World Academy of Science, Engineering and Technology
International Journal of Bioengineering and Life Sciences
Vol:9, No:02, 2015

Klippel Feil Syndrome: A Case Report and Review of Literature

Authors : Rim Frikha, Nouha Bouayed Abdelmoula, Afifa Sellami, Salima Daoud, Tarek Rebai

Abstract : Klippel-Feil Syndrome (KFS) is characterized by congenital vertebral fusion of the cervical spine resulting from faulty segmentation along the embryo's developing axis. A wide spectrum of associated anomalies may be present. This heterogeneity has complicated elucidation of the genetic etiology and management of the syndrome. We report a case of an isolated Klippel-Feil Syndrome with C5-C6 fusion on the cervical spine. It's the rarest form of congenital fused cervical vertebrae which is predisposed to the risk of spinal cord injury and neurologic problems. The aim of this paper was to review clinical heterogeneity; radiographic abnormalities and genetic etiology in Klippel-Feil Syndrome. We insist in comprehensive evaluation and delineation of diagnostic and prognostic classes.

Keywords : Klippel–Feil anomaly, genetic, clinical heterogeneity, radiographic abnormalities
Conference Title : ICHG 2015 : International Conference on Human Genetics
Conference Location : London, United Kingdom
Conference Dates : February 16-17, 2015