GENETICS AND GENODERMATOSES

WHEN THE SUN CAUSES THE PROBLEMS OR DOUBLE CASE OF XERODERMA PIGMENTOSUM IN A FAMILY

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Background: Xeroderma pigmentosum (XP) is one of the rare autosomal recessive disorder that occurs due to inactivation of the xeroderma pigmentosum protein, which is an important DNA damage recognition protein involved in DNA nucleotide excision repair (NER) and characterized by hypersensitivity to sunlight and is associated with a high predisposition to UV-induced skin cancers. UVR induces dipyrimidine photoproducts such as cyclobutane pyrimidine dimer (CPD) and 6-4 pyrimidineepyrimidone photoproduct (6-4PP), which cause distortions in the double helix (Lagerwerf et al., 2011). Because of NER or translesion synthesis deficiency, patients with XP cannot remove or overcome the dipyrimidine photoproducts.

Observation: The siblings of 16 and 12 years of age were admitted to Dermatology department of Tashkent Pediatric Medical Institute, Uzbekistan, with 15- and 11-years lasting lesions continuing to grow and transform. The family anamnesis revealed a close-up marriage. The patients have photosensitivity and easily develop sunburns with erythema, edema and vesicles following sun exposure. From 1 year of age the solar lentigines have been developed. By the age of 16 the elder brother has 3 focal of squamous cell carcinomas (SCCs), 2 of which was successfully treated by cryosurgery. The younger sister has the similar pigmented rash on a face, however, in addition presented nodular hyperkeratotic mass on her lower lip. The histological findings showed signs of SSC. There are no neural and ocular abnormalities was found during examination. The management included rigorous photoprotection, CO2 laser desiccation of skin formations and topical application of 5% imiquimod.

Key message: the abovementioned clinical case of double incident of XP demonstrates the actuality of genodermatoses and importance of genetic consultations before planning a child for a prophylaxis.