Cases of Maturity onset diabetes of the young (MODY)

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Introduction: Maturity-Onset Diabetes of the Young (MODY) refers to a collection of inherited disorders characterized by non-autoimmune diabetes mellitus that develops at a young age. Here, we present two cases of MODY, specifically MODY1 (HNF4a) and MODY2 (GCK). Notably, we have identified a novel mutation in the GCK gene, which has not been previously reported.

Case report: Case 1: MODY 2 with a novel mutation The first case involves a 21-year-old black female with no significant medical history, who visited an outpatient clinic for a health screening conducted by the US Army. Her HbA1c level was found to be 6.2%. Her father had a history of diabetes, while her mother did not. The patient had a body mass index of 19.33 kg/m^2 and had not experienced recent weight changes. Initial laboratory tests revealed a glucose level of 113 mg/dL and an HbA1c of 6.4%. The C-peptide level was 1.7 ng/ml. Genetic testing was performed and revealed a c.74T>C p.Leu25Pro mutation in the GCK gene, specific to the MODY subtype. This mutation was determined to be likely pathogenic. Importantly, this mutation had never been reported previously and represents the first identification of its kind in relation to GCK-MODY. Case 2: MODY 1 with diabetic complications The second case involves a 64-year-old male who was diagnosed with type 2 diabetes at the age of 20. His father, mother, and brother were all diagnosed with diabetes, and his son was diagnosed at the age of 26. The patient had a normal body mass index of 21.8 kg/m^2. Laboratory tests revealed a glucose level of 93 mg/dL, an HbA1c of 7.1%, and a C-peptide level of 1.7 ng/ml. GAD Ab was negative. Genetic testing revealed a heterozygous c.200G>A p.Arg67Gln mutation in the HNF4A- gene, which was determined to be likely pathogenic. He had mild nonproliferative diabetic retinopathy and the urine microalbumin/creatinine ratio was 45mg/g.

Discussion: The GCK MODY case described here highlights the discovery of a novel mutation that functions as a causal gene. Genetic testing plays a crucial role in accurately diagnosing atypical cases of diabetes.

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Pathogenic/Likely pathogenic variant: Detected*
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- Observed Genetic Variants
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Gene cDNA change AA change Zygosity Variant Category

GCK c.74T>C p.Leu25Pro Heterozygous Likely Pathogenic
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=[판독결과]==========
- Observed Genetic Variants
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Gene cDNA change AA change Zygosity Variant Category

HNF4A c.200G>A p.Arg67Gln Heterozygous Likely Pathogenic
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