HAIR DISORDERS

A HEREDITARY HYPOTRICHOSIS SIMPLEX IN A CHINESE FAMILY

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Background: Hereditary hypotrichosis (HH) is a heterogeneous group of inherited hair loss disorders which can
essentially be divided into syndromic and nonsyndromic forms. Hereditary hypotrichosis simplex (HHS; OMIM146520 /605389) is a rare heritable autosomal dominantnon-syndromic hereditary hypotrichosis disorder which is characterized by progressive loss of hair beginning in the middle of the first decade of life.

Observation: Two affected individuals, one unaffected relatives from a Chinese family clinically characterized as HHS. The affected boy and his mother had normal scalp hair density at birth. At approximately 4 months of age hair loss began and progressed gradually with age until near complete loss of scalp hair. Sequence analysis revealed that DNA obtained from the boy and his affected mother had the same recurrent p.Leu9Arg mutation of the PRL21 gene while that was negative in his unaffected aunt. CDSN and APCDD1, failed to detect sequence variants in either affected or unaffected individuals of the family. We obtained genetic diagnosis of HHS. It is also not known whether RPL21 is involved in the Wnt signaling pathway. Functional study of this gene will provide important insights into the molecular and cellular basis of hair growth.

Key message: Heterozygous missense mutation in the RPL21 gene on chromosome 13q12.12-12.3 with the generalized form of HHS. Here, we report a recurrent missense mutation c.95G>A in the RPL21 gene in a Chinese family with HHS.