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# A novel large deletion mutation of *FERMT1* gene in a Chinese patient with kindler syndrome

**Key words:** Kindler Syndrome (KS), FERMT1 gene, Mutations

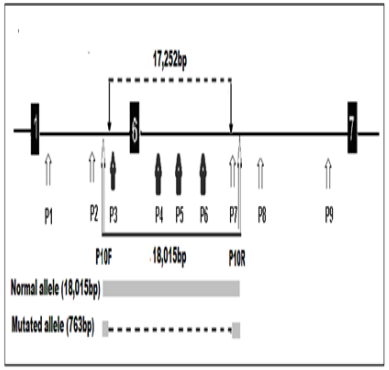
# *Research Summary*

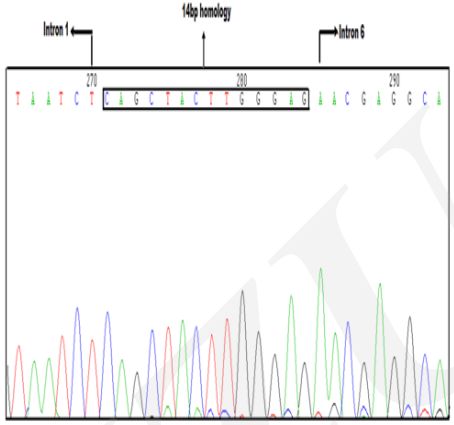
**This article was aimed to determine the mutation in the FERMT1 gene associated with a 7-year-old Chinese patient who presented clinical manifestation of KS.**

- **Mutation analysis of *FERMT1* gene in the family members.**
- **Clinical characterization of the patient with KS.**

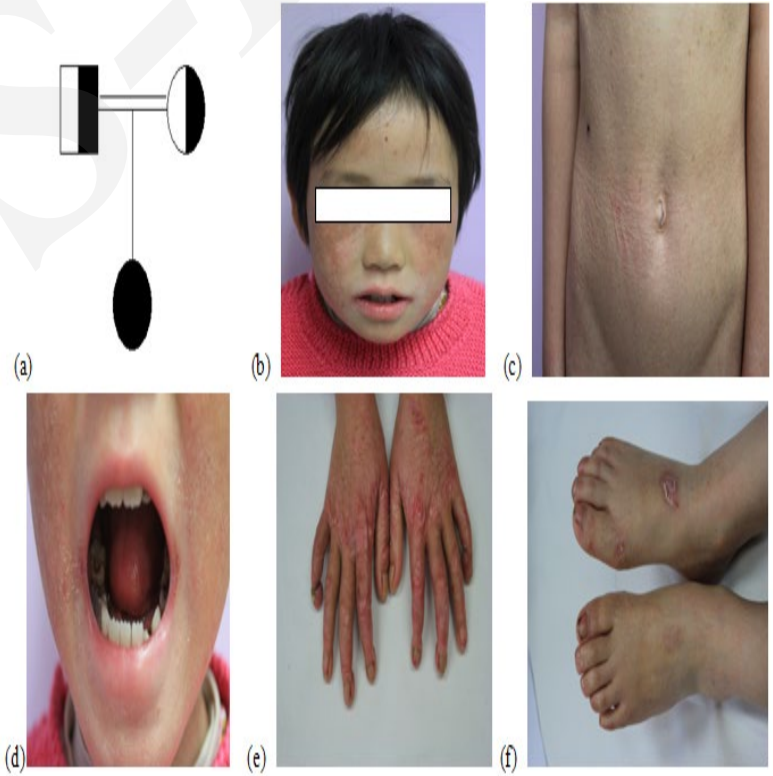
# *Innovation points*

- **Identification** of a new large deletion mutation of *FERMT1* gene.




- **Summary** of clinical characterization of the patient with KS.




- **Emphasis** of a rational diagnostic procedure of *FERMT1* gene mutations, especially for the patients with large deletion in this gene