## **Case Study**



# Cases of ambiguous genitalia: possibility of congenital adrenal hyperplasia

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#### Introduction

Ambiguous genitalia isn't a disease. It's a sign of a condition that affects sexual development, and it's referred to as a disorder of sexual development. Most common cause of ambiguous genitalia is CAH [1-3].

In our institute we came across 3 paediatric cases of ambiguous genitalia, which were planned for surgery and during their investigation work up they were discovered to be cases of congenital adrenal hyperplasia. So, we would like to discuss some of the presentations of CAH as ambiguous genitalia [4].

#### Materials and method

Case 1: A 8-month-old female patient weight 6 kg presented in opd with single opening in vestibule (no separate urethral and vaginal opening) (Figures 1-5).

Case 2: A 6-year-old patient presented to opd with complaint of ectopic urethral opening with undescended testes (Figures 6 and 7).

Case 3: A 3-day-old, 3 kg, patient presented with ambiguous genitalia (Figure 8).

Hormonal assessment of 17 hydroxy progesterone along with karyotyping was done in all these cases besides routine haematological and preanaesthetic work up (Table 1).



Figure 1. Image showing Karyotype.





Figure 2. Genitoscopic finding.



Figure 3. Before genitoplasty.

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#### Result

All these cases were discovered to be of CAH. They were started on steroid treatment as per paediatrician and endocrinologist and 2 of them were operated for genitoplasty.

#### Discussion

All cases of ambiguous genitalia should be evaluated for medical disorder CAH because the treatment plan changes with apparent sex of the patient and after karyotyping whether to consider baby or child as a male or female. We in our study tried to highlight these features only. Our results like previous studies also suggest early and keen observation in patients of ambiguous genitalia.



Figure 4. Immediate post operation period.



Figure 5. After 3 months of operation.



Figure 6. A 6-year-old patient presented to opd with complaint of ectopic urethral opening with undescended testes.

Death Cares	REPORT	PathCare Labs Private Limited General Laboratory - By No. 34. Chestry of (V), Keese General Comparison (V), Keese Ph. Date 71(2)/721	
Patient Name I Master. DLS Age and Sex : DYS / Male Referring Doctor : SODANI HOS Referring Castomer : SODANI HOS Vial ID : G0768463 Sample Type : WB-Sodium F Cliver Address : LG-1, Norya	IAN KHAN ADDHA PITAL <i>teparin</i> <i>Centre, 16/1 Race Course R</i>	Neg. No.     00491709310085       CCC code     00491709310085       Stock Code     201480-300       Stock Code     211580-2010 02:10 pte       Stock Code     211580-2017 02:10 pte       Stock Code     20140-2017 03:16 AM       Stock Code     05-061-2017 10:51 AM       Stock Code     05-061-2017 10:51 AM	
	CYTOGENI	ITICS	
Chromosomal Analysis- Blood			
CYTOGENETICS NUMBER	PCL/CG/2417/17		
CLINICAL HISTORY	Mild renal hypospadias with h	Internet more and internet in the	
NO OF CELLS SCORED	30	terminate prepable Undescended tests	
NO OF METAPHASES ANALYZED	30		
NO OF METAPHASES KARYOTYPED	0 10		
BANDING METHOD	GTG Banding		
BAND RESOLUTION	550	1 0 000	
KARYOTYPE (ISCN-2013)	46,XX[30]	and the	
DIAGNOSIS	A Male Karyotype with XX chromosmes [30]		
NTERPRETATION	Chapteria and the distribution of the second strength and the second strength		
ECOMMENDATIONS	derivets documentation by the Unotage is a suggested Physical as index should be checked Montaria of Thyroid, relation and generations and generation Montaria of Thyroid, relation and generations suggested FISH for Sex Charmonarias in accommendation to rule out low level monorais		

Figure 7. Image showing patient's report.



Figure 8. A 3-day-old, 3 kg, patient presented with ambiguous genitalia.

Table 1. Table showing comparative investigations.

	Case 1	Case 2	Case 3
Usg abdomen	normal	An elongated isoechoic soft tissue posterior to urinary bladder and anterior to rectum.	normal
Urine (R &M)	normal	normal	normal
karyotype	44+xx female	44+xx male	44+xx female
17 hydroxy progesterone	increased	increased	increased

### Conclusion

In each case presenting with ambiguous genitalia in children, CAH should be kept as a differential diagnosis.

#### References

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