underlying molecular mechanisms of dysglycaemia in FBS and other GLUTs deficiencies.