

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

Linda Mahfouz El Nachar (1), Djalal Rekik (2), Nicolas De Roux (3), Matthieu Peycelon (4), Juliane Léger (1,3,5), Annabel Paye-Jaouen (4), Marianne Alison (2), Alaa El Ghoneimi (4), Jean Claude Carel (1,3,5), Laetitia Martinerie (1,5)

(1) Department of Pediatric Endocrinology, Centre de Référence des Maladies Endocriniennes Rares de la Croissance, Robert Debré Hospital, Assistance Publique-Hôpitaux de Paris, F-75019 Paris France. (2) Department of pediatric Radiology, Robert Debré hospital, Assistance Publique-Hôpitaux de Paris, France. (3) Institut National de la Santé et de la Recherche Médicale (Inserm), Unité 1141, DHU Protect, F-75019 Paris, France. (4) Department of visceral surgery, Robert Debré hospital, Assistance Publique-Hôpitaux de Paris, France. (5) Paris Diderot University, Sorbonne Paris Cité, F-75019 Paris, France

PARIS  
DIDEROT

## 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)

46, XX DSD with no palpable gonad (n= 10)  
CAH (n= 8)/46, XX Ovotestis (n= 2)

Non-DSD etiologies or DSD without abnormal external genitalia (n=140)

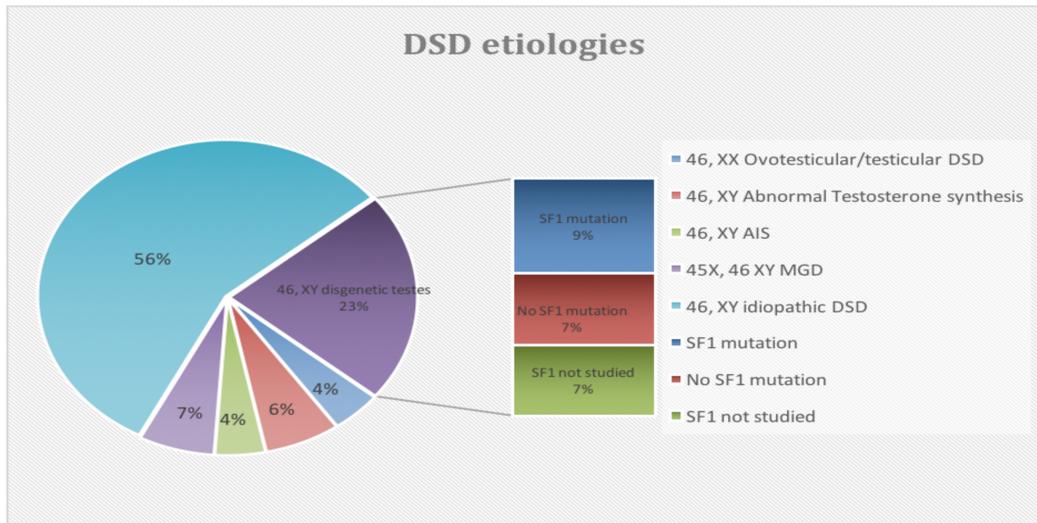
46, XY DSD, not operated (n= 3)

Operated DSD with at least one palpable gonad (n= 46)

### Population description (N=46)

	N	%	Mean ± SD	Min-max
<b>Consanguinity</b>	5/44	11,4		
<b>Related familial history</b>	5	10,9		
Prematurity	15	32,6		
Prematurity and SGA	11	23,9		
SGA	17	36,9		
<b>Karyotype</b>				
46 XY (SRY +)	41	89,1		
46 XX (SRY -)	2	4,3		
45 X, 46 XY (SRY +)	3	6,5		
<b>Sex of rearing</b>				
M/ F	45/ 1	98/ 2		
<b>Age at first evaluation</b>				
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		
<b>Urethral meatus</b>				
Posterior/ Median	44/ 2	96/ 4		
<b>Genital bud length (mm)</b>				
- Neonatal period			20,1 ± 5,8	8 - 32
- Minipuberty			25,8 ± 3,9	20 - 30
<b>Gonad localization</b>				
- 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**



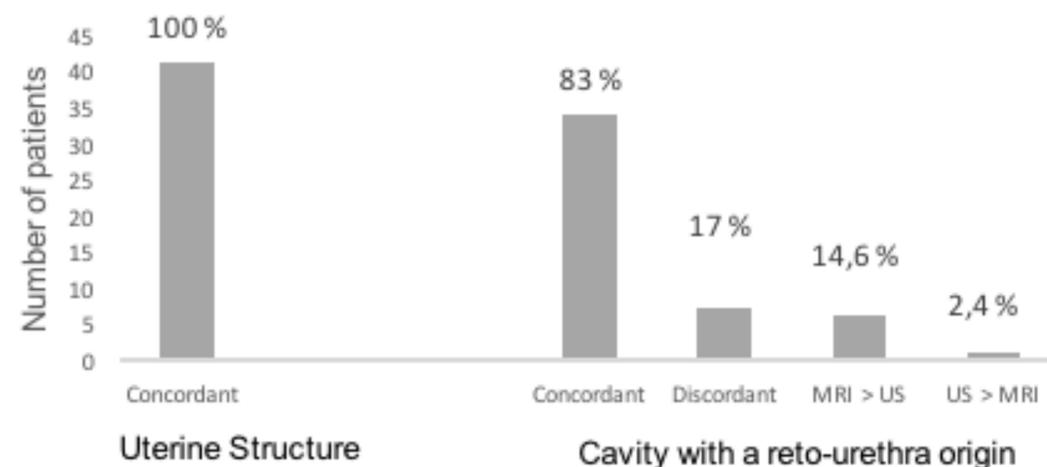
### Iconographic features

- 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.

