"Bone Block" and Congenital Spine Deformity

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What do you see in these images?

a. Juvenile rheumatoid arthritis with idiopathic scoliosis
b. Fibrodysplasia ossificans progressiva
c. Klippel-Feil syndrome with congenital scoliosis
d. Ankylosing spondylitis
e. Diffuse idiopathic skeletal hyperostosis with hemivertebrae

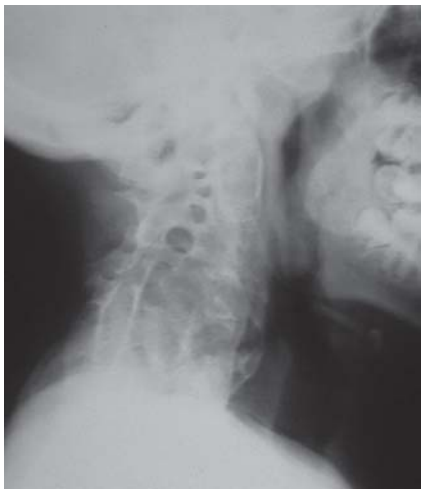

Fig. 1. Lateral plain radiograph illustrating complete fusion of the cervical spine.

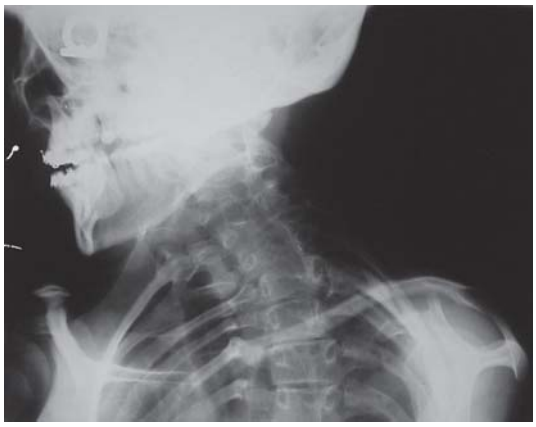

Fig. 2. Anteroposterior plain radiograph demonstrating deformity of the spine.

A 14-year-old female who presented with a limited range of neck motion with various congenital vertebral anomalies that included improper segmentation or extensive "fusion" of the cervical spine, hemivertebrae, and a right rigid convex thoracolumbar scoliosis (Figs. 1 and 2). She was diagnosed with Klippel-Feil syndrome (KFS) with congenital scoliosis. A hemivertebrectomy was eventually performed with spinal fusion to correct her spinal curvature.

KFS is a relatively uncommon congenital condition characterised primarily as congenital fusion of 2 or more cervical vertebrae with or without additional spinal manifestations, such as scoliosis, diastematomyelia or spinal dysraphism. The true aetiology of KFS is speculative and its phenotypic expression varies between individuals. The majority of young individuals with KFS are asymptomatic, however, various fused cervical patters present an increased risk of developing symptoms and potential spinal cord injury caused by even minor trauma or, as is in some cases, iatrogenic-related. KFS is also associated with various extraskeletal malformations, such as cardiac and genitourinary abnormalities, renal agenesis, cleft palate, hearing deficiencies, upper and lower extremity malformations and Sprengel's deformity. Therefore, thorough clinical and radiographic examination of the spine and associated organ systems is warranted to rule out similar conditions to KFS, identify patients at high risk of spinal cord injury, and to help determine appropriate management options.

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