Peer reviewed article

Prenatal diagnosis of facial clefts: evaluation of a specialised counselling

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Summary

Questions under study/principles: Prenatal diagnosis of cleft lip and palate has raised many questions concerning the best way to inform the parents and offer appropriate support during the remaining pregnancy to help prepare a positive birth experience. Prenatal counselling differs according to who is responsible, and to that person's knowledge of the practical aspects of care and the impact of the malformation on the child's quality of life. The aim of the study was to evaluate our current techniques and modify them when appropriate.

Methods: 29 couples having experienced prenatal diagnosis were asked to respond retrospectively to a questionnaire. Personal obstetric and family history, timing and quality of information provided and their impressions at birth were considered.

Results: The parents used several sources of information: the obstetrician, the counsel by the cleft team surgeon, the parents support groups and the Internet. 93% of the parents felt well prepared psychologically for the birth of their child and concerning the practical aspects of care. 54% felt relieved that their child was less affected than imagined. 96% considered prenatal diagnosis a benefit.

Conclusion: Basic information should be provided soon after ultrasound confirmation, followed by progressively more detailed technical discussions over the remaining pregnancy.

Key words: cleft lip and palate; prenatal diagnosis; counselling

Introduction

Cleft lip with or without an associated cleft palate (CL/P) is a relatively common congenital birth defect occurring in about 1:750 births in the European population. There exist many different types of clefts, from isolated partial cleft lip or isolated velar (soft palate) cleft to complete unilateral or bilateral cleft (lip, alveolus and hard and soft palate), including many intermediate variations. Prenatal diagnosis of cleft lip by ultrasound scan can be performed as early as 12 weeks into pregnancy, and the child's prognosis is generally considered to be dependent upon the presence and type of associated anomalies. Amniocentesis is generally proposed for karyotype determination as well as a complete morphological work-up in order to inform the future parents completely and correctly.

When the ultrasonographer discovers a cleft, he or she usually turns to the local multidisciplinary cleft team for counsel and care of the expectant parents. This prenatal collaboration is useful since care is adapted to individual needs according to the type of cleft. The quality of the counselling is of great importance for many reasons. First, the discovery of any malformation is a shock and disappointment to the parents, and initiates the grieving process necessary for the ultimate acceptance of their different child. Second, most of the general population has very little knowledge of the malformation and what they do know is often biased by negative feelings: the memory of a classmate with a facial scar or who had abnormal speech, or who was inhibited in his behaviour. Parents are afraid for many reasons, and the aim of prenatal counselling is to provide them with a clear vision of what can and will be done for their child so that he can live a fulfilling life. The aim of this study was to retrospectively analyse our counselling of the parents, to identify weak points and provide propositions. The question of course is no longer as it once was "Should we tell the parents?" but how and when, in order to keep the burden of the prenatal diagnosis from being a "poisoned gift".

No financial support declared.

Patients and methods

Since 1989, the Obstetrics and the Paediatric Surgery departments of the University Hospital in Lausanne have collaborated in the counselling of parents whose child to be born presents a malformation. Those presenting a cleft anomaly are referred to the paediatric surgeon specialised in cleft surgery leading the multidisciplinary team who will evaluate and provide care for the child composed of a specialised orthodontist, speech therapists, paediatric ENT specialists, a craniofacial surgeon and a psychiatrist.

Counselling consisted of discussions with both parents and lasted an average of 2 hours. The following aspects were considered:

- Types of clefts and possible causes
- In case of a cleft palate: feeding procedures, description of the orthodontic plate and its manipulation, breastfeeding techniques.
- Surgical correction, timing and operations, hospital stay details (duration, rooming-in)
- Multidisciplinary follow-up: speech, ear problems, orthodontics, and psychology
- Insurance coverage
- Before and after surgery photos of children with similar clefts
- Written information booklet, team website address and brochure from parent support group
- Offer of psychological counselling by specialist

A second session was planned 4 weeks later and grandparents and siblings were invited to participate according to the parents' wish. Appointments were then planned every 4 weeks until birth. It was strongly advised not to modify the planned birth place, and the parents were reassured that the surgeon would be available on the phone and visit within 48 hours of the birth.

The first postnatal visit took place in the hospital where the child was born. Confirmation of the morphological type of cleft was done and explanations concerning the feeding techniques given to the parents and the nursing staff. Surgical repair of the cleft was described and once again, before and after photos of children presenting the same type of cleft were shown.

From 1989 to 2001 thirty-seven children born with facial clefts had been diagnosed by prenatal ultrasound scan. Two early cases did not benefit from prenatal counselling because the referring obstetricians decided not to inform the parents. In this retrospective study, all 35 families were sent an evaluation questionnaire to be completed at home and returned anonymously. The questionnaire was made of 70 semi-open questions concerning the pregnancy, circumstances surrounding the diagnosis, information they had received and their reaction to it, care of the newborn and their own propositions towards improving the quality of the prenatal counselling. For each question the parents could answer as a couple or separately. The author of the study was presented as a resident in paediatric surgery conducting a quality-control study of prenatal care and it was specified that all responses would remain strictly anonymous. Parents had the opportunity to contact the author to clarify certain questions when necessary. A psychologist was made available to the parents if they felt that participation in the study revived negative feelings or conflicts within the couple. Out of the 35 couples 1 had left the country, one refused without offering a reason and 4 did not participate due to language comprehension difficulties. The results of the study are therefore based on 29 questionnaires. This study was approved by the local Ethics Committee.

Results

Among the 29 children included in this study, 17 were boys and 12 girls. Ten (35%) were born in the university hospital, 18 (62%) in a regional or private hospital and 1 (3%) at home. Three mothers (10%) had decided to change the place of birth and delivered in the referral centre.

In 13 cases (44%) the mother was a primipara. Mean maternal age was 31 years (range: 23–42) and mean parity was one (range 0–3). The mean gestational age at the time of diagnosis was 23 weeks (range 16–36). In 19 cases (66%), diagnosis was obtained during routine ultrasound screening. The remaining 10 were performed because of positive family history of facial cleft or another malformation (n = 3), obstetric pathology (n = 6) or motor vehicle accident (n = 1). The total number of ultrasounds performed during pregnancy varied from 3 to 10 (mean 6) depending upon the gestational age at diagnosis and the associated anomalies. Twenty couples (68%) were referred to the university centre for confirmation. Fifteen patients (52%) underwent amniocentesis, fourteen declined the offer, and one fœtus underwent MRI for suspicion and confirmation of familial median cleft.

Ten cases (34%) had a positive family history of facial cleft, and in 4/10 it was a first-degree relation (parent or sibling). In another 9 cases (31%) there was a positive family history for another congenital malformation not related to facial clefts.

The cleft was bilateral in 9 cases (31%), 6 of which involved the lip, alveolus and hard and soft palate (CLP), one cleft lip and alveolus (CLA), one isolated cleft lip (CL) and one oro-ocular cleft (Tessier IV). The cleft was unilateral in 18 cases (61%), with 13 CLP, 2 CLA, and 3 CL. The study also included one isolated cleft palate and one median cleft.

Associated malformations were observed in 7 children (24%) and included isolated cases of renal dysplasia, interventricular or interauricular communication, Simpson-Galabi-Behmen syndrome, Van der Woude syndrome, congenital deafness and congenital cataract. Only the diagnoses of renal dysplasia, Simpson-Galabi-Behmen and Van der Woude syndromes were made before birth.

In 62% of the cases, the private obstetrician announced the diagnosis, and the remaining 38% received the information from the ultrasonographer performing the referral examination. The

majority of the parents (93%) said they had trusted the diagnosis, and 4 (14%) asked for a second opinion. Only one (3.4%) couple said they had not wanted to know at the time and regretted having been informed at all, even later.

Parental reactions at the time of diagnosis varied greatly but most expressed severe psychological shock. Feelings of guilt, anxiety, fear and sadness were common, and some couples admitted feeling revolted and "destroyed". Four admitted initial rejection (three considering pregnancy termination without proceeding with the request) and three feared the child would suffer. Concerning medical details, parents expressed curiosity as well as incomprehension (n = 3), trust (n = 3) and often relief (n = 5) that the malformation was not life-threatening. Only two couples solicited psychological support, but five others later expressed regrets for missing the opportunity.

The first contact with the specialist was organised in 38% of the cases by the mother's obstetrician, in 52% by the ultrasonographer and in 10% by friends or acquaintances. Time between the diagnosis and the first meeting was extremely variable, from a few hours to more than a month, with a majority (59%) within two weeks. This interval was deemed adequate for 73% and too long for 27%. The same interval was considered acceptable for certain patients and too long for others, different opinions co-existing within couples. The first contact lasted an average of 2 hours, but varied from one to four hours. On average, parents had the opportunity to meet the surgeon again once or twice during the pregnancy depending on the gestational age at diagnosis. After the first discussion, parents were free to cancel or reschedule according to their individual needs, and 2 did cancel, feeling they had enough general information. In spite of the differences, 86% of the parents thought the number of discussions was adequate and that they were very important.

The specialist's availability was considered satisfactory by 95% of the parents. All couples felt they had obtained clear answers to their questions concerning the cleft's aspect and aetiology. Fourteen percent felt they had not received definitive answers to their questions concerning feeding due to the possibility of a cleft palate, and that this had worried them. It had been explained that the existence of a cleft palate was impossible to confirm or exclude based on 2-D ultrasound and for some parents (34%) this was a source of severe anxiety.

All families were satisfied by the opportunity to see drawings of the surgical procedures and photographs of children presenting different forms of clefts before and after surgical repair. Two couples (7%) said afterwards that they disliked some of the medical close-up pictures that were described as shocking. The operations and their timing based on anesthesiological and future growth considerations were well understood by 97% of the parents. However, 21% would have preferred an earlier lip correction to avoid difficult social situations for themselves.

For 17% of the parents, Internet was a good way to obtain further information. Twenty-one (71%) of the mothers did not think that the visit of the specialised nurse or midwife at home in the post-partum period helped with feeding problems. Meeting other parents and their children was often (79%) considered useful and reassuring.

Overall, 93% of the parents considered that they had been well prepared for the birth. Seventeen (59%) couples were relieved to see their child at birth and see that the malformation seemed much less serious than they had imagined. Three couples (10%) had thought about pregnancy termination before prenatal counselling, but decided against it afterwards. Seven families admitted that the experience of having a child with a facial cleft had changed their minds regarding the size of the family, principally because of the energy, time and organisation required in the post-natal period.

Discussion

Overall, in this study, prenatal counselling satisfied the parents who were informed of the presence of a facial cleft during the antenatal period. It seemed important that the study be conducted by a non-member of the cleft team so that the parents could feel free of feelings of loyalty to the team who treated their child and give more critically constructive answers. Presenting the author as a paediatric surgeon in training encouraged parents to answer candidly, informing someone who solicited their expertise as parents rather than answering research questions from a professional psychologist. Ninety-three percent of the parents said they trusted the diagnosis and only 14% asked for a second opinion. Diagnosis was made between 16 and 36 weeks of gestation, and the two cases diagnosed at 16 weeks were bilateral forms, confirming previous studies [1–3] showing that severe forms are diagnosed earlier. Associated malformations were found in 7/29 infants (24%), as compared to 43% in Fischer's study [4], 21% in Cockell's series [5] and 45% in Perrotin's [1].

Two families had to be excluded from the study since they had not been informed in spite of a positive diagnosis for a cleft. Both mothers thought that this was probably done in their so-called best interest to avoid upsetting them, and insisted that the prenatal diagnosis should be given to avoid the shock of discovery at birth and the feeling of being betrayed by their obstetrician. In Belgium accord-

ing to Maes in 1998 [6], it seems the diagnosis is not systematically given for fear that the psychological shock could be harmful to the pregnancy. It is now widely accepted that the parents must be informed of any finding. One couple said they would have preferred not to have known the diagnosis before birth. They thought that the information kept them from enjoying the rest of the pregnancy and the mother needed psychiatric support.

In our cases, the diagnosis was given to the parents by the gynaecologist or by the referral ultrasonographer. Explanations given at the time were mostly simple and reassuring, according to most parents. Many parents (38%) said they felt shocked when first informed of the diagnosis. Some parents felt revolted and three couples (10%) said they had considered pregnancy termination but none requested one. These results differ from those of Bronshtein [7] describing 14 cases of termination in 15 prenatally diagnosed cases. Life conditions, culture and available care facilities probably contributed to these differences, but like others [3, 8] we are convinced that good counselling helps avoid unnecessary pregnancy termination. Fear that the child would suffer in 10% of the parents shows that they need to be informed that the cleft is not an open wound, but surfaces covered with skin or mucosa having normal sensitivity. In our study, most parents who had previously seen a person with a facial cleft remembered a very visible scar, a person with normal growth and intelligence but with a strange voice and suffering from social exclusion. Parents not having known anyone before, often (51%) feared that the child's intelligence would be impaired.

The first meeting occurred within 2 weeks of the diagnosis for 58% of the parents. Some parents considered a 2 days delay as too long, whereas others were satisfied to have had a week to "digest" the news and to prepare their questions. Some parents wanted to talk, share their frustration and anger, reassure themselves and obtain precise information immediately in order to cope with questions from family and friends. Others preferred to gather information themselves and see the specialist when under less stress, being better able to listen and understand. The parents asked two types of questions: vital, urgent ones requiring immediate short answers concerning mostly survival and feeding, followed by more precise technical ones that appeared after the initial discussion and concerned surgical procedures and treatment of sequelae. An ideal time for the first consultation does not appear to exist, but it seems important to have a first meeting quickly and then remain available for the parents, responding to their individual needs. The delay between the diagnosis and the specialised counselling was judged satisfactory by 73% and the number of prenatal consultations as sufficient for 86%, although the same delay or number of consultations was evaluated differently by different couples, hence, reflecting very different needs. Finally, 93% of the parents felt well prepared for the birth.

Counselling during the antenatal period was considered essential by all the parents; this finding has been confirmed by other groups [3, 5, 6, 9–11]. All parents appreciated availability and particularly the possibility of telephone conversations. The explanations were qualified as clear and reassuring. Parents felt listened to, understood and relieved to discover they were not alone in this situation. Questions arose between the consultations and written information allowed them to find quick answers. Written information is also helpful in informing siblings, family and friends. The question that never received a definitive answer before birth and remained a major source of worry was the possible association with a cleft palate. This explains why the subject of neonatal feeding was not always clear to the parents. The uncertainty surrounding the existence of an associated cleft palate might be diminished by the use of 3-dimensional imagery [12].

Positive family history can influence the parents' perception and the great differences between the present standards of care and the situation 20 to 40 years ago must be explained in detail. Contacts with the local parents' association was very much appreciated by the majority (79%) of the future parents, some parents (10%) however preferred not to participate in group meetings and felt that the malformation did not need to play a central role in their lives. Photographs of similar cases and drawings of the surgical procedures involved in the reconstruction helped 97% of the couples to better understand the various possible forms and the results after surgical correction. Seventeen (58%) couples were relieved at birth to see that the cleft was a less serious problem than they expected, showing that the imagined problem, isolated from the child as a whole, always seems worse than reality. Some parents expressed the fact that close-ups showing only the cleft can be very shocking and they preferred pictures of the child in his normal environment. Similarly, although ultrasound helps the parents understanding the nature of the defect [10, 13], it might be wise to exercise prudence with respect to the 3-D ultrasound now becoming more easily available.

Internet has become an important way for parents to obtain information, contacts and advice. It has become routine for us to provide a list of website addresses including our own, and many parents expressed satisfaction with the information provided.

Twenty-eight of the 29 couples thought that prenatal diagnosis had given them enough time to work through their feelings and accept the child at birth. They had time to warn and prepare family and friends, in particular grandparents and siblings. Even if this process is extremely difficult, the vast majority (96%) felt it was easier in the antenatal than in the postnatal period, as observed by others [3, 6, 9]. While all the parents were offered

professional psychiatric counsel, few contacted the referral specialist (7%) but several (17%) regretted afterwards not having done so. This raises the difficult question of how much encouragement should be given to overcome initial reticence towards psychological support.

Planned birthplace was only modified in favour of the referral centre in 3 cases. During the prenatal discussions, the parents were encouraged not to change their plans so as to not dramatise the event and to allow normal postnatal visits for family and friends. Among the mothers who did deliver in regional hospitals or private clinics, five (17%) deplored the lack of knowledge and tact of the medical staff. They also were often excluded from care taking and feeding and sometimes even separated from other mothers. All of these situations threatened the building up of a normal parent–child bond and were resented by the parents who felt they knew more about the care of their child than the professionals did.

Timing of the operations was well understood by the parents. Six out of 29 would have preferred earlier (before 5 months) surgical correction, admittedly for their own social comfort as observed by others [4, 13]. Facial clefts are still poorly understood malformations in the Swiss population. Reactions in public places often made parents wish the surgical repair could be done sooner. Clear explanations concerning the procedures and their impact on the long-term global result convinced all the parents to wait. Paradoxically, some mothers expressed distress after lip closure, because they had to readapt to the baby's new facial features and expressions. Many mothers insisted on the importance of including the parents in postnatal care and wanted to proceed with their usual care taking throughout post-operative hospital stay.

Seven (24%) couples said that their experience with a child having a cleft influenced their family planning. Trips, hospitalisations, surgery, orthodontic treatment, speech therapy, etc. are very demanding. These are described as major stress factors for the family as well as the concerned child in Bradbury's study [11]. Negative societal reactions and the great amount of energy needed for treatment [3, 9, 13] were cited as the principal reasons for limiting family size. However in our study 20 (69%) of the parents still considered the possibility of further pregnancies.

Early feeding problems could be attenuated with the help of a specialised nurse whose systematic postnatal visit would reassure the parents. She could also make sure that administrative steps with the state insurance are properly taken so that the parents worry less about the financial aspects of care.

In spite of rapid counselling, parents' expectations were not always satisfied. Their needs differ greatly and are difficult to assess. The satisfaction of the parents in our study may be more related to their impression of being listened to rather than to the intrinsic quality of the information provided. Empowering the parents by making them experts in cleft care as soon as possible helps them overcome their initial fear and sense of helplessness. In understanding their child's problem they can recover their role as an active caretaker.

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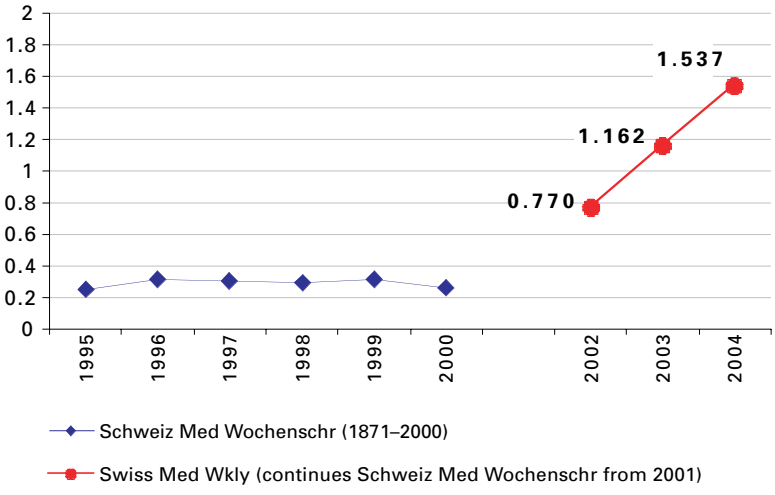

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