

# 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), caused by mutations in the glucokinase (GCK) gene is rare in a Chinese population.

## Objective and hypotheses:

We report three Chinese families with MODY2 and sequenced the GCK gene to find novo mutation.

## Method:

Three unrelated Chinese families with MODY2 and 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 old. His sister had IFG. His father and grandmother had diabetes mellitus at 22 years old and 25 years old, respectively. In Family 3, the proband was a boy who had IFG and IGT at 12 years old. His sister had diabetes mellitus at eight years old. 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 missense heterozygous mutation in exon 5 (c.571 C>T, p.Arg 191Trp) was detected in Family 3.

## Conclusion:

A thorough investigation of the three Chinese families with MODY2 revealed two novel mutations and one known mutation. GCK gene sequencing helps in MODY2, especially when there is uncertain IFG or IGT.

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