## Cosmetic & Pharmaceutical Research > Cosmetic & Pharmaceutical Research > Hair and adnexia

# 1 | Novel D323G mutation of DSG4 gene in a girl with localized autosomal recessive hypotrichosis clinically overlapped with monilethrix.

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**ABSTRACT**

BACKGROUND: Localized autosomal recessive hypotrichosis (LAH) is an inherited rare disease caused by DSG4 mutations, characterized by short, sparse, brittle hair affecting restricted areas such as the scalp, trunk, and extremities. To date, DSG4 mutations have been reported in 14 pedigrees of LAH overlapping with monilethrix. METHODS: To clarify the etiology of hair defects for a 2-year-old Chinese girl, peripheral blood, skin, and hair samples were collected, and skin immunohistochemistry, electron microscopy (scanning and transmission types), Vivascope confocal microscopy, and DSG4 sequencing were investigated. RESULTS: The patient presented sparse hairs of various length and follicular hyperkeratotic papules. Eyebrows and lashes were also involved (broke or shed). The biopsy specimen revealed curled ingrown hair shafts within the hair follicle and keratin-filled hair follicles. Scanning electron microscopy revealed hair cuticle loosely and irregularly arranged, as well as a marked warping, curling, cracking, and detachment of hair cuticle. Transmission electron microscopy indicated notable dysadhesion between cells of the outer root sheath. A homozygous mutation A1103G in exon 8 of DSG4 was identified in the patient, resulting in the substitution of an aspartic acid by glycine (D323G) and reduced DSG4 expression in the affected scalp epidermis. CONCLUSIONS: The homozygous A1103G mutation in DSG4 was responsible for the disease development. © 2015 The International Society of Dermatology. PMID:26173648 DOI:10.1111/ijd.12889