Variability of ribosomal cistron activity in type 2 diabetes

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Abstract

Type 2 diabetes mellitus (T2D) is a polygenic metabolic disorder, the risk of which is determined by a combination of genetic and environmental factors. The heritability is estimated at approximately 30–70%. Chromosomal abnormalities are rare in T2D; however, several microdeletions or duplications affecting genes involved in pancreatic development or determining β-cell function have been described as factors increasing disease risk. For example, structural alterations in the 15q region and the 11p15 locus have, in some cases, been associated with insulin resistance and early-onset diabetes manifestation. Although such chromosomal anomalies are not detected in the majority of T2D cases, their identification is important for differentiating rare forms and for genetic counseling.

Despite extensive investigation of genetic markers, the role of chromosomal structural changes in the pathogenesis of T2D remains incompletely defined. Cytogenetic analysis, including the detailed study of microdeletions and duplications, may become an important tool for identifying rare and diagnostically challenging forms of the disease. Particular importance is given to the study of nucleolus organizer region (NOR) activity, as it is associated with the intensity of protein synthesis and the regulation of cellular metabolism, which may in turn influence insulin production and glucose homeostasis. Research in this direction will not only deepen the understanding of the molecular basis of the disease but also create the groundwork for planning personalized preventive and therapeutic approaches.

Based on the above, the aim of the present work was to investigate genomic parameters and the activity of nucleolus organizer regions in lymphocyte cultures from individuals with type 2 diabetes. Analysis of the results demonstrated that the activity of ribosomal cistrons varies depending on the chromosome, which may reflect alterations in the regulation of cellular metabolism and protein synthesis characteristic of this pathology.