Naevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin syndrome, is a rare, autosomal dominant disorder with systemic involvement (1). It is caused by germline mutations in the \( PTCH1 \) gene (locus 9q22.3–q31), which produce a tumour suppressor protein (2).

The prevalence of NBCCS varies from 1/30,000 to 1/256,000 (3), depending on the geographic location. NBCCS is characterised by multiple basal cell carcinomas (BCCs) which usually develop at a young age, palmar or plantar pits, odontogenic keratocysts that appear in the first, second and third decades (4) of life and ectopic calcification of the falx cerebri. However, many other anomalies, such as macrocephaly, hypertelorism, cleft lip and/or palate, skeletal and eye abnormalities have been described in patients with NBCCS. Several low-frequency neoplasms, such as medulloblastoma, meningioma, ovarian and cardiac fibroma have also been reported in these patients.

The diagnosis of NBCCS is based on clinical findings and familial history of the patient and can be made if 2 major and 2 minor criteria are met (4). Genetic counselling must be considered (3). Furthermore, during pregnancy, ultrasound scans can be performed to detect developmental malformations.

Patients with NBCCS usually present to a dermatologist because of skin lesions; however, in addition to a periodic dermatological evaluation, management of the disease may require a wide range of specialists such as dentists, cardiologists, oncologists, and orthopaedic surgeons.

**CASE REPORT**

A 22-month-old girl with a 2-month history of a thoracic cage mass (Fig. 1A) was referred to our institution for investigation and treatment of this lesion. Prior to surgery, she was referred to the dermatology unit for the evaluation of several cutaneous lesions previously diagnosed as melanocytic naevi.

At physical examination she showed multiple small, pigmented papules involving the face, trunk (Fig. 1D), and limbs. Macrocephaly, moderate hypertelorism (Fig. 1B) and palmar pits (Fig. 1C) were also observed. Furthermore, the patient’s mother presented with multiple facial lesions clinically consistent with BCCs, and reported that in the past she had undergone several excisions of BCCs and that she had been diagnosed as suffering from NBCCS.

Whole body CT scan showed no alterations in the CNS and ribs but revealed the presence of a solid oval shaped mass measuring \( 7 \times 4 \times 2 \) cm in the thoracic right cage with evident enhancement after contrast. The thoracic mass was excised. Histological examination showed proliferation of rhabdomyoblasts resembling either ganglion cells with vesicular nuclei and prominent nucleoli, or ribbon or strap-like cells with deeply eosinophilic cytoplasm and cross striations. Both types of cells had a benign appearance. These findings were consistent with fetal rhabdomyoma (Fig. S1D).

A total of 7 cutaneous lesions were also removed (one on the face, 4 on the trunk and 2 on the left hand). These lesions were detected by dermoscopy that revealed blue-gray globules.
inside all the lesions, as described by Feito-Rodríguez et al. (5), except for one on the left hand that had a round shaped cystic appearance. Histology analysis revealed 5 lesions that appeared to be basal cell carcinomas (Fig. S1A and B) corresponding to a tumour nodule located in the deep dermis. The nodule was encapsulated and composed of microfollicles lined by cuboidal or basa
dloid component, mimicking an eccrine adnexal tumour. The latter skin tumour was located in the deep dermis of the palm and was encapsulated and composed of microfollicles combined with cuboidal and basa
do cells.

Foetal rhabdomyoma is an extremely uncommon benign neoplasm with skeletal muscle differentiation that has already been reported in association with NBCCS (14). Quite recently a thoracic foetal rhabdomyoma has been reported in association with the mutation c.585-1G>A (15), as in our case.

The authors declare no conflict of interest.

REFERENCES