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CASE STUDY

Osseous bilateral pseudotumour of the thumb in severe haemophilia A – A case report

Prakas K Mandal, Rishu Vidhrati, Debasis Gantait

Pseudotumour is a rare complication of haemophilia, categorised as osseous or non-osseous (soft tissue) lesions based on anatomic location. The bones most frequently involved are the larger ones; pseudotumours of small bones are rare. Here we present a rare case of pseudotumours of both thumbs in a 10-year-old male with severe haemophilia A, successfully treated with factor replacement therapy. This case highlights the possibility of treating such cases conservatively, but also the need for education to enable early intervention to prevent potential complications that could be life-threatening.

Keywords: *Haemophilia A, Pseudotumour, Thumb, Case report*

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Although rare, haemophilic pseudotumours are potentially life- and limb-threatening and occur more frequently in countries where access to factor concentrates is limited. In this case study, conservative treatment and initiating low-dose prophylaxis have so far been effective in managing a paediatric case of pseudotumour of both thumbs

Pseudotumours are a rare complication, occurring in 1–2% of people with haemophilia (PwH) [1,2]. A pseudotumour is a haematoma surrounded by a thickfibrous capsule (pseudocapsule), presenting as a progressive cystic swelling of soft tissue (non-osseous) or bone (osseous) [2]. Pseudotumours can develop as a complication linked to inadequate treatment of bleeds and are the result of multiple episodes of bleeding into bones or soft tissue [3,4]. If left untreated, the encapsulated lesion gradually enlarges, putting pressure on and potentially causing damage to adjacent structures [3,4,5]. This can have a serious impact on limb function and viability and may be life-threatening. Most cases of osseous pseudotumour are seen in the femur, pelvis, tibia, and bones of the

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hand, while soft tissue pseudotumours are mostly associated with the iliopsoas, quadriceps, triceps, soleus, and gluteus muscles [6].

Although pseudotumours are a rare complication of haemophilia, there are some countries– including India – where they are more common due to difficulties in accessing the factor concentrates used to treat haemophilia, either for acute bleeding episodes or as prophylaxis. Effective early intervention is important to prevent serious complications. Few cases of pseudotumour of the thumb have been reported in the literature [7,8] and treating them effectively in a resource-constrained environment can be challenging. Here, we present the case of a child who presented at our hospital in Kolkata with osseous pseudotumours in both thumbs.

CASE SUMMARY

A 10-year-old boy presented to us with swelling and pain in bilateral thumbs (left>right) which he was reported to have had for three years, and which had increased in size in the previous six weeks. He was a diagnosed case of severe haemophilia A without inhibitors and was on intermittent 'on demand' factor replacement therapy (FRT). He had a previous history of bleeds in the right elbow and right ankle which were treated successfully at the hospital with FRT and did not have any chronic sequelae or deformities. He had not presented previously in relation to issues with either of his thumbs.

On examination there was swelling and mild tenderness of both thumbs; the overlying skin appeared normal (Figure 1). There was no history of trauma. Differential diagnosis included connective tissue disorder, abscess caused by tuberculosis, fracture and true tumour. The child had no history of fever or any skin rash, and radiological findings were not suggestive of any infective or inflammatory pathology. Involvement of the thumb in disseminated tuberculosis without the involvement of any other system was unlikely. Fracture was unlikely due to the lack of trauma, and tumours involving bilateral thumbs is extremely rare. Straight X-ray of the hand showed a lytic bone lesion with prominent internal trabeculation in the bilateral proximal phalanx of the thumb. Magnetic resonance imaging (MRI) showed an expansile altered signal intensity in the proximal phalanx of thumb on both sides (left>right) with outward bulging of subcutaneous fat all around with maintained adjoining joint spaces suggestive of pseudotumour (Figure 2).

Orthopaedic opinion was taken for further management. Surgical intervention in the form of curettage and bone filling was ruled out in the first instance in favour of conservative management with FRT. It was agreed that aggressive surgical management would be planned in the case of recurrence of the pseudotumour.

As per World Federation for Haemophilia (WFH) guidelines [9], the patient was started on FRT with plasma-derived factor VIII (FVIII) in a dose of 50 units/kg twice daily for two weeks, for which he was admitted to our hospital. This was followed initially by thrice weekly prophylaxis with plasma-derived FVIII, followed by long-acting FVIII (Eloctate) twice weekly at a dose of 20 units/kg administered at the hospital. On follow-up after six months, the swelling in both thumbs had significantly reduced both clinically and radiologically. The patient is continuing to receive regular twice weekly prophylaxis as an outpatient. The child's parents have been counselled regarding the need for regular prophylaxis therapy to prevent any future complication. Adherence to prophylaxis therapy is satisfactory.

DISCUSSION

The clinical presentation of pseudotumours varies according to anatomical location. Gilbert et al. describe different clinical features for proximal and distal

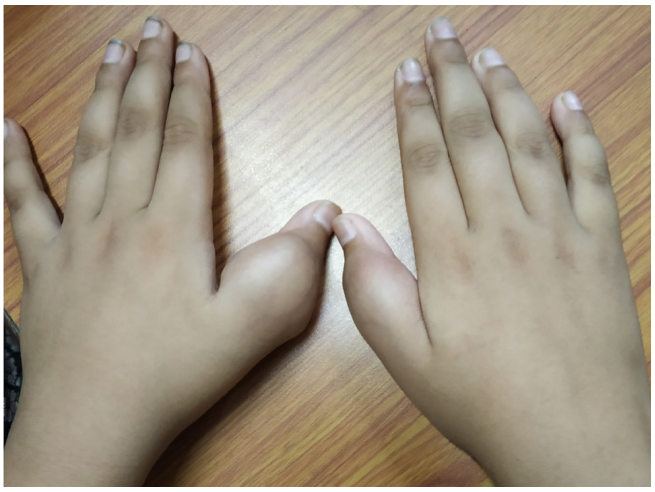


Figure 1. Pseudotumour of thumbs (bilateral bulbous swelling of proximal thumb)

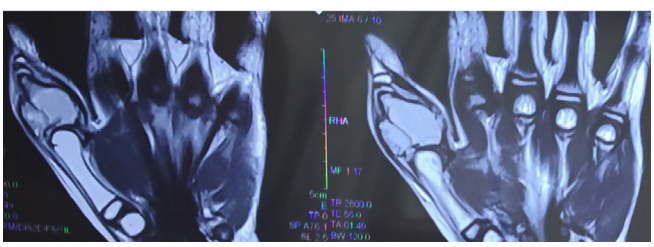
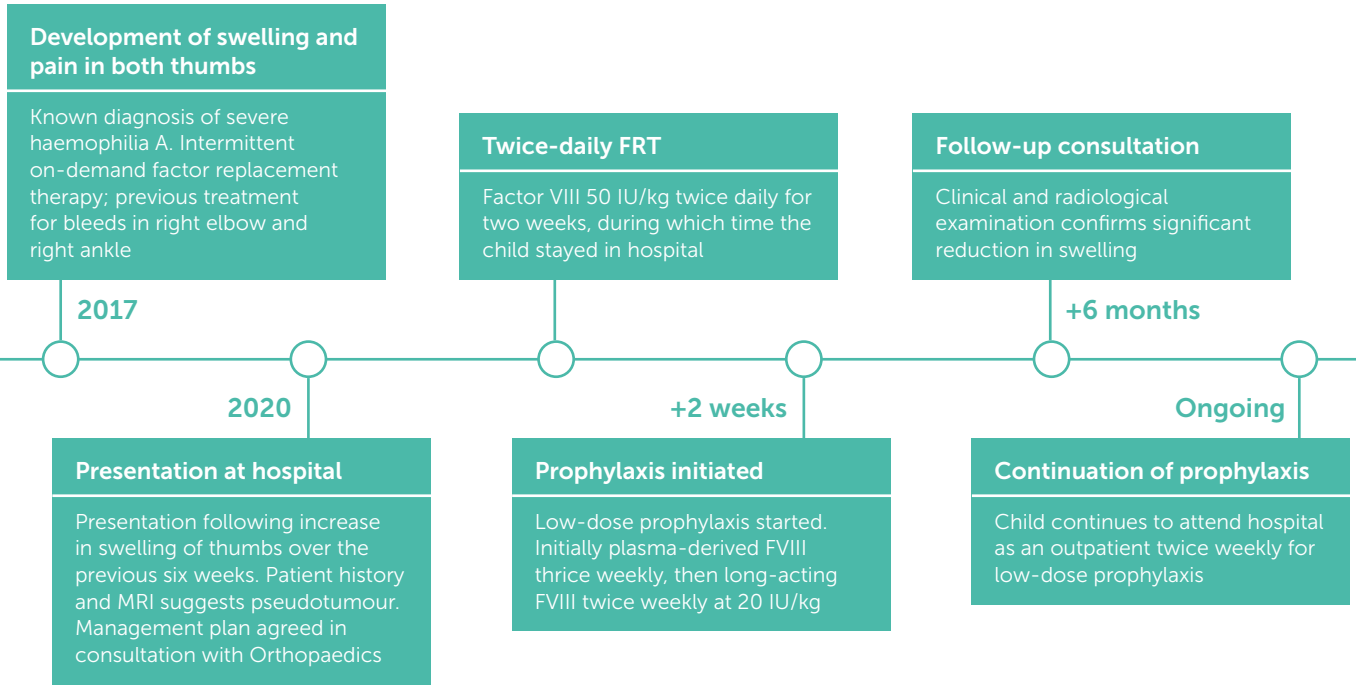


Figure 2. Magnetic resonance imaging of right thumb (hyperintense lesion of proximal phalanx with liquefacation)

Figure 3. Timeline of presentation and treatment



pseudotumours [10]. Proximal pseudotumours occur more frequently in adults around the femur and the pelvis, starting in the soft tissues and eroding the bone from the outside. They present as a painless mass until the patient later presents with fracture, and they do not respond to conservative treatment. Distal pseudotumours affect skeletally immature younger patients and are usually seen distal to the wrist and the ankle. They develop rapidly and secondary to intraosseous haemorrhage and are commonly seen in the small cancellous bones (calcaneus, talus, and metatarsals of the feet, but rare in carpals and metacarpals). Osseous pseudotumours can present in their 'simple' state as a painless cystic lesion but in severe forms may present in the form of joint dysfunction or bone fracture [8]. In the case reported here, the presentation was a thumb swelling that had been painless for a long period of time, but which had become painful more recently, possibly due to acute internal bleeding.

Conventional radiography, sonography, computed tomography (CT) and MRI each play an important role in the diagnosis and management of pseudotumours [7]. Management is complex and depends on the site, size, growth rate, and effect on adjoining structures. Options include FRT, radiation and surgical excision. The World Federation of Hemophilia (WFH) recommends a short course (6-8 weeks) of FRT for early pseudotumour (before acquiring a pseudocapsule), with continuation of therapy if serial ultrasound evaluations reveal the mass is shrinking and repeat evaluation after 4-6 months [3]. For patients with large pseudotumours with pseudocapsule, surgical excision is recommended, followed by close monitoring and long-term prophylaxis to prevent recurrence of bleeding. Surgical excision is the treatment of choice but should only be carried out in major haemophilia centres by a multidisciplinary surgical team [11]. Khubrani AM et al. from Riyadh, Saudi Arabia, reported a case of haemophilic pseudotumour of the thumb in a young boy that was managed surgically with curettage and a bone graft [8]. Issaivanan et al. from New Delhi, India, reported a case of haemophilic pseudotumour of the thumb in a child who responded to combined treatment with radiation therapy and factor VIII replacement, resulting in complete resolution of the lesion [7]. The case reported here was also treated conservatively with FRT and responded well. Conservative treatment has been recommended for osseous pseudotumours located in distal parts of the limbs [12].

Despite the outcome of the episode reported here being positive at the time of writing, there is a high risk of recurrence. Prevention is essential to avoid the complications that pseudotumours can result in, so the best course of action in the long term is to ensure that the child remains on prophylactic therapy [12,13]. Rodriguez-Merchan suggests that the

occurrence of pseudotumours is an indirect measure of country's haematological prophylaxis [13]. India has a large population but accounted for only 4% of the FVIII concentrates used around the world in 2020 [14]. However, even low-dose prophylaxis has been shown to be effective at reducing the occurrence of bleeding and to have clinical benefits [15-17]. Prophylaxis therapy has not been a possibility for PwH in India until recently but this scenario is starting to change. Some haemophilia centres are now offering low and intermediate dose prophylaxis [18] and there is a national recommendation that children with haemophilia in India are managed with prophylaxis [19]. This was not the case for the patient reported here, who had experienced multiple bleeding episodes and reported to the hospital after three years of living with swelling to his thumbs. His parents had not been aware of the potential long-term consequences and were not able to access the haemophilia treatment centre easily. As the factor concentrates provided at our hospital are supported by WFH humanitarian aid and home therapy is not an established form of support for PwH in India, it is necessary for patients to attend our clinic for treatment, including prophylaxis therapy. Now that the child's parents have been counselled regarding importance of prophylaxis therapy and risk of recurrence of life-threatening complications like pseudotumours, they are attending hospital regularly for their son's treatment. We are hopeful that starting the child on a low-dose prophylactic regimen and helping his parents to understand the importance of both this and accessing treatment for any suspected bleeding at an early stage will help to reduce the risk if not prevent pseudotumour recurrence.

CONCLUSION

Pseudotumours are a rare complication in haemophilia and the involvement of both thumbs is reported very rarely. Each case of pseudotumour should be managed individually; clinical and radiological evaluations are helpful in planning further management. Although surgical intervention can be recommended, this case highlights the feasibility of a conservative approach with regular factor replacement therapy in a resources-constrained setting. This case also highlights the importance of access to prophylaxis in developing countries and the need to raise awareness and educate PwH and the parents of children with haemophilia in these countries about the importance of early intervention to prevent long-term and potentially life-threatening complications.

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Informed and written consent has been obtained from the legal guardian of the individual reported in this case report. The case report has been evaluated and approved by the Institutional Ethics Committee (IEC) of Nil Ratan Sircar Medical College and Hospital, Kolkata.

AUTHOR CONTRIBUTIONS

PKM designed the study. PKM, RV and DG contributed to the manuscript writing, literature search, and manuscript editing. All authors reviewed the manuscript and gave final approval.

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