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A Young Girl Presented with Short Stature with Sprengel Deformity: A Case Report

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Abstract

AKlippel-Fiel syndrome (KFS) is a rare congenital disorder characterized by the presence of a triad of short neck, limited neck mobility, and low posterior hairline. A 5-year-old girl of consanguineous parents had limited neck movement and poor growth for three years. Her height was 87cm (SDS -4.6), and her weight was 11kg (SDS-2.8), indicating significant growth retardation. Intelligence was normal for age, but a clinical examination revealed a short neck, limited neck movement, and Sprengel deformity. Although her thyroid function was normal, the growth hormone stimulation test (using clonidine) was borderline low. By karyotyping, a normal female pattern was identified (46, XX). While the echocardiogram was normal, the ultrasound revealed that the left kidney was absent, along with an infantile uterus. An X-ray of the cervical spine revealed a Sprengel deformity and cervical ribs. Her radiological bone age was delayed. The diagnosis of Klippel-Feil Syndrome was made by using clinical presentation, physical examination, and laboratory investigations. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S59]*

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