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Case report

C1 C2 spinal cord compression in hereditary multiple exostoses: case report and review of the literature

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ABSTRACT

Introduction and importance: Hereditary multiple exostoses (HME) is a benign disease, usually encountered in the metaphysis of long bones. On the opposite, spinal localizations are very rare. We herein describe a case of HME with a spinal cord compression.

Case presentation: A 31-years-old patient with a history of HME, presented with signs of cervical spinal cord compression that progressively appeared in the last 10 years. Cervical MRI and CT scan showed a compressive osseous tumor arising from the surface of the right side of the dorsal arch of the axis. Our patient underwent C1C2 laminectomy, but no clinical improvement has been recorded postoperatively.

Clinical discussion: Spinal cord compression is an extremely serious complication of hereditary multiple exostoses (HME). The cervical and thoracic areas were predominantly affected, with the symptoms usually developing slowly. Early surgery is correlated to good postoperative outcomes.

Conclusion: Despite its rarity, HME should be considered as a possible cause for spinal cord compression.

1. Introduction

Hereditary multiple exostoses (HME), also called osteochondromatosis, is a rare dominant autosomal skeletal disease. It is usually a benign condition, but associated to a considerable risk of malignant degeneration to chondrosarcoma. Bony exostoses typically occur at the tips of long bones, iliac crests and scapula, whereas spinal osteochondroma are much rarer [1]. Moreover, spinal cord compression due to osteochondroma is an unusual complication of this disease. We report the case of a young man with a history of hereditary multiple exostoses who presented a C1C2 osteochondromas responsible for cervical myelopathy. This work has been reported in line with the SCARE criteria [2].

2. Case report

A 31-year-old man, smoker of 10 units-pack-year with a familial history of two siblings affected with multiple hereditary exostoses, has also been diagnosed with this condition at the age of 10.The diagnosis was suspected as the patient presented exostoses on the left lower limb, related to masses that developed from the proximal left tibia. He underwent surgery for removal of these lesions, and pathologic exam concluded to osteochondromas related to an HME. He also presented an intra-abdominal mass arising from the right iliac wing. The patient presented to our department due to the progressive onset of a weakening on right arm and leg, as well as numbness in his left side. He also reported difficulties in bladder control. During the few past months, he also reported silent aspiration. These symptoms began in the last 10 years, and tended to a very progressive worsening. Physical examination showed an inequality of the length of the lower limbs, multiples exostoses in his hands and feet, limp walk, right brown Sequard syndrome, brisk reflexes and bilateral Babinski sign. He also presented a left glossopharyngeal palsy. The range of motion of the cervical spine was normal.

Multiple imaging studies have been performed. Standard radiograph of the cervical spine showed, on profile view, an exophytic bony mass originating from C2 (Fig. 1). Chest X ray showed a large exophytic osseous mass involving the second right rib (Fig. 2).

Spinal CT scan (Fig. 3) and MRI (Fig. 4) revealed an osseous tumor originating from the inner surface of the right-sided dorsal arch of the axis. This tumor extended into the upper cervical canal, compressed and deviated the spinal cord to the left. This mass was hyperintense on T1-weighted images (WI) and hypointense on T2-WI.

The patient's perspective about the treatment options was discussed

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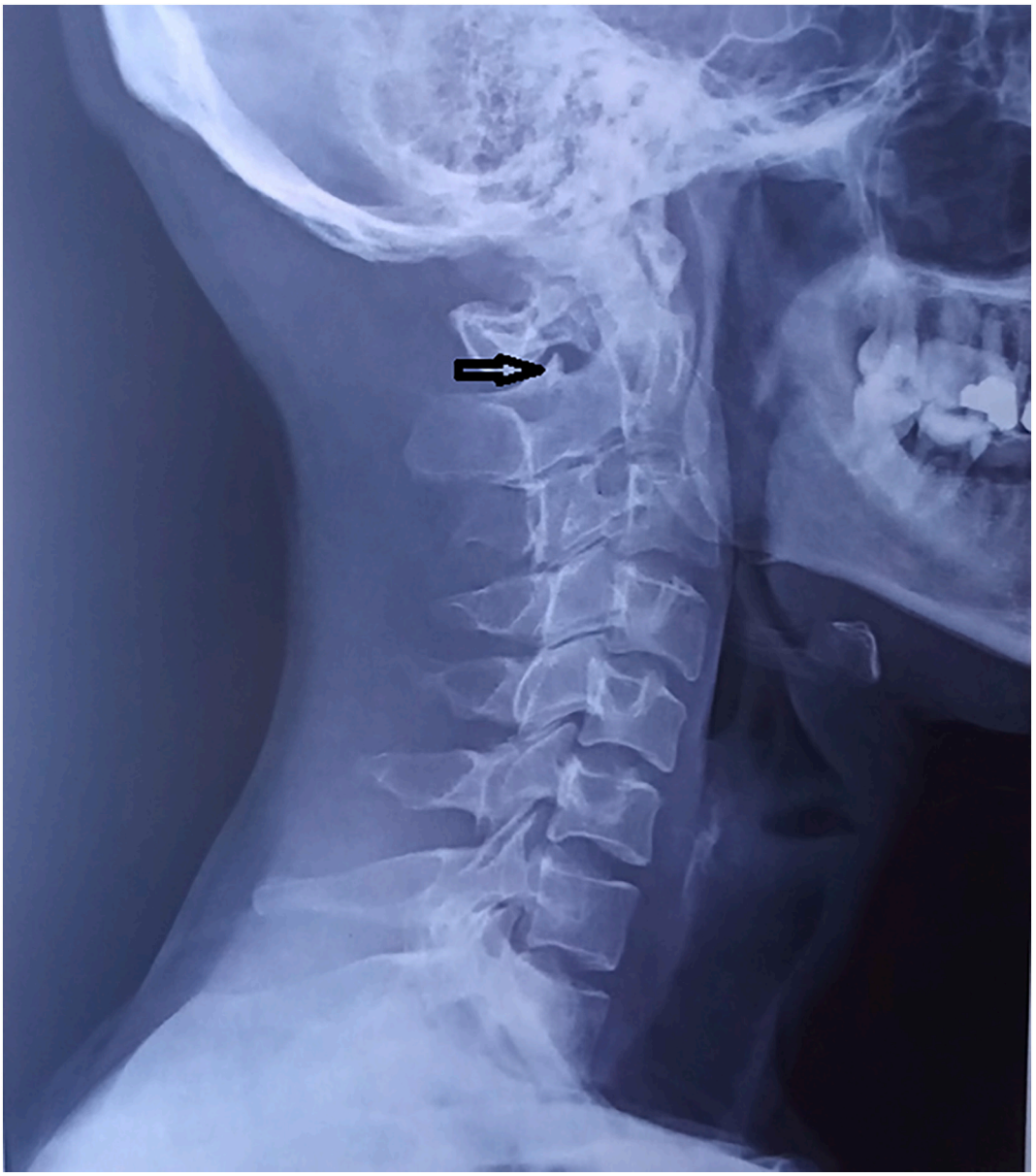

Fig. 1. Lateral radiograph of the cervical spine demonstrating an exophytic bony mass originating from C2 (arrow).

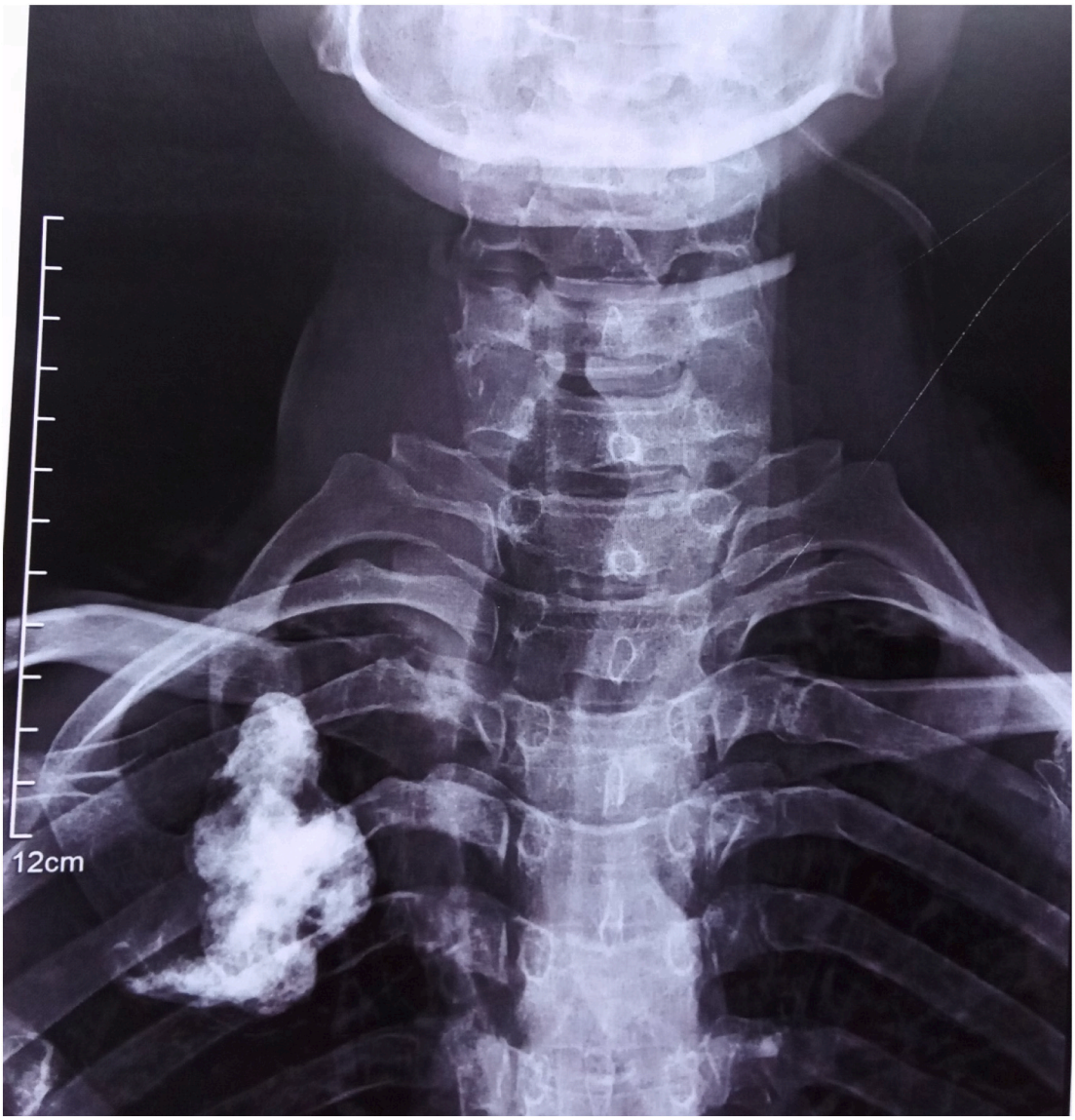

Fig. 2. Chest X ray showed a large exophytic osseous mass involving the second right rib.

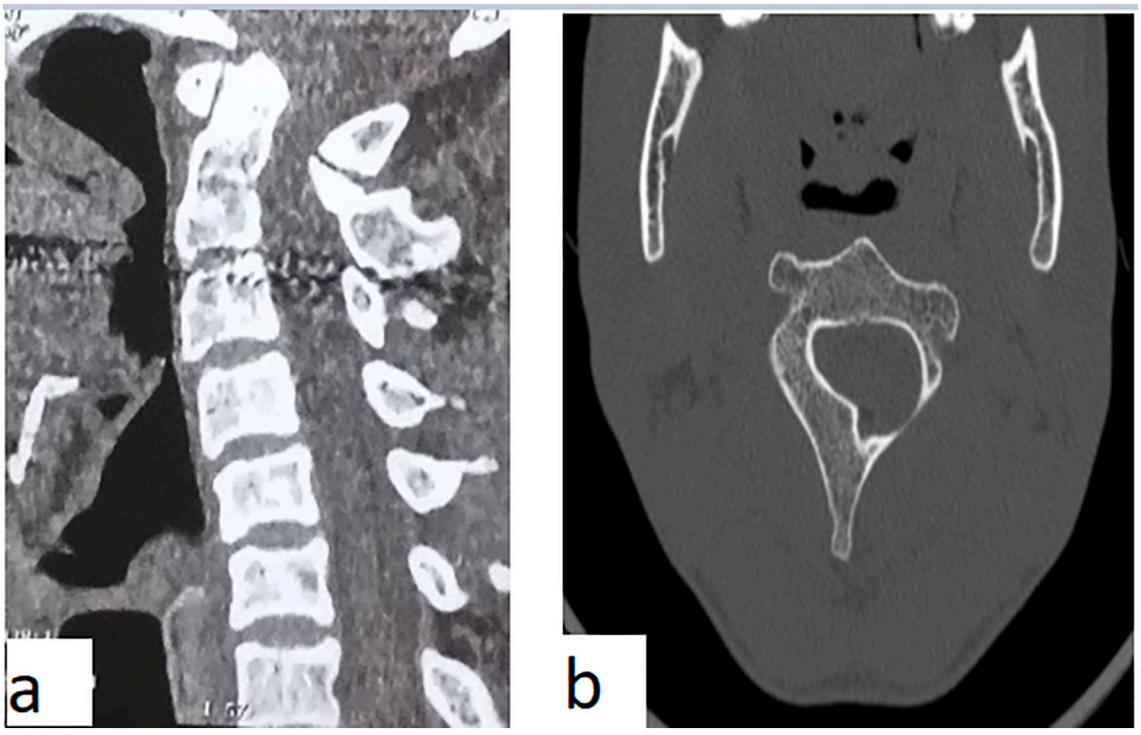

Fig. 3. Sagittal (a) and axial (b) bone window computed tomography scan of the cervical spine showing a bony outgrowth growing anteriorly and filling the spinal canal, arising from the inner posterior arch of C2 at the right side.

and the final decision was to operate him.

A careful intubation using video laryngoscope was performed by a senior physician. The patient underwent a C1-C2 laminectomy. A complete removal of the C1/2 exostoses was performed. The resection was performed through drilling until a complete decompression of the nervous structures could be obtained. A senior surgeon operates the patient.

No stabilization has been proposed as preoperative imaging showed no evidence for instability, and as decompression did not require an arthrectomy.

Postoperative course was uneventful. A foam cervical collar is used for 10 days to reduce pain after surgery. Postoperative cervical CT scan showed a complete excision of exostoses and decompression of the cervical spine cord (Fig. 5). Although the patient had a physical rehabilitation for 6 months, neurologic examination at discharge and half a year after surgery was the same as preoperative clinical features. Histopathological examination confirmed the diagnosis of osteochondroma without signs of malignant degeneration.

3. Discussion

HME also known as diaphyseal aclasis, multiple osteochondromas, Ehrenfried disease, hereditary deforming chondrodysplasia, and osteogenic disease. This pathology was first described by Boyer in 1814 [3]. It is characterized by the development of multiple osteochondromas within long bones (Fig. 6). The incidence of HME is about 1 out of 50,000 living births [4]. It is related to an autosomal dominant disorder, with full penetrance and both genders can be affected with a slight male predominance [5]. A family history was established in approximately 65% of patients with multiple osteochondromas [1]. Osteochondromas are related to growth plates, which explains the fact that their development rate may be increased at puberty. Schmale et al. reported that multiple hereditary exostoses have a penetrance of 50% by the age of 3.5 years and nearly 100% by the age of twelve years [6]. This suggests that most lesions are present at around the age of 10. In almost 92% of HME patients, 2 genes EXT1 and EXT2 located respectively at 8q24 and 11p11-p12, have been isolated [7].

Although most of osteochondromas are located around the proximal humerus and the knee, 20% arise from the axial skeleton [8]. However, Roach reported that 68% of patients with MHE had a spinal exostosis and 27% had lesions encroaching into the spinal canal [9].

Cervical spine is the predilection site for osteochondromas, followed by thoracic and lumbar levels [10]. The most frequent site is the C2 vertebral bone, followed by C3 and C6 [11]. Most of the lesions are found in the posterior elements of the vertebra, with a tendency to arise from the tips of spinous, transverse, and articular processes [12]. Predilection for the atlantoaxial joint, may be related to its important mobility, which makes them predisposed to greater stress and microtraumas. All these factors promote exostic growth [13].

The risk of malignant degeneration into chondrosarcoma is 10–25% [14]. Usually, the origin of malignancy is the cartilaginous cap [15]. Osteochondromas are benign, slow-growing tumors. This is the cause for the slow onset of clinical impairment. But in some cases, acute symptoms may develop following a direct or indirect trauma of the cervical spine [16]. .An anterior encroachment can lead to less common symptoms such as dysphagia, cranial nerve palsy, hoarseness, Horner's syndrome [17].

A careful routine clinical examination within patients with HME looking for neurological symptoms is a mandatory screening tool, in order to diagnose early spinal localizations that may induce neurologic impairment [18]. MRI is the gold standard for detecting spinal cord or roots compression, but CT scan allows a more accurate study for compressing bony elements.

Intraspinal exostoses causing spinal cord compression should indicate surgical excision, whose adequate timing is fundamental to prevent neurologic impairments and sequelae. Surgery aims to obtain decompression of the nervous elements through a laminectomy, associated to resection of the osteochondromas. Usually, after gross resection of superficial bony structures, a drilling is performed in order to minimize aggression towards nervous structures. Stabilization may be discussed

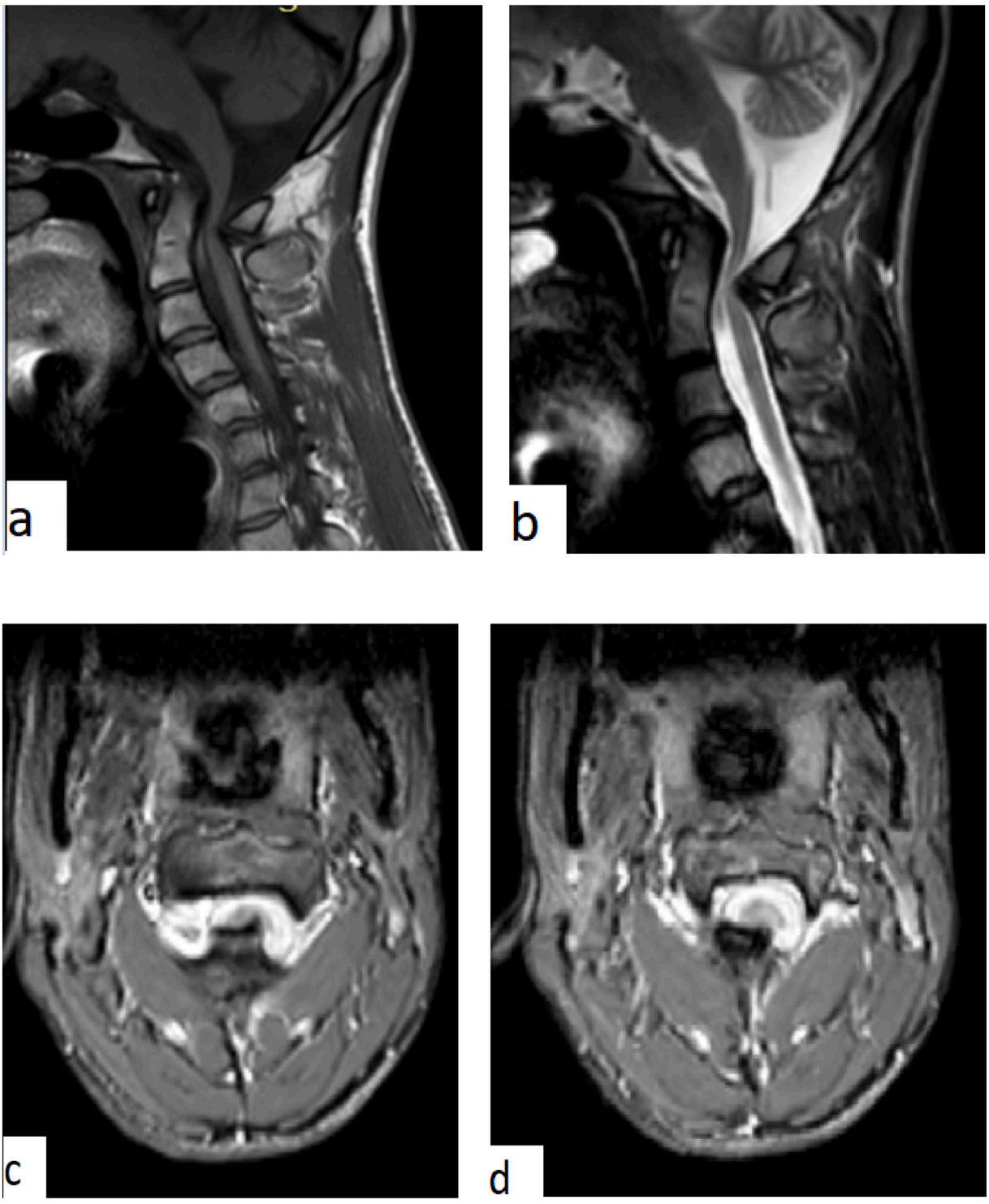

Fig. 4. Sagittal T1 (a) and T2 (b) weighted MR images of the cervical spine demonstrate the marked narrowing of the spinal canal at the C2 level. At the compressed level, the axial T2 (c, d) weighted images demonstrating a C2 posterior mass severely compressing the cord.

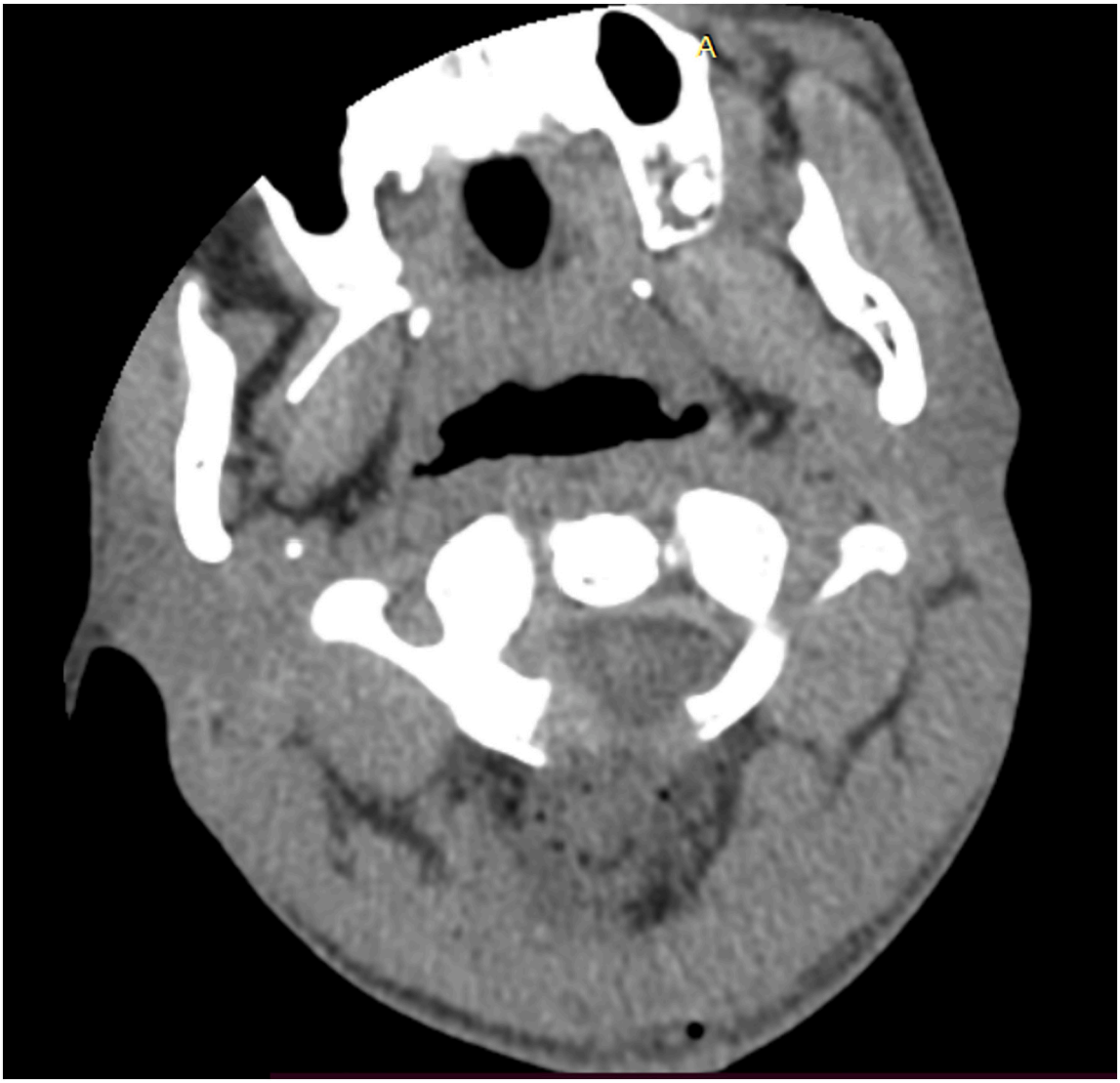

Fig. 5. Post-operative cervical CT scan showing complete decompression of the cervical spine canal at C2 level.

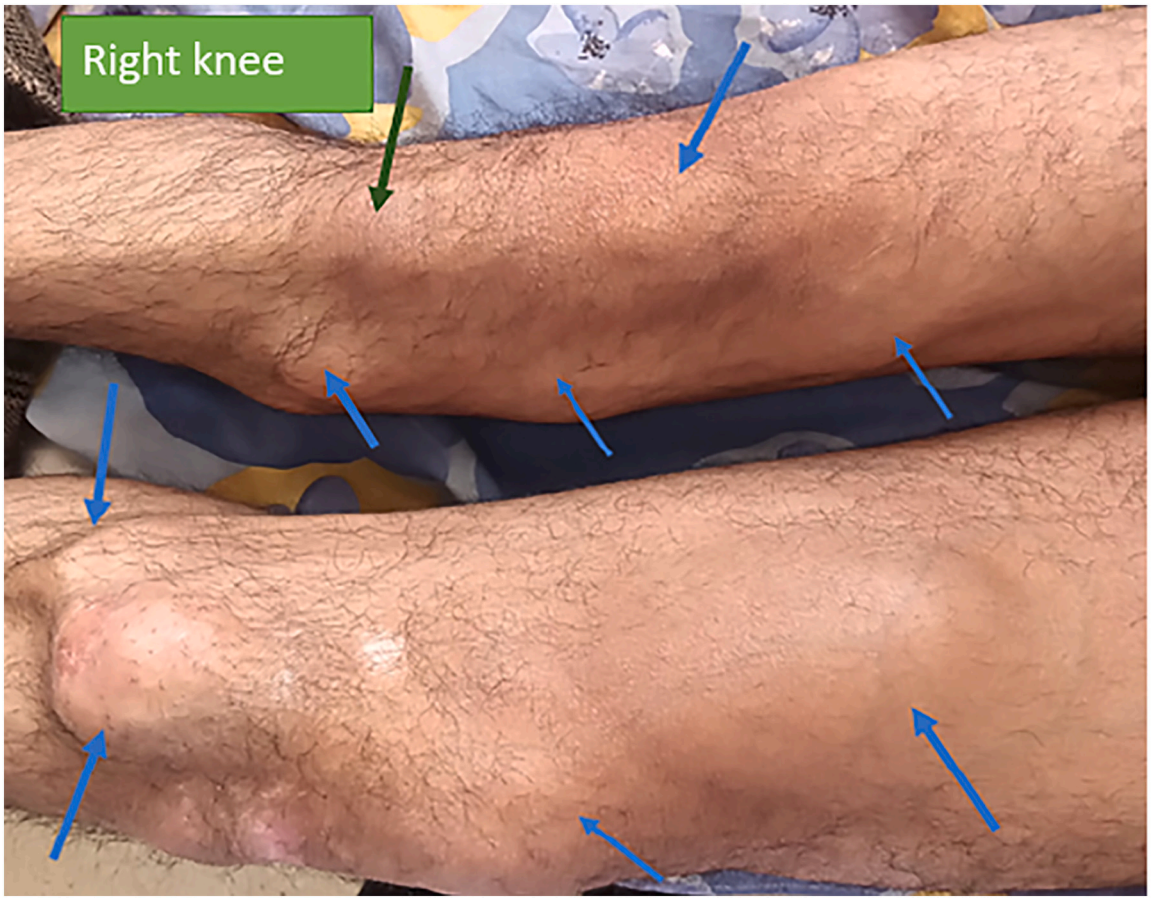

Fig. 6. Multiple exostoses of the lower limbs (blue arrows).

referring to the affected levels, the origin of the exostoses, the age of the patient, and a substantial preoperative instability. The stabilization was not necessary in our case because the facet joint was preserved. Post-operative neurological evolution depends from the timing of the surgery. The recurrence rate is low, as approximately 2–4% of spine osteochondroma recur after an average follow up of 4.2 years [19].

4. Conclusion

Although hereditary multiple exostoses is a rare genetic disorder, it needs a particular attention as treatment and solving the issues of this pathology may be a real challenge. HME patients have generally a worse quality of life than the general population. An early screening of spinal osteochondromas is important to improve this quality of life and to avoid neurological sequelae. Guidelines for the follow up of HME affected patient should be more deeply discussed through larger series, in order to emphasize therapeutic strategies.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Ethical approval

This paper as a case report, therefore does not require ethics approval.

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Jemel Nesrine wrote the paper.
Gader Ghassen edited and reviewed the article.
Bedioui Aziz collected the data.
Zammel Ihsen supervised.
Badri Mohamed supervised.

Declaration of competing interest

None.

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