CYTOGENETIC INVESTIGATIONS IN PATIENTS WITH CONGENITAL ABSENCE OF UTERUS

by

D. KRISHNA KUMARI
C. KUSUMA KUMARI

and

P. P. REDDY

SUMMARY

Chromosome investigations were done in 23 cases of primary amenorrhoea who were diagnosed with absence of uterus by ultrasonographic scanning. Of the 23 subjects, 16 showed a 46, XX complement, the remaining 7 showed abnormal chromosome complements. One subject had testicular feminization and the other 6 had gonadal dysgenesis with different chromosome complements. We conclude that there is an unexpectedly high frequency of aberrations in patients without a uterus.

Introduction

Congenital absence of uterus with or without ovarian agenesis is one of the causes of primary amenorrhea. The other causes include congenital malformations, metabolic derangements, selective anterior pituitory failure, genetic defects and other malignant tumors. The purpose of the present paper is to report chromosome investigations in 23 cases of primary amenorrhea with congenital absence of uterus.

Material and Methods

Twenty-three individuals having primary amenorrhea who showed absencs of uterus on ultrasonic scanning were selected for the study. Ultrasonographic scanning was done with Gray scale compound contact scanner (Sonograph EPTM, (Unirad CZD Model 849) using 3.5 m Hz transducer. Chromosome analyses were carried out on metaphase figures obtained after a modified technique of Moorhead *et al* (1960). The slides were stained according to Seabright (1971) in order to facilitate individual chromosome identification. Confirmatory staining methods like 'Q' bending (1960), 'C' banding (1974) and late replication of X chromosome (1981) were carried out in selective cases to confirm the nature of anomaly. X chromatin analysis was a routine procedure in all the patients.

Observations

Table I shows the chromosome findings

TABLE I

Cytogenetic Findings in Patients with Absent Uterus

Chromosome constitution	No. of cases	Percentage
46, XX	16	69.56
45, X	4	17.39
46, XY	1	4.35
46, XX/47, XXY	1	4.35
45, X/46, X, frag	1	4.35

From: Institute of Genetics, Begumpet, Hyderabad-500 016.

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TABLE II

Clinical Details of the Patients With Chromosome Abnormalities

Case Regn. No.	Age (Yrs.)	Com-plaint	Height (cms)	Secondary sex characters	Breast development	Vagina	X-chro-matin	Karyotype
970	30	P.A.	137.2	Absent	—	+	—	45, X
3950	17	P.A.	137.5	Absent	—	+	—	45, X/46, X, frag
1048	19	P.A.	138.2	Absent	Slight	+	+	46, XX/47, XXY
1569	18	P.A.	134.0	Absent	—	+	—	45, X
2317	18	P.A.	158.5	Normal	—	+	—	46, XY
1059	15	P.A.	132.5	Absent	—	+	—	45, X
3296	20	P.A.	135.0	Poor	—	+	—	45, X

P.A. = Primary amenorrhea.

of the 23 cases with absence of uterus. Seven (30.43%) cases were found to have chromosome abnormalities. Remaining 16 (69.56%) had normal XX karyotype. Table II shows the clinical details of the patients with chromosome abnormalities.

Conclusions

There are no reports in the literature on cytogenetic investigations in patients with congenital absence of uterus. Azoury and Zones (1966) have detected normal XX chromosome constitution in patients with congenital absence of vagina. In our investigations, we have found that 16 cases had normal XX karyotype, and the remaining 7 cases showed abnormal chromosome pattern which include: 45, X Turner syndrome in four cases, 46, XY testicular feminization syndrome in one case, 46, XY|47, XXY gonadal dysgenesis in one case, and 45, X|46, X, fragment Turner syndrome variant in one case.

Congenital absence of uterus can occur as an isolated embryological defect. If it is associated with chromosome abnormality, screening of family members, especially parents and siblings helps for genetic counselling.

References

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