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Case Report

Corneoscleral locally aggressive fibrous histiocytoma in Xeroderma Pigmentosum patient: A case report

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Abstract

Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder that has been found in all continents and racial groups in relation to faulty repair of DNA with sun exposure. Several cutaneous and ocular tumors have been described in relation to XP including fibrous histiocytoma (FH). The diagnosis of conjunctival FH is challenging owing to the rarity of this tumor and the diversity of its classification into benign, locally aggressive and malignant. We are describing a recurrent FH exhibiting a locally aggressive behavior in a child with history of XP. Detailed histopathological features are presented with literature review.

Keywords: Fibrous histiocytoma, Xeroderma pigmentosum, Fibrohistiocytic, Fibroxanthoma

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Introduction

Xeroderma Pigmentosum (XP) is a rare autosomal recessive disorder and has been found in all continents and racial groups. It is a disorder of DNA repair characterized by sun sensitivity and UV radiation-induced skin and mucous membrane cancers. Although patients with XP develop several cutaneous malignancies during their disease, cutaneous angiosarcoma and fibrous histiocytoma (FH) are extremely rare. We describe a case of corneoscleral FH in a 7-year old female patient who is known to suffer from XP.

Case report

A 7-year-old Saudi female diagnosed with Xeroderma Pigmentosum (XP) presented initially in the year 1996, to our institution because of redness, irritation and photophobia; and a pterygium-like growth in her left eye. Protection from sunlight was recommended with no indication for surgical intervention. At the age of 12 years, her nasal limbal conjunctival mass was excised elsewhere with removal of corneal base, and cryotherapy of the affected area. The specimen's size was 5 mm in greatest dimension grossly and histopathologically showed active spindle cell growth, with fibrohistiocytic features, a fascicular and storiform growth pattern, focal xanthoma cells and frequent mitoses but minimal anaplasia. The diagnosis was active spindle-cell proliferation consistent with a low-grade fibrohistiocytic lesion, with potential for recurrence.

By the age of 14, she presented with a large irregular nodular minimally vascularized conjunctival mass obscuring the entire left cornea and limbus with no further visualization of the anterior segment details. Her best corrected vision was light perception with good projection. Her right eye was unremarkable. The facial skin demonstrated the typical features of XP without skin tumor (Fig. 1).

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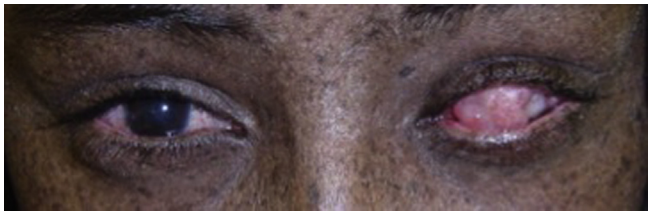

Fig. 1. The clinical appearance of our 14-year old patient with XP and a large conjunctival mass growing over her left cornea.

CT scan showed a corneal mass protruding into the anterior chamber with no evidence of intraocular or retro bulbar extension. Enucleation was performed and the globe showed an irregular corneal lesion extending from limbus to limbus. Histopathologically, the peripheral corneal epithelium was variable in thickness and separated from Bowman's layer by an infiltrative tumor. The epithelium was absent over the main surface of the tumor, which has replaced the corneal stroma with formation of a plaque-like protruding area towards the anterior chamber without disrupting Descemet's membrane (Fig. 2A & B).

The tumor was composed of two cell types; arranged in storiform pattern: spindle-shaped cells with elongated nuclei and larger cells with rounded nuclei and prominent nucleoli that were presumably histiocytic. Mitoses were rare in the deeper tissue, the cells were generally fibroblastic in appearance with a fascicular distribution while in the superficial tissue, numerous mitoses (5–10/40 higher power field) were present, as well as few multinucleated giant and pleomorphic tumor cells (Fig. 2C & D). Immunohistochemical stains were performed and the tumor cells strongly expressed Vimentin with focal expression of CD68 (Fig. 2E & F). The proliferation index with Ki67 was low. The tumor cells failed to express Desmin, Cytokeratin, SMA, Melan A, CD34. The histopathological diagnosis was recurrent left eye limbal fibrous histiocytoma, locally aggressive.

Patient was followed up with no evidence of recurrence for 18 months' post-enucleation then she lost follow up since then.

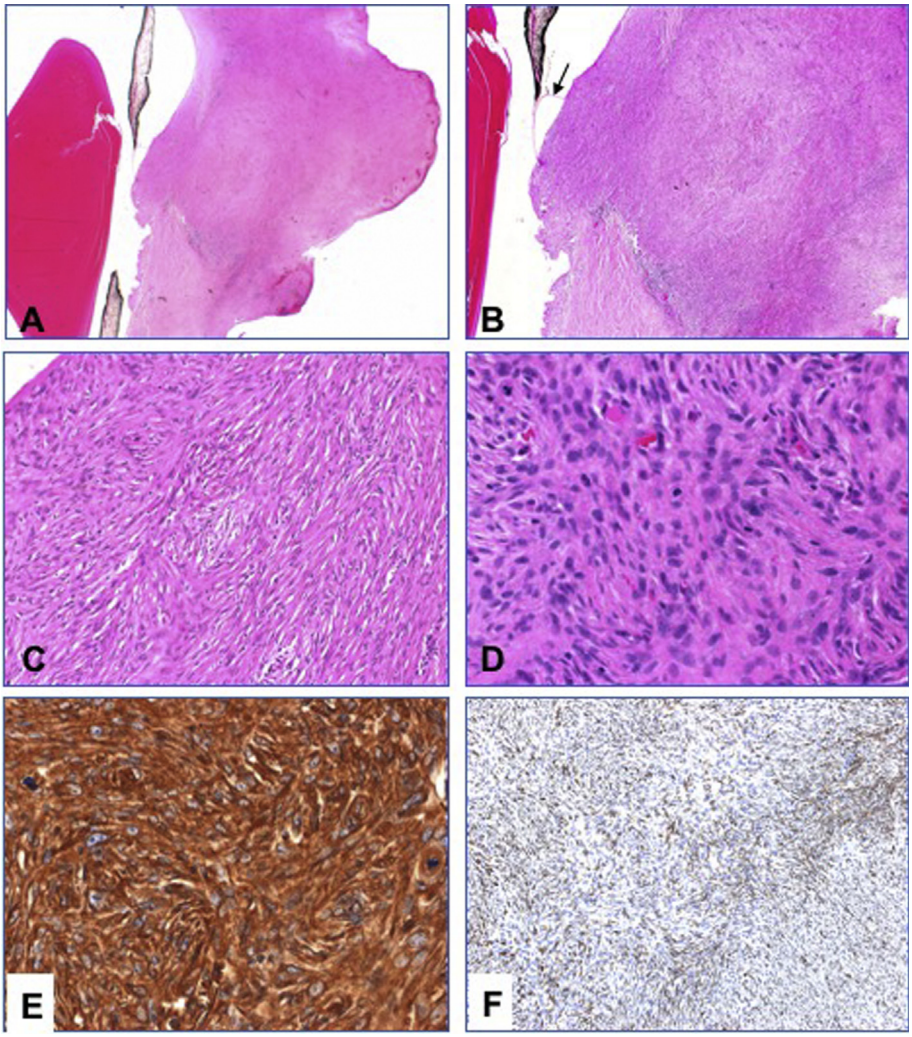

Fig. 2. A. Low power histopathological photo of the anterior part of the enucleated globe sowing the extending corneal mass from the adjacent limbus (Original magnification 12.5× Hematoxylin & Eosin). B. Slightly higher magnification of the corneal mass with protrusion into the anterior chamber and intact Descemet's membrane (black arrow). (Original magnification 25× Hematoxylin & Eosin). C. The deeper part of tumor with spindle-shaped cells in a storiform pattern (Original magnification 200× Hematoxylin & Eosin). D. The superficial part of tumor with pleomorphic cells and mitotic figures (Original magnification 400× Hematoxylin & Eosin). E. Immunohistochemical staining showing positive tumor cells with Vimentin (Original magnification 400×). F. Focal positive tumor cells with CD68 (Original magnification 200×).

Discussion

XP is a rare autosomal recessive disorder of DNA repair characterized by sun sensitivity and UV radiation-induced skin and mucous membrane cancers. The disease begins in early life with the first exposures to sunlight, the median age of onset being 1–2 years of age, with skin rapidly exhibiting the signs associated with years of sun exposure. This includes extreme sensitivity to sunlight, triggering severe sunburn, abnormal lentiginosis (freckle-like pigmentation due to increased numbers of melanocytes) on sun-exposed areas, xerosis, skin aging and multiple skin cancers, with the lack of proper sun protection.[1,2] Skin tumors include basal cell carcinoma, squamous cell carcinoma, and malignant melanoma, where the patient may ultimately die from the consequences of their tumors. Although patients with XP develop several cutaneous malignancies, cutaneous angiosarcomas and fibrous histiocytoma are extremely rare.[3] In Saudi Arabia, XP has been recently studied and the estimated incidence was found to be like what has been reported in the United States and Europe (1:1000,000), however, consanguinity was consistent and almost all parents in that series were first degree relatives.[4] Ocular involvement was reported including squamous cell carcinoma in 19% and limbal melanoma in 4%.[4]

On the other hand, fibrous histiocytoma (FH) formally known as fibroxanthoma encompasses a heterogeneous group of soft tissue tumors that are composed of cells that resemble fibroblasts and histiocytic cells, which are often arranged in a cartwheel or storiform pattern.[5] Conjunctival FH can be benign, locally aggressive or malignant based on the presence of pleomorphism and atypical mitoses histopathologically. Malignant FH is known as pleomorphic undifferentiated sarcoma and has a local recurrence rate of 19–31%, a rate of metastasis up to 35%, thus a low five-year survival rate of 65–70%.[6] In that review, it has been concluded that the mean age was 39 years with no gender predilection.[6] The lesion typically shows vessels across the surface rather than feeding vessels at the base which was observed in our case.[7] Clinically, the lesion is often described as fleshy-pink or erythematous conjunctival lesion, however it can be yellowish owing to the presence of histiocytes.[8–10] It can clinically resemble scleritis and/or episcleritis, xanthogranuloma, solitary fibrous tumor or even pterygium like in our case initial presentation.[11]

Microscopically, FH comprises a variable mixture of spindle-shaped fibroblasts and ovoid histiocytes. Immunohistochemistry, although not specific enough to distinguish FH from other mesenchymal lesions, serves to exclude other spindle cell tumors and is essential for the diagnosis of FH. FH cells are expected to express Vimentin (such as in our case), factor XIIIa, and CD163. The mitotic activity found near the limbus like our case, might explain the pathogenesis of this tumor as being originating from the corneal epithelial stem cells.[6,8,9]

Regarding the age, benign FH has been reported in childhood irrespective of the absence or the presence of XP.[12]

Atypical fibroxanthoma, which consists of myofibroblastic cells is another similar entity that can be confused with malignant FH and has been reported in association with XP. Regarding such association, three cases have been reported of FH/fibroxanthoma of the ocular surface in XP patients. Pe'er in 1999 reported the first case of malignant fibrous histiocytoma of the skin and bulbar conjunctiva in a 3-year-old girl from Malawi diagnosed with XP.[13] The second case was reported by Shao in 2007 in a 5-year-old boy with the diagnosis of XP and atypical fibroxanthoma of the conjunctiva.[14] then the most recent case of atypical fibroxanthoma was described in a 24-year old male with XP.[15] Our case also presented in childhood, which can be explained by the fact that XP is a predisposing factor for FH to occur before adulthood.

The most appropriate management of FH at any site, particularly the conjunctiva, is complete surgical excision with tumor-free margins to avoid local recurrence.[10,11]

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Declaration statement

This case report has been approved by the Research department at KKESH. The authors have no conflict of interest or financial declaration in relation to this case report.

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