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Fibrodysplasia Ossificans progressive (FOP): A case report

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Abstract:

Fibrodysplasia ossificans progressiva (FOP) is a very rare, and disabling genetic disease characterized by the development of heterotopic ossification and big toe deformities. The main target is the axial musculature, but ectopic bone formation occurs in the ligaments, the fascia, the tendons and the joint capsules. Small soft tissue traumas and intramuscular injections can lead to flare ups. Diagnosis mainly depends on clinical examination and radiological findings. There is no treatment for FOP, but preventive and conservative measures are important to enhance life quality of patient and prevent flare ups. We present an 8-year-old male patient who has osseous lumps and limb deformities and restricted movement since the age of 4 years. Diagnosis was based on clinical examination and radiological findings. The patient was the first case of FOP to be diagnosed in our unit.

Eight years old boy presented with history of multiple lumps then progressive movement disabilities and deformities affecting limbs, neck and trunk with inability to open his mouth started from four years old age, otherwise since birth till the age of four years the child was normal. The caregivers had medical consultation since then with no improvement. No history of previous operations or similar cases in the family. The patient has one older sister with no congenital medical problems. No consanguinity between parents and no specific medications are taken by mother during pregnancy.

On examination, the patient was thin and looks pale, he was able to stand supported but cannot take upright position even with assistance. The child was mentally sound but cannot speak well due to difficulty in mouth opening. There was kyphoscoliosis the back, stiffness and torticollis of neck, and very limited rotation, forward bending and lateral bending of cervical spine. There was a hard painless mass felt on the back of the neck. The temperomandibular joint was little swollen and stiff with difficulty to open the mouth.

The examination of upper limbs showed bilateral severe limitation of range of motion (ROM) of both shoulders and bilateral adducted arms, right elbow with full normal ROM and normal right forearm and hand, while left forearm was pronated and flexion deformity of wrist. There was palpable painless hard mass felt on the lower left axilla extending from medial aspect of left arm to the chest wall.

The lower limbs examination revealed small hard lump felt on the inner proximal right tibia referring to an exostosis and calcified tendon origin on radiography, there was bilateral hallux valgus with bilateral short first ray (big toe microdactyly), and the left hip and knee are in fixed flexion of about 40 degrees. Irregular painless bony solid lump about 10 cm diameter in the upper medial thigh was present. We noticed that the severity of findings is more on the left side which is more affected than right side.

All laboratory findings (Complete blood picture, erythrocytic sedimentation rate, C reactive protein, liver and kidney function tests, serum electrolytes) were within average normal ranges and that was quite interesting. The Roentgenographic studies showed ankylosis of temporomandibular joints. Sheet extra-skeletal ossifications extending on back of the neck to the ligamentum nuchae and on both left and right axilla from medial aspect of humerus to chest wall (Figure 6). Flexion contracture of left elbow and left knee, kyphoscoliosis of the cervical and thoracolumbar spines.

There are no definite guidelines for treatment of FOP till now .Attempts to remove this heterotopic bone usually lead to episodes of explosive new bone formation. Physical therapy, intramuscular injections, biopsies from lesions, nerve blocks and orotracheal intubations are contraindicated as stretching of the soft tissues around a joint and muscle trauma can lead to a painful flare-up. Short course of steroids, analgesics and bisphosphonates helps to relieve symptoms of acute pain. Preventive measures are useful to prevent flare ups and devastating course of disease as regular follow-up, avoidance of trauma, avoidance of chest infections and regular audiometric evaluation. The most promising approaches currently under investigations are angiogenesis inhibitors as thalidomide, mast cell inhibitors as montelukast, immunosuppressive drugs as Rapamycin and BMP4 inhibition (experimental treatments).