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 perineoscutal hypospadias. He was raised as a male and underwent urethreplasty 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 hernia. He expressed male gender identity. At 17 years he came to Switzerland for mastectomy.

Evaluation by DSD team

1. Clinical assessment:
   \[ P_1, P_2, G_1 \]
   Penis size: 6.5x2.5 cm
   Testicular volume: 2ml

2. Biochemical analyses
   - LH (2-9 UI/l): 12.6
   - FSH(2-12 UI/l): 7.7
   - Testosterone (9-32nmol/l for P4): 4.4
   - ßestradiol (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
   - Inhibine B (67-304 pg/ml for P4): 35.1

   \[ P_1, P_2, G_1 \]
   Penis size: 7.0x2.8 cm
   Testicular volume: 5ml

Follow up 3 month after surgery

Ovarian histopathology:
   normal ovarian tissue.

Testicular ultrasonography:
   normal tissue, no focal lesions.

Discussion

- Chimerism is a rare condition with few reported cases.
- Spontaneous fertility has been reported in both sexes.
- The functional tests induced spontaneous virilisation 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 tests.
- 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).