Clinical Observations

Trisomy 18 and Neural Tube Defects

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ABSTRACT

BACKGROUND: Trisomy 18 or Edwards syndrome is a chromosomal abnormality characterized by a broad clinical picture and a limited survival. More than 130 different abnormalities have been described in these patients—among them are neural tube defects. METHODS: We verified the frequency and types of major neural tube defects observed among patients with trisomy 18. Our sample consisted of consecutive patients evaluated by a clinical genetics service of a referral hospital in southern Brazil between 1975 and 2008. Fisher’s exact test (two-tailed) and chi-square test with Yates’ correction were used to compare frequencies (P < 0.05 values were considered as significant). RESULTS: During the period of evaluation, we identified 50 patients with trisomy 18; 33 (66%) were female and age at the first evaluation ranged from 1 day to 16 years (median 14 days). One cell line with full trisomy 18 was the predominant cytogenetic finding (90%). Three patients (6%) had major neural tube defects, all females. These were two patients (4%) with encephaloceles and one (2%) with myelomeningocele. This last patient underwent correction surgery on her first day of life. CONCLUSIONS: Our data, in accordance with the literature, support the idea that the presence of neural tube defects among patients with trisomy 18 is not coincidental (i.e., these defects are actually part of the spectrum of abnormalities presented in trisomy 18). Thus, the diagnosis of trisomy 18 should be considered in children with major neural tube defects, especially in the presence of other abnormalities or dysmorphisms.

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Introduction

Trisomy 18 or Edwards syndrome is a chromosomal abnormality characterized by a broad clinical picture and a limited survival. More than 130 different abnormalities have been described in these patients, and among them are neural tube defects [1].

We verified the frequency and types of major neural tube defects observed among patients with trisomy 18.

Materials and Methods

Our sample consisted of consecutive patients evaluated by a clinical genetics service of a referral hospital in southern Brazil between 1975 and 2008. The results of cytogenetic analysis and clinical data were collected from medical records of patients, with emphasis on findings related to neural tube defects. Fisher’s exact test (two-tailed) and chi-square test with Yates’ correction were used to compare frequencies (P < 0.05 values were considered as significant). This study was approved by the Research Ethics Committee of the Universidade Federal de Ciências da Saúde de Porto Alegre.

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Results

During the period of evaluation, we identified 50 patients with trisomy 18; 33 (66%) were female, and age at the first evaluation ranged from 1 day to 16 years (median 14 days). In relation to the cytogenetic findings, the presence of one cell line with full trisomy 18 was predominant (90%). Three patients (6%) had major neural tube defects—all females—these were two patients (4%) with encephaloceles and one (2%) with myelomeningocele. The patient with myelomeningocele was one of the two individuals in the sample who had chromosomal constitution with double aneuploidy: trisomy 18 associated with triple X. The patient with myelomeningocele underwent corrective surgery on her first day of life. This surgical procedure was performed before the diagnosis of trisomy 18.

Discussion

Despite central nervous system abnormalities being common among patients with trisomy 18, neural tube defects have been considered an uncommon finding [2-4]. Despite this, when we compare the frequency of neural tube defects found in our sample with that observed in the general population [5], we observe a significant relationship ($P < 0.0001$).

The frequency of neural tube defects found in our sample (6%) was similar to that described in other studies of trisomy 18 patients (6% to 12%) [2-4]. Myelomeningocele has been considered the defect most often reported [2-4]. Encephalocele is a rare finding among postnatal patients with trisomy 18. Interestingly, this defect was the neural tube defect most observed in our sample. Although not verified in our study, examples of anencephaly and craniarachischisis have also been described in the literature among individuals born with trisomy 18 [4].

Conclusion

Our data, in accordance with the literature, support the idea that the presence of neural tube defects among patients with trisomy 18 is not coincidental (i.e., these defects are actually part of the spectrum of abnormalities presented in trisomy 18). Thus, the diagnosis of trisomy 18 should be considered in children with major neural tube defects, especially in the presence of other abnormalities or dysmorphisms. This may have important implications on the management, prognosis, and genetic counseling of patients.

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