Introduction

Xp21 contiguous gene deletion syndrome is a rare X-linked recessive disorder resulting in glycerol kinase deficiency, congenital adrenal hypoplasia, intellectual disability and/or Duchenne muscular dystrophy. In this study, Xp21 contiguous gene deletion syndrome was confirmed in one family with three boys by clinical and genetic analysis.

Subjects and Methods

- The first child of the family was a boy. He showed dark skin and development delay at the age of one month, and died at the age of 1 year without definite etiological diagnosis.
- The second boy had similar clinical features. Congenital adrenal hyperplasia was suspected. Elevated serum creatine kinase and myohemoglobin were noticed. He was also died aged 1 year because of a febrile illness.
- The third boy was hospitalized for dark skin and hypotonia at the age of 7 days with the suspected diagnosis of congenital adrenal hyperplasia.

Results

- Significantly increased serum creatine kinase and myohemoglobin were found in the third boy of the family, indicating rhabdomyolysis.
- Elevated plasma adrenocorticotropic hormone with continuous low 17-OHP, severe hyperkalemia, hypertriglyceridemia and glyceroluria were observed.
- Xp21 contiguous gene deletion syndrome was confirmed by a SNP array for the boy.

After the supplements of hydrocortisone and fludrocortisone, the patient improved rapidly. But the serum creatine kinase was not decreased.

Conclusions

A family affected by Xp21 contiguous gene deletion syndrome was reported. Three boys experienced misdiagnosis. The differential diagnosis should be paid attention to for the patient with early-onset hypoadrenocorticism.