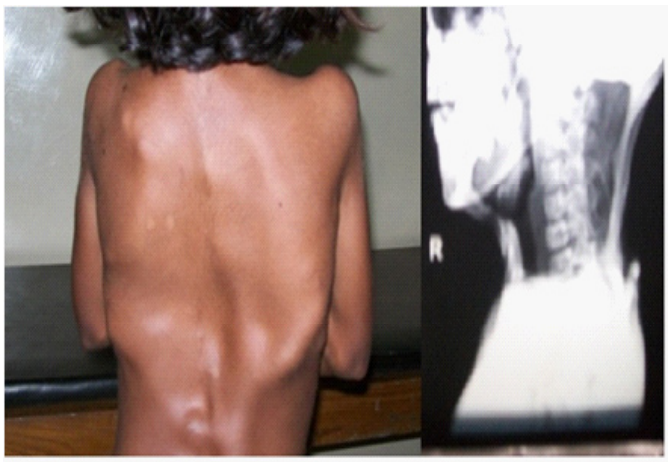
SPOT DIAGNOSIS (IMAGE GALLERY)



MUTLITPLE SOFT TISSUE CALCIFICATIONS WITH TOE DEFORMITIES

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A ten years old female child, born of consanguineous marriage, presented with bilateral toes deformities since birth and multiple bony swellings over neck and trunk increasing progressively in number and size for three years.

What is the diagnosis?

Fibrodysplasia ossificans progressiva (FOP) is a rare disabling hereditary disorder of connective tissues characterized by symmetric congenital anomalies of the great toes and thumbs with progressive post-natal heterotopic ossification of soft tissues including those related to the striated muscles, leading to permanent disabilities. (1) It affects men and women equally, with a current worldwide prevalence of approximately 1 case in 2 million individuals. (2) Calcification of soft tissues starts during the first decade of life at 5 years on average. (3) Episodes of painful soft tissue swellings occur which are often precipitated by soft tissue injury, intramuscular injections, viral infection, muscular stretching, falls or fatigue. These flare-ups transform skeletal muscles, tendons, ligaments, fascia, and aponeuroses into heterotopic bone, rendering movement impossible. (3) Although most cases of FOP are sporadic (non-inherited mutations), a small number of inherited FOP cases show germline transmission in an autosomal dominant pattern. Heterozygous activating mutations in activin receptor IAperctivin-like kinase-2 (ACVRI, ALK2), a bone morphogenetic protein (BMP) type I receptor, exist in all sporadic and familial cases of FOP. (1,2) Differential diagnosis includes progressive osseous heteroplasia, osteosarcoma, lymphedema, soft tissue sarcoma, desmoid tumors, aggressive juvenile fibromatosis, and non-hereditary (acquired) heterotopic ossification. Skeletal survey is required to find out extent of involvement. (4) Current management is focused on early diagnosis, assiduous avoidance of injury or iatrogenic harm, symptomatic amelioration of painful flare-ups, and optimization of residual function. Short course steroid is helpful during acute exacerbations. Treatment with bisphosphonate and isotretinoin help to some extent but disease is slowly progressive and most of the patients are bed ridden by 30 years. (5,6) Death occurs by second to fourth decade usually due to respiratory failure. (3)

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