GENETICS AND GENODERMATOSES

ULTRASTRUCTURAL INVESTIGATIONS IN AN AUTOSOMAL RECESSIVELY INHERITED CASE OF DYSCHROMATOSIS UNIVERSALIS HEREDITARIA- A CASE REPORT

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Background: Dyschromatoses are the presence of asymptomatic mottled hyperpigmented macules admixed with hypopigmented macules. They are divided into dyschromatosis symmetrica hereditaria (DSH) and dyschromatosis universalis hereditaria (DUH). There are specific mutations in ADAR1/DSRAD and ABCB6 genes in DSH and DUH, respectively. It is usually transmitted in an autosomal dominant (AD) pattern and very few in autosomal recessive (AR).

Observation: 23-year old unmarried male, product of consanguineous marriage, presented to us with mottled dyspigmentary patches since the age of 15 years. Dermatological examination revealed 1–10 mm sized hypopigmented and hyperpigmented macules. Ultrastructural and molecular analysis and electron microscopy demonstrated morphologically normal melanocytes containing melanosomes of all stages and number of melanosomes showed more than two fold rise in both melanocytes and keratinocytes in hyperpigmented lesions. Further analysis based on their distribution pattern in keratinocytes revealed significantly higher number of single as well as clusters of 2–4 melanosomes in hyperpigmented skin. On investigating the regulation of melanogenesis pathway in DUH, we compared the transcript levels of 3 key melanogenic enzymes– DCT (dopachrometautomerase), TRP1 (tyrosinase related protein 1) and Tyr (tyrosinase) in the epidermis of the biopsies.

Key Message: In this form of DUH, melanocyte numbers are not affected, however, the melanosome synthesis and maturation are affected. There was more than two-fold up-regulation of DCT and TYR in hyperpigmented epidermis.