Cite this as: Ying Gao, Jin-li Bai, Xiao-yan Liu, Yu-jin Qu, Yan-yan Cao, Jian-cai Wang, Yu-wei Jin, Hong Wang，Fang Song, 2015. A novel large deletion mutation of *FERMT1* gene in a Chinese patient with kindler syndrome. *Journal of Zhejiang University-Science B (Biomedicine & Biotechnology)*, **16**(11):957-962. [doi:10.1631/jzus.B1500080]

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

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

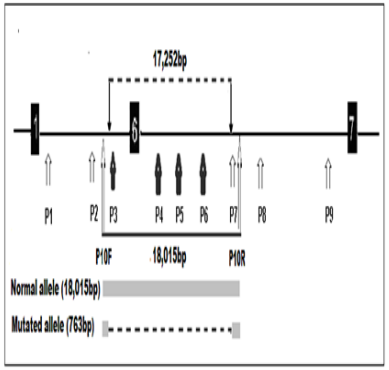
Research Summary

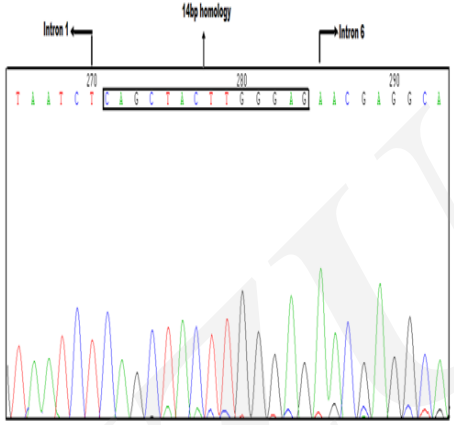
This article was aimed to determine the mutation in the FERMT1 gene associated with a 7-year-old Chinese patient who presented clinical manifestation of KS.

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

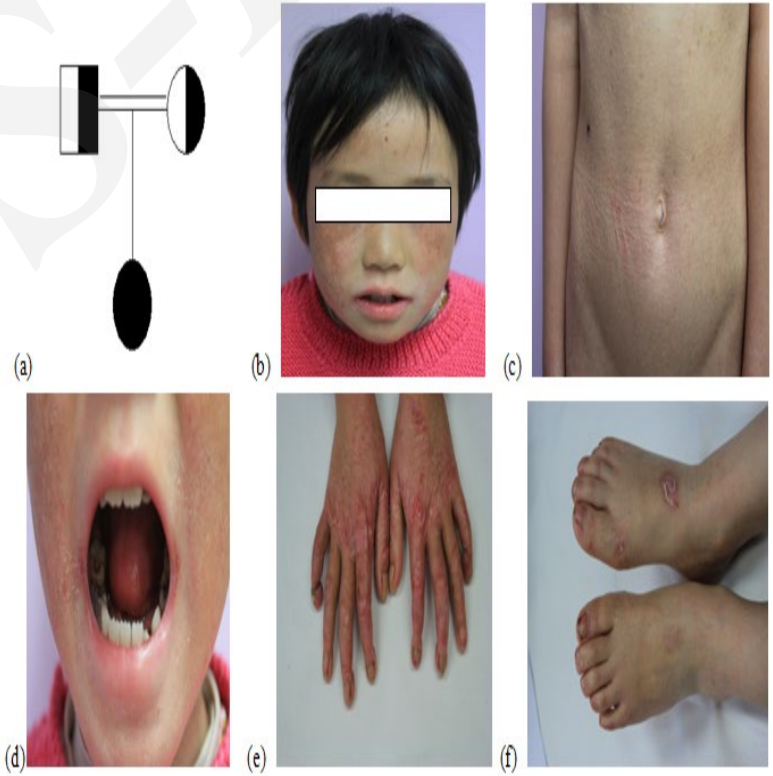
Innovation points

- **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