SPOT DIAGNOSIS (IMAGE GALLERY)

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This 13 years child presented with progressive deformity over occipital region since birth. On examination she had short neck, restriction of neck movements, low hair line and bony prominence over occipital region. There were no focal neurological deficits or other abnormalities. X-ray cervical spine, cranio-vertebral junction and open mouth view are depicted in the figure.
What is the diagnosis?

X-rays show extensive fusion of bodies and deformity of cervical vertebrae and hypoplastic dense suggestive of extensive Type 1 Klippel-Feil anomaly. Klippel-Feil syndrome (KFS) is a rare and complex disorder that is mainly characterized by congenital fusion of the cervical vertebrae with a short neck, limitation of the movement of the head or neck and a low posterior hairline. This syndrome is a result of failure of the normal segmentation of the cervical somites during the 3rd to 8th weeks of gestation. Its incidence is estimated as 1:40,000-42,000. Type 1 is characterized by cervical spine fusion in which elements of many vertebrae are incorporated into a single block. Type 2 has cervical spine fusion in which there is failure of complete segmentation at only one or two cervical levels and may include an occipito-atlantal fusion. Type 3 has Type 1 or type 2 fusion with co-existing segmentation errors in the lower dorsal or lumbar spine

E-published: February 2009