THE ANDROGYNY OF MONGOLS

By

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IT is now accepted that the underlying cause of mongolism is a trisomy of the 21st chromosome (Denver classification). This finding has given a new impetus to research into this condition. It is hoped by investigating these people to be able to find out what genetic information is carried on this extra chromosome, this would be of great interest to all students of human biology, as well as being of use in the more general field of human medicine. It is very unlikely that the extra chromosome in every case will carry the same genetic information, and this would account for the within species differences in the syndrome of mongolism.

This paper is concerned with one aspect of the growth and sexual development of the male mongol. It is a study of the "Androgyny Score" which is derived from a special formula expressing a relationship between shoulder width and bi-iliac diameter. This score is affected by many things and there is normally a significant difference between the score for adult males and females. (Tanner, 1951).

THE MATERIAL

Only male mongols have been studied. The mongols were compared with a control group of similar life age, the controls were drawn from non-mongol patients in the same hospital for the mentally handicapped. No cases of doubtful mongolism were included and the diagnosis of this condition was made on clinical grounds alone without any reference to chromosome status.

THE METHODS

Androgyny. The method of Tanner (1951) was used and conversion to an androgyny score was made by the formula given by the same author, but in the present survey no constant was used as a discriminant.

ANDROGYNY SCORE=3 × Biacromial diam. in cm. — Bi-iliac diam. in cm.

Height. This was measured in the standing erect posture as described in previous communications (Dutton, 1959a). Measurements were made in the metric system.

Sexual Development. Pubic hair and genital development was assessed according to the schedule described by Tanner (1955).

THE RESULTS

Sexual Development. It will be seen from Fig. 1 that there is no difference in the time of onset of puberty in the two groups. The final completion of sexual development is, however, often not completed in the mongols. This bears out observations made by others. (Rundle *et al.*, 1959). There is no difference between the pubic hair and genital development in the mongol group. Of the adult mongols aged 23 and over, 41 per cent. have reached stage 5 for genital development (adult development) and 59 per cent. have attained stage 5 for pubic hair development; in the adult controls 93 per cent. have reached stage 5 for both pubic hair and genital development. A life age of 11 was the earliest that signs of puberty were noted in the mongols, and for the control group the earliest

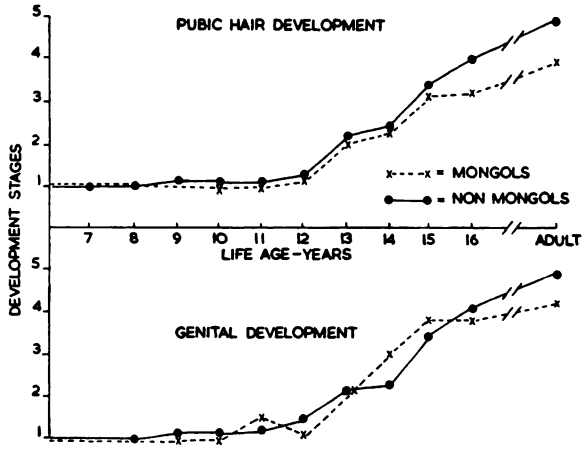

FIG. 1.—Sexual Development of Male Mongols and Controls.

age was 9 years, both these figures are within the normal range. By the age of 13 years all the control group were pubescent whilst 25 per cent. of 13-year-old mongol boys were still pre-pubescent.

Androgyny Score. When the mean androgyny score for each year of life age is compared in the two groups, it will be seen that there is little to choose between the mongols and the control group up to the age of 14, and then they diverge and by the time adult years are reached, the difference between the mean androgyny scores for the two groups is significantly different.

Table I gives the means for the two groups. Applying the "t" test, "t"= 3·0663 which shows that the difference between the means is significantly different at P=0·05.

TABLE I

Mean Androgyny Scores in Adult Mentally Handicapped Males

	Mean	S.D.
Mongols	78·8	6·5
Non-Mongols ..	85·13	4·6

Table II shows the means for different life ages and Figure 2 shows a graphical representation of this data.

TABLE II

Life age, years	6	7	8	9	10	11	12	13	14	15	16	Adult
MONGOL:												
Number	2	1	2	2	4	7	10	8	5	4	5	18
Mean androgyny score	51·0	48·4	51·1	58·0	57·6	57·4	62·0	64·3	69·7	71·8	70·5	78·8
NON-MONGOL:												
Number	3	4	4	5	6	6	11	10	15	14	15	20
Mean androgyny score	54·9	55·1	59·8	51·4	58·6	59·5	64·1	65·6	69·3	73·5	80·5	85·2

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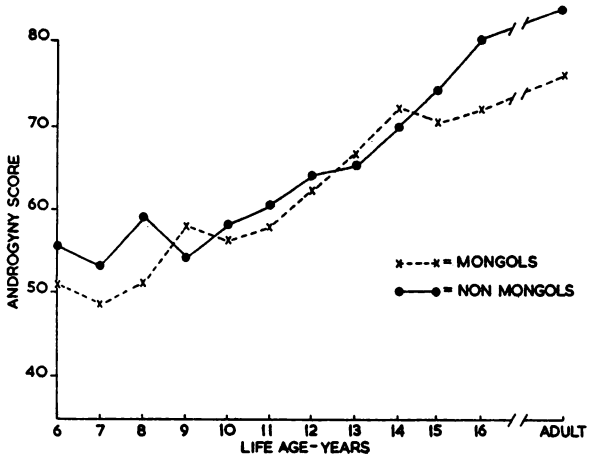

FIG. 2.—Androgyny Score *v.* Age in Male Mongols and other Subnormal Male Controls.§

Since there is a positive correlation between age and height, it was felt that as mongols are shorter than normal (Dutton, 1959a) it might be that the difference in androgyny scores would be removed if androgyny was compared directly with height rather than age. It is shown that there is a positive correlation between height and androgyny score in both groups, but the linear correlation is better in the control group than in the mongols. For male mongols "r"=0·596 and for the male controls "r"=0·622.

The linear regression equations for the two groups are:

MONGOLS:

Androgyny=0·436 × Height in cm. + 7·6.

NON-MONGOL CONTROLS:

Androgyny = 0·35 × Height in cm. + 20·3.

The lines of regression are shown in Figure 3.

The difference between the correlation coefficients for mongols and controls is statistically significant. The standard errors of the regression coefficients are: Mongols = ± 0·355 and for non-mongol controls = ±0·120.

DISCUSSION

Tanner (1951) found that the androgyny score had a normal distribution with means and standard deviations of: Males 90·1±4·7 and females 78·9± 4·6. He also found that the formula only misclassified 12 per cent. of the 490 men and women in his survey. The figures obtained in the present survey for the controls are just more than 1 standard deviation below Tanner's mean, the mongol group on the other hand are more than 2 standard deviations below Tanner's mean for men. The mongol mean is actually the same as the mean given by Tanner for females. The standard deviation of the controls in the present series was 4·6 as compared with Tanner's 4·7, but there is greater scatter amongst the mongol men who have a standard deviation of 6·5. The androgyny score is considered to be a discriminant function and enables the most effective differentiation between the sexes to be made using the formula employed here.

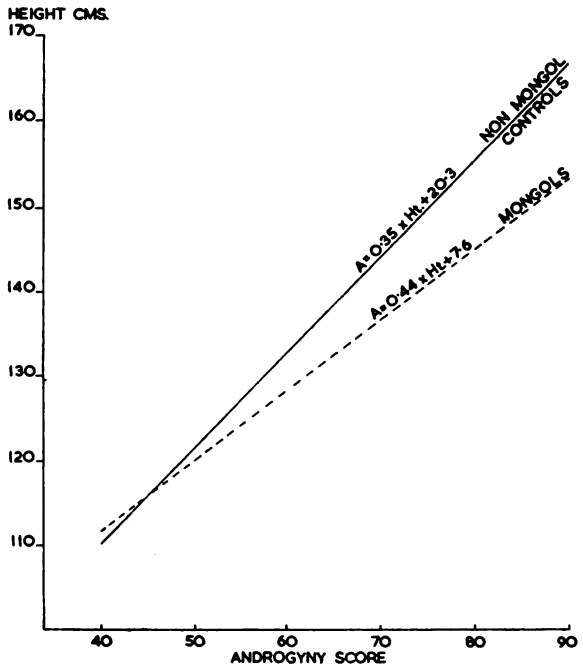

FIG. 3.—Height *v.* Androgyny Score in Male Mongols and Controls.

Ferriman *et al.* (1957) found a significantly wider bi-acromial diameter, and so a higher androgyny score, in women with hirsutes and oligomenorrhea. Lindegard (1956) found a significant correlation between the size of the penis and the androgyny score. Coppen (1958) found a more masculine androgyny score in women with toxaemia of pregnancy, and there is some evidence that there is a correlation between androgyny and psychosexual development. Coppen and Cowie (1960) found a raised androgyny score in the mothers who had given birth to a mongol at the age of 27 years or younger. Rundle *et al.* (1961) confirmed the work of others that in mothers who had given birth to a mongol child early in their reproductive career, there is a raised androgyny score, these young mothers (27 or under) of mongol children also had an increased output of dehydroepiandrosterone, as compared with mothers aged 28 and over who had given birth to mongol children.

The difference between androgyny in men and women is said to be due to changes occurring at puberty under the influence of the sex hormones, the bi-acromial diameter increasing as a direct effect of androgen production. Rundle, Dutton and Gibson (1959) investigated the testicular function of mongols from the point of view of androgen production, and showed that the mongol testis appears to produce androgens and undergoes the same pubertal spurt in excretion as did a control group, furthermore they showed that there was no difference in the excretion rates of individual ketosteroids. The mean daily excretion of 17-kerosteroids in mongol adults was found to be lower than the control group, but higher than in female mongols. Dutton (1959) has shown that amongst mongol boys there is no difference in the total daily excretion of 17-keto and ketogenic steroids.

The sexual development of the present group of mongol boys confirms the finding stated earlier (Dutton, 1959a) that although there was considerable development of the genitalia and pubic hair, often there was arrest before complete development had been achieved; this latter finding was also shown to be a fact by Rundle *et al.* (1959) when they were considering the development of adult male mongols in relation to androgen production. The present series seems to point out that puberty starts at around the right time, but in the majority of cases full sexual development is never achieved by the time adult age is reached. This timing of the onset of puberty is also in agreement with the steroid excretion studies on mongol boys (Dutton, 1959b) and in mongol boys and adults (Rundle *et al.*, 1959).

Benda (1947) states that the sexual development is retarded and maintains that 50 per cent. of male mongols have undescended testicles at birth and never will descend. This is not the finding of Rundle and Dutton (1962), in their series of 23 male mongols aged 16 to 23 years they found one case of bilaterally undescended testicles, and 3 with unilateral undescended testicle. They also reported that 7 had both inguinal canals closed, and 15 with one or more canals open. Benda also says that the penis is mostly infantile and short, but may sometimes be long and thin. In the present survey this was not confirmed: often the penis grew normally in length, but there was poor development of the glans. Benda also considers that the pubic hair is long and silky, and that the distribution is female. In the present series the distribution was not found to be typically female, but the pubic hair was found to be sparser than normal. The most remarkable thing is that the mongol's pubic hair has a marked lack of curl and is very straight; this may account for the apparent sparseness.

The androgyny score cannot therefore be due solely to androgen excretion, for there is in the male mongol a female androgyny score, but a male pattern of androgen excretion, and one must look for other factors controlling this aspect of sex differentiation. Rey and Coppen (1959) have reviewed some of the factors controlling the androgyny score and have pointed out that the female is the basic state, deviation to the male starts *in utero* under the influence of the foetal hormones, and for these to be present in the normal ratio the foetus must have the correct complement of sex chromosomes. The Klinefelter syndrome of hypogonadism and female sex chromatin has a smaller biacromial diameter, and therefore a low androgyny score (Raboch, 1957). The mongol is considered to have normal sex chromosomes and the sex differentiation at birth is normal, it is only at the time of puberty when the second phase of sex differentiation occurs that the mongol boy diverges from the control group as far as the androgyny score is concerned. Vague (1956) has drawn attention to the intensity with which various parts of the body respond to the sex differentiating hormones, and this may be an important factor in the mongol.

The onset of puberty in the mongol occurs at the normal time. The timing of puberty in the normal male is not well understood; it may be due to a variation in the sensitivity of the gonads or an alteration in the feedback mechanism of the gonad-hypothalamic-pituitary axis. Animal experiments have shown that if an immature gonad is transplanted into an adult animal pubertal changes occur in the gonad and vice versa. It may also be that in the hypothalamus there are two centres regulating sexual development, one stimulatory and the other suppressive. It is also known that the male pituitary is depleted of its hormones just prior to puberty; in the female no such depletion occurs at this time; this difference can be abolished by castration in the male.

The possible causes for the failure of full sexual development and low

androgyny score would appear to be either abnormally low sensitivity of parts of the mongol body to differentiating hormones, or an abnormality of the hormones produced by the pituitary. Dutton (1959a) suggested that the pituitary may be at fault in the causation of the specific failure of longitudinal growth with a normal skeletal maturation.

Summary

The androgyny score and sexual maturation of male mongols was compared with a control group from non-mongol mentally subnormal males. The mongols were found to have a female androgyny score, normal onset of puberty but incomplete sexual development as regards pubic hair and genitalia. The possible causes of these findings were discussed.

Acknowledgments

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