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.

Results

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

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

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

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, seen on both. Pelvic MRI findings were concordant with US in identifying vagina/urethra 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 uteruses with a specificity calculated at 100% in both cases; while sensitivities to identify vagina or large utricles 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 müllerian 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 utricle 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 müllerian structures in children with DSD.