PATHOLOGIC FINDINGS OF NEURAL TUBE DEFECTS*

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SUMMARY

This report presents the congenital malformations found in a prospectively collected series of 10 infants with neural tube defects. Nine of these were stillborns and one was a neonate who died within 24 hours after birth. All of the infants were singletons. Five of them had craniorachischisis totalis, 2 had anencephaly, 2 had encephalocele, and one had very severe form of cerebro-facial dysplasia.

Key Words: Neural tube defects, craniorachischisis totalis, anencephaly, encephalocele, cerebro-facial dysplasia.

INTRODUCTION

Neural tube defects (NTDs) are among the most common congenital afflictions of man. Their epidemiologic characteristics are not clear, but it may be assumed that both genetic and environmental factors contribute to their pathogenesis (1-5). They are known to exhibit wide geographic, ethnic, and sex, and secular variation (6-8). The aim of this study was to evalute the frequency and the types of NTDs in Diyarbakır.

MATERIALS AND METHODS

During a prospective study all of the 1,500 pregnant women who admitted to the Diyarbakır Maternity Hospital and the University Hospital, University of Dicle and their offsprings were carried out between January and September 1988. One infant and 9 stillborn showed NTD. All the autopsies and pathological examinations were performed by the author.

RESULTS

Of the 10 aborted children, 5 had craniorachischisis totalis, 2 had anencephaly, 2 had encephalocele, and one had the severest form of cerebro-facial dysplasia. The detailed findings are summarized in Table I.

DISCUSSION

10 cases of NTD were found in 1,500 births. The frequency of cases with NTD at the Maternity Hospital and at the University Hospital seems to be rather high, comparad to the other parts of Turkey such as 1.8% and 2.6% in Ankara (9,10) and 2.2% in İstanbul (11), and 1.9% in İzmir (12). Of the 10 infants with NTD, 9 were stillborn, compatible with the literature (9,13).

The sex distribution, showed one male, and 9 females similar to the results of other studies (7,14,15). All the infants with NTD were singletons.

In regard to gestational age, one infant was pre-term while the other 9 were of normal gestational age.

Though in NTDs familial occurence is known (16), one of these 10 infants had positive familial history of NTD.

Although maternal zinc deficiency was shown to be one of the factors responsible for NTDs in Turkey (10,17-20), in this study maternal zinc status was not known.

The association of NTDs with some maternal diseases such as organic heart diseases, chronic lung diseases, diabetes mellitus (15,21-23), polyhydramnios, abrabtio placentae, maternal exposure to medications including sulfonamides, diuretics, and antihistamines (14,24,25) is well-known. In this study there was no history about these diseases or medications with the exception of the infant with encephalocele. The mother of this infant received anti-depressants during the first trimester. Although the association between maternal fever and NTDs has been reported both in human population (26-30), and in other species (31,32) in this study such an association was not noted.

Knox's fetus-fetus interaction hypothesis (33,34), was not obvious. In this study 2 probands were from first pregnancies. The other infants' mothers have had multiple pregnancies. Multiparity may suggest the fetus-fetus interaction which could not be considered in these cases.

As shown in Table I, other organ system malformations were common in this study. These findings are in agreement with other studies (2,13,35,36). On the other hand some of the associated congenital malformations in our cases were unusual in nature such as dextrocardia, single ventricle, right-sided arcus aorta, double thymus, thymic hypoplasia, and asplenia.

No previous studies in this subject in Diyarbakır could be found for the comparison. The study is in progress, and the mothers are followed-up by obstetricians for future pregnancies.

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TABLE I: PATHOLOGIC FINDINGS OF THE CASES WITH NEURAL TUBE DEFECTS

Case No.	Sex	Type	Malformations in Other Organ Systems
1	F	CFD	—
2	M	E	cleft palate, hypoplastic adrenals, and left ectopic adrenal
3	F	CT	cleft plate, low-set and rotated ears, goiter, thymic enlargement, lung hypoplasia, diaphragmatic hernia, ectopic kidneys, accessory spleen, adrenal agenesis.
4	F	E	fusion defect of the nose, cleft lip and palate, goiter, double thymus, left polycystic kidney and hydroureter, adrenal agenesis.
5	F	CT	hypoplastic adrenals.
6	F	CT	exophthalmos, corneal clouding, low-set and rotated ears, cleft palate, thymic hypoplasia, lung hypoplasia, single lobe (left lung), biatrium triloculare, aortic stenosis, right-sided arcus aorta, asplenia, hypoplastic adrenals.
7	F	A	exophthalmos, low-set and rotated ears, cleft palate, goiter, thymic hypoplasia, lung hypoplasia, single lobe (both right and left lung), dextrocardia, right-sided arcus aorta, aortic atresia, diaphragmatic hernia, intestinal malrotation, hypoplastic adrenals.
8	F	A	low-set and rotated ears, thymic enlargement, short bowel, hypoplastic adrenals.
9	F	CT	exophthalmos, corneal clouding, low-set ears, thymic enlargement, lung hypoplasia, hypoplastic adrenals.
10	F	CT	exophthalmos, corneal clouding, low-set and rotated ears, thymic hypoplasia, lung hypoplasia with incomplete lobation, hypoplastic adrenals.

CFD. cerebro-facial dysplasia.
CT. craniorachischisis totalis.
E. encephalocele.
A. anencephaly.

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