Cardiovascular Diabetology
Volume ٨, ١٠ December ٢٠٠٩, Article number٦٣

Mutations in the coding regions of the hepatocyte nuclear factor ٤ alpha in Iranian families with maturity onset diabetes of the young

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Abstract

Background: Hepatocyte nuclear factor ٤α (HNF٤α) is a nuclear receptor involved in glucose homeostasis and is required for normal β cell function. Mutations in the HNF٤α gene are associated with maturity onset diabetes of the young type ١ (MODY١). The aim of the present study was to determine the prevalence and nature of mutations in HNF٤α gene in Iranian patients with a clinical diagnosis of MODY and their family members. Twelve families including ٣٠ patients with clinically MODY diagnosis and ٢١ members of their family were examined using PCR-RFLP method and in case of mutation confirmed by sequencing techniques. Fifty age and sex matched subjects with normal fasting blood sugar (FBS) and Glucose tolerance test (GTT) were constituted the control group and investigated in the similar pattern. Single mutation of V٢٥٥M in the HNF٤α gene was detected. This known mutation was found in ٨ of ٣٠ patients and ٣ of ٢١ individuals in relatives. Fifty healthy control subjects did not show any mutation. Here, it is indicated that the prevalence of HNF٤α mutation among Iranian patients with clinical MODY is considerable. This mutation was present in ٢٦,٦٪ of our patients, but nothing was found in control group. In the family members, ٣ subjects with the age of ≤٢٥ years old carried this mutation. Therefore, holding this mutation in this range of age could be a predisposing factor for developing diabetes in future. © ٢٠٠٩ Taghavi et al; licensee BioMed Central Ltd.

Reaxys Database Information

Indexed Keywords

EMTREE drug terms: hepatocyte nuclear factor ٤; HNF٤A protein, human; glucose; hepatocyte nuclear factor ٤alpha

EMTREE medical terms: adolescent; adult; article; Asian; blood; case control study; ethnology; female; genetic predisposition; genetics; glucose blood level; glucose tolerance test; human; Iran; male; mutation; non insulin dependent diabetes mellitus; nucleotide sequence; onset age; open reading frame; pedigree; phenotype; polymerase chain reaction; risk factor; clinical article; controlled study; family; gene mutation; gene sequence; maturity onset diabetes mellitus; pathogenesis; prevalence; restriction fragment length polymorphism

MeSH: Adolescent; Adult; Age of Onset; Asian Continental Ancestry Group; Blood Glucose; Case-Control Studies; Diabetes Mellitus, Type ٢; DNA Mutational Analysis; Female; Genetic Predisposition to Disease; Glucose Tolerance Test; Hepatocyte Nuclear Factor ٤; Humans; Iran; Male; Mutation; Open Reading Frames; Pedigree; Phenotype; Polymerase Chain Reaction; Risk Factors; Young Adult
Medline is the source for the MeSH terms of this document.

Chemicals and CAS Registry Numbers: Blood Glucose; HNF٤A protein, human; Hepatocyte Nuclear Factor ٤;glucose, ٥٠-٩٩-٧, ٨٤٧٧٨-٦٤-٣