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Imaging Prenatal Diagnosis of Cleft Lip and Palate in Brazil: Frequency and Familial Impact

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ABSTRACT

Background and aim: The ultrasound evaluation of the fetal face for the detection of orofacial clefts is the most common diagnosis. This study analyzed the frequency of patients whose orofacial clefts diagnosed prenatally. Concordance of prenatal diagnosis and postnatal outcomes, and the impact of prenatal diagnosis.

Materials and methods: The sample was composed of 200 children (age of 0-36 months) relatives, with any orofacial cleft, regardless of ethnicity, gender, or socioeconomic background. A self-administered questionnaire responded, and descriptive statistics and Fisher exact test analyzed the results.

Results: Among the 200 children, 25.5% had diagnosed prenatally. There was concordance between prenatal diagnosis and outcome in 62.7% of cases, which is similar for all clefts (p=0.81). Less than entirely accurate prenatal diagnosis occurred in 37.2%. Under the 51 families that had a prenatal diagnosis, 66.6% considered the opportunity as favorable. Among the convenient reasons, 58.8% of the families mentioned the possibility to prepare themselves.

Conclusion: One-quarter of patients diagnosed prenatally and less than entirely accurate findings occurred in more than a third of the cases diagnosed. Despite that, families often report prenatal diagnosis as helpful for the acceptance of the condition and psychological preparation before the child is born.

1. Introduction

It is known that more than 20% of pregnancies of fetuses with congenital anomalies end in spontaneous abortion; the remaining 80% are either liveborn or stillborn, resulting in 3% to 5% of newborns with congenital disabilities[1]. Among the diverse fetal malformations, 90% occur in babies born to parents without a known risk factor. Thus the prenatal follow-up with ultrasound examination is recommended for the entire population of pregnant women[2]. Ultrasound examination is a useful diagnostic screening method in the prenatal period for identifying anomalies and is fundamental for neonatal and obstetric counseling and planning[3]The inclusion of ultrasound examinations in the prenatal period for the detection of orofacial clefts was first reported in 1981[4]. The accuracy of ultrasonography for prenatal diagnosis of CLP is correlated with the experience of the sonologist, maternal body type, fetal position, the amount of amniotic fluid, and the type of cleft[5]. Nowadays, three-dimensional (3D) ultrasonography[6], 3D-printed models[7], and prenatal magnetic resonance imaging (MRI)[8] are playing an important role in the accuracy of prenatal diagnosis of orofacial clefts.

Since then, the ultrasound evaluation of the fetal face for the detection of orofacial clefts has received increasing attention, and several classification systems have developed. In general, the type of orofacial cleft determined by the location (midline, unilateral or bilateral) and extent (an only lip, lip, and palate, or single palate)[9]. With this examination, the visualization of the nose and lips is possible around 15 weeks of pregnancy. Christ and Meininger4 reported that cleft lip and palate might have detected between 28 and 33 weeks of pregnancy, yet the study of Rey-Bellet and Hohlfeld[10] mentioned that the cleft might have observed as early as 12 weeks of pregnancy.

By the prenatal diagnosis of a cleft, all parents may prepare themselves to accept the child's condition and educate themselves about the future care of their child. Although there is no intrauterine treatment for CLP, prenatal diagnosis also allows mothers to prepared for a cleft's individual needs[11]. The proper and timely counseling helps instill a sense of preparedness for the family, which significantly improves the quality of postnatal treatment received by the child enabling a near-to-normal quality and standard of life[12]. The detection of an orofacial cleft may be shocking for the family. Parents of children with orofacial clefts face a period of coping, requiring the understanding of health professionals involved in this process[13,14]. Rey-Bellet and Hohlfeld[10] reported that informing the parents helps overcome the initial fear and feeling of impotence, with a better understanding of the child's problem.

Considering the lack of information in the literature on the rate of prenatal diagnosis of cleft lip and/or palate, this study evaluated the percentage of individuals assisted at a tertiary rehabilitation center whose cleft had

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diagnosed during the prenatal period. We assessed the range of professionals involved in the diagnosis, the concordance of prenatal ultrasound and postnatal outcomes, and the impact of prenatal diagnosis for the family. The study analyzed the null hypotheses that (1) prenatal diagnosis would be similar in clefts affecting the lip, palate, or both; and (2) prenatal diagnosis would have a positive impact on the family.

2. Materials and methods

This study was approved by the Institutional Review Board (protocol n. 196/2011). All families were educated about the research and voluntarily signed an informed consent form agreeing to participate.

This was an observational, retrospective, cross-sectional study comprising application of a questionnaire, which was given to biological relatives of 200 children with cleft lip and/or palate attending a Brazilian tertiary care craniofacial center for surgical treatment or follow-up during the study period (October 2011 to January 2012), with any type of orofacial cleft, regardless of ethnicity, gender or socioeconomic background, aged 0 to 36 months. The sample size was calculated considering the highest percentage found in the literature (88% by Liou et al., 2011[15]), assuming a range of error of 5% and confidence interval of 95%, which yielded a sample size of 162 participants.

The individuals were invited to participate consecutively, as they attended the institution for routine care. All invited individuals agreed to participate. Children attending the hospital with adoptive parents or relatives that could not provide detailed information on the prenatal period were not invited to participate.

The questionnaire was adapted from a study conducted in the United States by Strauss16 and contained open and multiple-choice questions addressing prenatal diagnosis's social aspects. The questionnaire was translated into Portuguese, adapted for cultural reasons (especially eliminating questions related to pregnancy termination, which is not allowed in the country), and initially applied it to 30 caretakers to check the understanding of the questions. During this stage, the examiner observed that the questions were clearly understood, and the participants had no doubts about it. A single investigator conducted the recruitment of families. The questionnaires were parent self-reports and were completed in the clinic reception room. The researcher was available for the participants to elucidate any uncertainty about the questions asked. The actual type of cleft checked on the children's hospital records.

The results were evaluated using percentages and with descriptive statistics. The t-test and Fisher exact test analyzed comparison and associations between findings.

3. Results

Sample characteristics

The mean age of children was 18.7 months (standard deviation (SD) 11.27), ranging from 10 days to three years. Among the respondents, there were 185 mothers, six fathers, six grandmothers, and three aunts. The mean age of respondents was 32.8 years (SD 9.18), ranging from 17 to 71. The mean number of people living in the same house as at the child was 3.83 (SD 1.41), ranging from one to 10 people. The mean income of families was US$ 565.89 (SD 656.38), ranging from US$ 50.54 to US$ 5730.00, with three cases of unemployment. Further information on the sociodemographic characteristics of the sample presented in Figure 1.

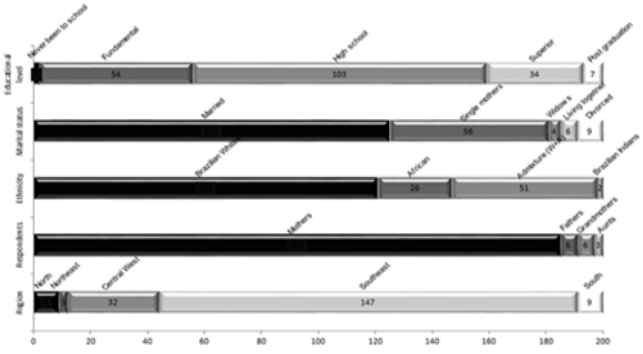

Figure 1. Sample distribution concerning the different sociodemographic aspects.

Cleft lip and palate, other anomalies and recurrence

Among the 200 children, 150 had siblings, ranging from one to nine, including two cases of twins and one case of triplets. A total of 75 participants reported other cases of cleft lip and/or palate in the family, affecting more than one relative in several cases, including 9 cases of orofacial clefts in mothers, four in fathers, 10 in siblings, 10 in grandmothers, two in grandfathers, two in great grandmothers, 18 in uncles and aunts, plus 44 cases in cousins of diverse degrees.

The cleft affected only the lip in 42 cases, the palate in 54 cases, and both lip and palate in 104 cases. Nineteen children presented with associated malformations, including tracheomalacia (1), agenesis of the corpus callosum (1), dwarfism (1), mandibular micrognathia (1), eye malformations (1), claw-like hands and feet (1), bone malformation with hydrocephaly (1), rare facial clefts (2), Pierre Robin sequence (6), and Down Syndrome (1), Treacher Collins Syndrome(1), Patau Syndrome (1) and Stickler (1) syndrome.

Prenatal care and diagnosis

Among the 200 children in the sample, 51 had been diagnosed prenatally. Besides these cases, two families stated they suspected that the doctor knew about the cleft on the prenatal examination yet did not inform them. The distribution of prenatal diagnosis about the number of prenatal consultations, ultrasound examinations, and pregnancy period on the determination presented in Figures 2 and 3.

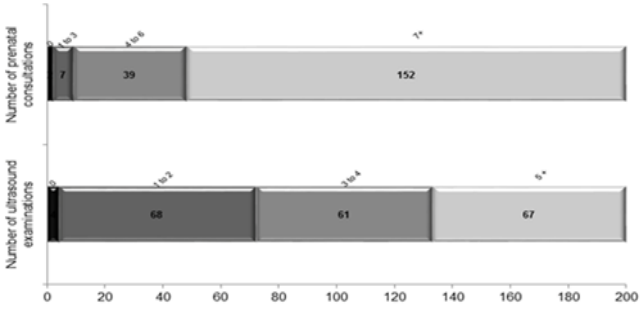

Figure 2. Sample distribution according to prenatal care.

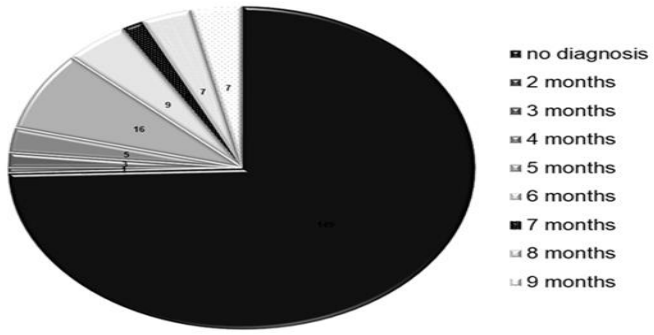

Figure 3. Period of prenatal diagnosis.

There was no association between prenatal diagnosis or orofacial cleft and mean family income (p=0.43), several prenatal consultations (p=0.27), and several ultrasound examinations (p=0.20). There was an association between the prenatal diagnosis of an orofacial cleft and the type of cleft (p=0.000, Figure 4).

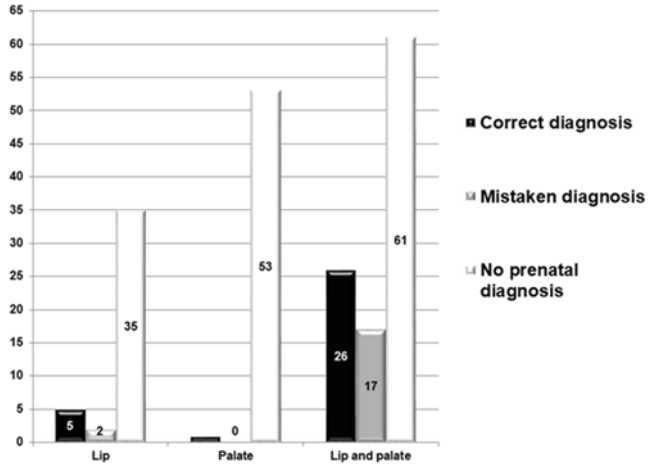

Figure 4. Distribution of individuals as to the type of cleft, prenatal diagnosis, and concordance between the type of cleft diagnosed prenatally and the postnatal outcome.

There was concordance between the type of cleft diagnosed prenatally and the postnatal outcome in 32 of 51 cases, without a statistically significant relationship with the type of cleft (p=0.81, Figure 4). In fourteen cases of cleft lip and palate, the families reported prenatal diagnosis only of cleft lip. Also, two cases of unilateral cleft lip and palate had been diagnosed as bilateral, one case of bilateral cleft lip and palate had been diagnosed as unilateral, and two cases of isolated cleft lip had been diagnosed as cleft lip and palate.

Family perception of prenatal diagnosis

One family did not reply to some questions of this section because the mother said she could not remember; thus, some items were responded only by the families of 50 individuals.

Among the 51 cases with prenatal diagnosis, most considered the opportunity as good (34), followed by bad (8), very good (7), and very bad (1). Not all families provided the reasons to consider the prenatal diagnosis as good or bad. Among the favorable reasons, the families mentioned the possibility to prepare themselves (28 cases), get information on the issue (2), one mother reported she had been expecting the prenatal diagnosis because she also had a cleft, one mother prepared to undergo a tubal ligation with the child's delivery due to the diagnosis. One family requested the possibility of attending a craniofacial center consultation to receive information before the child was born. The families that considered the prenatal diagnosis as bad or very bad mentioned the impossibility of taking any measure before birth (3), fear (1), not finding treatment at their cities (1), one mother said she felt ill during her pregnancy because of the diagnosis, and one reported occurrence of hemorrhage and the need to remove her uterus and ovaries during delivery, which she assigned to the psychological stress caused by the prenatal diagnosis.

4. Discussion

Considering the present results, the first hypothesis was rejected. The second hypothesis was accepted, since (1) no statistically significant difference was found for prenatal diagnosis between different types of clefts; and (2) most families considered the prenatal diagnosis as a pleasant experience. Notwithstanding, some slight differences were observed between types of clefts, and a small proportion of families found the prenatal diagnosis a negative experience. The prenatal diagnosis of cleft lip and palate is essential because it allows for informed counseling of biological parents and the planning of postnatal treatment[17]. The high frequency of other cleft cases in the family of these children (75 reports), affecting relatives of different degrees, confirms reports of the family history of cleft[18].

Nineteen children in the sample presented additional malformations, associated or not with syndromes. Babcook et al.[9] and Nyberg et al.[19] highlighted the importance of identifying the specific type of orofacial cleft, due to the strong correlation between some types of cleft and the presence of other fetal anomalies. A reliable diagnostic method to distinguish these anomalies is essential because some countries allow the family to choose to interrupt the pregnancy. When the family decides to continue the pregnancy, the prenatal diagnosis offers an excellent opportunity for emotional and psychological preparation before birth[13,14].

Despite the high level of prenatal care demonstrated by the number of professional consultations and ultrasound examinations in this sample, there was a relatively low prevalence of prenatal diagnosis (25.5%) compared to higher rates in other studies, of up to 88% [15]. Besides other factors, this may be related to the utilization of bidimensional ultrasound in the Brazilian health system, since the three-dimensional ultrasound allows better visualization of orofacial clefts[20].

Unlike North American and European countries and similar to other Latin American countries, abortion is legally forbidden in Brazil.[21]Recently, abortion has been allowed only in cases of prenatal diagnosis of anencephaly confirmed by two independent sonographers.. Therefore, this aspect was not addressed in this investigation; thus, this differs from other studies on this subject[22]. The percentage of terminations by abortion varies between countries according to the malformation involved, being up to 100% in cases of chromosomal anomalies, metabolic disorders, and spina bifida in countries as Australia, Switzerland, and United States[23]. It is estimated that 1,800 legal pregnancy terminations are performed due to congenital anomalies per year in the United Kingdom[24]. Comparatively, the percentage of completion after prenatal diagnosis of cleft lip and palate is lower[25], being more significant in the presence of multiple associated anomalies[26,27].

In this study, no association was observed between prenatal diagnosis and mean family income, number of prenatal consultations, and number of ultrasound examinations, demonstrating a similar possibility of prenatal diagnosis regardless of the frequency of prenatal care visits. The study was conducted in a public health center assisting individuals of all socioeconomic backgrounds, as evidenced by the variable income. The type of cleft

diagnosed prenatally was confirmed at birth in 32 of 51 cases, without differences between cleft types. In fourteen cases of cleft lip and palate, the prenatal diagnosis was reported only for cleft lip. Amazingly, two children with cleft lip had been prenatally diagnosed with cleft lip and palate, suggesting a possible bias on the sonographer when a cleft lip is detected.

In this study, among cases with prenatal diagnosis, most families considered the experience as generally positive, especially since it allowed for the possibility for the family to prepare themselves. The information on the extent of the defect and absence of associated anomalies should always be offered whenever possible to enhance parent counseling. Counseling should include referral to treatment programs or multidisciplinary teams28. However, considering that this was a retrospective study, there may have been inherent biases concerning the reliability of outcomes, especially regarding the acuity of respondents' memories about the prenatal follow-up, and the emotional aspect involved since all respondents were relatives of young children still undergoing major steps of their rehabilitation process.

Counseling and guidance of parents by the medical team at the time of diagnosis enhances the psychological aspect of treatment and leads to a positive approach. Contact with knowledgeable professionals about the treatment process helps decrease anxiety[29].

According to Rey-Bellet and Hohlfeld[10] and Marilyn and Jones[30], counseling during the prenatal period is fundamental for the families, corroborated in this study. The parents appreciate the availability of other parents who had been through the same situation and especially the possibility to share their experiences, feeling that they are not alone in that situation.

This study revealed that essential sources of information for the families after the diagnosis include the craniofacial centers, followed by other health professionals, the internet, different families of children with cleft lip and palate, and associations of parents and individuals with clefts. This study demonstrates the wide variability of sources of information and support mobilized by the families soon after the orofacial cleft diagnosis. This study reinforces the need to establish solid strategies for support and information to affected families. Such support should be widely offered and diffused throughout public and private systems of prenatal and perinatal care.

It should be emphasized that the family's reactions to the birth of a child with special needs are widely variable depending not only on the birth country but also on cultural and religious beliefs, previous experiences of the family, besides a myriad of factors. Therefore, the family's comments presented in this manuscript may not necessarily reflect the opinions observed in other communities, either locally or worldwide.

5. Conclusion

The prenatal detection of cleft lip and palate is still low in Brazil, especially for cleft palate alone. Prenatal ultrasound diagnosis aids families' psychological preparation, allows for the referral of pregnant women to specialized centers, for follow-up by a multidisciplinary team, and the planning of the child's delivery.

Conflict of Interest

The authors declared that there is no conflict of interest.

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