Case Report

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Disastrous evolution of ollier disease: a rare case report

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Introduction: Ollier disease is a rare genetic disorder characterized by the development of multiple enchondromas. The clinical manifestations of the disease vary widely, but patients often present with bone deformities and an increased risk of developing chondrosarcoma. Here, the authors present a case report of a 25-year-old male patient with a devastating and historic evolution of Ollier disease.

Case presentation: At the age of 10, the patient developed a sub-centimeter mass in the first phalanx of the left middle finger, which subsequently grew in size. A biopsy was performed at the age of 14, which confirmed the diagnosis of chondroma. At the age of 14, the patient developed multiple large masses on the left hand, resulting in the amputation of his left hand. At 25 years old, the patient developed new masses in his contralateral hand and left foot.

Discussion: Ollier disease is caused by somatic mutations in the PTH/PTHrP receptor gene, leading to the formation of multiple enchondromas. Patients with Ollier disease are at an increased risk of developing chondrosarcoma, which can be life-threatening. The diagnosis of Ollier disease is usually made based on clinical and radiographic findings, and genetic testing can confirm the diagnosis. Treatment is typically focused on managing the symptoms and preventing the development of chondrosarcoma.

Conclusion: The authors presented a case report of a patient with a devastating and historic evolution of Ollier disease. This case highlights the importance of early diagnosis and management of this disease to prevent the development of chondrosarcoma and minimize the risk of complications. Further research is needed to better understand the underlying mechanisms of the disease and develop effective treatments.

Keywords: case reports, disease progression, enchondromatosis, multiple/diagnosis, ollier disease/complications

Introduction

Ollier disease, also known as enchondromatosis, is a rare disorder that affects the skeletal system. It is characterized by the development of multiple enchondromas. This disease occurs in less than 1 in 100 000 people and is nonhereditary[1].

Enchondromas typically appear in the hands and feet during early childhood, but they can also develop in other bones of the body. They can cause limb-length discrepancies, skeletal abnormalities, and fractures. The severity of the symptoms varies depending on the location and number of enchondromas present in the body[2].

The diagnosis of Ollier disease is made through a combination of clinical and radiographic evaluations. X-rays can reveal the presence of enchondromas and their location[3].

One of the major complications of Ollier disease is the potential for malignant transformation of the enchondromas into chondrosarcomas. This transformation is rare, but it can occur and is a serious concern for individuals with Ollier disease. Therefore, regular monitoring of the enchondromas is important to detect any signs of malignant transformation early[4].

HIGHLIGHTS

- This case report presents a unique and unprecedented evolution of Ollier disease, with the development of multiple enchondromas extending to the lower limbs.
- While Ollier disease is a rare disorder, this case presents a particularly rare and devastating manifestation of the disease, highlighting the need for early diagnosis and management.
- This case underscores the importance of vigilant monitoring and early intervention to prevent the development of complications.
- The need for further research to better understand the underlying mechanisms of Ollier disease and develop effective treatments to improve outcomes for patients.

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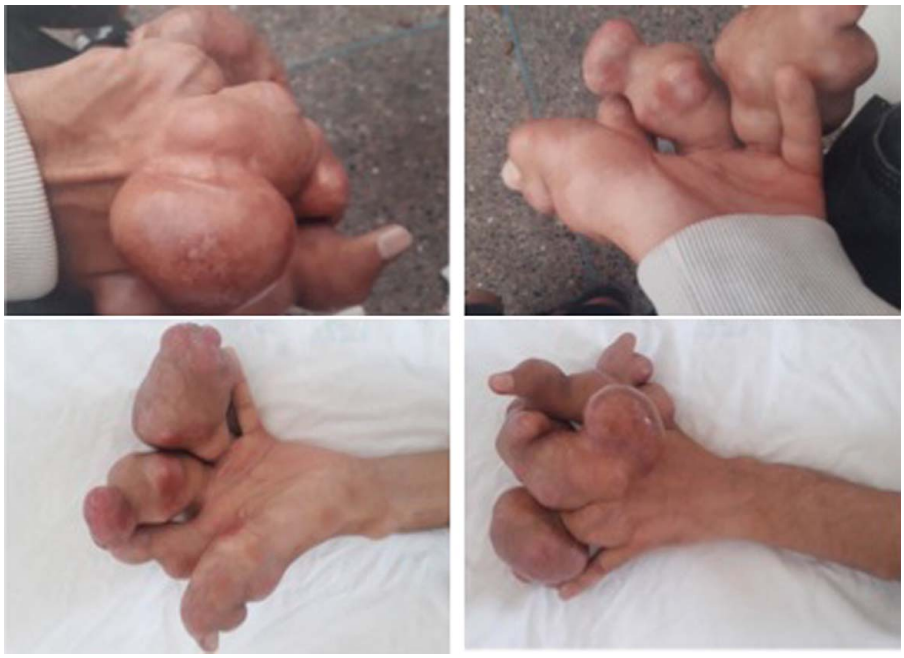

Figure 1. Clinical aspect showing the disastrous evolution of the lesions invading the left hand with the deformity and the collateral venous circulation.

Treatment for Ollier disease is typically focused on managing the symptoms associated with the enchondromas. This can include surgery to correct limb-length discrepancies or fractures, as well as regular monitoring to detect any signs of malignant transformation. In some cases, radiation therapy or chemotherapy may be necessary to treat chondrosarcomas that develop from enchondromas[4].

We report a case of a 25-year-old male with a rare and disastrous evolution of Ollier disease with a 15 years follow-up. This manuscript has been reported in line with Surgical CAse REport (SCARE's) 2020 Criteria[5].

Case presentation

A 25-year-old right-handed male with no significant medical history. His symptoms had begun at the age of 10 when a small mass was noted in the first phalanx of the left middle finger. The mass was 3 cm long, renitent, painless, and infiltrating the interphalangeal joint. The patient benefited of periodic surveillance with physical and radiographic examination. At the age of 15, he developed other masses of the first four fingers with a deformation of the left hand associated to massive collateral venous circulation with asymmetrical distribution. The evolution was very fast, random, and disastrous (Fig. 1).

The radiological aspect of the hand showed a massive evolution of multiple, calcified, geographic, heterogenous lesions with an elongated shape and a typical notch-like image associated with a severe osteolysis and growth abnormality of the carpe and distal extremity of the radius and ulna. Also, a destruction of the metacarpals and the phalanges with a significant disorganization of the bone architecture (Fig. 2). This radiological aspect suggests primitive and benign multiple cartilaginous lesions reminiscent of enchondromas, which was confirmed on histology after a biopsy of a phalangeal lesion in the left hand.

Faced with the destructive aspect of the lesions and the histological confirmation of Ollier disease, a trans-metacarpal amputation was indicated and performed at the age of 21. The fifth finger was spared (Fig. 3).

At the age of 25, he developed new masses on the contralateral limb affecting the fourth finger, as well as on the left foot involving the toes (Fig. 4). Radiographs of the right hand showed multiple lytic lesions with endosteal scalloping, and the radiographs of the left foot showed a lytic lesion in the destructing and deforming metatarsus and phalanges of the toes. Histopathological examination of the masses showed benign cartilage tumors consistent with multiple enchondromas evoking their connection with the general evolution of Ollier disease. These lesions are currently under clinical and radiological surveillance.

Discussion

Ollier disease is a rare disorder that usually presents in childhood with multiple enchondromas. It is an uncommon, nonhereditary

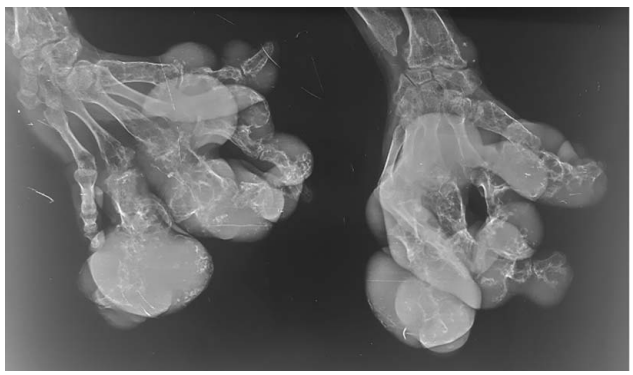

Figure 2. A standard AP and lateral radiograph of the left hand reveals lesions of enchondromatosis with complete disorganization of the hand architecture.

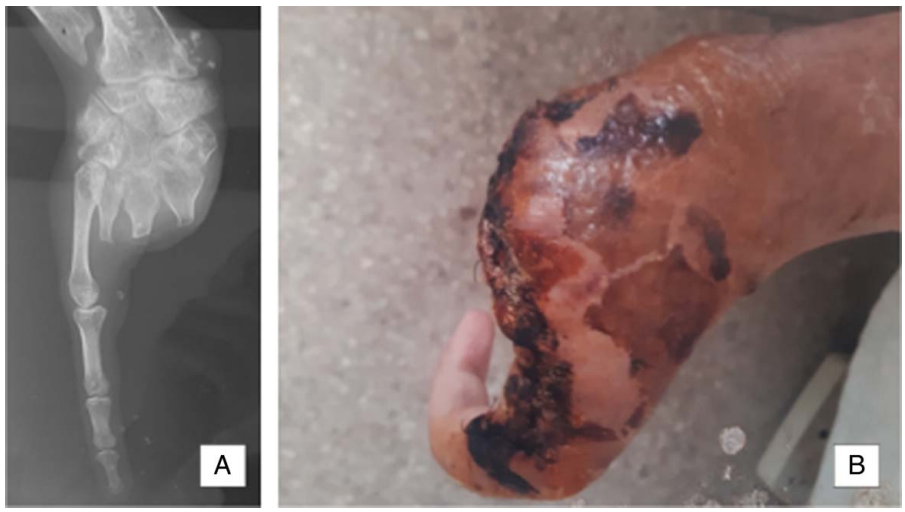

Figure 3. A standard AP and lateral postamputation radiograph of the left hand (A). Clinical aspect of the left hand after the amputation (B).

skeletal disorder characterized by the presence of multiple enchondromas or cartilaginous masses in the metaphysis and diaphysis of long bones or short tubular bones of the hands and feet. The tumors can cause deformities, fractures, and limb-length discrepancies[6]. The diagnosis of Ollier disease is usually made by clinical and radiographic examination. The enchondromas can undergo malignant transformation to chondrosarcoma in rare cases[4]. In our case, the patient had a disastrous and historic evolution of the disease, with multiple masses developing in various locations, leading to significant loss of hand function and amputation.

Given the patient's extensive history of multiple enchondromas, a diagnosis of a massive evolution of Ollier disease was established. It is a rare nonhereditary disorder characterized by the development of multiple enchondromas, which are benign cartilage tumors, throughout the bones of the body[6]. The disease usually appears in childhood and can affect any bone, but commonly involves the long bones of the limbs[7].

The pathogenesis of Ollier disease is not well understood. It is thought to be caused by a somatic mosaic mutation of the PTH/PTHrP receptor gene that occurs during embryonic development and affects the differentiation of mesenchymal cells into chondrocytes[8]. The mutations lead to the formation of abnormal cartilage nodules or enchondromas. The multiple enchondromas can cause a variety of problems such as limb-length discrepancy, joint deformities, and pathological fractures.

The hand's small tubular bones are the most commonly affected by Ollier disease, followed by the femur, tibia, fibula, humerus, radius, and ulna[8]. Size, location, number, age of onset, and diagnosis of enchondromas can all vary. It represents a typical chondrosarcoma form. Patients with the condition may have skeletal abnormalities as bending, shortening, pathological fractures, and asymmetric deformity. These patients typically exhibit an angular deformity or growth disparity for the first time when they are still young, peaking before the age of 10. Several enchondromas in adults can lead to skeletal complications as arthritis, deformity, or malignant change[7]. Both of the aforementioned two situations fit these qualifications.

There is currently no cure for Ollier disease, and treatment is focused on managing the symptoms and complications associated with the disease. Treatment options include surgical excision of symptomatic enchondromas, orthopedic management of limb deformities, and regular monitoring for the development of malignant transformation, which occurs in 10–20% of cases[9,10].

Our case report highlights the devastating and historic evolution of Ollier disease in a 25-year-old male patient with a long history of multiple enchondromas. The patient had an early onset of the disease at the age of 10 years with the appearance of a small mass on the left middle finger. The diagnosis was confirmed with a combination of clinical, radiological, and histopathological findings. Over time, the patient developed multiple large masses on his left hand and eventually lost all function of his left hand, leading to the need for amputation. The disease continued to progress, with the appearance of new masses on the contralateral limb and foot.

The rarity of Ollier disease and the severe complications associated with it make this case report particularly relevant. The case emphasizes the need for regular monitoring of patients with Ollier disease for the development of malignant transformation and the importance of early intervention to prevent severe complications. It also highlights the need for more research into the pathogenesis of Ollier disease and the development of more effective treatments for this rare disorder.

This case highlights the importance of considering Ollier disease as a differential diagnosis in patients presenting with multiple enchondromas.

The management of Ollier disease involves close monitoring of the lesions for malignant transformation, as well as management of complications such as limb deformities, fractures, and nerve compressions. Surgical intervention is reserved for cases with symptomatic or aggressive lesions[11]. In our case, amputation of the left hand was necessary due to the extensive involvement of multiple fingers and loss of hand function. Unfortunately, despite surgical intervention, the patient continued to develop new lesions in other limbs[12].

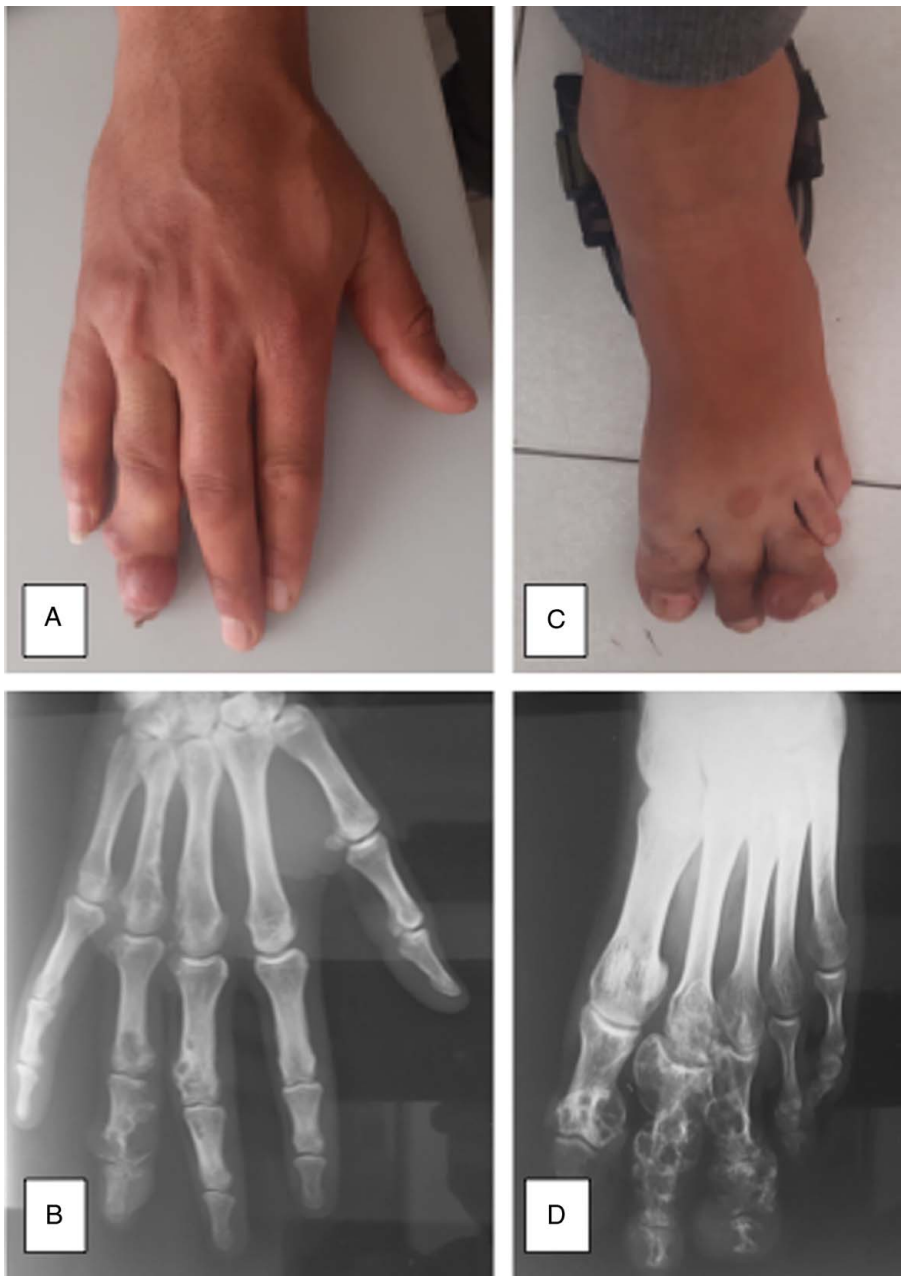

Figure 4. Topography and radiography of enchondromas with extension to the contralateral hand (A–B) and left foot (C–D).

Conclusion

In conclusion, our case report describes a rare presentation of Ollier disease, with an extensive and debilitating evolution affecting multiple limbs of a young male patient. It is a rare disorder characterized by multiple enchondromas and can be challenging to diagnose, especially in a typical presentations[11]. It can cause severe complications such as limb deformities, joint problems, and pathological fractures. The disease is not well understood, and there is currently no cure. Our case report emphasizes the devastating and historic evolution of Ollier disease and highlights the need for regular monitoring and early intervention to prevent severe complications. More research is needed to understand the pathogenesis of the disease and to develop more effective treatments for this disorder.

Ethical approval

This study is exempt from ethical approval from the institution.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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None.

Provenance and peer review

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