Case Series/Case Reports
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External Occipital Protuberance Enlargement in Children: A Benign Anatomic Variant with Rare Associations

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Abstract

Objective – The aim of this study is to raise awareness about External Occipital Protuberance (EOP) enlargement in children, its typically benign anatomical variant, and the importance of considering associated conditions in diagnosis and management. **Case Report** – We reviewed the cases of four male patients, presenting with EOP enlargement. Among the cases, one involved a newborn diagnosed with Menkes disease, a rare genetic disorder affecting copper transport, which was associated with occipital exostosis. The other three cases involved adolescents with varying degrees of EOP enlargement, linked to factors such as prior trauma and excessive screen time. **Conclusion** – While EOP enlargement is generally a benign anatomical variant, this study underscores the importance of differential diagnosis, particularly in the pediatric population. Clinicians should be aware of the potential, albeit rare, associations with serious conditions such as Menkes disease. A comprehensive approach to diagnosis and management is recommended, especially in symptomatic cases.

Key Words: External Occipital Protuberance ▪ Pediatric Exostosis ▪ Occipital Knob ▪ Occipital Bun ▪ Skull Abnormalities.

Introduction

The External Occipital Protuberance (EOP) is a normal anatomical prominence located on the posterior surface of the occipital bone (1-3). The EOP can be classified into three types: smooth (type 1), crest (type 2), and spine (type 3) (3, 4, 11)**.** An EOP is considered enlarged (EEOP) if it exceeds 10 mm (1-4, 7).

EOP enlargement is often seen in late adolescence, more commonly in males. In the study by Shahar and Sayers, 41% of the total population had an enlarged EEOP, and males were significantly more affected (67.4%) compared to females (20.3%) (1-3). The nuchal ligament attaches to the EOP, and this attachment plays a crucial role in the mechanical stress exerted on the bone (3, 6). Recent studies suggest that this connection, combined with poor posture and increased use of smartphones, can contribute to the enlargement of the EOP in younger populations (1, 2, 6, 7). Although typically benign, other conditions such as trauma, tumors, inflammation, or genetic factors must be considered when diagnosing occipital protuberances (3, 7).

This article discusses four cases of EOP enlargement in children and adolescents, exploring the etiology, clinical significance, and diagnostic considerations.

Results

Case 1

A 12-year-old Caucasian boy was admitted to the Emergency Department with a painful occipital tumefaction, noted one day before admission. His medical history was insignificant. The examination

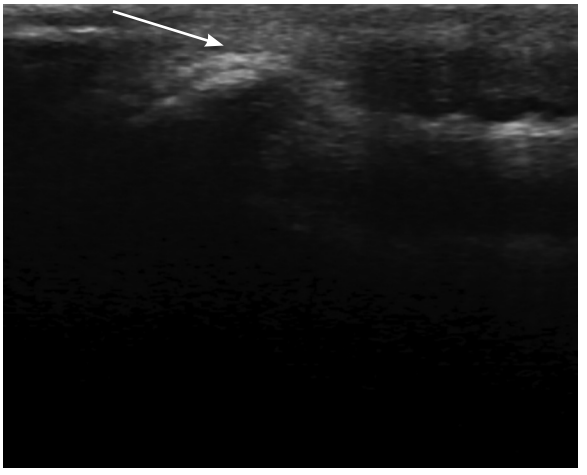

Fig. 1. *Ultrasound view showing a prominent external occipital protuberance in a pediatric patient.*

revealed the presence of a tender and painful bony swelling. An ultrasound was performed and revealed a prominent occipital protuberance (Fig. 1). His excessive screen use and poor posture were identified as potential contributing factors, and he was discharged without further investigation.

Case 2

A 15-year-old Caucasian boy was admitted to the Emergency Department with complaints of progressive occipital tumefaction, aggravated over the previous two years. He had a previous history of head trauma seven years before, which resulted in a hematoma and incisive wound with the need for suture. X-ray imaging revealed an exostosis of 18.6 mm at the occipital protuberance (Fig. 2). The boy was later referred for a pediatric appointment. Blood tests were performed, including levels of ceruloplasmin and copper. The results were normal, and he was discharged from hospital follow-up.

Case 3

This 15-year-old boy had been monitored for mild cognitive impairment and attention-deficit hyperactivity disorder. During a neurodevelopmental appointment, he reported an occipital tumefaction. X-ray revealed an EOP enlargement of 20.5 mm (Fig. 3). Extensive blood tests and genetic analysis were conducted, including levels of Ceruloplasmin, copper, ATP7A gene, array and X-fragile, to rule out any underlying genetic disorders, all of which returned normal results.

Case 4

A male newborn with hypotonia and hair discoloration was evaluated. Initial blood tests showed

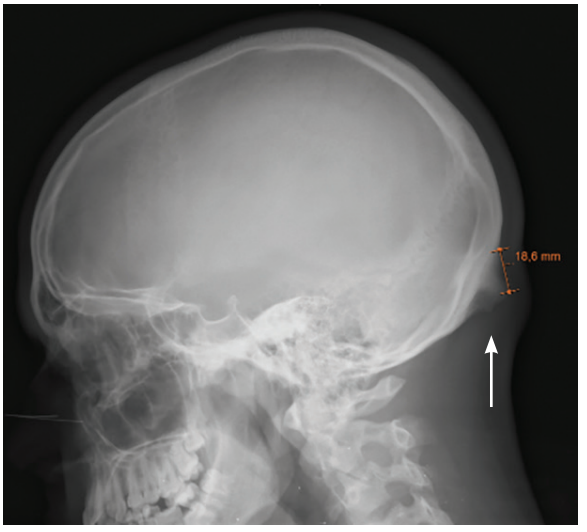

Fig. 2. *Lateral X-ray views of the skull showing a prominent external occipital protuberance in a pediatric patient.*

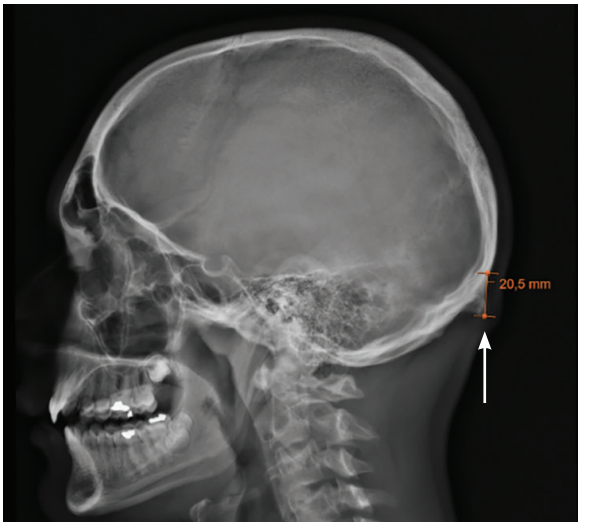

Fig. 3. *Lateral X-ray views of the skull showing a prominent external occipital protuberance in a pediatric patient.*

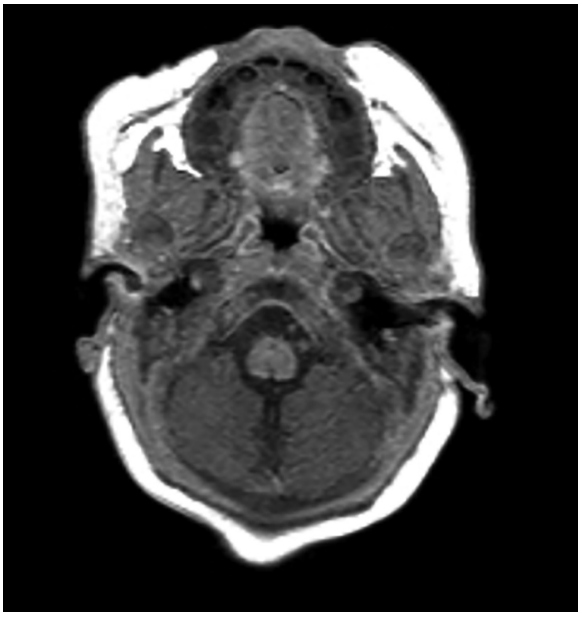

Fig. 4. *CT scan of the skull showing a prominent external occipital protuberance in a pediatric patient.*

low levels of copper and serum ceruloplasmin. On suspicion of Menkes disease, evaluation of the hair by electronic microscopy was requested, which showed *pili torti,* and molecular testing enhanced a mutation on the ATP7A gene. The diagnosis of Menkes disease was confirmed. Subsequent imaging revealed an occipital exostosis (Fig. 4). This patient experienced progressive neurological decline and passed away at the age of six years.

Discussion

EOP enlargement is typically considered a benign anatomical variant, commonly found in late adolescence and more frequently in males. Despite its usually asymptomatic or mildly symptomatic nature, the recent increase in prevalence warrants further investigation. It has been theorized that sustained head tilt and protracted postures related to use of modern technology, such as smartphones and tablets, place mechanical stress on the EOP. This stress could lead to bony remodeling over time, although further research is required to confirm the causal link (3, 6, 7).

While most cases of EOP enlargement are innocuous, healthcare providers should be vigilant and consider other potential causes in the differential diagnosis, such as ATP7A-Related Copper Transport Disorders, of which Menke´s disease represents the most severe presentation. It may manifest with both neurological decline and EOP enlargement (8). Occipital bone osteoma, a benign bone tumor, can also present with similar symptoms. Other conditions, such as fibrous dysplasia and hematomas may also mimic EOP enlargement (3, 9). Therefore, a thorough differential diagnosis is essential to ensure accurate management.

Although the gold standard for the study of skull anomalies is X-ray, the radiation inherent to X-ray image acquisition has prompted the use of alternative non-radiation imaging techniques with pediatric patients, such as ultrasonography (10, 11). Conservative treatments such as physiotherapy, ergonomic adjustments (e.g., soft pillows), and analgesics are first-line approaches. In persistent cases, surgical recontouring may be an option, though it should only be considered when conservative management fails (3, 5).

Conclusion

Since the current lifestyle, related to the indiscriminate use of screens from an early age, with sustained postures, can cause structural changes to the external occipital protuberance, we aim to raise awareness among healthcare professionals about this condition and its rare associations, emphasizing the importance of a comprehensive approach to differential diagnosis and management.

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References

1. Shahar D, Sayers MGL. Prominent exostosis projecting from the occipital squama: More substantial and prevalent in young adults than older age groups. Sci Rep. 2018;8:3354. doi:10.1038/s41598-018-21625-1.
2. Shahar D, Sayers MGL. A morphological adaptation? The prevalence of enlarged external occipital protuberance in young adults. J Anat. 2016;229(2):286-91. doi:10.1111/joa.12466.
3. Cheng Q, Xiong X, Li Y, Xiao C, Han S, Fan P, et al. Enlarged occipital spur and craniofacial morphology: a cephalometric analysis. Oral Radiol. 2023;39:743-9. doi:10.1007/s11282-023-00694-7.
4. Gunacar DN, Gonca M, Kose TE. Occipital spurs on lateral cephalometric radiographs: morphologic and morphometric features. Oral Radiol. 2021;38:416-421. doi:10.1007/s11282-021-00574-y.
5. Varghese E, Samson RS, Kumbargere SN, Pothen M. Occipital spur: understanding a normal yet symptomatic variant from orthodontic diagnostic lateral cephalogram. J Contemp Dent Pract. 2017.
6. Grubmuller A, Botkin C, Frye S, Muzaffar R, Osman M. Occipital exostosis resulting from excessive screen time. J Nucl Med. 2020;61(suppl 1):3047.
7. Porrino J, Sunku P, Wang A, Haims A, Richardson ML. Exophytic external occipital protuberance prevalence pre- and post-iPhone introduction: A retrospective cohort. Yale J Biol Med. 2021;94(1):65-71.
8. Orphanet. Paris: INSERM US14; ©1997–2024 [updated 2023 Oct 15; cited 2024 Oct 23]. Available from: https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=198.
9. Şenol N, Kapucuoğlu FN, Aydın YS. Occipital Bone Osteoma. J Nerv Syst Surg. 2014;4(3):134-137. doi:10.5222/sscd.2014.134.
10. Domingo AB,Godino AG. Ultrasound assessment of the external occipital protuberance in young adults: Clinical implications. Rev Fisioter Invasiva. 2020;3(1):45-48. DOI: 10.1055/s-0040-1712928.
11. Gómez Zubiaura A, Alfageme F, López-Negrete E, Roustan G. Type 3 External Occipital Protuberance (Spine Type): Ultrasonographic Diagnosis of an Uncommon Cause of Subcutaneous Scalp Pseudotumor in Adolescents. Actas Dermosifiliogr (Engl Ed). 2019;110(9):761-4. doi:10.1016/j.adengl.2019.06.001.