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Neurofibromatosis type 1 associated with multiple internal and external anterior abdominal wall defects. A case report

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ABSTRACT

Neurofibromatosis type 1 is a relatively common autosomal dominantly inherited genetic disorder. It is characterised by variable clinical manifestations, including café-au-lait spots, axillary or inguinal freckling, cutaneous neurofibromas, plexiform neurofibroma, bony lesions, optic glioma, and iris Lisch nodules, which constitute the clinical diagnostic criteria. In addition, a wide range of systemic abnormalities, including skeletal deformities, cardiovascular anomalies, neurocognitive deficits, as well as nervous system and non-nervous system tumours, have been described in patients with NF1. We present a previously unreported systemic association in NF 1 in an adolescent male: the presence of bilateral congenital hydrocele, divarication of rectus abdominis muscles and umbilical hernia, all external and internal defects of the anterior abdominal wall.

INTRODUCTION

Neurofibromatoses (NF) are a relatively common genetic disorders with autosomal dominant inheritance[7]. They are a heterogenous group of diseases with high predisposition to the development of benign and malignant tumours affecting different parts of the body[9, 21]. Three types of NF, neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2), and shwannomatosis, have been described[15]. NF1 is the most common type accounting for about 96% of the cases[14, 17]. It occurs in 1 in 3500 live births and is characterized by variable expression of clinical manifestations including café-au-lait spots, axillary or inguinal freckling,

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cutaneous neurofibromas, plexiform neurofibromas, bony lesions, optic glioma, and iris Lisch nodules[20]. A number of other abnormalities including skeletal deformities, cardiovascular anormalies, neurocognitive deficits, and several nervous and non-nervous system tumours have been described in patients with NF1[9]. However, to the best of our knowledge, co-existence of bilateral congenital hydrocele, divarication of rectus abdominis muscles (DRAM) and umbilical hernia in a patient with NF1 has not been previously reported in the English literature. One other example of such rare associations, albeit intracranial, reported from Africa not too long ago was that of pleomorphic xanthoastrocytoma in another boy with NF1[2].

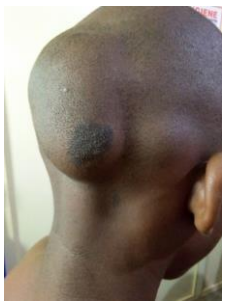
In this report we present an 11 year old boy with NF1 and a novel spectrum of systemic associations involving both internal and external anterior abdominal wall defects.

Case Description

An 11 year old boy first presented at our hospital aged 2 years with painless bilateral scrotal swellings. The swellings were first noticed about 18 months earlier and had been increasing in size progressively. He had been a product of term pregnancy given birth to at gestational age of 39 weeks and 2 days. The birth weight was 3.2kg. Examination at this first hospital presentation in childhood revealed non-tender, brilliantly transilluminating bilateral scrotal masses. In addition, there were a 2-cm separation of the rectus abdominis muscles, a 1-cm umbilical defect, and a 2cm right sided occipital mass. A clinical diagnosis of bilateral congenital hydrocele with divarication of the recti abdominis and umbilical hernia was made. He underwent surgical procedures of bilateral herniotomy and hydrocelectomy; while the divarication of the abdominal recti and the umbilical hernia were managed non-operatively.

The boy presented in our hospital 9 years later with a progressively enlarging occipital swelling (Fig. 1). The swelling was first observed at birth as a < 2cm painless occipital skin mass, and was the same one that had been documented at the first hospital presentation for the abdominal wall defects. The mass gradually increased in size over time till review, necessitating presentation because of the cosmetic blemish it had become. There was a paternal family history of dark pigmented skin patches and neurofibromas. Clinical examination showed a soft 10cm by 8cm by 3cm largely right sided, painless, occipital mass with overlying dark skin patch; it had no attachment to the underlying bone. He also had multiple cafe-au-lait spots over the skin of the limbs and the trunk (Fig. 2), as well as iris harmatomas. There were bilateral hairline inguinal scars (Fig 3). The DRAM had resolved spontaneously while the umbilical defect has reduced significantly (about 3mm residual defect).

A clinical diagnosis of plexiform neurofibroma in a patient with neurofibromatosis type 1 was made. He subsequently underwent excisional biopsy of the occipital mass which was histologically confirmed to be a plexiform neurofibroma (Fig 4).



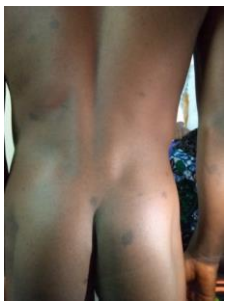


Figure 1. 10cm by 8cm occipital plexiform neurofibroma in our patient.

Figure 2. Photograph of the patient showing multiple cafe-au-lait spots.

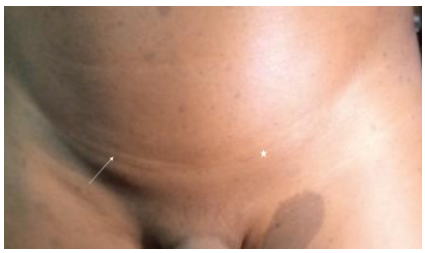


Figure 3. Photograph of the patient showing the well-healed scars of the previous bilateral herniotomy (arrow on the right, star on the left).

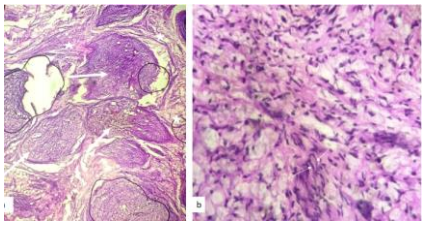


Figure 4. (a) low power (x4) photomicrograph of the lesion's histology specimen showing spindle cells (arrows) separated by fibrocollagenous stroma (stars). (b) high power (x40)

photomicrograph showing elongated spindle cells with wavy nuclei (arrows).

Discussion

Neurofibromatosis type 1 (NF1), the most frequent of the neurofibromatoses, is a relatively common genetic disorder[7, 15]. It is largely diagnosed clinically using well defined criteria[9, 12]. A wide range of abnormalities including cutaneous lesions, skeletal deformities, cardiovascular defects, neurocognitive deficits, as well as nervous system and non-nervous system tumours have been described in patients with NF1[9]. Not previously reported however, to the best of our knowledge, is the occurrence of multiple anterior abdominal wall defects both external and internal in a patient with NF 1. We present one case of such, an 11-year-old boy who was managed at our hospital at the age of 2 years for divarication of rectus abdominis muscles, umbilical hernia and bilateral congenital hydrocele but presented 9 years later with an occipital mass that was increasing in size progressively; surgical pathology of this lesion post-excision was reported as plexiform neurofibroma.

Neurofibromatosis type 1 results from a germline mutation in the NF1 gene, a tumour suppressor gene located on the long arm of chromosome 17 (17q11.2). It encodes neurofibromin which is a negative regulator of the Ras proto-oncogene, a key signalling molecule in cell growth and differentiation[8, 10, 18]. Divarication of rectus abdominis muscles (DRAM) is very rare and is usually congenital when present[11]. Congenital DRAM can occur as autosomal dominant transmission without associated syndrome or sequence; or as a part of some clinical syndromes like the prune-belly, Beckwith-Wiedemann, Simpson-Golabi-Behmel, and some other midline-defect syndromes[3, 4, 5, 16].

In contrast, the exact cause of umbilical hernias is unknown, although higher incidence has been reported in African, and Africa-American infants compared to Caucasians[1]. Greater risks of umbilical hernia has also been documented in premature and low-birth-weight infants, children with metabolic disorders like hypothyroidism; autosomal trisomies like trisomy 18 and 21; as well as syndromes like Beckwith-Wiedemann, Marfan and Simpson-Golabi-Behmel[22, 23]. As for congenital hydroceles, they result from failure of closure of the Processus Vaginalis, the exact pathogenesis of which is still a subject of controversy[6]. Familial inheritance[6, 13], and association of hydrocele with the FG syndrome (Opitz-Kaveggia syndrome)[19], have however been documented.

Apart from the clinically documented family history of NF1 in the patient presented in this report, we are not in the position to provide any other genetic / familial explanation for the associations with the internal and external anterior abdominal wall defects seen in him. Perhaps it is a mere coincidence or maybe there is an as yet unidentified genetic or familial predispositions. Most non-syndromic DRAM, paediatric umbilical hernia, and congenital hydroceles resolve spontaneously, surgery being indicated in the latter if the hydrocele persists beyond the age of 2 years. Our patient had surgery for the hydrocele at age 2 years and was managed non-operatively for the DRAM and umbilical hernia to a good outcome.

Plexiform neurofibromas develop in about 30-50% of patients with NF1 and carry a lifetime risk of malignant transformation[9]. Indication for surgery in plexiform neurofibroma include pain referrable to the lesion, functional compromise, and cosmetic disfigurement[20]. Our patient presented and had excision of his occipital lesion because of the latter reason.

Conclusion

NF1 is a relatively common inherited disorder with a wide range of associated anomalies. We have presented what must be a novel clinical spectrum of associations of NF 1 in an 11-year-old adolescent boy from Nigeria: a rare case of divarication of rectus abdominis muscles, umbilical hernia and bilateral congenital hydrocele in a child who fulfilled the clinical criteria for NF1. He was managed earlier in childhood for the associated abdominal wall lesions but presented 9 years later for the management of an occipital plexiform neurofibroma because of its cosmetic disfigurement.

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