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# Rhabdomyosarcoma and rhabdomyoma associated with nevoid basal cell carcinoma syndrome: Local treatment strategy


## Abstract

This article presents the case of a child presenting with a rhabdomyosarcoma associated with a fetal rhabdomyoma in the setting of nevoid basal cell carcinoma syndrome. Oncologic strategy is discussed.


## 1 | INTRODUCTION

Rhabdomyosarcoma (RMS) is the most common pediatric soft tissue sarcoma.[1] Fetal rhabdomyoma (RM) represents less than 2% of all primary skeletal muscle tumors.[2] Nevoid basal cell carcinoma syndrome (NBCCS), resulting from PTCH1 mutations,[3] is characterized by a wide range of developmental abnormalities and a predisposition to neoplasms. We present the first reported case of RMS associated with a fetal RM in the setting of NBCCS and discuss the oncologic strategy.

## 2 | CASE REPORT

A 1-month-old baby boy was referred for two lesions: a 4-cm-diameter retroauricular soft, mobile tumor and a 1-cm-diameter occipital tumor (Figure 1A). Cystic lymphangioma was suspected. The

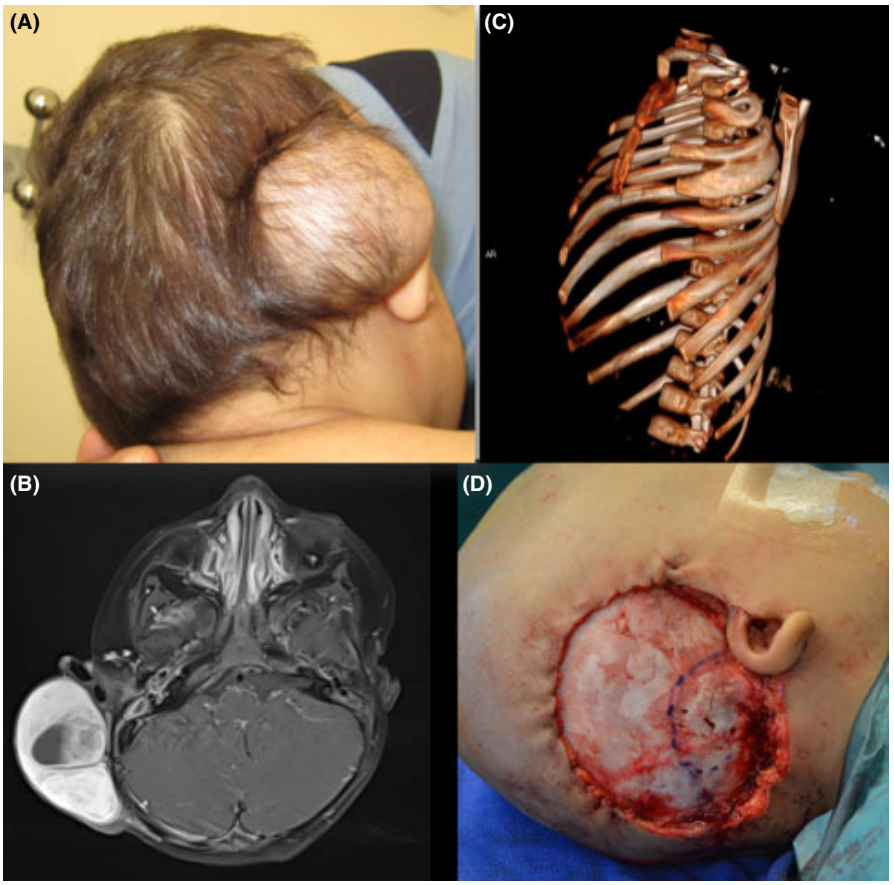


**FIGURE 1** A, Clinical lateral photograph of the tumor. B, Preoperative MRI, axial view, T1 sequence after gadolinium injection. The tumor enhanced the gadolinium. C, Thoracic CT scan, 3-dimensional reconstruction showing a fusion of the 2nd and 3rd left rib. D, Preoperative view of the surgical extension

  

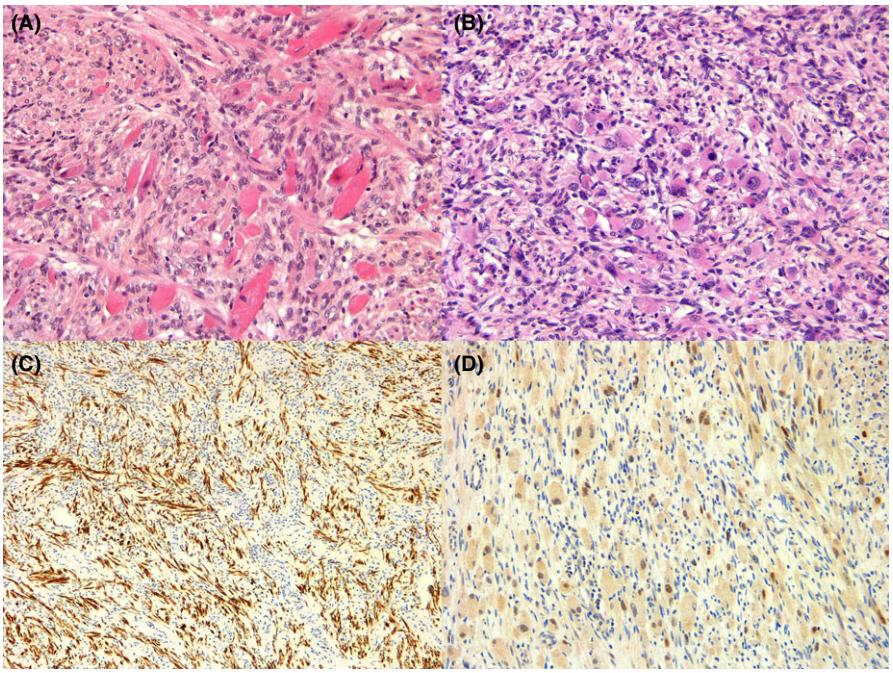


**FIGURE 2** Pathological and immunohistochemical examination. A, Nonalveolar rhabdomyosarcoma pattern observed in the central part of the mass, composed of atypical rhabdomyoblasts (HES ×400). B, Fetal rhabdomyoma pattern observed at the periphery of the mass, composed of well-differentiated rhabdomyoblastic cells and less-differentiated fusiform cells (HES ×400). C, Diffuse expression of desmin antibody in rhabdomyosarcoma pattern. D, 15% of the cells expressed myogenin in nonalveolar rhabdomyosarcoma

patient's 14-year-old brother had macrocephaly, rib and vertebral anomalies, multiple nevi, and dental malposition, but PTCH1 mutation was negative in 2007.

At the age of 20 months, the lesion enlarged. Magnetic resonance imaging showed three heterogeneous tissular tumors: two behind the ear and one on the back of the head. The tumor appeared inhomogeneous generally iso-intense with the brain in T1 and T2; of higher signal intensity on T2 weighted images in some areas; and enhanced after gadolinium injection (Figure 1B). Pathologic examination from surgical biopsy showed RMS on the retro-auricular lesion and fetal RM on the occipital lesion. Molecular biology did not show sarcoma specific fusion transcript. Diagnosis was nonalveolar RMS, Intergroup Rhabdomyosarcoma Study (IRS) III, associated with congenital rhabdomyoma.

Thoracic computed tomography showed rib fusions and posterior fusions of thoracic vertebrae (Figure 1C). The baby had macrocephaly (+2 standard deviation). Genetic examination confirmed NBCCS (deletion of exon 20 on PTCH1). The same mutation was discovered in his brother and mother.

The patient was included in the RMS 2005 protocol (European Paediatric Soft Tissue Sarcoma Study Group, a protocol for nonmetastatic rhabdomyosarcoma). Given the risk of radiotherapy with NBCCS, it was decided to surgically resect the initial RMS tumor volume (Figure 1D).

Pathologic examination showed a bizonal pattern, with RMS mainly in the central part. Atypical rhabdomyoblasts were intermingled with fusiform, stellate, and round cells. Cellular density varied within a predominantly myxoid stroma (Figure 2A, B). At the periphery were coalescent, well-circumscribed nodules of fetal RM. This lesion was composed of well-differentiated rhabdomyoblastic cells and less-differentiated fusiform cells. Immunohistochemistry showed diffuse expression of desmin in both lesions (Figure 2C). Approximately 15% of cells expressed myogenin in RMS and RM (Figure 2D). The proliferation index (Ki67) was approximately 10% in RMS and less than 5% in fetal RM. Surgical margins were complete. The patient remained in remission 2.5 years after diagnosis.

## 3 | DISCUSSION

Radiation therapy is classic treatment for RMS. In individuals with NBCCS, radiation therapy confers a high risk of early basal cell carcinoma[3] and should be avoided.

Surgery is a valuable treatment for local therapy in RMS.[4] The IRS group showed that survival depends on surgical margins[4] and International Society of Paediatric Oncology–Malignant Mesenchymal Tumor committee demonstrated that radiotherapy could be avoided when complete excision could be achieved.[5] Earlier diagnosis with neonatal biopsy could have reduced the extension of the surgical resection.

To our knowledge, this association had never been described in the literature. In children, the presence of two distinct tumors in the same patient should prompt consideration of genetic conditions and developmental anomalies.

We have also demonstrated the importance of radical surgery for NBCCS-RMS in avoiding radiotherapy and the risk of radiotherapy-induced tumor.

**Keywords**
gorlin syndrome, nevoid basal cell carcinoma syndrome, rhabdomyoma, rhabdomyosarcoma

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