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Staghorn Calculus in Pediatric Congenital Myotonic Dystrophy

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Introduction

Congenital myotonic dystrophy (CMD) is an inherited genetic disorder characterized by multiple organ involvement, such as cataracts, cardiac conduction defects, cognitive impairment, endocrinologic, gastrointestinal, and respiratory diseases, as well as the muscular system. Staghorn calculus, a large kidney stone that occupies the entire renal pelvis and calyces, is unusual in pediatrics. Although renal involvement is not well known in CMD, there has been a suggestion of a possible association between cystinuria (one of the causes of urinary stones) and myotonic dystrophy via dysfunction of amino acid cell transportation. We report a pediatric case accompanied by these two rare conditions.

Case report

A 13-year-old male patient was diagnosed with CMD at two months of age due to perinatal asphyxia and hypotonia. A radio-opaque signal was incidentally identified in the whole spine X-ray taken for scoliosis evaluation (Fig. 1). Further evaluation with an abdominal CT scan showed giant calculus occupying the left renal pelvis. The patient was admitted to the urology department and underwent percutaneous nephrolithotomy under general anesthesia (Fig. 2). Following surgery, he could not be weaned from the mechanical ventilator and received treatment in the intensive care unit. After removing the endotracheal tube, non-invasive ventilation was applied via an oronasal mask. The duration of non-invasive ventilation was gradually reduced, leading to complete weaning after seven days. The patient recovered without any further complications.

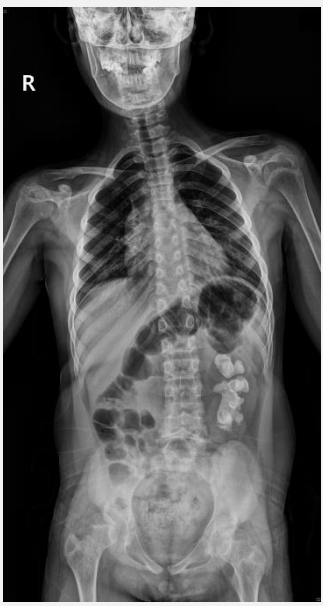

Fig. 1. Pre-operative whole spine x-ray. A huge kidney stone occupying the left renal pelvis was observed.

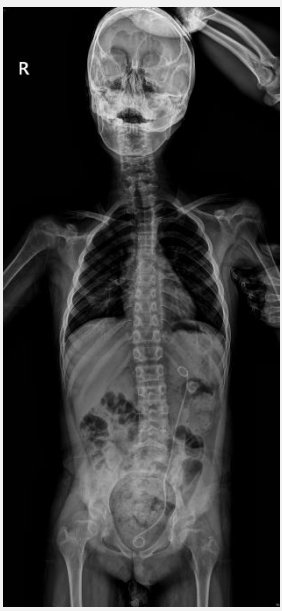

Fig 2. Post-operative whole spine x-ray. Most of the kidney stones have been removed and a ureteric stent has been placed.

Conclusion

The development of urinary stone at early age in CMD patients is not yet known well. It seems that the association between two rare conditions need to be elucidated.