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A Newborn with Klippel-Feil Syndrome in Association of Annular Pancreas and Diastematomyelia

Anüler Pankreas ve Diastematomiyeli Saptanan Tip III Klippel Feil Sendromlu Bir Yenidoğan Olgu Sunumu

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ABSTRACT

Klippel–Feil syndrome (KFS) is a complex syndrome associated with osseous and visceral anomalies that include the classical clinical triad of short neck, limitation of head and neck movements and low posterior hairline. Multiple congenital anomalies have been associated with this disease. We reported the first case of KFS presented with the symptoms of duodenal obstruction and annular pancreas detected intraoperatively.

Key Words: Annular, Diastematomyelia, Klippel-Feil syndrome, Newborn, Pancreas

ÖZET

Klippel-Feil Sendromu; kısa boyun, baş ve boyunda hareket kısıtlılığı, düşük saç çizgisi klinik triadı ile tanımlanan, kemik ve organ anomalilerinin eşlik ettiği kompleks bir sendromdur. Multiple konjenital anomaliler bu sendromla birlikte görülebilir. Duodenal obstruksiyon ile başvuran klippel-feil sendromu tanımlanan bir yenidoğanda, bilindiği kadarıyla bu sendrom ile ilk kez birlikteliği tanımlanmış anüler pankreas saptanan bir olgu sunulmuştur.

Anahtar Sözcükler: Anüler, Diastematomiyeli, Klippel-Feil sendromu, Yenidoğan, Pankreas

INTRODUCTION

Klippel-Feil syndrome (KFS) was first described in 1912 and is characterized by the congenital fusion of two or more cervical vertebrae. The associated clinical findings include short neck, low posterior hairline, and severe restriction of cervical motion; the classical clinical triad that is seen in less than 50% of the patients. The disease is relatively rare, with an approximate incidence of one in 42,000 births, and 65% of those affected are female (1-3). KFS is classified into three types according to the degree of involvement.

Type I is the classic form with extensive cervical and upper thoracic vertebral fusion. Type II represents fusion at 1st or 2nd cervical levels. Type III consists of cervical fusion associated with lower thoracic or lumbar vertebral fusion (4).

Patients with KFS may have other features in addition to their vertebral abnormalities. Hearing difficulties, genitourinary abnormalities such as malformed kidneys, neural tube defects, cleft palate or heart abnormalities have also been reported. Affected individuals may have underdeveloped shoulder blades that sit abnormally high on the back, a condition called 'Sprengel deformity'. This heterogeneity requires comprehensive evaluation of all patients and treatment regimes that can vary from modification of activities to extensive spinal surgeries (2). In this report, we described a newborn with KFS in association of annular pancreas and duedonal obstruction that wasn't reported previously.

CASE REPORT

A 2 days of old female neonate born to a 23 year old unconsanquineous mother second gravida was admitted to our clinic with no meconium passage within the first 24 hour and recurrent bilious vomiting after breastfeeding. Her birth weight was 2200 g, and gestational age was 34 weeks. The physical examination of the patient showed short neck, limitation of head movement, low posterior hairline and a hairy patch at the base of lumbar spine (Figure 1A, B). KFS was thought with the clinical findings of our patient appropriate with classic triad of KFS fenotype. Her laboratory analysis revealed hypokalemic hypochloremic

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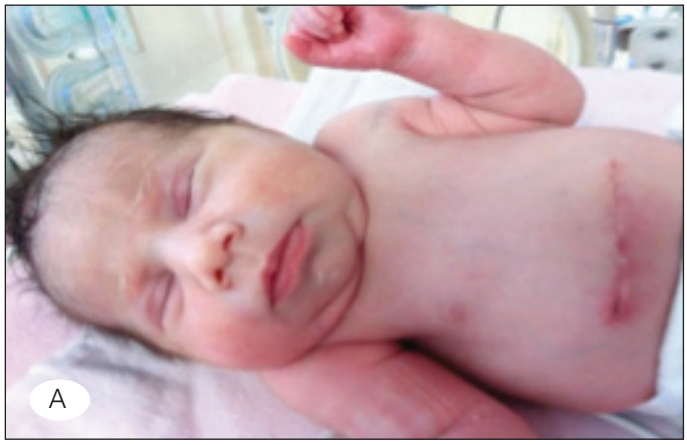
metabolic alkalosis and acute prerenal failure. There was a "double-bubble sign" on the plain abdominal radiograph. Abdominal ultrasound revealed no abnormality in the liver, gall bladder, kidney, spleen and other intraabdominal structures. An osephago-gastro-duodenoscopy has been made and there was not any contrast passage beyond the second segment of the duodenum. On the spinal magnetic resonance imaging (MRI), there was vertebral fusion at C5-T1 and T12-L1 regions (Figure 2). MRI also showed diastematomyelia at T12-L2 vertebrae and scoliosis (Figure 3). The cranial MRI was normal. Together with the considereration of radiologic and clinical findings, our case was classified as KPS type III.



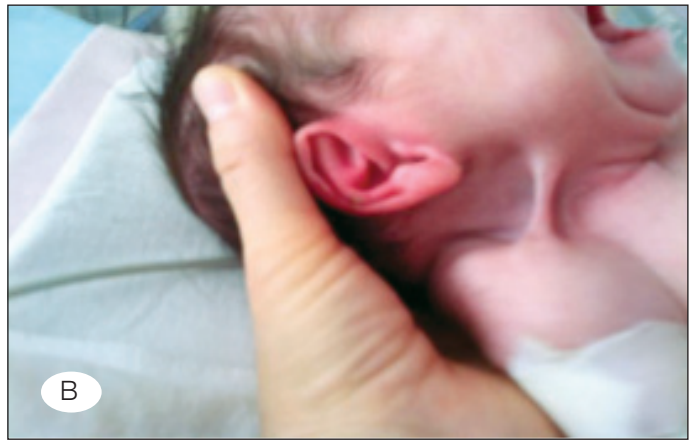


Figure 1A, B: The appearence of the face and neck of our patient.

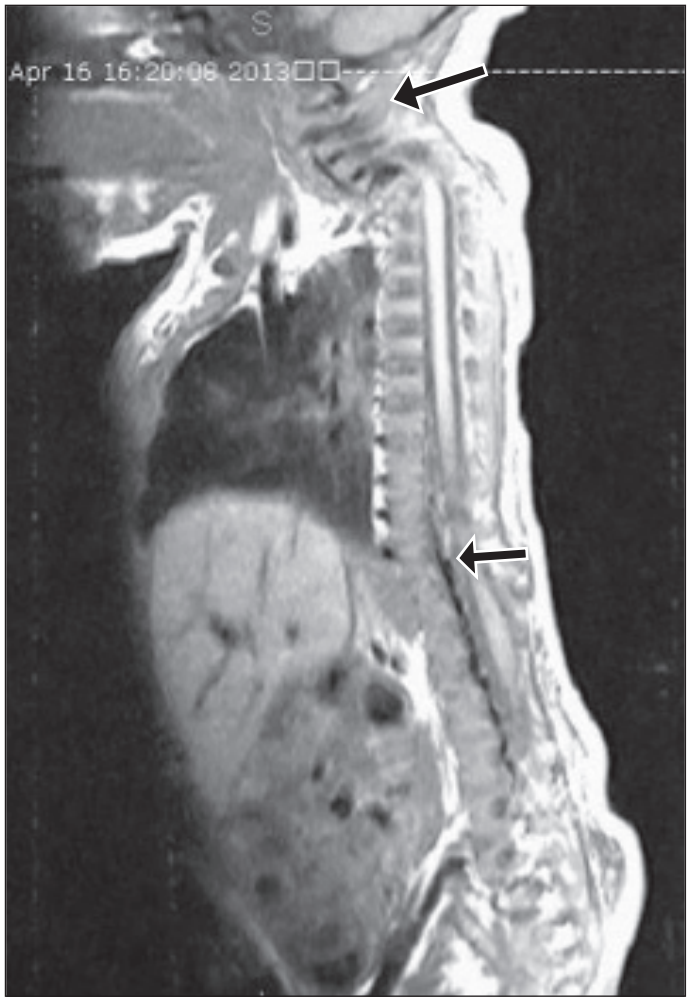


Figure 2: The appearence of magnetic resonans imaging showing cervical and thoracolumbar vertebral fusion.

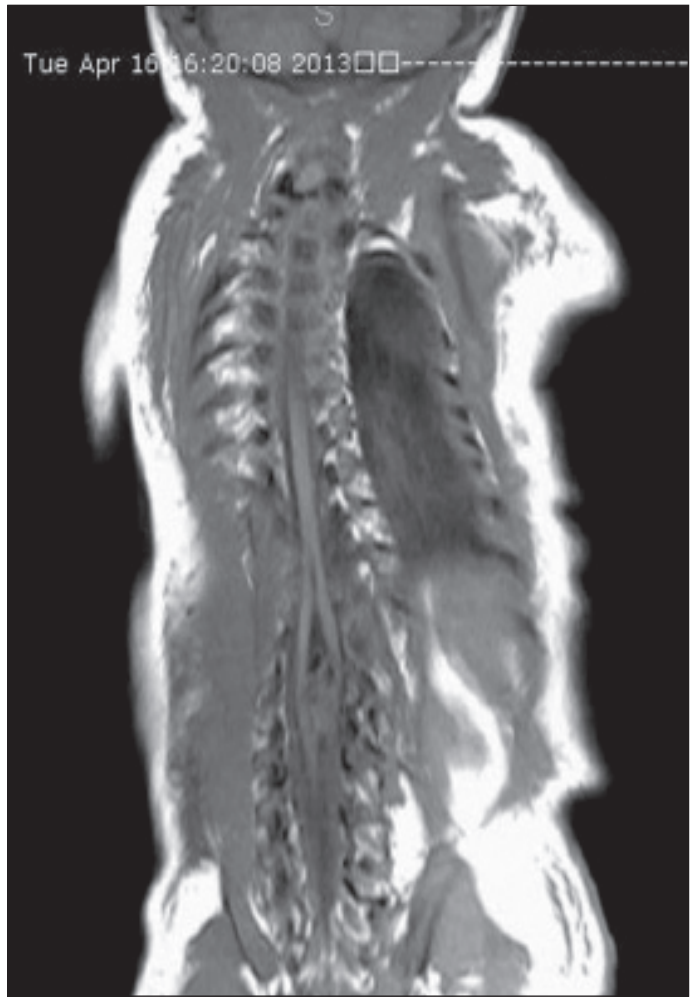


Figure 3: The appearence of magnetic resonans imaging showing diastometamyelia and scoliosis.

The patient was followed in the neonatal intensive care unit. After the stabilization of patient, laparotomy was performed at 7th day postnatally. An annular pancreas was observed intraoperatively. Following the procedure for releasing of the pancreas around duedonum, enteral nutrition was started on the third post-operative day and no intolerance to feeding was observed. The neurosurgery and orthopedic departments decided an operation 3 months later. We discharged the patient on breastfeeding with a multidisciplinary follow-up programme including neonatology, pediatric surgery, orthopedics, neurosurgery, and physical therapy departments.

DISCUSSION

KFS is a rare disease associated with numerous abnormalities of the organ systems. Patients affected with the KFS have to undergo a complex examination of whole body in order to treat any life threatening problems (1-4). Our case had gastrointestinal abnormality with the initial symptoms of duedonal obstruction except classical triad of the KFS and diagnosed annular pancreas intraoperatively.

Annular pancreas is a rare congenital abnormality characterized by a ring of pancreatic tissue surrounding the descending portion of the duodenum. In symptomatic neonates a plain abdominal X-ray or ultrasound will show the classic 'double bubble' sign suggesting duodenal obstruction. The definite diagnosis was possible at the time of laparotomy (3,5,6). The most common gastrointestinal abnormalities in association of KPS reported in the literature was duodenal web (7). According to us, this is the fisrt case report diagnosed annular pancreas in association of KFS.

Diastematomyelia is defined as a double cord malformation that mainly occurs in the lower thoracic and lumbar regions. The association of thoracolumbar diastematomyelia with KFS occurs in approximately 20% of patients (8,9). Our patient also had thorasic diastematomyelia. Cervical examples of diastematomyelia are very rare but often associated in cases with KFS (10,11).

The cases of KFS are sporadic; uncommon familial forms, showing autosomal dominant or autosomal recessive inheritance have also been reported. Due to there was no familial history of our case, we assumed this case was a sporadic form. Recently, growth differentiation factor 6 (GDF6) and 3 (GDF3) genes were demonstrated to be mutated in some individuals of KFS (1,12).

The follow-up of our patient after discharge is maintained in our hospital outpatient clinics of neonatology, orthopedics, neurosurgery and physical therapy departments. Genetic testing for the possible mutations for KFS was planned. This is the first case of KFS together with annular pancreas reported in the literature. More case series are needed to support if it is a gastrointestinal abnormality associated with this syndrome or it is just an incidental togetherness.

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