X Chromosome number and mental health status in Sri Lanka

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Summary

The X chromosome number of 143 undergraduates, 104 children from the Preethipura Home for the mentally handicapped and 484 admissions to the Psychiatric Ward of the General Hospital, Kandy has been assessed by the buccal smear method. No discordance with sex was observed for the undergraduates and the mentally handicapped children. In the psychiatric patients the prevalence rates for numerical X chromosomal aberrations was 0.005 for males and 0.01 for females.

Key words: Buccal smear, X chromosome number, mentally handicapped, psychiatric patients

Introduction

The study of X chromatin bodies in oral mucosal cells in buccal smears is a reliable method of predicting the number of X chromosomes in humans (1). X chromatin surveys in the West have shown that approximately 0.05% of all new borns have numerical aberrations of the X chromosomes (2, 3). The prevalence of these aberrations has been reported to be higher in inmates of institutions for the mentally defectives (4) and mental hospitals, also in the West (5). Neither the incidence at birth nor the prevalence of numerical X chromosomal aberrations in mental defectives or psychiatric patients is known for Sri Lanka. This paper is based on a study of X chromatin in buccal smears in three groups comprising both genders differing in mental health status.

Populations and Method

The three groups studied consisted of

i. undergraduates at the Faculty of Medicine, Peradeniya (n = 143);

ii. children of Preethipura Home for the mentally handicapped, Hendala (n = 104);

iii. patients of the Psychiatric Ward, General Hospital, Kandy (n = 484).

A modification of the method of Barr was used (6). The subjects were requested to rinse their mouths. The buccal smears were made by firmly scraping the mucosa of the cheek with a metal spatula. The scraping was smeared gently on a clean albuminised slide and fixed immediately in freshly prepared fixative consisting of equal volumes of ether and ethyl alcohol. Each slide was identified by a code number only. Permanent preparations were made after staining them with carbol fuchsin. Fixation time varied from 1/2 hr to 48 hrs. In reading the slides 100 large cells with smooth unbroken nuclear membranes were scored for the number of X chromatin bodies. Larger than normal or smaller than normal X chromatin bodies were also looked for. Using this method an X chromatin positive count of 20 ± 10 was observed in buccal smears of the majority of normal females. Smears with less than 2 chromatin bodies in 100 cells were scored as X chromatin negative. Stained rod-shaped bacteria made scoring difficult in some smears. A certain proportion of slides could not be read due to bad preparation.

In the case of the undergraduates the buccal smears were made during practical classes for the demonstration of X chromatin. No slides were discarded from this group. The slides of the mentally defective children at Preethipura were made during two visits to Hendala in February and October 1969. At the first visit slides from the 54 younger children, both boys and girls, were prepared. Those from the 50 older boys were made on the second visit.

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Eleven out of these 104 slides (approximately 10%) had to be discarded. The slides from the admissions to the psychiatric ward of the General Hospital were made by visiting the ward twice a week from February 1969 to June 1970 on clinic days. Out of a total of 550 admissions for this period 484 were sampled. Thirty four slides (approx 7%) had to be discarded from this group.

Results

Fig. I shows an X chromatin positive nucleus as demonstrated by this method. No buccal smears with smaller than normal or larger than normal X chromatin bodies were observed. Table I gives the results for the males in the three groups studied. In the undergraduate and mentally defective groups no discordance of X chromatin with sex was observed. In the admissions to the psychiatric ward one male had 18 X chromatin positive cells in the 100 counted.

Table II gives the results for the females in the three groups studied. In the undergraduate and mentally defective groups no discordance of X chromatin with sex was observed. In the admissions to the psychiatric ward one female had no X chromatin positive cells and another had 10 cells with two X chromatin bodies in the 100 counted. There was no follow up with karyotyping due to financial constraints in this study. However, it has been demonstrated that X chromatin positive males were XXY and X chromatin negative females XO, when X chromatin studies were followed up with karyotyping (2). Therefore the presumptive chromosomal sex of the discordant individuals in this study has been reported as XXY for the male and XO for the female.

Discussion

Undergraduates form a group highly selected for mental fitness. It was not surprising

Table I – The X chromatin status and the presumptive chromosomal sex in males in each group

Group	Number	X Chromatin		Presumptive Chromosomal sex
		Negative	Positive	
Undergraduates	86	86	0	XX
Mentally defective children	68	68	0	XY
Psychiatric patients	233	232	–	XY
			1	XXY

Table II – The X chromatin status and the presumptive chromosomal sex in females in each group

Group	Number	X Chromatin		Double Positive	Presumptive Chromosomal sex
		Negative	Positive		
Undergraduates	57	0	57	0	XX
Mentally defective children	25	0	25	0	XX
Psychiatric patients	217	0	215	0	XX
		1	–	–	XO
		–	–	1	XXX

therefore that no numerical abnormalities of the X chromosome were detected in them.

In studies in the West a higher prevalence of numerical chromosomal aberrations has been reported for high grade mental defectives with intelligence quotients (IQ's) around 75(7) but not from the low grade mental defectives with IQ's around 25(8). No diagnosis cards or IQ assessments were available for the mentally handicapped at Preethipura Home. There was a marked difference between the younger children, ages ranging from 3 – 9 years, and the older boys, ages ranging from 10 – 22 years. The younger children had been admitted to the Home as the parents were unable to provide the special care necessary when looking after a low grade mentally defective at home. Around 75% of them had to be fed, washed and clothed and could not be toilet trained. Around 25% of the older boys on the other hand were helping out with the cleaning of the premises, in the preparation of food and in gardening. They appeared mentally normal. Buccal smears of all the 104 inmates were made but only 93 of these could be read. Therefore the small sample size and the inclusion of low grade mental defectives and mentally normal children could contribute to the failure to observe numerical X chromosomal aberrations in the inmates at Preethipura.

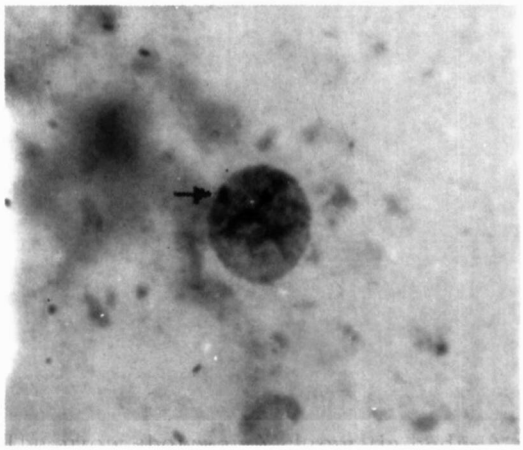
The inmates of mental hospitals on whom studies are available from the West were long-standing chronic patients. The prevalence of XXY males and XXX females were four to five times greater in the mentally retarded and psychotic than their background incidence in the newborn population. In contrast XO women were not found with increased frequency (9). Generalising from the findings of these studies, XXY men were stereotyped, mentally retarded and XXX women as psychotic. XO women on the other hand were expected to have little difficulty in their personal adjustment. However, longitudinal psychological studies on individuals detected at birth have demonstrated mental dysfunction and maladjustment in all three categories (10).

In the present study one XXY in 233 males and one XO and one XXX in 215 females were detected among admissions to a psychiatric ward in Sri Lanka, giving a prevalence rate of 0.005 for males and 0.01 for females. As admissions to this ward were mostly cases of recent acute episodes of mental illness, no comparisons can be made with studies from the West. It is probable that the low prevalence rates may reflect the low incidence rate of X chromosomal aberrations in this part of the world. In a study from India no sex chromatin discordance was found among 2,058 male and 1832 female consecutive live births (11). These results have been omitted when pooling results from the West "because it was made in India and it is not known whether there is a difference in frequency of sex chromosomal abnormalities in various racial groups" (5).

It is regrettable that no other studies on the incidence of numerical sex chromosomal aberrations in this part of the world have been reported. A concerted effort must be made to establish the incidence of sex chromosomal aberrations, and their prevalence in high grade mental defectives and inmates of mental hospitals in Sri Lanka.

The results of this study were communicated as part of "Sex chromatin studies in population groups" (Proceedings of the Ceylon Association for the Advancement of Science, 1971; p. 22).

Fig. 1



The arrow shows an X chromatin body in a nucleus of a mucosal cell in a buccal smear.

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