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Unilateral renal agenesis and ipsilateral absence of the vas deferens in a cryptorchid infant with Klippel-Feil syndrome

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ABSTRACT

We report an exceedingly rare case of Klippel-Feil syndrome (KFS), compounded by ipsilateral absence of the vas deferens, renal agenesis, and diaphragmatic hernia. Unilateral absence of the vas deferens was found incidentally during orchidopexy. To the best of our knowledge, no case of unilateral absence of the kidney and vas deferens has been reported in children with KFS.

1. Introduction

Klippel-Feil syndrome (KFS), first described by Klippel and Feil in 1912,[1] is a rare congenital cervical vertebral defect, characterized by the triad of a short neck, low posterior hairline, and restricted cervical range of motion. Although KFS is known to manifest a broad spectrum of comorbidities spanning musculoskeletal, central nervous, cardiovascular, ophthalmological, and urogenital systems, reports of unilateral renal agenesis and ipsilateral absence of the vas deferens in association with this syndrome are rare and limited only in adults. Herein, we present the first pediatric KFS case compounded by unilateral renal agenesis and ipsilateral absence of the vas deferens with cryptorchidism.

2. Case report

A 4-month-old boy with an uneventful prenatal and familial medical history was referred to our department after an undescended left testis was detected during his 4-month checkup. The left testicle was palpable in the inguinal region, with no other discernible abnormalities in the external genitalia. We planned to perform orchidopexy at 6-12 months old. Concurrently, an asymmetry in neck alignment was noted, prompting a visit to our center's orthopedic surgery department. Imaging revealed vertebral fusion and left scapula elevation (Sprengel's deformity) (Fig. 1A), and a diagnosis of KFS was made. Incidentally, imaging studies also revealed a left diaphragmatic hernia and left renal agenesis (Fig. 1B). Technetium-99m-labeled dimercaptosuccinic acid scintigraphy taken later showed no accumulation in the left kidney (Fig. 2). He underwent transabdominal diaphragmatic hernia repair in the pediatric surgery department at 5 months of age. When he turned 9 months, left orchidopexy was performed, and intraoperative findings revealed the absence of the left vas deferens and its epididymis structures (Fig. 3). At 2 years and 4 months, he underwent orthopedic left scapular descent. Each postoperative course was uneventful, with no difference in testicular size or position between the right and left testes, and right renal function was well maintained at the 2-year follow-up.

3. Discussion

KFS is a congenital cervical vertebral fusion disorder, characterized by a triad of clinical features, including a short neck, low posterior hairline, and restricted cervical range of motion. This condition arises due to abnormal segmentation of the cervical spine during fetal development. Although the presence of all three classical features is observed in <50% of cases, the presence of any of them should prompt consideration of this syndrome.[2] The estimated prevalence ranges from 1 in 40,000 to 42,000 live births, with a slightly higher occurrence (65%) reported in females.[2] KFS is classified based on variations in the morphology of the fused vertebrae, resulting in three subtypes: type I, characterized by extensive cervical and upper thoracic spine fusion; type II, involving fusion between two vertebrae; and type III, where fusion extends to the lower thoracic or lumbar spine, in addition to the cervical spine.[2] Although the causative genes for each subtype have been reported in only a few cases, it is assumed that genetic mutations that have not been clarified are involved in KFS pathogenesis. The fused vertebrae themselves often remain asymptomatic and require only observation. However, complications and comorbidities involving the

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musculoskeletal, central nervous, cardiovascular, ophthalmologic, and urogenital systems frequently manifest as primary symptoms of the disease.

The complication rate of congenital urogenital diseases in this syndrome has been reported to be 25–35%.[2] It is well-established that urinary tract malformations frequently accompany spinal malformations, and the incidence of urogenital complications is also high in this syndrome, particularly unilateral renal agenesis, which has been reported frequently. The bone and urogenital systems share a common origin in the primitive mesodermal tissue. At the end of the fourth week of fetal life, the blastemas of the cervicothoracic somite, pronephros, and mesonephros are close. Coinciding with this, ureteric buds sprout from the Wolffian ducts at approximately 5 weeks of gestation, while the notochord develops into the vertebral body. It is reasonable to infer that abnormalities during this period could contribute to musculoskeletal and urogenital malformations.[2]

Embryologically, the development of the vas deferens and kidneys is also closely intertwined. Congenital unilateral absence of the vas deferens, a malformation resulting from Wolffian duct developmental defects, has been associated with unilateral renal agenesis in 70% of cases.[3] In some cases, the contralateral vas deferens could be absent as a part of the congenital bilateral absence of the vas deferens. Although the present case is our only known pediatric case of KFS with unilateral renal agenesis and ipsilateral absence of the vas deferens, two adult cases of KFS with unilateral renal agenesis presenting azoospermia have been reported.[4] Both cases were diagnosed with obstructive azoospermia possibly caused by bilateral absence of the vas deferens. Routine abdominal ultrasound examination should be recommended for male infants with KFS, and appropriate counseling of the parents for possible infertility is required when unilateral renal agenesis is detected. As normal spermatogenesis is expected in cases with KFS, successful testicular sperm extraction and intracytoplasmic sperm injection have been reported.[4]

In this particular case, an ipsilateral diaphragmatic hernia was also observed. We could identify only two cases of diaphragmatic hernia associated with KFS,[5] and it appears to be exceedingly uncommon for this syndrome to co-occur with renal agenesis, absence of vas deferens, and diaphragmatic hernia.

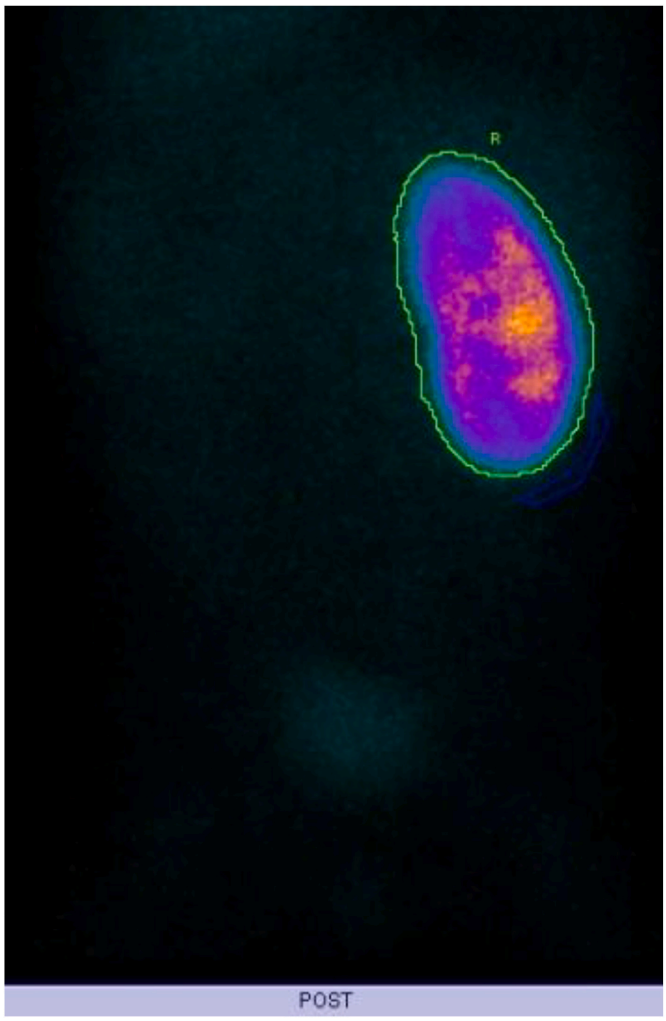

Fig. 2. Technetium-99m-labeled dimercaptosuccinic acid scintigraphy showed no accumulation in the left kidney.

4. Conclusion

We presented an exceedingly rare case of KFS, compounded by ipsilateral renal agenesis, absence of vas deferens, and diaphragmatic hernia. Since the spine and genitourinary system are closely involved in embryology, various complications must be considered in this syndrome.

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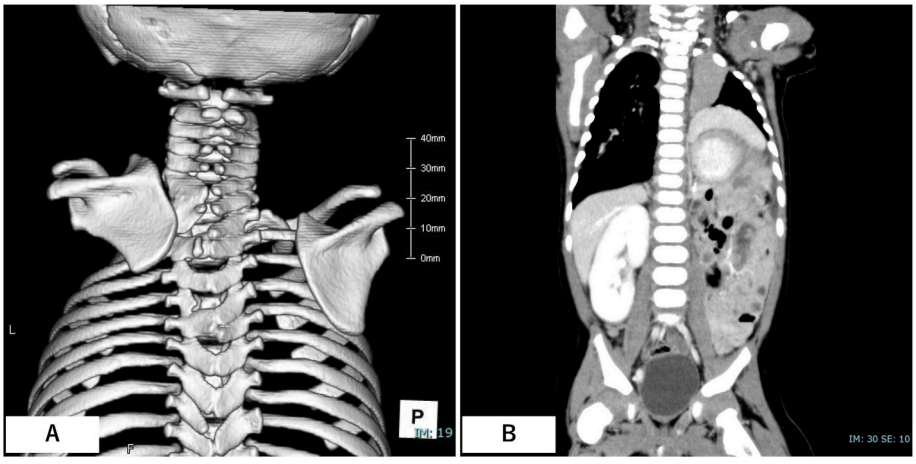

Fig. 1. Computed tomography at presentation showed an elevation of the left scapula and vertebral fusion of right C6-7 and left Th1-2 (A). A left diaphragmatic hernia and left renal agenesis were detected incidentally (B).

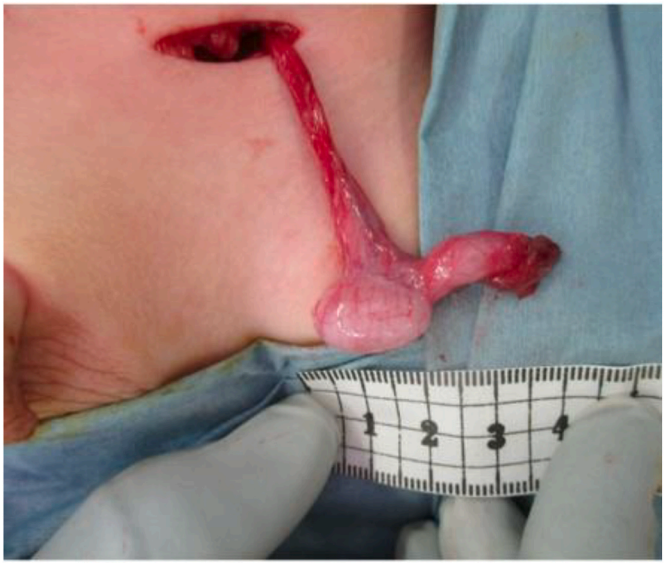

Fig. 3. Intraoperative findings showed the absence of the left vas deferens and its epididymis structures.

agencies in the public, commercial, or not-for-profit sectors.

Credit author statement

Toshimitsu Tanaka: Investigation, Data curation, Visualization, Writing- Original draft preparation. Fumi Matsumoto: Writing-Reviewing and Editing. Satoko Matsuyama: Reviewing and Editing. Futoshi Matsui: Reviewing and Editing.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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