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Bloom Syndrome About One Cases

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Abstract **Case Report**

Bloom syndrome is an immune-compromised disease with autosomal recessive inheritance secondary to BLM gene mutation with a high risk of malignant degeneration. The stunting delay and the predisposition to cancers of different histological types and multiple localizations are constant. The diagnosis may be unrecognized until the onset of complications and is essentially based on the caryotype. No treatment is known to date and the long-term prognosis is poor with a mean age of 21 years.

Keywords: Gene mutation, caryotype, malignant degeneration, chemotherapy.

INTRODUCTION

Bloom syndrome is an immune-compromised disease with autosomal recessive inheritance secondary to BLM gene mutation.

The high risk of malignant degeneration requires knowledge of this pathology. We report in this study an observation about a case collected at the Pediatric Traumatology Orthopedics Unit at Mohammed VI University Hospital, Marrakesh, Morocco.

CASE REPORT

15-year-old girl, of low socio-economic level. From a first degree consanguineous marriage. Follow-up for BLOOM syndrome and bilateral bronchial dilatation. Consult for a swelling of the left shoulder that has appeared for two months and is rapidly increasing in volume.

Antecedents

Thin-weight delay. Followed since the age of 7 for branchial dilatation. Repeated digestive infections. Hypochromic microcytic anemia. No similar case in the family.

Clinic: weight delay at - 4 DS, conjunctival blades.
T °: 38c °.

Narrow face, slight dolichocephaly, with hypo-pigmentation spots spread all over the body (Figure1,2).

Fig-1: Narrow face, slight dolichocephaly

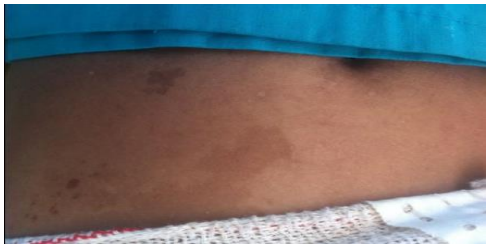

Fig-2: Hypo-pigmentation spots spread all over the body

Cough with greenish sputum. Slight dyspnea. Alteration of the general condition. Painful hot swelling at palpation with collateral circulation and inflammatory signs extended to the shoulder and upper extremity of the right arm (Figure 3).

Fig-3: Collateral circulation and inflammatory signs extended to the shoulder and upper extremity of the right arm

Radiography

Radiography of the shoulder (Figure 4): lytic image of the upper extremity of the humerus with fuzzy boundaries, with osteolysis, rupture of the cortex and invasion of the soft parts, complicated pathological fracture.

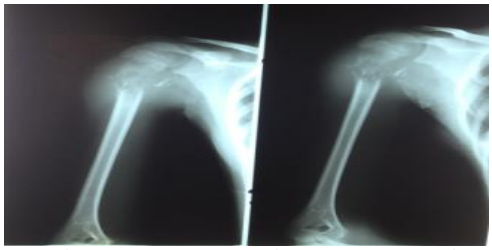

Fig-4: Radiography of the shoulder : lytic image of the upper extremity of the humerus with fuzzy boundaries, with osteolysis, rupture of the cortex and invasion of the soft parts, complicated pathological fracture

 MRI (Figure 5): metaphyso-diaphyseal tumor process of 9x8x7 cm with involvement of the cortex and infiltration of the soft parts, involvement of the conjugation cartilage, the humeral arteria is pushed in and the basilar vein is envelopped by the tumor, no skip metastases.

Biopsy

Well-differentiated and keratinizing squamous cell carcinoma with infiltration of the bone, starting from the skin. Extension assessment: X-ray of the thorax showing interstitial syndrome and bilateral basal opacities, an abdominal ultrasound showing hepatic hilar adenopathies measuring for the largest 5x2.23cm, vascularized with color Doppler.

Decision

First chemotherapy treatment, then oncologic amputation with assessment of hepatic tumor localization.

Evolution

Patient died during her chemotherapy treatment in a septic shock table on medullary aplasia and immunodepression field.

DISCUSSION

First described en 1954 by American dermatologist DAVIS BLOOM [1]. Autosomal recessive disease [2]. It is due to the mutation of the BLM gene located at position 15q 26.1., Causing a non-sister chromatid exchange of homologous chromatids factor greater than 10 times normal [2].

Incidence: 1 / 559,000 in Germany, 1 / 63, 331, 000 in the United States, 1 / 10,836,000 in Japan, 2 cases described in Morocco [3].

The stunting delay and the predisposition to cancers of different histological types and multiple localizations are constant [4,5].

Ocular manifestations (photosensitivity), sexual disorders, endocrine disorders (diabetes) and multiple malformations have been described including flat feet, bone hypoplasia, polydactyly, absence of a toe, stenosis of the ureter and meatus ... [6].

The diagnosis may be unrecognized until the onset of complications and is essentially based on the caryotype. In the literature, 168 cases of BLOOM syndrome have been described, of which 100 cases have developed cancers, ie 59.5% of cases, viz, 51 carcinomas (51%), including 8 carcinoma of the skin and 21 myeloid and lymphoid leukemia, 23 lymphomas and 5 rare tumors (2 tumors of WILLIS, 2 osteosarcoma and 1 medulloblastoma [7].

Surgical treatment of tumors is reserved for localized forms of early discovery that respond well to chemotherapy. Infectious episodes of the disease are treated symptomatically (antibiotic therapy, analgesic treatment, physiotherapy and a good nutritional balance. The long-term prognosis is poor with a mean age of 21 years

CONCLUSION

The bloom syndrome is a rare genetic disorder of which no treatment is known. A genetic counsuling with reducing consanguineous marriages and performing an antenatal diagnosis still needed. Long-term surveillance should be performed in order to early detect cancers and treat repetitive infections.

REFFERENCE

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