LATE CLINICAL PRESENTATION, BIOLOGICAL ASSESSMENT AND MANAGEMENT OF PAIS IN A DEVELOPING COUNTRY

D Gorduza[1], FT Mouafo[2], CL Gay[1], I Plotton[1], J Birraux[3], S Dahoun[3], P Morel[1], P Mouriquand[1], C Le Coultre[3], PY Mure[1]

[1] Hôpital Femme-Mere-Enfant, Hospices Civils de Lyon, Lyon, France

[2] Yaounde Gynaeco-Obstetric and Paediatrics Hospital, Yaounde, Cameroon

[3] University of Geneva Children's Hospital, Geneva, Switzerland

Background:

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

Objective and hypotheses:

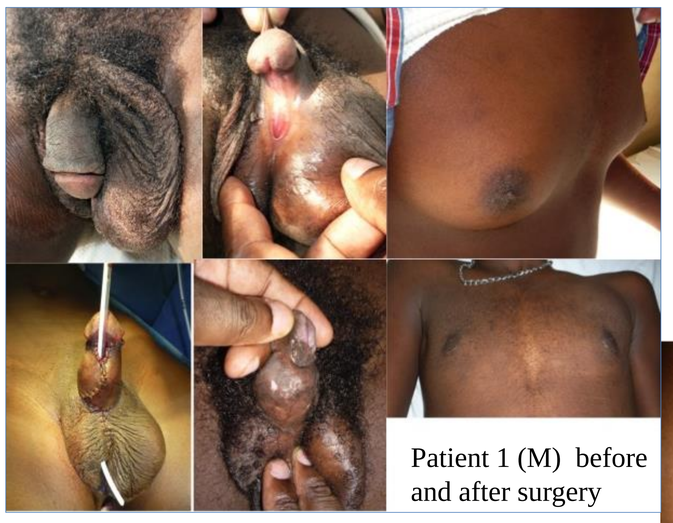
Three families with PAIS (6 patients) are reported, focusing on their phenotype and treatment depending on sex of rearing. Biological investigations and surgical management are described.

Method:

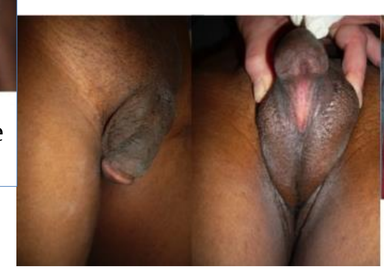
Between 2009 and 2015 a consultation for uro-genital malformations in pediatric patients was set up in Yaoundé (Cameroon).
Data on patients with PAIS were retrospectively reviewed.

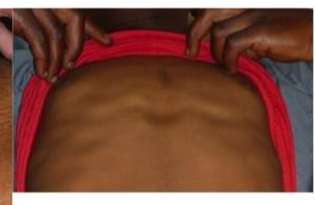
Results:

- 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)
  - variable degree of genital masculinisation
  - palpable testes
- 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 (Patients 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)



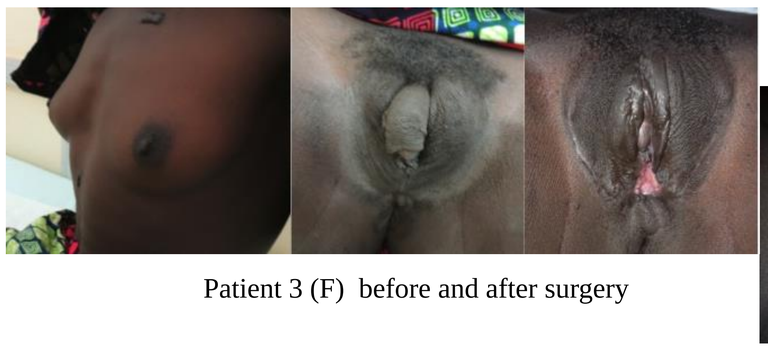
Patient 1 (M) before and after surgery



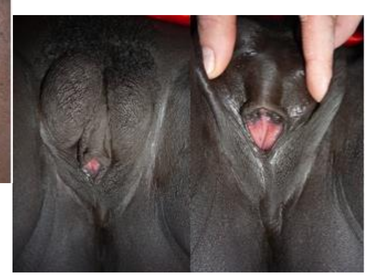


Patient 2 (M)

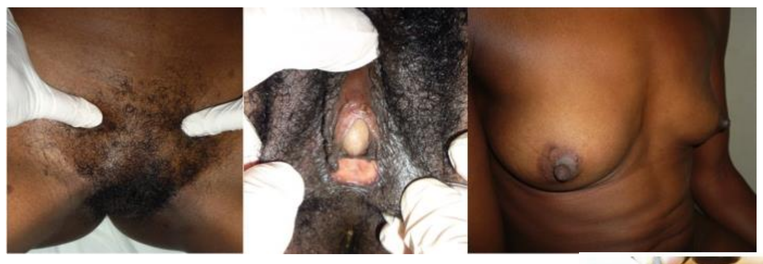
Patient		Sex of rearing	Age	Phenotype					Biological assessement				
				Size of the genital tubercle (cm)	Meatus Position	Chordee	Gonads	Pilosity	Testosterone (nmol/l)	AMH (pmol/L)	FSH (UI/L)	LH (UI/L)	AR mutation
Family 1 Cousins (the mothers are sisters)	1	M	14	8	Perineal	+++	Left scrotal Right not palpable	++	19	300	13	4,7	pAla597Thr
	2	M	18	8	Perineal	+++	Inguinal bilateral	++	22	19	46	20	
Family 2 Sisters	3	F	16	7	Perineal	+++	Right scrotal Left inguinal	+	31,21	292	4,6	22	pSer889
	4	F	24	4	2 perineal orifices (uretral and vagina) vaginal length 6 cm		Scrotal bilateral	+	48	114	16	18	
Family 3 Siblings	5	F	17	4,5	Perineal	+	Inguinal bilateral	++	74,58	45,5	7	18,06	pArg856HIS
	6	M	25	5,5	Perineal	+	Inguinal bilateral	+++	61,66		23,09	21,85	



Patient 3 (F) before and after surgery



Patient 4 (F)



Patient 5 (F)

Patient 6 (M)

Conclusion:

- Even with the same mutation the genitalia phenotype are very different
- The classically descripted gynecomasty 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 sexuality 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

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Gonads & DSD

Daniela GORDUZA

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