

## Congenital alveolar rhabdomyosarcoma in a newborn.

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### Summary

We report a neonatal case of congenital alveolar rhabdomyosarcoma, presenting as a purple-red tumor on the left upper eyelid. The patient was treated with partial excision, chemotherapy and, finally, at the age of one year, radiotherapy. He was free of recurrences during 18 months of follow-up.

### Key words

Rhabdomyosarcoma, newborn.

**R**habdomyosarcoma (RMS) is a malignant mesenchymal neoplasm that exhibits striated muscle differentiation. This tumor is the most common soft tissue sarcoma of childhood accounting for over half of all cases.

Congenital alveolar rhabdomyosarcoma in a newborn is extremely rare and invariably fatal with current therapy.

### Case report

A 13-day-old boy was born by normal spontaneous vaginal delivery at 38 weeks of gestation. His birth weight was 2.5 kg. The infant was referred for evaluation of a rapidly enlarging tumor in the left upper eyelid, which was already present at birth.

On physical examination, there was a purple-red non tender nodule, measuring 4 x 3 cm in diameter. The lesion was not movable on the underlying tissue (Fig. 1). There were no other associated skin findings. Except for the skin lesion, the general clinical examination and the routine laboratory tests were otherwise unre-

markable. A computed tomography scan of the head revealed an intraorbital soft tissue mass, which caused slight exophthalmos. There was no evidence of bone or ocular involvement. The presumed diagnosis was capillary hemangioma. Because of its continuous and rapid enlargement, an excisional biopsy was performed.

The histopathologic examination showed infiltration of the reticular dermis and subcutaneous tissue with aggregates of round blue cells with scant clear cytoplasm in an alveolar growth pattern, separated by trabecular connective tissue (Fig. 3).

By immunohistochemical studies the tumor cells were positive for vimentin, muscle specific actin, myogenin and desmin (Fig. 4). These features were consistent with the diagnosis of alveolar rhabdomyosarcoma. Molecular cytogenetic analysis specific for alveolar RMS is not available in our country nor FISH.

Treatment included partial surgical resection and chemotherapy consisting of vincristine, actinomycin D, and cyclophosphamide. Radiotherapy was added at the age of 1 year. 18 months after the diagnosis the tumor completely regressed leaving a depressed scar (Fig. 2).



Fig. 1



Fig. 2

Fig. 1, 2: Purple-red, non tender, 4 cm x 3 in size tumor of the left upper eyelid (Fig. 1). The same boy, when aged 18 months (Fig. 2), presented an atrophic scar following the regression of the tumor.

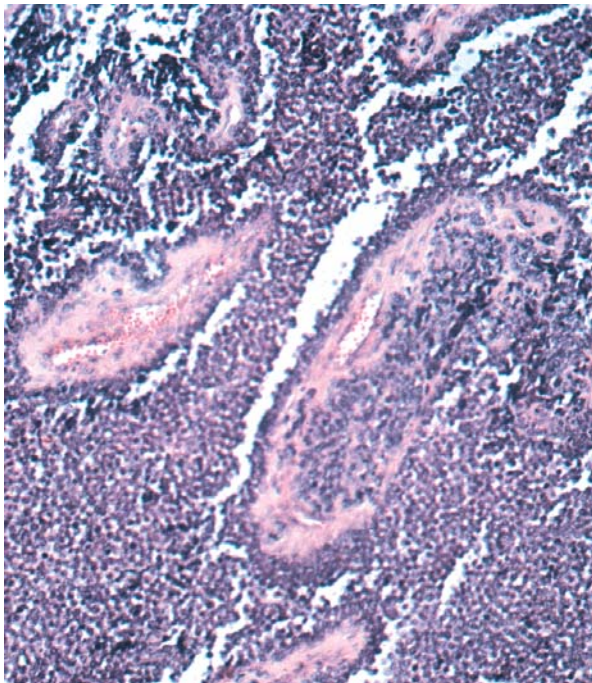


Fig. 3

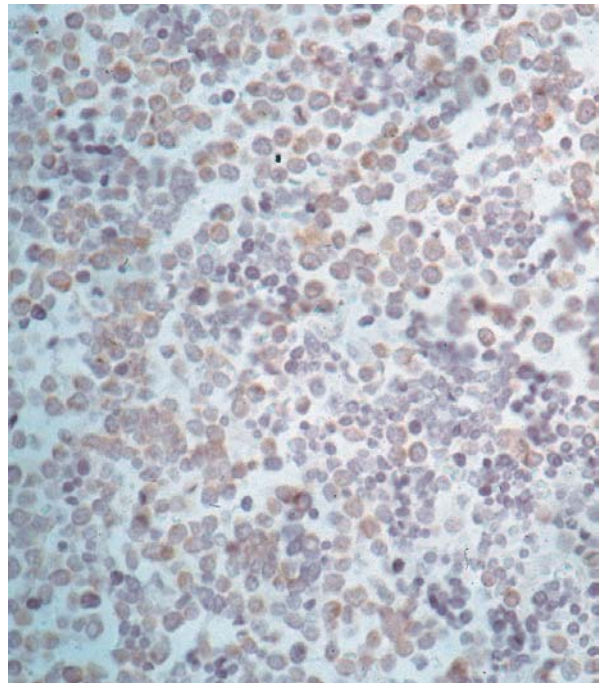


Fig. 4

Fig. 3, 4: The histological examination shows infiltration of the dermis (Fig. 3, 25x, H&E) with small, round, blue cells with scant cytoplasm in an alveolar growth pattern, separated by trabecular connective tissue. The tumor cells are desmin+ (Fig. 4, 45x, immunoperoxidase).

## Discussion

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma of childhood. The median age at diagnosis is 5 years and almost two thirds of all patients are diagnosed before 10 years of age, the tumor being rare in adults. Two percent of cases are present at birth (7). Head and neck tumors are most common in children younger than 8 years of age and, when arising in the orbit, are almost always of embryonal variety (8). RMS accounts for 4-8% of all malignant solid tumors in children (3). In most cases RMS presents as a rapidly enlarging and ill-defined mass, which may compress vital structures. It tends to occur most often in the head, neck, extremities and genitourinary tract. Cutaneous presentation of RMS is uncommon. The dermal nodules were thought to be the result of RMS arising within the dermis or secondarily invading the dermis from the underlying soft tissue (7, 11, 12).

Orbital RMS is a high-grade malignant tumor usually presenting at 7-8 years of age. When managed by surgery alone it is characterized by high mortality rate, but recent advances in chemotherapy and radiotherapy improved significantly its prognosis. Orbital involvement occurs in approximately 10% of the patients and alveolar histology is seen in only 9% of cases (6, 10). The latter pathological findings imply a less favorable prognosis, because no survivors were so far reported (4). The congenital alveolar cases are rare and seem to be different from alveolar RMS in older children (1, 9).

The typical pathological finding of RMS is characterized by the presence of small, round and blue tumor cells. Therefore, it should be emphasized that in case of a round cell tumor, immunohistochemical and ultrastructural studies are mandatory in order to identify the tumor and differentiate it from other forms, including RMS, neuroblastoma, Ewing's sarcoma, malignant lymphoma and small cell carcinoma.

Four main pathological subtypes of RMS are recognized: embryonal, alveolar, pleomorphic, and botryoid. The embryonal variety is the most frequent in infants and young children (11). The alveolar subtype is characterized by the presen-

ce of alveolar-like spaces lined with small, round neoplastic cells and with tumor cells floating within these spaces.

Immunohistochemical studies may show undifferentiated cells to be positive only for vimentin indicating a mesenchymal origin. More differentiated cells may be positive for desmin, actin, myogenin and myoglobin, indicating a muscular differentiation.

Molecular cytogenetic analysis shows that alveolar RMS is consistently associated with the characteristic translocations: t (2; 13) (q 35; q 14) and t (1; 13) (p 36; q 14) which respectively encode for the PAX 3-FKHR and PAX 7-FKHR fusion oncoproteins. The patients having PAX 3-FKHR positive alveolar RMS are characterized by a worse prognosis as compared to patients with alveolar RMS containing the less common translocations PAX 7-FKHR (2, 4).

Metastases may occur via lymphatics to regional lymph nodes or hematogenously most commonly to lungs, bone and bone marrow (3, 5).

Treatment options include radical or partial surgical resection, chemotherapy, which is recommended in all cases because of the poor prognosis due to local recurrence and, finally, radiotherapy, which is necessary when the tumor cannot be completely removed (10).

In conclusion, we report a newborn with a congenital alveolar rhabdomyosarcoma in the left upper eyelid. This tumor was responsive to surgery, chemotherapy and radiotherapy and experienced good response, being free of recurrences during 18 months of follow-up. The better prognosis of our case could be related to PAX 7-FKHR fusion, although we were not able to confirm this hypothesis.

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