GENETICS AND GENODERMATOSES

# XERODERMA PIGMENTOSUM : CHALLENGE OF DIAGNOSIS IN WEST AFRICA

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Xeroderma pigmentosum is related to a defect of the enzymes involved in repairing the oncogenic effects of ultraviolet exposure. The condition is found all over the world, in all ethnicities and races. This rare genodermatosis is often unknown in countries lacking specialist in dermatology. This scarcity and insufficiency of qualified personnel give rise to difficulties in diagnosing this pathology, especially in West Africa where XP is wrongly diagnosed for other pathologies. Objective: To share with colleagues the problem of diagnosis of Xeroderma pigmentosum in countries with insufficient number of dermatologist and poor technical platform. Observation: 21-year-old man, with no pathological history of dermatosis was seeking for medical consultation because of cutaneous dryness and photophobia. Previous visits at several health centers failed to diagnose the condition and no noticeable improvement was seen from given treatment. Verbal questioning found the notion of consanguinity (his father and his mother are cousins, and belong to the same Dogon group). Cutaneous pigmentary disorders were absent at birth but appeared at the age of 12 years. Physical examination highlighted photophobia in addition to specific clinical and paraclinical signs allowed to diagnose Xeroderma pigmentosum varying type. Conclusion: in West Africa, the challenge of diagnosis of Xeroderma pigmentosum is undoubtedly related to a lack of qualified personnel and technical means of diagnosis, which makes its frequency underestimated.

Key words: Xeroderma pigmentosum, West Africa, challenge of diagnosis