

Accuracy of pelvic MRI in evaluating internal genitalia in patients with Disorders of Sex Development "DSD" and at least one palpable gonad

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Background

Patients with disorders of sex development (2/10,000 live births, unknown molecular diagnosis in 80% of cases) require multidisciplinary management for etiology identification and gender assignment. Identification of mullerian structures is a crucial part of the evaluation process. Ultrasonography remains the first-line imaging modality to delineate the internal reproductive organs; while the importance of magnetic resonance imaging is insufficiently studied.

Aim

To evaluate the **diagnostic accuracy of pelvic MRI** in the assessment of internal genitalia in patients with **DSD and at least one palpable gonad at diagnosis**.

Subjects and Methods

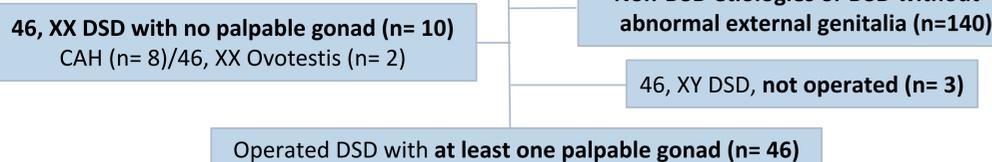
Retrospective descriptive comparative single-center study (Robert Debré hospital 2008-2014) in **DSD patients having at least one palpable gonad, who underwent pelvic MRI and surgical management**. Pelvic MRI of included patients were selected from a radiologic database. Clinical, biological, radiographic, cytogenetic and histopathologic data were evaluated. Imaging were reviewed blindly by the same radiologist. Pelvic MRI findings were compared to US and per-operative cystoscopy whenever performed.

→ **Uterine structure V/S ----> Cavity with a retro-urethral origin (Vagina/ Large utricule)** Sagittal T2 TSE MRI (thickness 2 mm)



Results

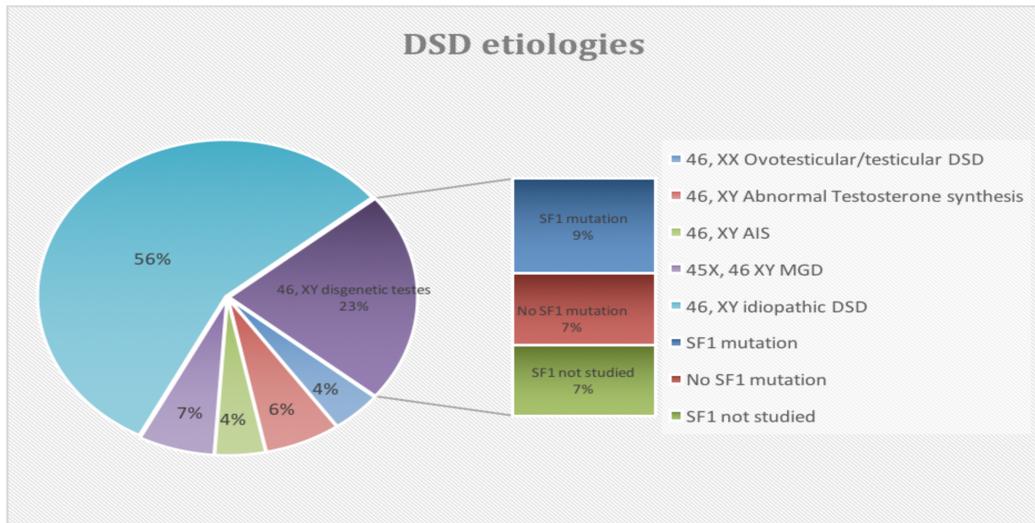
Pelvic MRI (2008-2014) prescribed by pediatric endocrinologist or visceral surgeon (n=199)



Population description (N=46)

	N	%	Mean ± SD	Min-max
Consanguinity	5/44	11,4		
Related familial history	5	10,9		
Prematurity	15	32,6		
Prematurity and SGA	11	23,9		
SGA	17	36,9		
Karyotype				
46 XY (SRY +)	41	89,1		
46 XX (SRY -)	2	4,3		
45 X, 46 XY (SRY +)	3	6,5		
Sex of rearing				
M/ F	45/ 1	98/ 2		
Age at first evaluation				
Neonatal period (< D15)	30	65,2	2 ± 2,1 D	D0 -D11
Minipuberty (D15 - 6M)	11	23,9	3,4 ± 2,3 M	D21-M6
Early childhood (10M - 7Y)	2	4,3		
Late childhood (8Y - 14Y)	3	6,5		
Urethral meatus				
Posterior/ Median	44/ 2	96/ 4		
Genital bud length (mm)				
- Neonatal period			20,1 ± 5,8	8 - 32
- Minipuberty			25,8 ± 3,9	20 - 30
Gonad localization				
- Scrotal/ inguinal (Bilateral)	35/ 2	76/ 4,3		
- Scrotal and inguinal	3	6,5		
- Only one palpable gonad	6	13		

Genetic aspects (After re-analyzing hormonal, clinical and histopathological data) **NGS (puce DSD) being currently performed for all patients with no molecular diagnosis**



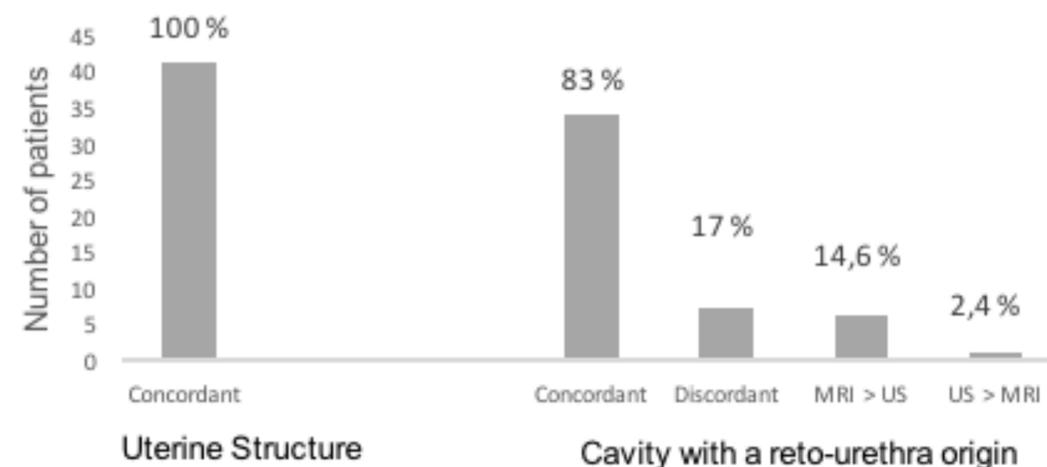
Iconographic features

- Pelvic MRI (n=46)
- Pelvic US (n=41)
- Genitography (n=9)
- Per operative cystoscopy (n=13)

MRI performed at 6 ± 4,4 months (D22 - M20) during minipuberty, 4 during neonatal period
 * 7 Genito-IRM (1 catheter failed to introduce)
 * 39 without Genito-IRM
 -venous overhydration + Lasilix (n=1)
 -Venous overhydration (n=1)

Comparison between pelvic MRI/US findings

- **According to genitography, surgery findings and per-operative cystoscopy: 6 Cavities with a retro-urethral origin were seen only on MRI (Small cavities), 1 seen only on US, 1 not seen on both; One Uterine structure was not shown on both, 3 seen on both. Pelvic MRI findings were concordant with US in identifying vagina/ utricule and uterine structures in 83% and 100% respectively.**



- **According to per-operative cystoscopy considered as a gold standard:** Sensitivities of pelvic MRI and US were **equal (80%)** in identifying uterus with a specificity calculated at **100%** in both cases; while sensitivities to identify vagina or large utricule were estimated at **86,7%** and **80%** for pelvic MRI and US respectively.

Discussion

Advantages of our study

Population size and clinical homogeneity at diagnosis/ Blind review of imaging by one operator/ First study to assess the reliability of pelvic MRI in the evaluation of mullerian structures

Limitations

Retrospective review/ Heterogeneity of exams quality due to several MRI protocols (Depending on urinary flow during exam)

Conclusion and perspective

Pelvic MRI evaluation for internal genitalia appears **complementary to US solely for vagina/ large utricule cavities (mostly small cavities without any infectious complications or surgical removal)**. Thus, its **cost effectiveness** should be reviewed to reduce costs on public health (Hospitalization/ Sedation/ Long delay/ Performed as a **part of the preoperative workup**, rarely involved in the choice of sex of rearing). A **larger prospective study** is required to a **strong imaging protocol consensus** for the evaluation of mullerian structures in children with DSD.