GENETICS AND GENODERMATOSES

TWO CASES OF XERODERMA PIGMENTOSUM INVOLVING SIBLINGS

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Background: Xeroderma Pigmentosum is a rare genetic autosomal recessive disease characterized by sensitivity to UV-light. I present two siblinges' cases of XP.

Observation: Patient D.C. is 9-year-old male child, patient C.C. is 13-month-old boy. D.C. had the skin redness and lesion appeared at the age of six months; the younger one - at the age of eight months. Both children had skin lesions occurred after short time sun exposure. D.C had been observed by local dermatologist with the diagnoses Photodermatitis, Atopic dermatitis for several years. Parents are healthy and deny consanguineous marriage.
Patient D.C. had dry skin. Erythema plaques with erosion and serum crusts were prominent on the cheeks and jaw skin. On the skin of the face, limbs, trunk hyper-pigmented spots found varying 2-6 millimeters in diameter and slightly raised above the surface. The spots had no definite shape; some were oval while others were irregular in shape. Spots were denser in both color and distribution over cheeks, temples and extender aspects of extremities; spots present over the body .
C.C. has erythema on the face, forearms, legs. There are some irregular-oval shaped mocca-colored spots on the back.

Investigation: Identical investigation was performed for both patients; EKG, CBC, Chem-7, UA, EKG, BUN, serum creatinine, ELISA, Hepatitis B and C, Uroporphyrin, Echo-cardiography.
D.C. tests showed mild Hypochromic Anaemia.
C.C. tests showed a PFO. Examination of other systems both children did not reveal abnormalities.
Abnormalities were not find by Oncologist.
Considering the similar histories and clinical tests conveyed the suggestion that both patients were suffering XP. The diagnoses were confirmed by Kazakhstan Central Institute of Dermatology.

General message: XP is a rare disease and can be taken as Photodermatitis and Atopic Dermatitis. Therefore, patients should be fully examined. Have to bear in mind this genetic disease.