shorter than pressure ulcer and all ulcers with SGD patients were healed completely. As elderly people gradually increase worldwide, it is important that these ulcers should be recognized in SGD patients and distinguished from pressure ulcer.

In conclusion, the ulcers developed with SGD are thought to be produced by mainly friction more than pressure force in elderly peoples, and their locations different from that of pressure ulcers. And it is important to notice that these SGD ulcers have relatively shorter duration and better response of treatment than that of pressure ulcers, so it is necessary to avoid excessive treatment.

CONFLICTS OF INTEREST

The authors have nothing to disclose.

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Fig. 1. (A∼C) The skin lesions of the proband. (D) Pedigree of the Chinese family in this study.

Fig. 2. Chromas diagrams of this family. The upper one: the proband and her father’s. The lower one: her mother and brother’s.
forelock. The severe form shows a typical white forelock on frontal scalp and relatively large leukoderma on the chest, abdomen, and extremities. The moderate phenotype is the intermediate type of these two forms. Missense and frameshift mutations of the KIT gene are responsible for a range of phenotypes with piebaldism.

We collected one Chinese family with piebaldism. The proband, a 16-year-old girl, born with a white forelock, irregular 1 ~ 5 cm-sized white patches on her abdomen and multiple large areas of white patches asymmetrically scattered over both knees with intrapatch freckling (Fig. 1A~C). Her father showed similar lesions. The other members of this family were normal. Based on clinical phenotype classification of piebaldism, a severe form of piebaldism was defined.

The family members voluntarily joined the genetic screening after informed consents were obtained. All exons, exon-intron flanking regions of KIT gene were analyzed by polymerase chain reaction and Sanger sequencing. A novel heterozygous missense variant (c.1993C>T, p.P665S) of KIT gene was identified from the proband and her father (Fig. 2). This mutation was absent in her mother and brother and 100 unrelated healthy people as well. By PolyPhen-2, this mutation was predicted to be probably damaging with a score of 0.999 (sensitivity: 0.09, specificity: 0.99). All of these results supported that this mutation probably had pathogenic significance.

Generally, loss of functional mutations of KIT gene is responsible for almost all cases of piebaldism. The cytoplasmic region of KIT contains a juxtamembrane domain and a TK (tyrosine kinase) domain that is subdivided into a TK domain 1 (TK1), a kinase insert domain and a TK domain 2 (TK2). The severe phenotypes of piebaldism usually result from mutations involving the intracellular tyrosine domain, whereas the mild phenotypes are usually caused by mutations involving the amino-terminal extracellular ligand binding domain. Wasag et al. demonstrated that one mutation (p.T619A) in the TK1 domain led to much weaker phosphorylation level of KIT protein than wild type KIT. In addition, one mutation (p.G664R) was demonstrated to cause severe phenotype of piebaldism. As the novel mutation (p.P665S) is in the TK1 region and adjacent to the above missense mutation, we infer that this mutation (p.P665S) may probably affect the autophosphorylation of KIT and thus result in significantly decreased proliferation and migration of melanocytes in the lesions.

In summary, a novel missense mutation (p.P665S) in TK1 region of KIT was reported in a Chinese family with severe piebaldism. Mutations in cytoplasmic TK region are mostly associated with severe phenotypes; however, the pathogenicity of this variant remains to be fully elucidated.

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CONFLICTS OF INTEREST

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