Ultrasound of ambiguous genitalia: Challenges and Pitfalls

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Introduction

Ambiguous genitalia or Disorder of Sexual Development (DSD) is when a baby is born with genitalia that look different, this represents a difficult time for the parents, as they may have been aware of the gender of their baby since the 20 week anomaly scan or earlier.[1] Karyotyping of the infant and review by a paediatric endocrinologist and Multidisciplinary team (MDT) is an essential first step.[2] It is important to identify an infant with congenital adrenal hyperplasia (CAH), as this has implications for their health (dehydration, low blood pressure), additionally a chromosomal female infant (46XX) will have ambiguous genitalia due to having increased amounts of testosterone antenatally.[3] The role of the ultrasound practitioner is to identify the uterus and any gonads (ovaries or testes), although care must be taken when communicating the findings with the parents, as identification does not mean that they are functional.[2,4]
Some cases may not become evident until puberty starts and it is important that the ultrasound practitioner understands the common appearances that they may come across in clinical practice, see chart 1.[5]

Chart 1

Condition	Presentation	Gonads	Uterus	Vagina	External genitalia
Sex chromosome DSD					
Turner syndrome (45,X)	Female; other features (1:2000)	Streak ovaries	Yes	Yes	Female
Klinefelter syndrome (47,XXY)	Male; not usually diagnosed (1:700)	Testes	No	No	Male (may be small penis)
Mosaicism (45,X/46,XY)	Newborn (or male) (common)	Variable, dysgenetic testes or streak	Variable; may be hemi-uterus	Variable	Variable (or male); asymmetry
46,XY DSD					
Complete testicular dysgenesis	Female; adolescent, no puberty (rare)	Streak	Yes	Yes	Female
Partial testicular dysgenesis	Newborn (rare)	Testes, dysgenetic testes, streak	Variable	Variable	Variable
Complete androgen insensitivity (CAIS)	Female adolescent, no periods; girl with hernia or lump (rare)	Testes (often inguinal or in hernia)	No	Blind ending	Female
Partial androgen insensitivity (PAIS)	Newborn (rare)	Testes (usually inguinal or high scrotal)	No	Variable	Variable
Penoscrotal hypospadias	Newborn (common)	Testes (often inguinal)	No	No	Hypospadias
46,XX DSD					
Ovotestis	Newborn (rare)	Ovotestis, ovary, testis	Variable	Variable	Variable
Congenital adrenal hyperplasia	Newborn (common)	Ovaries	Yes	Variable	Variable

Pitfalls

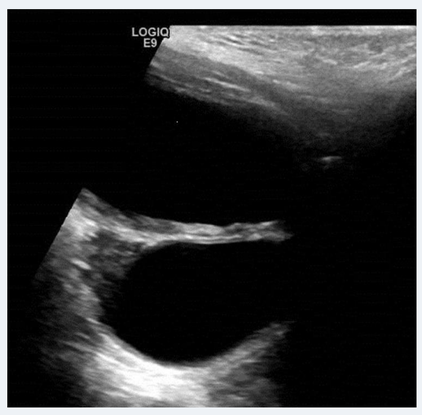
•Utriculus/utricular remnant (see case 1 image) versus genuine uterus (see cases 2, 3 and 5 images) – a prepubertal uterus looks like a utricular remnant (midline structure between the rectum and bladder) unless under the influence of maternal/endogenous hormone stimulation.[5]

•Testes maybe incorrectly identified as lymph nodes when present within the groin area as they have a similar ovoid appearance (see case 3 image). If the lymph node is large enough you should be able to identify a hilar blood supply within versus a mediastinum testis within the testis.[5]

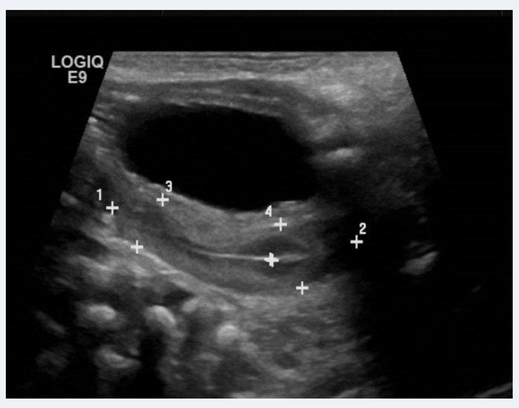
•As with all transabdominal ultrasound scans, sometimes ovaries may not be seen due to overlying bowel gas and incomplete bladder filling (difficult in infants) and then their presence or absence remains unknown. Or they may be difficult to image (see case 4 image). Parents need reassurance and repeat scans or MRI may be useful in these cases.[2,4,5]

•Identifying cysts as an epididymal abnormality when an ovotestis is present and the cysts are ovarian follicles (see case 5 image).

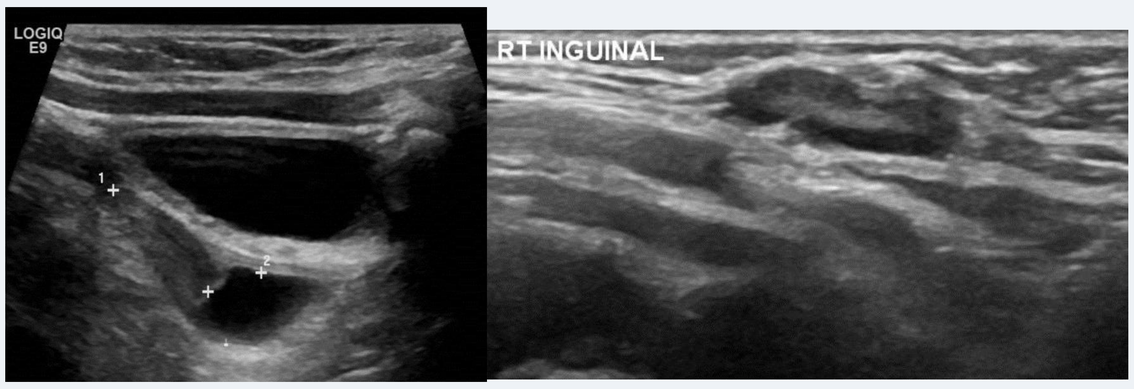
Cases



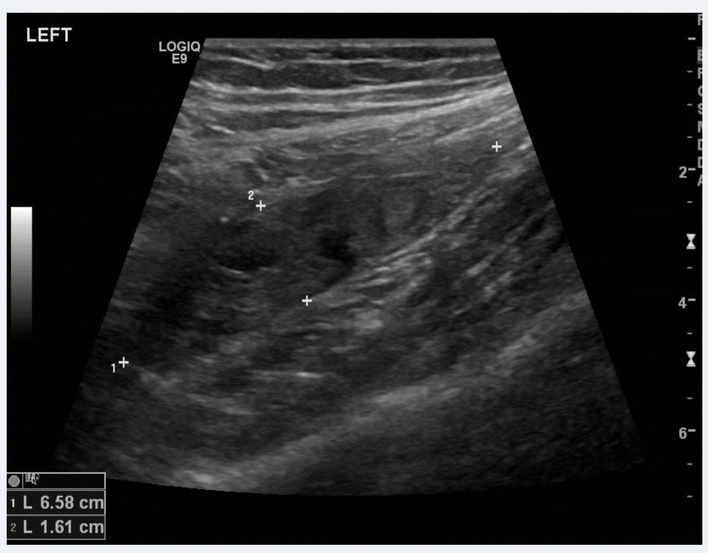
Case 1: Utriculus in patient with mixed gonadal dysgenesis 45XO/46XY



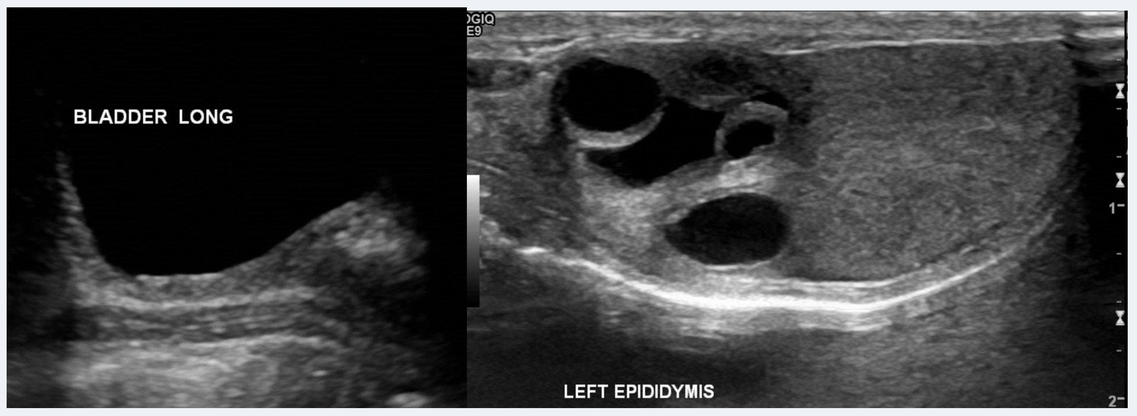
Case 2: Normal plump neonatal uterus in patient with CAH 46XX



Case 3: Uterus + vagina present with small hydrocolpos in patient with gonadal dysgenesis 45XO/46XY and a lymph node in inguinal canal in the same patient (no gonadal tissue identified on ultrasound)



Case 4: Ovotestis in iliac fossa in patient with cloacal anomaly



Case 5: Uterus in patient with DSD 46XX Karyotype and phenotypically male, ovotestes in scrotum with ovarian follicles mistaken for epididymal cysts

Conclusion

Ultrasound plays an important role in the identification of the gonads in infants and older children. The importance of communication with the parents is highlighted, as understanding of the limitations of the ultrasound and often the necessity for serial scans is needed. An MDT and collaborative approach are needed to care for the patient holistically and to determine the sex rearing of the infant. The ultrasound practitioner needs to be as accurate as possible in their diagnosis.

References:
1.Dsdfamilies.org (2014) 'When your baby is born with genitals that look different … The first days'. Available at : https://www.dsdfamilies.org/application/files/3115/3787/2514/English_DSD_booklet.pdf. [accessed: 22 October 2018].
2.Ahmed, S.F., Achermann, J. C., Arlt, W., Balen, A., Conway, G., Edwards, Z., Elford, S., Hughes, I. A., Izatt, L., Krone, N., Miles, H., O'Toole, S., Perry, L., Sanders, C., Simmonds, M., Watt, A. and Willis, D. (2016) 'Society of endocrinology UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development (revised 2015)'. *Clinical Endocrinology*, 84, pp. 771-788.
3.El-Maouche, D., Arlt, W. and Merke, D. P. (2017) 'Congenital adrenal hyperplasia'. *Lancet*, 390, pp. 2194-2210.
4.De Bruyn, R. (2010) *Pediatric Ultrasound: How, why and when*. 2nd edn. Edinburgh: Churchill Livingstone.
5.Achermann, J. (2018) 'Ultrasonography for children with potential differences/disorders of sex development' GOSH protocols.