Chimerism in a teenager with ovotesticular disorder of sexual development (DSD)

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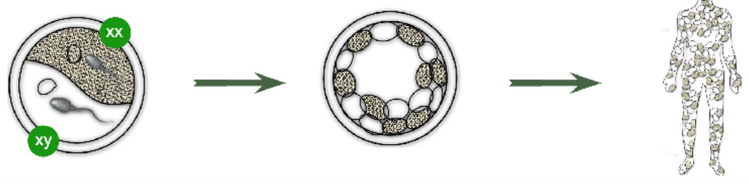
Background

Chimerism is characterised by the presence of two or more genetically distinct cell lines (originating from two or more zygotes) in the same individual.
In situations when the sex of the fertilized eggs is disparate, it can lead to intersex phenotypes.



Case report

A 17 year-old adolescent from Togo presented at birth with perineoscrotal hypospadias. He was raised as a male and underwent urethroplasty at age 10. His twin sister was phenotypically normal and healthy.
At 12 years he developed severe gynecomastia. He expressed male gender identity.
Between 12-17 years he developed severe gynecomastia and periodic hematuria. He expressed male gender identity.
At 17 years he came to Switzerland for mastectomy.

Evaluation by DSD team

1. Clinical assessment:

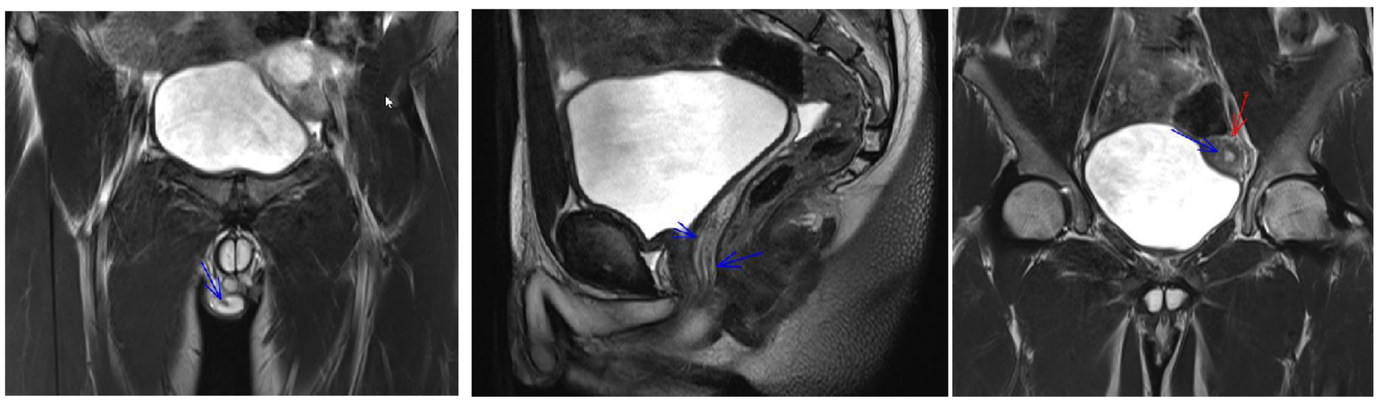
	A_2 PP_4G_4	A_2 PP_5G_4
	Penis size: 6.5x2.5 cm	**Penis size: 7.0x2.8 cm**
	Testicular volume: 2ml	**Testicular volume: 5ml**

2. Biochemical analyses

LH (2-9 UI/l)	12.6	27.2
FSH(2-12 UI/l)	7.7	45
Testosterone (9-32nmol/l for P4)	**4.4**	**10.1**
Œstradiol (0.035-0.13nmol/l for P4)	0.39	
17-OH hydroxyprogesterone (< 3 nmol/l)	1.7	
AMH (4.1-75.7 pmol/l)	10.9	9.6
Inhibine B (67-304 pg/ml for P_4)	35.1	25.5

3. Imaging studies

MRI revealed small right testis, intraabdominal left ovary (2.8 x 2.7 cm) and presence of both an uterus (9.3 ml) and the proximal two thirds of a vagina.



4. Karyotype test: 46 XX [82], 46 XY [18]

5. Diagnostic: **Chimeric ovotesticular DSD**

Management

Surgery: 2nd stage of Bracka's hypospadias surgery, bilateral mastectomy, laparotomy: left ovariectomy and hysterectomy, left testicular prosthesis.

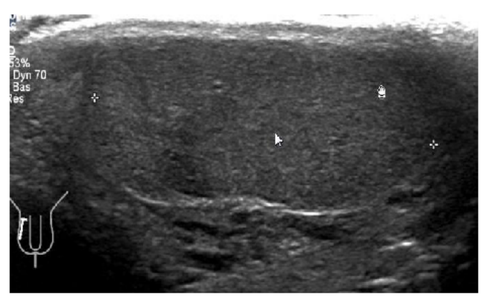
Follow up 3 month after surgery

Ovarian histopathology:

normal ovarian tissue.

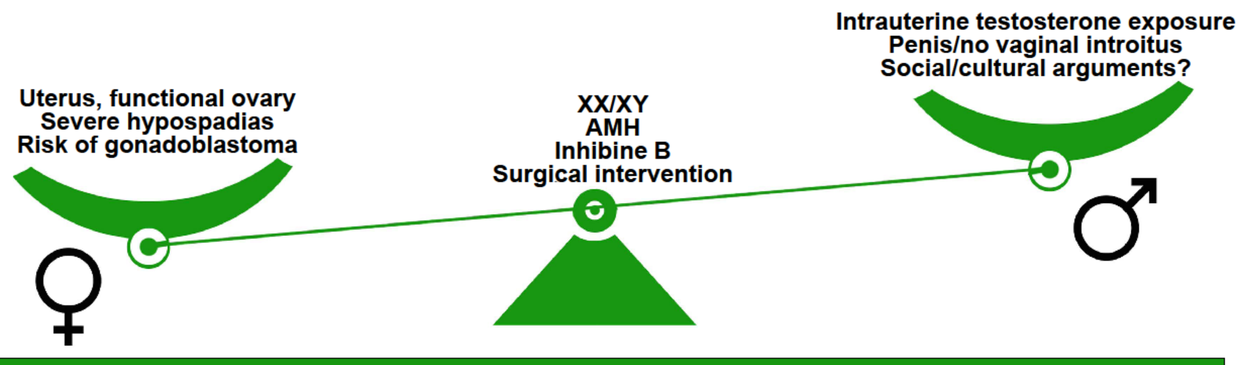
Testicular ultrasonography:

normal tissue, no focal lesions.



Discussion

- **Chimerisme is a rare condition with few reported cases.**
- **Spontaneous fertility has been reported in both sexes.**
- **The functional testis induced spontaneous virislisation after ovarectomy.**
- **Post surgical inhibine B indicates low fertility potential.**
- **Evaluation of risk of gonadoblastoma:**
  For the moment: clinical observation has been choosen.
  For the future: testosterone substitution is probably indicated (gonadectomy ?).



Conclusions

- **Chimerism can cause DSD with broad phenotypic spectrum including possibly functional ovary and testis.**
- **Surgery should be based on gender identity and not on sex of rearing. This argues for later intervention.**
- **One must consider gonadectomy for the risk of gonadoblastoma vs conservative approach (spontaneous virilisation/potential fertility).**

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