Background:
Partial androgen insensitivity syndromes (PAIS) are rare 46,XY DSD (disorder of sex development).

Objective and hypotheses:
Three families with PAIS (6 patients) are reported, focusing on their phenotype and treatment depending on sex of rearing. Biological investigations and surgical management are described.

Method:
Between 2009 and 2015 a consultation for uro-genital malformations in pediatric patients was set up in Yaoundé (Cameroon). Data on patients with PAIS were retrospectively reviewed.

Results:
- 3 families with atypical genitalia
  - 2 cousins raised male (Patient 1 and 2)
  - 2 sisters (Patients 3 and 4)
  - 2 siblings: one raised female (Patient 5) and the other raised male (Patient 6)
- Mean age at the first consultation: 19 years [14 - 25 years]
- Gender assignment done by parents in the neonatal period without any investigation (3 males, 3 females)
- Clinical examen at presentation: breast development (5/6)
- Biological assessment confirmed 46,XY with androgen receptor (AR) mutation
- All the patients requested surgery supporting of sex of rearing
- Four patients already underwent genital surgery (2 girls and 2 boys):
  - For girls: feminising genitoplasty or vaginal dilatation and orchidectomy
  - For boys: mastectomy, masculinising genitoplasty and orchidopexy
- Only one patient (Patient 4) had sexual activity before surgery
- The operated patients was very satisfied:
  - For 2 males patients (Patient 1 and 2): voiding correctly, good cosmetic penile appearance, not sexual activity yet
  - For female patient (Patient 3): start sexual activity, good clitoral sensitivity and cosmetic aspect
- Patients non operated yet are looking forward for the surgery
- Hormonal therapy (hormonal substitution for girls)

Conclusion:
- Even with the same mutation the genitalia phenotype are very different
- The classically described gynecomastia is not constant
- There was no gender dysphoria despite the phenotype
- Corrective surgery was performed using actual standards
- All patients regretted the late diagnosis and late surgical correction
- The potential of sexual activity and fertility for the boys need to be assessed.
- The management of PAIS patients requires an experienced multidisciplinary team to allow a full clinical and biological assessment