To report a case of concomitance of diabetes mellitus, severe acanthosis nigricans, short stature in a 6-year boy.

Methods

Retrospective review of medical records of a patient with diabetes, acanthosis nigricans and short stature followed at the Departments of Endocrinology and Metabolism, Children’s Hospital of Fudan University. This rare case was undergoing whole exome sequencing.

Results

This boy 6 months old was diagnosed with severe acanthosis nigrican due to symptoms. When he was 6 years old, he was diagnosed with diabetes due to symptoms, laboratories work up and metformin therapy was started. His height was 106cm (<P3) and his weight was 17kg (P3-P10). His C-peptide was over 20 ng/ml with negative autoantibodies of GADA, IAA, IA-2A and ICA. His family history was very unique. His brother was diagnosed with neonatal diabetes and died at the age of one-month. His sister with severe acanthosis nigrians died of diabetes ketoacidosis during adolescent period. A homozygous mutation (p.I348F) was found in exons 4 of the INSR gene.

Conclusions

This case report showed that a homozygous mutation INSR mutation contributed to diabetes mellitus, severe acanthosis nigricans and short stature in this Chinese child with unique family history.

Nothing to disclose

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