A Novel Heterozygous Pathogenic Variant in PORCN Gene Causing Focal Dermal Hypoplasia with Short Stature: Case Report and Literature Review

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**Objective** To explore the clinical features and the genetic cause of a multiple malformation patient with short stature. **Methods** The clinical data was collected in Beijing Children’s Hospital in November 2017. The disease-causing variant was identified using exome sequencing and confirmed with Sanger sequencing. Related literature was searched from Wanfang and Pubmed databases using the key word of “PORCN gene” to identify the clinical features and gene mutation. **Results** The patient is a 10-year-old girl, she was referred to hospital because of short stature. She presented multiple deformities such as patchy skin hypoplasia, syndactyly and right ear malformation. In exon 2 of PORCN gene, genetic sequencing revealed a de novo heterozygous variant c.49_80delTGTCTCCTGCCTACTGCCCAGCAGGGCCTTGA (p.C17fs*84). The variant is novel and classified as pathogenic. The patient was diagnosed with focal dermal hypoplasia (FDH). **Conclusion** FDH is a multi-system affected birth defect. In addition to typical skin damage and bone malformation, it can also cause short stature.

**Figure 1** Right ear malformation
**Figure 2** Blaschko linear pigmentation and depigmentation along the arms
**Figure 3** Fat hernia of leg
**Figure 4** Left middle finger and ring finger syndactyly after operation