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Case Report

Intraosseous Haemophillic Pseudotumour of distal digital phalanx – A rare imperceptible diagnosis

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ABSTRACT

Introduction: Digital distal phalanx lytic lesion is a rare presentation, posing diagnostic dilemma due to its varied etiology. Haemophillic intraosseus pseudotumour is one of the rare etiology causing such lesion. It is present in <2% cases with severe haemophillia. Case Presentation :We report a case of 13-year boy without any past history of bleeding dyscrasias, who presented with a fungating mass of right thumb and lytic &expansile lesion of distal phalanx of thumb on X-ray. On routine investigation he was diagnosed with mild haemophillia A. The patient was managed conservatively with Factor VIII level replacement and daily dressingusing tranexamic acid locally. Patient recovered completely in 3-months duration with good functional outcome. Conclusion: Swelling with lytic lesion in distal phalanx can be first presentation of mild Haemophillia without any previous history of bleeding dyscrasia. In such cases obliviou biopsy should be avoided.

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Introduction

Digital distal phalanx osteolytic lesion with swelling is a rare presentation. Neoplastic disorders like enchondroma, keratoacanthoma, epidermal inclusion cyst, glomus tumor, GCT and metastasis can present with such lesion along with rare etiologies like pyogenic osteomyelitis, tubercular osteomyelitis, hyperparathyroidism and renal osteodystrophy [1,2]

Intraosseous haemophillic pseudotumour, though an uncommon yet serious complication of both hemophilia A and B, can also present as an osteolytic lesion and mimic neoplastic or infective etiologies.[3]Mis-diagnosing such lesion may lead to avoidable catastrophic outcomes.
We present a similar rare case of haemophillic intraosseous pseudotumour of distal digital phalanx of thumb which developed as fungating mass secondary to oblivious biopsy though managed judiciously but still faced the brunt of misdiagnosis.

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Case Report

We present a case of a 13-year-old male patient presented in our OPD with the complaint of pain, swelling and fungating mass on the right thumb. Patient gave history of trivial trauma 3 months back following which he developed pain and swelling on right thumb for which he consulted local practitioner. Initially he was diagnosed as case of soft tissue infection and was given antibiotics.

Patient symptoms persisted despite antibiotic course hence plain X-ray handwas done by local practitioner which showedlytic lesion of distal phalanx of thumb (Fig 1), due to which initial diagnosis of neoplastic lesion was made by the local practitioner.

Without detailed haematological investigation, a biopsy was done to confirm the diagnosis. Unfortunately, swelling got converted to fungating mass secondary to unwarily done biopsy which was without keeping haemophilic pseudotumour as one of the differential diagnosis. Furthermore, the patient was advised amputation as the treatment option because biopsy came out to be inconclusive.

About 3 months after the first symptoms,the patient presented to our hospital with ulcerated fungating mass of size 4x4 cm over the terminal part of right thumb with areas of necrosis and scarred tissue. Proper history was taken specially to rule out constitutional symptoms like weight loss and history of smoking and other vascular/bleeding disorders. Local examination revealed tender mass with normal temperature with irregular margins, with no peripheral erythema without active bleeding.

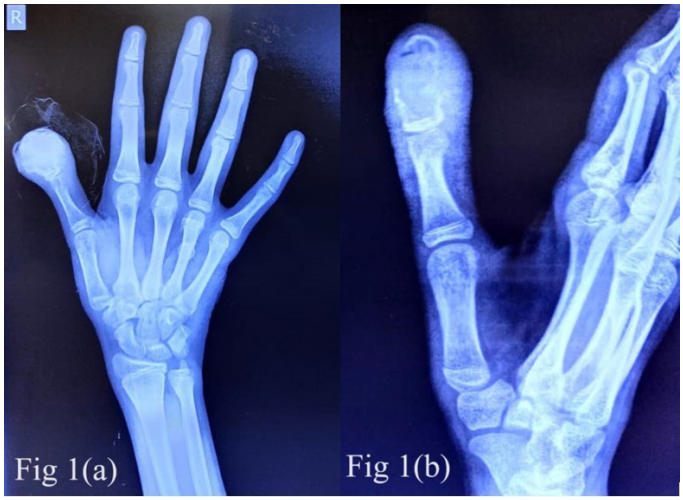


Figure 1 : Radiograph showing lytic lesion of distal phalanx at initial symptoms (a) AP and (b) Lateral view

Fresh X-rays done at our hospital revealed much more destruction, lysis and expansile corticle margins of distal phalanxsoft tissue shadow over it(Fig .2)

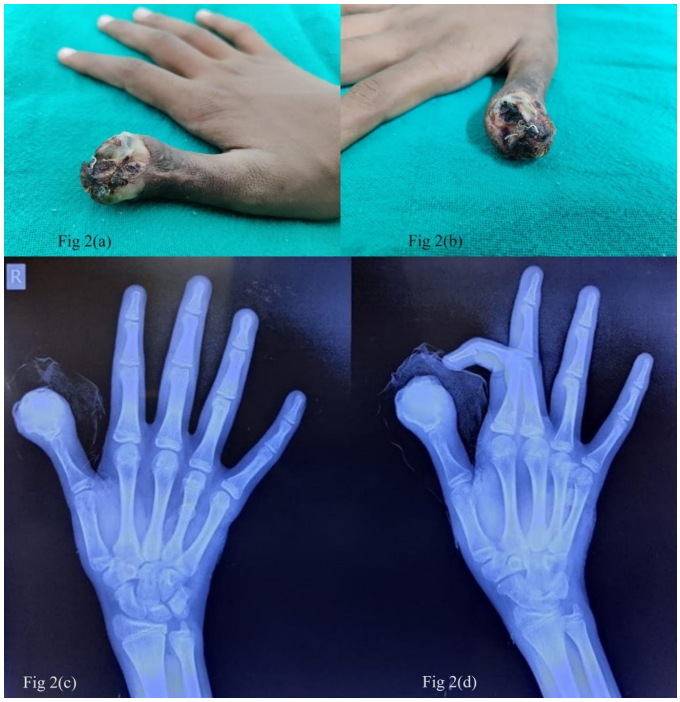


Figure 2 : Post-biopsy 3 months after initial symptoms showing clinical images having fungating lesion (a) lateral and (b) frontal aspect. Radiograph showing more destruction of distal phalanx (c) AP and (d) lateral view

Routine blood investigations were within normal limit but history lead us to the suspicion of Haemophilia. Prothrombin time was 13.1 seconds and activated partial thromboplastin time (aPTT) was 49.5seconds. Raised aPTTled to further investigation of factor VIII and IX levels in which Factor IX level was found to be normal butFactor VIII level was 7.6%..

The patient was managed conservatively. Factor VIII level was replaced according to standard protocol (40 IU/kg per dose thrice a week for 10 days). Daily dressing was done using tranexamic acid locally.

Patient recovered completely in 3-months duration with complete healing of wound and reconsolidation of partial distal phalanx with good functional and aesthetic outcome. (Fig 3 and 4)

Discussion

Distal phalanx lytic lesion is a rare entity which always poses diagnostic challenge leading to delay in management and at times unfavourable outcomes. Many disorders like keratokanthoma, glomus tumour, epidermal inclusion cyst etc. may present with osteolytic lesion which further poses diagnostic dilemma. Many researchers like Rajakulasingam et al[4]and Jamshidi et al[5] described paradigm of diagnosis of such lesion which may range from benign bony lesions like enchondroma to malignant tumour like squamous cell carcinoma.

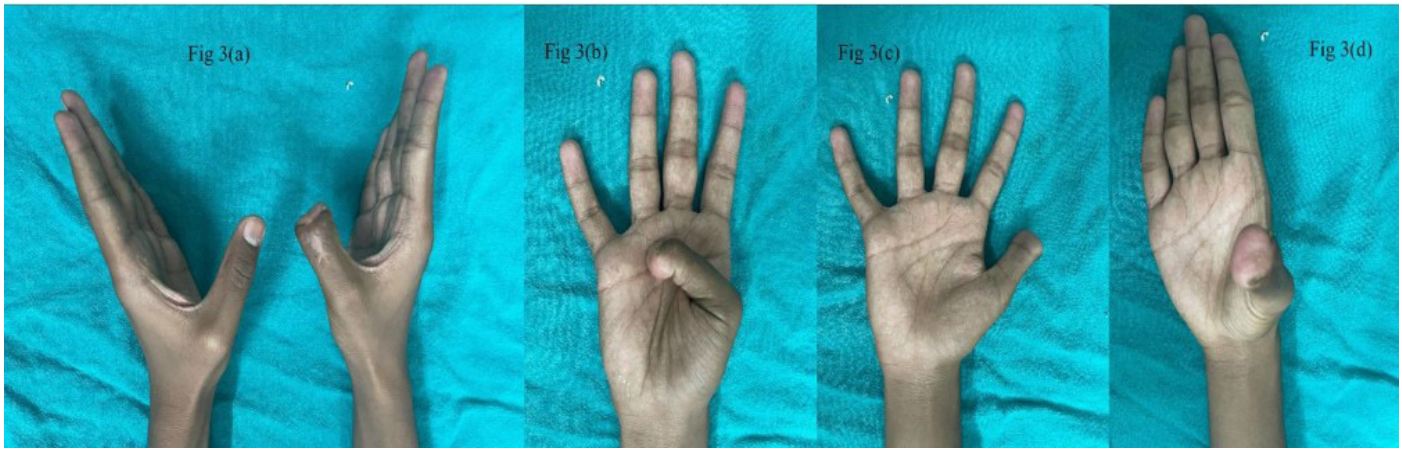
Despite an elaborate description of various diagnosis, haemophillicpseudotumour as a cause of lytic lesion is often overlooked, which is also seen with this patient led to oblivious biopsy by local practiotioner without heamatological investigation which is actually a contraindication in case of haemophillia.

Hemophilic pseudotumor occurs in 1 to 2% patients with severe haemophilia[6]. The severity of the disease is classified on the basis of the patient's baseline level of factor VIII or factor IX. Severe hemophiliahave less than 1.0 U/dL of the specific clotting factor, and with spoantaneous bleeding. Moderate hemophiliahave 1–5 U/dL and require mild trauma to induce bleeding. Individuals with mild hemophilia have levels greater than 5 U/dL, may go undiagnosed for many years.

Repeated bleeding with formation of hematomas and hemarthrosis are the most frequent musculoskeletal manifestations of the disease.[7] In our case although patient have level of 7.6 U/dL, his first presentation was phalanx intraosseous pseudotumour, with no history of any bleeding diathesis or hemarthrosis.

Gilbert described pseudotumours of two varieties proximal and distal. Proximal pseudotumours with more adult presentation occurs mainly in the proximal axial skeleton. Originating in the soft tissues with slow growth, they erode the bones from the outside. Theypresent as painless mass; later as pathological fracture occurs more frequently in adults. Distal pseudotumoursoccur in short bonesof extremities, as in the hands and feet, especially in younger patients with open physes and are due to intra-osseous bleeds with rapid growth.[8]

The exact pathogenesis of pseudotumor of bone is not well understood.



Three probable mechanisms have been suggested including extension from a hemarthrosis under pressure, bleeding into adjacent soft tissue with secondary bone invasion or subperiosteal bleeding with cyst formation.[9]

Because of its rarity, there is lack of consensus on management and no standardized treatment protocol has been developed.[10] Treatment ranges from Conservative management, embolization, Percutaneous Currettage with Fibrin or hydroxyapeptite filling [11,12] Radiation therapy[13], Surgical Excision with/ without Prosthetic Replacement[14].

Thomas et al proposed non surgical management of pseudotumour size < 3cm along with factor cover and enlarging lesion size > 3cm, excision should be considered.[15] But, Afshar et al performed amputation of distal phalanx despite the size of pseudotumour lesion was < 3cm (27 x17 x16 mm3).[16]Sharma et al yielded good result in distal radius pseudotumourlesion of size7.5cm x 6cm with factor replacement and local wound therapy.[17]

It is pertinent to preserve the maximum possible length of thumb where ever possible to provide best functional outcome. Hence we managed this patient conservatively with good functional outcome and radiographic evidence of reossification of distal phalanx remanent.

Conclusion

Swelling with lytic lesion in distal phalanx can be first presentation of mild Haemophilliawithout any previous history of bleeding dyscrasia. Such patients should always be investigated to rule out haemophilic pseudotumour before proceeding for invasive procedure like biopsy to prevent unfavourable outcomes.

Conflict of Interest Statement

The Author(s) declare(s) that there is no conflict of interest.

Ethics approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Informed consent was obtained from the individual participant included in the study.

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Reference

[1] Azzopardi C, Chisholm C, Vidoni A, Crighton J, Davies M, James SL, Botchu R. Lesions of the distal phalanx: Imaging overview. European Congress of Radiology-ESSR 2017.

[2] Goel A, Gupta A, Bhatia N, Goyal R, Mehndiratta A, Yadav A. Giant Cell Tumor of the Ring Finger Distal Phalanx. The Journal of Hand Surgery. 2021 Mar 1;46(3):249-e1.

[3] Purkait R, Mukherji A, Dolai TK, Bhadra R. Intraosseous pseudotumour in a child with mild hemophilia B: report of a rare case and brief review of literature. Indian Journal of Hematology and Blood Transfusion. 2014 Sep;30:366-8

[4] Rajakulasingam R, Azzopardi C, Murphy J, Davies M, Toms A, James S. Lesions of the distal phalanx: imaging overview. Indian J MusculoeskeletRadiol. 2019;1(1):47-56...

[5] Jamshidi K, Mazhar FN, Jafari D. Lytic lesion in the distal phalanx of the hand. Shafa Ortho J. 2015 Feb 10;2(1):e441.

[6] Rodriguez Merchan EC. The haemophilic pseudotumour. Int Orthop 1995;19(4):255–260

[7] Scott JP, Montgomery RR (2012) Hereditary clotting factor deficiencies (bleeding disorder). In: Kliegman RM, Stanton BF, St. Geme JW, Schor NF, Behrman

[8] RE (eds) Nelson textbook of pediatrics, 19th edn. Saunders, Philadelphia, p 1699–1703

[9] Gilbert MS. The hemophilic pseudotumor. Prog Clin Biol Res 1990; 324: 263–8.

[10] Geyskens W, Vanhoenacker FM, Van der Zijden T, Peerlinck K (2004) MR imaging of intra-osseous hemophilic pseudotumor: case report and review of the literature. JBR-BTR 87:289–293

[11] Rodriguez-Merchan EC. The haemophilic pseudotumour. Haemophilia. 2002 Jan;8(1):12-6.

[12] Caviglia HA, Fernandez-Palazzi F, Galatro G et al. Percutaneous treatment of haemophilic pseudotumours. In: Rodriguez-Merchan EC, Goddard NJ, Lee CA, eds. Musculoskeletal Aspects of Haemophilia Oxford: Blackwell Science Ltd, 2000: 97–104.

[13] Sagarra M, Lucas M, de la Torre E et al. Successful surgical treatment of haemophilic pseudotumour, filling the defect with hydroxyapatite. Haemophilia 2000; 6: 55–6.

[14] Brant EE, Jordan HH. Radiologic aspects of hemophilicpseudo¬tumors in bone. Am J Roentgenol Radium Ther Nucl Med 1972;115(3):525–539

[15] Buchowski JM, Cascio BM, Streiff MB, Frassica FJ. Resection and reconstruction of a massive femoral hemophilic pseudotumor. Clinical Orthopaedics and Related Research (1976-2007). 2005 Jan 1;430:237-42.

[16] Thomas BP, Fouzia NA, Raveendran S, Pallapati SR, Abraham A, Srivastava A. Management of hemophilic cysts and pseudotumors of the hand in bleeding disorders: a case series. The Journal of Hand Surgery. 2018 May 1;43(5):486-e1.

[17] Afshar A, Tabrizi A. Hemophilic Pseudotumor of the Distal Phalanx. Shiraz E-Medical Journal. 2023 Apr 30;24(4).

[18] lSharma A, Lodhi JS, Lingaiah P, Kumar A, Arora S. An Unusual Presentation of Haemophilia A: Pseudotumor of Distal End Radius-Masquerading as Giant Cell Tumor. J Hand Microsurg. 2019 Oct;11 (Suppl 1):S11-S15. doi: 10.1055/s 0038-1646774. Epub 2018 May 28. PMID: 31616120; PMCID: PMC6791804.

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