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Challenge in Diagnosing Osteopoikilosis: A Case Report

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Abstract

BACKGROUND: Osteopoikilosis (OPK) is a rare benign osteosclerotic dysplasia and occurs in 1/50,000 people. OPK is inherited in an autosomal dominant and associated with several clinical manifestations. At present, there is no agreement on diagnosing OPK. In this case report, we describe a 24-year-old female patient complaining of a lump and pain in the sole of the right foot.

CASE PRESENTATION: A 24-year-old female complained of a painful lump on the right pedis for 1 year. On physical examination, a mass was found at the sole of the right foot with no sigsn of inflammation or trauma, well-marginated and a dimension of 1cm x 0,5 cm x 0,5 cm. We perform a radiographic examination including bone survey and found multiple homogenous sclerotic lesions were spread over almost all visualized bony structures with oval to round in shape, varied in size, and well-defined borders. The laboratory examination shows normal results. Based on the findings described above, we diagnosed the patient with OPK. The patient was provided with analgesics as therapy and periodic observation.

CONCLUSION: OPK is a rare case and is generally found incidentally on radiographic examination. The combination of history taking, clinical manifestations, and typical radiographic findings is sufficient to establish the diagnosis. This can prevent unnecessary examinations or invasive procedures.

Introduction

Osteopoikilosis (OPK) (OPK; osteopathia condensans disseminated, spotted bones) is a rare benign osteosclerotic dysplasia and was first described by Alberg Schonberg in 1915 [1], [2]. OPK occurs in about 1/50,000 people, there is no difference between men and women and can occur at any age, but is mostly found in young adults [3]. OPK is inherited in an autosomal dominant associated with SMAD regulation in the LEMD3 gene [4]. OPK is associated with several clinical disorders such as dermatofibromas, scleroderma, keloids, syndactyly, dacryocystitis, bone disorders, endocrine dysfunction, renal malformations, and facial or dental abnormalities [5], [6].

Approximately 15–20% of cases came with main complaint of pain but the majority are asymptomatic and diagnosed incidentally on radiographic examination. Radiographic findings of OPK are homogeneous, well-defined, and multiple osteoblastic symmetrical sclerotic lesions 1–10 mm in size and round or oval [1], [3], [7]. At present, there is no agreement on diagnosing OPK. In this case report, we describe a 24-year-old female patient complaining lump and pain in the sole of the right foot.

Case Description

A 24-year-old female student complained of a painful lump on the right pedis for 1 year. The lump felt soft and did not get bigger but was getting more painful. The pain worsens with strenuous activities such as walking and relief with analgesics. She denied any unexplained weight loss, fever, and history of trauma. At this time, the patient could walk and move without limitation.

On physical examination, a mass was found at the sole of the right foot with no sigsn of inflammation or trauma, well-marginated and a dimension of 1cm x 0,5 cm x 0,5 cm. A 1 cm × 0.5 cm × 0.5 cm painful lump was found in the right pedis in the second metatarsophalangeal digits of the plantar during palpation. The lump was warm, soft, well-demarcated border, smooth surface, and immobile. We suspected that the mass was located under the subcutaneous tissue. The range of motion of the joint is obtained without limitation, either actively or passively.

A radiographic examination was performed for further investigation. On plain radiograph examination of the right foot with anteroposterior and

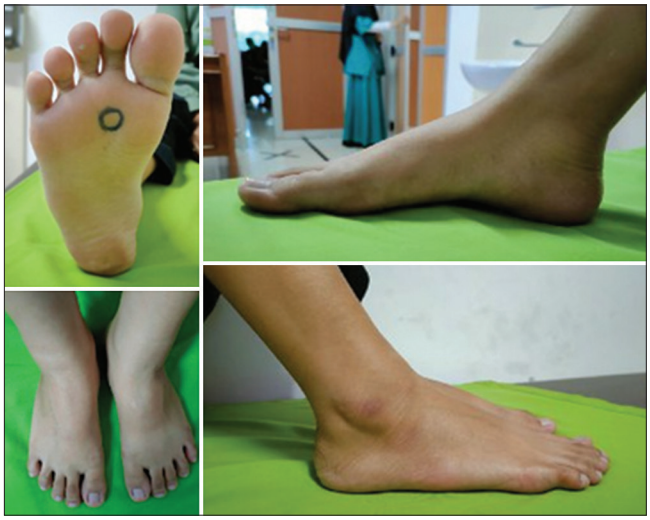

Figure 1: Clinical findings of the patient. Painful lump in second digit metatarsophalangeal of the plantar

oblique views, multiple homogenous sclerotic lesions were found. The lesions were spread over almost all visualized bony systems with oval to round in shape, varied in size, and well-defined borders (Figure 2). The examination was continued with a plain X-ray examination of the pelvis and a bone survey with suspicion of metastases.

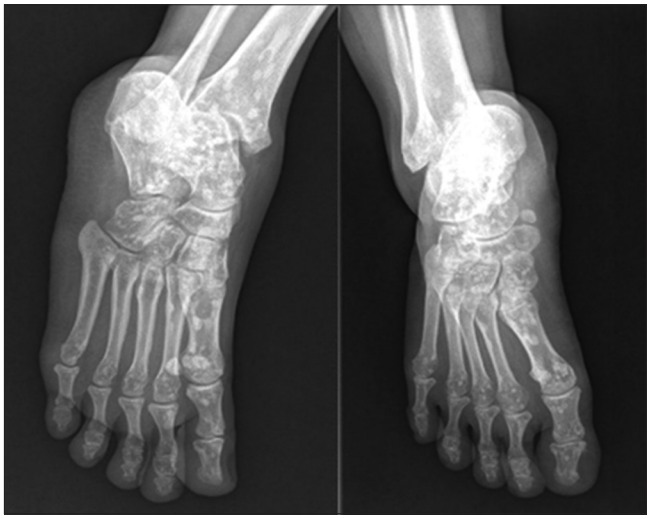

Figure 2: Plain radiograph of the right pedis in anteroposterior and oblique positions. Multiple homogenous osteoblastic lesions, oval to round in shape with varying in size, and firm boundaries

Plain radiographs of the pelvis and bone surveys showed identical results to plain radiographs of the right pedis (Figure 3).

In the laboratory examination, complete blood counts, markers of inflammation, and blood chemistry were performed with insignificant results. There was a slight increase in CEA and ESR with normal results in other parameters. Based on the findings described above, we diagnosed the patient with OPK. The patient was provided with analgesics as therapy and periodic observation of the patient.

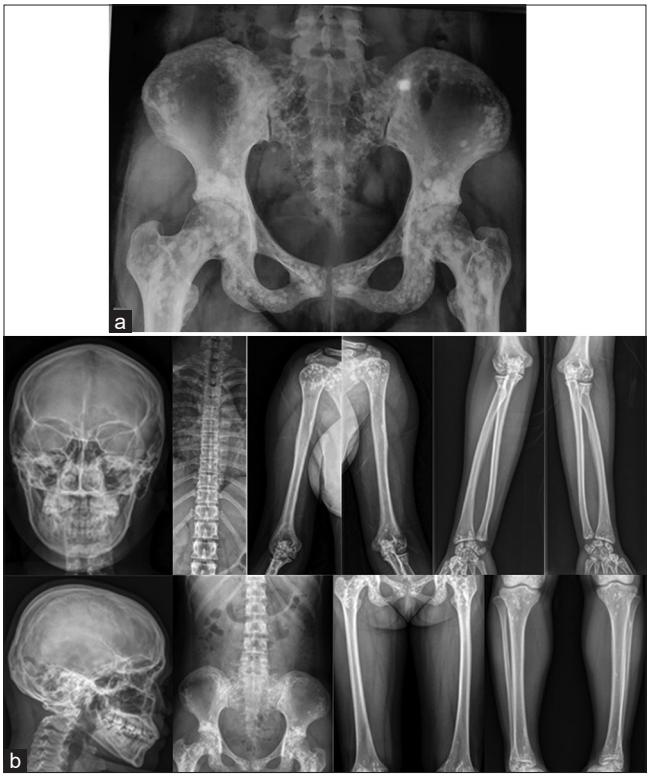

Figure 3: (a) Plain radiograph of pelvic anteroposterior positions. (b) Bone survey of the result was identic with Figure 2. Multiple homogenous osteoblastic lesions, oval to round in shape, varying in size with well-defined borders on the pelvis, bilateral femurs, bilateral cruris, bilateral pedis, bilateral humerus, bilateral antebrachia, and bilateral manus

Discussion

OPK has an incidence of about 1/50,000 but the diagnosis of OPK occurs incidentally so the incidence rate is estimated to be underreported [2], [6], [8], [9]. OPK is benign and inherited in autosomal dominant patterns [1], [2], [8], [10]. This inheritance is related to SMAD regulation of the LEMD3 gene [10]. This is thought to have caused the failure of secondary spongious bone resorption [2]. In the study by Benli *et al.*, 1992, a follow-up investigation of the families of four patients who had OPK found that 53 of the 95 people who were investigated were diagnosed with OPK [1], [10]. There are several clinical conditions associated with OPK. Buschke-Ollendorff syndrome is a condition associated with dermatofibromas and OPK. This is thought to be due to the over-proliferation of fibroblasts which is in line with the pathogenesis of dermatofibromas. Approximately 25% of OPK patients present with skin manifestations [1], [2]. Cardiac abnormalities, urogenital system abnormalities, facial abnormalities, diabetes mellitus, and skeletal system

disorders have also been reported to be associated with OPK. Approximately 15–20% of patients with OPK present with mild pain [1], [2], [9]. In the case, we reported, a painful lump on the patient's right foot was the only complaint, although according to the literature, the majority of OPK patients were asymptomatic. There were no other symptoms or other clinical conditions associated with OPK in the patient we reported. We also did not carry out further investigations into the patient's family caused by insurance issues, so there were no data that showed us similar diagnostic information in the patient's family.

The sclerotic lesions of OPK usually develop in children and persist, the lesions rarely disappeared when adults but their size and number may change [1], [2]. Characteristics of plain radiographic findings on OPK are multiple symmetrically sclerotic lesions, with sizes varying between 1 and 10 mm, well-defined, homogeneous circular, or ovoid [1], [8], [9], [11]. The sclerotic lesions of OPK are more common on the appendicular skeleton rather than on the axial skeleton, more common in the epiphyses and metaphysis of long bones, phalanges, tarsals, carpals, and pelvis, and are bilateral [1], [2], [8], [10]. Based on the characteristic plain radiography appearance, the most common condition resembling OPK is osteoblastic metastatic lesion. Findings of osteoblastic metastatic lesion characterized by asymmetric sclerotic lesions, generally found in the axial skeleton, periosteal destruction, and reaction, and increased uptake on the bone scan [1], [2], [8]. Accordingly, OPK and osteoblastic metastases can be distinguished by their characteristics and predilection for bone lesions. Other conditions that have characteristic radiological findings similar to OPK are mastocytosis, sclerosis tuberoses, melorheostosis, and osteoma. OPK with mastocytosis can be distinguished by several characteristics including their symptoms and radiographic findings. In mastocytosis, the common symptoms are skin manifestations such as urticaria pigmentosa. Bone lesions are only found in 70–75% of cases and are commonly found in axial bones. Bone lesions are generally mixed types (sclerotic and lytic) and accompanied by pain [12]. The findings of history taking, physical examination, and radiographic examination including bone survey in this patient were consistent with the characteristics of the OPK lesion compared to an osteoblastic metastatic lesion or mastocytosis.

Bone scan examination can also be done if the diagnosis cannot be established. A negative bone scan can exclude a metastatic lesion, although a positive bone scan does not exclude a diagnosis of OPK. We did not perform histopathological examination because the diagnosis of OPK was already established. We propose that the diagnosis of OPK can be made by a combination of history taking, clinical manifestations, and radiographic examination. This is supported by the cases we reported and the literature we encountered.

Therapy given to patients with OPK is symptomatic treatment. Analgesics are commonly used in patients with OPK. Bone targeting therapy such as bisphosphonates in patients with OPK is still controversial [2], [8]. We provide pain relief for the patient in the first 5 days and suggest regular monitoring due to OPK associated and postulated can change into malignancy such as osteosarcoma, giant cell tumor, and chondrosarcoma [10]. We decide to stop the follow-up the patient after 3 months.

Conclusion

OPK is a rare case and is generally found incidentally but should still be considered in the differential diagnosis of patients with radiographic findings of sclerotic lesions. The combination of history taking, clinical manifestations, and typical radiographic findings is sufficient to establish the diagnosis. This can prevent unnecessary examinations or invasive procedures.

Additional Information

Human subject

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