

LETTERS TO THE EDITOR

The usefulness of detection of sex chromatin in babies with uncertain sex in resource limited settings

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Ambiguous genitalia in a new born baby is a major problem to the health care worker as well as the family of the affected (1). 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 (1, 2). The gold standard for such cases in karyotyping, and this may pose a problem since access to chromosomal analysis is not readily available to all institutions. Identification of sex chromatin is a simple first line investigation that can be used in resource limited settings. The term 'sex chromatin' is used to describe two structures namely the Barr body, present in epithelial and other tissue cells and the drumstick chromosome of the polymorphonuclear leucocytes (3,4,5). The Barr body was described by Dr. Murray Barr in 1949 and is produced by the inactivation of the X chromosome as explained in the Lyon hypothesis (3, 6). Drumstick chromosome is also produced by the same inactivation mechanism but is seen in 3 out of 100 circulating neutrophils in the blood of normal females (4). The Barr body examination in a buccal smear and identifying the drumstick chromosome in a peripheral blood smear are relatively simple and inexpensive tests and can give the clinician an idea

about the chromosomal sex of the baby when karyotyping is unavailable.

A sample of 18 babies with ambiguous genitalia referred to the department of Anatomy University of Peradeniya, for detection of sex chromatin from January 2006 to August 2009 was examined. Babies were examined for phenotypic characteristics of their genitalia and categorized into male type or female type according to the Prader staging system characteristics (7). Size of the clitoris or phallus and labioscrotal separation was taken as deciding factors.

A smear from the buccal mucosa was obtained and stained with basic fuchsin and a drop of blood obtained by heel prick was stained with Giemsa. Prepared smears were examined under light microscopy for the presence of Barr bodies and the drumstick chromosome.

Babies referred were between the ages of 5 days and 2 years with the majority being less than 1 month of age (72%).

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.

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Eleven of the babies had female type external genitalia. Nine of these babies were positive for Barr bodies and the drumstick chromosome. Seven of these had a probable diagnosis of adrenal hyperplasia after hormone essays, suggesting a female genotype with virilised female external genitalia. Congenital adrenal hyperplasia is known to be the commonest cause for ambiguous genitalia worldwide (8). One of these babies had multiple congenital anomalies consisting of ventricular septal defect, extra digits in the hand and abnormal palmar creases. This child was being investigated for a syndrome. The other child had normal hormone levels but a uterus had not been detected at ultrasound and was scheduled for further investigations.

Out of the babies with female type genitalia two babies were negative for Barr bodies and the drumstick chromosome. One of these babies could be having Turner's syndrome while the other who had male levels of testosterone could be insensitivity to male sex hormones but require further investigations for a definitive diagnosis.

Five babies with male type external genitalia had negative Barr bodies and drumstick chromosomes, suggesting a male genotype. Out of these none had palpable testes and ultrasound examination had not been able to detect the testes in the abdomen most probably due to the young age of the babies. These babies are likely to be having hypospadias with undescended testes with or without sex hormone imbalances. One of these babies also had

Downs syndrome with the typical facis and a atrial septal defect.

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 (9).

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 in resource limited settings where karyotyping maybe unavailable.

References

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