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# Molecular study of intron 2 of calreticulin gene (CALR) in type-2 diabetic patients

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Type 2 diabetes mellitus (T2DM) is a complex polygenic disease. Genetic factors play major role in the pathogenesis of T2DM. Calreticulin is a 46 kD $Ca^{2+}$ binding protein and ER chaperon. Calreticulin (CALR) was found in high concentration in pancreas. Genomic analysis and detection of variants related to type-2 diabetes can help to determination T2DM pathophysiology and its familial pattern of inheritance. Imbalance in $Ca^{2+}$ concentration and dysfunction of the chaperone system are speculated to be linked with type-2 diabetes mellitus (T2DM). Two-dimensional protein profiling of pancreatic beta cells in T2DM subjects has shown that the Ca2+ binding chaperone, calreticulin (CALR), plays a role in the pathophysiology of this disease. In a case/control study design, we performed mutation screening of the promoter region, 9 exons and exon/intron boundaries of CALR by PCR-SSCP and sequencing in 120 patients afflicted with T2DM and 530 controls. Two novel mutations were detected in T2DM patients, which were absent in the control gene pool (Mid P exact <0.01). The first mutation was a G>T transversion in intron 2 conserved polypurine tract at IVSII-142. The second mutation was a 9-bp deletion in the highly conserved exon 9 encompassing amino acids 402-404. Exon 9 encodes the low affinity, high capacity $Ca^{2+}$ binding domain of CALR. This case is the first instance of a microdeletion in a gene coding sequence reported in T2DM. To our knowledge, the current study reports for the first time, CALR gene mutations that co-occur with T2DM.

## Biography

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