

DETECTION OF SEX CHROMATIN IN BABIES WITH AMBIGUOUS GENITALIA

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Introduction

Ambiguous genitalia in a new born baby is a major problem to the health care worker as well as the family of the affected. To minimize psychosocial problems that may arise in the families of affected babies, early detection and quick determination of the sex of the individual is essential. However this may pose a problem since access to chromosomal analysis is not readily available to all institutions. The Barr body examination and identifying the drumstick chromosome are two relatively simple methods of detecting the inactive X chromosome. This can give the clinician an idea about the chromosomal sex of the baby.

Materials and Methods

The sample included 18 babies with ambiguous genitalia referred to the Department of Anatomy for detection of sex chromatin from January 2006 to August 2009. A smear from the buccal mucosa was obtained with a wooden spatula after cleaning the mucosa with cotton wool. Smears were taken after 30 to 45 minutes of breast feeding. The smear was stained with basic fuchsin. A capillary smear was obtained by adding a drop of blood obtained by heel prick on to a clean

slide. Smear was stained with Giemsa. Prepared smears were examined under light microscope for the presence of Barr bodies and the drumstick chromosome.

Consent was sort from the mother, and the baby was examined under good light. Abnormalities detected were recorded.

Results and Discussion

The results are given in Table 1. It was observed that the defects in the genitalia had been detected at birth in all babies, and that health personal had explained the problem to the parents. However it was seen that the parents had formed their own opinion and had named their babies giving them what appeared to be the most likely gender. Only one baby had other congenital anomalies.

Seven of the babies had been identified as having congenital adrenal hyperplasia and were already on treatment. Congenital adrenal hyperplasia is known to be the commonest cause for ambiguous genitalia worldwide. Barr bodies and drumstick chromosomes were positive, suggesting these babies to be of female genotype.

Table 1: Sex chromatin findings of 18 babies

No	Age in days	Birth weight (grams)	Abnormalities in genitalia	Probable diagnosis	Named as	Barr body	Drumstick
1	10	2600	Female type	Adrenal hyperplasia	Not named	Positive	Positive
2	6	3105	Female type	Adrenal hyperplasia	Not named	Positive	Positive
3	14	2800	Male type	Mixed gonadal dysgenesis	Boy	Positive cells and negative cells	Few positive cells
4	21	2600	Female type	Adrenal hyperplasia	Girl	Positive	Positive
5	30	2250	Female type	Adrenal hyperplasia	Girl	Positive	Positive
6	60	2600	Male type	Hypospadias and undescended testes	Boy	Negative	Negative
7	7	2550	Male type	Hypospadias and undescended testes	Boy	Negative	Negative
8	9	2300	Female type	Syndromic	None	Positive	Positive
9	18	2500	Female type	Normal	Girl	Positive	Positive
10	5	2700	Male type	Virilised female	Boy	Positive	Positive
11	150	2550	Male type	Syndromic	Boy	Negative	Negative
12	6	3100	Female type	Insensitive to male hormones	Girl	Negative	Negative
13	14	3200	Male type	Hypospadias and undescended testes	Boy	Negative	Negative
14	365	3200	Female type	Adrenal hyperplasia	Girl	Positive	Positive
15	730	3100	Female type		Girl	Negative	Negative
16	830	2500	Female type	Adrenal hyperplasia	Girl	Positive	Positive
17	45	3000	Female type	Adrenal hyperplasia	Girl	Positive	Positive
18	27	2550	Male type	Hypospadias	Boy	Negative	Negative

Five babies with male type external genitalia had negative Barr bodies and drumstick chromosomes, suggesting a

male genotype. These babies are likely to be having hypospadias with

undescended testes with or without sex hormone imbalances.

One baby was identified as having few cells with clear Barr bodies and few cells with drumstick chromosomes. On examination the baby had male type external genitalia, but a uterus had been identified on ultra sound. This could be a case of mixed gonadal dysgenesis which is known to be the second most common cause.

Conclusions

Ambiguous genitalia appear to be detected at birth by health professionals. Congenital adrenal hyperplasia is the commonest cause for ambiguous genitalia in this sample. It can be stated that Barr body and drumstick chromosome analysis give good supportive evidence in deciding the sex of babies with ambiguous genitalia. However karyotyping and chromosome analysis is essential for definitive diagnosis.

References

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