Case Report

Hemophilic pseudotumor of the skull: A rare presentation in a young child

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Abstract

Hemophilic pseudotumors are rare complications of severe hemophilia and typically involve muscles and bones. We present the case of a 1-year-old 5-month-old boy with severe hemophilia A, who developed progressively enlarging left occipital scalp swelling. A computed tomography (CT) scan revealed a significant left occipital osteolytic calvarial mass causing a mass effect and displacing the 4th ventricle. The absence of acute neurological compromise allowed for conservative management in this case. This case highlights the importance of considering hemophilic pseudotumors in patients with hemophilia, even in atypical locations. Early recognition and individualized management are crucial for optimal outcomes in patients with this challenging complication.

Keywords: Hemophilia, Hemophilic pseudotumor, Scalp, Skull.

Introduction

Hemophilia is a condition in which a deficiency in clotting factors causes coagulation defects. Hemophilia pseudotumors are rare complications of hemophilia [1]. It usually involves muscles of the pelvis or lower limb as well as bone involvement, most commonly involving the femur, tibia, pelvis or small bones of the hands [1]. Skull involvement is a rare occurrence [2]. We report a rare case of skull hemophilic pseudotumor and review its presenting illness, diagnosis and treatment.

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Case summary

A 1-year-5-month-old Malay boy underlying severe hemophilia A and a history of recurrent intracranial bleeding complicated by right-sided hemiparesis and seizures presented to the hospital with a complaint of progressively enlarging left occipital scalp swelling (Figure 1). The swelling had been present for two months, initially growing rapidly but stabilizing in size for the past month prior to presentation. Despite the swelling, the child remained active and alert, without fever or signs of increased intracranial pressure.

Ultrasound was initially performed to evaluate the swelling. Ultrasound revealed a heterogeneous hypo-hyperechoic mass in the left occipital region with anechoic components. There was no intralesional vascularity. (Figure 2). It involved the subperiosteal region, with extension to the diploe of the occipital skull.

A computed tomography (CT) scan of the brain was subsequently performed in view of the presenting complaint. The findings revealed a significant left occipital osteolytic calvarial mass with inhomogeneous hyperdense attenuation associated with curvilinear deformed bone/calcification at the outer margin of the lesion and bone resorption in this region (Figure 3). This hematoma caused a mass effect, displacing and compressing the 4th ventricle with resultant hydrocephalus. Additionally, frontal lobe encephalomalacia was noted as a sequela of previous intracranial bleeding (Figure 4).

These imaging findings, which are correlated with underlying hemophilia, are suggestive of hemophiliac pseudotumors. Given that the patient remained active with no sign of increased intracranial pressure, the patient was treated conservatively and monitored for any sign of increased intracranial pressure. This patient did not receive treatment for factor replacement due to a financial constraint. The patient was subsequently on a palliative treatment. No follow-up imaging was performed.

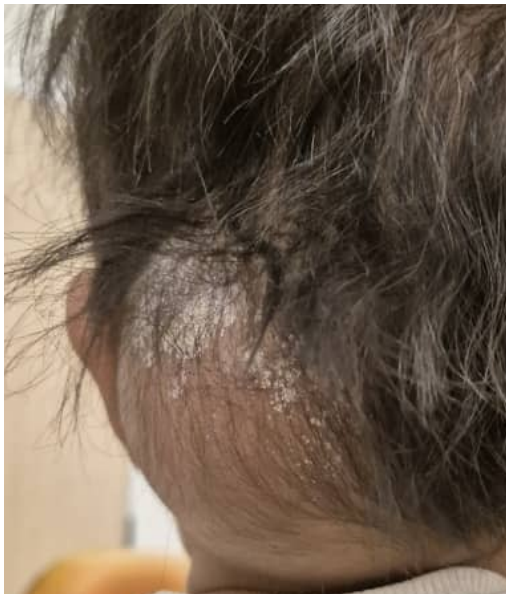

Figure 1. *Large left occipital scalp swelling on presentation.*

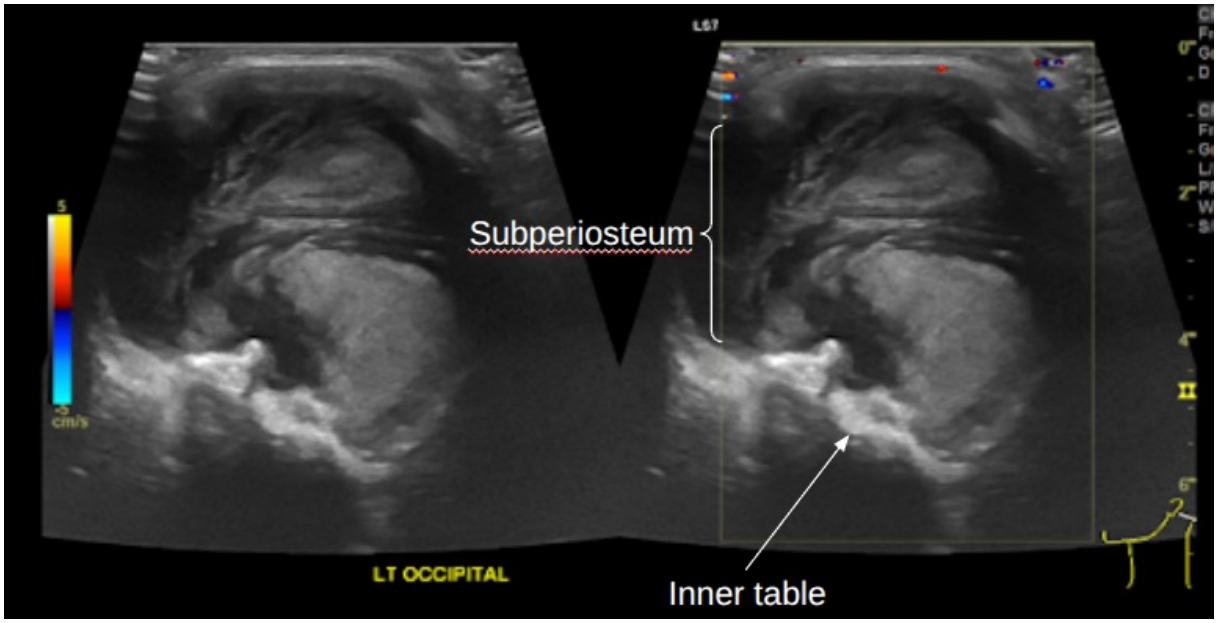

Figure 2. *Ultrasound image shows (A) grey scale, (B) with color Doppler showing a heterogeneous hypo-hyperechoic mass in the left occipital region with an anechoic component involving the subperiosteal region (shown by bracket), with extension to the diploe of the occipital skull. There was no intralesional vascularity.*

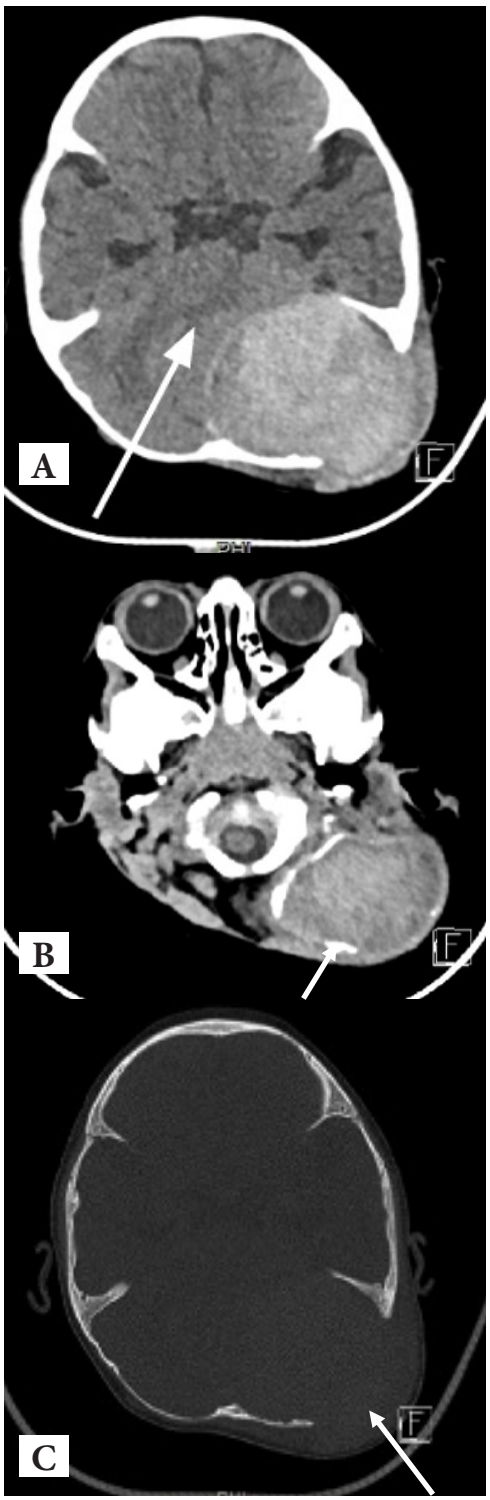

Figure 3. *(A) Large hyperdense mass in the left occipital region suggestive of a pseudotumor causing compression of fourth ventricle (white arrow) (B) curvilinear deformed bone/calcification (white arrow) noted at the outer margin of the mass (C) Expansion of the inner and outer table at the left occipital region with associated cortical thinning and bony resorption (white arrow) around the mass.*

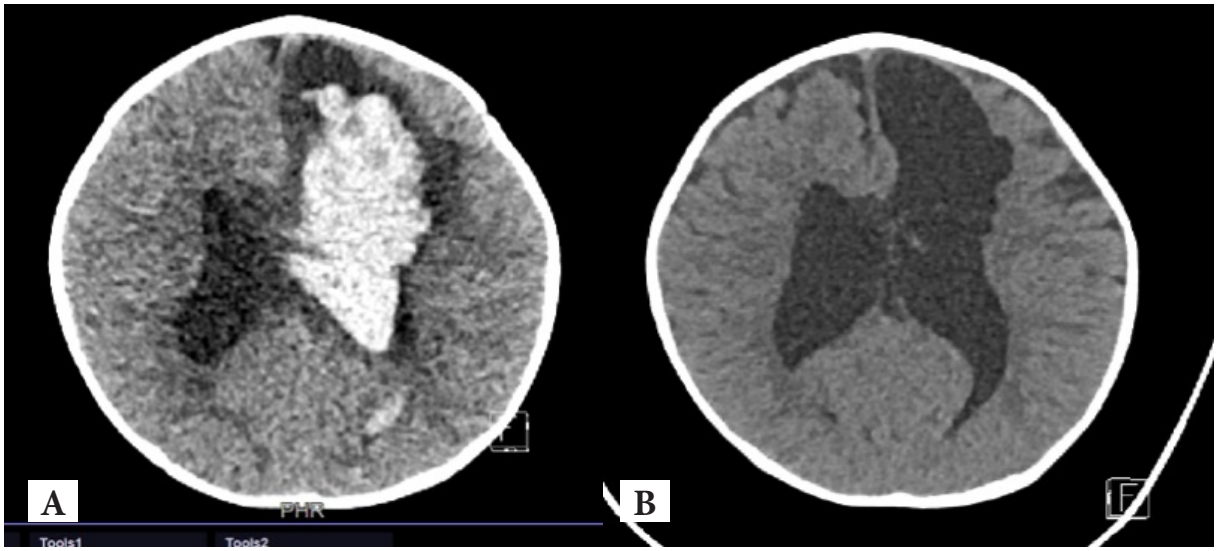

Figure 4. *(A) CT brain performed 17 months prior to current presentation shows left frontal intraparenchymal bleeding (white arrow) with intraventricular extension and hydrocephalus (black arrows). (B) Current CT noted evidence of left frontal encephalomalacia in the corresponding region.*

Discussion

Hemophilic pseudotumors are rare complications of severe hemophilia, arising from the insidious accumulation of blood within the muscle or bone due to recurrent bleeding episodes [1]. These conditions, affecting less than 2% of individuals with severe hemophilia, manifest clinically with a wide range of symptoms depending on their location and size [2]. It usually involves muscles of the pelvis or lower limb as well as in the case of bone involvement, most commonly involving the femur, tibia, pelvis or small bones of the hands [2]. Skull involvement is a rare occurrence [3,4,5]. There are limited number of case reports on skull hemophilic pseudotumor especially in pediatric population. The youngest patient with this condition in our literature review is reported by Horton et al (1993) which involve a 12-month-old American Indian boy [6]. Depending on their location, these pseudotumors can cause various complications. As their sizes

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increase, they can compress into nearby anatomical structures, causing symptoms. In our case, it presented as a left occipital 'mass', which is a rare site of involvement that causes a mass effect on the surrounding brain structure.

Radiological imaging, encompassing X-rays, CT and magnetic resonance imaging (MRI), plays a crucial role in diagnosing and characterizing these lesions. X-rays are particularly valuable for accessing intraosseous pseudotumors, revealing their characteristic lytic, expansile nature, which is often accompanied by cortical changes and periosteal reactions [7-8]. Ultrasound can demonstrate the presence of fluid within the pseudotumor [1] as well as progression after treatment, particularly in the case of soft tissue pseudotumor [2]. In our case, ultrasound revealed a heterogeneous mass with anechoic components. CT and MRI offer comprehensive insights into its extent, composition, and relation to adjacent structures [1]. In the case of intraosseous involvement, CT is particularly useful for evaluating crossing trabeculae, cortical changes, and periosteal reactions. MRI has a remarkable ability for assessing intramedullary portions and nearby soft tissue (neurovascular bundle) as well as monitoring the therapeutic response. The characteristic MRI appearance is an intramedullary cystic lesion containing fluid components, which have complex signal intensities reflecting the effects of remote and recurrent hemorrhage and clot organization [2]. In our case, CT was able to characterize the lesion, revealing its extension and mass effect on the surrounding structure and eventually providing an accurate diagnosis that was correlated with the underlying condition.

Other possible differential diagnoses with solitary soft tissue lesions with bone erosion include neuroblastoma one metastasis as in Hutchinson syndrome. However, this condition usually appears as a multiple area of bone metastasis [9]. Figure 5 depicts an instance of neuroblastoma metastasis, manifesting as multiple osseous lesions [10]. In terms of imaging, MRI for hemophilic pseudotumor shows T1W/T2W peripheral hypointense rim representing fibrous tissue that contains hemosiderin which is not seen in Hutchinson syndrome. A combination of the given history of hemophilia and imaging features, biopsy should be avoided in these patients [11]. In our case, the presence of underlying Hemophilia A with a

solitary soft tissue mass in the skull best fit the diagnosis of hemophilic pseudotumor even though no MRI imaging was performed.

The management approach primarily focuses on prevention through meticulous control of bleeding episodes with a factor replacement therapy and other conservative measures [1]. However, for established pseudotumors, a multipronged therapeutic strategy may be needed, encompassing options such as low-dose radiotherapy, percutaneous curettage, surgical resection, or even filling the resultant cavity with a bone graft or other suitable materials [7]. In our case, the patient was treated conservatively with further monitoring for any sign of increased intracranial pressure.

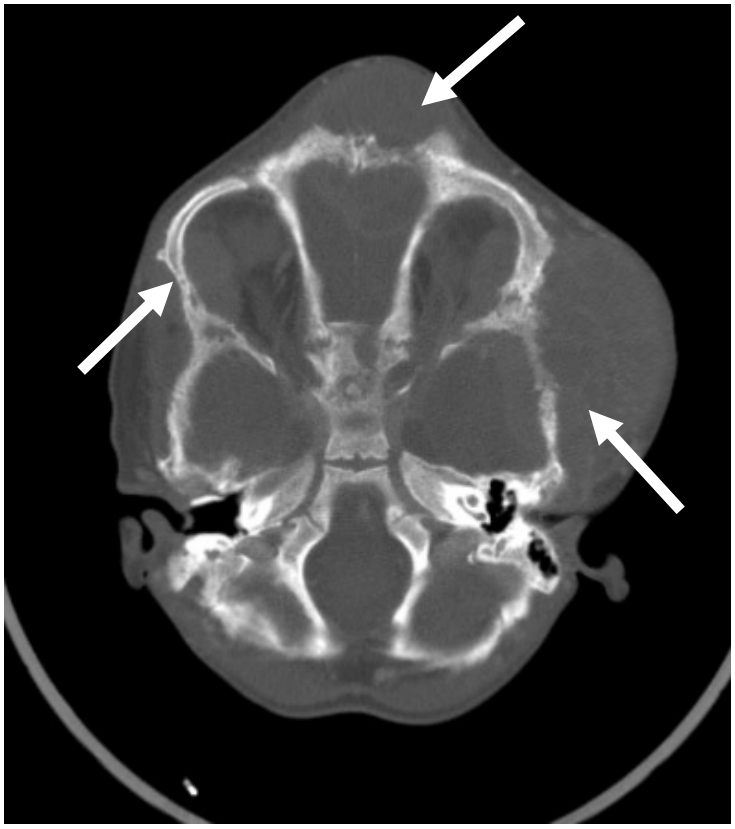

Figure 5. *Multiple osseous lytic metastases of neuroblastoma in skull base and calvarium (arrows) - Case courtesy of Ahmed Abdrabou, Radiopaedia.org, rID: 32540 [10]*

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Conclusion

This case highlights the rare but significant complication of hemophilia pseudotumor in a young boy with severe hemophilia A. His presentation with progressively enlarging scalp swelling and subsequent CT findings emphasized the importance of considering this diagnosis in patients with hemophilia, even in unusual locations such as the skull. While rare, hemophilic pseudotumour remains a challenging complication, necessitating a high index of suspicion, prompt diagnosis, and multidisciplinary management to optimize patient outcomes. However, with the presence of a history of Hemophilia, typical presentation and imaging findings, particularly MRI which shows peripheral rim of T^1W and T^2W hypointensity, biopsy should be avoided. Further research and awareness are essential to improve the understanding and management of this rare entity.

Conflict of interest: The authors declare that they have no conflicts of interest.

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