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GCK mutations in Chinese MODY2 patients: A family pedigree report and review of Chinese literature

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Background: Maturity-onset diabetes of the young type 2 (MODY2) is caused by mutations in the glucokinase (GCK) gene and is rare in the Chinese population. We report three Chinese families with MODY2 and the sequencing of the GCK gene.

Methods: Three unrelated Chinese families with MODY2 and their pedigrees were investigated. In Family 1, the proband was a seven year old girl with impaired fasting glucose (IFG) and impaired glucose tolerance (IGT). Her mother and maternal grandfather had IFG. In Family 2, the proband was a boy who had diabetes mellitus at 11 years. His sister had IFG. His father and grandmother had diabetes mellitus at 22 and 25 years, respectively. In Family 3, the proband was a boy who had IFG and IGT at 12 years. His sister had diabetes mellitus at 8 years. His father and grandfather had IFG and/or IGT. The GCK gene was directly sequenced.

Results: Diabetes mellitus or IFG/IGT was found among three consecutive generations in three families. One novel nonsense heterozygous mutation in exon 5 (c.556 C>T, p.Arg 186 stop) was detected in Family 1. Another novel frameshift mutation in exon 4 (c.367-374dupTTCGACTA, p.Ile 126 fs) was found in Family 2. A previously reported, a missense heterozygous mutation in exon 5 (c.571 C>T, p.Arg 191Trp) was detected in Family 3.

Biography

Yan Lan Fang is the Associate Professor and Associated Chief Physician of Dept., of Pediatrics in the First Affiliated Hospital Zhejiang University. She is the Member of the Ethics and Doctor-Patient Communication Professional Committee of Chinese Medical Doctor Association of Neonatologist branch. She was awarded Second Prize in Zhejiang Provincial Science and Technology Award. She is engaged in clinical, teaching and scientific research work more than 10 years. Her research interest is in molecular and genetic mechanism of pediatric endocrinology and metabolic diseases, such as thyroid binding globulin deficiency, children obesity, diabetes mellitus and disorder of sex differentiation, supported by grants from The Medical Science and Technology Program of Zhejiang province. She has published 2 SCI indexed papers in international journal as the first or corresponding author.

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