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Clinical Image

CLINICAL IMAGE

A 'Y-shaped' soft tissue ossification in fibrodysplasia ossificans progressiva

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CASE REPORT

A 16-year-old girl developed multiple painful and tender soft tissue swellings over her neck, trunk and extremities since 2 years of age. These swellings appeared spontaneously or after trivial injury and either used to resolve spontaneously or they ossified into mature bone. These flares of heterotopic ossification resulted in gradual restriction of movements of neck, trunk, shoulder, elbow, hip and knee joints. There was associated congenital hallux valgus bilaterally (Fig. 1a). There was no history of similar illness in her family. The radiographs of the girl showed certain characteristic features of fibrodysplasia ossificans progressiva (FOP). The cervical skiagram showed characteristic anomalies of the cervical spine in form of large posterior elements and fusion of the facet joints between C2 and C7 (Fig. 1b) [1]. A CT scan of dorso-lumbar spine showed a peculiar

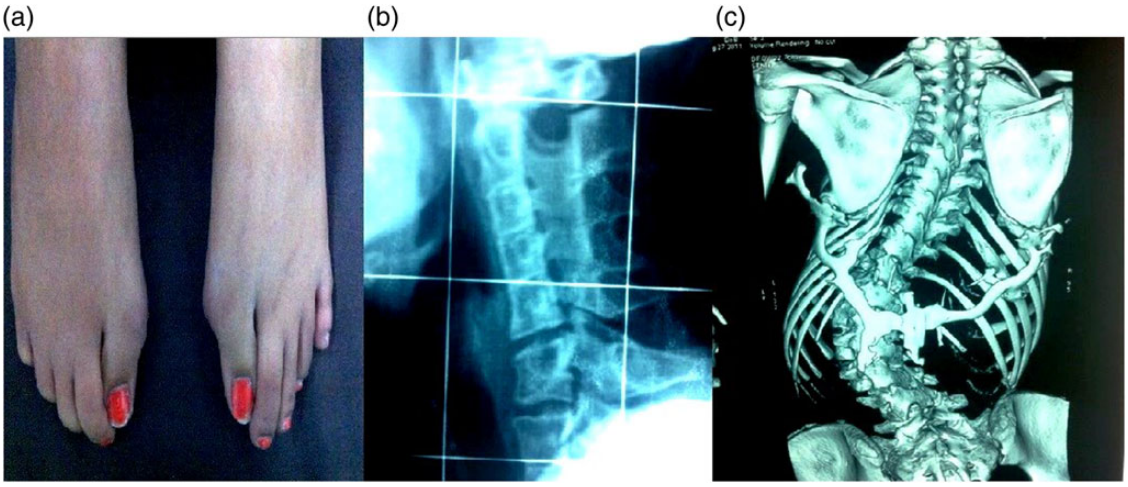

Figure 1: (**a**) Characteristic malformed great toe and hallux valgus. (**b**) The cervical skiagram shows large posterior elements and fusion of the facet joints between C2 and C7. (**c**) A CT scan of dorso-lumbar spine shows Y-shaped soft tissue ossification extending from the inferior angle of bilateral scapula to the midline soft tissue of posterior back (lattisimus dorsi muscles and thoracolumbar fascia), dorso-lumbar scoliosis with convexity to left, squaring of thoracic and vertebral bodies and fusion of posterior elements of thoracic and lumbar vertebrae.

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'Y-shaped' soft tissue ossification extending from the inferior angle of bilateral scapulae to the midline soft tissue of posterior back (lattisimus dorsi muscles and thoracolumbar fascia). It also demonstrated dorso-lumbar scoliosis and fusion of posterior elements of thoracic and lumbar vertebrae (Fig. 1c).

DISCUSSION

FOP is a rare genetic disorder of connective tissue characterized by congenital malformed great toes and progressive heterotopic ossification leading to severe disability. Classic FOP is caused by a recurrent activating mutation (617G>A; R206H) in the gene ACVR1/ALK2 encoding Activin A receptor type I/Activin-like kinase 2, a bone morphogenetic protein type I receptor [2]. This genetic defect which leads to the formation of a heterotopic skeleton involves normal skeletal morphogenesis at heterotopic sites [3]. During the first decade of life, sporadic episodes of painful soft tissue swellings (flare-ups) occur which are often precipitated by trivial injury. These flare-ups transform skeletal muscles, tendons, ligaments, fascia and aponeuroses into heterotopic bone causing restriction of movements of various joints. The flares may partially respond to a short course of steroids, but overall prognosis is reduced with median lifespan of 40 years. Most patients die of complications of thoracic insufficiency syndrome [4].

CONFLICT OF INTEREST STATEMENT

None declared.

REFERENCES

1. Schaffer AA, Kaplan FS, Tracy MR, O'Brien ML, Dormans JP, Shore EM, *et al*. Developmental anomalies of the cervical spine in patients with fibrodysplasia ossificans progressiva are distinctly different from those in patients with Klippel Feil syndrome: clues from the BMP signaling pathway. *Spine (Phila Pa 1976)* 2005;**30**:1379–85.
2. Kaplan FS, Xu M, Seemann P, Connor JM, Glaser DL, Carroll L, *et al*. Classic and atypical fibrodysplasia ossificans progressiva (FOP) phenotypes are caused by mutations in the bone morphogenetic protein (BMP) type I receptor ACVR1. *Hum Mutat* 2009;**30**:379–90.
3. Kaplan FS, Strear CM, Zasloff MA. Radiographic and scintigraphic features of modeling and remodeling in the heterotopic skeleton of patients who have fibrodysplasia ossificans progressiva. *Clin Orthop Relat Res* 1994;**304**:238–47.
4. Pignolo RJ, Shore EM, Kaplan FS. Fibrodysplasia ossificans progressiva: clinical and genetic aspects. *Orphanet J Rare Dis* 2011;**6**:80.