EAS Journal of Orthopaedic and Physiotherapy
Abbreviated Key Title: EAS J Orthop Physiother
ISSN 2663-0974 (Print) | ISSN 2663-8320 (Online)
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-4 | Issue-4 | Sept-Oct, 2022 | DOI: 10.36349/easjop.2022.v04i04.002

Case Report

Congenital Pseudo-Arthrosis of the Tibia in Patients with Neurofibromatosis Type 1: Case Series and Review of Literatures

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Article History
Received: 10.09.2022
Accepted: 17.10.2022
Published: 26.10.2022

Journal homepage:
https://www.easpublisher.com

Quick Response Code

Abstract: The term congenital pseudo-arthrosis of the tibia is a misnomer, as it is usually not present at birth but clinically manifest within the first decade of life. It is nonetheless a rare disease with yet a poorly understood natural history having varying presentations, which possess a great challenge of both diagnosis and management. It is however, a documented fact that 50% of these patients have neurofibromatosis type 1 and thus, a careful study and understanding of this condition as it present in patients with neurofibromatosis type 1 will also expose more about the disease entity itself, aid clinicians in making early diagnosis and tailor down a definitive treatment scheme which will help in limb salvage and prevent subsequent limb amputations amongst other complications. Here, we present a case series of two patients each with features of neurofibromatosis type 1, presenting with pseudo-arthrosis of the tibia. First patient is a 14-year old girl who presented with right leg deformity, multiple skin freckles and café au lait macula lesions on the body. Lower limb deformity was noticed since patient was 6-years of age following a minor fall at home, she has had multiple trial of local splinting of the limb and conservative non-surgical management of the limb deformity/fracture also in a peripheral hospital which has also failed and the deformity has persisted since then, she subsequently develops freckles which was noticed initially from the hand and progresses to cover the whole body including the palms and sole of the foot. On examination, she was stunted for age, not in any distress, anicteric not pale, with freckles covering the whole body, and macula café au lait spots around the upper back and thoracic region. There is an obvious deformity of the right leg, pseudo-arthrosis above the ankle joint and the right foot is under developed compared to the left foot. The second patient is a 4-year old.

Keywords: congenital pseudo-arthrosis, clinically manifest, diagnosis.

INTRODUCTION

Congenital pseudo-arthrosis of the tibia is a rare condition with an iceberg phenomenon as it houses behind it high probability of association with neurofibromatosis type 1, as high as 50% of those with congenital pseudo arthrosis of the tibia have neurofibromatosis type 1 [1, 2], so although this condition is uncommon, with an incidence varying between 1:140 000 and 1: 250 000 [3] the burden and challenges of managing this condition on the background of neurofibromatosis type 1 with all its antecedent complications is enormous [3] and delay in diagnosis and management can lead to myriads of problems including limb length discrepancy, mechanical axis deviation, close joint stiffness and pathological fractures all of which can ultimately cost the patient the limb [3-5]. Two patients present to our facility, each of which has spent years managing a tibia fracture that has refused to heal, both of which also has features of neurofibromatosis type 1 evident by multiple skin freckles, café au lait spots and lisch nodules in the eyes. Although rare and uncommon, these patients are still present and presents to our facilities. It is therefore paramount for clinicians' index of suspicion to be raised in this conditions in other to achieve earlier diagnosis and prompt definitive intervention so as to prevent the cascading event that follows late diagnosis and institution of management which can cost the patient a great amount of money in failed attempt at treatment and the depressing possibility of limb loss.

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CASE SERIES

A 14-year old girl who presented with right leg deformity, multiple skin freckles and café au lait macula lesions on the body. Fig. 1 Lower limb deformity was noticed since patient was 6-years of age following a minor fall at home with subsequent inability to use the affected limb, there was no history of any noticed discrepancy or abnormal bowing of the limbs prior to the event of the fall and no history of deformity in any of the other limbs or inability to use any of the other limbs. No history suggestive of abuse or non-accidental trauma. She has had multiple trial of local splinting of the limb at home by traditional bone setters (TBS) which have all failed. The persistence of the limb deformity necessitated her presentation to some hospital facilities where conservative non-surgical management of the limb deformity/fracture with cast was also attempted, with no satisfactory outcome. She was noticed to have freckles which were said to have begun initially from the hand and has progressed to cover the whole body including the palms and sole of the foot Fig. 1- 2. There was no history of nodular lesions noticed on the body, no history of easy bruisability or fractures in other parts of the body, patient was not on any medications and has no history of any known drug allergies. On examination, we found a girl that was stunted for age, not in any distress, anicteric, acyanose not pale, with freckles covering the whole body but more around the axilla and pubic region, there were macula café au lait spots around the upper back and thoracic region. On musculoskeletal examinations, patient has an obvious deformity of the right leg, pseudo-arthrosis above the ankle joint and the right foot is under developed compared to the left foot Fig. 3-4. Ophthalmology review reported lisch nodules observed in both eyes. The review of the X-ray shows the pseudoathrosis on the distal tibia and fibula, both of which appear thin and atrophic with pointed distal fragments Fig. 5-6.

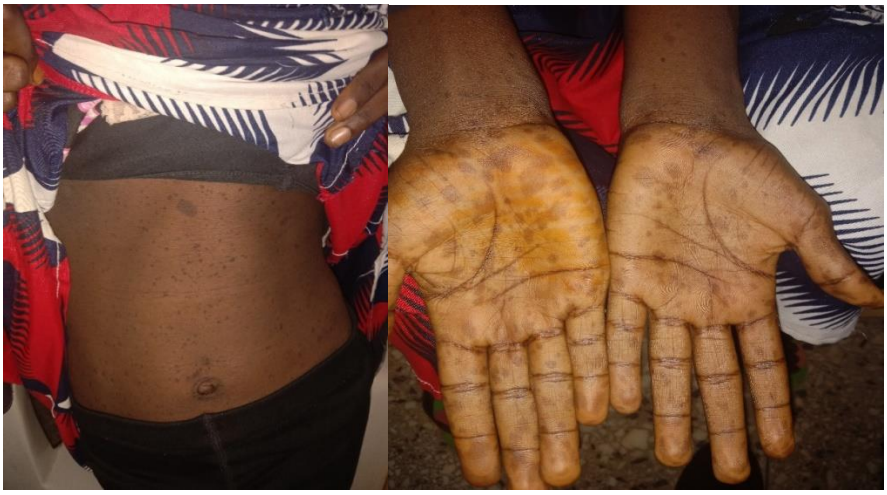

Fig. 1-2: Clinical photograph showing the freckles and café au lait spots on the body and palms of the patient

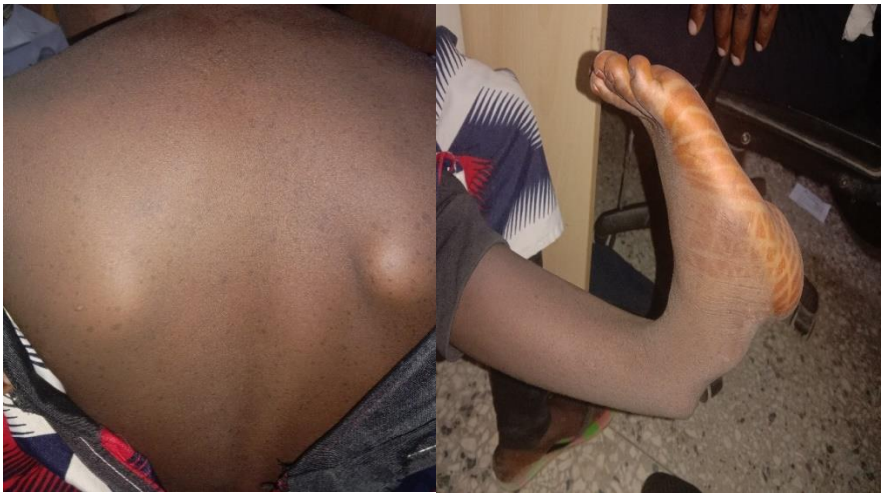

Fig. 3-4: Clinical photographs showing the back of the patient with freckles and the pseudoarthrosis of the distal tibia and the less develop limb

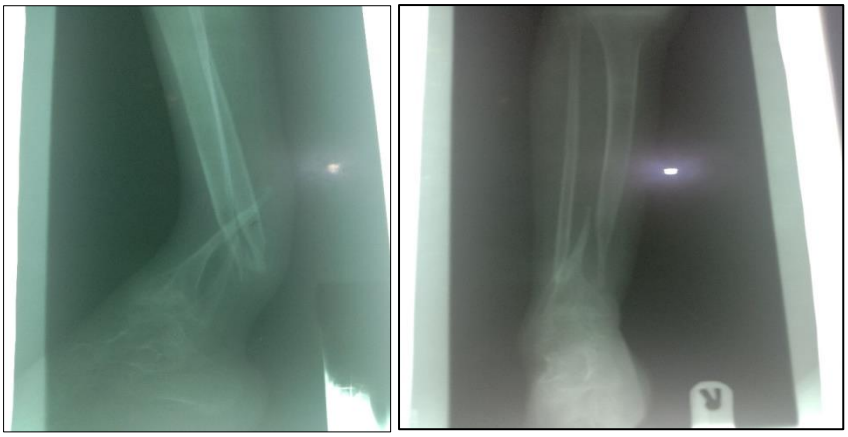

Fig. 5-6: X-ray showing distal fracture of the tibia and fibula, with associated pseudoarthrosis

DISCUSSION

Congenital pseudoarthrosis of the tibia (CPT) is a rare disease that is usually intimately related to neurofibromatosis type 1, as much as 40-80% of this patients have the condition [6] and such children affected are noticed to show some early features such as anterior lateral bowing of the tibia (kyphotic tibia) which may progress to pathologic fracture, the pseudoarthrosis and higher risk of refracture even after initial union has been achieved [7]. The tibia will also show some areas with segmental dysplasia, this dysplasia has been implicated in the initial bowing seen on the tibia, the nonunion and reduced growth in the distal tibial epiphysis that subsequently result in shortening of the limb [8]. Other features noticed of the affected tibial bone are the marked reduction in mechanical strength and osteogenic capability [9]. There is a higher cellular and fibro vascular tissue growth at the expense of the bony cortex, this promotes in-coordinate osteoclastic bone resorption and a reactive change that occurs simultaneously also at the medullary aspect leading to deposition of excess trabecular bone and medullary sclerosis [10] the major histopathologic changes found are highly cellular fibro vascular tissues and paucity of vascular growth [11], some have suggested that the pathologic features are more on the periosteum than the bone itself [12]. There are many other theories that try to explain this condition, many of which are either discarded or not fully proven; vascular insufficiency propagated by Codvilla, genetic defects by Gaenslem, nutritional deficiency by Henderson, endocrine imbalance by Duraiswami and infection during pregnancy by Pagella etc. Andersen has tried to classify this condition based on the radiological findings noticed before the pseudoarthrosis arose and their response to treatment- into cystic type, dysplastic type, clubfoot type and sclerotic type [13]. The clubfoot type has been also noticed to have the most favorable prognosis [14], while union occurred most among those with sclerotic type, refracture has also been found to be more amongst this group. The dysplastic types have the poorest results, and those cases in which the fracture of the fibula has not occurred before their first operative procedure have better prognosis that those that have [15]. Boyd *et al.,* identifies six different types of congenital pseudoarthrosis of the tibia; type I are those with anterior bowing and tibial defects, type II have hourglass constrictions of the tibia, type III are those with observed bony cysts while in type IV there is pseudoarthrosis with sclerotic segments and march fracture. Type V has in addition to findings in type IV a dysplastic fibula, and in type VI, an intraosseous neurofibroma or schwannoma is obvious [16]. Currently the most frequently used classification system is that of the Crawford [17] which has the advantage of being descriptive and also being able to identify the different stages of the disease. There are four types here viz: type I, with anterior bowing and increase in cortical density with a narrow medulla, type II has anterior bowing and narrow sclerotic medulla, type III has anterior bowing in association with a cyst or sign of fracture while type IVS has anterior bowing with a clear fracture and pseudoarthrosis often involving both the tibia and fibula. The numerous classification systems noticed in this disease condition is also a pointer to its marked heterogeneity and the difficulty in summing up etiology, natural history and treatment of this condition in one classification [18]. CPT becomes conspicuous in a child at his first year of life. Although, Andersen has described a rare type with late onset which develops in a tibia that was normal at birth but start showing anterior lateral bowing between the ages of 4 and 12 [8]. Age of the patient at surgery is a clear factor in the rate of healing achieved. Joseph *et al.,* got consolidation in as high as 92% of patients treated under the age of 3-years old whereas patients after the age of 3-years he got 72% [19]. Gouron *et al.,* and Pannier *et al.,* have even reported bone union in patients as young as 14-months of age [20, 21]. The perculiarities of the patients in this case series is that, all presented after the age of 4 years with NF type I, hence they all had Crawford type IV. It is therefore paramount for clinicians in our environment to be well informed on the reality of the presence of this condition despite its rarity. A higher index of suspicion is required to be able to arrive at an early diagnosis, so as to prevent all the complications

that comes with delay in treatment and proffer a better prognosis and chance of recuperation for the child.

CONCLUSION

CPT though rare, is however a disabling condition in children with a challenging track of management, seen more commonly among those with NF type 1. The high variability in its presentation makes it difficult to pave a natural history and a routine pathway of presentation that can aid in earlier diagnosis, thus, clinicians are required to have a high index of suspicion especially among children presenting with features of NF type 1 in other to arrive at its diagnosis earlier enough for an intervention by the orthopedic surgeon to commence in good time.

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Citation: Ibrahim S, Musa A, Stephen Yusuf, Timothy E, Aremu A (2022). Congenital Pseudo-Arthrosis of the Tibia in Patients with Neurofibromatosis Type 1: Case Series and Review of Literatures. *EAS J Orthop Physiother*, *4*(4): 26-29.