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# A NOTE ON MONGOLISM IN TWINS

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It has often been thought that if one member of a pair of twins is a mongol, the other member is always affected if the pair is monozygous, and rarely affected if the pair is dizygous. This view is based on the absence of any report of monozygosity in a discordant pair. However, little evidence is available, since few of the pairs that have been recorded may be accepted unreservedly as monozygous. In the absence of direct evidence the matter can be examined by considering (*a*) the frequency of concordance among affected pairs, and (*b*) the numbers of affected pairs that are of like and of unlike sex.

The present report is based on 129 pairs in which sex, number affected, and diagnosis are believed to be reliably recorded. One hundred and seven were assembled by Øster (1953) and sources of a further 22 pairs are given in the Appendix. Nineteen of the latter were reported in the literature; the remaining three pairs are now reported for the first time:

(*a*) Table I shows the distribution according to sex of 152 affected in 129 pairs. Acceptable evidence of dizygosity is recorded for three of 23 pairs in which both twins are affected (MacKaye, 1936; Gordon and Roberts, 1938; Jervis, 1943). Even if it is assumed that all other concordant pairs are monozygous, the proportion (20/129 = 15·5 per cent.) is lower than estimates usually given for the proportion monozygous among representative twin pairs. Since the proportion of monozygous twins varies with maternal age, reaching its lowest value at the age quinquennium 35–39 years (Waterhouse, 1950), it is conceivable that this result may be due to the late maternal age at which many mongols are born. Maternal age is known for only 55 of the 129 pairs.

TABLE I
DISTRIBUTION OF AFFECTED TWINS

| Type of Pair | | Number in Discordant Pairs | Number in Concordant Pairs | Total Number of Affected |
|---|---|---|---|---|
| Like Sex | Males | 31 | 20 | 51 |
| | Females | 31 | 26 | 57 |
| Unlike Sex | Males | 25 | — | 25 |
| | Females | 19 | — | 19 |
| Total | Males | 56 | 20 | 76 |
| | Females | 50 | 26 | 76 |

However, it is possible to estimate the maternal age distribution of affected twins from knowledge of the age distributions of unaffected twins and of a representative series of mongols. An estimate has been calculated from the distribution of 241 mongols born in the City of Birmingham during the 11 years 1942–52 (Smith and Record. 1955a) and from the Registrar-General's figures for the maternal age distribution of twin maternities in England and Wales during the same years. Comparison of this estimated distribution with that for the small series of affected twins for which maternal age at birth was recorded (Table II) shows reasonable agreement.

TABLE II
MATERNAL AGE DISTRIBUTION OF AFFECTED TWINS

| Maternal Age (yrs) | Under 20 | 20–24 | 25–29 | 30–34 | 35–39 | 40–44 | 45 and over | Total |
|---|---|---|---|---|---|---|---|---|
| Expected per cent. | 0·5 | 5·1 | 13·9 | 18·9 | 34·1 | 26·1 | 1·4 | 100 |
| Observed per cent. | 1·8 (1) | 5·5 (3) | 14·5 (8) | 9·1 (5) | 36·4 (20) | 25·5 (14) | 7·3 (4) | 100 (55) |

Expected proportions of monozygous twins at each maternal age quinquennium have been estimated by Weinberg's method from data published by the Registrar-General for the 11 years for which data on mongol births were available. Application of these proportions to the estimated distribution by maternal age for affected twins gives a value for the expected proportion of monozygous pairs among pairs selected by the presence of an affected member. This proportion (26·3 per cent.) is greater than the proportion of concordant pairs for which there is no evidence of dizygosity (15·5 per cent.), the difference (10·8 per cent.) being more than 2·5 times its standard error (3·9 per cent)..

The above procedure is open to the criticism that twin pairs reported in the literature may be unrepresentative. It is probable, however, that selection will tend to favour publication of concordant rather than of discordant pairs.

(*b*) The expected numbers of like and unlike sexed pairs can be estimated in 109 presumptively dizygous pairs composed of 106 discordant pairs

(assumed to be dizygous) and three concordant pairs whose dizygosity was established. The sex ratio of affected twins is 50 per cent., so that the expected proportion of like sexed pairs is also 50 per cent. Table III shows that there is a significant excess of like sexed pairs. It therefore seems unlikely that all the discordant pairs are dizygous.

TABLE III
DISTRIBUTION OF PRESUMPTIVELY DIZYGOUS TWIN PAIRS

| Sex | Like Sexed | Unlike Sexed | Total |
|---|---|---|---|
| Observed (*a*) .. | 65 | 44 | 109 |
| Expected (*b*) .. | 54·5 | 54·5 | 109 |
| Difference (*a*)–(*b*) | +10·5 | −10·5 | — |

$\chi^2 = 4{\cdot}05$, $n = 1$, $p < 0{\cdot}05$

It is possible to estimate the probable number of discordant monozygous pairs by subtracting the number of unlike sexed from the number of like sexed discordant pairs. If the difference (18) is added to the number of concordant pairs for whom there is no evidence of dizygosity (20), the total (38) may be regarded as an estimate of the number of monozygous pairs. The proportion of monozygous pairs calculated from this estimate (38/129 = 29·5 per cent.) is very near the expected proportion of monozygous pairs (26·3 per cent.). The fact that it is slightly larger may be due to the selective reporting of concordant pairs.

An alternative explanation which should be considered is that foetal loss might account for the low proportion of concordant pairs. Some support for this possibility is provided by the observation (Smith and Record, 1955b) that threatened abortion may be more common in pregnancies that result in a mongol than in pregnancies that result in unaffected births. If the presence of one malformed foetus predisposes to abortion, the presence of two might increase the risk. Foetal loss in concordant pairs cannot, however, account for the excess of like sexed pairs among discordant pairs, and it is concluded that the data suggest that mongolism sometimes occurs in only one of a monozygous pair.

## SUMMARY

To 126 reports of mongolism in one or both members of a twin pair are added three new pairs. The 129 pairs contained 152 affected. Reliable evidence of zygosity is available for very few.

The proportion of concordant pairs for whom no evidence of dizygosity is reported is significantly less than the expected proportion of monozygous pairs. If it is assumed that all discordant pairs are dizygous, they contain a significant excess of like sexed pairs. When the proportion of monozygous pairs among the reported pairs is estimated by Weinberg's method, it agrees well with the expected proportion of monozygous pairs.

It is suggested that the most likely explanation of these results is that some of the discordant pairs are monozygous.

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## APPENDIX

SOURCES OF 22 TWIN PAIRS CONTAINING AT LEAST ONE MONGOL, NOT INCLUDED IN THE SERIES OF 107 PAIRS ASSEMBLED BY ØSTER (1953)

| Author | Date | Sex of Twins | Remarks |
|---|---|---|---|
| Wieland .. .. | 1927 | **f f** | Stated to be monozygous |
| Arancibia and Fumasoli .. .. | 1935 | **m f** | |
| Jenkins .. .. | 1936 | **f m** | |
| Doxiades and Portius | 1938 | **f f**<br>**f f** | |
| Gordon and Roberts | 1938 | **f f** | Dizygous (finger prints) |
| Harker .. .. | 1942 | **f f** | |
| Déchêne .. .. | 1947 | **f f** | |
| Cook .. .. | 1950 | **m f**<br>**f m** | |
| Posteraro .. .. | 1951 | **m f** | |
| Robertson .. | 1952 | **m f** | |
| Lang-Brown, Lawler, and Penrose .. | 1943 | **f f**<br>**f f**<br>**m m**<br>**m m** | Monozygous (blood groups)<br>Dizygous (blood groups)<br>Dizygous (blood groups)<br>Dizygous (blood groups) |
| Morris and MacGillivray .. | 1953 | **f f**<br>**m f** | |
| Young .. .. | 1954 | **m m** | Monozygous (blood groups and finger prints) |
| Not previously reported .. | | **m m** | Dizygous (blood groups and finger prints) |
| Not previously reported .. | | **m m** | Zygosity not examined |
| Not previously reported .. | | **f f** | Zygosity not examined |

Sex of affected twins is given in heavy type.