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Case report

Posterior urethral valves and Down syndrome

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Abstract
The broad range of renal and urinary tract abnormalities associated with Down syndrome are not well known. We present two cases from a single institution of posterior urethral valves associated with Down syndrome. The cases illustrate the potential for delayed diagnosis and the management challenges. The literature is reviewed and a discussion of the need to screen Down syndrome children for urinary tract anomalies is presented.

Introduction

The first description of Down syndrome (DS) was in 1866 by the British physician Dr. John Langdon Down [1]. Despite this long history, the association of DS with congenital anomalies of the kidney and urinary tract (CAKUT) have received little attention. While DS is the most common chromosomal anomaly, and it is cardiac, gastrointestinal, eye, hearing and thyroid associations are well known; the associated CAKUT have received little attention in the literature [2]. In 1960, Berg et al. were the first to describe an association between DS and CAKUT [1].

This lack of clinical awareness may result in a delayed diagnosis and thus poorer outcomes. We present two cases that illustrate this problem. Additionally, a review of CAKUT's broader association with DS is presented and a consideration of whether screening of DS children for CAKUT should be the standard of care.

Case 1

A 5-year-old boy, diagnosed at 18 months with DS, presented to paediatric urology services with urinary incontinence. His incontinence had previously been ascribed by the parents to delayed toilet training due to the associated developmental delay. Physicians attending to the child had likewise not taken the symptom seriously. History additionally revealed that no antenatal sonar had been done; he had recurrent UTIs and a poor urinary stream. Abdominal examination revealed a palpable bladder. KUB sonar showed gross bilateral hydroureteronephrosis and a thick walled bladder (Fig. 1). A cystogram confirmed a diagnosis of posterior urethral valves (PUV). His creatinine was elevated at 71 μmol/L (normal < 42 μmol/L). He underwent endoscopic fulguration of his PUV and is due for a protocol relook cystoscopy at 3 months.

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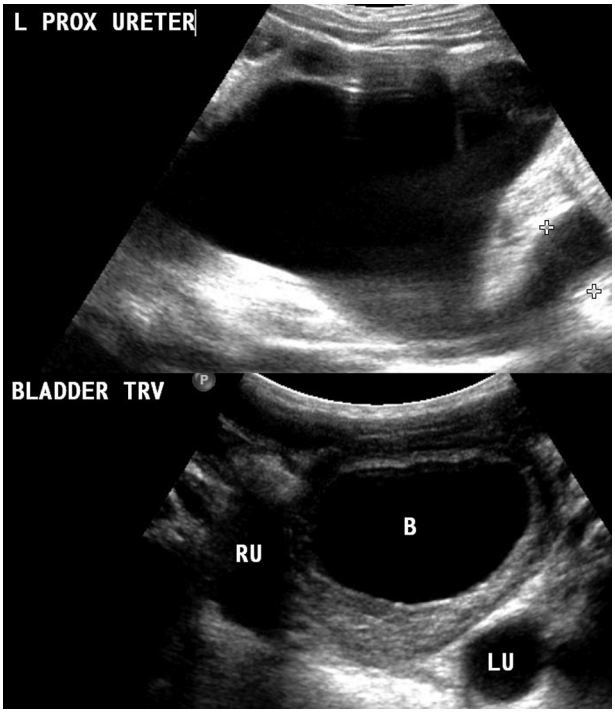

Figure 1 Gross bilateral hydroureteronephrosis with a thick walled bladder was demonstrated on KUB sonar.

Case 2

A 10-year-old boy known from birth with DS presented at 6 months of age with urosepsis. No antenatal sonar had been performed, but post-natal KUB sonar revealed bilateral hydroureteronephrosis, a thick walled bladder and a dilated posterior urethra. Cystogram confirmed PUV. His admission was complicated by a perinephric abscess which required percutaneous drainage. Additionally catheter related trauma and resultant scrotal urinary extravasation necessitated a vesicostomy rather than primary endoscopic ablation. He was then lost to follow-up and represented at age 6 when his vesicostomy was closed and the PUV was ablated.

Thereafter he developed incontinence and worsening hydronephrosis. Urodynamics assessment demonstrated a small capacity, poorly compliant bladder. He was commenced on timed voiding and anticholinergics. This has resulted in improved continence, but a renogram demonstrated new cortical defects and a reduced GFR of 68 ml/min/1.73 m^2. We have thus recently elected to attempt to institute clean intermittent catheterisation.

Comment

Kupferman reported that the prevalence of a broad range of CAKUT in DS children was 3.2%. This is roughly five times higher than in unaffected individuals [2]. Children with DS are also at increased risk of having PUV [1,2]. It has been noted that CAKUT is the 3rd most common association in DS children after cardiac and gastrointestinal anomalies [2]. Additionally, a recent review of over 100 DS children showed a 27% prevalence of lower urinary tract symptoms, with males and the young particularly affected [3].

The two cases presented here illustrate the significant potential morbidity associated with CAKUT in DS children. In addition to the potential requirement for surgery, these children may need to adhere to demanding bladder regimes including behavioural, drug and intermittent catheterisation interventions.

The delayed diagnosis in Case 1 undoubtedly contributed to the child's degree of renal impairment. Based on the available literature and the experience with these two cases, non-invasive ultrasound screening to identify CAKUT can, we feel, be justified as standard of care during the initial evaluation of DS children [1,2].

Conflict of interest

The authors assert that there is no conflict of interest.

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