

EDITORIAL : VAN DIE REDAKSIE

SEX CHROMOSOME ABNORMALITIES IN GROUP SURVEYS

When Moore and Barr (1955)¹ introduced their method of detecting abnormalities of nuclear sex by examining cells of the oral mucosa, surveys of large population groups became possible. Mental defectives provided one group in which abnormalities were expected to occur more frequently than in the general population, since both Klinefelter's syndrome and the triple X 'super female' were occasionally found to be associated with mental defect, and also some young 'women' with gonadal dysgenesis (Turner's syndrome) seemed to have rather poor cerebral equipment. Barr and his colleagues² themselves reported that 14 chromatin-positive patients were found among 1,506 institutionalized male mental defectives (0.94%). Three of the patients had duplicated sex chromatin. Two of these and 11 patients with typical chromatin-positive nuclei belonged to the Klinefelter syndrome clinically (0.86%).

Several surveys have been undertaken to determine the nuclear sex of boys attending special schools for the mentally backward, and of both male and female inmates of institutions for the mentally defective. One such survey was reported in a combined Edinburgh and Dartford effort in 1962, with summaries of previous work in this field;³ 4,514 inmates of mental defective institutions were examined. Twenty-eight out of 2,607 males were found to be chromatin-positive—23 with a single sex-chromatin body, 4 with some cells in which 2 bodies were seen, and 1 with 3 bodies in a proportion of cells. The over-all frequency of chromatin-positive males was 10.7 per 1,000. Among 1,907 females, 8 were found to have 2 sex-chromatin bodies in a proportion of their buccal cells—a frequency of 4.2 per 1,000—while only 1 was chromatin-negative.

Chromosome studies on the 28 males showed 17 to have an XXY complement (6.5 per 1,000), 4 an XXXY complement (1.5 per 1,000), 2 an XXYY complement (0.8 per 1,000), and 5 were mosaics (1.9 per 1,000). One of the subjects with an XXY complement (case 21) was also trisomic for autosome 21.

Nine abnormal females were found, of whom 8 had double sex-chromatin bodies in a proportion of their buccal cells, while the 9th was chromatin-negative. Of the 8 with double sex chromatin, 7 were shown to have an XXX sex-chromosome complement, while the 8th was found to be a mosaic of XX/XXX type. The 1 chromatin-negative patient was 7 years old and had an XO sex-chromosome complement.

In summary, the frequency of abnormal males was 10.7/1,000, of XXX females 4.2/1,000 and of XO females 0.5/1,000. The available evidence indicated a significant association between major mental deficiency and an increased complement of X chromosomes, in both phenotypic males and females. On the other hand, the limited data suggest that women with an XO sex-chromosome complement are not appreciably mentally handicapped. Hamerton *et al.*⁴ at about the same time commented that it was unlikely that the association of intellectual subnormality with sex-chromosome aberrations was due specifically to genes or gene complexes on the sex chromosomes, but rather to a generalized imbalance of the chromosome set as a result of aneuploidy. Furthermore, it seemed probable that the relatively mild malformations resulting from abnormalities of the sex chromosomes, when compared with malformation caused by autosome anomalies, were due to the relative genetic inertness of the sex chromosomes when compared with the autosomes.

More recently Court Brown and his group⁵ have published the results of over 20,000 buccal smear examinations in newborn babies. Twenty-one of the male babies were chromatin-positive (1.96/1,000), 4 of the females were chromatin-negative (0.4/1,000), while 12 females had cells with 2 sex-chromatin bodies (1.2/1,000).

Chromosome studies were made in 18 male and 13 female liveborn infants. Twelve of the males had XXY and 1 had XXYY sex-chromosomes; the remaining 5 were examples of XY-XXY mosaicism. Nine of the females had XXX sex-chromosomes, 3 had an XO sex-chromosome constitution, and 1 was an example of XO/XX mosaicism.

At least in Britain it would appear that the genetic abnormalities associated with Klinefelter's syndrome are comparatively common (about 2 per 1,000); the 'triple X' syndrome is less common, and Turner's syndrome comparatively rare. While the first 2 anomalies are rather frequently associated with mental defect, Turner's syndrome does not appear to be so to any measurable extent.

1. Moore, K. L. and Barr, M. L. (1955): *Lancet*, **2**, 57.
2. Barr, M. L., Shaver, E. L., Carr, D. H. and Plunkett, E. R. (1960): *J. Ment. Defic. Res.*, **4**, 89.
3. Maclean, N., Mitchell, J. M., Harnden, D. G., Williams, J., Jacobs, P. A., Buckton, K. A., Baikie, A. G., Court Brown, W. M., McBride, J. A., Strong, J. A., Close, H. G. and Jones, D. C. (1962): *Lancet*, **1**, 293.
4. Hamerton, J. L., Jagiello, G. M. and Kirman, B. H. (1962): *Brit. Med. J.*, **1**, 220.
5. Maclean, N., Harnden, D. G., Court Brown, W. M., Bond, J. and Mantle, D. J. (1964): *Lancet*, **1**, 286.

PROBLEME VAN DIE MIDDELJARE

Nie net die probleme van die toenemende aantal bejaardes in die samelewing geniet vandag besondere aandag nie, maar ook die probleme van die groot groep mense in die middeljare, d.w.s. mense van ongeveer tussen die ouderdomme van 45 - 64 jaar. Dat daar baie spesiale probleme

is waarvoor hierdie groep mense te staan kom, weet ons. Ons is egter nie altyd bewus van hoe groot die omvang van dié probleme is nie, ook nie van al die implikasies vir die toekomstige geluk en gesondheid van hierdie groot groep mense wat die onvervangbare spil uitmaak van die

wiel waarom die hele geweenskapslewe draai nie. Om hierdie rede sal dit goed wees vir almal wat met mense en hul gesondheid te doen het, om ernstige aandag te gee aan 'n interessante en prikkelende onlangse publikasie: *Problems of the Middle-aged*,¹ onder die redaksie van Prof. Clyde Vedder van die Departement van Sosiologie en Antropologie van die Northern Illinois University, De Kalb, Illinois.

Die sentrale tema van hierdie publikasie is dat die groot uitdaging van die studie van die ouderdomsverskynsels (die gerontologie) vandag eintlik in die middeljare lê. Hierdie middeljare, wat ongeveer van 45 tot 64 strek, is die fase van die lewenssiklus waarvan ons op stuk van sake die minste weet. En tog val nagenoeg 20% van die hele bevolking (en miskien 80% van die dinamies-produktiewe deel van die gemeenskap) in hierdie kategorie.

Die besprekings wat in *Problems of the Middle-aged* bymekaargebring is, is gekies met die bedoeling om lig te werp op die betekenis en gevare van die middeljare, sodat feite beskikbaar gestel kan word wat ons kan help om onredelike opvattinge, vooroordele en nodelose vrese af te breek. Om die gebied wat gedek word so omvattend moontlik te maak, sluit die boek deskundige artikels in uit die verwante gebiede van die medisyne, die sielkunde, die sosiologie en die ekonomie—in soverre as wat hulle betrekking het op die sentrale fasette van die middeljare, b.v. houdinge ten opsigte van ouer word, oefening na 40, probleme ten opsigte van werkaangeleenthede, beraad met betrekking tot plasing in werk, gesinsverhoudinge en

-spanninge, maatskaplike gedragspatrone, spanning en drukte, emosionele konflikte, migraine en ander disritmiese versteurings, en algemene geneeskundige en chirurgiese stellingsmetodes.

In die verlede het maar net enkele skrywers spesiale aandag gegee aan die probleme van die middeljare. So vestig Barron, byvoorbeeld, in *The Aging American* die aandag op sosio-sielkundige en gesondheidsprobleme van hierdie tydvak. Tibbits and Donahue toon aan in *Aging in Today's Society* dat die middeljare 'n tyd van geleenthede en uitdagings is. Gervant, in *You're Older than you Think*, toon aan dat die uitdaging van die middeljare daarin lê om voor te berei vir die jare wat voorlê en *nie* om tevrede te wees met 'n skemerkelkie voor ete, 'n slapie na die maaltyd, die aandkoerant en ander eksterne vermake nie. Hierdie tydrawende roetine kan 'n belemmering vir geestelike groei en ontwikkeling word en kan lig ontwikkel in die mees onproduktiewe en ontwrigtende stel gewoontes waarin 'n mens kan verval. En Cabot in *You Can't Count on Dying* stel een van die belangrikste vrae van ons tyd deur te vra: waarom moet middeljarige mense *uit 'n werk* aftree en waarom kan hulle nie ook *terwille van iets anders* en iets nuuts aftree nie?

Die publikasie waarna ons hier veral verwys, *Problems of the Middle-aged*, lewer 'n beduidende en belangrike nuwe bydrae tot hierdie probleemgebied. Dit behoort 'n wye leserskring onder geneeshere en welsynswerkers te vind.

1. Vedder, C. B. red. (1965): *Problems of the Middle-Aged*. Springfield, Illinois: Charles C. Thomas.