ORIGINAL ARTICLE

Eye Manifestation of Extrarenal Malignant Rhabdoid Tumour

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SUMMARY

Extrarenal malignant rhabdoid tumour (EMRT) is very rare and aggresive childhood neoplasm with a rapid progression. The prognosis is still very poor with 80 % mortality rate. We report a case of a newborn baby with extrarenal malignant rhabdoid tumour of an upper eyelid. An EMRT was diagnosed based on the histological examination. This case report highlights the clinical presentation, radiological features and difficulty in diagnosis. The purpose is to underline the importance of its inclusion in the differential diagnosis of any aggresive lesion in a child.

Key words: retinopathy of prematurity, screening, timing of the examination

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INTRODUCTION

Malignant rhabdoid tumour (MRT) is a rare, highly malignant tumour in childhood age. It was classified as an independent unit in 1978 (1, 2). Its tissue origin has not yet been specified, and a therapeutic protocol has not yet been stipulated either (2). The tumour occurs in three main localisations: in the kidneys, in the central nervous system and in soft tissues. It very often metastasises, and even despite a combined therapy, the mortality rate reaches 80% (1). Extrarenal malignant rhabdoid tumour (EMRT) is a subgroup of MRT.

CASE REPORT

The authors describe a case of a newborn girl, who was transferred immediately after birth to the department of neonatology at the University Hospital Brno, due to suspected haemangioma of the upper left eyelid. The family anamnesis was without any unusual features, the infant originates from the 6th physiological pregnancy.
The clinical picture showed a semi-solid projecting formation of the upper left eyelid, with a size of approximately 2 cm (Fig. 1). The bulb was intact, without deviation. The finding on the anterior segment and the fundus did not manifest pathological changes. Suspicion of haemangioma of the upper left eyelid was stated. We immediately conducted an MR examination (Fig. 8) and ultrasonography. Both examinations evidenced a haemangioma of the upper eyelid.

As a result we applied an adequate therapy for haemangioma – oral therapy with propranolol and corticoids. Due to progression of the tumour, DepoMedrol 40 mg was applied via injection into the tumour, as well as 3 IV boluses of SoluMedrol, and a salvage therapy was commenced for infants with rapidly progressing haemangioma (1x Avastin, 1x IV Taxol).
In the differential diagnostics we considered the two main diagnoses: congenital haemangioma and sarcoma. As the progression clinically continued (Fig. 2), and haemorrhage from the formation newly appeared, an examination was conducted by positron emission tomography, which demonstrated spreading into the eye socket and increased metabolic activity, which was within the range of malignancy. We conducted control ultrasound and MR examinations (Fig. 9,

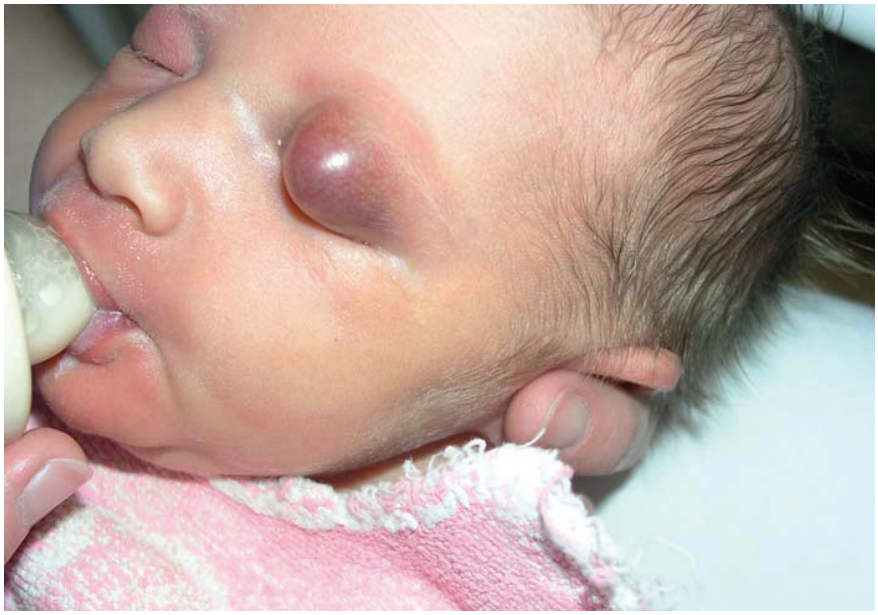

Fig. 1 Condition 2nd day after birth.

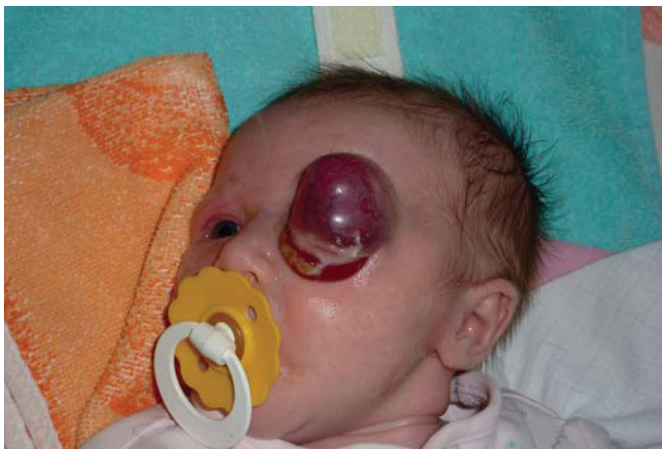

Fig. 2 Condition 2nd week after birth

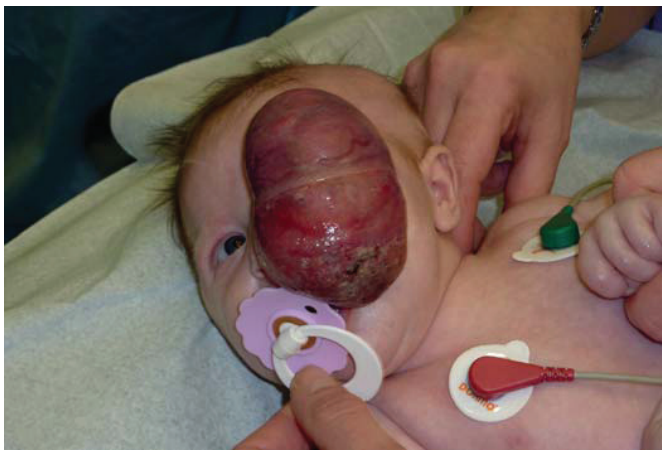

Fig. 3 Condition 3rd week after birth

Fig. 4 Condition 1st day after surgery

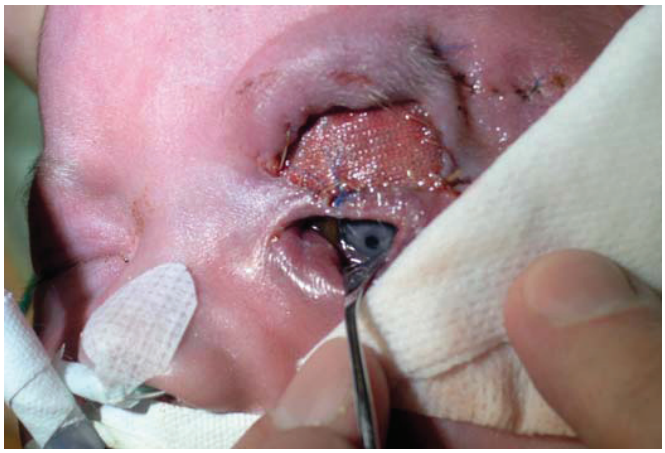

Fig. 5 Condition 2nd day after operation

10, 11), which showed a pronounced enlargement and progression of the malignant tumour.

Conservative therapy was unsuccessful, the focus grew very rapidly and protruded (Fig. 3), closing the slit lid gradually and exerting pressure upon the left nostril. As a result we indicated a biopsy from the cranial part in the area of the superciliary ridge and a histological examination. The preliminary result of the histological examination indicated juvenile congenital fibrosarcoma. A surgical procedure was subsequently indicated: majority marginal resection of the tumour and reconstruction of the upper eyelid (partially by moving the soft tissues with partial reconstruction of the conjunctiva and tarsal plate and temporary coverage of the upper eyelid with artificial skin cover until the definitive determination of the biological activity of the tumour (Fig. 4, 5, 6).

The final result of the histology from the resected tumour was EMRT. The initial staging of the disease was without demonstration of remote foci of the disease. MR of the brain and the orbit did not demonstrate a measurable residue of the tumour. Full-body MR and full-body scintigraphy of the skeleton were without pathological foci of increased activity. As a result, the oncologists commenced therapy according to the protocol of the European Paediatric Society for sarcoma of soft tissues from 2005 (NRSTS EpSSG 2005), which incorporates surgical resection, chemotherapy and radiotherapy. The chemotherapy involved 10 cycles of application of cytostatics: Vincristine, Doxorubicin, Cyclophosphamide, and Cyclophosphamide, Carboplatin, Etoposide. With regard to the necessity of the fastest possible commencement of chemotherapy, we relinquished the reconstruction of the upper eyelid by a more complex method, and the artificial skin cover was replaced by a rotated move of skin flap from the left temporal region, leaving a functionally deficit coloboma medially. The healing took place without fundamental complications, the coloboma was gradually stabilised and the infant learned to adapt movements of the bulb according to the lighting, if the eye is not covered.

With regard to the age, the potential adverse effects and the general remission of the disease, radiotherapy was not conducted.

Our patient is now 10 months after the reconstruction of the upper eyelid and 2 months from the end of chemotherapy. Her overall condition is favourable; the local finding is relatively satisfactory from a cosmetic perspective following plastic surgeries (Fig. 7). The lid slit is smaller, with a pronounced coloboma of the upper eyelid, which afflicts as much as 75 % of the eyelid. For this reason we must prevent onset of exposure keratitis by the application of lubricants. The bulb is in hypotropia, the dynamics of the bulb are limited when looking upwards. The conjunctiva is slightly hyperaemic; the visible part of the cornea is smooth and transparent. The definitive solution of the reconstruction of the upper eyelid has not been planned yet. However, the prognosis for the future is infaust according to the literary data.

DISCUSSION

Malignant rhabdoid tumour was first described as an independent clinical

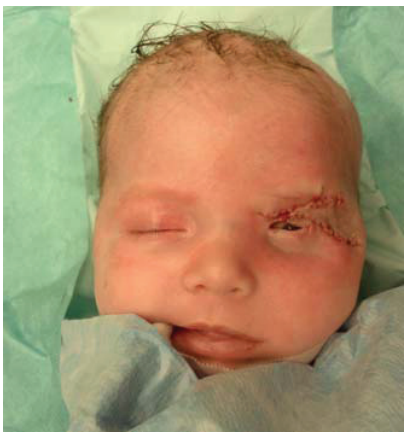

Fig. 6 Condition after reconstruction of upper eyelid

unit in 1978 (1, 2). Previously it had been confused with other oncological disorders according to its localisation. Its tissue origin has not been specified yet (2).
Determination of the diagnosis is difficult also with regard to its very low incidence (incidence rate of 0.1 per million infants per year – however, this number is considered to be underestimated precisely due to the difficulty

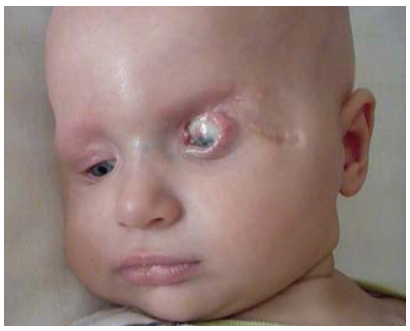

Fig. 7 Condition 10 months after surgery

of diagnosis). It is based above all on histology, immunohistochemistry and molecular genetics (2).
Imaging methods do not enable us to determine the diagnosis precisely. Due to the rare incidence of EMRT, the characteristic radiological features are not known. In our case it was not possible to differentiate haemangioma, sarcoma and EMRT on the basis of MR and ultrasound examinations.
Histopathological diagnostics are based on the presence of characteristic cellular characteristics. These are large oval to polygonal cells with a quantity of eosinophilic cytoplasm, a large vesicular nucleus with prominent nucleoli and filamentous cytoplasmic inclusions. From an immunohistochemical perspective, a positive immune response to vimentin, cytokeratin and epithelial membrane antigen is important (1).
Molecular genetics has detected a mutation of the gene INI 1/SMARCB 1. This gene codes protein, which has the function of a tumour suppressant. It is a component of the BAF complex, which plays an important role in cellular proliferation and differentiation, in cellular antiviral activities and inhibition of the formation of tumours. Defects of the INI 1 gene are linked to rhabdoid tumours. The INI 1 gene lies on the chromosome 22, in the locus 22q11 (3, 4). In our patient no known mutation was demonstrated by the available molecular genetic methods.
There is still no unified strategy of therapy for EMRT. Patients are treated according to various therapeutic protocols with regard to localisation

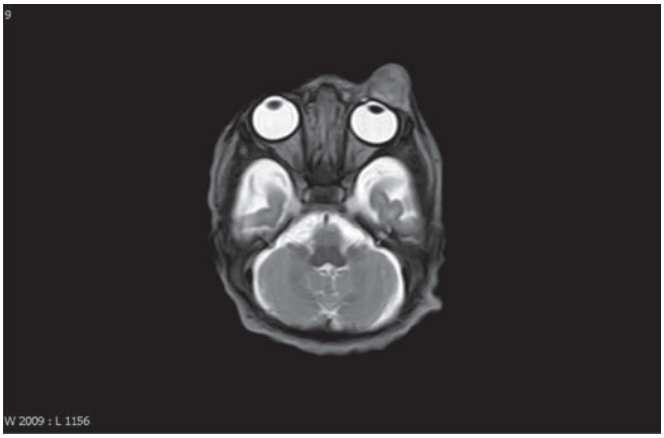

Fig. 8 MR of brain – 2nd day after birth, size of tumour 10 x 4 x 6 mm.

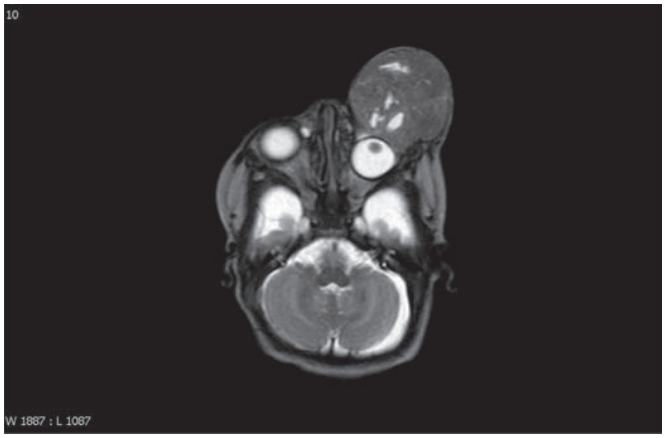

Fig. 9 MR of brain – 22nd day after birth, size of tumour 47 x 42 x 59 mm.

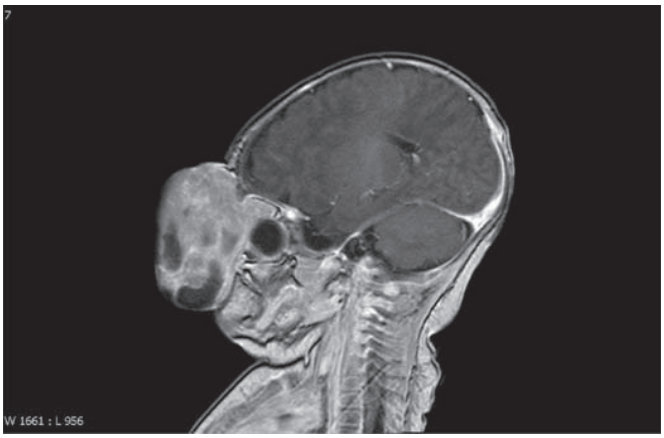

Fig. 10 MR of brain – 22nd day after birth.

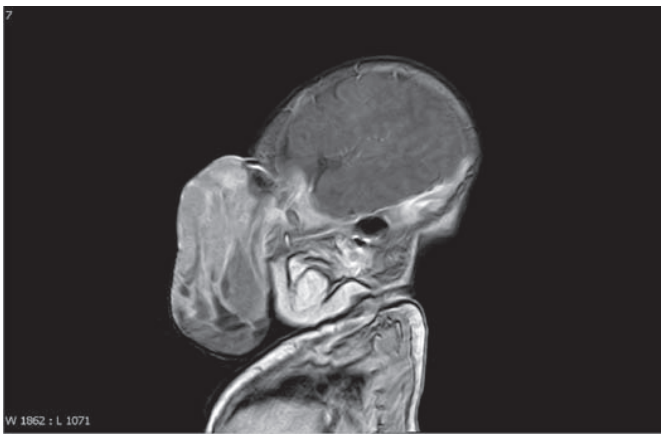

Fig. 11 MR of brain – 29th day after birth.

of the tumour. In the majority of cases, surgical resection, radiotherapy and chemotherapy are combined. Despite the combined therapy, the survival rate of patients with a malignant rhabdoid tumour of soft tissues within the first 5 years is less than 20-30%.

CONCLUSION

Extrarenal malignant rhabdoid tumour is a rare, aggressive tumour with an infaust prognosis. It may occur in any part of the human body. The efficacy of therapy is weak and the prognosis is unfavourable in comparison with other paediatric tumour disorders. In differential diagnostics of haemangioma and aggressive paediatric lesions, it is also necessary to consider rare diseases such as EMRT, because imaging methods are not yet capable of specific differentiation of this malignancy.

LITERATURE

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