0021-7557/11/87-03/225
Jornal de Pediatria

Time of diagnosis of oral clefts: a multicenter study

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Abstract

Objective: To determine the time of diagnosis of typical orofacial clefts in different Brazilian regions and its influence on age at surgical correction.

Method: This was a prospective, descriptive, cross-sectional study conducted in medical centers in the Southeast, South, and Northeast of Brazil. Trained speech therapists and geneticists interviewed the parents of affected children using a previously validated questionnaire. Epi-Info and SPSS were used for data analysis. Significance level was set at 5% ($p \leq 0.05$).

Results: The sample consisted of 215 interviews conducted in the following regions: 21.9% (47) in the Southeast, 51.1% (110) in the South, and 27% (58) in the Northeast. Monthly family income was higher in the Southeast ($p \leq 0.05$). Cleft lip and palate were found in 61.4% (132) of cases, cleft palate in 20.9% (45), and cleft lip in 17.7% (38). Diagnosis occurred in the maternity ward in 75.3% (162) of cases, during the prenatal period in 14% (30), and after hospital discharge in 10.2% (22). The Southeast had a higher frequency of prenatal diagnosis (27.7%), possibly related to greater purchasing power in this region and greater availability of prenatal investigation. Of all cases diagnosed in the maternity ward, 74.4% occurred in the Northeast. However, no significant difference was found when comparing time of diagnosis, region, and age at first surgery.

Conclusion: Considering that diagnosis is more common in the maternity ward, local health care teams should be trained in order to effectively improve the initial care of these patients. Although time of diagnosis did not affect age at surgery, it favors the planning of neonatal care and treatment of affected infants.

J Pediatr (Rio J). 2011;87(3):225-230: Cleft lip, cleft palate, diagnosis, public health.

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No conflicts of interest declared concerning the publication of this article.

Financial support: Fundação de Amparo à Pesquisa do Estado de São Paulo (FAPESP), Brazil.

Suggested citation: Amstalden-Mendes LG, Xavier AC, Antunes DK, Ferreira AC, Tonocchi R, Fett-Conte AC, et al. Time of diagnosis of oral clefts: a multicenter study. J Pediatr (Rio J). 2011;87(3):225-230.

Manuscript submitted Oct 21 2010, accepted for publication Dec 23 2010.

doi:10.2223/JPED.2084

Introduction

Cleft lip and/or palate, also called typical orofacial clefts (TOCs), are the most common craniofacial birth defects and their formation occurs during embryonic development.[1,2] TOCs affect one in every 600 newborn babies[3,4] and manifest alone or associated with other birth defects.[5] About 300 syndromes appear to have TOC as one of their characteristics.[6-8]

Early diagnosis of TOCs makes it possible to investigate other defects and prevent and/or minimize complications.[9-12] Difficulties in feeding are the most common complications, such as insufficient sucking, presence of milk within the nasal cavity, suction, and decreased food intake, affecting the infant's nutritional status and resulting in poor weight gain.[13,14]

Neonatal care is a complex approach and the involvement of the health care team since diagnosis may help parents understand the implications of that birth defect and the potential for esthetic and functional correction, in addition to taking measures to minimize comorbidities.[15-17] Immediate postnatal pediatric management requires critical decisions, such as measures regarding infant hygiene and feeding practices, investigation of associated anomalies, and referral to clinical and genetic assessment and to surgical correction in reference centers.[16,18,19]

The objective of the present study was to determine the time of diagnosis made in different Brazilian regions.

Method

This prospective, descriptive, cross-sectional study was approved by the Research Ethics Committee (no. 438/2002). Parents or legal guardians of children with TOCs, aged 0 to 12 years, from eight health care facilities were invited to participate in the study. Of these facilities, five were specialized centers (three in the South, one in the Northeast, and one in the Southeast) and three were non-specialized centers (two in the Northeast and one in the Southeast). Specialized centers were considered as those in which cleft care was provided by a multidisciplinary team of specialists, regardless of the specialist skills of all team members.

Data were collected using a validated structured interview and through voluntary statements. The interviews were recorded; a single researcher listened to all recordings and tabulated the data.

Statistical analysis was performed using Epi-Info (version 6.04d, Centers for Disease Control and Prevention, Atlanta, USA) and SPSS version 16.0 (SPSS Inc., Chicago, USA). The chi-square test and analysis of variance (ANOVA) were used to compare mean values. Significance level was set at 5% ($p \leq 0.05$).

Results

Of 230 interviews received for analysis, 15 were excluded. The final sample consisted of 215 (100%) valid interviews: 51.1% (110) from the South, 27% (58) from the Northeast, and 21.9% (47) from the Southeast. There were no participating centers from the North and Midwest. Specialized centers accounted for 86.51% (186) and non-specialized centers for 13.49% (29) of interviews.

Of all cases, 62.8% (135) were male and 37.2% (80) were female. The patients' mean age at interview was 4 years, and 75% were under 8 years of age.

According to data reported by the families, associated anomalies were investigated in 89.3% (192) of cases. Clefts alone were found in 85.42% (164) of cases, and in 14.58% (28) of cases clefts were associated with other anomalies. In the Northeast, in 10.7% (23) of cases the diagnosis had yet to be established.

Regarding cleft type, cleft lip and palate (CLP) were found in 61.4% (132) of cases, cleft palate (CP) in 20.9% (45), and cleft lip (CL) in 17.7% (38). There was a significant difference in the frequency of cleft type per region ($p = 0.040$). CLP was more frequent in the Northeast, South, and Southeast, respectively (Table 1). The Northeast showed a lower proportion of CL cases in relation to the Southeast and South, which showed similar distribution.

Table 1 - Distribution of subjects with cleft lip and/or palate according to Brazilian regions*

	Cleft type		
Region	**CL (%)**	**CLP (%)**	**CP (%)**
Northeast	4 (6.9)	41 (70.7)	13 (22.4)
South	25 (22.7)	67 (60.9)	18 (16.4)
Southeast	9 (19.1)	24 (51.1)	14 (29.8)
Total	38 (17.7)	132 (61.4)	45 (20.9)

CL = cleft lip; CLP = cleft lip and palate; CP = cleft palate.
* Chi-square test ($p = 0.040$).

Regarding time of diagnosis, clefting was diagnosed after birth in the maternity ward in 75.3% (162) of cases; during the prenatal period in 14% (30); and after hospital discharge in 10.2% (22).

Data analysis demonstrated significant differences between time of diagnosis and cleft type ($p < 0.0001$) (Table 2).

The diagnosis of CL occurred mainly during the prenatal period, and that of CP mainly in the maternity ward or after hospital discharge ($p = 0.007$). Among cases detected in the maternity ward and after hospital discharge, in the latter, almost all patients had a diagnosis of CP ($p < 0.0001$).

Regarding socioeconomic status, the average monthly family income was 3.72 times the minimum wage in the Southeast, 2.22 in the South, and 1.57 in the Northeast. Participants from the Southeast reported higher incomes than those interviewed in the Northeast and in the South ($p < 0.001$). The Northeast showed a trend toward lower incomes than the South, but without statistical significance ($p = 0.080$).

An analysis of the effect of monthly family income on the time of diagnosis revealed that the average monthly income of cases diagnosed during the prenatal period was significantly higher than that of cases diagnosed in the maternity ward or after discharge ($p < 0.0001$). However, among cases of postnatal diagnosis, there was no difference between cases diagnosed in the maternity ward and after discharge ($p = 0.6223$).

In 81.9% (176) of cases the physician was the professional to disclose the presence of oral clefts to the parents; the nurse in 8.8% (19); family members or friends in 4.7% (10); and other professionals in 4.2% (9). In 0.5% (1) of cases, the information was not disclosed.

Regarding time of diagnosis per region, TOCs were diagnosed in the maternity ward in 81.8% (90) of cases in the South, in 74.4% (43) in the Northeast, and in 61.7% (29) in the Southeast (Table 3). No significant differences were observed when time of diagnosis and regions were correlated ($p = 0.005$). Prenatal diagnosis was significantly higher in the Southeast, whereas diagnosis in the maternity ward was higher in the Northeast ($p = 0.007$). There was no significant difference ($p = 0.094$) between regions when diagnosis in the maternity ward and after discharge were correlated.

Lip surgery was performed in 87.65% (149) out of 170 subjects with cleft lip (38) or cleft lip and palate (132). Half of the cases diagnosed during the prenatal period, in the maternity ward, and after hospital discharge underwent surgery, on average, at 4.5, 6, and 9 months of age, respectively. However, the difference found in age at lip

Table 2 - Distribution of subjects with cleft lip and/or palate according to time of diagnosis*

	Cleft type		
Time of diagnosis	**CL (%)**	**CLP (%)**	**CP (%)**
Prenatal care	11 (29)	19 (63.3)	0 (0)
Maternity ward	27 (71)	110 (84)	25 (55.5)
After hospital discharge	0 (0)	2 (1.5)	20 (44.4)
Total	38 (100)	131 (100)	45 (100)

CL = cleft lip; CLP = cleft lip and palate; CP = cleft palate.
* Chi-square test ($p < 0.0001$).

Table 3 - Distribution of subjects with cleft lip and/or palate according to Brazilian regions and time of diagnosis*

	Time of diagnosis			
Region	**Prenatal care (%)**	**Maternity ward (%)**	**After hospital discharge (%)**	**Total (%)**
Northeast	4 (7.1)	43 (74.4)	10 (17.6)	57 (100)
South	13 (11.8)	90 (81.8)	7 (6.4)	110 (100)
Southeast	13 (27.7)	29 (61.7)	5 (10.6)	47 (100)

* Chi-square test ($p = 0.005$).

surgery between groups at different times of diagnosis was not significant (p = 0.185).

Palate surgery was performed in 69.5% (123) out of 177 cases with cleft palate (45) or cleft lip and palate (132). Half of the cases diagnosed during the prenatal period, in the maternity ward, and after hospital discharge underwent surgery, on average, at 14, 19, and 16 months of age, respectively. There was no significant difference in age at palate surgery between groups at different times of diagnosis (p = 0.937).

Of 312 clefts eligible for surgical correction in individuals who, according to the parents, had the minimum age to undergo surgery (162 between CL and CLP; 150 between CP and CLP), 51.11% (160) were not corrected within the scheduled time. The reasons were: other diseases (mainly ear infections and pneumonia) in 22.5% (36) of cases, service problems (scheduling delays, lack of beds) in 20.62% (33), anemia in 18.12% (29), and poor weight gain in 18.12% (29). The centers followed different surgical protocols; thus, delay to surgery was based on information obtained from parents or legal guardians, according to surgical planning at each service.

Discussion

This is the first multicenter study conducted within the Skull/Face Project Brazil, which aims to contribute to the improvement of care delivered to individuals with craniofacial anomalies. Health care facilities located in the Southeast, South, and Northeast of Brazil participated in this study. Centers located in the North and Midwest were not interested in participating despite attempts to contact them. Coincidentally, these are the regions providing less specialized care.[19]

There was a prevalence of CLP in the three regions, which is consistent with data published by Mossey & Little.[3] Similar results were found by Loffredo et al.[20] and by Nunes et al.,[21] who also detected a larger proportion of cases among men,[3] in agreement with our findings.

There was a prevalence of clefts alone, and in 14.58% of cases clefts were associated with other anomalies. This result is consistent with the findings by Cohen et al.[7] and Mossey & Little,[3] who revealed a mean of 15% of syndromic clefts. A Brazilian study conducted in the city of São José dos Campos, southeastern Brazil, found 9.1% of syndromic clefts.[22]

In the Northeast, there were 23 cases without an established diagnosis. Difficult access to a geneticist has already been documented by a previous study within the Skull/Face Project Brazil.[19] That health care facility provides care to the population of the state of Ceará, northeastern Brazil, and has a geneticist on staff, but staff members have no direct access to genetic laboratory tests, factors that delay diagnosis. Moreover, dysmorphologic evaluation is an evolutionary process and may require investigation of other organs and systems and monitoring of neuropsychomotor conditions before a diagnostic conclusion is reached.[11,18]

TOCs were diagnosed in the maternity ward in 75.3% of cases. A similar result was reported by Di Ninno et al.,[23] who found 80% of postnatal diagnosis in a sample from the city of Belo Horizonte, southeastern Brazil.

Only 17.64% of individuals with CL and CLP were diagnosed during the prenatal period, and 10.2% of affected subjects, mainly with CP, were diagnosed after hospital discharge. Thus, such finding reinforces the need for attention to an ultrasound diagnosis during prenatal care, particularly of CL and CLP, and pediatric evaluation during the first examination of the newborn.[17]

Examination of oral structures is essential, especially due to difficulties in feeding experienced by individuals with TOCs. Such findings require prompt intervention, such as appropriate feeding practices and guidance on posture and oral hygiene, thus ensuring adequate nutrition and weight gain.[13]

The prenatal diagnosis found in only a few cases in our sample is consistent with that found by Di Ninno et al.[23] Jones[24] believes that this diagnosis may cause emotional problems for parents. Di Ninno et al.[23] and Johnson & Sandy[25] concluded that prenatal diagnosis is beneficial and that families want to be informed of the results. This diagnosis allows parental counseling and effective postnatal surgical, feeding, and treatment planning.[26]

Bunduki et al.[11] state that, after the diagnosis is confirmed by ultrasound, parents should be referred to specific follow-up programs in reference centers. According to Chitty & Griffin,[16] findings of structural or chromosomal abnormalities determine prognosis and indications for chromosome investigation and fetal echocardiography.

CL can be easily diagnosed by ultrasound, whereas CP is diagnosed more often after birth by clinical examination of the newborn.[9,10,12] These findings were confirmed in our study, with significance between cleft type and time of diagnosis.

Although there was an overall prevalence of diagnosis in the maternity ward across the three regions, prenatal diagnosis was more frequent in the Southeast. The average monthly family income of cases diagnosed during the prenatal period was significantly higher than that of cases diagnosed in the maternity ward or after discharge. This finding suggests that the higher income of southeastern families allowed greater access to prenatal care and ultrasound examination.

In our study, the physician was the main person responsible for delivering information on the presence of clefts to parents, followed by the nurse. Di Ninno et al.[27] concluded that health professionals have a generally low

level of knowledge on TOCs. As a result, many parents return home with unanswered questions. These aspects were also considered deficient by Schardosim et al.,[28] who suggested that health professionals should have greater commitment to provide comfort and helpful guidance to parents. Diagnosis disclosure by family members, friends, and other professionals found in 19 cases in our sample reflects the lack of training among health care teams to address a common birth defect associated with high morbidity. In addition to technical clarification by the health care team, emotional support to family members is also of paramount importance.[18]

The delay to surgery observed in our sample was mostly due to factors related to the health status of affected subjects (anemia, diseases, poor weight gain), in addition to service problems (scheduling delays, lack of beds). Amstalden-Mendes et al.[13] showed similar results, with poor weight gain as the main factor leading to delay to surgery in their sample, reinforcing that attention to nutritional status is crucial to surgical correction within appropriate time. Other important aspects in approaching subjects with TOCs include the correct indication of feeding practices, care provided by specialized staff, and referral of patients to reference centers.[13,29]

In fact, a Brazilian study of normal newborn infants found that preventive measures are likely to minimize interruption of exclusive breastfeeding in the infant's first month of life.[30] These measures, together with a longitudinal follow-up performed until the establishment of feeding practices and other resources tailored to the peculiarities of each infant with clefts, may promote appropriate weight gain.

Age of affected individuals at corrective surgery was not dependent on early diagnosis of TOCs. Although necessary, surgical correction is not the only treatment. Early diagnosis allows contact of parents with specialists, favors early introduction of different feeding practices, preventing weaning whenever possible, and facilitates overall treatment planning and neonatal care tailored to the peculiarities of this congenital defect.

Despite the need for a highly complex structure for rehabilitation treatment, child care and treatment of morbid complications should be carried out in primary and secondary health care facilities,[13] which justifies staff training and organization of the public health system in order to improve attention to this important group of congenital defects.

Across Brazilian regions, clefts appear to be more often diagnosed in the maternity ward. Therefore, hospital health care teams should be able to provide initial care, thus contributing to future health care required. Considering the prevalence and morbidity associated with TOCs, staff training is suggested for multidisciplinary management of affected subjects as part of health care policies, starting at the maternity unit.

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