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Case Report
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About One Case of Primitive Embryonal Rhabdomyosarcoma

KA Aly Mbara[1], DIAGNE Jean Pierre[1], DIENG Mamour[3], GUEYE Abou[1], SOW Aboubacry Sadekh[2], SY El Hadji Malick[1], NDIAYE Joseph Matar Mass[2], MBAYE Soda[1], DE MEIDEROS Marguerite Edith Quenum[1], SAMRA Audrey[1], GAYE Serigne Sohibou[1], BADGI Mariama[2], WANE Aissatou Magatte[3] and NDIAYE Papa Amadou[1]*

[1]University Ophthalmological Center of Abass Ndao Hospital, Senegal

[2]Ophthalmology Department of Aristide le Dantec Hospital, Senegal

[3]Diamniadio Children's Hospital Ophthalmology Department, Senegal

Abstract

Aim: in order to underline the rarity as well as the diagnostic, therapeutic and prognostic aspects of this affection, we report one case of an embryonic orbital rhabdomyosarcoma in a newborn.

Observation: It was a 3 months-old female baby, full-term by vaginal delivery, of a well-followed pregnancy. The mother was 19-years old, primigravida, primiparous and without parental inbreeding. The baby was admitted at the 5th day of life for a left exorbitism at her birth.

Examination found a tumoral orbital syndrome with a beginning corneal necrosis. A head CT-scan found a dense tissue process occupying the left orbit and discharging the orbital content outside, with a contrast enhancement. It was developed on medial rectus muscle. The alpha-fetoprotein level at 8th day of life was high, about 400 ng/ml. An exaltation was realized at the 9th day of life with a pathological examination evoking a rhabdomyosarcoma. After a month and a half, there was a tumor recurrence. The blood smear showed more than 25% of blasts. The baby has received courses of chemotherapy.

Discussion and Conclusion: The diagnosis of rhabdomyosarcoma must be evoked in any child with orbital evolutive signs. An imaging must be performed urgently. The embryonal form is the more common, but its occurrence in newborn period is particular.

Keywords: Rhabdomyosarcoma; Newborn; Vital prognosis

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Correspondence:
KA Aly Mbara, Department of Ophthalmology, Abass Ndao National Hospital Eye Center, Senegal, E-mail: kaam75@hotmail.com

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Introduction

Rhabdomyosarcoma (RMS) is a striated muscle differentiation tumour that develops at the expense of non-bony supporting tissue and can occur in any part of the body. Its orbital location represents 12% of all possible locations. It is the most common malignant mesenchymal tumour in children after retinoblastoma and accounts for 5% of all pediatric malignant tumors [1,2]. It is extremely virulent hence the need for early diagnosis that greatly improves survival and prognosis. The latter is related to histology, lack of metastasis at diagnosis, age of the child, localization, volume and operability of the tumor. It is a radiochemosensible tumor, whose surgical treatment is less and less indicated [2]. We report an embryonic-like orbital rhabdomyosarcoma in a newborn to highlight the rarity as well as the diagnostic, therapeutic and prognostic aspects of this condition.

Observation

It was a 3-month-old female infant born on full-term, by vaginally delivery, from a well-attended pregnancy. The mother was a 19-year-old woman, primigravidous, primiparous and without any notion of parental inbreeding. She was received at day 5 of life for an exorbitism of the left eye at birth. The initial examination showed an orbital tumor about 5 cm long axis, exorbitism, chemosis, and onset of corneal necrosis (Figure 1). The somatic examination was without particularity. A CT scan of the head and face (Figure 2) was performed. It showed a tissue density process occupying the left orbit pushing the orbital contents out, strongly enhanced after contrast injection. It was developed at the expense of medial right muscle, with a reactive inflammation of the homolateral ethmoid. There was no intratumor calcification or damage to the cerebral and orbital contralateral stage. An extension check-up, including clinic, chest X-ray, abdominal ultrasound and echocardiography, was normal. The level of alpha-foetoprotein on the eighth day of life was 400 ng/ml. An exenteration was

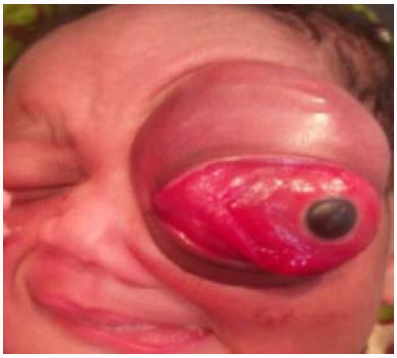

Figure 1: Exorbitism, chemosis, corneal necrosis in the left eye.

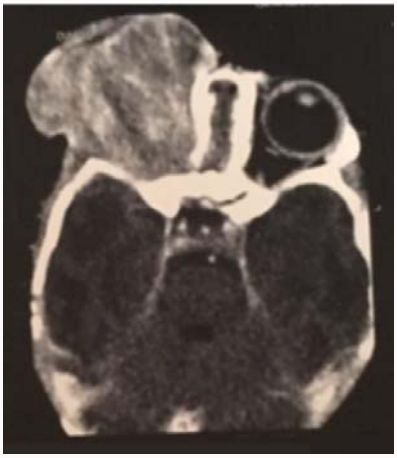

Figure 2: Left orbital tissue density process pushing the orbital contents out, strongly enhanced after contrast injection.

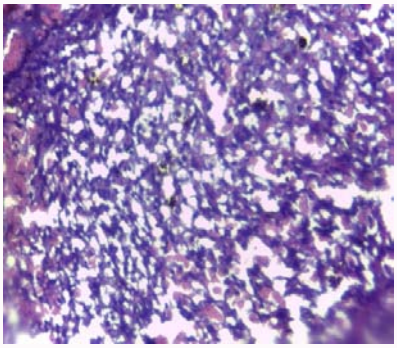

Figure 3: Histological section shows a carcinomatous tumor proliferation of round and elongated cells resembling rhabdomyoblats, morphological aspect in favor of rhabdomyosarcoma.

made at day 9 of life and a pathological examination was performed, suggesting a rhabdomyosarcoma (Figure 3).

At one and a half months of life, the child had a tumour recurrence (Figure 4). Alpha-foetoprotein was 298.2 IU/L and normal beta HCG. The blood smear showed more than 25% blasts and the cytopuncture was noncontributory. The child had received chemotherapy in the pediatric oncology unit (Figure 5).

Discussion

This tumor, which mimics striated muscle tissue and develops from mesenchymal embryonic cells, can be at the expense of striated

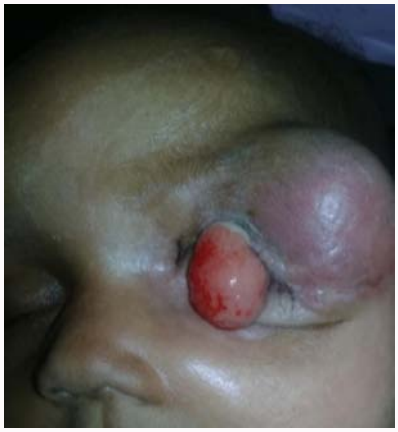

Figure 4: Tumour recurrence at one month and a half of life.

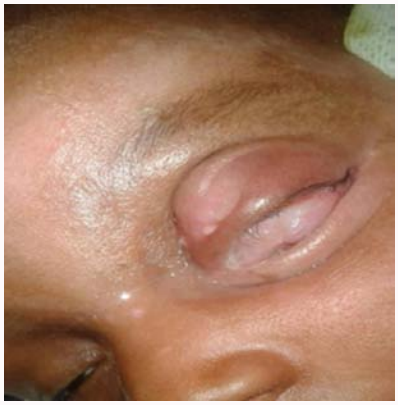

Figure 5: Left eye tumor regression of after chemotherapy.

muscle as well as in an area where it is absent [3]. 76% of orbital locations are in the orbital cavity; 12% conjunctival, 9% intraocular, and 3% palpebral. Intraocular locations are exceptional and develop from the ciliary body or iris [3]. This is cavitary localization for our case.

Rhabdomyosarcoma is an early childhood tumor; it is the most common primary malignant orbital tumor in children. The average age at diagnosis is 7 to 8 years [2]. Rare cases were described during the neonatal period [4,5]. This makes our case unique. Neonatal forms are rare, with an estimated frequency of 0.4% to 2% [6].

The classical clinical presentation is a very rapidly evolving unilateral exophthalmia [2]. As we observed in our patient, we often note an explosive character of the symptomatology. The fundus may be normal or have papillary edema. Rhabdomyosarcoma can be revealed by metastases (liver, lung, bone, brain) [3,5]. In our patient the extension assessment did not reveal metastases.

Moreover, the imaging is not pathognomonic, but provides arguments in favor of the diagnosis: Tissue density of the lesion, enhancement after injection of iodine or gadolinium, osteolysis aspects of the orbital walls. It makes it possible to visualize the lesion, to locate it in the orbit, sometimes to affirm its original structure, to measure it, to identify its relationships and to specify its orbital or encephalic extension [3,7,8]. For our case the computed tomography showed characteristics in favor of this tumor.

Histology classifies rhabdomyosarcomas into two subtypes,

embryonic and alveolar. Embryonic rhabdomyosarcoma is more common, about 80% of cases, and has a better prognosis than alveolar rhabdomyosarcoma, with 94% and 74% survival respectively [2].

The contribution of immunohistochemistry is valuable in this type of tumour [9]. It makes it possible to correct certain erroneous diagnoses by the dosage of anti-desmine AC, anti-myogenin and smooth anti-muscle. Indeed, the distinction between rhabdomyosarcoma and other mesenchymal tumors is sometimes difficult especially when it comes to undifferentiated forms, as in our observation.

The main differential diagnoses of embryonic rhabdomyosarcoma are orbital teratoma, lymphangioma, hemangioma, meningocele and epithelial cyst [5].

Management that may include chemotherapy (vincristine, actinomycin D, cyclophosphamide) and/or radiotherapy requires multidisciplinary consultation [5,9]. Surgery is not systematic [5]. The Intergroup Rhabdomyosarcoma Study Group I (IRSG I) give priority to preserving the function over a complete resection [9,10]. Since the introduction of multimodal chemotherapy and/or radiotherapy for localized rhabdomyosarcoma in children and adolescents, the percentage of survival has improved significantly from 25% in the 1970s to over 74% today [2,9,11,12]. This management therefore requires a multidisciplinary approach, as we have done, involving the pediatric oncologist, the pediatric ophthalmologist, the radiotherapist, the radiologist and the pathologist.

It is important to consider from the moment of diagnosis the possibility of healing after a well-conducted treatment, but also acute and late toxicities of therapies. In our case, the diagnosis of teratoma was first mentioned motivating an exenteration followed by a pathological examination and five adjuvant chemotherapy cures because of the initial tumor volume which was very important. But it is after one month that the diagnosis of embryonic rhabdomyosarcoma was obtained under the basis of the histopathology report.

Current treatments have now improved 5-year survival to 92% of patients [5]. Poor prognosis factors are age less than 12 months, alveolar or undifferentiated forms, tumour size (>5 cm), lymph node involvement, delayed diagnosis and the presence of metastases at the time of diagnosis [5]. Considering our patient's age, undifferentiated tumor histology, the large size of the tumor and the extra-orbital invasion of the tumor, our patient had a derogatory vital prognosis with tumor recurrence noted around the fortieth day after surgery and death sometime later. This could also be related either to incomplete exenteration, or to a late and secondary chemotherapy or a need for untreated radiotherapy.

Conclusion

The diagnosis of rhabdomyosarcoma should be mentioned in any child who presents an evolutionary orbital symptomatology. Imaging is performed in an emergency (computed tomography or nuclear magnetic resonance). When a suspicious mass or infiltration is detected, a biopsy is quickly programmed. The amount of tissue collected must be sufficient to allow a classical histology which will be supplemented by an immunohistochemical examination.

Once the diagnosis is made the treatment must be undertaken urgently by chemotherapy combined more or less with radiotherapy or surgery.

The embryonic form being the most common, but its occurrence in the newborn makes the particularity of our observation because the congenital form is little described in the literature.

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