

CONCORDANCE AND DISCORDANCE OF ANENCEPHALY IN 109 TWIN PAIRS IN JAPAN

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Summary One hundred and nine pairs of twin with anencephaly were ascertained during the period from 1969 to 1976 in Japan. The rate of concordance in anencephalic twins was 8.3% and seems to be higher than those of previous reports.

INTRODUCTION

The indications of genetic factors in the etiology of anencephaly come from an influence of parental consanguinity, a specific familial aggregation and twin studies (Carter, 1974). The recurrence risk after the birth of the first anencephalic child in a sibship was about 4%, but after two affected sibs the risk rose to 13% (Masterson, 1962). On the other hand, the concordance rate for anencephalic twins was very low (Lemire *et al.*, 1977), although published data on anencephalic twins were not enough (Rogers and Weatherall, 1976).

In Japan the twin concordance rate for anencephaly was reported to be zero in a previous study (Imaizumi, 1974). The present communication deals with further analyses of observation on such twins including additional data thereafter obtained.

MATERIAL

Data on anencephalic twins were obtained from the fetal death and postnatal death certificate records reported during the period from 1969 to 1976. Details about the certificate records have been reported elsewhere (Imaizumi, 1974).

RESULTS AND DISCUSSION

Table 1 shows twin concordance and sex of twin sets with anencephaly. Data on anencephalic twins for the 38 among 109 pairs were published by Imaizumi (1974). Nine among 109 pairs (8.3%) of twin were concordant. The sex of twins was ascertained for the 62 among 109 twin pairs. The number of like-sexed twin pairs were 56 and the number of unlike-sexed twin pairs were six. Sex of one

Table 1. Twin concordance of anencephalic cases.

Years	Concordant			Discordant									Total
	Sex of twin sets			Sex of twin sets									
	MM	FF	MF	MM*	FF*	MF*	FM*	M?*	F?*	?M*	?F*	??*	
1969-1971	0	0	0	7	10	2	0	8	7	1	0	3	38
1972-1974	3	3	1	11	4†	0	1	10	9	0	0	0	42
1975-1976	0	1	1	11	6	1	0	2	6	0	1	0	29
Total	3	4	2	29	20	3	1	20	22	1	1	3	109

M*, unaffected male; F*, unaffected female; ?*, unaffected unknown sex; †, one twin pair has anencephaly and congenital hydrocephaly.

member or both of a twin pair was unknown in the remaining 47 pairs. The proportion of unlike-sexed twins in the general population was 16.11% in Japan (Ministry of Health and Welfare of Japan, 1974), and the value in this study was 9.68% (6/62). The difference between both values is statistically significant at the 0.1% level. Namely, the deficiency of unlike-sexed twins was observed in anencephalic twins.

James (1976) mentioned that concordance rates are higher in the like-sexed than in the unlike-sexed pairs. On the other hand, in the present study, concordance rates were 13% (7/56) in like-sexed and 33% (2/6) in unlike-sexed pairs. However, in the latter case, the sex distribution of 47 twin pairs among 109 pairs were unknown.

Rogers and Weatherall (1976) have assembled from the literature a series of 681 twin pairs with anencephaly or spina bifida. Rogers (1976) used these data and estimated that the concordance rates were 17.8% (26/146) and 1.6% (8/535) for monozygous and dizygous twins, respectively. However, the value for dizygous twins was very low compared with the recurrence risk in sibs born after an affected child. Table 2 shows twin concordance and sex of anencephaly which were obtained from the literature. Nineteen among 422 pairs of twins are concordant, and the concordance rate is 4.5%. The sex distribution of six twin pairs among 422 pairs in Table 2 were unknown. It is generally accepted that approximately 70% of like-sexed twins are dizygous (Rogers, 1976). When the total of 299 like-sexed pairs (284 discordant and 15 concordant) is divided into 70% dizygous and 30% monozygous twins, 209 dizygous and 90 monozygous twins are obtained in the like-sexed pairs. From these numbers, the 15 concordant like-sexed pairs were estimated to be composed of 7 dizygous twins ($4 \times 209/117$) and 8 monozygous twins. Therefore, the concordance rate is 8.9% (8/90) for monozygous and 3.4% (11/326) for dizygous twins.

Overall concordance rate in the present study was 8.3% (9/109) and seems to be higher than those in other investigations (see Rogers and Weatherall, 1976; James, 1976; Janerich and Piper, 1978). However, the results are not conclusive and further investigation is needed.

Table 2. Anencephaly in twins.

Authors	Total cases of anencephaly	Discordant pairs affected				Concordant pairs affected				Total of affected pairs
		Like sex		Unlike sex		Like sex		Unlike sex		
		Like sex	Unlike sex	Not stated	Like sex	Unlike sex	Like sex	Unlike sex		
Böök and Rayner (1950)	67	3	0	0	0	0	0	0	3	
Record and McKewen (1951)	?	17	7	0	0	0	0	0	24	
Wilson (1955)	?	1	0	1	0	0	0	0	2	
Coffey and Jessop (1957)	137	1	1	0	0	0	0	0	2	
Neel (1958)	40	1	0	0	0	0	0	0	1	
Dumoulin and Gordon (1959)	?	2	1	0	0	0	0	0	3	
Lewis (1960)	?	0	0	0	0	0	1	1	1	
Towell (1961)	?	0	0	0	0	1	0	0	1	
Alter (1962)	29	0	0	0	0	1	0	0	1	
Frézal <i>et al.</i> (1964)	402	5	1	1	0	0	0	0	7	
deSa' <i>et al.</i> (1964)	?	1	0	0	0	0	0	0	1	
Williamson (1965)	29	2	0	0	0	0	0	0	2	
Symonds (1965)	?	0	1	0	0	0	0	0	1	
Gittelsohn and Milham (1965)	1,504	24	14	0	0	4	1	1	43	
DeHaan (1966)	?	0	0	0	0	1	0	0	1	
Stevenson <i>et al.</i> (1966)	382	8	2	0	0	0	0	0	10	
Carter <i>et al.</i> (1968)	364	5	1	0	0	0	0	0	6	
Yen and MacMahon (1968)	782	9	4	1	0	0	0	0	14	
Butler and Alberman (1969)	494	7	3	2	0	0	0	0	12	
Horowitz and McDonald (1969)	948	24	16	1	0	0	0	0	41	
Czeizel and Révész (1970)	360	1	5	0	0	0	0	0	6	
Hay and Wehrung (1970)		57	14	0	0	1	0	0	72	
Overbeke (1971)		88	30	0	0	4	1	1	123	
Elwood and Nevin (1973)	175	3	1	0	0	0	0	0	4	
Carter and Evans (1973)	425	6	3	0	0	0	1	1	10	
Janerich and Piper (1978)		19	9	0	0	3	0	0	31	
Total		284	113	6	15	4	4	4	422	

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