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Case Report

Xeroderma pigmentosum and rhabdoid tumor of the kidney: A very rare case report association ☆

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ABSTRACT

The aim of this observation was to report an exceptional association of xeroderma pigmentosum and rhabdoid renal tumor in a 7-year-old girl, diagnosed with imaging and treated by adjuvant chemotherapy.

Introduction

Xeroderma pigmentosum (XP) is defined by extreme sensitivity to sunlight, which translates into sunburn, pigment changes in the skin, and a very high incidence of skin cancer [1]. It is a rare autosomal recessive disorder of nucleotide excision repair [2]. Prevalence is estimated at 1/300,000 in the United States and Europe. Certain populations have a higher prevalence (Japan, North Africa, Egypt, and the Middle East). Prevalence is increased, especially in communities in which consanguinity is common [3]. The diagnosis of xeroderma pigmentosum is established in a proband on the basis of clinical findings and family history and/or by the identification of biallelic pathogenic variants in one of the genes [4].

The most commonly reported causes of death were skin cancer (34%), neurological degeneration (31%), and internal cancer (17%). The occurrence of extra-dermatological cancers in patients followed by XP has been rarely reported in the literature [4].

Observation

A 7-year-old girl from a consanguineous marriage, the second of 2 siblings, was followed in the dermatology department for XP since the first year of her life, the diagnosis of XP was made on clinical criteria: skin pigmentation, skin ulceration after sun exposure and photosensitivity. There is no similar

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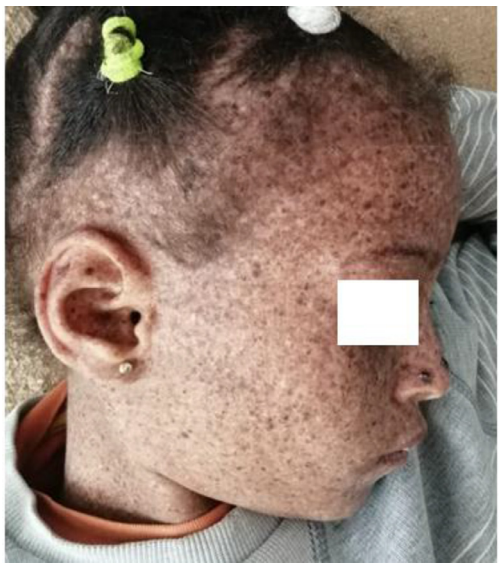

Fig. 1 – Hyperpigmentation skin lesions.

case in the family of the 2 parents. The patient admitted to the hospital for right back pain which evolved over 4 months.

Clinical examination revealed a good general condition; diffuse hyperpigmented skin lesions (Fig. 1). An abdominal examination revealed a firm, palpable mass situated in the right lumbar. The biological assessment showed no renal failure, cytobacteriological urinalysis was normal, no red blood cells, with a negative culture.

For initial evaluation, abdominal ultrasound revealed a right renal mass poorly limited, heterogeneous, containing hypoechogenic zones, vascularized in color Doppler, with hilarious renal lymphadenopathy presenting the same characteristics as the mass. A thoraco-abdomino-pelvic CT was performed in order to better characterize this renal mass and to rule out metastases. It confirmed the presence of an upper right polar renal mass, poorly defined, multiloculated, with irregular contours, measuring 50×20×10 mm, invading the renal pedicle and segments VI of the liver, but was separated from the right adrenal gland, associated with retroperitoneal lymphadenopathy (Figs. 2 and 3). The contralateral kidney was

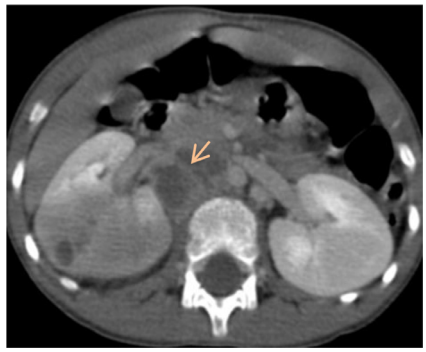

Fig. 3 – Retroperitoneal lymphadenopathy.

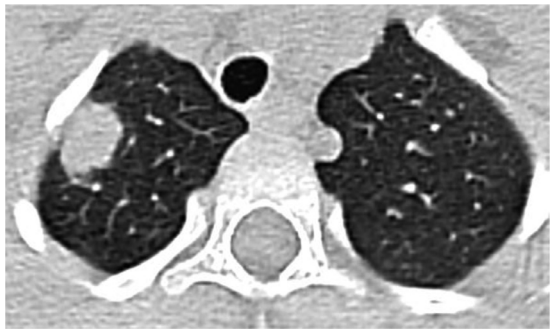

Fig. 4 – Spiculated mass of the right superior lobar lung.

unharmed. CT showed tumor extension into the right superior lobar lung (Fig. 4), with spiculated contours mass, and left subclavicular lymphadenopathy.

An ultrasound-guided biopsy of the subclavicular lymphadenopathy was decided in view of neoadjuvant chemotherapy. Histological and immuno-histochemical analysis of the biopsy revealed a rhabdoid tumor of the kidney (Figs. 5 and 6).

Adjuvant chemotherapy was indicated, and the patient died after her first chemotherapy session.

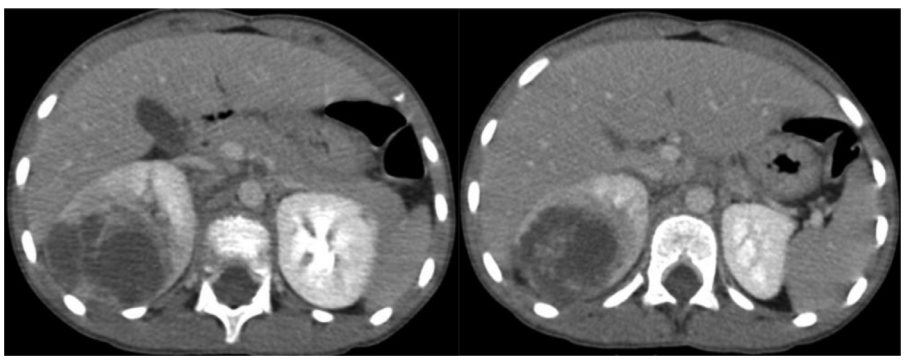

Fig. 2 – Axial contrast-enhanced CT image showing heterogeneously enhancing renal right masse, multiloculate, with irregular contours.

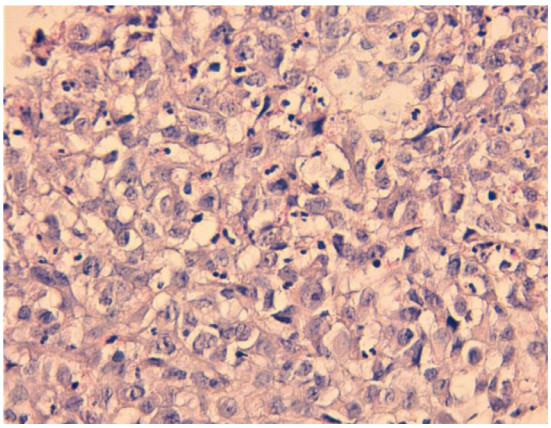

Fig. 5 – High magnification showing a tumoral proliferation made up of spindle-shaped cells, with a focally rhabdoid morphology. The tumor cells presented marked cyto-nuclear atypia, with a hyperchromatic nucleus surrounded by an eosinophilic cytoplasm, sometimes clarified. Numerous mitosis were present (magnificationx40, hematoxylin-eosin staining).

Discussion

Xeroderma pigmentosum patients have sun sensitivity and an increased skin cancer risk, this hypersensitivity is due to genetic abnormalities responsible for a decrease in DNA repair capacity [5].

This disease is related to a defect in genes within the nucleotide excision repair system for the first 7 genetic groups (A-G), and to an abnormality in transcription groups for the eighth group (xeroderma pigmentosum variant-XPV). This genetic heterogeneity is responsible for the great diversity in the clinical presentation of XP [2].

In addition to skin tumors, there is a risk of extra dermatological cancers. This risk has been evaluated between 10 and 20 times more than the general population. Several types of tumors have been reported: acute leukemia, thyroid carcinomas, uterine sarco-mass, lung, and brain tumors [3]. The pathophysiological mechanisms of the occurrence of tumors in internal organs not exposed to UV rays have not been elucidated in patients suffering from XP.

The association between xeroderma and kidney tumors is not current. The histologic types of these tumors were: nephroblastoma (n = 1) [8], mixed epithelial and malignant stromal renal tumor (n = 1) [9], and 2 cases of leiomyosarcoma associated with these tumors were described [6,7]. The occurrence of renal rhabdoid tumor in the case of XP is exceedingly rare, and our case represents the first case reported in the literature.

Rhabdoid tumor of the kidney is a rare, highly aggressive malignancy of early childhood, extremely rare over the age of 5 years, recognized as a distinct neoplasm from Wilms tumor. However, the origin of the tumor is still unclear. The median age at diagnosis is 11 months [10]. The major initial symptoms of a rhabdoid tumor of the kidney are comparable to those of other renal tumors, including hematuria, abdominal pain, and palpable mass [11]. Several CT findings were suggestive of renal rhabdoid tumor: calcifications, heterogeneous and lobulated renal mass, tumor necrosis, enlarged vessels, subclavicular hematoma [12]. Metastases are most frequently seen in the lungs, lymph nodes, the liver, and the brain. Invasion of the renal vein is common [10].

The treatment of choice consists of radical nephrectomy and resection of adjacent lymph nodes followed by chemotherapy. Survival is poor, with an 18-month survival rate of only 20% [13].

Conclusion

To our knowledge, this is the first case associating Xeroderma pigmentosum and renal rhabdoid tumor. Survival is poor, making the study of this association difficult. Radical nephrectomy is the treatment of choice.

Patient consent

The patient's parents agreed with a written informed consent to anonymously publish their daughter's medical informations.

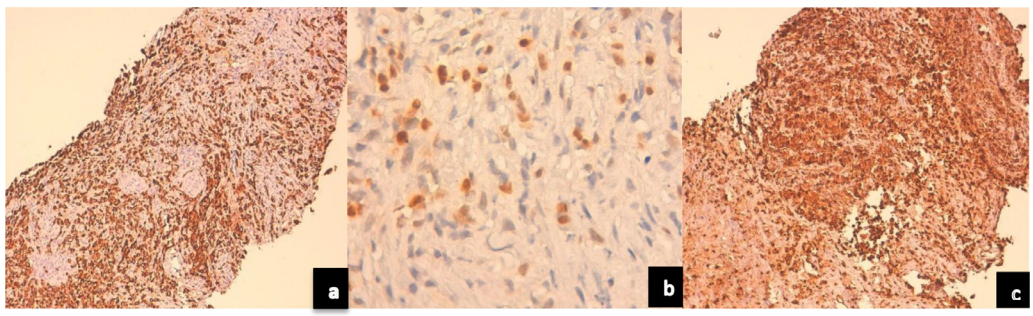

Fig. 6 – Immunohistochemically the tumoral cells expressed, (A) Cytokeratine (Magnification x10), (B) Cycline D1(Magnification x40), (C) Vimentine (Magnification x20).

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