

# Cases of ambiguous genitalia: possibility of congenital adrenal hyperplasia

Eeshansh Khare<sup>1</sup>, Ashok Kumar Laddha<sup>2</sup>, Brijesh Kumar Lahoti<sup>3</sup> and RK Mathur<sup>4</sup>

<sup>1</sup>P G resident, MGM Medical College Indore, India

<sup>2</sup>Assistant Professor, MGM Medical College Indore, India

<sup>3</sup>Associate professor, MGM Medical College Indore, India

<sup>4</sup>Professor and Head of Department, MGM Medical College Indore, India

## 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).

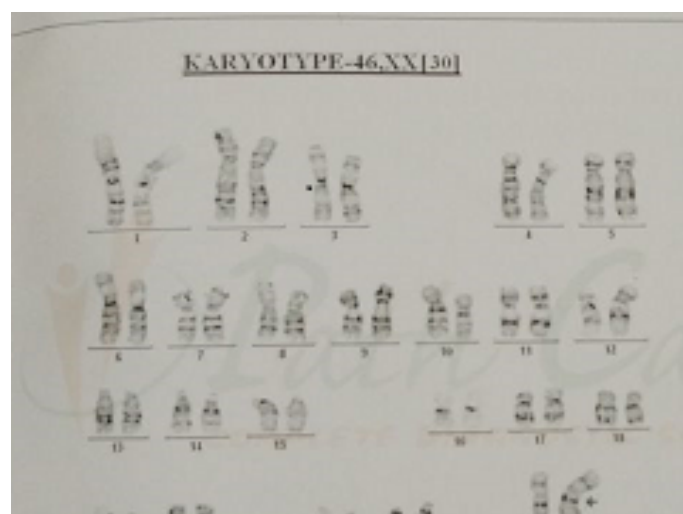


Figure 1. Image showing Karyotype.

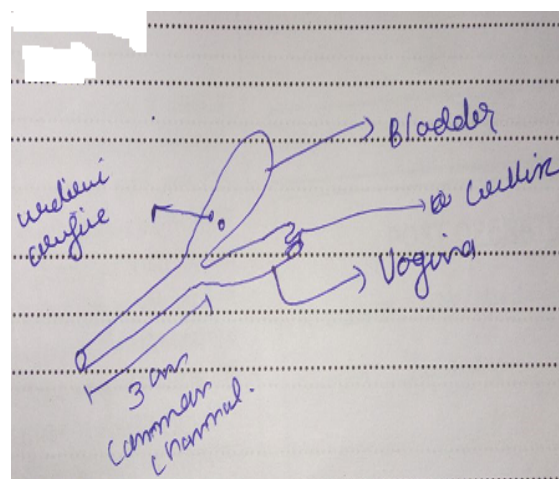


Figure 2. Genitoscopic finding.

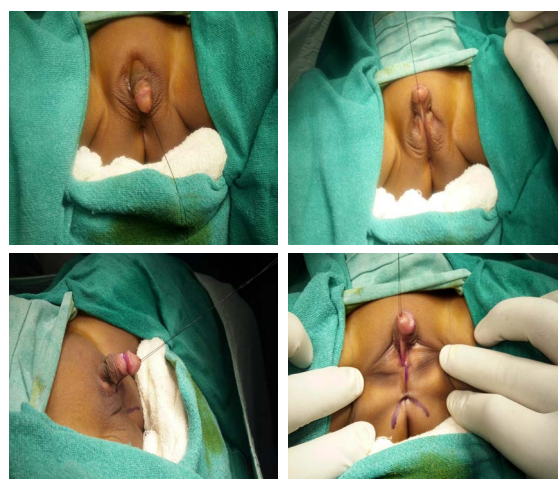


Figure 3. Before genitoplasty.

**Correspondence to:** Eeshansh Khare, P G resident, MGM Medical College Indore, India, Tel: 9669104717; Email: eeshanshkhare29@gmail.com

**Key words:** ambiguous genitalia, CAH (congenital adrenal hyperplasia), karyotyping, 17-hydroxyprogesterone

**Received:** April 02, 2018; **Accepted:** April 11, 2018; **Published:** April 14, 2018

## 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.

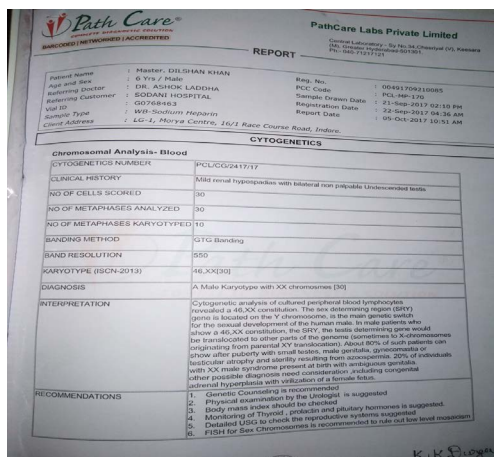


Figure 7. Image showing patient's report.



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

- Ogilvie CM, Crouch NS, Rumsby G, Creighton SM, Liao LM, et al. (2006) Congenital adrenal hyperplasia in adults: A review of medical, surgical and psychological issues. *Clinical Endocrinology (Oxf)* 64: 2-11.
- Hughes I (2005) Prenatal treatment of congenital adrenal hyperplasia: Do we have enough evidence? *Treatments in Endocrinology* 5: 1-6.
- Merke DP, Bornstein SR (2005) Congenital adrenal hyperplasia. *Lancet* 365: 2125-2136. [Crossref]
- Krishnan S and Amy BW (2015) Ambiguous genitalia in new born.

**Copyright:** ©2018 Khare E. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.