复旦大学附属儿科医院

DIABETES MELLITUS, AUTOIMMUNE HEMOLYTIC ANEMIA, HEPATOSPLENOMEGALY AND LYMPHADENOPATHY: A RARE ASSOCIATION IN CHINESE CHILDREN

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OBJECTIVES

To report a case of concomitance of diabetes mellitus, autoimmune hemolytic anemia, hepatosplenomegaly and lymphadenopathy in a 7-year girl.

METHODS

Retrospective review of medical records of a patient with multiple autoimmune diseases followed at the Departments of Endocrinology and Metabolism, Children's Hospital of Fudan University. This rare case was undergoing whole exome sequencing.

RESULTS

This girl 2 year was diagnosed with hepatosplenomegaly and lymphadenopathy due to symptoms. When she was 7 years old, she was diagnosed with diabetes due to symptoms, laboratories work up and multiple dose injection insulin therapy was started. Her sister died of diarrhea during infant period. Her Hemoglobin fluctuated between 95 and 123 g / L with positive autoantibodies ANA and ANCA. Her family history was very unique. Her brother was diagnosed with neonatal diabetes and died of diarrhea at the age of one-month. A compound heterozygous mutation (p.E22X and p.Q114X) was found in exons 1 and 3 of the *IL2RA* gene.

CONCLUSIONS

This case report showed that a compound heterozygous *IL2RA* mutation contributed to autoimmune phenomena in this Chinese child.

NOTHING to DISCLOSE

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