Xeroderma pigmentosum with Ophthalmological Symptoms

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Xeroderma pigmentosum is a rare skin disease with autosomal-recessive heredity, which is characterized by hypersensitivity to sunlight. The skin areas exposed to the sun exhibit poikiloderma, teleangiectasis, atrophy of the epidermis and hyperkeratosis. Moreover, multiple benign and malignant tumors of the skin may develop, even at an early age [3]. Xeroderma pigmentosum may be associated with ophthalmological symptoms such as photophobia, keratoconjunctivitis and tumors of the eyelids, cornea and limbus.

The hypersensitivity to sunlight is probably due to an enzymatic defect. Ultraviolet light brings about chemical changes of DNA chains which changes may impair mitosis and other cellular functions. In normal cells, such DNA defects may be recognized and eliminated, following which the continuity of the DNA chain is repaired by supplementation of DNA bases in the correct sequence. This process is called repair replication. Cells of patients suffering from xeroderma pigmentosum show a decreased capacity of repair replication after irradiation with ultraviolet light [1,2].

Below, one case will be concisely reported, with reference to some of the ophthalmopathological and dermatopathological findings.

The patient, a woman aged 20 years, had healthy parents. Her only brother suffers from the same disease. The patient's hypersensitivity to direct sunlight had already been observed when she was a baby. As early as the 6th year of life, a few malignant skin tumors, including one squamous cell carcinoma, had been removed by Dr. Doormaal, a dermatologist in Breda. Early in 1972, Dr. Smals, an ophthalmologist in Breda, had removed a leiomyoma of the limbus of the left eye. In view of the

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rapid recurrence of this tumor in the same localization, the patient was referred to the Ophthalmological Hospital in Rotterdam. In September 1972, part of this tumor was removed and submitted for examination to the Department of Pathology of the Medical Faculty of Rotterdam. It was diagnosed as a mesenchymal malignancy, probably a leiomyosarcoma. Orbital exenteration was then performed. Ophthalmopathological examination of the eye and the other orbital contents was performed under the direction of Dr. Manschot. In addition to the above-mentioned mesenchymal tumor, this examination revealed a small melanoma of the choroid. In a period of 6 months after the exenteration, a plastic surgeon took 34 biopsy samples of the skin which not only showed the characteristic histopathological picture of xeroderma, but proved also to contain benign and malignant tumors of various natures such as: a few melanomas, basocellular carcinomas, squamous cell carcinomas, haemangiomas, a xanthoma and a kerato-

acanthoma. The patient died suddenly after extensive skin transplantation of the face. Previously she had exhibited severe neurological abnormalities, which had probably been due to a cerebral melanomous metastasis brought to light by the post-mortem examination.

References

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