PRENATAL SEX DETERMINATION BY Y-FLUORESCENT BODY

by

NOOR AFSHAN SIDDIQI
ZAKIA ARHAD
and
S. MOHSIN

SUMMARY

The present study mainly aimed at prenatal sex determination of foetuses from percentage of Y-fluorescent body positive cells. In present study one hundred amniotic fluid cells were counted for Y-fluorescent body in each case. In cases of male foetuses 30-80% Y-body positive cells were seen. While in case of female foetuses, Y-fluorescent body was not seen in any case. Prenatal sex determination by Y-body method is rapid, practical and easy.

Knowledge of the foetal sex before delivery has aroused interest of laymen and scientists since time immemorial. The subject has received renewed interest because of availability of scientifically based methods for sex diagnosis and enhanced knowledge about a number of sex linked diseases e.g. Duchenne type of muscular dystrophy, haemophilia, etc. In these conditions determination of foetal sex in early pregnancy would be valuable in deciding whether to interrupt the pregnancy or not (Fuch, 1966; Abbo and Zellweger 1970).

Zech (1969) visualised Y-chromosome in interphase nuclei by fluorescent microscopy after staining with quinacrine dihydrochloride. Later, various scientists used this technique to identify the Y-chromosome in interphase nuclei of amniotic fluid for prediction of foetal sex in utero. The object of present study was to evaluate the Y-fluorescent body technique for determination of foetal sex.

Material and Method

Liquor amnii was studied in a total of 48 cases for prenatal sex determination by detection of Y-body, using quinacrine mustard.

Amniotic fluid was collected either by transabdominal route (abdominal amniocentesis or at the time of caesarean section) or by vaginal route (aspiration through bulging membranes or through cervical os).

Techniques: About 5-10 ml of amniotic fluid was centrifuged for 10 minutes at 2000-2500 r.p.m. Cell pellets were obtained and resuspended in 2 ml. of fixative (methanol and glacial acetic acid in 3:1 ratio) for at least two hours. Then samples were recentrifuged for 10 minutes

From Department of Obstet. & Gynaecology, J.N. Medical College, Aligarh Muslim University, Aligarh-202 001.

Accepted for publication on 14-3-1983.

at 2000 to 2500 r.p.m. Fixed cell sediments were resuspended in fixative. A slide was made from 2-3 drops of this suspension and heat dried.

Slides were stained with 0.5% quinacrine mustard for 25 minutes, later dipped in distilled water for few seconds, then in buffer solution of pH 5.6 for 5 minutes. The slides were seen under fluorescent microscope through 10 x and 100 x oil immersion objectives at 365 U.V. light with barrier filter 0/50/0. The slides were seen within two hours of preparation. One hundred cells were scored from each case. The cells were labelled as positive for Y-body if a single bright spot was seen (Fig. 1) and negative if no such fluorescent body was found.

Observation and Discussion

Forty-eight cases were studied by the Y-fluorescent body method, of which 23 were predicted as males and 25 as female foetuses. Apparent sex at birth was male in 24 cases and female in 24 cases. One male foetus was wrongly predicted as female, while rest of the predictions were correct. Total percentage of correct prediction was 97.8, comparable to results of Kapsoon *et al* (1976). Prediction in cases of male foetuses was 95.8% accurate, and cent per cent in cases of female foetuses (Table I).

In cases of female foetuses, Y-body positive cells were not found at all, while in male foetuses 30-80% liquor amnii cells were positive for Y-body. The variation in values in male foetuses may be due to the presence of different types of cells in amniotic fluid (Khudr and Benirechke 1971).

The above finding is comparable to that of some other authors (Table II).

Other authors have found fluorescent body in female foetuses also, though low in number (Table III). They suggested

TABLE I
Prediction of Sex

Total No. of cases	Phenotypic Sex	Y-Fluorescent		No. of cases with		% of correct prediction
		+ve	—ve	Correct diagnosis	Wrong diagnosis	
48	24 Male	23	1	23	1	95.8
	24 Female	—	24	24	—	100.0
Total correct prediction 97.9 per cent						

TABLE II
Range of Y-body

Author's name	Year	Range of Y-body		% of accuracy
		Male	Female	
Cervenka et al.	1971	68-87	Zero	100
Rook and Hsu	1971	3-9	Zero	100
Khudr et al	1971	25-50	Zero	100
Aggarwal et al.	1976	40-60	Zero	99
Peter and Amber	1977	60-00	Zero	Not given
Present study	1982	30-80	Zero	97.8

TABLE III

Author's name	Year	Range of Y-body		% of accuracy
		Male	Female	
Fergusson-Smith et al.	1971	30-80	0-10	100
Valenti et al.	1972	22-89	0-9	94
Nelson	1973	Above 20 weeks		
		59.7	2.9	91
		Below 20 weeks		
		56.1	2.6	90
Joseph et al.	1975	65-95	0-4	100
Kapsoon et al	1976	50-100	20	99

that small proportion of cells in female nuclei might have fluorescence of X-chromatin, especially D group and chromosome number 3. They could be wrongly predicted as Y-body.

In the present study, there was complete correlation between predicted female foetal sex and phenotypic sex at birth while there was one wrong prediction in cases of male foetuses. That male foetus was wrongly predicted as female as no fluorescent body was seen. It might have been absent due to presence of an unusually small Y-chromosome. An unusually small Y-chromosome is due to deletion of the distal heterochromatic segment of Y-chromosome and probably it has no demonstrable phenotypic effect (Court-Brown *et al* 1966). The failure of fluorescence due to small and deleted Y-chromosome did present a problem. To alleviate this problem, Y-body determination should be conducted upon the buccal smear of the father for excluding hereditary non-fluorescing Y-chromosome. In our false negative cases, it was not possible to study the buccal smear of the father for Y-body.

References

1. Abbo, G., Zellweger, H.: Lancet, **1**: 216, 1970.
2. Aggarwal, S., Devi, P. K., Mahajan, R. C.: J. Obstet. Gynaec. India. **26**: 249, 1976.
3. Cervenka, J., Gorlin, R. J., Bendel, R. P.: Obstet. Gynec. **37**: 912, 1971.
4. Court-Brown: Eugen. Lab. Memoirs (London) XL **11**: 91, 1966.
5. Ferguson-Smith, M. E. and Ferguson-Smith, M. A.: Brit. Med. **4**: 69, 1971.
6. Fuchs, F.: Clin. Obstet. Gynaec. **9**: 565, 1966.
7. Hamilton, S.: Aggarwal, N. Hingorani: J. Obstet. Gynaec. India. **26**: 848, 1976. 848, 1976.
8. Joseph, R., Verma, I. C., Verma, K.: Indian Paed. 12 (ii): 1063, 1975.
9. Kapsoon, J. U., James, F., Facog, E.: Obstet. Gynec. **42**: 287, 1976.
10. Khudr, G. and Benirechke, K.: Am. J. Obstet. Gynec. **110**: 1091, 1971.
11. Nelson, M. M.: Clin. Genetics, **4**: 252, 1973.
12. Peter, J. and Amber, L.: Indian Pediatric, **14(4)**: 259, 1977.
13. Rook, A. and Hsu, L. Y.: Nature, **280**: 53, 1971.
14. Valenti, C., Linn, C. C, and Baum, A.: Am. J. Obstet. Gynec. **112**: 89, 1972.
15. Zech, C.: Exp. Cell. Res. **58**: 463, 1969.

See Fig. on Art Paper II