Case Report

Neonatal Paratesticular Scrotal Mass: A Rare Case of Atypical Neurofibromatous Neoplasm with Uncertain Biologic Potential

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Abstract

Atypical neurofibromatous neoplasm with uncertain biologic potential presenting as a paratesticular scrotal mass in a neonate with congenital giant melanocytic nevus is rare. Only one such case of neonatal scrotal neurofibroma has been reported earlier. We report an additional case and its management.

Keywords: Atypical neurofibromatous neoplasm with uncertain biologic potential, congenital melanocytic nevus, neonate, Paratesticular mass

Introduction

Neurofibromatosis type 1 is an autosomal dominant condition with a birth incidence of approximately 1:3000.[1] It presents as intraneural neurofibroma or plexiform neurofibroma depending on the site and the involvement of the nerve trunks. Small subset of these lesions undergoes malignant transformation. Growing, painful, or otherwise troublesome lesions are common indications for surgical excision or diagnostic biopsy. Based on the histologic features and immunohistochemistry (IHC) findings, the neurofibroma is classified as neurofibroma with cytologic atypia or hypercellularity, Atypical neurofibromatous neoplasm with uncertain biologic potential (ANNUBP) and malignant peripheral nerve sheath tumour (MPNST)[2] Although ANNUBP has been sometimes labelled as low-grade MPNST, they have been associated with mostly a low risk of recurrence and essentially no risk of metastasis. An unusual case of ANNUBP presenting as a paratesticular scrotal mass in a neonate with giant congenital melanocytic nevus (CMN) is discussed.

Case Report

A 13-day-old male neonate presented with complaints of dark pigmentation of the skin in the lower half of the body sparing the left lower limb and inguinoscrotal swelling since birth [Figure 1]. The swelling was firm and non-tender and both the testes could not be palpated separately from the swelling. There was no documented reporting of such swelling on any of the antenatal scans.

The nevus was found to be involving both the dorsal and ventral aspects of the body [Figure 1]. On the ventral aspect, it extended from below the umbilicus up to the right lower limb while sparing the left lower limb, and on the dorsal aspect, it was extending from below the lower half up to the right foot. There were also multiple satellite lesions in the bilateral upper limbs. The central nervous system examination and serum alpha fetoprotein levels were normal. An ultrasound (USG) study was done, in which there was hyperechoic mass in the right hemiscrotum and the right testis could not be separately visualised from the swelling. The left testis was noted to be in the left hemiscrotum.

USG of the abdomen showed normal study. The child was taken up for surgery for inguinoscrotal exploration. Intraoperatively, it was a firm mass closely adherent to the

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Figure 1: Pre-operative and intra-operative pictures. (a-c) Giant congenital melanocytic nevus with huge scrotal mass. (d-f) Intraoperative pictures showing mass separate from both testes

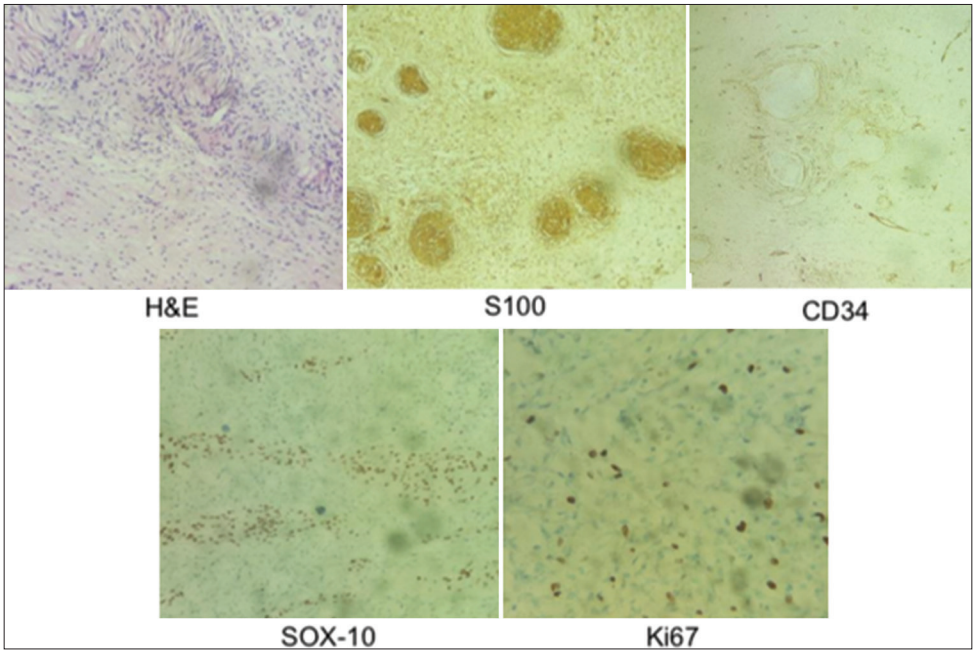


Figure 2: Histopathology and immunohistochemistry. IHC showing e/o atypical neurofibroma/atypical neurofibromatous neoplasm of uncertain biological potential (ANNUBP). H and E: e/o diffuse neurofibroma, S-100: Immunoreactive in tumor cells, CD34: Scattered immunoreactive in tumor cells, SOX-10: Scattered immunoreactive in tumor cells, Ki-67: 14%. 40X objective & 10X eyepiece (X400 magnification)

penile shaft and the scrotal skin. With due diligence, the dissection was done and both the testes were preserved. With the remaining skin, scrotoplasty was done [Figure 1]. Urinary catheter was removed on the 5th post-operative day.

Histopathological findings were suggestive of atypical neurofibroma/atypical neurofibromatous neoplasm of uncertain biological potential (ANNUBP) [Figure 2] while the skin lesion biopsy with IHC was suggestive of intradermal nevus. The child is under follow-up for 6 months and is doing well.

Discussion

Atypical neurofibromas and atypical neurofibromatous neoplasms of uncertain biologic potential (collectively AN/ANNUBP) refer to neurofibromas with atypical histological features.[2] They are

considered premalignant with an increased risk of progressing to a MPNST.[3] Nuclear atypia, hypercellularity, variable loss of neurofibroma architecture and mitotic activity are the histological characteristics to be seen on biopsy while in IHC, Ki-67, p53, S100, SOX 10 and CD 34 are looked for.[2]

Neurofibromatous tumours with nuclear atypia, hypercellularity, variable loss of neurofibroma architecture (e.g., herringbone or storiform fascicular growth and/or loss of CD34 positive network), and/or mitotic activity beyond isolated mitotic figures (>1/50 high power field [HPF] and <3/10 HPF) should be designated as having uncertain malignant potential when at least two of these features are present.[2] Labelling it malignant can lead to overly aggressive treatment. ANNUBP does not have a specific IHC/genetic testing but variable to complete loss of S100 protein/SOX10 expression, loss of the CD34-positive fibroblastic network, high Ki-67 (MIB1)-labelling index and loss of the CDKN2A locus at 9p21 could hint towards early malignant transformation. The presentation in our patient could be easily confused with the similar presentation of CMN with brain involvement which is then termed as neurocutaneous melanosis (NCM) when it meets certain criteria.[4] The difference between ANNUBP and NCM is histological and is based on the IHC and is important to make as ANNUBP with higher Ki-67 labelling index (>10%) may assist in the detection of malignancy in a neurofibroma. In our index case, the Ki-67 index was reported as 14% which indicates that the lesion might be premalignant.[2] Our patient had CMN, but IHC features showed features of ANNUBP.

Along with the dilemma in the diagnosis of ANNUBP, the management guidelines are also not well defined. Nevus and neurofibroma involving such large surface area of the body in a neonate limits the treatment options. In order to excise the firm mass, obtain the tissue for diagnosis and to achieve cosmesis, surgery is considered. In our case, we could also not palpate the testes clinically or identify it separately from the neurofibroma which was then found separate intra-operatively.

The occurrence of seizures in such patients warrants further investigations to rule out melanin deposition in brain parenchyma or meningeal involvement. The complete workup of such patients with the requirement of IHCs, good histological reports and magnetic resonance imaging (MRI) in a neonate could be challenging in resource-limited settings. The parents should be counselled well for regular follow-up for any late recurrences or any recurrent episodes of seizures for which the child should be further investigated. The parents were also counselled for the risk of similar lesion in future progeny and then the antenatal scans could be supplemented with fetal MRI to make informed decisions about the pregnancy.

Motiwala *et al*. reported a similar case of neurofibroma in a month old child.[5] However, no further IHC results were reported. Singh *et al*. reported the development of neurofibroma in a case of Giant Congenital Becker's nevus over back.[6]

What could be natural course of such cases of CMN? Goyal *et al*. reported a case of giant melanocytic nevus presenting as a huge vulval mass in a 15-year-old girl with features of a compound nevus on histopathological examination.[7] Elkiran *et al*. discussed the development of giant lymphoedema involving the scrotum and penile skin in a case of CMN.[8] Belysheva *et al*. described two cases of melanoma in patients having giant congenital melanocytic nevi, although these lesions did not affect the scrotum.[9] Because of late occurrences of such pathologies, long-term follow-up is mandatory in these patients.

Conclusion

Neonatal para-testicular masses in children suffering from CMN may be ANNUBP; IHC is indispensable for diagnosis. Although complete surgical excision is usually possible, long-term follow-up is needed due to uncertain malignant potential.

Patient consent

Consent was taken from the Institutional Ethics Committee and the parents of the child for using clinical pictures and data for publication.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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