International Journal of Scientific Research Studies

ISSN(print): 3050-6905, ISSN(online): 3050-6913

Volume 02 Issue 02 February 2025

DOI: 10.58806//ijsrs.2025.v2i2n01

Page No: 17-20

A Clinical Case of Ambiguous Genitalia in A New Born

Dr Sanchayan Sinha[1], Anannya Ghosh[2], Sharmistha Chatterjee[3]

[1]Consultant Biochemist, Suraksha Diagnostics PVT LTD, Newtown, Kolkata

[2] Demonstrator, Department of Biochemistry, College of Medicine and Sagore Dutta Hospital, Kolkata

[3] Associate Professor, Department of Biochemistry, College of Medicine and Sagore Dutta Hospital, Kolkata

ABSTRACT

Introduction: Ambiguous genitalia, also called as atypical genitalia is a rare condition where a child is born with outer genitals that do not clearly look either male or female. Lack of production of certain hormones can cause the embryo to develop with a female body type, regardless of genetic sex.

Methodology: a 20 days old baby came in OPD with persistent vomiting and dehydration. She had hypernatremia, hypokalemia, cardiac arrhythmia. Genital examination showed ambiguous genitalia. Karyotyping report XY genotype. With conservative management she recovered and went home but returned back in emergency within few days in poor condition. There was severe depletion of serum potassium level and persistent arrhythmia for which the baby was transferred to NICU and put on ventilator. The baby finally died due to persistant arrythmia

Result: on first admission the baby had hypokalemia (2.6 mmol/L), hypernatremia (168 mEq/L), moderate dehydration and metabolic acidisis (pH 7.31, Bicarbonate 18 mmol/L). Baby managed conservatively and Discharged. On 2nd admission the baby had poor general condition, persistant metabolic acidosis (pH 7.28 and Bicorbonate 12 mmol/L) and persistant hypokalemia

Conclusion: with all the above findings we can conclude that the baby was suffering from congenital adrenal hyperplasia probably due to 17 alpha hydroxylase deficiency.

KEYWORDS: Day Case Surgery, Pediatric, Anesthesia, Caregiver.

ARTICLE DETAILS

Published On:
07 February 2025

Available on:
https://ijsrs.org/

INTRODUCTION: Ambiguous genitalia, also called as atypical genitalia is a rare condition where a child is born with outer genitals that do not clearly look either male or female. They may have features of both sexes or not be fully developed. The characteristics of the child's genitals may not match their internal sex organs or their genetic sex. [1]

Sex determination and sex differentiation can be divided into chromosomal sex or karyotype (46 XX, 46 XY, or variants), gonadal sex (presence of a testis or ovary) and phenotypic sex (external genitalia and internal structures). [1]

Human embryo is bipotential with genetic sex determined at conception. The male and female reproductive organs and genitals both come from the same tissue in the fetus. Male differentiation requires specific molecular and biochemical stimuli at specific times. Female differentiation occurs in absence of male determinant genes. [2]

Ambiguous genitalia can develop if the process that causes this fetal tissue to become "male" or "female" is disrupted. The physical appearance of people with this condition can vary widely. Rarely, the physical appearance may be fully developed as the opposite of the genetic sex. For example, a genetic male may have developed the appearance of a female. [2]

The common disorders of sexual development are Klinefelter Syndrome (47XXY), Turner Syndrome (45X), Mixed gonadal dysgenesis, Congenital adrenal hyperplasia (CAH), Androgen insensitivity syndrome and Cryptorchidism [3]

Lack of production of certain hormones can cause the embryo to develop with a female body type, regardless of genetic sex. Androgen insensitivity syndrome is such a condition where even if the body makes the hormones needed to develop into a physical male, the body does not respond normally to those hormones. This may produce female characteristics, even though the genetic sex is male. [3]

Here we presented a case of a case of a newborn with ambiguous genitalia

THE CASE: 20 days old female (previously diagnosed at the time of birth as per birth certificate) patient came to the emergency with complaints of intermittent vomiting for 3 days. On examination the baby was moderately dehydrated, irritable and semiconscious. mother gives history of premature birth delivered by SVD at 28 weeks. Birth weight was 1.7 Kg. baby was kept in NICU for first 72 hours where he was treated with fluid, oxygen and phototherapy (for 1 day) for physiological jaundice. No history of sepsis or antibiotic estimation had been seen according to the previous treatment record. The baby along with the mother was discharged on her Day 5 of life. Mother also complained of history of mild respiratory distress since but which gradually aggravated for last 6 to 7 days for which she took the baby to a local health centre where she was treated with bronchodilator, fluid, expressed milk, nebulization and antibiotics. For last 3 days because of intractable vomiting and altered sensorium the baby had been referred to the emergency of College of Medicine and Sagore Dutta Hospital.

MATERIALS AND METHODS: the baby was then admitted in neonatal intensive care unit. On admission she had severe respiratory distress with RR 84/m, chest retraction, mild cyanosis was present. Heart rate was 142/m, BP was 70/40 mmHg, the baby was irritable but lethargic, with moderate dehydration. Pallor and jaundice were absent.

The baby was primarily treated with fluid, oxygen, nebulization. Blood sample was collected at the admission. Blood reports showed normal hemogram, hypernatremia, hypokalaemia, normal liver function test and thyroid function test reports.

The arterial blood gas report at 12 hours of admission showed metabolic acidosis with normal sodium and persistent hypokalaemia. Baby was then put on Na-bicarbonate to combat metabolic acidosis and potassium chloride injection for hypokalemia.

With 4 days of supportive treatment of hypokalaemia the baby stabilized, respiratory distress improved, electrolyte imbalance corrected.

But the systemic examination the baby had persistent tachycardia. Genital examination showed enlarged clitoris/penis.

The chest X ray and ECG was normal, echocardiography was within normal limit.

Then the paediatrician suggested for Karyotyping in which 46XY was found.

After 8 days of homestay, the baby was again hospitalised and for persistent respiratory distress. On admission the baby was diagnosed again to have hypernatremia, hypokalemia and metabolic acidosis. The baby was shifted to the NICU for better management and was put in mechanical ventilation. Gradually the baby's condition became poor and with persistent acidosis and after 5 days of hospital stay the baby died.

RESULTS

Table 1: investigations done on the 1st time admission as follows:

Parameters	Method	Value	Unit	Reference range
Bilirubin (Total)	Photometry	1.6	Mg/dl	
Bilirubin (direct)	Photometry	0.6	Mg/dl	
Urea	Photometry	12.5	Mg/dl	
Creatinine	Photometry	0.82	Mg/dl	
Sodium	Indirect ISE	168	mEq/L	135-145
Potassium	Indirect ISE	2.6	mmol/L	3.6-5.2
Random blood sugar	Photometry	52	Mg/dl	
Hemoglobin	Photometry	14.9	g/dl	11.5-15.5
Total leukocyte count	DC detection method	8600	*10^3/µL	5 - 13
Differential count	DC detection method	N: 42% L:52% M:3% E:2% B 1%	%	N-40-60% L-20-40% M-2-5% E-1-4% B-0-0.9%
ESR	Westergren method	18	Mm/hr	3-13
Platelet	Microscopy	260*10^3	*10^3/µL	170-450*10^3
CRP	Immunoturbidimetry	19	Mg/dl	<10

Abbreviation: RBC – Red Blood Corpuscles, WBC – White blood corpuscle N – Neutrophil L-Lymphopcyte E- Eosinophil B-Basophil M – Monocyte, PCV-Packed cell volume

Table 2: ABG report on 1st admission

Parameter	Value	Unit	Reference range
pH	7.31		7.35-7.45
pCO2	46	mmHg	35-45
pO2	78	mmHg	75-100
HCO3	18	Mmol/L	22-26
BE	-4	Mmol/L	±2

Abbreviation: BE: Base Excess

Table 3: ABG report on 2nd admission

Parameter	Value	Unit	Reference range
pH	7.28		7.35-7.45
pCO2	48	mmHg	35-45
pO2	74	mmHg	75-100
HCO3	12	Mmol/L	22-26
BE	-10	Mmol/L	±2

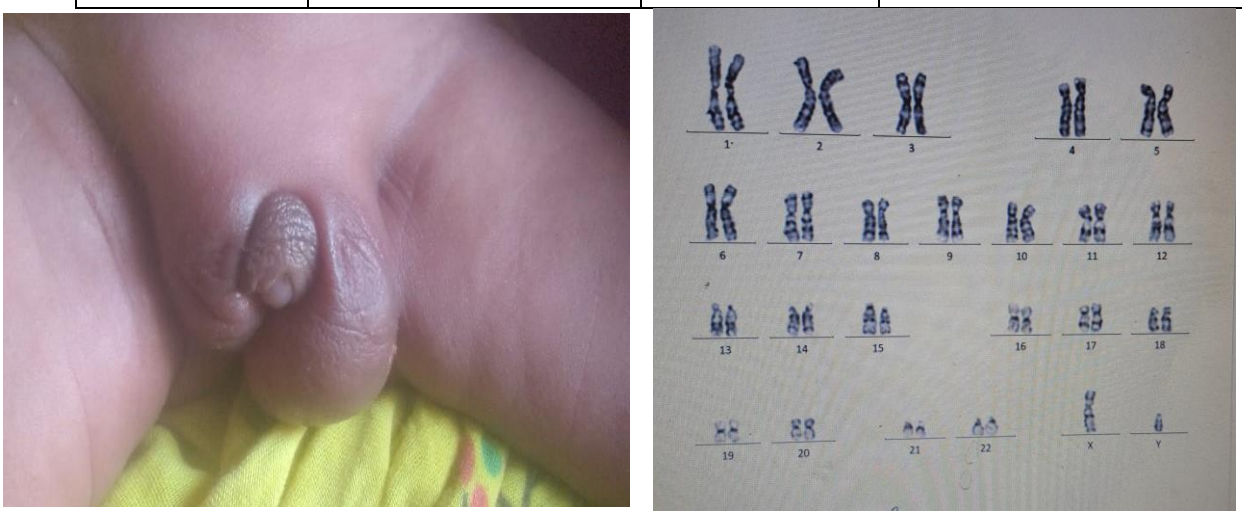

Fig 1: external genitalia and the karyotyping profile of the baby

DISCUSSION

17-alpha-hydroxylase deficiency (17OHD) is a rare autosomal recessive disorder caused by mutations in the *CYP17A1* gene, which occurs in 1 in 50 000 live births and only 1% of congenital adrenal hyperplasia (CAH).[4] There is decrease in cortisol, androgen, and estrogen production with a subsequent increase in adrenocorticotrophic hormone (ACTH) and gonadotropin levels.[5] High ACTH levels contribute to increased production and accumulation of 17-deoxycorticosteroids, especially deoxycorticosterone. Excessive levels of mineralocorticoids lead to volume expansion, hypertension, hypokalemia, and high, normal, or suppressed aldosterone, with oligo/amenorrhea in females and pseudo-hermaphroditism in males[6]

Androgen and estrogen production is also affected because 17-hydroxylase/17 in the gonads, plays an important role in sexual maturity throughout fetal life and puberty. [7] This results in ambiguous or female external genitalia in affected male individuals and absent or delayed pubertal development in female patients.[8]

In the present case, the new born baby presented with hypernatremia, hypokalaemia, ambiguous genitalia. The baby was dehydrated during hospitalisation. During the course of treatment, there was persistent hypokalaemia, hypernatremia, metabolic acidosis and arrhythmia.

The karyotyping profile of the baby shows presence of Y chromosome (genotypically male). But the external genitalia is poorly developed (small sized penis resembling to clitoris).

In 17OHD there may salt and water wasting and hypokalemia where in the present case we got the presence of hypokalemia and dehydration. But in our case, the baby had hypernatremia may be due to excessive vomiting and dehydration.

The cardiovascular defect in the present case can be explained by possibility of arrhythmia asnd sudden cardiac arrest in congenital adrenal hyperplasia as per the study of Agarwal S et al in 2005. [9]

The persistent hypokalemia and the arrhythmia which is a well established complication in the congenital adrenal hyperplasia has also been found in our case and the baby died due to cardiac arrest with persistent arrhythmia

CONCLUSION

with all the above findings we can conclude that the baby was suffering from congenital adrenal hyperplasia probably due to 17 alpha hydroxylase deficiency

LIMITATION: due to lack of time, and resources we could not perform the assay of hormones like ACTH, CRH, and enzyme like17 alpha hydroxylase directly

REFERENCE

1) Fischer KM, Kolon TF. Embryology and differences of sex development. In: Guzzo TJ, Wein AJ, Kovell RC, Weiss DA, Ziemba JB, eds. Penn Clinical Manual of Urology. 3rd ed. Philadelphia, PA: Elsevier; 2024:chap 25
2) Sorbara JC, Wherrett DK. Disorders of sex development. In: Martin RJ, Fanaroff AA, Walsh MC, eds. Fanaroff and Martin's Neonatal-Perinatal Medicine. 11th ed. Philadelphia, PA: Elsevier; 2020:chap 89
3) Yu RN, Diamond DA. Disorders of sexual development: etiology, evaluation, and medical management. In: Partin AW, Dmochowski RR, Kavoussi LR, Peters CA, eds. Campbell-Walsh-Wein Urology. 12th ed. Philadelphia, PA: Elsevier; 2021:chap 48
4) Wong SL Shu SG & Tsai CR. Seventeen alpha-hydroxylase deficiency. Journal of the Formosan Medical Association 2006105177–181. (10.1016/S0929-6646(0960342-9) [DOI] [PubMed] [Google Scholar]
5) Philip J Anjali Thomas N Rajaratnam S & Seshadri MS. 17-Alpha hydroxylase deficiency: an unusual cause of secondary amenorrhoea. Australian and New Zealand Journal of Obstetrics and Gynaecology 200444477–478. (10.1111/j.1479-828X.2004.00275.x) [DOI] [PubMed] [Google Scholar]
6) Kim SM & Rhee JH. A case of 17 alpha-hydroxylase deficiency. Clinical and Experimental Reproductive Medicine 20154272–76. (10.5653/cerm.2015.42.2.72) [DOI]
7) Miller WL. The syndrome of 17,20 lyase deficiency. Journal of Clinical Endocrinology and Metabolism 20129759–67. (10.1210/jc.2011-2161) [DOI] [PMC free article] [PubMed] [Google Scholar]
8) Larson A Nokoff NJ & Travers S. Disorders of sex development: clinically relevant genes involved in gonadal differentiation. Discovery Medicine 201214301–309.
9) Sudden cardiac arrest in a neonate with congenital adrenal hyperplasia. Agarwal S, Deshpande G, Agarwal D, Dave A, Shah JJ. Pediatr Cardiol. 2005;26:686–689. doi: 10.1007/s00246-004-0858-3.