

Original article:

Clinical study of ambiguous genitalia in pediatric cases

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Abstract:

Introduction: For families the birth of a child is a long anticipated, exhilarating event. The first question in the delivery room is "Is it a boy or girl?" The first thing that interested family members and friends inquire about is the sex of the baby. The attending doctor is faced with a dilemma of how to deal with the situation when he is confronted with a newborn with ambiguous genitalia.

Material and methods: This is the study of thirty cases of ambiguous genitalia at this institution for two years duration

Results: The first few moments, of parents, after the delivery of a new baby are filled with joy. The first announcement made after the delivery is of the gender of the baby and this determines the sex of rearing. Sometimes, however, the gender assignment is difficult because of ambiguity of the external genitalia.

Conclusion: From this study we may conclude that intersex is a rare but important group of disorder.

INTRODUCTION

For families the birth of a child is a long anticipated, exhilarating event. The first question in the delivery room is "Is it a boy or girl?" The first thing that interested family members and friends inquire about is the sex of the baby.¹ The attending doctor is faced with a dilemma of how to deal with the situation when he is confronted with a newborn with ambiguous genitalia. It is traumatic for parents to hear that the sex of child cannot be determined. It is essential to explain to the parents that their child has a birth defect that interferes with the usual way of determining the sex of a baby. Parents may feel as though they have given birth to a "Freak".

MATERIAL AND METHODS

This is the study of thirty cases of ambiguous genitalia at this institution for two years duration. The cases were categorized & divided into the following group

Female pseudohermaphroditism

Male pseudohermaphroditism

True Hermaphroditism

Mixed gonadal dysgenesis

The first two have only ambiguous external genitalia where as the last two have ambiguity of both internal and external genitalia and chromosomes. The patients are evaluated in the newborn period or when they come in to the O.P.D jointly by pediatric endocrinologist and pediatric surgeon. Mainly the patients are referred to pediatric surgeon after the initial investigations for proper gender assignment. Once an appropriate sex assignment has been made, the next critical step is performance if needed of a reconstructive procedure in a timely fashion. In this study

thirty patients of ambiguous genitalia were treated surgically. Diagnostic workup included blood, urine steroid measurements, x- ray, and ultrasound.

OBSERVATION & RESULTS

There were 30 patients of ambiguous genitalia studied in period of two years duration. After investigations the cases were divided in to 4 broad groups as follows:

Table No: 1 GROUPS

Group	No of patients	Percentage
Female Pseudohermaphroditism	13	43.33 %
Male Pseudohermaphroditism	9	30.00 %
Mixed gonadal dysgenesis	2	6.67 %
True hermaphroditism	6	20.00 %
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Table No: 2 AGE AT PRESENTATION:

Group	No of patients	Percentage
Less than 1 month	3	10.00 %
1 month - 1year	16	53.33 %
1year - 5years	6	20.00 %
More than 5years	5	16.67%

Table No: 3 The patients presented with different complaints

• REASON FOR CONSULTATION:

Group	No of patients
Ambiguous genitalia	20
Undescended testis	3
Abnormally sited urethral orifice	2
Clitoromegaly	3
Diarrhea, Vomiting	2

Table No: 4 FEMALE PSEUDOHERMAPHRODITISM:

Patient No.	Age at evaluation	Presenting symptoms	Karyotype
1	9 months	Ambiguous genitalia	46XX
2	7 years	Ambiguous genitalia	46XX
3	1 months	Vomiting + Diarrhea	46XX

4	1 ^{1/2} months	Clitoromeagaly	46XX
5	2 years	Vomiting + Diarrhea	46XX
6	3 years	Ambiguous genitalia	46XX
7	6 years	Bilateral UDT	46XX
8	10 years	Ambiguous genitalia	46XX
9	3 months	Ambiguous genitalia	46XX
10	5 years	Ambiguous genitalia	46XX
11	7 years	Ambiguous genitalia	46XX
12	10 days	Clitoromeagaly	46XX
13	2 months	Ambiguous genitalia	46XX

Table No: 5 EXAMINATION OF EXTERNAL GENITALIA OF FEMALE PSEUDOHERMAPHRODITES

Pt No.	Gonad		Phallus length (cm)	Perineal opening	Labio Scrotal Folds	Hyper pigmentation
	Left	Right				
1	NP	NP	3.0	Two	Fused	+
2	NP	NP	2.5	Single	Bifid	+
3	NP	NP	3.0	Two	Normal	+
4	NP	NP	3.0	Two	Normal	+
5	NP	NP	2.0	Two	Fused	+
6	NP	NP	3.5	Single	Fused	+
7	NP	NP	3.0	Single	Bifid	+
8	NP	NP	3.5	Single	Bifid	-
9	NP	NP	3.0	Single	Fused	+
10	NP	NP	3.0	Two	Bifid	+
11	NP	NP	3.5	Two	Bifid	-
12	NP	NP	3.0	Single	Fused	+
13	NP	NP	2.5	Single	Bifid	+

NP – Nonpalpable

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Table No: 6 INVESTIGATIONS AND TREATMENT IN FEMALE PSEUDOHERMAPHRODITES

Pt No.	Hormonal Study	Genitogram	Operative procedure	Sex of rearing
1	Increase 17 OHP	Short Common	Clitoroplasty	Female

		channel		
2	Increase 17 OHP Decrease S.Cortisol	Long Common channel	Clitorovaginoplasty	Female
3	Increase 17 OHP	Cervical dimple +	Clitorovaginoplasty	Female
4	Increase 17 OHP Increase S.11 Deoxycortisol	Cervical dimple +	Clitoroplasty	Female
5	Increase 17 OHP	Short Common channel	Clitorovaginoplasty	Female
6	Increase 17 OHP	Cervical dimple +	Clitorovaginoplasty	Female
7	Increase 17 OHP	Lost to follow up	Lost to follow up	-
8	Urinary 17 OHP Increase	Short Common channel	Clitorovaginoplasty	Female
9	Increase 17 OHP	Long Channel Cervical dimple	Clitorovaginoplasty	Female
10	Increase 17 OHP	Cervical dimple +	Clitoroplasty	Female
11	Increase 17 OHP	Cervical dimple +	Clitoroplasty	Female
12	Increase 17 OHP	Lost to follow up	Lost to follow up	-
13	Increase 17 OHP	Awaited	Awaited	Awaited

OHP – Hydroxyprogesterone

Table No: 7 MALE PSEUDOHERMAPHRODITISM

Pt no.	Age at evaluation	Presenting symptom	Kartype
1	10 months	Ambiguous genitalia	46,XY
2	3 months	Ambiguous genitalia	46,XY
3	3 months	Ambiguous genitalia	46,XY
4	11/2 years	Hypospadias	46,XY
5	4 months	Ambiguous genitalia	46,XY
6	4 months	Ambiguous genitalia	46,XY
7	9 months	Undescended testis	46,XY
8	10 months	Hypospadias	46,XY
9	11/2 years	Ambiguous genitalia	46,XY

Table No: 8 EXAMINATION OF EXTERNAL GENITALIA IN MALE
PSEUDOHERMAPHRODITES

Pt. No.	Phallus length (cm)	Gonad	
		Left	Right
1	3.5	SIP	P
2	2.0	P	P
3	3.0	P	P
4	4.2	P	P
5	1.5	P	P
6	2.5	P	P
7	3.0	SIP	P
8	1.5	P	P
9	1.5	P	P

SIP - Superficial Inguinal Pouch

NP - Non-Palpable, P - Palpable

DISCUSSION:

The first few moments, of parents, after the delivery of a new baby are filled with joy. The first announcement made after the delivery is of the gender of the baby and this determines the sex of rearing. Sometimes, however, the gender assignment is difficult because of ambiguity of the external genitalia.³

The infant born with ambiguous genitalia requires accurate and prompt diagnosis, so that management plan can be formulated to address the separate requirements of the child and parents. The child may need urgent medical treatment of a life threatening endocrine disorder such as congenital adrenal hyperplasia, or surgical treatment for the ambiguous genitalia.⁴The first few days for many parents, are horrifying. It is devastating for them to be told that physicians are having problem in determining the sex of the baby. Because there are genetic reasons for child's abnormality, the parents may have to face similar experience in the future.⁵Not surprising, parents become desperate for emotional support, feel intensely guilty and need a clear explanation of their baby's condition. In the recent years, scientific understanding of the pathogenesis of many disorders of sexual differentiation has increased greatly.⁶

In this bewildering, confusing and complex problem of intersex, the pediatric surgeon is one of the 4 wheels of a chariot, in its total management. The surgeon is all the more important as he has to be involved in the management from the time of diagnosis till the child is positioned in society with complete redesigning, reconstruction, of the anatomy as found.

CONCLUSION:

From this study we may conclude that intersex is a rare but important group of disorder.

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