



# DIABETES MELLITUS, SEVERE ACANTHOSIS NIGRICANS AND SHORT STATURE: A RARE ASSOCIATION IN CHINESE CHILDREN

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## OBJECTIVES

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 nigricans due to symptoms. When he was 6 years old, he was diagnosed with diabetes due to symptoms, laboratory 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 nigricans 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 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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