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An Unusual Skin Lesion Presenting To an Orthopaedic Surgeon.

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ABSTRACT

Neurofibroma is genetic disorder characterized by tumorous growth from the neural crest cells. They not only involve various systems of interest to departments like neurological, dermatological, orthopaedic, pediatric but their manifestations also slowly evolve with different manifestations presenting at different ages. Thus they present difficulty in diagnosis and management. Here, we report a case of neurofibroma that had no specific orthopaedic symptom except a swelling.

Keywords: skin lesion, othropaedic, neurofibroma

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Case Report

A 13year-old boy (a school student) came to our outpatient clinic with complaints of a painless swelling over his right ankle. He had neither pain anywhere over his limbs nor had any difficulty in micturition. His past medical history revealed no specific chronic disease. Earlier his parents took him to a physician who diagnosed this case as a vascular malformation and advised drugs and preferred a biopsy. The boy had no hearing or learning disability. On examination, the boy's eyes were normally spaced and he was normally built and nourished to his growth proportion .His blood pressure was 110/70 mm of Hg. His physical examination revealed two soft swellings one above the other in the lower lateral aspect of right leg with diffuse margins Figures 1 to 3.The upper swelling was smaller than the lower one, both merging together smoothly. There was no warmth or tenderness over these swellings. They were passively movable from side to side (figure 4), but not up and down (figures 5 and 6). On moving the proximal smaller swelling the lower swelling also moved suggesting that these two arose from the same longitudinal structure,the right sural nerve There were no deformities in the spine or lower limbs. He had both his testes descended well into the scrotum. His secondary sexual characters were proportionate to his age..There was no neurological deficit of the limbs.His examination after undressing revealed that he had multiple café au lait spots of sizes 1.5 cm to 6 cm involving the front and back and sides of the trunk and in the upper and lower limbs also (figures 7, 8.9 and 10). The physical status of the patient is presented in figure 11.

Routine haemogram and biochemical tests were within normal limits. His right ankle X-ray revealed soft tissue shadows extending from lower fibular area to near the lower end of the lateral malleolus and the right lower fibula was found to be slightly bowed (fig.12 and 13). There was no destruction of the fibula or lower tibia. MRI of his right ankle revealed increased soft tissue component marked in arrow in figures 14, 15 and 16.

There were few bright lesions in MRI of the patient's brain (figure 17) which suggest an aberrant myelination, which are pathognomonic to NF1.The child was diagnosed to have NF1 by clinical means by the diagnostic criteria. [1] He was sent for ophthalmic examination which revealed that he did not have any abnormality like Lisch nodules. After we were sure that the boy had type 1 neurofibroma, no biopsy was done and we counseled to his parents to bring him for a regular follow up and to report if he develops danger signs like incontinence of mictrurition, weakness of limbs or visual disturbances. Since the child was comfortable and had no symptoms pertaining to the swelling, his parents were given the option of excision of the ankle neurofibroma only if it gives problems.

Discussion

Many people with NF1 from India especially from rural background tend to over look it until any systemic involvement is there. In this child, the time of occurrence of the clinical manifestations correlated with the general timeline of such occurrences. For example at 13 years he had café au lait spots (99% of which occur between birth to 12 years) and cutaneous fibromatosis (99% of them occur after 7 years). [1] Also as per guidelines [the] boy was not subjected to any biopsy of the swelling[1]. A definite instruction was given to his parents to bring him every 6 months for a regular follow up or at any time in between if he develops a specific complaint like mental development and performance in studies, visual symptoms, normal pubertal development, appearance of new neurofibromata, spinal deformity [1].

We have also considered the other differential diagnosis of this presentation. The size and the number of café au lait spots is useful only for diagnosing but has no bearing on the severity of the disease. Since there are many manifestations of this disease and there are varying degrees of expression at different ages, the best practices of management is yet to evolve [2]. The patient did not have any other feature other than the café au lait spot . he had normal genital, absence of pulmonary stenosis or retarded growth or deafness [3]. A clinical diagnosis can be only considered if lentigines occurs with two other symptoms like ECG abnormalities and ocular hypertelorism Or if there was no lentigines, three other above conditions are found along with a first-degree relative with a clinical diagnosis [4]. This assumes significance since acute myelogenous leukemia is reported in these patients [5]. While there are also instances when only after evaluation of their parents of the leopard syndrome patients found to have NF1 [6].

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In lips and oral mucosa our patient had no café au lait spots and submucosal neuromas clinically ruling out Peutz Jegher syndrome [7,8] and MENs2a respectively [9]. Since there was a localized soft tissue involvement pertaining to the lower leg lateral aspect, we also thought about the possibility of a Klippel–Trénaunay syndrome [10].

Neuro fibromin is a(tumor suppressor) suppresses Ras GTPase proteins. Hence when neurofibromin is reduced, unopposed activity of the cells increases the growth of cells leading to NF1. In small animal models apart from external manifestations of tumours, learning disability was also produced with suppression of neurofibromin [2]. In such animal models, where neurofibromin is suppressed, cholesterol rate limiting enzyme inhibitor – lovostatin, was found to reverse these attention deficits and visual spatial impairment [1].

These NF1's cutaneous neuro fibromas were shown to contain axons, Schwann cells , fibro blasts, perineural cells mast cells and increased extra cellular matrix. Since there is no specific marker for the perineural cell, its distribution is difficult to judge. Recently an analysis of the tight junctions in cell interfaces found claudin -1 to be a marker of perineural cells [11].

In cases presenting early especially in pediatric and young adult patients with NF 1, lumbar spine bone mineral apparent density (LS BMAD) was more affected than femoral neck bone mineral apparent density (FN BMAD) or whole body (bone mineral content (BMC)/ height [12]. The bright lesions in MRI of brain (formerly called UBO -unknown bright objects) appeared in the predicted age of 8-16 years in this child and are caused by aberrant myelination and are also pathognomonic to NF1[1].

Though it is a case of neurofibroma with no atypicality, to those who are familiar with the various skin presentations, the interesting angle to the presentation is the point that this was not looked upon as a neurofibroma by the previous physician. He diagnosed this case as a vascular malformation. . Clinical feature of NF1are known, what is new is that it presented to orthopaedic surgeon since the skin was not seen at first instance after undressing. The telltale evidence of café au lait spots might have the diagnosis clear at the earlier instance.

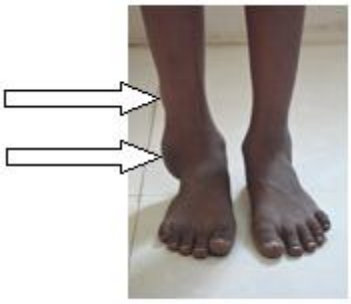

Figure 1: The front view of the right ankle and foot showing the two swellings marked by arrows

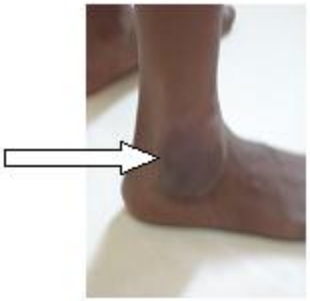

Figure 2: The lateral view of the right ankle and foot showing the swellings marked by arrow

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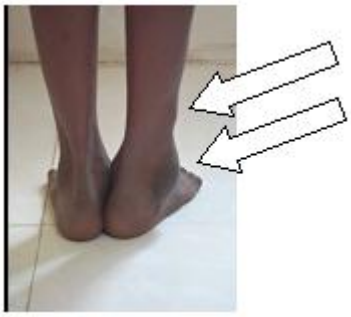

Figure 3: The view of the right ankle and foot from behindshowing the swellings marked by arrow

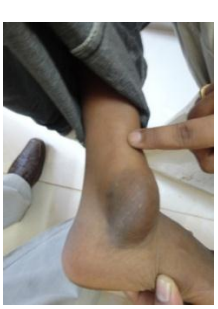
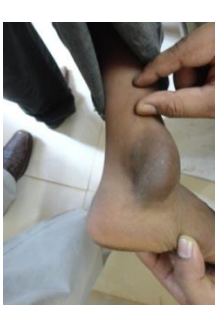
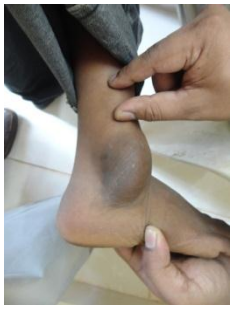

Fig 4 **Fig 5** **Fig 6**

In figure 4, the upper swelling is pushed from side to side.
In figures 5 and 6 the upper swelling is pushed up and down.

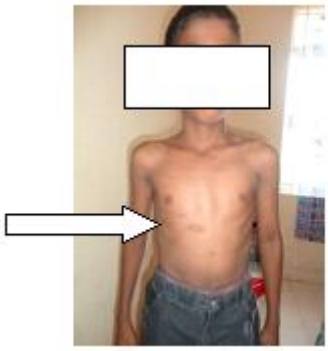

Figure 7: The clinical picture of the patient showing café au lait spot in fornt of the chest with arrows pointing to it.

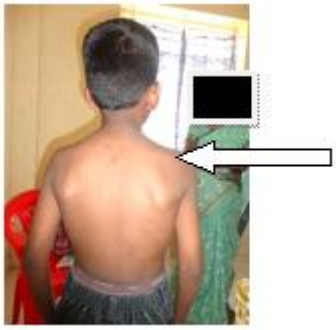

Figure 8: The clinical picture of the patient showing café au lait spot in his back with arrows pointing to it.

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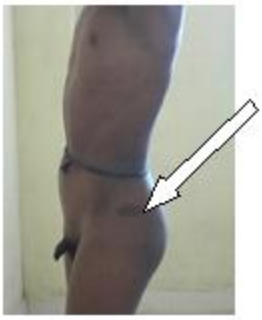

Figure 9: Café au lait spot on the lateral aspect of the left thigh marked with X-rays

Figure 10: Café au lait spot on the anterior aspect of the right thigh marked with X-rays

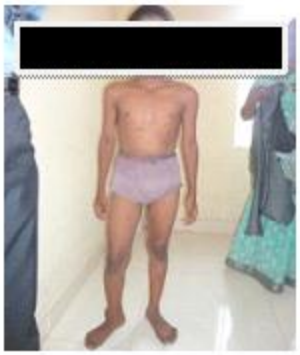

Figure 11: Clinical picture of the patient standing

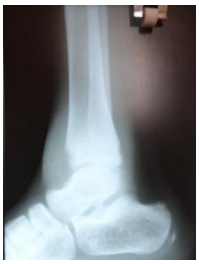

Fig 12

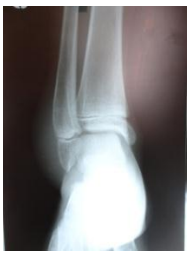

Fig 13

Figure 11 and 12 shows lateral and antero posterior views of the patient's right ankle.

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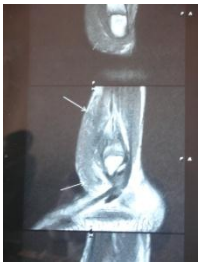

Figure 14: LS of the patient's right lower leg showing the soft tissue component in the anterior aspect marked with arrows.

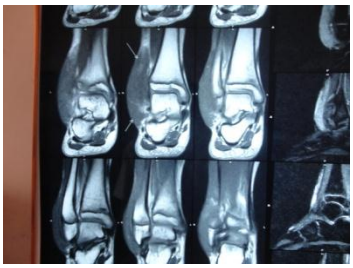

Figure 15: CS of the lower leg showing the soft tissue component in the lateral aspect of the patient's right ankle.

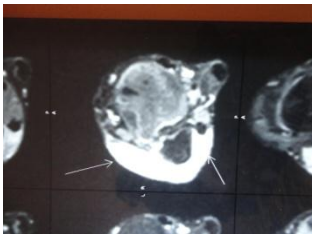

Figure 16: TS of the lower right leg of the patient showing the soft tissue component in the anterolateral aspect marked with arrows.

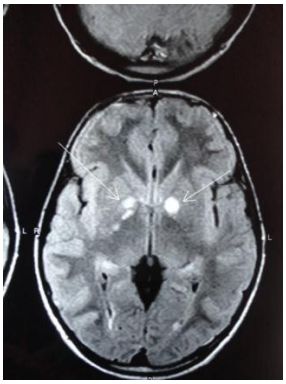

Figure17: The patient's MRI showing the bright lesions marked with arrows.

CONCLUSION

Most of the findings in the case reported concur with the literature. NF-1 is well known to be associated with bone deformities. This case report shows that NF1 with typical skin lesions can even present primarily to an orthopedic surgeon .As varying degrees of expression happen at different ages of the same patient, a thorough knowledge of the diagnostic criteria of NF1 is also a must and unnecessary biopsies and temptation of interfering should be resisted. Also proper undressing of the patient is essential to for the diagnosis of NF1.

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REFERENCES

[1] J Med Genet 2007 ; 44(2): 81-88
[2] Williams VC, Lucas J, Babcock MA, Gutmann DH, Korf B, Maria BL . Pediatrics 2009;123 :124-133
[3] Gorlin RJ, Anderson RC, Blaw M. Am J Dis Child 1969;117(6): 652–62.
[4] Voron DA, Hatfield HH, Kalkhoff RK. Am J Med 1976;60(3): 447–56.
[5] Uçar C, Calýskan U, Martini S, Heinritz W. J Pediatr Hematol Oncol 2006;28(3):123.
[6] Digilio MC, Sarkozy A, de Zorzi A et al. Am J Med Gen 2006;140(7):740–6.
[7] James, (mucosa cafeau lait)William; Berger, Timothy; Elston Dirk. 2005. Andrews' Diseases of the Skin: Clinical Dermatology (10th ed.). Saunders. p. 857. ISBN 0-7216-2921-0.
[8] Bouquot Jerry E, Neville Brad W, Damm Douglas D, Allen Carl P. 2008. Oral and Maxillofacial Pathology. Philadelphia: Saunders. p. 16.11. ISBN 1-4160-3435-8
[9] Pujol RM, Matias-Guiu X, Miralles J, Colomer A, de Moragas JM. J Am Acad Dermatol 1997;37(2 Pt 2): 349–52.
[10] James, William Berger Timothy, Elston Dirk. 2005. Andrews' Diseases of the Skin: Clinical Dermatology (10th ed.). Saunders. p. 585. ISBN 0-7216-2921-0.
[11] Pammi KP, Aho HJ, Laato MK, Peltonen TK, Peltonen SA . J Histochem Cytochem 2006; 54(1):53-61
[12] Lodish MB, Dagalakis U et al. Endocrine Rel Cancer 2012;19:817-25