Incidental diagnosis of familial osteopoikilosis in a patient firstly complaining of ankle pain

Abstract ID : 1211
Submitted by : Sergio Figueiredo the 2016-02-11 17:58:14
Category : Others
Typology : Poster
Status : Validated
Authorisation to disclose : Yes/Oui

Introduction and Objectives: Familial osteopoikilosis is a rare, autosomal dominant, benign sclerosing bone dysplasia. Its diagnosis is mostly done when an X-ray is requested for any other reason. In plain films, small round shaped bone cysts can be found around major joints, with predilection for long tubular bones. Our aim is to describe the diagnosis process of an atypical presentation of familial osteopoikilosis through an observed clinical case.

Methods: A single case report.

Results: We hereby report the case of a 32 year-old woman, complaining of left non-traumatic ankle pain. Foot and ankle X-rays revealed anterosuperior calcaneotalonavicular fusion, but also several hyperlucent cystic lesions around the articular surface of both ankle and foot bones. In fear of a malignant disease, the patient was further studied through additional films and an ankle magnetic resonance imaging scan. All of them revealed the same lesions all around wrists, hands, hips, knees, ankles and feet. The same were then found in both siblings but not in her parents, none of which presented any kind of signs or complaints. Genetic testing revealed a mutation in the LEMD3 gene, thus confirming the diagnosis.

Discussion/Conclusion: Diagnosis confirmation of osteopoikilosis is mandatory, as malignant threats may appear similar on imaging studies. Inspite of the great aid provided by genetic testing, sibling resemblance and knowing disease penetrance as not being complete would allow us to safely keep disease vigilance without major fears of severe prognosis.

Keywords : Familial Osteopoikilosis, Buschke-Ollendorff syndrome, LEMD3 gene mutation, Myelopathy, Benign Dysplasia

Authors

Sergio Figueiredo 1, Ana Daniela Pereira 2, Luis Machado 2, Joao Andre Silva 2, Pedro Cardoso 3, Antonio Sa 2,

1. Ortopedia 2. Centro Hospitalar de Leiria, Leiria, PORTUGAL
2. Orthopaedics 2, Centro Hospitalar de Leiria, Leiria, PORTUGAL
3. Orthopaedics, Centro Hospitalar do Porto, Oporto, PORTUGAL

Authors (raw format)

Figueiredo Sergio - email : sergio.figueiredo@gmail.com Institution : Centro Hospitalar de Leiria Department : Ortopedia 2 City : Leiria Country : PORTUGAL Speaker : Yes
Pereira Ana Daniela - email : analousada@live.com.pt Institution : Centro Hospitalar de Leiria Department : Orthopaedics 2 City : Leiria Country : PORTUGAL Speaker : No
Machado Luis - email : luis.miguel.machado@gmail.com Institution : Centro Hospitalar de Leiria Department : Orthopaedics 2 City : Leiria Country : PORTUGAL Speaker : No
Silva Joao Andre - email : joaioandresilva@netcabo.pt Institution : Centro Hospitalar de Leiria Department : Orthopaedics 2 City : Leiria Country : PORTUGAL Speaker : No
Cardoso Pedro - email : pfcardosoc@gmail.com Institution : Centro Hospitalar do Porto Department : Orthopaedics City : Oporto Country : PORTUGAL Speaker : No
Sa Antonio - email : antoniosa933@gmail.com Institution : Centro Hospitalar de Leiria Department : Orthopaedics 2 City : Leiria Country : PORTUGAL Speaker : No