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Case Report

Solitary intrascrotal neurofibroma in a child: A case report

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ABSTRACT

Background: Neurofibroma (NF) remains a rare entity among various scrotal masses in the pediatric age group. We report a case of solitary intrascrotal extra testicular plexiform neurofibroma in a 6-year-old child with the clinical management of this rare entity and review of the literature.

Case Summary: A 6-year-old male presented with incidentally noticed intrascrotal swelling. Clinical examination confirmed the presence of solitary intrascrotal swelling with normal testis. Radiological imaging showed the presence of a hypoechoic mass in the left hemiscrotum with normal both testes. Surgical exploration showed the presence of unencapsulated soft tissue mass and histopathological examination (HPE) confirmed the diagnosis of plexiform neurofibroma. Thorough evaluation ruled out features of neurofibromatosis 1 thus confirming the diagnosis of solitary intrascrotal extratesticular plexiform neurofibroma. Complete surgical excision resulted in satisfactory recovery with no recurrence on follow-up

Conclusion: Neurofibroma although rare should be considered as a differential in the diagnosis of an intrascrotal mass in the pediatric age group. The benign nature of the lesion and extratesticular origin usually makes testis sparing surgical excision feasible which remains the treatment of choice with excellent prognosis and minimal chances of recurrence. A thorough histopathological examination is mandatory to rule out neurofibromatosis.

Keywords: Neurofibroma, Intrascrotal, Children

INTRODUCTION

The presence of a scrotal mass in the pediatric age group is usually associated with parental and physi cian anxiety owing to a variety of possible pathologies both benign and malignant. Most pediatric scrotal tumors are mesenchymal in origin arising more frequently in extra-testicular tissue than in gonads. Among commonly reported pediatric scrotal masses like leiomyoma, fibroma, lipoma, hemangioma, and epidermoid cyst, etc., neurofibroma remains an extremely rare entity.[1] A review of available English literature suggests that only a handful of cases of isolated intra-scrotal neurofibroma has been reported to date with less than 5 cases reported in the pediatric age group with the youngest patient being 14 years of age.[1-8] Herein, we describe the clinical, radiological, pathological presentation and management of extra testicular intrascrotal isolated neurofibroma in a 6-year-old boy which appears to be the youngest patient to be reported in the literature to date.

CASE REPORT

A 6-year-old boy presented in a pediatric surgery clinic as a case of scrotal swelling noticed incidentally while taking a bath. There was no history of trauma,

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pain, or any surgical intervention in the past. On clinical examination, there was soft non-tender swelling in the left hemiscrotum approximately 5 cm in craniocaudal and 3 cm in transverse dimension with both testes good-sized and palpable separately. MRI showed the presence of heterogeneously enhancing intrascrotal extratesticular mass measuring about 5.5x3cm in size appearing isointense on T1 (Fig.1A) and hyperintense on T2-weighted images (Fig.1B).

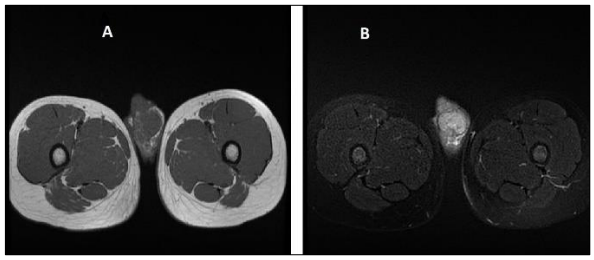

Figure 1: A: T1 image showing target sign and isointense scrotal mass. B: T2 image showing hyperintense scrotal mass.

The patient underwent scrotal exploration which revealed the presence of unencapsulated soft tissue mass in the left hemiscrotum separated from the testis and arising from scrotal soft tissue having extension in perineum with one of the nerves forming contents of mass (Fig.2).

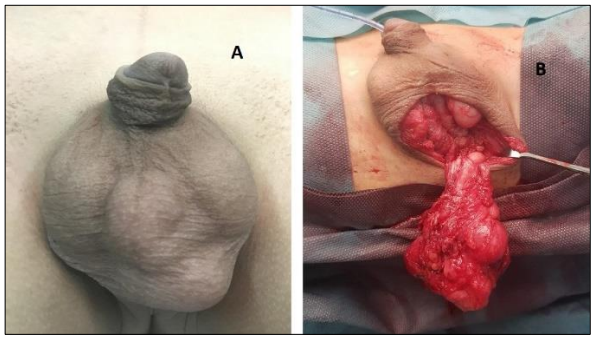

Figure 2: A: Preop image showing scrotal mass. B: Intraop finding showing unencapsulated mass

HPE reported soft tissue mass 53 gm in weight and 6cm in maximum dimension with two elongated cylindrical segments consistent with nerve segments are seen on one side of the resection. Microscopic examination of resected specimens showed the presence of a tortuous mass of expanded hypercellular nerve branches composed of spindle cells of Schwann origin confirmed by positive expression to S100 and SOX10 markers, while negative for desmin (Fig 3). Since the patient had features suggestive of plexiform neurofibroma, he was further evaluated for Neurofibromatosis type 1(NF1). The genetic, ophthalmic, dermatological, and neurological evaluation confirmed the absence of NF 1 thus confirming the diagnosis of isolated intrascrotal plexiform neurofibroma. Follow up after 1 year the patient was doing well with no evidence of recurrence clinically or radiologically.

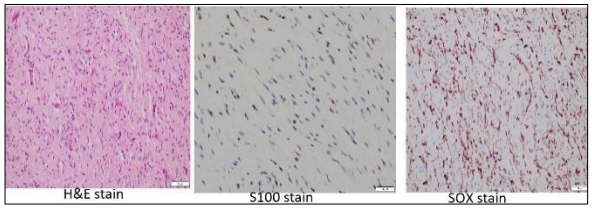

Figure 3: Micrograph of the tumor with bland spindle cells and loose collagenous stroma staining positive for S100 and SOX10 immuno histochemical maker

DISCUSSION

Neurofibroma (NF) is an uncommon benign tumor originating from the Schwann cells present in the nerve sheath.[1,2] NF present clinically either as a solitary or as multiple lesions with a systemic condition termed NF1 which is caused by a germline mutation in the NF1 tumor suppressor gene located at 17q11.2. [1,4,5] The World Health Organization classifies NF into two broad categories: Dermal and plexiform. Dermal NFs arise from a single peripheral nerve, while plexiform NFs are associated with multiple nerve bundles. [1,5.6] The solitary NF can occur in any region of the body but is commonly seen in the neck, thorax, cranium, retroperitoneum, and flexor aspects of the upper and lower extremities with genital NF being rare in all age groups. [1,4]

Neurofibromas are rare benign tumors among common benign scrotal masses like leiomyomas, lipomas, fibromas, hemangiomas, and epidermoid cysts. [1,5,6] Grossly neurofibroma remains unencapsulated tumors and soft in consistency.[1,5] Microscopically neurofibroma is characterized by the presence of combined proliferation of all the elements of peripheral nerve axons, Schwann's cells, fibroblasts, and perineural cells with Schwann's cells being the predominant cellular element. [1,2,5] The cells are arranged in a biphasic architecture made of Antoni A and Antoni B areas. Antoni A areas refer to dense bundles of spindle cells with palisading nuclei (Verocay bodies). Antoni B areas are characterized by loose-textured tissue, fewer cells, and myxoid changes.[7] Histological diagnosis is further confirmed by immunohistochemistry which shows uniform staining with S 100 protein.[1-6]

Clinically scrotal neurofibroma is usually extratesticular in location arising from tunics and subcutaneous neural tissue. As experienced in the present case, NFs are usually separate from the testis, vas deferens, and epididymis commonly originating from the genital branch of the genitofemoral nerve lying posteriorly to the spermatic cord.[6] Asymptomatic extratesticular scrotal mass remains the most common presentation among presenting symptoms

like hydrocele, painful scrotal swelling, and scrotal discomfort usually between 8-77 years of age. [1,5,6,8] The present case appears to be the youngest to be reported in the literature to date. The tumor can be solitary or plexiform in nature as a part of NF1. As experienced in present and two reported cases, isolated scrotal plexiform neurofibroma in absence of NF 1 remains an extremely rare entity. [1,8] The presence of plexiform nature of tumor as found in present case warrants thorough clinical evaluation to confirm the association of NF 1 in the form of cafe-au-lait macules, sphenoid dysplasia, Lisch nodules, optic glioma, positive family history, freckling of the axillary and inguinal region, long bone cortical thinning, or pseudoarthrosis. [1-6]

Ultrasound scan remains the gold standard in the evaluation of pediatric scrotal masses. Neurofibroma on ultrasound scan imaging appears as well defined, relatively homogeneous, predominantly hypoechoic masses with poor to moderate vascularization on color Doppler. [1,2] PET-CT although not usually recommended in the pediatric age group is often performed in adults and an absence of increased uptake of radiotracer confirms the benign nature of neurofibromas. [2,5,6] MRI is the modality of choice for the characterization of neurofibromas as it allows exquisite anatomical analysis of the lesion and its relation to the surrounding soft tissues. On MRI, neurofibromas usually appear as an isointense mass on T1W1 and hyperintense on T2WI in relation to muscle. The presence of a hyperintense rim and central area of low signal ("target sign") on the T2WI image is highly suggestive of neurofibroma. [2,5,6,8] On CT scan NF appears as well-demarcated masses with attenuation equivalent to skeletal muscle. [2]

Surgical excision with preservation of testis remains the treatment of choice for scrotal NF.[1,4-9] Since these lesions are usually unencapsulated and infiltrating the nerve fascicles so the parent nerve is usually sacrificed which might result in groin pain. Solitary neurofibromas which are not associated with NF1 are usually treated by surgical excision. As experienced in present cases most of these lesions are extra testicular and hence salvation of testis is usually feasible although orchidectomy has been reported in two cases one with intratesticular NF and the other with sharing blood supply. [6,10] Frozen section where available is usually recommended especially with advancing age of presentation. Malignant change may not be apparent in short-term follow-up. [1,4-8]

In conclusion, solitary neurofibroma needs to be considered in the differential diagnosis of a mass of the male inguinal canal despite the extreme rarity of this entity. MRI is the modality of choice to confirm the presence and to characterize a tumor in this location due to its ability to exquisitely depict soft tissues and its multiplanar capabilities. Since malignant degeneration of solitary neurofibromas and local recurrence after resection are extremely rare in the absence of NF1, surgical management is curative.

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