PIGMENTATION

HYPOMELANOSIS OF ITO WITH A LOSS OF HETEROZYGOSITY IN THE 7Q31.31-7Q32.1 REGION AND KRT5/14 MUTATION

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Background: Hypomelanosis of Ito (HI) is characterized by hypopigmentation following the Blaschko's lines, usually associated with chromosomal mosaicism. Keratin (KRT) 5 and 14 are structural proteins in basal layer participating in modulation of cell differentiation and migration. Mutation of KRT5/14 are related to multiple congenital dermatoses involving pigmentary abnormality.

Observation: A 19-year-old male presented with asymptomatic patches and streaks of hypo- and hyperpigmentation on the right side of face, trunk and extremities. The lesion began with a congenital erythema on the right forehead which turned into a hypomelanotic patch and extended to the right face and neck 3-month afterbirth. By the age of 1, additional hypomelanotic patches and streaks developed on the right back and extremities. 6 years later, multiple brown hyperpigmented macules spontaneously appeared within the former hypomelanotic patches and streaks. The tone and contour of the lesions stabled thereafter. The patient had no congenital malformations or delays in mental and physical development. No related family history was mentioned. Physical examination revealed hypopigmented patches and streaks on the left face, trunk and extremities following the Blaschko's lines. The dark-brownish, hyperpigmented spots were superimposed within the hypomelanotic lesions and confined to them. Histological findings of the hypomelanotic patch revealed few melanocytes and decreased pigmentation in basal layer. Electron microscopic found dysfunctional melanocytes with vacuolated mitochondria and immature melanosomes. The hyperpigmented lesion exhibited increased pigmentation in basal layer manifested as massive caps consist of numerous mature melanosomes on the nuclei of basal cells. Microarray analysis with DNA isolated from both peripheral blood cells and hypomelanotic lesions showed no mosaicism but a loss of heterozygosity in the 7q31.31-7q32.1 region. Further whole exome sequencing found two novel variants at KRT5 (c.115G>T, p.Gly39Trp) and KRT14 (c.580C>T, p.Arg194Cys).

Key message: KRT5/14 mutation accompanied by chromosome abnormality might be a new pathogenesis of HI.