

FIBRODYSPLASIA OSSIFICANS PROGRESSIVA

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Fibrodysplasia ossificans progressiva (FOP) is a rare disorder. The striking features of the disorder are the replacement of muscles, tendons, and aponeuroses by masses of bone, and the presence of certain skeletal abnormalities. In this report, we describe a woman who has literally grown up in bed since the age of three years.

Case Report

A 38-year-old female, product of a non-consanguineous marriage, developed progressive weakness, generalized pains, and stiffness of muscles around the third year of her life. This steadily worsened and she became bedridden within a year, and has grown up in bed for the last three-and-a-half decades. She had normal pubertal development and was menstruating regularly. During this period, she had multiple admissions in various hospitals, with excision of hard masses from her legs performed on many occasions.

Clinical examination revealed a young female with pulse rate of 80 beats per minute and BP of 120/70 mm Hg. She was 140 cm tall, weighed 39 kg, and had mild hirsutism. She could not sit up in bed and moved like a log of wood with help. She had a short neck, thick broad hands, small feet with bilateral hallux valgus, and short digits. She had multiple scars (from previous surgeries) and multiple areas of induration. She also had flexion contractures of both elbows, and hard bony swellings in multiple places. Examination of chest, cardiovascular system, abdomen and nervous system was normal. Tuning fork tests revealed a normal hearing pattern. Investigations revealed mild hypochromic microcytic anemia, but normal kidney and liver functions. Her serum calcium, phosphorus, alkaline phosphatase and albumin were normal on three occasions. Chest x-ray, 12-lead electrocardiogram, and ultrasonography of abdomen were normal. Radiological screening revealed diffuse muscular calcification in pelvis (Figure 1), thighs, and legs and feet (Figure 2). Histopathology of the muscle biopsy specimen from one



FIGURE 1. X-ray of pelvis showing diffuse muscular calcification.

of the hard masses revealed features consistent with fibrodysplasia ossificans.

Discussion

Gay Patin in 1692 described the first case of FOP as a woman who turned into a log of wood.¹ This disorder is characterized by formation of bone tissue around the muscles, and specific skeletal abnormalities with or without the presence of some other somatic abnormalities.

It is a disorder which generally occurs in the first three decades of life, and a majority of patients have onset of symptoms by the age of four years, but there can be a delay of many months before the diagnosis is made.² The disease predominantly affects boys. There is some evidence that the disorder is transmitted as a dominant trait.³ The affected person becomes stiff as wood, and has to be supported in order to turn from supine to standing position.² New bone deposits are laid in connective tissue between the muscle fibers, with characteristic sparing of the muscles of facial expression, the diaphragm, laryngeal muscles, and tongue. Ossification is often precipitated by minor trauma, including intramuscular injection or a biopsy procedure.

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FIGURE 2. X-ray of foot showing sheets of muscular calcification.

The swelling at the traumatic site is gradually replaced by bone.⁴

Associated skeletal abnormalities include defects of the great toe (hallux valgus and monophalangeal great toe), short first metacarpal bone, microdactyly, malformation of the little finger, reduction defects of all limbs, and abnormalities of the cervical spine.⁵ Abnormalities of the cervical spine are usually noted in childhood. These include fusion of cervical vertebrae and abnormal vertebrae with small bodies and enlarged pedicles. All these abnormalities may or may not be associated with cervical calcification, but neck movements are severely affected.⁵

Certain systemic anomalies have been noted to be associated, though less frequently. These are maldevelopment of central nervous and cardiovascular systems, sexual infantilism, hypospadias, ectopic testes and the thickening of femoral neck. Lung function tests reveal a restrictive pattern.⁶ Electrocardiography may reveal right

bundle branch block, left axis deviation with sinus tachycardia, ST segment changes, and supraventricular tachycardia in a few patients.⁷ Deafness has been reported, which may have important negative implications in the total management of these patients.²

A number of treatment modalities have been tried from time to time in the past. These include surgical removal of the ectopic bone, low calcium diet, decreased vitamin D intake, chelating agents, calcitonin, steroids and bisphosphonates. Etidronate disodium has been claimed to reduce the postoperative recurrence of heterotropic ossification.⁸ Treatment with all these modalities has been found to be disappointing.⁴ Death usually occurs secondary to a respiratory problem between the second and fourth decades, however, survival up to the seventh decade has been reported.⁸

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