

LENGTH OF THE Y CHROMOSOME IN A GENERAL MALE POPULATION

EVA ZEUTHEN, JOHANNES NIELSEN

SUMMARY

Measurement of the Y chromosome has been made in 508 males examined for military service. There was a normal Gaussian distribution of the length of the Y chromosome.

The frequency of mentally retarded males was significantly higher than expected among those with a comparatively large Y ($Y/F \geq 1.00$). It is concluded that there may be an association between the length of the Y chromosome and certain types of mental retardation.

A number of studies have indicated that increase in length of the Y chromosome may effect behaviour and personality development (Nielsen 1968, Kahn et al. 1969, Nordland 1969, Nielsen 1971, Nielsen and Henriksen 1972, Nielsen and Friedrich 1972, Friedrich and Nielsen 1973, Christensen and Nielsen 1973).

The present study of the length of the Y chromosome in relation to conscription, education, and intelligence level, is part of a prevalence investigation of chromosome aberrations in a Danish population sample from males examined for military service.

MATERIAL AND METHODS

Examination for military service is compulsory in Denmark. All males are registered for this examination, and all but severely handicapped persons are required to be examined.

The present study comprises a representative sample of all males registered to be examined first time for military service, during autumn 1969 and spring 1970, in one (No. IV) of the seven conscription areas of Denmark. Area No. IV comprises the mid-east part of Jutland centered around the second largest city of Denmark (Århus). The population in this area, which was 781 573 in 1965, can be considered representative of the Danish population outside Copenhagen. The sample comprised 3 840 males. Chromosome examination was made in males with a stature of 181 cm or above, males with testes ≤ 12 ml, measured with Prader's orchidometer (Prader 1966), as well as all males required to be examined for military service on account of severe physical or mental disability: these groups comprised 1 115 males, 20 of whom refused to have a blood sample taken.

The length of the F chromosome is stable, but the length of the Y chromosome is variable, and several studies have shown that the distribution of the Y chromosomes according to length in the male population is a normal Gaussian distribution (Unnérus et al. 1967, Lins and Sundequist 1971, Nielsen and Friedrich 1972). That the length of the Y chromosome is stable and inheritable has also been shown in several family studies (among others, by Chapelle et al. 1963, Borgaonkar et al. 1969, El Alfi 1970, Genest et al. 1970).

Measurements of the Y and the F chromosomes and calculation of a Y/F index give a reliable estimate of the length of the Y chromosome. The length of the Y chromosome has been measured in relation to several other chromosomes such as No. 2, 17-18, and 21-22; but Cohen et al. (1966) tested different methods and concluded that the most reliable one was to compare the length of the Y chromosome with the length of the F chromosome, and Court Brown et al. (1966) used the Y/F measurements in their studies of chromosome aberrations in the general population.

We have measured the Y and the F chromosomes according to the method described by Nielsen and Friedrich (1972) in all males who had a chromosome examination made in the population sample examined during the first half part of the study, i.e., autumn 1969, comprising a total of 508 males.

Educational level is registered at the examination for military service in nine groups, and intelligence is recorded by a group test with a classification in nine groups.

RESULTS

The mean Y/F ratio was 0.88 ± 0.003 , the median was 0.87, and the range was 0.73-1.11, the 95% confidence limits was 0.87-0.89, and there was a normal Gaussian distribution as seen in Fig. 1.

We found no association between the intelligence-educational level among those tested and length of the Y chromosome, but there was a significantly higher frequency of mentally retarded males with $Y/F \geq 1.00$ (24%), compared with those having a $Y/F < 0.90$ (5.0%) ($\chi^2 = 14.021$, $P < 0.001$: Table I), and this difference was also significant when comparing males with $Y/F \geq 0.90$ (12.8%) with those having $Y/F < 0.90$ (5.0%) ($\chi^2 = 9.690$, $P < 0.005$).

The frequency of males conscripted for military service at the examination was lower

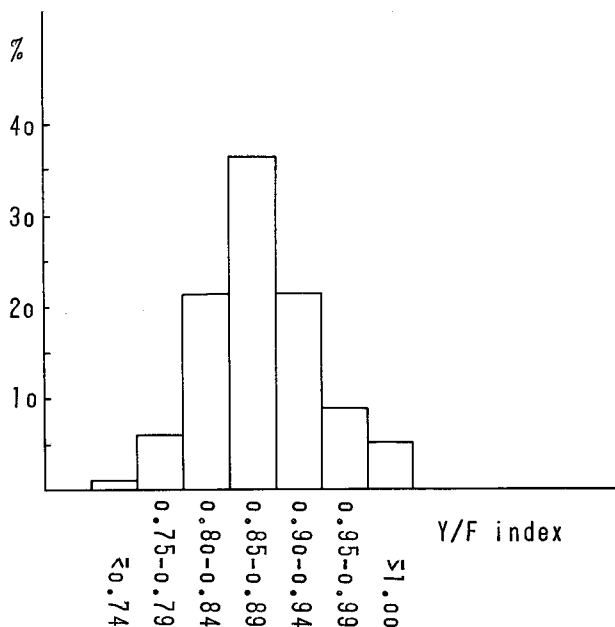


FIG. 1. Distribution of Y/F index for the 508 males.

TABLE I
INTELLIGENCE-EDUCATIONAL LEVEL AND LENGTH OF THE Y CHROMOSOME

Intelligence and educational level	Y/F index						Statistically significant difference
	< 0.90		0.90-0.99		≥ 1.00		
	Total	%	Total	%	Total	%	
Mentally retarded	16	5.0	17	11.0	6	24.0	$\chi^2 = 14.021, P < 0.001$
1-9	304	95.0	138	89.0	19	76.0	
Total	320	100.0	155	100.0	25	100.0	

The 8 physically handicapped males rejected without examination are not included.

TABLE II
EXAMINATION FOR MILITARY SERVICE AND LENGTH OF THE Y CHROMOSOME

Results of examination for military service	Y/F index					
	< 0.90		0.90-0.99		≥ 1.00	
	Total	%	Total	%	Total	%
Conscripted	230	74.2	103	73.6	12	63.1
Rejected	50	16.1	24	17.1	4	21.1
Postponed	30	9.7	13	9.3	3	15.8
Total	310	100.0	140	100.0	19	100.0

The 39 mentally retarded males rejected without examination are not included.

among males with $Y/F \geq 1.00$ (63.1%) than among those with $Y/F < 1.00$ (74.0%); the difference is, however, not statistically significant (Table II). Criminality was no more frequent among males with $Y/F \geq 1.00$ (4.2%) than among those with $Y/F \leq 0.99$ (5.0%).

DISCUSSION

The distribution of the Y/F index in the present randomly selected population of males with a stature ≥ 181 cm or testes ≤ 12 ml, was similar to that of a 10% randomly selected sample of newborn boys (Nielsen and Friedrich 1972). The mean Y/F ratio in this study was, however, somewhat higher (0.88 ± 0.003), compared with that of the newborn boys (0.84 ± 0.004). The difference is, however, not significant.

The distribution was a normal Gaussian, as seen in Fig. 1, which correlates with what has been found in other population studies (Unnérus et al. 1967, Torre and Giménez-Martin 1970, Lins and Sundquist 1971, Nielsen and Friedrich 1972).

The finding of an increase in the frequency of mentally retarded males from 5.0% in the Y/F group < 0.90 , to 11.0% in the Y/F group 0.90-0.99, to 24.0% in the Y/F group ≥ 1.00 ($P < 0.001$, as referred to the increase from 5% to 24%), indicates that there may be an association between the length of the Y and mental retardation.

We found otherwise no association between the length of the Y and intelligence-educational level, nor was there any association between length of the Y and criminality.

The frequency of males conscripted for military service was higher among those with a Y/F < 0.90 (74.2%) than among those with a Y/F ≥ 1.00 (63.1%), but the difference is not significant.

Increase of the Y chromosome above a certain length is most probably due to duplication of part of the strongly fluorescent material of the distal part of the long arms Y, as indicated from studies by Sperling and Lackmann 1971, Wahlström 1971, Knuutila and Gripenberg 1972, and Tishler et al. 1972.

There are strong indications that genes concerning the male gonadal development are located in the short arms of the Y chromosome (Fraccaro et al. 1966, Jacobs and Ross 1966, Nielsen et al. 1972). Genes in the strongly fluorescent part of the long arms of the Y may mainly have influence on mental function, most probably mainly concerning personality function, but also to a certain extent concerning certain factors in the intellectual function.

REFERENCES

- Borgaonkar D.S., McKusick V.A., Herr H.M., De Los Cobos L., Yoder O.C. 1969. Constancy of the length of human Y chromosome. *Ann. Genet. (Paris)*, 12: 262.
- Chapelle A. de la, Hortling H., Edgren J., Riainen R.K. 1963. Evidence for the existence of heritable large Y chromosomes unassociated with developmental disorder. A cytogenetical and clinical study of 4 males with hypogonadism, one with mongolism and their relatives. *Hereditas*, 50: 351.
- Christensen K.R., Nielsen J. 1973. Incidence of chromosome aberrations in a child psychiatric hospital. *J. Clin. Genet.* (In press).
- Cohen M.M., Shaw M.W., MacCluer J.W. 1966. Racial differences in the length of the human Y chromosome. *Cytogenetics*, 5: 34.
- Court Brown W.M., Buckton K.E., Jacobs P.A., Tough I.M., Kuenssberg E.V., Knox J.D.E. 1966. *Chromosome Studies on Adults*. Cambridge University Press.
- El-Alfi O.S. 1970. A family with a large Y chromosome. *J. Med. Genet.*, 7: 37.
- Fraccaro M., Lindsten J., Klinger H.P., Tiepolo L., Bergstrand C.G., Herrlin K.M., Livaditis A., Pehrson M., Tillinger K.G. 1966. Cytogenetical and clinical investigations in four subjects with anomalies of sexual development. *Ann. Hum. Genet.*, 29: 281.

- Friedrich U., Nielsen J. 1973. Chromosome studies in 5049 consecutive newborn children. *Clin. Genet.*, 4: 333.
- Genest P., Laberge C., Poty J., Gagné R., Bouchard M. 1970. Transmission d'un petit « Y » durant onze générations dans une lignée familiale. *Ann. Genet. (Paris)*, 13: 233.
- Jacobs P.A., Ross A. 1966. Structural abnormalities of the Y chromosome in man. *Nature (Lond.)*, 210: 352.
- Kahn J., Carter W.I., Dernley N., Slater E.T.O. 1969. Chromosome studies in remand home and prison populations. In West D.J. (ed.): *Criminological Implications of Chromosome Abnormalities*. Cambridge: Institute of Criminology.
- Knuutila S., Gripenberg U. 1972. The fluorescence pattern of a human Yq+ chromosome. *Hereditas*, 70: 307.
- Lins L.-E., Sundequist U. 1971. Y/E index in a Swedish population. *Acta Genet. Med. Gemellol. (Roma)*, 20: 211.
- Nielsen J. 1968. Y chromosomes in male psychiatric patients above 180 cm tall. *Br. J. Psychiatry*, 114: 1589.
- Nielsen J. 1971. Prevalence and 2½ years incidence of chromosome abnormalities among all males in a forensic psychiatric clinic. *Br. J. Psychiatry*, 119: 503.
- Nielsen J., Friedrich U. 1972. Length of the Y chromosome in criminal males. *Clin. Genet.*, 3: 281.
- Nielsen J., Henriksen F. 1972. Incidence of chromosome aberrations among males in a Danish youth prison. *Acta Psychiatr. Scand.*, 48: 87.
- Nielsen J., Friedrich U., Christensen A.L., Godt H.H., Strömberg L.S. 1972. A phenotypic male with karyotype 45,X/45,X,ace+(?Yq—). *Humangenetik*, 15: 319.
- Nordland E. 1969. En undersøkelse av gutter med problematferd og stort Y-kromosom. *Nordisk Tidsskrift for Kriminalvidenskab*, 57: 74.
- Prader A. 1966. Testicular size: assessment and clinical importance. *Triangle*, 7: 240.
- Sperling K., Lackmann I. 1971. Large human Y chromosome with two fluorescent bands. *Clin. Genet.*, 2: 352.
- Tishler P.V., Lamborot-Manzur M., Atkins L. 1972. Polymorphism of the human Y chromosome: fluorescence microscopic studies on the sites of morphologic variation. *Clin. Genet.*, 3: 116.
- Torre C. de la, Giménez-Martín G. 1970. The human Y chromosome in the Spanish population. *J. Genet. Hum.*, 18: 235.
- Unnérus V., Fellman J., de la Chapelle A. 1967. The length of the human Y chromosome. *Cytogenetics*, 6: 213.
- Wahlström J. 1971. Are variations in length of Y chromosome due to structural changes? *Hereditas*, 69: 125.

RIASSUNTO

È stata effettuata una misurazione del cromosoma Y in 508 maschi visitati per il servizio militare. La lunghezza del cromosoma ha presentato una distribuzione normale. La frequenza di ritardi mentali è risultata significativamente più elevata dell'atteso negli individui con un Y relativamente grande ($Y/F \geq 1.00$). Se ne conclude che potrebbe esservi un'associazione fra lunghezza del cromosoma Y e certi tipi di ritardo mentale.

RÉSUMÉ

Une mensuration du chromosome Y a été effectuée chez 508 mâles visités pour le service militaire. La longueur du chromosome a présenté une distribution normale. La fréquence d'arriérés mentaux était significativement plus élevée chez les individus avec un Y relativement grand ($Y/F \geq 1.00$). L'on en conclut qu'il pourrait y avoir une association entre longueur du chromosome Y et certains types de retard mental.

ZUSAMMENFASSUNG

Bei 508 jungen Männern, die für den Militärdienst untersucht wurden, wurde eine Messung des Y-Chromosoms vorgenommen. Die Länge des Chromosoms war dabei normal verteilt. Bei den Personen mit relativ grossem Y war das Vorkommen von geistig zurückgebliebenen Individuen bedeutend höher als zu erwarten wäre ($Y/F \geq 1.00$). Daraus wird geschlossen, dass vielleicht eine Beziehung zwischen der Länge des Y-Chromosoms und gewissen Typen geistiger Zurückgebliebenheit besteht.

Johannes Nielsen, M.D., Chief of Service, The Cytogenetic Laboratory, Århus State Hospital, DK-8240 Risskov, Denmark.