

Urinary Tract Anomalies in Down Syndrome

To screen or not to screen?

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Genitourinary Anomalies

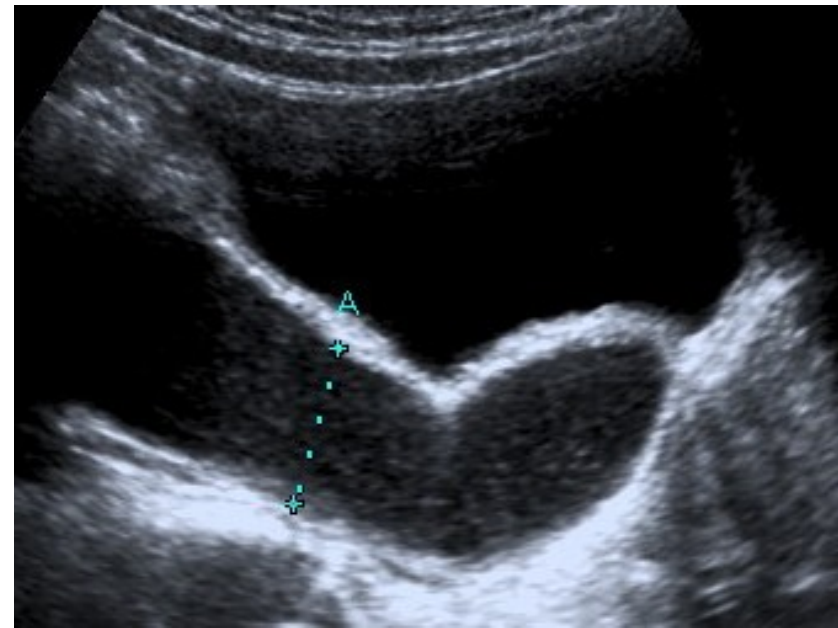
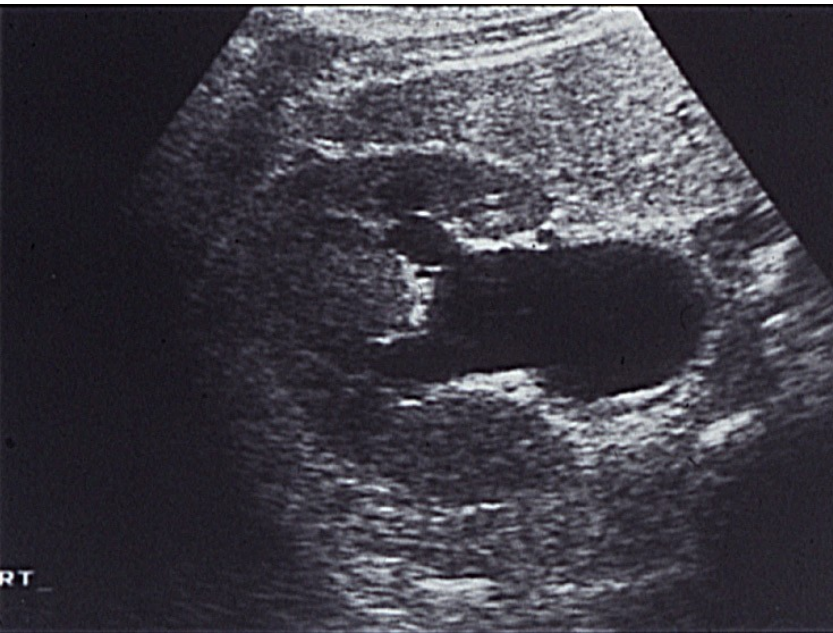
3% - 16%

- **Upper Urinary Tract – most common**

Renal agenesis/hypoplasia/cysts

Horseshoe kidney/Renal duplication

Pelviureteric/Vesico-ureteric junction obstruction/VUR



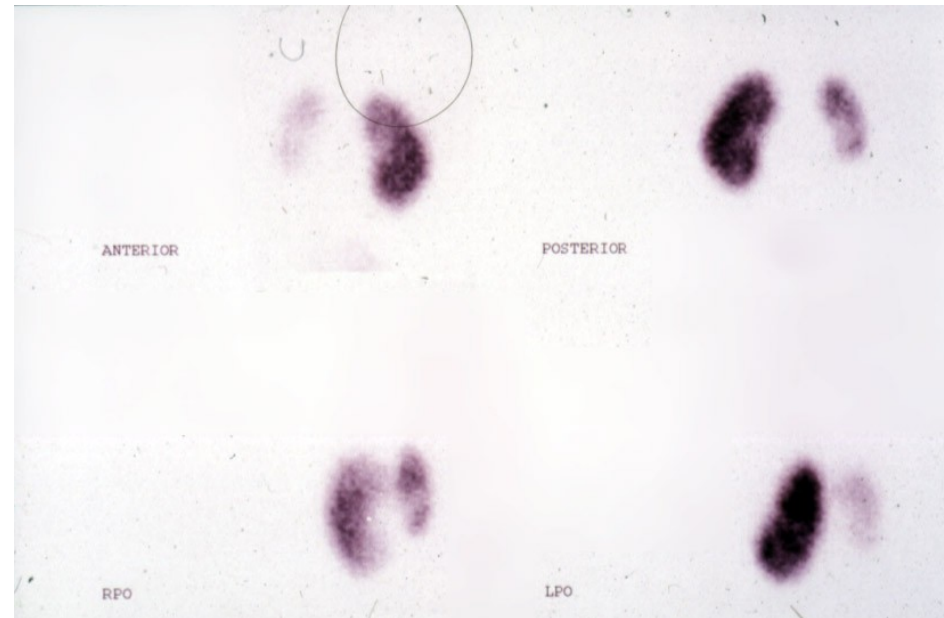
Upper Urinary Tract

- **Most detected on antenatal scans**
- **Usually unilateral**
- **If symptomatic presentation – US Scan**
- **Routine screening not indicated**

Bladder Outlet Obstruction

- Effects both kidneys
- May cause renal failure

In PUV ESRD in approx 30%



To screen or not to screen?



ELSEVIER

Journal of Pediatric urology

Is there an association between functional bladder outlet obstruction and Down's syndrome?

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KEYWORDS

Down's Syndrome;
Functional bladder
outlet obstruction

Abstract *Objective:* The incidence of urinary tract abnormalities in patients with Down's syndrome (DS) is estimated to be 3–7%. Abnormalities included are renal hypoplasia, renal cysts, ureterovesical and ureteropelvic junction obstruction and, more recently, an association between males with DS and the non-neurogenic neurogenic bladder has been reported. Based on clinical experience, the hypothesis is tested that patients with DS have functional bladder outflow obstruction secondary to detrusor sphincter dyssynergia.

Methods: This study comprised three parts: an initial retrospective review of case notes of

Down Syndrome Bladder Outlet Obstruction Retrospective Study

- **8 patients; 7 males, mean age 3 ½ yrs**
- **UTI: 5**
- **Incontinence/Poor urinary stream: 3**
- **Antenatal Hydronephrosis: 1**

J Pediatr Urol 2007; 3: 369

Down Syndrome

Bladder Outlet Obstruction

Retrospective Study

- **US Scans/IVU**

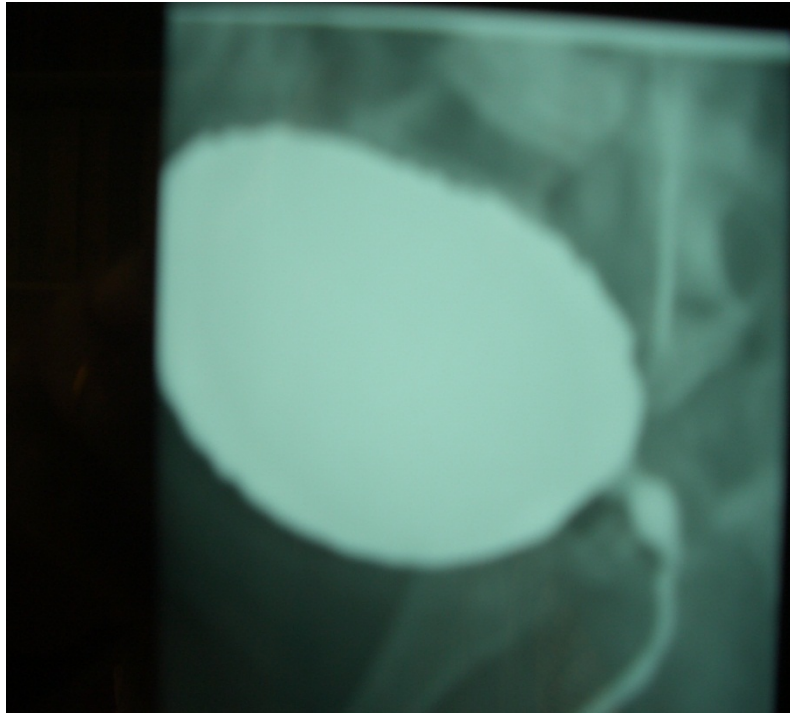
Hydronephrosis

Bladder wall thick/PVR



Down Syndrome Bladder Outlet Obstruction Retrospective Study

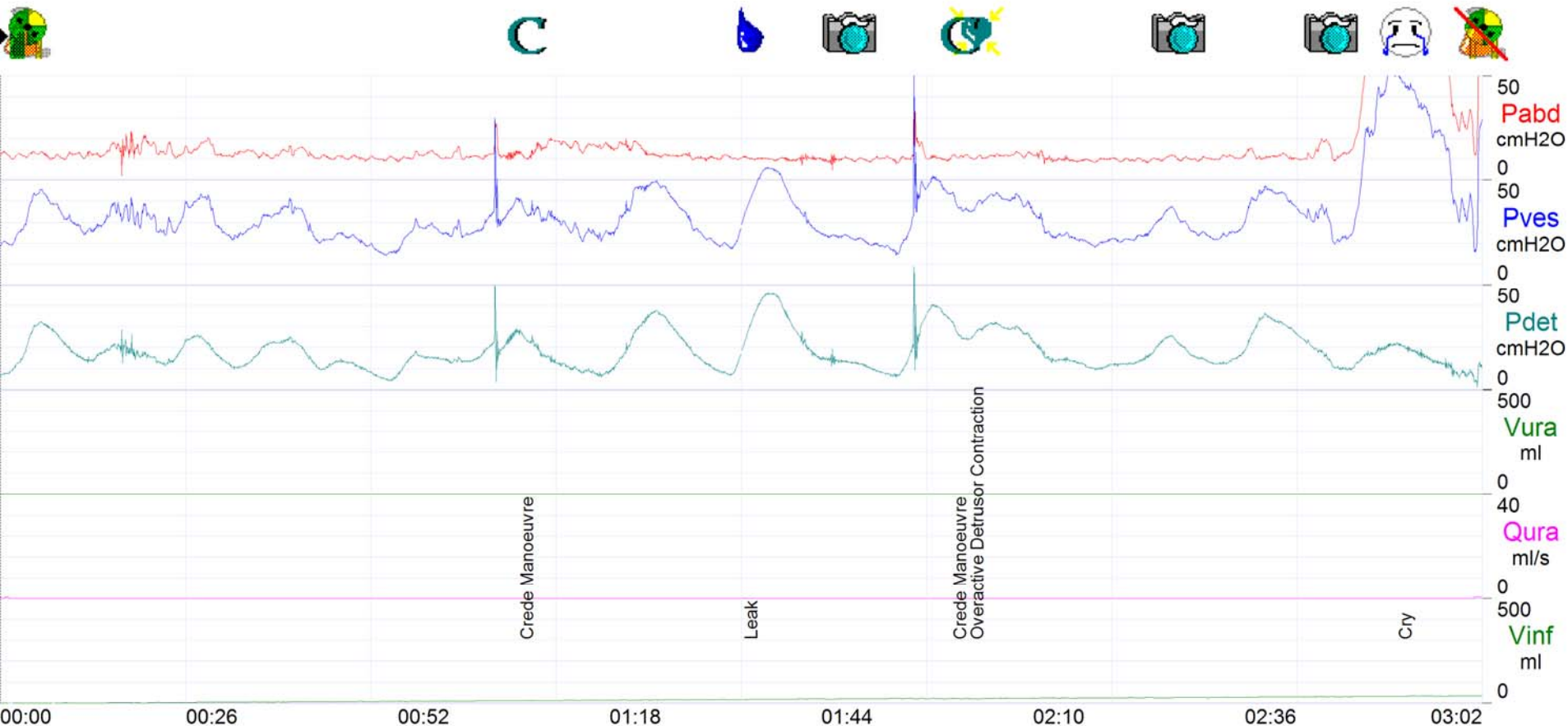
- **Normal spine/Neurology**
- **No anatomical obstruction**
- **? Detrusor Sphincter Dyssnergia**



Down Syndrome Bladder Outlet Obstruction Retrospective Study

- Videourodynamic studies

VUR/Poor compliance/*Detrusor overactivity*



Treatment

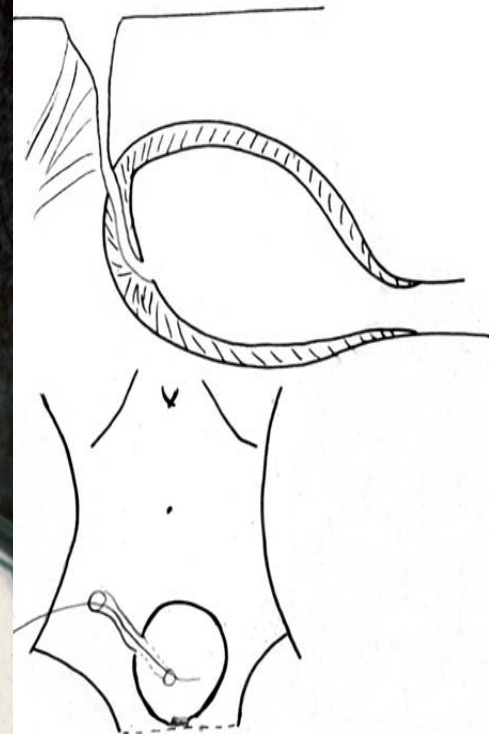
- **3 Urinary diversion**

 - 1 Ileal conduit/2 Mitrofanoff*

- **4 alpha-blockers/anticholinergics**

- **1 Observation**

Mitrofanoff Procedure 1980



MITROFANOFF PROCEDURE

Prospective Study

- A DS patients in local community 5-16 yrs
- Screening postal questionnaire

if positive

- Detailed questionnaire
- Bladder scan/Flow rates

Results

- **23/44 (55%) families responded**
- **74% bladder dysfunction**
 - 65% Incontinence*
 - 48% Weak/Intermittent stream*
- **Flow rate/Bladder scan only in 3**
 - 1 Obstructive flow/Residual*

Conclusions

- **Data not sufficient to recommend routine screening**
- **Enough data to raise concern in those presenting with UTI/Incontinence**
- **Low threshold to investigate those**

NICE Guidelines August 2007
Imaging > 6 Months

Test	Responds in 48 Hrs	Atypical UTI	Recurrent UTI
Acute US	No	Yes	No
Us in 6 weeks	No	No	Yes
DMSA 4-6 months later	No	Yes	Yes
MCUG	No	No	No

Change in Practice

- **Perform US in all with UTI, an exception to NICE guidelines**
- **Maybe not all the incontinence is due to the developmental delay!**
- **Investigate more aggressively with Flow rates/Renal US ?**