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Polyostotic Fibrous Dysplasia Affected Craniofacial with Thoracic and Lumbar Spine in Adolescents Treated with A Long Segment Spinal Fusion: A Rare Case

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ABSTRACT

Fibrous dysplasia (FD) is a benign tumor-like bone condition. It affects the predilection of long and craniofacial bones, and the prevalence of spine involvement is very low. Most patients with fibrous dysplasia are asymptomatic. However, fibrous dysplasia may be painful or cause swelling. Invasion of medullar bone with fibrous tissue leads to a fragile structure of bone with a risk of fracture and multiple complications [1]. We report a case of polyostotic fibrous dysplasia of craniofacial with spinal involvement affecting the thoracic and lumbar spine simultaneously with severe back pain that came to the emergency department. A Male 13 years old presented with severe back pain that started intermittently four weeks before he came to an orthopedic surgeon. His symptoms were both diurnal and nocturnal, worsening with fatigue and everyday activity. His parent's patient started showing enlargement of the maxilla and mandible at seven years old, and he started to show a kyphotic spine deformity at 8. On physical examination, tenderness of the thoracal and lumbar spine was noted. In the thoracolumbar MRI, there were multiple lesions on the thoracal and lumbal vertebra. The patient was treated with posterior laminectomy and long posterior stabilization and fusion. Satisfactory results were achieved, and there were no complications. The extreme rarity of the disease and its presentation imposes rigorous investigations to rule out malignancies. Surgical treatment was indicated for severe back pain with posterior stabilization and decompression. The surgical treatment gave a good functional outcome and improved the visual analog score.

1. INTRODUCTION

Fibrous dysplasia (FD) is a benign tumor-like bone condition. It is characterized by replacing normal cancellous bone in the medullary canal with immature fibrous tissue. Clinically, FD is usually asymptomatic, and when symptoms occur, they manifest as pain, swelling, and deformity [2]. It affects the predilection of

long and craniofacial bones, and the prevalence of spine involvement is very low [3]. In general, FD presents three forms monostotic, polyostotic, and polyostotic. FD with endocrinopathies, which can be associated with hyperpigmentation and endocrinological disorder, is called McCune–Albright syndrome [2], [3].

Fibrous dysplasia most commonly presents in the teens or twenties. It may occur in any bone but is most common in the proximal femur, tibia, ribs, and skull. It affects slightly more males than females. Most patients with fibrous dysplasia are asymptomatic. However, fibrous dysplasia may be painful or cause swelling. It can cause repeated pathologic fractures or severe bone deformity, such as the "shepherd's crook" varus deformity of the proximal femur.

Involvement of the spinal column in either monostotic or polyostotic form is uncommon. Most cases of polyostotic fibrous dysplasia of the spine involve the appendicular skeleton. Findings on imaging can be highly suggestive of the diagnosis when they show typical features. However, spinal lesions can rarely show up as lytic lesions and can mimic metastasis [1], [3].

Invasion of medullar bone with fibrous tissue leads to a fragile structure of bone with a risk of fracture and multiple complications [1]. We report a case of polyostotic fibrous dysplasia of craniofacial with spinal involvement affecting the thoracic and lumbar spine simultaneously with severe back pain that came to the emergency department. This article was compliant with the SCARE guidelines 2020 [4].

2. CASE REPORT

A Male 13 years old presented with severe back pain that started intermittently four weeks before he came to an orthopedic surgeon. His symptoms were both diurnal and nocturnal, worsening with fatigue and everyday activity. He sought analgesics, went to general orthopedics in Meulaboh Region Aceh, and already performed both femur x-ray. He had no particular family or personal history of inflammatory diseases, and there was no history of trauma or falls. Four days before surgery, the patient felt severe back pain with a visual analog scale of 8 out of 10, and he came to the emergency department because the pain was out of proportion and did not subside with oral analgetics.

From the history taken from his parents, the patient started showing enlargement of the maxilla and mandible when he was seven years old, and he started to show a kyphotic spine deformity at eight years old, but they did not bring him to seek medical advice. However, the patient was able to perform the daily activity without any limitations. There was unremarkable family history.

Upon inspection, he had a deformity and asymmetry of craniofacial slight, kyphotic posture in the thoracal and lumbal region, and there was a café au lait spot skin lesion on the right thoracolumbar region. On palpation, the thoracic and lumbar spine were tender. Thoracal and lumbar spinal movements were very limited because of severe back pain. There was no radicular pain, and the neurologic examination was normal.

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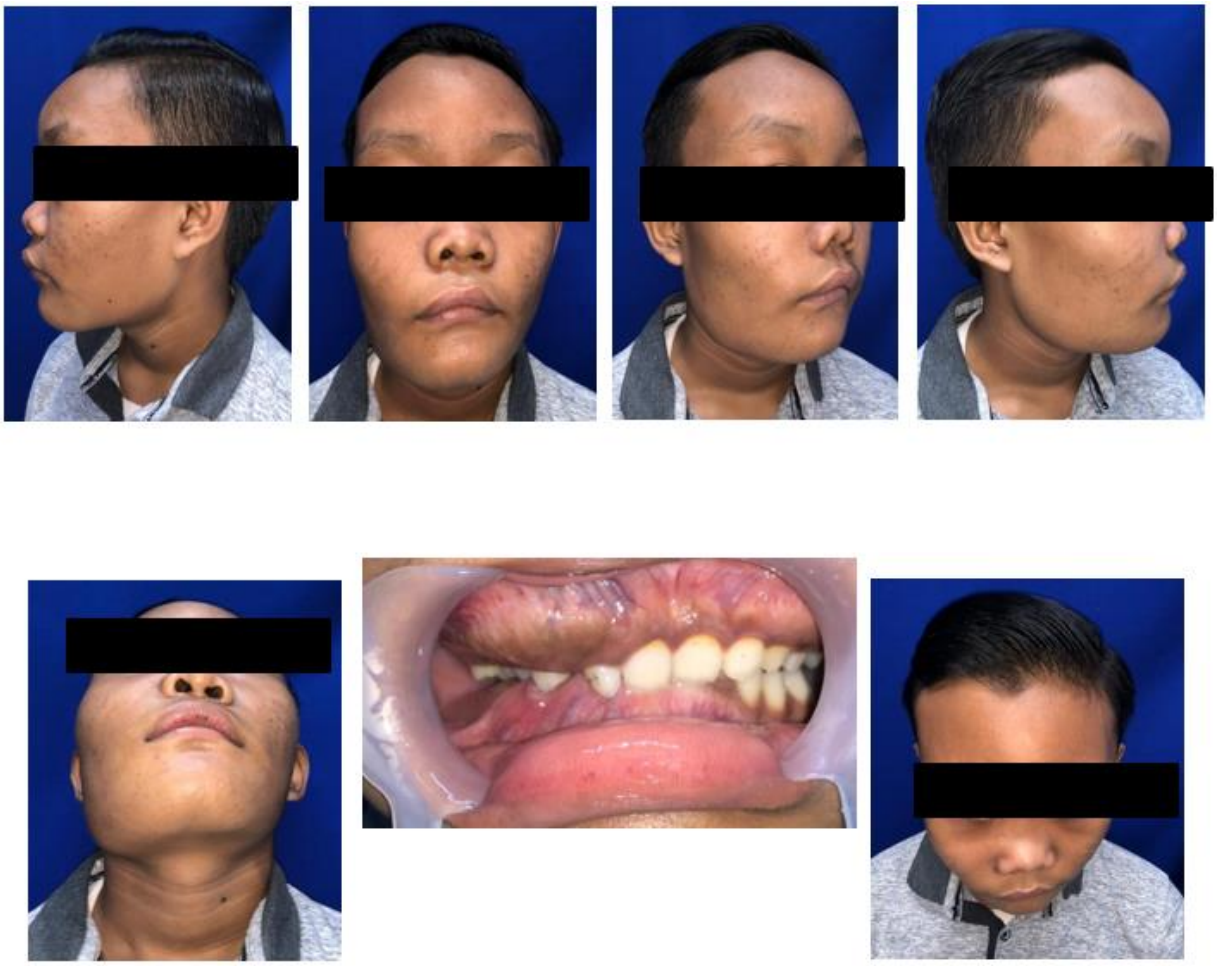


Fig 1. Craniofacial Clinical Picture

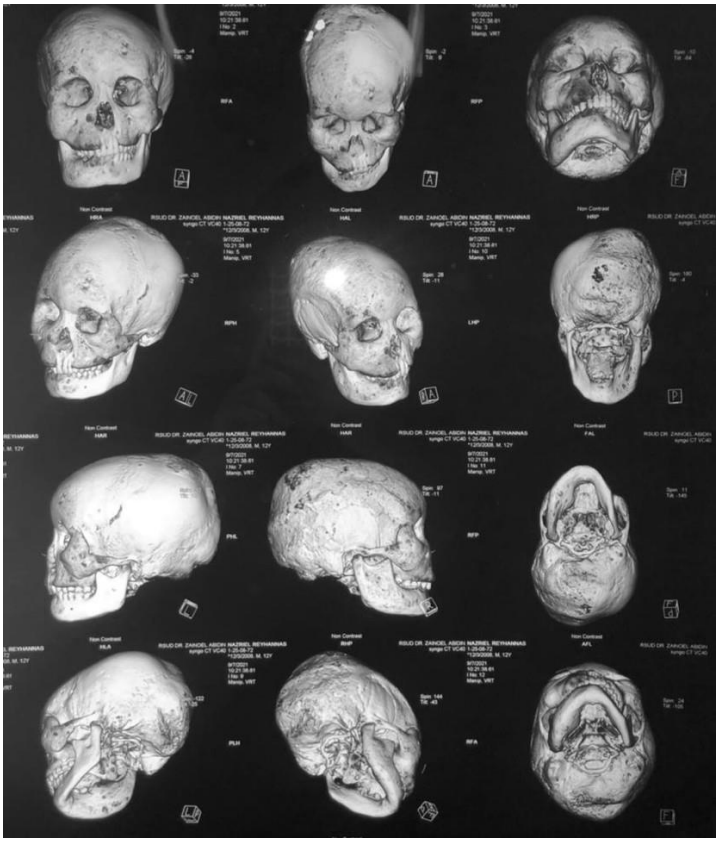
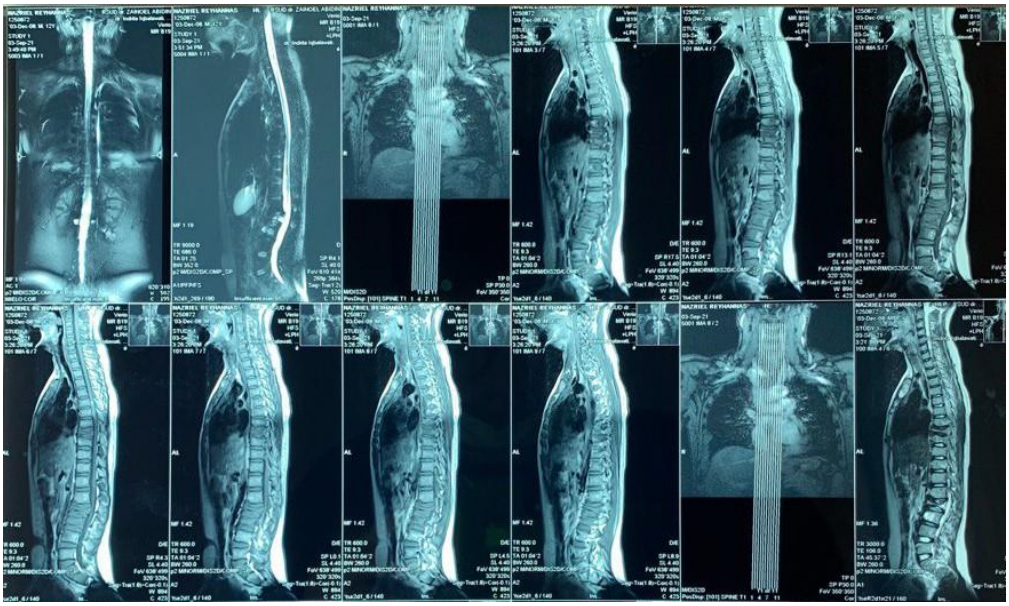


Fig 2. CT-Scan 3D Craniofacial



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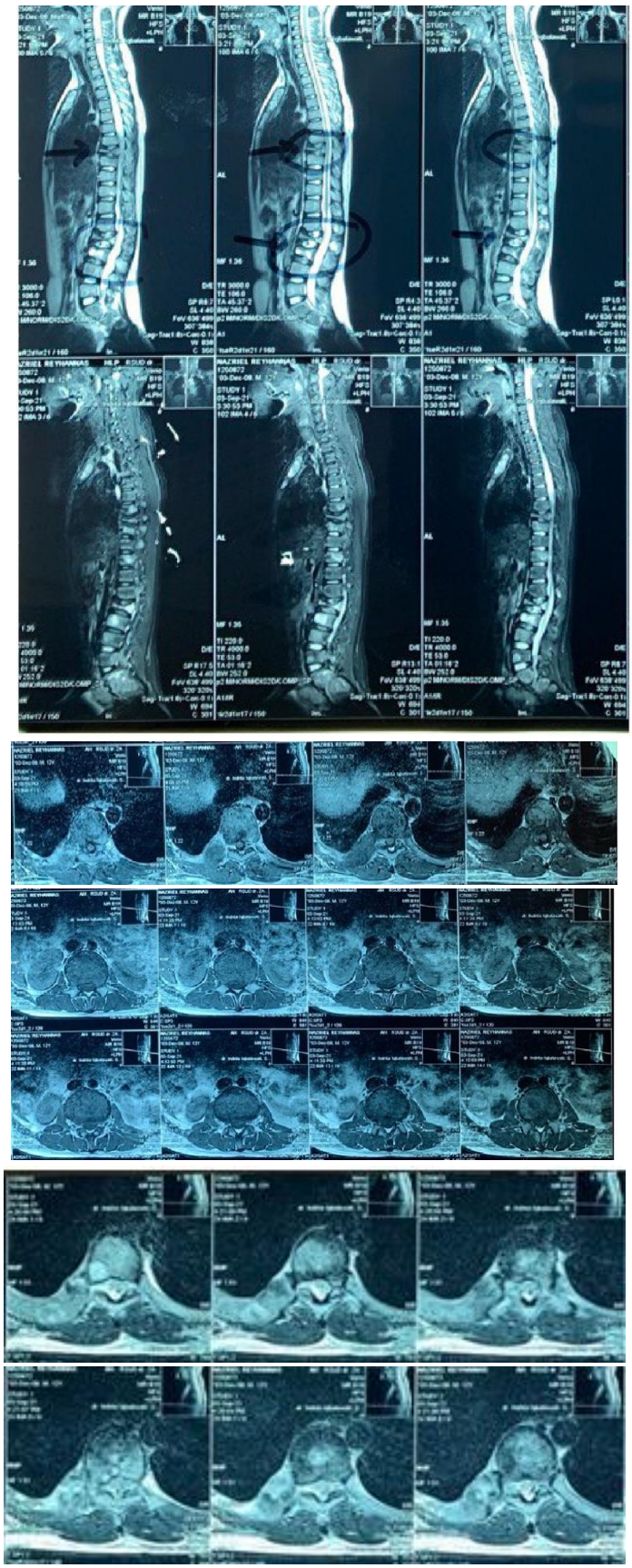

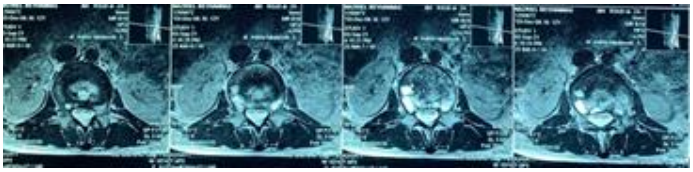

Fig 3. MRI Pre Operation

The patient performed an MRI of the spine, and on T2-Weighted Images, there was a burst fracture on the Th9 and L2 vertebra; on T1-Weighted Images, there were multiple lesions on the thoracal and lumbal vertebra. Sagittal T1 weighted magnetic resonance imaging (MRI) showed low to iso-signal intensity lesions of thoracic and lumbar vertebrae with a hypointense peripheral signal. Sagittal T2 weighted MRI showed iso to high-intensity lesions of the vertebrae body.

The patient underwent laminectomy decompression on the Th9 and L2 and long segment posterior stabilization from Th5 until L5 by the experience orthopaedic surgeon.

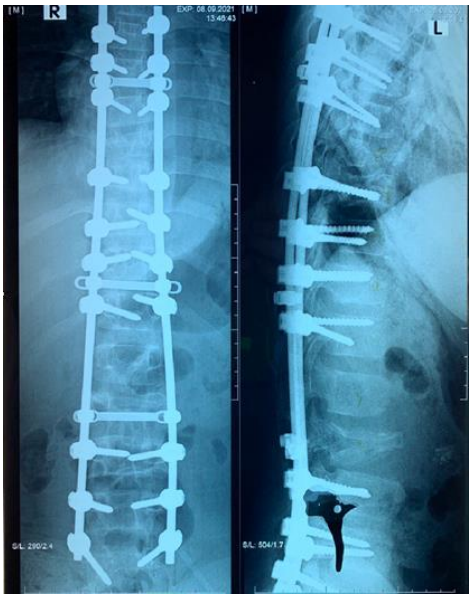

Fig 4. Xray Post Operation

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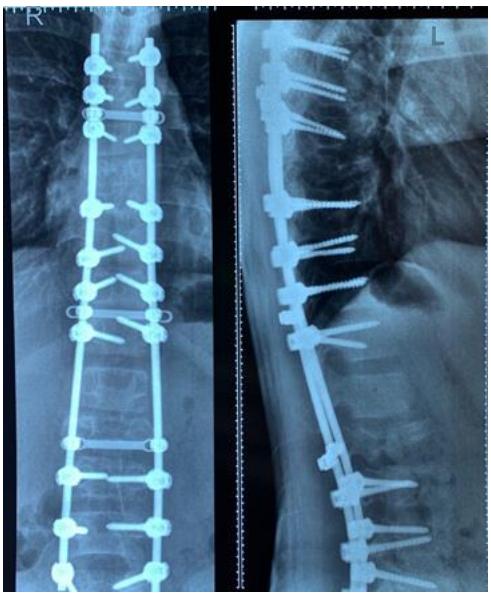
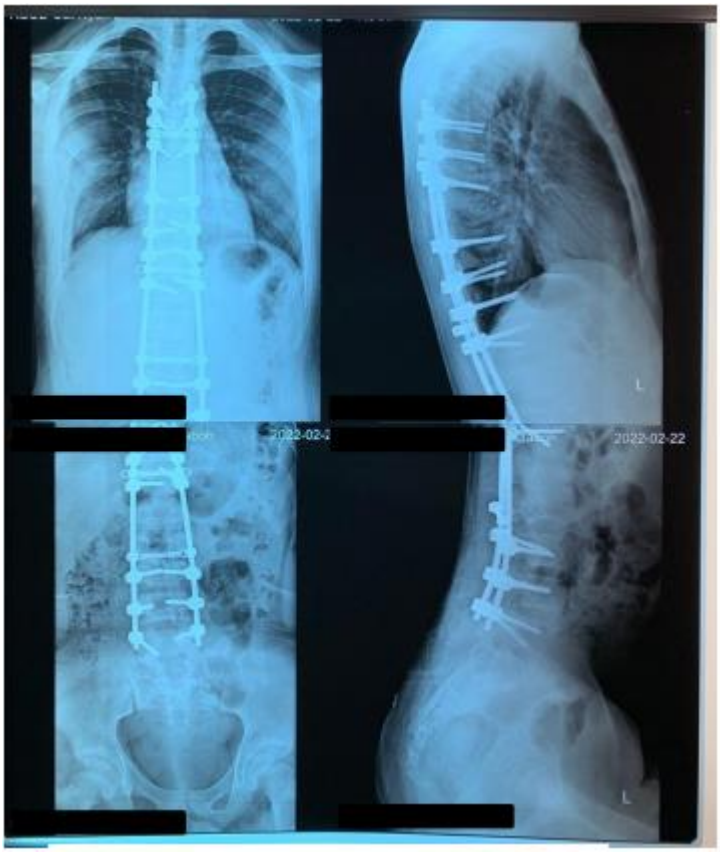


Figure 5. X-ray 2 months post-op



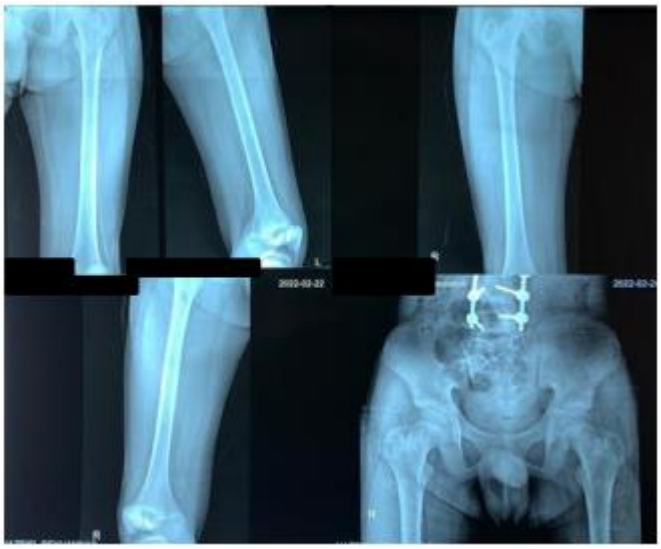


Fig 7. X-ray 6 months post operation

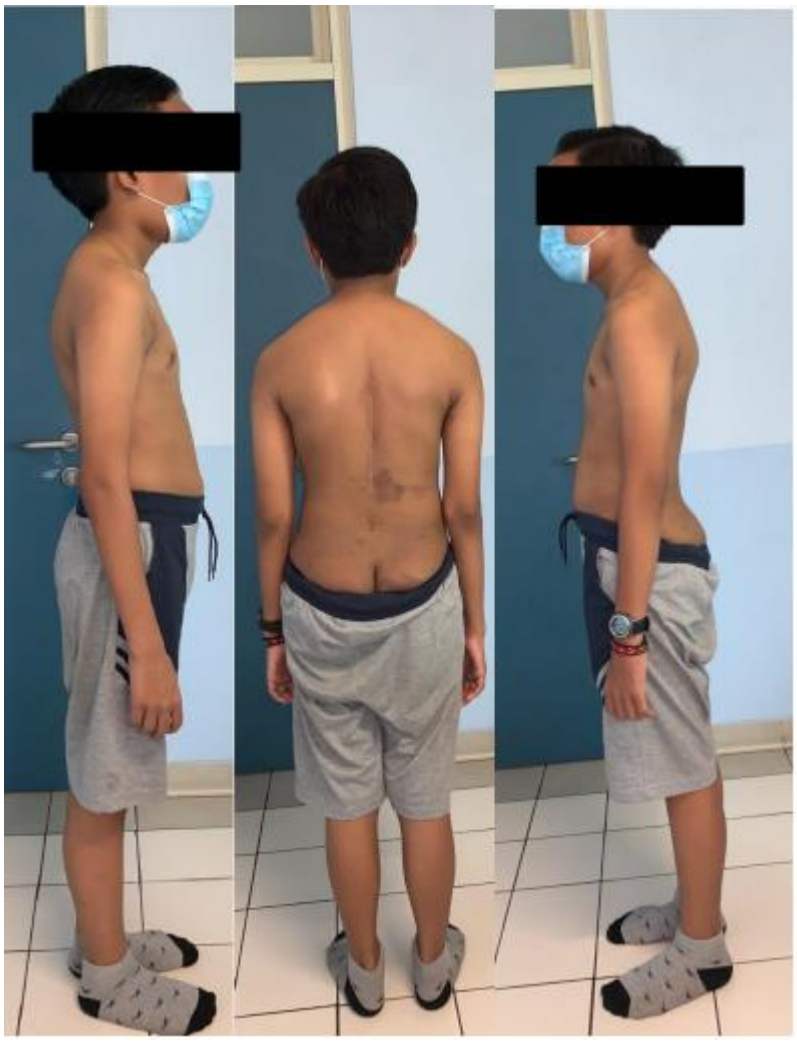


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Fig 8. Clinical Picture 9 Months Post Operation

The patient was treated with posterior laminectomy and long posterior stabilization and fusion. Physical therapy was performed. Therefore, a satisfactory results were achieved, and there were no complications. The patient was able to return to normal activity three weeks after the operation.

3. DISCUSSION

Fibrous dysplasia is a genetic non-inherited bone disease accounting for 7% of all benign bone lesions [5]. The etiology of FD is unknown but has been linked to an activating mutation of the Gs alpha gene on chromosome 20, leading to increased cyclic adenosine monophosphate availability [6]. FD is commonly seen in ribs, proximal femur, tibia, humerus, and craniofacial bones, and it presents as a single lesion 70% of the time [7]. Spinal FD accounts for only 2.5% of cases and is divided into two subtypes: monostotic and the polyostotic forms [1], [2]. While the polyostotic form is the most common presentation in the spine, the monostotic form is unusual [8]. FD can affect all compartments of the spine; the lumbar spine is by far the most affected, with an incidence of 14%, followed by the cervical spine at 7% of the time, whereas lesions of the thoracic spine remain rare [9]. This case represents a rare presentation affecting both the thoracic and lumbar spine. Polyostotic FD can be associated with café- au-lait pigmented spots, precocious puberty, and other hyperfunctioning endocrinopathies such as hyperthyroidism, growth hormone excess, Cushing syndrome, and renal phosphate wasting realizing a unique disease called McCune-Albright syndrome [2], [5]. Such conditions were successfully ruled out in our patient.

The radiographic characteristics of FD in long bones have been largely reported in the literature. However, imaging features of spinal FD are seldom. Findings on imaging can be highly suggestive of the diagnosis

when they show typical findings. Otherwise, differential diagnosis with multiple malignant and benign tumors becomes difficult. Commonly found lesions on plain radiographs and CT scans are a decrease in the body of vertebrae and osteolytic lesions that are characterized by a delicate, closely meshed, fine specular component within

The lesions impart to it a characteristic appearance that has been termed GGO. A surrounding sclerotic rim is also described. Expansible lesions characterize spinal FD but concern the adjacent intervertebral disc. In almost all cases, involvement of the pedicles has been associated with the presence of vertebral body lesions. The posterior elements of the vertebra can also be affected uncommonly. MRI findings are non-specific, showing various patterns on T1 and T2 weighted images depending on the number of collagen cells, cystic lesions, and hemorrhage forming the trabecular bone matrix. The peripheral sclerotic border of FD lesions is seen as a black ring on T1, and T2 weighted images called a rind. After gadolinium administration, the degree of enhancement is variable. [5], [7], [10] as it was shown cased in our patient. In cases of FD with lytic lesions, the differential diagnosis based on radiology should include haemangioma, giant cell tumor, and aneurysmal bone cyst. In cases of FD with blastic lesions, it should include Paget's disease and osteoblastoma. However, osteolytic metastasis rarely presents as expansible vertebral lesions [1].

Diagnosing spinal FD in adults is difficult because cases involving only the spine are rare. Radiological presentations can be suggestive but sometimes atypical in adults. The accuracy of CT-guided percutaneous biopsy for diagnosis is up to 90% depending on the lesion's components, the specimen's size, and the equipment used, whereas some scholars prefer open biopsy [3]. Typical pathological findings show irregularly structured trabecular bone without osteoblastic borders surrounded by a fibrous stroma [6]. Differential diagnosis with other lytic lesions relies mainly on various arguments, including clinical presentation, laboratory results, and radiological findings. In this patient, the radiological aspect was very suggestive of FD. Normal calcium, phosphate, alkaline phosphatase, and parathormone allowed ruling out brown tumors, and bone biopsy ruled out malignancies.

The presence of the symptoms determined the treatment of fibrous dysplasia. In the present case, the spinal cord was compressed and stretched by the kyphotic deformity. Surgery is indicated for a patient with persistent pain, neurologic impairment, vertebral collapse, instability, and/or cord compression [11]. We performed a surgical treatment regarding this condition to achieve the following goals: decompression of the spinal cord, maintenance of spinal stability, eradication of symptomatic lesions, and prevention of further pathological fractures. In our case, the patient underwent an emergency surgical treatment by

Performing T5-L5 stabilization, decompression, and fusion because of visual analog scale pain was 8 of 10. Three weeks after surgery, the patient could see activity without any complaint or limitation, with a visual analog score of 1-2. One year after the surgery, the patient came to the outpatient department without back pain, weakness, and neurological deficit.

4. CONCLUSIONS

This case of fibrous dysplasia simultaneously involves the thoracic and lumbar spine, a rare case presentation. The extreme rarity of the disease and its presentation imposes rigorous investigations to rule out malignancies. Surgical treatment was indicated for severe back pain with posterior stabilization and decompression. The surgical treatment gave a good functional outcome and improved the visual analog score.

ACKNOWLEDGEMENT
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