

A QUALITATIVE DESCRIPTION OF RECEIVING A DIAGNOSIS OF CLEFTING IN THE  
PRENATAL OR POSTNATAL PERIOD

by

Rachel H. Malinowski

BS, James Madison University, 2001

Submitted to the Graduate Faculty of  
Graduate School of Public Health in partial fulfillment  
of the requirements for the degree of  
Master of Science

University of Pittsburgh

2005

UNIVERSITY OF PITTSBURGH

Graduate School of Public Health

This thesis was presented

by

Rachel H. Malinowski

It was defended on

April 12, 2005

and approved by

Thesis Advisor:

Robin E. Grubs, PhD, CGC  
Assistant Professor of Human Genetics  
Co-Director, Genetic Counseling Program  
Department of Human Genetics  
Graduate School of Public Health  
University of Pittsburgh

Thesis Advisor:

Mary L. Marazita, PhD, FACMG  
Associate Dean for Research  
Head, Division of Oral Biology  
Director, Center for Craniofacial and Dental Genetics  
Professor, Oral and Maxillofacial Surgery  
School of Dental Medicine  
Professor, Human Genetics  
Graduate School of Public Health  
Professor, Psychiatry  
School of Medicine  
University of Pittsburgh

Committee Member:

Patricia I. Documét, MD, DrPH  
Visiting Assistant Professor  
Doctoral Program Coordinator  
Department of Behavioral and Community Health Sciences  
Graduate School of Public Health  
University of Pittsburgh

## ACKNOWLEDGEMENTS

The participants of this study deserve an enormous thank you for offering both their time and insight for this research study. I appreciate the frankness and honesty with which these participants spoke to me. The lessons I have learned from them reach far beyond the pages of this document. I am thankful for their participation; without them, this study could not have taken place.

I would also like to say “Thank you!” to the Cleft-Craniofacial Center, especially Dr. Joseph Losee, Dr. Carla Weidman and Mr. Matthew Ford for allowing me to conduct this study and for all their help with recruiting participants.

Thank you to my thesis committee, Dr. Patricia Documet, Dr. Robin Grubs and Dr. Mary Marazita, for their encouragement and ability to open my mind to new ideas. I thank Dr. Marazita for her support and for giving me the independence to conduct this study. A sincere and gracious thank you goes to Dr. Robin Grubs for her guidance and perseverance. She has taught me the value of qualitative research, a priceless gift.

To my classmates, you are true friends with hearts big enough to fill an ocean. We could not have made it through these two years without the ability to lean on one another. I know each of you will have an impact on your corner of the world. You have each certainly touched mine.

Mom, Dad and Danielle, your love and guidance have allowed me to fulfill my dream of becoming a genetic counselor. I would not be here today without the support of my incredible family.

To Curt, your forever optimism and sincere belief that I can do whatever I put my mind to have provided me with the motivation to succeed, day in and day out. Say goodbye to the Turnpike and hello to the next step of our lifelong journey together!

Robin E. Grubs, PhD, CGC  
Mary L. Marazita, PhD, FACMG

A QUALITATIVE DESCRIPTION OF RECEIVING A DIAGNOSIS OF CLEFTING IN THE  
PRENATAL OR POSTNATAL PERIOD

Rachel H. Malinowski, MS

University of Pittsburgh, 2005

**Background:** Advances in ultrasound technology have revolutionized obstetrical care and have resulted in a greater number of cases of cleft lip with or without cleft palate (CL+P) diagnosed prenatally by ultrasound. This study investigated the experience of receiving a diagnosis of clefting in the prenatal or postnatal period.

**Methods:** Open-ended interviews were conducted with 20 parents of children with CL+P. Of these parents, 12 experienced a prenatal diagnosis of their child's cleft, while eight received the diagnosis at birth. Interviews were transcribed and analyzed using a qualitative description approach with an emphasis on thematic analysis. Common themes emerged from participant's responses to questions regarding the delivery of the diagnosis, preparation for the birth of their child, advantages and disadvantages of prenatal diagnosis, use of the Internet, views on abortion, interaction with other parents, among other issues.

**Findings:** Preliminary findings were synthesized into themes that included "shared parental experiences," "coping," "preparation," "disadvantages," and "alternative perspectives." These overarching themes were divided into subthemes. Advantages cited for prenatal diagnosis included having the time to psychologically adjust to the diagnosis, to become informed, to educate other children, to opt for additional testing, and to plan for the baby's needs. Some participants felt a drawback of prenatal diagnosis was an emotional disruption of the pregnancy,

while other participants found no disadvantages. All participants in the prenatal group indicated they were glad they learned of the cleft before the birth of their child. Some participants in the postnatal group would have rather received the diagnosis prenatally, while others were satisfied learning of the diagnosis in the delivery room.

Interpretation: There seemed to be greater similarities than differences between the two groups of participants. Parents seemed to be affected more by how the diagnosis was delivered than the timing of the diagnosis. A prenatal diagnosis of a cleft may have a negative impact on the pregnancy, nonetheless parents seemed to want this information. High-resolution ultrasound has become standard of care for many pregnant women. Understanding the consequences of prenatal diagnosis is an important contribution to the field of public health.

## TABLE OF CONTENTS

1.	INTRODUCTION .....	1
2.	REVIEW OF THE RELEVANT LITERATURE .....	2
2.1.	CLEFT LIP WITH OR WITHOUT CLEFT PALATE (CL+P).....	2
2.1.1.	Psychosocial issues associated with CL+P.....	4
2.1.2.	Prenatal diagnosis of CL+P .....	5
2.1.3.	Parental experience of a prenatal cleft diagnosis.....	9
2.1.4.	Rationale for studying the experiences of parents who receive a prenatal diagnosis of an oral cleft.....	14
2.2.	QUALITATIVE RESEARCH.....	15
2.2.1.	Defining qualitative research .....	17
2.2.2.	Paradigms of Qualitative Research.....	19
2.2.3.	Interviewing.....	20
2.2.4.	Quality and credibility in qualitative research.....	21
2.2.5.	Qualitative Description .....	22
3.	METHODS .....	27
3.1.	PARTICIPANTS .....	27
3.2.	INTERVIEWS .....	28
3.3.	DATA ANALYSIS.....	30
3.3.1.	Contact summary sheet.....	30
3.3.2.	Transcription.....	31
3.3.3.	Coding.....	31
3.3.4.	Thematic Analysis .....	34
3.3.5.	Addressing quality and credibility .....	35
4.	RESULTS .....	37
4.1.	THEMES.....	37
4.1.1.	Shared parental experiences.....	37
4.1.1.1.	Receiving the diagnosis .....	38
4.1.1.2.	Shock.....	41
4.1.1.3.	Cleft cause and embodied knowledge (Lippman, 1999) .....	42
4.1.1.4.	Parent to parent support .....	44
4.1.1.5.	Not a special needs child.....	46
4.1.2.	Prenatal diagnosis .....	48
4.1.2.1.	Coping.....	48
4.1.2.2.	Preparation .....	52
4.1.2.3.	Disadvantages .....	56
4.1.2.4.	Alternative perspectives.....	59
4.1.3.	At birth diagnosis.....	62
4.2.	OTHER ISSUES.....	65
4.2.1.	Pregnancy termination .....	66

4.2.2. Genetic testing .....	67
4.2.3. Internet use.....	68
5. DISCUSSION .....	70
6. CONCLUSION.....	83
APPENDIX A.....	85
INSTITUTIONAL REVIEW BOARD LETTER OF APPROVAL .....	85
APPENDIX B .....	87
PARTICIPATION REQUEST SCRIPT.....	87
APPENDIX C .....	89
SCRIPT FOR FOLLOW-UP CALL.....	89
APPENDIX D.....	91
LETTER PACKAGED WITH INFORMED CONSENT .....	91
APPENDIX E .....	93
INTERVIEW GUIDE FOR PRENATAL GROUP.....	93
APPENDIX F.....	96
INTERVIEW GUIDE FOR POSTNATAL GROUP .....	96
APPENDIX G.....	99
DEMOGRAPHICS OF PARTICIPANTS.....	99
BIBLIOGRAPHY.....	100

## LIST OF TABLES

Table 1: Recurrence risks for isolated oral clefts.....	3
Table 2: Sensitivity of routine transabdominal ultrasound screening for clefts .....	6

## LIST OF FIGURES

Figure 1: Procedural map of data organization and analysis .....	30
Figure 2: Coding example.....	33

## 1. INTRODUCTION

The purpose of this study was to describe the experience of receiving a diagnosis of clefting in the prenatal or postnatal period. Cleft lip with or without cleft palate is a common congenital birth defect that occurs in about 1/700 babies born in the United States (Abramowicz, Cooper, Bardi, Weyant, & Marazita, 2003). Improvements in ultrasonography have led to the prenatal diagnosis of a higher percentage of these anomalies.

Previous studies have examined the attitudes of parents of children with nonsyndromic oral clefts with respect to prenatal diagnosis. The majority of these studies were performed by asking parents to complete a mailed questionnaire. Several have concluded that parents find prenatal diagnosis to be useful in preparing them for their child's birth. A qualitative study might highlight issues that were not previously identifiable by constrained responses to mailed questionnaires. For instance, one participant's interpretation of "preparation" may be different than another participant's. Qualitative studies might give insight to the experience of these parents as well as identify their specific needs. Additionally, this study may provide information for the health care professional caring for these families.

Specifically, we proposed to:

Aim 1: Develop a better understanding of the experience of receiving a prenatal diagnosis of clefting compared to the experience of receiving the diagnosis at birth.

Aim 2: Determine the most important issues to assess regarding prenatal diagnosis of clefting in order to plan a larger, more focused study in the future.

Aim 3: Determine what is most useful and important to parents when they meet with the doctors and staff at the Cleft Craniofacial Center.

## **2. REVIEW OF THE RELEVANT LITERATURE**

### **2.1. CLEFT LIP WITH OR WITHOUT CLEFT PALATE (CL<sub>±</sub>P)**

CL<sub>±</sub>P is the most common craniofacial condition occurring at an incidence of approximately 1/700 births (Abramowicz et al., 2003). CL<sub>±</sub>P may occur as an isolated finding or as part of a spectrum of anomalies associated with a specific syndrome or chromosome abnormality. Most often CL<sub>±</sub>P is found in isolation without any additional abnormalities. Infants with CL<sub>±</sub>P have a cleft lip which usually is accompanied by cleft palate. In isolated cleft palate (CP), the cleft palate occurs by itself, without cleft lip or other abnormalities. CL<sub>±</sub>P and isolated cleft palate are types of oral-facial clefts and are considered separate birth defects. Both cleft lip and cleft palate are a result of a failure of fusion. Clefts of the lip are the result of the failure of the lateral nasal and median nasal processes, part of the frontonasal prominence, and the maxillary prominence to merge (Bender, 2000). The lip is usually closed by 35 days of gestation. If the lip does not fuse the palatal shelves shift and become separated by the tongue, which in turn can result in cleft palate. Palate development begins during the fifth week of gestation, after fusion of the upper lip, and is complete at the end of the twelfth week. Clefting may occur on one side of the face (unilateral) or both sides of the face (bilateral). Oral clefts may also be incomplete,

involving the lip and the anterior part of the maxilla or complete, which includes the lip, the anterior part of the maxilla and the hard and soft palate (Bender, 2000).

Males are two times more likely to be born with a cleft lip and palate than are females. The incidence of CL+P also varies according to ethnicity, with a lower frequency in the African American population. (Abramowicz et al., 2003) The exact cause of isolated CL+P remains unknown, though many studies have been conducted which examine environmental factors, such as alcohol, tobacco, and medication exposures during pregnancy, as well as genetic influences (Abramowicz et al., 2003). Isolated CL+P is considered a multifactorial trait, caused by an interaction of both environmental and genetic factors. This model assumes a threshold effect in which there is a critical mass of contributory genetic and environmental factors that have to be present before a cleft occurs. However, recent studies have proposed a greater degree of heterogeneity in etiology than previously thought, with at least some familial oral clefts being the result of the presence of one or at the most a few, major predisposition genes with incomplete penetrance (Stadter, 2002, p. 408). If, after a thorough evaluation by a clinical geneticist and a careful family history, the patient is diagnosed with an isolated oral cleft the family should be provided with recurrence risks which are based on empiric data. In general, the recurrence risk increases with increasing number of affected family members and with increasing severity of the cleft. Recurrence risks for isolated cleft lip +/- palate (the risks in the table are for CL+P and not CP) are described in Table 1 (Stadter, 2002, p.409).

**Table 1: Recurrence risks for isolated oral clefts**

<b>Affected family member</b>	<b>CL+P (%)</b>
1 Sibling	3-7
2 Siblings	8-14
1 Parent	2-5

These risks are most accurately applied to families who do not have multiple affected individuals. If the family history shows a number of individuals with CL<sub>±</sub>P, then a possible syndrome or possible Mendelian inheritance should be considered.

### **2.1.1. Psychosocial issues associated with CL<sub>±</sub>P**

One of the most important responsibilities of the health care professionals working with families that have a child with an oral cleft is providing emotional and medical support. In an article by Nicholson, a parent of a child with an oral cleft eloquently states, “After months of anticipation, we look forward to seeing and holding that newborn baby. The pain of childbirth will be over, but there can be a very noticeable difference in the tone of the birthing room when a child is born with CL<sub>±</sub>P” (Nicholson, 2002, p.474). The birth of a child with any type of physical abnormality can bring on a grieving process as parents grieve for the “loss of a perfect baby” they had imagined during the pregnancy. Forth (2003) argues, “Many parents use the term ‘devastation’ to describe their feelings when they recover from the initial shock, and realize their baby is in some way ‘handicapped’” (Forth, 2003). It is important to help parents work through the stages of grieving to make sure that they eventually come to the acceptance of their child. Each family member’s reaction to the child with the oral cleft may be influenced by his or her unique experiences. Some individuals will view the cleft as merely a structural defect that is amenable to surgery, but other family members might react with greater intensity (Stadter, 2002, p.410). Stadter states, “Having other members of the family with oral clefts can be a double-edged sword in regards to how it may impact the family’s reaction” (Stadter, 2002, p.410). The

way the family reacts might depend on the experience of the other family member with the cleft. If this person had a successful repair with few complications, then this may help to allay many fears of the family. On the other hand, an unsuccessful or complicated repair might frighten the family.

Families can feel overwhelmed with the birth of a baby with an oral cleft. Consultation with a comprehensive craniofacial team can help parents understand the medical needs of their child. Initially counseling should focus on the immediate concerns for the first year of life including feeding (addressing breast feeding), lip and palate surgery, and ear problems (Jones, 2002). A discussion of issues related to pain and insurance may also be useful. Contacting a family soon after diagnosis might reassure them that they are not alone and that there are experienced people available to help them care for their child. When parents have a child with CL<sub>±</sub>P, they may experience feelings of guilt at possibly having caused the cleft. Parents can usually be reassured that there was nothing they did that caused the cleft. Explaining to parents that we do not have any control over the genes we pass on to our children can sometimes be a useful approach. Other families that have a child with an oral cleft can be a great resource for families with a newly diagnosed baby because they are the experts in the daily care of these children (Jones, 2002).

### **2.1.2. Prenatal diagnosis of CL<sub>±</sub>P**

Advances in ultrasound have truly revolutionized prenatal care. Today's detailed scans are able to better assess fetal anatomy and therefore increase the detection rate of malformations. In light of this, more cases of CL<sub>±</sub>P are being diagnosed prenatally, especially when women are seen at

tertiary centers. The sensitivity and specificity of a prenatal cleft diagnosis is affected by several factors including the type of equipment used, the experience of the ultrasonographer, maternal body habitus, fetal positioning and whether the pregnancy is high-risk. Several studies conducted suggest that although the specificity (proportion of negative cases that are correctly identified by the test) is high, the sensitivity (proportion of positive cases that are correctly identified by the test) is generally low, averaging 20% (Johnson & Sandy, 2003). A summary of clefts diagnosed by ultrasound in several large studies is shown in Table 2 (Johnson & Sandy, 2003). A direct comparison between these various studies is difficult because of the differences in methodology. Additionally, two of the studies (Stoll, Dott, Alembik, & Roth, 2000; Shaikh, Mercer, Sohan, & Soothill, 2001) included individuals with cleft lip with or without cleft palate, while the other studies included individuals with cleft lip with or without cleft palate and individuals with isolated cleft palate (Johnson & Sandy, 2003).

**Table 2: Sensitivity of routine transabdominal ultrasound screening for clefts**

<b>Study</b>	<b>Sensitivity (%)</b>	<b>False positives</b>	<b>Number of patients scanned</b>	<b>Gestational age at detection (weeks)</b>
Levi et al., 1991	5/29 (17)	1	16,072	-
Chitty et al., 1991	2/9 (22)	0	8,785	-
Stoll et al., 2000a	28/214 (13)	-	131,760	-
Hafner et al., 1997	5/8 (63)	8	5,407	Mean 21
Boyd et al., 1998	12/25 (48)	0	33,376	-
Grandjean et al., 1999	57/316 (18)	0	-	Mean 28
Stoll et al., 2000b	55/309 (18)	-	265,679	Mean 23
Climenti et al., 2000	161/751 (21)	-	709,027	Mean 20-25
Shaikh, et al., 2001	23/130 (18)	0	-	-
Sohan et al., 2001	14/28 (50)	0	-	-
<i>Total</i>	<i>362/1819 (20)</i>			

(-): Information is not available

An accurate diagnosis is critical in counseling families with regards to the long-term significance of the anomaly including both prognosis and recurrence risk information (Jones, 2002). The identification of an oral cleft on ultrasound should lead to a careful ultrasound examination for the detection of any other abnormalities. Fetal karyotyping should also be considered, especially if other anomalies are present. When a cleft lip with cleft palate is diagnosed prenatally, it is the responsibility of the genetic counselor, or other member of the cleft-craniofacial team, to provide “balanced counseling” (Saal, 2002). Saal interprets “balanced counseling” to mean that the family should be advised about the number and types of surgeries that will be required as well as the potential for other associated complications, including velopharyngeal insufficiency, conductive hearing loss, and potential psychological problems. Most children with cleft lip and cleft palate do extremely well physically and socially and this may provide reassurance to parents. As Saal notes, “The key is to offer balanced counseling so the couple can make an informed decision regarding their pregnancy” (Saal, 2002).

Most couples are concerned as to the severity of the cleft. One large study reported an inaccurate diagnosis of severity in 38% of cases (Shaikh et al., 2001). Ultrasound can provide an accurate assessment of fetal anatomy but the information obtained may be incomplete, in terms of severity of a cleft lip and the involvement of the palate (Jones, 2002).

Although the lips fuse by eight weeks gestation, an accurate view of the face by ultrasound is not reliable until about 15 weeks gestation. Prenatal diagnosis of cleft palate is more difficult to

achieve because of the acoustic shadow from the facial bones. Optimal timing of a prenatal cleft diagnosis is thought to be around 20 to 22 weeks gestation (Johnson, 2003).

The use of three-dimensional ultrasound has also increased the accuracy of prenatal diagnosis of oral clefts (Stadter, 2002, p. 409). Three-dimensional ultrasound presents facial images with greater clarity than conventional two-dimensional imaging and allows the sonographer to identify precisely the facial location of the planar view (Jones, 2002). The better visualization of facial structures with the three dimensional ultrasound may lead to increased sensitivity. However, three-dimensional ultrasound is not yet widely available and suffers from the same potential impairments to visualization as the two dimensional ultrasound (Johnson & Sandy, 2003).

Within the existing literature, both advantages and disadvantages of a prenatal cleft diagnosis have been described. Potential advantages include psychological preparation, an opportunity for parent education, planned neonatal care and preparation for feeding, an opportunity for the investigation of other anomalies and possible chromosome studies, as well as increased reproductive choices for the parents including termination of pregnancy (Johnson & Sandy, 2003). The main disadvantages of a prenatal oral cleft diagnosis are the emotional disturbance of the pregnancy, inability to correct the problem, imprecision of the diagnosis, and cost effectiveness (Matthews, Cohen, Viglione, & Brown, 1998). Additionally, the controversy over antenatal surgery and the threat of abortion of otherwise normal fetuses are issues of concern (Forth, 2003). A number of studies, examined in the following section, have been published in which parental opinion of the advantages and disadvantages were assessed.

### **2.1.3. Parental experience of a prenatal cleft diagnosis**

Davalbhakta and Hall (2000) performed a retrospective questionnaire study of parents of children with clefts which sought to answer whether the cleft was diagnosed prenatally, what week in pregnancy the cleft was diagnosed; how soon after the diagnosis the parents were counseled by the cleft team, if the parents were given adequate information, and whether the prenatal diagnosis prepared them. Of those who had a prenatal diagnosis 85% (23/27) felt that it prepared them psychologically for the birth of their child with a cleft. A total of 11% of parents (3/27) felt they would rather not have known about the diagnosis prior to the birth of their child. A similar evaluation by Matthews et al. (1998) surveyed families regarding their feelings about the desirability and circumstances of prenatal diagnosis and the role of the cleft team in prenatal diagnosis. All nine families who responded to the survey felt that the prenatal diagnosis of the cleft made the adjustment to the diagnosis easier, however only one third of these families felt they were given enough information about clefting at the time of the diagnosis. Preparation and education were quoted as advantages of a prenatal cleft diagnosis. A total of six families felt that there were not any disadvantages to prenatal diagnosis, while three families felt that the diagnosis was anxiety provoking. All parents who met with the cleft team felt the experience was valuable and would recommend it to a friend. The majority of families (8/9) would have an ultrasound to look for a cleft in a subsequent pregnancy, but none of these families would consider pregnancy termination for an isolated cleft.

A Swiss study by Rey-Bellet and Hohlfeld (2004) used “semi-open questions” to evaluate the specialized counseling received by parents after the prenatal diagnosis of an oral facial cleft.

Much like the preceding investigations, this study also found that the majority of parents (93%) felt well prepared for the birth of their child. Only one couple (out of 29) regretted having been informed of the cleft diagnosis prenatally. This particular couple felt that the information kept them from enjoying the rest of the pregnancy and the mother needed psychiatric support. This study was able to document parental reactions at the time of the diagnosis and found that they varied greatly but “most expressed severe psychological shock” (Rey-Bellet & Hohlfeld, 2004). Common feelings included guilt, anxiety, fear and sadness (Rey-Bellet & Hohlfeld, 2004). The majority of parents (79%) felt that contact with the local parents’ association was of great benefit. Interestingly, some mothers in this study expressed distress after their child’s surgery for lip closure as they were forced to readapt to the baby’s new facial features and expressions. This study concluded that “empowering the parents by making them experts in cleft care as soon as possible helps them overcome their initial fear and sense of helplessness” (Rey-Bellet & Hohlfeld, 2004).

Another survey study, performed by Sagi, Shiloh and Cohen (1992) in Israel, sought to understand parents’ intentions to use prenatal diagnosis of cleft by ultrasound in subsequent pregnancies. Seventy-seven parents of a child with an oral facial cleft were sent a questionnaire which was developed to measure intentions to utilize prenatal diagnosis for a cleft and to terminate an affected pregnancy as well as to assess health beliefs such as perceptions of the malformation and the diagnostic procedure involved. Results from the questionnaire revealed several disadvantages associated with a prenatal diagnosis, such as false positive and negative results, inconclusive results, inability to predict the severity, and anxiety from the result, which were described by 18% of respondents. This study also found that 78% of participants would

probably or definitely utilize prenatal diagnosis in a subsequent for a cleft and 57% of them would probably not or definitely not terminate the pregnancy if a cleft was identified.

When a birth defect or health problem is identified, health care professionals are in the position of having to share this difficult news with parents regarding the health of their unborn baby or newborn. The information provided and the manner in which it is given is called, by some, the informing interview (Byrnes, Berk, Cooper, & Marazita, 2003). Research has shown that certain aspects of the informing interview have long-term effects on a family's ability to accept and adjust to a diagnosis, and the relationship that the parents have with their children. Additionally, the parental acceptance of their child has an impact on the child's psychological and social development (Byrnes et al., 2003). The person delivering the news to parents has the opportunity to either positively or negatively impact the experience.

Research has suggested that the extent of psychological trauma experienced by a parent is influenced by the manner in which bad news is delivered when an ultrasound abnormality has been identified (Alkazaleh et al., 2004). Several studies have been performed which examine parents' experiences of receiving a prenatal or postnatal diagnosis (Byrnes et al., 2003; Alkazaleh et al., 2004; Schuth, Karck, Wilhelm, & Reisch, 1994). One study that examined the postnatal informing interview experience of parents that have a child with CL±P found a theme of dissatisfaction with health care professional's knowledge of oral-facial clefts and treatment (Byrnes et al., 2003). Parents in this study wanted health professionals to be in control of informing conversations, to show caring and confidence, to show their own feelings, to make an effort to comfort parents and to provide referrals to other parents. Another study sought to find

out the relative importance women placed on a variety of issues when receiving bad news as a result of an abnormal ultrasound scan (Alkazaleh et al., 2004). This study found that “women value immediate, clear information with different options explained, enough time to ask questions, information regarding follow-up care, privacy and the sympathy of the person giving the bad news” (Alkazaleh et al., 2004). Schuth et al. (1994) described the desire for emotional support from women who are confronted with a malformation in their child. The majority of women in this study felt that, during the examination and afterwards, the physician should wait before giving recommendations, especially about terminating a pregnancy. Schuth argues, “Medical and psychosocial competence on the physician’s part will not cancel the catastrophe for the patient but may help her to come to terms with it” (Schuth et al., 1994). The women in this study expected the informing doctor to be a professionally competent expert who can take into account the current psychosocial functioning of the woman and her partner when informing them of the exact diagnosis, therapy, and prognosis of the malformation, and who allows for questions from the parents.

Termination of pregnancy following a prenatal diagnosis of a cleft is an issue which has been debated in the literature. If a facial abnormality is identified at a time in gestation when termination of the affected pregnancy is a realistic option, the woman and her partner are offered the choice of whether to carry the pregnancy to term (Jones, 2002). A clear understanding of treatment outcome for treatment of oral clefts seems to be a major factor in this decision. Blumenfeld, Blumenfeld, & Bronshtein (1999) state that 24 out of 25 prenatal cleft lip diagnoses, detected between 14 and 16 weeks gestation, were electively terminated. Twelve of these 24 fetuses had isolated cleft lip with or without cleft palate, and the remaining twelve had associated

anomalies. This study took place in Israel and utilized transvaginal sonography. In contrast to this study, an analysis in the UK conducted transabdominal ultrasound scans at 20 weeks gestation and identified 23 cases of CL±P, two of which ended in termination (Shaikh, 2001). As a follow-up to the Blumenfeld et al. study (1999), Jones (1999) published an article which examined the rate of termination after a prenatal diagnosis of a cleft in a southern California medical center. She found that two women out of eight terminated their pregnancies based solely on the cleft finding (Jones, 1999). In yet another investigation, also performed in the UK, three out of 30 women with prenatal cleft diagnoses opted for termination of their pregnancies. Two of these cases had multiple additional abnormalities including holoprosencephaly. One woman terminated her pregnancy based solely on the diagnosis of an isolated cleft lip and palate (Davalbhakta & Hall, 2000). Matthews et al. (1998) found in their survey study of a US population of nine families that none of them considered terminating their pregnancy following a prenatal diagnosis. In the Rey-Bellet and Hohlfeld study (2004) some parents admitted to feeling “revolted and destroyed” upon learning of the cleft diagnosis and three couples considered pregnancy termination, but none of the parents followed through with the procedure (n=29). Several factors appear to be important when parents are considering pregnancy termination based on an isolated oral cleft diagnosis. These include the perceived burden, expectation of recurrence, religious and cultural beliefs, professional advice and gestational age at diagnosis (Johnson & Sandy, 2003).

Use of the Internet has the ability to greatly impact a patient’s medical experience. Eysenbach (2003) estimates that “each day, more than 12.5 million health-related computer searches are conducted on the World Wide Web.” Increasingly, pregnant women and their families turn to

the Internet for information regarding ultrasound findings. Although some of the information is useful, none is filtered and some is grossly inaccurate (Jones, 2002). The widespread use of the Internet and the fear of some health care professionals that patients will stumble upon inaccurate and frightening information has led to many craniofacial centers creating handouts that list useful websites. The impact of the Internet with respect to prenatal diagnosis of oral-clefts has not been evaluated by many research studies. One study that examined this issue found that for 17% of parents who responded to a questionnaire regarding their prenatal cleft experience, the Internet was cited as a good way to obtain further information (Rey-Bellet & Hohlfeld, 2004). Clearly, families may search the Internet for information upon receiving a prenatal diagnosis of an oral cleft.

#### **2.1.4. Rationale for studying the experiences of parents who receive a prenatal diagnosis of an oral cleft**

As more cases of oral clefts are diagnosed prenatally it becomes essential for us, as health care professionals, to understand the parental perspective, but to also elicit the experiences of parents who received a diagnosis at birth. Adjusting to a cleft diagnosis, confronting the options of issues of termination and searching the Internet for accurate information are just a few of the concerns that parents face when they are informed that they are carrying a baby with a cleft lip with or without a cleft palate. In order to provide the best care for our patients, it is important to understand the experiences of parents who have actually been through the process. The quality of the counseling provided to parents at this vulnerable time will impact their coping and ability to maintain a sense of normalcy for the duration of the pregnancy. Prenatal counseling, if

approached with sensitivity and care, can make the birth of a baby with a cleft a positive experience for the parents (Matthews et al., 1998).

There is limited information regarding parental experience in the research which has previously been conducted on this topic. An experience which is filled with emotion and feelings, such as receiving a diagnosis of an oral facial cleft can be explored with more depth and detail using qualitative methods. For example, rephrasing the ‘yes or no’ question, “Did the prenatal diagnosis of your child’s cleft prepare you for his/her birth?” to “How did the prenatal diagnosis of your child’s cleft prepare you for his/her birth?” has the potential to elicit an entirely different response and allows parents to explain their experience in their own words. Additionally, we compared a group of parents who experienced prenatal diagnosis to a group who found out about their child’s cleft in the delivery room. Such a comparison allows a better assessment of the advantages and disadvantages of prenatal diagnosis. Though prior quantitative research has been conducted on this topic a qualitative approach may lead to a more in depth understanding of the experience of receiving a prenatal or postnatal diagnosis of an oral cleft. A greater understanding of this experience may also influence clinical practice for this particular group of patients.

## **2.2. QUALITATIVE RESEARCH**

Qualitative research has the potential to go beyond the numbers and statistics of its quantitative counterpart to delve into the lives and experiences of individuals and families. The real world of clinical practice involves intentions, meanings, intersubjectivity, values, personal knowledge,

and ethics, yet most published clinical research consists of observational epidemiology and clinical trial designs (Miller & Crabtree, 1998, p.295). These traditional quantitative studies create a separation of the variables of interest from their everyday setting, place them in a controlled research environment and then try to explain the results in the original context.

It has been suggested that clinical researchers should attempt to refocus their investigative questions that emerge from the clinical experience to pay attention to and reveal any underlying values and assumptions, and direct the results toward clinical participants (Miller & Crabtree, 1998, p.297). Many clinical research questions about body, life and power concern experience, meaning, patterns, relationships and values. Thus, in order to understand the impact of advances in the medical field and to make sound decisions when caring for patients it is important to attend to the experience of the people who have lived through these events.

Reproductive medicine is a field of rapid advancement which can benefit from alternative research methods, such as qualitative approaches, because “we don’t understand enough about the social dimensions of the phenomena under study to rely solely on quantitative methods” (Beeson, 1997). A prenatal diagnosis of a fetal structural abnormality may impose profound social dilemmas on patients and clinicians alike. Genetic counselors serve a unique role as professionals that understand both the complexities of human genetics and the dynamics of human behavior. Genetic counselors weave in and out of these two worlds of molecular and social sciences (Beeson, 1997). Though a number of studies have been conducted which have examined the utility of a prenatal diagnosis of a cleft, most of these studies used a closed-ended questionnaire design in which participants’ answers were constrained to pre-determined

responses. This type of research design does not allow researchers to explore the experience of participants. Questions that concern experiences, relationships, and values “weave the concerns of body, life, and power into a holistic narrative and call for the designs and methods of the qualitative clinical researcher” (Miller & Crabtree, 1998, p.298). A detailed description of how and why prenatal diagnosis of a cleft may or may not be useful has not been conducted. This experience for parents is filled with emotion and intensity that a simple yes or no question does not adequately address. A qualitative analysis is warranted to determine the specific concerns of these parents and to determine the precise benefits of prenatal diagnosis so that we, as health care professionals, may provide the best care possible for our patients.

### **2.2.1. Defining qualitative research**

Many definitions of qualitative research exist because the term is quite broad, incorporates many techniques and relies upon diverse epistemologies. Ritchie and Lewis (2003) describe the aims of qualitative research as being directed at providing an in-depth and interpreted understanding of the social world of research and participants by learning about their social and material circumstances, experiences, perspectives and histories. The emphasis of some qualitative research is on the participants’ lived experience and is well suited for uncovering the meanings people place on the events, processes and structure of their lives (Miles & Huberman, 1994, p.10). Qualitative researchers often attempt to interpret their data through the perspective of their study participants, in fact “seeing through the eyes of the people being studied” (Bryman, 2004). Beeson argues that “Qualitative research is productive because it enables us to discover and document aspects of reality that we cannot necessarily anticipate, and thus to transcend the limitations of our own perspective” (Beeson, 1997). The results of a qualitative analysis range

from descriptive summaries to detailed theories, some of which are inductively generated from the data.

Often, qualitative research studies include samples that are small in scale and purposely selected, data which are detailed and information rich, and analysis which may produce detailed description and classification to develop theories or explanations. Sampling is sometimes described as “purposeful” when it intends to select participants or sources on the basis of exploring meaning. Sampling can continue throughout the course of the study and is often closely linked to the “emergent nature of the research process” (Fossey, Harvey, McDermott, & Davidson, 2002).

Samples may be small in qualitative research studies as there is no fixed minimum number of participants. For some qualitative methods the aim of sampling is to refine ideas, generate concepts and not simply increase the sample size for the purpose of generalizing results. However, in order to fully describe the experience being studied there must be an adequate depth of information. Sampling often continues until the themes or concepts which have emerged from the data are fully developed. This phenomenon, referred to as “saturation”, means that further sampling would be redundant and would not generate new patterns or themes. Diverse qualitative analytical approaches exist but, in general, involve exploring the data, comparing and contrasting the data, and evolving to a more sophisticated understanding of the data.

Several approaches to qualitative data analysis involve coding, or attaching labels to segments of data, to identify concepts, patterns and themes. Visual displays can play a part of qualitative

research and selecting key quotes as exemplars is one method of communicating ideas visually to others. The researcher chooses segments of text (verbatim quotes from respondents) as exemplars of concepts, theories and also of negative cases (Ryan & Bernard, 2000, p.784).

### **2.2.2. Paradigms of Qualitative Research**

There are many ways to approach qualitative research and different methods used for gathering data. The philosophical underpinnings of qualitative research guide the collection of data, methodology used, and interpretation of the research. Rigorous qualitative research is characterized by congruence between the paradigm that informs the research questions and the research methods used (Fossey et al., 2002). These philosophical paradigms describe a system of ideas used by researchers to generate knowledge. A philosophical standpoint may help the researcher in developing and refining the particular methods of the study.

Soltis (1984) describe three different paradigms of research, namely the empirical, interpretive and critical paradigms. These views represent different ways of looking at the world, which influence the research approach used to observe and study experiences. The empirical paradigm, for example, is rooted in the scientific method, which relies on deductive logic in combination with observations and experiments to “refute propositions and confirm probabilistic causal laws, which are used to make generalizations about the nature of phenomena”. In contrast, interpretive inquiries focus on understanding and accounting for meaning of human experiences. Interpretive inquiry “goes beyond mere description of core concepts and essences to look for meanings embedded in common life practices” (Lopez & Willis, 2004). Finally, the critical paradigm stresses an awareness of the social, cultural, political and historical context of an experience. An

important aspect of critical inquiry is the on-going process of societal change. This approach is founded on the assumption that interpretation is influenced by socially acceptable ways of viewing reality (Lopez & Willis, 2004).

This investigation of the impact of a prenatal diagnosis of an oral cleft aligns most closely with the empirical paradigm. The empiric approach “depends on processes that are taken to be objective, neutral with respect to values, and reductionistic in that phenomena are studied as ‘component parts to describe, explain, and predict how these parts work, when and where’” (Fossey et al., 2002). Empiricism is not exclusively associated with experimentation and quantification and is actually defined as “originating in or based on observation or experience” (www.m-w.com). A strength of qualitative research is precisely its respect for the empirical world, that is, its potential for yielding verifiable knowledge of human group life and human conduct (Beeson, 1997). The experiences of parents of children with an oral cleft were analyzed with the application of a coding system with the goal of reducing the data to patterns and themes. The themes which were derived from the data were then utilized to describe and explain the experiences of these parents.

### **2.2.3. Interviewing**

Data collection for qualitative research is often directed towards discovering the who, what, and where of experiences. Qualitative data collection methods include, but are not limited to, in-depth, open-ended interviews, direct observation, written documents, and focus groups. A common method used to collect data, and the method utilized in this investigation, is the open-ended interview. “The job of the interview is to ensure that the relevant contexts are brought into

focus so that situated knowledge can be produced” (Mason, 2002, p.62). Informative results will be dependent upon whether or not the interviewer is able to create an environment where the participant feels comfortable being open and forthright. Open-ended questions are generally posed by the researcher who may or may not have generated a list of semi-structured questions.

This investigation employed an interview guide that outlined the issues to be explored during the interviews. Interview guides usually contain a list of questions and prompts designed to guide the interview in a focused, yet flexible and conversational, manner (Fossey et al., 2002). The interview guide was prepared to ensure that the same topics were covered with each study participant. The guide provided a point of reference, but the interviewer followed the conversation of the participants by asking follow-up questions and using probing techniques. Qualitative interviewing usually begins with general questions and then moves on to more specific questions. Shared language is an important concept in qualitative interviews and promotes an open relationship between the participant and the researcher. The qualitative interview acts as a “guided conversation” where a comfortable environment is created and shared language is promoted (Rubin & Rubin, 1995, p.126). Interviews are typically tape-recorded and transcribed to allow a more thorough examination of the interview and the opportunity for analysis by multiple researchers. The interview method was utilized in this study in order to generate in-depth, detailed accounts of participants’ experiences.

#### **2.2.4. Quality and credibility in qualitative research**

Patton claims, “Quality and credibility are connected in that judgments of quality constitute the foundation for perceptions of credibility” (Patton, 2002, p.542). Traditional scientific research

criteria for judging quality and credibility include objectivity of the inquirer, validity and reliability of the data, and the ability to generalize. The aim of qualitative research is typically not to generalize and researchers usually do not make a claim of generalizability of their findings to a larger population (Fossey et al., 2002). Miller and Crabtree (1998, p.307) state, “Local context and the human story, of which each individual and community story is a reflection, are primary goals of qualitative research, not ‘generalizability’.” Qualitative researchers emphasize that naturalistic inquiry should be judged by dependability and authenticity (reflexive consciousness about one’s own perspective, appreciation for the perspectives of others, and fairness in depicting constructions in the values that undergird them) (Patton, 2002, p.546). Subjectivity is embraced as a pathway to deeper understanding of the human dimensions of the world. Dependability of a qualitative study may be closely linked to the believability of the text. When the audience is drawn into the story and can put themselves in the “shoes” of the study participant, the conclusions make sense to the reader. Qualitative researchers are more interested in deeply understanding specific cases within a particular context than in hypothesizing about generalizations and causes across time and space (Patton, 2002, p.546). Qualitative clinical research is convincing when the methods are appropriate, the investigator’s relationship with the participants and data is explained, the audience recognizes itself in the findings, and the results matter to clinical participants (Miller & Crabtree, 1998, p.308).

### **2.2.5. Qualitative Description**

A qualitative descriptive study was undertaken in this investigation to examine the experience of receiving a diagnosis of clefting in the prenatal or postnatal period. Qualitative description offers a comprehensive summary of an event in everyday language (Sandelowski, 2000). This

type of research was chosen in order to describe in-depth the experience of parents who had been given a pre- or postnatal diagnosis of an oral cleft. Descriptive studies are useful in uncovering essences of phenomena that have been incompletely conceptualized by prior research. This particular population (parents of children with oral clefts) has, prior to this study, been primarily examined by quantitative studies. A qualitative descriptive study allows emotions and thoughts experienced by the parents to be characterized. Qualitative studies do not constrain the responses of the participant and therefore generate a more in depth view of this experience of an oral cleft diagnosis.

Additionally, qualitative description is “especially amenable to obtaining straight and largely unadorned (i.e., minimally theorized or otherwise transformed or spun) answers to questions of special relevance to practitioners” (Sandelowski, 2000). Examples of these types of questions include: What are the concerns of people about an event? What are people’s responses (e.g., thoughts, feelings, attitudes) toward an event? What reasons do people have for using or not using a service or procedure? What factors facilitate and hinder recovery from an event? We felt that our inquiry into the experience of receiving a prenatal or postnatal diagnosis of an oral cleft fit well into the types of questions postulated by researchers undertaking qualitative description. It is thought that by specifically addressing and describing the experience of these parents, clinicians and health care professionals who care for this unique population will better understand the needs of families who have a child with an oral facial cleft. Sandelowski argues, “Fundamental qualitative description entails a kind of interpretation that is low-inference, or likely to result in easier consensus among researchers” (Sandelowski, 2000).

Basic qualitative description is not highly interpretive in the sense that the researcher deliberately chooses to describe an event in terms of a conceptual framework. The data is typically presented in the everyday language of the participants. Sandelowski notes, “Researchers conducting this type of study seek descriptive validity, or an accurate accounting of events that most people (including researchers and participants) observing the same event would agree is accurate, and interpretive validity, or an accurate account of the meanings participants attributed to those events that those participants would agree is accurate” (Sandelowski, 2000). Researchers performing this type of research stay closer to their data and to the surface of words and events than researchers conducting grounded theory, phenomenologic, ethnographic, or narrative studies. “Surface”, as it is used in the statement above, conveys the depth of penetration into, or the degree of interpretive activity around, reported or observed events.

In terms of sampling in qualitative descriptive studies, virtually any type of purposeful sampling technique can be used. The goal of sampling is to obtain cases that are information-rich for the purpose of the study. The data collection is usually based on minimally to moderately structured open-ended individual or focus group interviews. Data collection may also include observation of events and examination of documents and artifacts. The expected outcome of this type of study is a straight descriptive summary of the contents of the data arranged in the best way that fits the data. Ways to arrange data may include chronological order of events, most prevalent to least prevalent theme, a day-, week-, month-, or year-in-the life approach or the Rashomon effect whereby the same event is described from the perspective of more than one participant (Sandelowski, 2000). Whichever way the investigator chooses to represent her data, the primary purpose is to produce a descriptive summary of an event, organized in a way that best contains

the data collected and that will be most relevant for the audience for which it was written. The summary may yield concepts and hypotheses that can be tested in future studies.

Qualitative descriptive studies may draw techniques from other approaches, including phenomenology and grounded theory. “Qualitative descriptive designs are typically an eclectic but reasonable and well-considered combination of sampling, and data collection, analysis and re-presentational techniques” (Sandelowski, 2000). For example, this study utilized the concept of constant comparison, typically associated with grounded theory, where the researcher categorizes data through a process of comparing new codes with previously identified codes. Additionally for this investigation, the process of thematic analysis, described by Boyatzis (1998) was drawn upon in the data analysis, and particularly coding of data. “Thematic analysis is a process for encoding qualitative information” (Boyatzis, 1998, p.4).

Encoding the data requires the development of a code, which consists of labeling the data. Code development leads to the identification of themes derived from the data. Boyatzis describes a theme as “... a pattern found in the information that at a minimum describes and organizes the possible observations and at a maximum interprets aspects of the phenomenon” (1998, p. 4). Generally, themes are interpreted from participants’ stories and illustrate a shared meaning of experiences that occur in varied contexts. Themes may encompass a number of categories and bring together ideas or experiences that would not have much meaning when viewed alone. Usually, themes are not stated by the participants, but are conceptualized by the investigator (DeSantis & Ugarriza, 2000). “Thematic analysis involves the search for and identification of common threads that extend throughout an entire interview or set of interviews (DeSantis &

Ugarriza, 2000). Leininger (1985, p.61) summarizes the process of thematic analysis in different steps which include identifying and listing observations and experiences, combining raw data and descriptors into meaningful units, known as patterns, identifying micropatterns and determining how they relate to patterns and themes, and synthesizing several patterns to obtain a broad, comprehensive, and holistic view of the data as themes and subthemes. The focus of the next section will be the methods used to carry out the investigation of the experience of receiving a diagnosis of clefting in the prenatal or postnatal period.

### **3. METHODS**

#### **3.1. PARTICIPANTS**

Participants were recruited from the patient population at the Cleft-Craniofacial Center at Children's Hospital in Pittsburgh. The goal for the study was to recruit an equal number of participants whose child was diagnosed prenatally and whose child was diagnosed at birth. The proposed study population included both men and women over the age of 18. All study participants have a child who was born with a non-syndromic cleft lip with or without cleft palate. The prenatal group consisted of 12 parents, while the postnatal group consisted of eight parents. Thirteen mothers were included in the study, while seven fathers participated. Of the 20 participants, 18 participated in phone interviews.

The University of Pittsburgh Institutional Review Board (IRB) approved the protocol and informed consent document for the study on August 4, 2004 (Appendix A). The IRB granted a waiver of informed consent to use medical record information (PHI) to identify potential research subjects before recruiting began. Dr. Carla Weidman, clinical psychologist at the Cleft-Craniofacial Center, accessed medical records to generate a list of potential research subjects. The specific criterion that was evaluated within the medical records included the timing of the child's diagnosis (prenatal diagnosis or diagnosis at birth), gender, type of cleft and whether or not the child was born with additional medical problems. Dr. Weidman then contacted the potential participants to ask for their permission to participate utilizing the participation request

script (Appendix B). For interested individuals, the investigator followed up this initial phone call with a second phone call within a few weeks. The purpose of the follow-up phone call was to determine if the potential participant was still interested, to introduce the investigator conducting the interviews, and to tell the potential participant that informed consent documents would be arriving in the mail for each parent (Appendix C). Interested individuals were then mailed two copies of the informed consent document with an explanatory letter (Appendix D). Within two weeks the participant was contacted again by phone. At that time, the investigator provided informed consent for interested participants allowing them to ask any questions or share possible concerns about the study. The investigator then reiterated the directions to initial each page and sign the last page. A self-addressed stamped envelope was included for participants to mail back the consent form. The investigator also asked the participant at this time if they would like to set up an interview date. Some participants planned an interview date during this phone call while others preferred to have the investigator call back to arrange an interview date upon receipt of the consent form.

### **3.2. INTERVIEWS**

Participants were given the option to have the interview conducted over the phone or in person at the Cleft-Craniofacial Center. Two parents chose in-person interviews while the rest of the participant interviews were conducted over the phone. When contacted for the interview (on the date and time that had been agreed upon by the investigator and participant) parents were reminded that the interview would be audiotaped. They were also told they could refuse to answer any question or request to erase an audiotaped response. An interview guide was

employed to carry out this investigation. The same demographic questions were asked of each parent. A slightly different interview guide was used for the parents who found out about their child's cleft prenatally compared to those parents who found out at birth. The interview guide listed sample questions to ask each participant, but the interviewer put each question into her own words for each participant, depending on the flow of the conversation. Most questions were open-ended, but others required only a yes or no response.

Each interview began with the statement, "Tell me the story of how you first found out about your child's cleft." With the exception of this first statement, the questions did not follow a specific order; instead the interviewer followed the participant's lead and asked follow-up questions, which were tailored to each participant. An exception to this interview guide method was the question that dealt with pregnancy termination. Due to the sensitivity of this topic, a standardized open-ended interview method approach was used for this question. The standardized open-ended interview approach requires carefully and fully wording the question prior to the interview. Participants were asked the question in this manner:

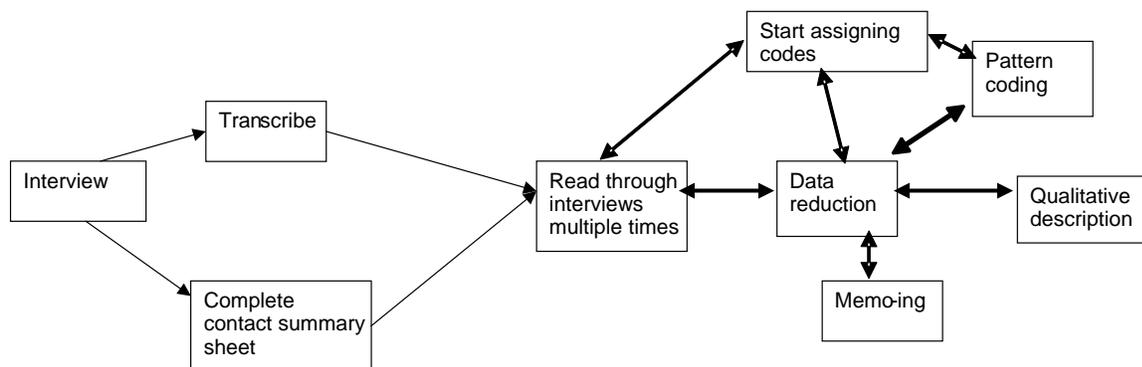
When parents are given a diagnosis that their child has a cleft lip, many thoughts about the pregnancy and raising a child who will need multiple surgeries crosses their mind. Some parents consider ending the pregnancy and some parents don't. Could you tell me your thoughts about this issue?

The interview guide was dynamic, meaning that as additional interviews took place, questions were added to the interview guide and ideas for different probing and follow-up questions were generated. This is part of the iterative process of qualitative research. In order to keep track of these changes a detailed record of interviews was created. This included a timeline of interviews as well as a copy of the interview guide used for each interview. When changes were made to

the interview guide they were done so in a different color and were written with an explanation of the new question or probe. This was done so that it was clear which questions had been updated or added. Usually the change was made after a study participant brought up a key point that the investigators felt was important enough to ask of all participants. Interview guides for both groups of participants can be found in the appendix (Appendix E and F).

### 3.3. DATA ANALYSIS

Prior to the organization and analysis of data a preliminary outline of this process was created by the investigator. This procedural map is shown in Figure 1.



**Figure 1: Procedural map of data organization and analysis**

#### 3.3.1. Contact summary sheet

A contact summary sheet was created for each participant following the interview. The contact summary sheet, as described by Miles and Huberman (1994, p.51) is a single page document, which contains questions about a particular participant. The sheet creates an overall summary of

the main issues from the interview and allows the investigator to reflect upon points of interest for the next interview. Contact type (phone or in person), date of the interview, site, name of participant and investigator were all included on the sheet. Also incorporated were main issues brought up during the interview, a summary of important information, and new questions to address with the next contact. The contact summary sheet was completed by the investigator as soon as possible following the interview and before transcription or coding began.

### **3.3.2. Transcription**

Transcribing is a point of transition between data collection and analysis. The investigator conducted her own transcription, instead of having it done by a transcriber, as an additional means to become immersed in the material. The actual quotations spoken by the participants were transcribed into Microsoft Word documents. Transcriptions included conversation placeholders such as “um” and “like”, however for the sake of clarity, the excerpts presented in the Results section do not include these placeholders unless they contribute to the meaning of the conversation.

### **3.3.3. Coding**

QSR NVivo software was utilized to assist in the coding of transcripts. Qualitative software programs, such as NVivo, facilitate data storage, coding, retrieval and comparison (Patton, 2002, p.442). Interview transcripts were imported into NVivo in rich text format. Before the process of coding began, each transcript was thoroughly read to familiarize the investigator with the interview. A codebook was then developed from the in-depth reading of the transcribed

interviews. Certain codes, initially derived from the literature review, such as “emotional disturbance of a placid pregnancy”, were also used (Forth 2003). Ryan and Bernard (2000) define codebooks as “...organized lists of codes (often in hierarchies).” Simply put, codes are labels for assigning meaning to the data that has been compiled. Codes were developed by the analyst with the intent of capturing the essence of the interviews. Initial codes closely reflected the spoken word of the participants, that is, abstract themes were not conceptualized in the early coding of the data. Similar phrases from different participants, relationships between various codes and common patterns in responses were noted. The codebook was continuously refined and altered as additional interviews were analyzed and rudimentary themes solidified. For example, if two codes were being used to describe the same phenomenon they were condensed into a single code. In the initial stages of coding there was a “become informed” code as well as a “knowledge is empowering” code. When the text that each of these codes represented was examined it was determined that these codes could be condensed into a single code. As new codes were generated they were constantly compared to interview text which had previously been coded. Some codes included large portions of text, while other codes consisted of a single word. For example, because many participants said the word “shock” during their interviews, this word was given its own label and was coded every time it was used. Certain text was coded with more than one label if the participant was expressing a complex thought, which, for example, occurred on a number of occasions as participants discussed the ways in which a prenatal diagnosis prepared them for the birth of their child. An example of the coding process is shown in Figure 2.

<b>Interview Transcript Excerpt from PNS9-Mom (Prenatal group)</b>	<b>Code:</b>	<b>Theme:</b>
<p><b>Interviewer:</b> And so how did you feel about the prenatal diagnosis?</p>		
<p><b>PNS9-Mom:</b> Oh, I couldn't imagine going through it without it. I mean it was so nice, you know when he was delivered they had a neonatal specialist there, who the first thing they did was check out the palate. You know the lip was obvious, but I guess they can't tell from the sonogram about the palate. I was relieved. You know I couldn't imagine having all of those emotions I had with him being diagnosed on the sonogram having them when I was laying on the table. You know there's just so many other things that you have to worry about. It would have been really, really difficult emotionally and I think that it would be really tough. We had the baby at [outside hospital] and they said, "We can kind of fix that up before you leave if you want" and we had already met with the people at Children's and we were like, "What?" So we called and we asked you know why would people do that and I guess some people do have that philosophy. And you know we probably would've said, "Ok you're the doctor", you know not knowing, but after we met with Children's we were like, "Wait, we'll do it this way". So who knows what we would've done. Yeah, just being prepared and it really only took us a day or two to say, "Phew, oh ok, now let's get back to normal and figure out what we are going to do about this." It was good to have that couple of days to kind of get the emotions, or the shock out of your system, and then to go forward with the research.</p>	<p>Plan for baby's needs</p> <p>Mental (self)</p> <p>Become informed</p> <p>Adjustment/ acceptance time period</p>	<p>Preparation</p> <p>Coping</p>

**Figure 2: Coding example**

A technique called memo-ing was used throughout the data analysis. Memos are ideas about codes and their relationships that come to mind to the analyst during the process of coding (Miles & Huberman, 1994). Memos are recorded as analytical notes which are not necessarily part of the codebook. Memo-ing allows the analyst to connect codes, categories (classification of codes) and theorize about how they relate to one another. Memos help researchers grapple with ideas about the data, set up an analytic course, refine categories and define the relationship among various categories (Charmaz, 2000).

### **3.3.4. Thematic Analysis**

Thematic analysis is the process of determining how the generated codes fit into patterns and themes, and synthesizing these themes to obtain a broad, comprehensive view of the data. Codes within each group of participants (prenatal diagnosis and at birth diagnosis) were closely studied for similarities, differences and recurring patterns. Pattern codes are explanatory or inferential codes that can identify an emergent theme or explanation (Miles & Huberman, 1994). Pattern coding helped to reduce the amount of data into more manageable units and laid the groundwork for analysis across cases. As codes were thoroughly examined larger patterns, or themes, were developed. Codes closely reflected the participant's spoken word while themes reflected a broader perspective or concept. The themes recognized in the prenatal group were then compared and contrasted with the themes from the postnatal group. Memos, which had been written during the coding process, were drawn upon for insights in this stage of analysis. These analyses crystallized the key dimensions of the data within each of the groups and emphasized the differences between the two groups. The analysis sought to relate and make sense of the different patterns and themes, which had initially been identified by the coding. Abstract interpretation of the results was kept to a minimum in order to align with the qualitative description study design. Qualitative description was chosen as the approach for this investigation in order to provide a comprehensive summary of the experience of receiving a cleft diagnosis.

### **3.3.5. Addressing quality and credibility**

Credibility of qualitative research is dependent upon the rigorous methods used for conducting an inquiry that yields high-quality data (Patton, 2002, p.553). “For observation or interview, prolonged engagement enhances the credibility of the findings” (Byrne, 2001). Rigor was established in this investigation by having prolonged exposure to the data and using the interview guide to systematically record participants’ views on similar topics. Prolonged exposure to the data included listening to the transcribed tapes and reading every transcript in its entirety at least twice before coding. A potential limitation of this study is that triangulation (combining multiple observers, theories, methods or data sources) and member-checking (review of the data by the study participants) were not utilized. The purpose of triangulation is to enhance the quality of the data by gathering information from different sources and/or various methods. This is believed to illuminate different facets of situations and experiences and helps to portray their complexity (Fossey et al., 2002).

Validity refers to the correctness or precision of the research. Questions the researcher should ask herself while conducting a qualitative study include: Was there any bias in the sample selection? Was the environment and quality of questioning during the interview effective for participants to fully express their views? Have the phenomena been identified, categorized, and named in ways that reflect the meaning assigned by the participants? And have the findings been portrayed in a way that remains true to the original data and allows others to see the analytic constructions that have occurred? (Ritchie & Lewis, 2003, p. 273). In this investigation there was not a known bias in the recruitment of participants for the study. The recruiter contacted each family from the list generated by examining the records at the Cleft-Craniofacial Center. If

a family could not be reached, she simply moved on to the next family from the list. She also spoke with the families of new patients as they arrived at the cleft clinic for their initial visit. Records were obtained only from births that occurred within the past two years. To address the issue of creating an environment during the interview where participants felt free to express themselves, the interviewer explained to each participant that he/she had the right to refuse to answer any question. Additionally, participants were told that a specific part of the tape could be erased if they wished to do so. The interviewer strived to establish rapport with the participants prior to the interview by phoning them for introductions, to discuss informed consent and to set up an interview appointment. Some participants spoke with the interviewer over the phone as many as three times prior to the interview itself. Careful attention was paid to the spoken words of the participants as interviews were transcribed. Internal validation was achieved by constantly comparing and checking codes within and across transcripts. The steps of analysis are also carefully outlined in the sections above so that reviewers can be aware of the analysis that occurred. The following chapter describes the results of the thematic analysis of the transcribed participant interviews.

## **4. RESULTS**

This study sought to examine the parental experience of receiving a diagnosis of clefting in the prenatal or postnatal period. Study participants took part in a phone interview or in-person interview. Demographics information was collected on each participant and is displayed in Appendix G. Participants were divided into two groups, those whose children were diagnosed prenatally and those whose children were diagnosed at birth. Twelve out of twenty participants (60%) consisted of the prenatal group while the other 8 participants made up the at birth group.

### **4.1. THEMES**

Transcripts of the interviews were analyzed by thematic analysis to generate a qualitative description. Generated themes provided insight into the parental experience of receiving and coping with a diagnosis of a cleft lip with or without cleft palate. Themes are not described in an order of importance and are not rated by the frequency in which the participants used them. This is due to the limited number of participants and because the exact same questions were not asked of each individual participant.

#### **4.1.1. Shared parental experiences**

All interviews began with the same question: “Tell me the story of when you first found out about your child’s cleft.” Each individual’s response to this question varied, but shared

experiences across the two groups were immediately evident. Common themes identified include: “receiving the diagnosis,” “shock,” “cleft cause and embodied knowledge” (Lippman, 1999), “parent to parent support,” and “not a special needs child”. Although there is a difference in timing of the diagnoses between the two groups of participants, the commonalities that parents experienced were striking. The sections below outline the themes commonly shared by both the pre- and postnatal participants.

#### **4.1.1.1. Receiving the diagnosis**

Parents often spoke in detail about receiving the diagnosis and the behavior of the individual who informed them of the cleft. Although some parents were informed of the cleft prenatally and others postnatally, many parents expressed similar sentiments regarding the lack of information they were given by the person who informed them of the cleft. When probed about the informing person’s mannerisms, one mother whose child was diagnosed prenatally said, “I felt it was very clinical, very matter-of-fact. The doctor said to me, ‘Well, you’re at that age.’ He tapped me on the leg and said, ‘They do wonders today.’” The individual providing the information sometimes may have felt that minimizing the cleft may help parents adjust to the diagnosis. “Our pediatrician that checked her out at that time, he was not very comforting. Maybe he thought he was being comforting because he just said, ‘Oh that’s no big deal, we’ll just get that fixed up no problem.’ And to me it was a big deal, this is my baby.”

Another mother expressed her frustration with the way the doctor handled her baby in the delivery room. Her baby was actually taken from the delivery room, without explanation, before she had the chance to even look at or hold her. In the midst of this her husband fainted, which added to the confusion of the moment:

The first words I heard from him [the doctor] were, “Oh my”. But as soon as they took the baby away they had a social worker come over and they just told me that she had the cleft lip and they didn’t tell me about the palate at the time. And they said it was something that was fixable and not something to be majorly worried about. I didn’t quite like it at all. I heard the, “Oh my”, my husband went down and I thought something was majorly wrong. I actually thought maybe the baby was dead or something. So I kinda wish they would have said everything’s fine and this is what it is, instead of waiting.

Another mother whose baby was also diagnosed at birth felt uncomfortable with the way her doctor handled the situation: “She wasn’t even all the way out when he [the doctor] told me. So maybe he could’ve waited a few minutes to let me get through what was going on and then say, ‘Ok, I don’t want to alarm you.’ But he just wasn’t real sensitive to the emotions of what was actually going on.” One mother who found out through ultrasound said her doctor’s first words were: “Oh honey, don’t worry about it.” She explained that the doctor’s approach was condescending and that she had no information: “All they could tell me is there’s a possible problem. They can’t tell you anything about who will deal with it, how you will deal with it, what will need to be done. For them just to have stuff from Smile Train, or any of the pamphlets, any actual pictures of the baby’s before and after surgeries. They just told me maybe he has a cleft and we don’t know.”

Not only was lack of information a problem but participants spoke of misinformation from informing physicians as well. One father in the prenatal group said: “The way it was explained to us, it made it sound as if whenever you cut an edge of a piece of paper with scissors and you know, you just have a little knick. They made it sound like a couple of stitches and when he was born, done and over with. So that’s what I was thinking for the longest time, (laughing) until we

found out otherwise.” Other participants expressed concern that they were not given any written information and were not referred to a cleft or craniofacial center

A few participants did express satisfaction with the delivery of the diagnosis. The main differences between these parents and those who were dissatisfied were in the behavior of the informing physician, the concrete information provided, and the prompt referral to the cleft clinic. As one participant, who was pleased with her informing physician, explains:

Well, we didn't find out until she was born. The doctor had, when she was born said, "You have a beautiful baby girl and she was born with a cleft and it's just something that didn't have time to get fixed and it's just part of the lip that's not connected. Nothing to worry about." He started with lots of statistics which he already knew about and told us that he'd get us a lot of information and that she was otherwise healthy. You know we really had a lot of admiration for the way he handled that.

A father whose child was diagnosed postnatally said that the doctor did not say anything upsetting, referred them to the cleft center and immediately gave them a pamphlet that was outdated, but had the correct phone number for the clinic. "We didn't wait around as soon as we left the hospital, we didn't even come home; we just left the hospital and went to Children's [Cleft-Craniofacial Center]."

Closely related to the pattern of "satisfaction" was the parental perception of how the delivery of the diagnosis should occur, coined "the ideal". In general, parents felt that providing additional information, referral to the cleft clinic and a compassionate, caring disposition in the physician would have improved the experience. As one participant mentioned, "I think if they had more information it would have been much more helpful. In the interim, you know I don't expect

them to have as much information about it as a genetic counselor but if they had something you could refer to, to calm you in that time until you meet with someone, it would have been much more helpful.” Another mother whose child was diagnosed prenatally expressed a similar attitude:

I think they should be, I don't know if they have the tools to do this, but a lot less clinical and a little more informative. Because they did tell me, 'Yes, it's unilateral, but like I said when I left there I didn't realize, was that both sides, one side? I think they could take a few minutes and really sit down and explain, they should have pictures on hand. I think you just need a little bit more; you leave there with this very empty feeling. I think the ideal picture would be to have some type of support system that maybe these women could come into the cleft clinic and they can talk to someone like me or others who have experienced this and you know there is a light at the end of the tunnel and they do wonders.

Parents felt that the informing physician should be as honest as possible, as one father put it, “point blank”. One mother acknowledged the difficult situation the health professional is placed in while giving the diagnosis by saying: “Maybe she doesn't have another way, how can she tell you? I don't know if she has another way to say it.”

#### **4.1.1.2. Shock**

Another common thread through both pre- and postnatal interviews was being shocked when receiving the diagnosis. Shock was a term that was spoken often and at times on numerous occasions during a single interview. Parents mostly used this term to describe their initial reaction to the news of the cleft: “She [the ultrasound technician] got to the lip and she kind of saw it, circled around and said, 'If you don't mind I'd like to bring the doctor in, I think I've detected a cleft lip.' And I was kind of very shocked. I mean I just remember being nervous and have a nervous look on my face, being shocked.” Another mother who experienced the prenatal diagnosis of her child's cleft echoed this mother's feelings: “They were able to get clear pictures

and almost while they were doing the sonogram, like I thought, as untrained as my eye is, ‘Hmm, kind of looks like there’s a space there.’ And then of course they said, ‘The doctor will come in and talk with you.’ I kind of suspected it, but at that point it was still a pretty big shock to me.” ‘Shock’ was also used in the context of coming to the realization that your child will not look like the average child: “You know, it was kind of horrifying. Because it’s a big shock, you know. I think just the shock of seeing that your kid’s gonna be born with something that’s, you know, not exactly normal.” Other parents were shocked by the fact that the cleft had not been diagnosed prenatally, yet they had received a number of ultrasounds: “We were kind of shocked because they got clear pictures of the face. We were surprised they didn’t pick it up. Now, we’re in a rural area, but the hospital, they never picked any of that up, which kind of shocked us.”

#### **4.1.1.3. Cleft cause and embodied knowledge (Lippman, 1999)**

Lippman (1999) conducted a qualitative study regarding prenatal testing in which she found that women’s stories went beyond the biomedical realm into, what she coined, their “embodied knowledge about risks, pregnancy and baby health, and of how this knowledge allowed them to negotiate and accommodate offers of testing”. In this study participants appeared to think about the cause of their child’s cleft by using biomedical knowledge while at the same time incorporating their personal values and experiences. In many families, ideas about what caused a child’s cleft appeared to be closely linked to feelings of guilt and blame. With certain parents this was related to a family history significant for cleft lip with or without cleft palate, but with other parents the guilt was linked to environmental factors. One woman whose child was diagnosed prenatally talked about sharing her child’s diagnosis with her mother: “My mom started crying and she said, ‘I prayed every time I was pregnant that my babies would be ok.’ No

one else in my family except my grandfather and [my son] have a cleft lip. So she thought they made it through, you know, like she didn't think that it would keep going." Another mother who found out about her child's cleft at birth said this: "Well, I kind of felt like maybe it was my fault in a way. I didn't really understand how it worked. My father was born with a cleft lip also and so I felt maybe that I had done something or there was something in me that caused it." In families in which there was not a family history of clefting parents searched for an explanation and many times pointed a finger at themselves: "You know, we don't have it in the family, what did we do wrong? I had an MRI at the beginning of the pregnancy without knowing I was pregnant so we were thinking, 'Was it the MRI, was it this, was it that?'" This theme of blame in the absence of a family history was present in other interviews as well:

I really want to believe it's genetic, [I] wasn't planning on getting pregnant and so I was smoking. Really, probably in my heart of hearts [I] feel that it is my fault. I know they don't know that, but it might be possible. It does me no good to sit here and beat myself up over it, I didn't do it on purpose. Had I been actively trying to conceive I would have stopped smoking, but by the time I found out they say it happens in the first seven weeks; I didn't even know I was pregnant until he had a cleft. It's in the back of my head that it's possible that it's my fault.

Another mother whose pregnancy was unplanned said this: "For awhile I kept thinking it was something I ate, something I did, but when you start looking at it and it happens so early you don't even know [you're pregnant]. The only thing I can think of is maybe I didn't have enough folic acid or something, because [my son] wasn't planned." Parents searched their memories to recall what they did during pregnancy that might have influenced the development of the cleft. One mother mentioned, "You know they say age isn't a big risk that it's more like a genetic thing and as far as we know, we don't have anybody on either side of the family. I've also heard about the folic acid thing and I felt like I ate really well during my first pregnancy and maybe I

was a little more lenient in my second one. I mean I still had a really good diet, I didn't have any coffee. Of course I'm reflecting on was it my fault, did I do anything?" A mother in the postnatal group was taking Zoloft and other medications at the time of conception: "I started blaming myself for being on the medicine and I took myself off of it, I really shouldn't have." A mother in the prenatal group had a stomach virus between the seventh and eighth week of pregnancy and questioned whether that was associated with the cleft. One mother felt the anxiety she experienced during her pregnancy was responsible: "What I read, other than it being genetic, [was] that it could be stress-induced early on in the pregnancy and I have to believe that's exactly what happened. Because we can't find it in the family anywhere and I was totally a basket case." Certain fathers felt that the potential environmental causes of an oral cleft were not valid: "They told us it might be something in the water. We're on well water where we live and after finding out about it I talked to some people that I work with and found out that their relatives had cleft lip and palate and they were all on city water, so that ruled that out." Another father also did some investigation into the environmental effects:

Now I hear people talking about it, in addition to heredity it being other factors, environmental. And at first we were thinking about when we had just conceived [my wife] had just gotten off the pill and I was on Vioxx at the time and you know, that's what I started thinking. But as I did research I found... you know they found statues of ancient Incas with a cleft lip. Now there was no Vioxx or birth control or electrical power lines or anything back in those days. So I honestly think its heredity, but I don't see any of that environmental crap.

#### **4.1.1.4. Parent to parent support**

An additional common theme identified in both pre- and postnatal participant interviews was the need for parent support. Many parents felt that other families who have a child with a cleft proved to be a unique resource, providing a different perspective than that of the health

professionals working with their child. In one mother's words, "there's nothing like talking to another mother." A mother whose child was diagnosed prenatally explained that she spoke with another couple after her child was born. Her comments were similar to those of the previous mother: "I think there's just something about a mother's point of view that the doctors didn't really have." Other parents provided a deep understanding of the experience because they have dealt with it themselves:

I think it would most helpful to any mom in this situation to have a support network of other mothers of cleft children. I have found that shared experiences are the most valuable and could help dispel a majority of the unknowns and anxiety. There are also so many things that could be shared to aid in caring for the special needs of these children.

Parents of children with oral clefts are able to share experiences that extended family members cannot provide. A mother who found out about her child's cleft at birth said: "With the parent of another child with a cleft they actually know what you're going through, where [your own] family is just kind of like, 'Oh everything's fine, don't worry about it.' You talk to other parents and they know what you're talking about, they understand things." Some fathers agreed as well: "They know what it's like to first find out and know nothing about it and they can really give you the ins and outs and really appease you." For those parents who did meet with other families, this was described as an uplifting experience. In many cases these meetings happened serendipitously: "There was a nurse who worked in the hospital that had a daughter who was born with a cleft 13 years ago and she came in and sat by my bed in the middle of the night, because she worked the midnight shift. She helped get me involved with the cleft palate center and told me, 'This is where you're going to go.' [She] brought in pictures of her daughter; she had gone home on her lunch hour and gotten those." Another mother in the prenatal group

described a chance meeting with an extended relative: “I actually did network with a couple of people that I met by just a fluke. One happens to be my cousin’s wife’s sister and her baby had a bilateral [cleft] two months before my son. And I mean her and I developed a support network that was fantastic for one another.” Other parents are able to discuss details of care and provide valuable tips, especially in the preparation for surgery, a time when many parents are extremely anxious. One mother talked about the change in her child’s personality after his first surgery and noted that he was completely off schedule. “You don’t know if that’s normal or not because you don’t have anyone else to talk to. No one really told me that every child is different.” Another participant felt that meeting with a parent would be useful because there are other medical issues associated with clefting, such as asthma and gastrointestinal problems that parents sometimes do not know about because physicians focus on the plastic surgery.

#### **4.1.1.5. Not a special needs child**

Some mothers from each group felt distressed when their child underwent his or her lip repair because of the drastic change in their appearance. A mother in the prenatal group expressed this by saying, “You get really accustomed to seeing them with the [cleft] lip and... it really grows on you. He just looks like a completely different child to me [after surgery].” Mothers became accustomed to their child’s cleft and missed the wide smile associated with the cleft. As one mother whose child was diagnosed at birth said, “ Looking at him... prior to his operation it was like, ‘Yeah, he had the cleft, he had it wide’, but I wasn’t looking at him as being a non-attractive child, let’s put it that way. I thought he was as cute as a button. Cause he had the widest, cutest smile, it was like I hated to give that up.” One mother claimed to miss the cleft after her child’s surgery: “They look so cute when they smile. You know, it’s so wide, you really miss it after the

surgery.” Some parents seemed to struggle with the lip repair because of the radical change to their child’s appearance:

At times I just felt really guilty for even pursuing it. The day before she had her surgery my husband and I were sitting there and we were saying, “Yeah, I don’t even know if we really want to do this.” Because like, this is how we got her and maybe we should keep her like this. You know of course reason comes to mind and you think well she’s going to have a hard time eating, she can’t drink, breathing and everything else, physically she needs to have it done. Cosmetic, yeah that’s nice, but physically it’s a help. Physically we wanted her to have her surgery, but cosmetically it was one of those, well, we got her this way who are we to change it?

No fathers verbally reported this attachment to their child’s cleft in their interviews.

This attachment to and acceptance of the cleft may be tied to another pattern observed in the data, that parents do not see their child as having a disability. As a mother whose child was diagnosed at birth puts it, “Even though it’s considered a disability I don’t really look at it that way because it’s fixable. I mean even if they’ll have surgeries, they’ll have speech problems, it’s something that is fixable.” This notion of the defect being ‘fixable’ was also noted in other interviews. One mother explained that her experience started out really ‘shaky’ because she and her husband did not understand what was going on, but at the same time they did not view the cleft as a disability. She states, “I believe she was born that way, but I didn’t see it as a defect. A defect is something you can’t fix.” Another participant shares a similar perspective: “I didn’t see him as a child with a special need. I knew that need was there, but I didn’t look at him that way.”

#### **4.1.2. Prenatal diagnosis**

All of the participants in the prenatal diagnosis group were glad they found out about their child's cleft before the birth. Parents cited different advantages and disadvantages of a prenatal diagnosis. Although some negatives associated with a prenatal diagnosis were mentioned by the prenatal participants all felt it was better knowing ahead of time. Participants in the postnatal group were also asked to describe their thoughts on prenatal diagnosis. Interestingly, some of these parents felt that a prenatal diagnosis of a cleft would be useful, but others did not agree. The themes generated from the transcribed interviews with prenatal participants are described in the next section.

##### **4.1.2.1. Coping**

A primary advantage quoted by the study participants for having a prenatal diagnosis of a cleft is the ability to cope and come to terms with the cleft diagnosis before the baby is born. By the time of the birth the parents have had time to adjust to the diagnosis of a cleft in their baby. The theme of "coping" was broken down into subthemes which include "coming to terms with the diagnosis," "acceptance time period," "loss of perfect baby," and "religion." Each is described in the following sections. This "coping" theme is only discussed in relation to the prenatal diagnosis participants. While participants in the postnatal group could imagine how a prenatal diagnosis would impact the birth experience, it was decided not to include their opinions in this section.

### *Coming to terms with diagnosis*

A prenatal diagnosis allows individuals to cope and process their child's condition before they ever actually see their baby at delivery. Many parents emphasized the shock associated with receiving a prenatal diagnosis of an oral cleft. How then do parents overcome this shock and move on to accepting their child's condition? One mother in the prenatal group stated, "It was very beneficial to know prior to birth." She was asked to expand upon what she meant by "beneficial" and she offered the following explanation, "The part of it being a shock was over, it was dealt with prior to birth, rather than having a baby and going through all the postpartum stuff on top of just finding out." A prenatal diagnosis may allow parents to "come to terms with the diagnosis" during the pregnancy. One mother explained that the first two weeks following the diagnosis were a very difficult, emotional time filled with tears and questions of "Why me?" When this participant was asked how her situation then improved, she responded, "You know, you just adjust, you read more about the cleft. ...We tried to talk and then things were sinking in." This sense of allowing the information to "sink in" was referred to by others as well: "It definitely gives you a chance to absorb it prior to just having a new baby and going through a delivery or a c-section, whatever it may be." Another mother discussed that her coping included becoming educated and having additional testing to find out if her baby had a chromosome condition. This lessened her anxiety by allowing her to view the cleft in a more favorable light. She explained, "Once we knew, we got educated, we met with people, it was a non-event. Once we knew [he] didn't have other developmental problems we were fine with it. We knew it wasn't going to be a piece of cake but it wasn't a horrible prognosis." One father expressed reassurance in knowing about the cleft ahead of time. "Well it was comforting to know what to expect; the worst-case scenario, the best case scenario."

### *Acceptance time period*

A prenatal diagnosis gives parents some time to digest the diagnosis of the oral cleft and, in general, individuals find this useful. This time allows them to “come to terms with the diagnosis,” the theme presented in the preceding section, and accept the fact that their child will be born with a cleft. One mother talked about how having a couple days to contemplate the news allowed them to move forward with a plan: “It really only took us a day or two to say, ‘Phew, ok, let’s get back to normal and figure out what we are going to do about this.’ It was good to have that couple of days to kind of get the emotions, or shock, out of your system and then go forward with the research.” Interestingly, this participant used the word shock as she described this “acceptance time period.” Other mothers appeared to feel the same way: “After it settled down a little bit, it took me about a week to adjust to the fact that he was going to be born with this.” Other participants were glad to have some time to deal with the diagnosis themselves before discussing it with family members. One mother said, “Actually now that I’m thinking about it I didn’t even really want to talk to people about it until it was like six weeks and then I started to really want to talk to other people. I think it takes a little bit of time.” Another mother said something very similar: “Once we did find out she was having that [the cleft] it took us awhile to kind of tell anybody. We had to kind of accept that for ourselves, you know.” It appeared that prenatal diagnosis gave these women time to accept the diagnosis. It also seemed to allow them to become accustomed to the idea before they told family members about it and before the birth of their child. One participant was explaining the pros and cons of prenatal diagnosis and provided a summary of her experience by saying: “I think overall knowing beforehand does help because it gives you those months to get through it.”

### *Religion*

Interestingly, a number of mothers and fathers talked about how their belief system helped them through the difficult time after their child's cleft was diagnosed by ultrasound. Those participants with a strong faith looked to God during this time for help. One mother found consolation in believing that God had chosen her and her husband to be the parents of a child with a cleft:

After leaving and searching on the web and everything, things start to settle down in your mind and then you get ready for it and then you think that maybe God thought that I am a pediatric dentist, my husband is a child psychiatrist, so maybe God planned that it would be better with parents like this than with other parents. You know, it's a consolation because we might be the right parents for a baby with a cleft.

One participant explained that his mother was having a difficult time dealing with the news of the cleft. He finally said to her, "Look, this has happened for a reason, God has a reason for this happening and we don't know what that is yet but we are going to trust in Him and we're all going to pray." Another father also spoke of the power of prayer when he said, "One of the big things for us was to have people praying for us. I told family and friends relatively soon after we had the news. I think that was the big motivation there, to have people holding us up in prayer."

### *Loss of the perfect baby*

A coping subtheme labeled, "loss of the perfect baby," was less prevalent than the previous subthemes, but emerged from the interviews with participants. This theme refers to the grieving process that some parents go through when they find out their baby is going to be born with a birth defect. Parents come to the realization that their baby is not going to be "perfect." As one mother puts it, "You know, obviously it's a shock when you find out your child's not going to be 100% normal." Another mother spoke about what the loss of a perfect baby meant to her,

“...thinking, ‘Oh my gosh, I’m not going to have the perfect baby’ because you do everything perfect while you’re pregnant and she’s not born perfect.” One mother delved into more detail as she described the mourning process she went through when the cleft was diagnosed on ultrasound:

I think the most important [thing] is like, you do your mourning... When somebody dies, you mourn for awhile. So doing that before really helps because after you really have the postnatal depression, or the baby blues. You’re tired, you’re sleep deprived, you cry for nothing and adding this I think would have been very hard.

The intense emotional responses to the prenatal diagnosis of the cleft these mothers expressed were quite poignant. Of note, no fathers addressed this theme of “loss of the perfect baby.”

#### **4.1.2.2. Preparation**

A prenatal diagnosis also allowed the parents to prepare themselves and their families for the birth of a child with special needs. Many participants had ample time to read extensively about feeding, surgeries and other challenges for children with oral clefts. Some met with the Cleft-Craniofacial Center and actually spoke with the surgeon who would be performing their baby’s operation before their baby was born. This theme was broken down into subthemes which included “knowledge is empowering,” “planning for the baby’s needs,” “telling family and friends,” and “additional testing.” As noted in the aforementioned section only responses from participants in the prenatal diagnosis group are included in this section.

##### *Knowledge is empowering*

This subtheme refers to the fact that most parents searched for as much information as they could find when they received the news of the cleft. Gathering information enabled them to feel empowered so that they understood the diagnosis and its implications. Simply stated, one mother said, “Knowing helps you be informed.” A number of participants discussed the use of

the Internet, which will be discussed in greater detail later in this document. Because of the unfiltered information on the Internet, the Cleft-Craniofacial Center at Children's Hospital in Pittsburgh has composed a list of websites that is frequently distributed to its families. One father was discussing this list and stated: "After we found out the doctor gave us some web sites to check into and we did some research on it, which was very helpful." A mother talked about how she prepared herself for the birth of her child by searching on the Internet and becoming familiar with the terminology the doctors were using, such as bilateral, complete, and incomplete. This information seemed to help her better understand the doctors at the clinic, enabling her to become a more active participant in her child's care. Another mother thought knowing was useful in preparing her for the birth of her child because "[I] had all the information about breast feeding, feeding the baby, how everything should be done. You know it's better to be aware than not." Many parents, when describing the experience of receiving a prenatal diagnosis, claimed that knowing ahead of time was better than the alternative. One mother in particular felt that arming herself with as much information as possible would aid in the preparation for the birth of her child:

I mean I have to tell you I found myself from the time I knew on looking at my sister's medical journals, I went to bookstores and went through all kinds of books and I searched for pictures. Because I hadn't known anyone with this, I was looking for the terminology, I was looking for success stories, I was looking for anything I could find. ...It gives you a chance to do your homework. I mean, I'm speaking from my perspective, how I handled it, but I think you don't really have a choice.

#### *Planning for the baby's needs*

A prenatal diagnosis allowed parents to educate themselves in terms of the physical and medical needs of their child before he or she was born. Parents were, in essence, able to line up a "game plan." As one mother described:

I mean, when I met with my pediatrician in an interview I was able to tell him, prior to my son's birth, this is what I was facing. Things like that I think it was helpful to basically line up a game plan. I met with the surgeon; I knew that I would retain him. I had met with the cleft clinic and gotten my bottles. In that sense I think it was helpful.

Other parents liked the fact that they could meet with the surgeon and the cleft team before their baby was born as well. One mother was describing how prepared she felt for the birth of her son and spoke of meeting with the surgeon. She said, "We were really very relaxed and very happy that he [the surgeon] was here. It was really great to know that we have experienced people around." Many parents interviewed in both the pre- and postnatal groups had to make long trips to Pittsburgh to the cleft clinic. To know ahead of time that they would be making the trip quite frequently was helpful to some parents. One mother explained this by saying, "Just getting prepared as far as we knew we would be making trips to Pittsburgh at least once a week for the first three months of his life." There was one father in particular that was extremely grateful for having known ahead of time and for understanding the surgeries that would take place after his child's birth. The delivery of their baby took place in a smaller hospital. This father felt had they not known about the diagnosis prenatally, their baby would possibly not have received optimal care. He said:

The big thing is, and I'm just going to elaborate on this, the big thing is I'm so glad that we knew a little bit more about it because we were able to make better decisions in the delivery room. We talked to different doctors, you know the best case, or the best procedure we found out was to wait a little while before you operate. Give him a chance to grow a little bit. There were people at the hospital [where he was born] that said 'Well, we could have him fixed up today.' We found out that's not necessarily the best way to handle that. Had we not known any better we may have just went along with that to the long term detriment of our baby. I'm thankful to be informed. That was a big, comforting thing to me.

### *Telling family and friends*

A clear advantage for parents was the ability to communicate the cleft diagnosis to family members and friends before the arrival of their baby. Many parents described telling “everyone,” as this mother did: “Everybody knew the baby had a cleft, like you know, the immediate family. So it wasn’t a shock to them. It made it easier because they could prepare as well. They can believe me, but they probably went on the Internet and educated themselves as well.” Another mother said telling family members before her child was born was much better than having them call her after the birth and being forced to tell them about the cleft. As parents educated themselves, they also educated their family. One father even spoke about telling the people at work. “They asked about it and I told them too.” For families with little children at home, the prenatal diagnosis allowed parents to prepare them. One mother described this situation in her home: “Being able to help my older son know what was going on, so he knew all along that [his brother] had a boo boo on his lip and the doctors were going to fix it; that actually helped our five year old a great deal.” Another mother also used the term “boo boo” to describe the cleft to her other child: “It helped us prepare our three year old son. He knew he was getting a baby sister and she was gonna have a boo boo on her lip. He pretty much told everybody that he had a baby sister coming. He knew, ‘Don’t touch, its an owie’. So that helped out a lot for him.” Parents in general felt that telling their other children, as well as young members of the extended family like nieces and nephews, allowed the children to understand and accept that the new baby would look different than other babies.

### *Additional testing*

Due to the possible association of a chromosome abnormality with a cleft diagnosis, some women chose to have an amniocentesis performed. For those that did, this provided reassurance

and aided in their preparation because they felt more certain of what to expect in their child. A normal chromosome result made parents feel more at ease with their child's cleft diagnosis:

We met with a genetic counselor and discussed it and we decided to do an amnio and ruled out there was no chromosome abnormality. So then it was just the lip and palate and we weren't worried because that could be corrected.

Typically, the decision-making regarding the amniocentesis was stressful for parents, having to weigh the benefits and the risks (there is a 1/200 chance of miscarriage associated with the test). Once parents received the good news that the chromosomes were normal they experienced a sense of relief. However, parents of children with anomalies other than an oral cleft were not included in this study, so a generalization that amniocentesis routinely relieves anxiety cannot be made.

#### **4.1.2.3. Disadvantages**

As parents, particularly mothers, described their experiences with ultrasound and prenatal diagnosis they often referred to a negative emotional response they felt as a reaction to the diagnosis. At the same time, other parents could not think of a shortcoming to the prenatal diagnosis even when specifically asked. There were a diverse range of responses in regards to negative aspects of a prenatal diagnosis of a cleft by ultrasound. Of important note is that most parents in the prenatal group would opt for ultrasounds again in a future pregnancy including those parents who described negative impacts of prenatal diagnosis.

#### *Emotional disturbance of the pregnancy*

During the interviews most of the participants in the prenatal group were asked about the way in which the diagnosis of the cleft impacted the pregnancy. As noted above, a wide range of responses emerged, but many women felt their pregnancy was negatively impacted by the news

of the cleft. When one mother was asked how she felt after the diagnosis, she said (while laughing): “Bad! Very, very bad. Too many things were going on.” It seemed she was laughing with relief that, retrospectively, everything had worked out well in that her son is healthy and has already undergone his initial surgery. She went on to explain that initially she and her husband felt like they were living in a fog. She was constantly crying and couldn’t sleep at night. The question, “why us?” plagued her mind. She said she was glad to have known before her son was born, but from her story it appeared that the course of her pregnancy was troubled by this news. Interestingly, this mother said, “You know, it is better to be aware than not. But the thing is I did not enjoy my pregnancy. I was not, you know, a very happy pregnant woman.” Other mothers similarly expressed this sentiment:

And then, the bad thing is it sort of for awhile puts a damper on the pregnancy, it makes you not be as excited. You think, “Why is something wrong?” You go through the whole, “Why does it have to be me, why does it have to be my baby?”

This constant questioning these mothers experienced seemed to reflect the turmoil of emotions they felt during this time. Other mothers also explained that they were upset, they cried, and they had unanswerable questions. One mother shared that her pregnancy changed because she became obsessed with her quest for information, as she put it, “the overkill of searching for information, reading.” She said she thought about it every day and felt “very nervous and anxious” though she did say she was still happy about being pregnant. That is in contrast to a father who explained that the diagnosis in his unborn child caused so much stress within his family that he almost wished his wife were not pregnant. His own mother underwent a very negative reaction to the diagnosis in her soon-to-be grandchild. This father said:

My mother's attitude on it got me to the point where I didn't (pause), I wasn't even excited about the pregnancy anymore. I didn't even, you know I wasn't thinking abortion or anything (laughs), but I was almost to this point and I was just wishing she wasn't even pregnant so we didn't have to deal with it.

During the interview he appeared to struggle to find the right words to explain his emotions, evidenced by the pauses in the flow of his conversation. Possibly his laugh was an attempt to lighten the weight of this issue. He indicated that his mother eventually accepted the diagnosis and before his child was born, he once again looked forward to the new addition to the family.

Women who went through multiple ultrasounds before their baby was diagnosed with a cleft experienced anxiety. Many times there was a "lag time" between the two ultrasounds and during this time period these women described feeling confused and worried. One mother who had two ultrasounds before they positively identified the cleft was concerned about the possibility that the baby had additional, more severe birth defects. She said:

So the hard part was I guess from the first one where [they said], "We're not sure, maybe, but you can't get an appointment from a week to ten days to confirm." That was a little tough. I guess during that time we were hoping that that's all it was. If it was there that's all it was.

This woman's description of hoping that the cleft lip was isolated without other birth defects emphasizes the worry she may have felt during those ten days between ultrasounds. Another couple who received their first ultrasound at a local hospital had a similar experience. It was not until a week later that they received a phone call that something was seen on the ultrasound and they were told they would have to travel to a tertiary center for a more detailed sonogram. Yet another woman was informed during her initial sonogram that everything looked fine, but she

then received a call the next day that her doctor wanted her to come in for an additional ultrasound. Apparently the facial pictures from the first scan were not very clear. The cleft was then identified during the second scan.

#### *No disadvantages*

There were parents in the prenatal group of participants who felt there were no disadvantages to learning that their child had a cleft before birth.

“Disadvantages, you know it’s kind of hard to say, is there a disadvantage? Because you find out, I mean it’s better to find out beforehand I would think.”

“Disadvantages, honestly, I can’t think of a disadvantage.”

“I don’t think there’s any disadvantages. I think it’s the more you know the more helpful it was.”

One father had a very interesting response that differed from other participants’ responses when they asked about disadvantages to prenatal diagnosis. Though his opinion does not reflect the majority it is an interesting perspective: “We’re pretty much a pro-life family, the only disadvantage I can see is for the baby if the parents weren’t. With the political climate and what you’re allowed to do now you know you can terminate the pregnancy. So the only disadvantage I see is for the baby.” Finally, one mother explained that she saw no disadvantages to prenatal diagnosis and she believed the diagnosis normalized her pregnancy. She said, “If anything it helped me to have a normal last couple of weeks. Of course that was in the back of my mind but it didn’t really affect the actual pregnancy”.

#### **4.1.2.4. Alternative perspectives**

Although the participants in the postnatal group did not experience a diagnosis through ultrasound, their opinion was sought and similarly, answers varied. Some parents felt that knowing ahead of time would have eased the shock they felt during the birth of their child, yet

most of the postnatal participants felt that they would have been anxious about the cleft throughout the pregnancy. Some parents felt that, although it would cause them worry, they would still want to know. Others felt that because it would only cause them to worry, they would not want to know. For example one mother in the postnatal group said that knowing ahead of time would have only made her upset. When asked about her thoughts on prenatal diagnosis she said, “I started thinking about that, I would have probably been upset the whole entire time, but then I started thinking if it would have been more serious, I would have been able to abort the baby.... If it was something more serious... would I even do that.... So I don’t know, maybe its better that I didn’t know at all.” Another mother also felt that a prenatal diagnosis would have induced anxiety. She felt that the birth of her child with the cleft had been “a breeze”:

I think for me it would have been nice to know [prenatally], but I’m glad we found out the way we did because everything has been you know, kinda like a breeze. We understood everything and they explained it to us at the cleft clinic and we were fine. But I think had I known early on in the pregnancy I would have been more worried about that than enjoying the pregnancy.

When asked, “So how do you think the prenatal diagnosis would have affected the pregnancy?”, she responded with:

I think as far as, I would have been so focused on that. You know because when we did come home from the cleft clinic we went on the Internet to check everything out. We tried to get as much information as we could. There’s such severe pictures on the Internet and I think that I probably would have done the same thing if I had found out when I was pregnant and I think that would have scared me more. I would have thought things were a lot worse than they were.

Some fathers also felt that knowing ahead of time would not have been advantageous. One father simply stated he would not like to know about the cleft lip before the baby's birth. He did not expand upon his response even when asked additional questions. Another father described the negative experience he and his wife had with the doctor who informed them of the cleft in the delivery room. This father felt that if the physician informing the couple was not sensitive and reassuring, then finding out ahead of time would be a disadvantage for the parents because it would only cause them concern. However, another father in the postnatal group felt that he would have liked to have known prenatally to ease some of the shock he felt initially in the delivery room. He said, "It wouldn't have been such a shock when he first came out. I could have researched it, known a little bit more, be a little more prepared. I guess more prepared mentally than when he was first born." Some mothers in the postnatal group also felt there were significant advantages to knowing ahead of time. These mothers generally felt that the knowledge would have caused them anxiety, but that they would have been able to prepare themselves.

If I had a choice to decide if I wanted to know or not I would want to know. Just because I am an information person. Although it would worry me sick, I would be probably be at a higher stress level than I was this time, but it was one of those things that I calmed myself down real quick once I got the information I needed. I think looking back on it I could've had a more favorable birth experience. Even if I didn't tell my family prior to the birth I could've had enough information where I could've helped them ease into it.

Another mother quoted preparation as an advantage of prenatal diagnosis. This family had difficult financial issues especially with health insurance after the birth of their baby. This mother said, "I would have liked to know early, that way we could have prepared financially and emotionally and getting doctors lined up rather than the hospital scrambling to figure out what

was going on... We could've kept up the insurance without having to beg, bargain and plead from other sources." Other mothers simply felt that knowing ahead of time would have eased the shock of the diagnosis at birth. One mother said, "You know, it wouldn't make any difference, we wouldn't have done anything different had we known. But, if we would have known...had it not been such a surprise... You know we didn't expect this at all. Just kind of the initial shock of the whole thing. That would've been nice if we would've known. I don't think there would be any disadvantages of knowing. Not in my situation."

#### **4.1.3. At birth diagnosis**

The experience of parents whose children were diagnosed with a cleft lip at birth was also elicited. As discussed in section 4.1.1.2, these parents spoke of the shock they felt at receiving the diagnosis at birth, but for the most part these participants seemed to come to terms with their child's diagnosis and quickly found resources available to them. One mother spoke in detail about her experience immediately following her child's birth. For her, not being informed about the cleft was the most frustrating and difficult aspect. She wished she could have been informed so that she could help her family understand the circumstance. She explained:

I felt very helpless. I am an information gatherer... I wasn't prepared for this. For all my family coming in, they too had a lot of questions and I felt very helpless because I couldn't answer them. I didn't know what her chance of survival was and was she ever going to be able to eat, was this something that was going to follow her for the rest of her life?

Despite the anxiety this mother expressed, she went on to explain that she and her husband had very high hopes for their child as soon as they met with the staff at the cleft clinic. She explained that this was difficult because the way she and her husband dealt with the situation was

different than what was expected of her by her family. She expressed that this left her feeling guilty:

I think after we left the hospital was the hardest because everybody expected my husband and I to have a nervous breakdown. We didn't. I felt kind of guilty because I didn't. After... visiting the Cleft Palate Center and meeting with the surgeon I had such high hopes and outlook on everything that I no longer saw this as a bad thing. It was just something I was chosen to deal with and move on.

Parents who are given the diagnosis at birth might not have the same length of time to speculate and dwell on what their child might be like as those who find out prenatally. Some parents even felt that the cleft was not a big deal. One father said he felt “a little bit surprised because we didn't know, but other than that it wasn't a big deal.” Another mother felt the same way: “I didn't know until he was born. I wasn't upset at all, I just asked the nurses if he would be able to breast feed. They were really surprised that I wasn't upset.” On the other hand, the birth of a baby who is different than what was expected did have an affect on some parents. The greatest concern of these participants was that they did not have much information on what a cleft lip actually was and how to treat it. As one father said, “When he first popped out I was wondering how he was going to breathe and how he was going to eat. I didn't know anything about it. I knew they could fix it, but I didn't know much about clefts.” Another mother explained that the experience in the hospital was scary because nobody knew how to feed him. One mother did say that she had feelings of depression following the birth of her child. This sounded similar to the emotional disruption of the pregnancy described by some mothers in the prenatal group, but was only voiced by a small proportion of participants in the postnatal group.

And then too after you have a baby there's a time period where you kind of go through, you have feelings of depression.... It seemed to be more extreme with [my daughter with the cleft] than it did with my first one.

Of note, one of the reasons cited as an advantage of prenatal diagnosis by participants in the prenatal group was being able to prepare their other children for the birth of their baby with the cleft. Interestingly, parents in the postnatal group felt that their other children handled the birth of the child with the cleft extremely well. A number of participants discussed this during their interview. One mother talked about her son's reaction to the new baby:

When he came in he was really excited. He said he just wanted to hold his new sister because he had sang to her every night when she was still in my belly. He would lay and pet my belly and sing to her and he just wanted to hold her because he had been waiting so long. And he looked at her and his look was not a look of horror or a look of concern, it was just pure love. He had no desire to, you know, he didn't want anyone to ask questions, just this was his little baby sister.

Another mother talked about a similar experience with her own daughter. She and her husband had explained to their daughter that the new baby looked different, but that he would be "fixed". This mother said, "We were kind of scared that she would be afraid to touch him, but she took to him, she didn't really see it at all."

Some participants in the prenatal group expressed their view of what it would have been like to find out about the cleft in the delivery room. The general consensus was it "would have been worse" than finding out prenatally. In fact, there were not any parents in the prenatal group that would have preferred a postnatal diagnosis. Certain participants tried to imagine what it would be like in the delivery room. One father stated, "I couldn't imagine what it would be like when

he was born if I would have only found out then and I knew nothing about anything. That would have been awful.” Another father agreed: “I feel like knowing ahead of time was a lot better than being surprised in the delivery room. I think that would have been a little bit more traumatic.” One mother recalled all of the emotions she felt during the prenatal diagnosis and explained that she could not imagine having all of those emotions when she was “laying on the table.” She felt it would have emotionally been more difficult. After one mother explained the negative impact the prenatal cleft diagnosis had on her pregnancy she was asked if she was still happy she had found out before her child’s birth. Her response was, “Oh definitely. Like now with the second baby I definitely want ultrasounds. I have no regrets, me or my husband or anyone in the family.” Another mother also explained that the prenatal diagnosis had negatively impacted her pregnancy, but when she thought of the alternative she said, “I think that would have been worse if I didn’t know. If they would’ve just handed me this baby and said, ‘Ok, what is that?’” Parents in the prenatal group seemed to feel that the shock of the experience would be significantly worse had it occurred in the delivery room.

## **4.2. OTHER ISSUES**

Parents were asked their opinion of specific topics during their interview including pregnancy termination, genetic testing, and use of the Internet. Because these questions were specifically posed to the participants we did not feel that they should be included with the other themes generated from the participants responses to more general questions about their experiences. Upon reviewing the literature, these issues appeared numerous times and we felt that it was important to address them.

#### **4.2.1. Pregnancy termination**

There was not a single participant in this study that mentioned he/she would consider terminating their pregnancy based solely on the finding of an oral cleft on ultrasound. Most participants in both the pre- and postnatal groups were asked this question:

When parents are given a diagnosis that their child has a cleft lip, many thoughts about the pregnancy and raising a child who will need multiple surgeries crosses their mind. Some parents consider ending the pregnancy and some parents don't. Could you tell me your thoughts about this issue?

Most participants indicated that they did not think about aborting the pregnancy. One mother explained that she was too far along to even consider abortion:

I never considered that. I was too far along, I knew that it wasn't a defect that was going to affect his quality of life so to speak. What did scare me is a lot of the reading I did said the cleft is sometimes involved in other syndromes. That scared me, but I never considered at 20 weeks terminating the pregnancy.

Another mother in the prenatal group felt that God would not have allowed her to become pregnant if He did not want her to have the baby. Some participants simply felt that a cleft was not significant enough to warrant consideration of abortion. A mother in the postnatal group said, "I know it's not a major deal. I would never consider ending a pregnancy over a cleft. I mean, even if they'll have surgeries, they'll have speech problems, its something that's fixable. My thought is you don't end a life because of that." Other participants also felt that an oral cleft is not serious in comparison to other birth defects.

A few participants did consider, or would consider in the future, termination if the anomalies involved more than a facial cleft. One couple was concerned about the risk of Down syndrome in their baby during the pregnancy. The wife explained that when Down syndrome was a possibility she and her husband did discuss the option of abortion. She went on to say, “But I think it was the anger of the moment. You think, we don’t want to have more trouble, it’s going to be complicating your life, the baby’s going to suffer a lot, there’s a lot you’re going to give up.” After the amniocentesis results came back normal, the couple commented that abortion was no longer discussed.

#### **4.2.2. Genetic testing**

Currently, research on craniofacial abnormalities is often focused on identifying genes that contribute to cleft lip and palate. Determining the genes involved with clefting will not only give insight into the etiology of the condition, but may also be used to develop genetic tests that would determine the probability for an individual to have a child with a cleft. Participants were asked whether or not they would be interested in such a test, possibly to aid in their future reproductive planning. The responses to this question were varied. Some parents felt that genetic testing results would have an impact on future reproductive planning, but would not be the only factor to consider. One father said he would be interested in a genetic test “only to see if that was a possibility.” He went on to say, “I don’t think that would change what we would do. We pretty much believe that it’s a little out of our hands. If God wants you to have a kid, you’re going to have a kid.” One mother was not sure she would want to be tested, but did say, “It wouldn’t deter us from having more kids, although it might be nice to know.” Another reason parents gave for a desire to undergo genetic testing was for preparation. A mother explained that it would be nice to know her risk, but “you don’t love them any more or any less,

it's just nice to know so you're prepared." Other participants were not sure if they would be interested in genetic testing. They did not see the value to being given a risk figure.

I'm not sure. If I would get pregnant and have another [child with] cleft lip and palate that would be fine with me. I don't know if I would want to know the percentages of that.

Another mother expressed a similar view: "Its probably going to come back with something that would say you have whatever percent chance. I don't know if that's of any value."

#### **4.2.3. Internet use**

Many sonographers and doctors advise against searching the Internet for information when parents are given a prenatal diagnosis of a cleft by ultrasound. Understandably, individuals may desire to find out as much information as possible and the internet provides easy access to information. Unfortunately, most of the information on the Internet is unfiltered and some of it is even inaccurate. Participants agreed that the pictures posted on the Web are usually the worst-case scenarios. When parents visit the Cleft-Craniofacial Center at the University of Pittsburgh, they are given a list of web sites that provide detailed, accurate information for parents. Most of the study participants searched the Internet for information; some found it useful, while others found the information and pictures to be frightening. Those parents that utilized the Internet frequently were selective in the sites they visited. One mother who appeared quite adept at searching the web said:

I am very aware of 'Don't believe everything you read.' To tell a person that is not comfortable with the Internet that that's where they should go for information, I'd be very leery about that. There's so much bad stuff on there and stuff that's really not positive and it may not even be true. You can put anything you want on the Internet. But some of the things that I did find were stories, personal acclamations of where they went, who they saw, where the best places were.

Some participants felt that the pictures they saw were frightening. The fear engendered by the pictures may have been compounded by not knowing how severe their child's cleft would be or the extent of the surgery their child would need. One mother explained that her husband got upset after looking at pictures on the Internet and said that he refused to look at any additional pictures. Some parents found the information and pictures on the Internet to be a somewhat distressing but also helpful. One father said, "Some of it was stressful because they showed you the worst case scenarios and you're like 'Oh my goodness!' but then it was also encouraging... because they showed the kind of work that they were able to do." Another mother also saw both positive and negative aspects of the Internet. When asked about her experience on the web she said, "Frightening, some of those web sites are very frightening. But some of them are very helpful." One mother in particular was very involved with Internet support groups and chat rooms. Originally, she sought answers to her questions regarding her child's surgeries and medical care, but now she actually answers other people's questions. Some parents suggested that other parents are often the best resources because they have firsthand experience. The Internet can be a good place to provide other parents with support. The next chapter provides a summary of the results, highlighting important conclusions.

## 5. DISCUSSION

The goal of this study was to explore the parental experience of a prenatal diagnosis of CL±P. This was achieved by recruiting two different participant populations, 1) parents that had received a prenatal diagnosis of their child's cleft (prenatal group) and 2) parents that found out about their child's cleft in the delivery room (postnatal group). Open-ended interviews were conducted with participants (mothers and fathers) to learn about their experience upon receiving the diagnosis of their child's cleft. Thematic analysis of the data identified a number of themes regarding this experience. Several themes identified were shared by both groups of participants. These included "receiving the diagnosis;" "shock;" "cleft cause and embodied knowledge;" "parent to parent support" and "not a special needs child". Specific themes identified in the prenatal group included "coping;" "preparation;" and "disadvantages". Each of these overarching themes was broken down into subthemes. "Coping" subthemes included "coming to terms with diagnosis;" "acceptance time period;" "religion" and "loss of perfect baby". "Preparation" subthemes included "knowledge is empowering;" "planning for the baby's needs;" "telling family and friends" and "additional testing". "Disadvantages" subthemes included "emotional disruption of the pregnancy" and "no disadvantages". Experiences of parents in the postnatal group were summarized under the themes "alternative perspective" and "at birth diagnosis". Other issues covered by the interviews included pregnancy termination, genetic testing, and Internet use.

In this investigation, 60% of the participants were given their diagnosis prenatally, while 40% of participants found out about their child's cleft in the delivery room. Themes identified from the interview transcripts gave insight to the parental experience of prenatal and postnatal diagnosis of a cleft. An interesting finding was that the experiences of participants within the two groups appeared more alike than different. What seemed to be the biggest difference in the experience between the two groups was the timing of the diagnosis, but parents otherwise seemed to go through similar reactions and emotions whether they found out about their child's cleft by ultrasound or at the time of delivery. Many parents discussed the time at which they received the diagnosis in depth.

Though a few of the participants were satisfied with the manner in which their physician informed them of their child's cleft, many participants expressed dissatisfaction with the way in which they learned about their child's diagnosis. This finding has been reported in the existing literature (Byrnes et al. 2003). Byrnes et al. found that, in terms of the informing interview, significant differences exist between parental experiences and parental preferences. In the present study parents expressed this incongruity by discussing their conversation with the physician and going on to explain how they would have liked it to occur. Parents' recommendations for receiving the diagnosis included allowing parents to see and hold their child immediately after delivery, providing a referral to the cleft clinic and providing a handout to give to parents with phone numbers or web sites to help them find more information. Participants also felt that a respectful, compassionate physician would make a positive impact on the experience.

In addition to discussing the delivery of the diagnosis by the physician, participants in both groups expressed the feeling of being shocked upon hearing their child's diagnosis. This shock reaction has been reported in the current literature (Sohan, Freer, Mercer, Soothill, & Kyle, et al., 2001; Davalbhakta & Hall, 2000). Sohan et al. (2001) reported that a prenatal diagnosis can help alleviate the shock which occurs when there is an unexpected cleft at birth. Another report stated "if the diagnosis is made correctly... it can help in decreasing the shock of having a baby with a cleft" (Davalbhakta & Hall, 2000). These findings were not fully supported by the current study, as participants in both groups expressed being in a state of shock upon hearing the diagnosis. However, it is possible that the type of shock or the extent varies depending on the context of the diagnosis (pre- or postnatal). For example, some of the participants in the postnatal group may have been shocked by the fact that their baby's cleft was not diagnosed by ultrasounds they had during their pregnancy rather than the presence of the cleft.

Lippman (1999) conducted a qualitative study to understand the decision-making process of women who had been offered an amniocentesis. She found that women's stories went beyond the biomedical realm into, what she coined, their "embodied knowledge about risks, pregnancy and baby health, and of how this knowledge allowed them to negotiate and accommodate offers of testing". In order to make a decision about having an amniocentesis these women made use of the resources of biomedicine, "while at the same time subverting their meaning and transforming them into strands woven into embodied knowledge which had meaning to their individual lives". The phrase "cleft cause and embodied knowledge" was taken from Lippman's work and used in this investigation to capture the participants' tendency to search for a cause of their child's cleft. As parents sought reasons for their child's cleft they made use of the medical information they

had available, but also wove their own feelings, experiences and values into the interpretation. Many parents knew of the multifactorial inheritance explanation of isolated orofacial clefts and applied it to their particular situation. Often times, especially in families with a significant history of clefting, feelings of guilt and blame were closely tied to explanations of cause. Parents who have a family member with a cleft may feel that it is somehow “their fault”. At the same time, participants who did not have a family history of a cleft searched for possible explanations and usually ended up pointing fingers at themselves. This process of searching for a cause may have had an impact on the coping process.

The theme “parent to parent support” is one that has been identified in the cleft palate literature. In her chapter entitled “Parents’ perspective on cleft lip and palate” Nicholson (2002, p.477) describes another family as being one of the most helpful resources for newly diagnosed families. She states that other families “can provide a wealth of information because they have been through the same things themselves”. Strauss et al. (1995) found that parents of a child with a cleft wanted more referrals to other parents than they had experienced. Additionally, the Cleft Lip and Palate Association (CLAPA) maintain a register of parents who are willing to contact new parents for support and practical advice (Forth, 2003). Existing literature in addition to the present study suggest that parents should be offered the opportunity to speak with another family following the diagnosis.

Although CL+P is usually referred to as a birth defect, many participants did not view their child in this way. The theme “not a special needs child” arose from these parental beliefs. Sagi, et al. (1992) referred to this “cognitive coping process” in their research as downward evaluation. The

process of downward evaluation enables an individual to restore their self-esteem, which has been overshadowed by the health problem (Sagi et al., 1992). Parents may perceive their own child's cleft as significantly less severe than they view clefting in general. Related to this parental perception was the report that some participants felt distressed after their child underwent his or her cleft repair because they had grown so accustomed to the cleft. Interestingly, Rey-Bellet and Hohlfeld reported this phenomenon in their study as well (2004). They reported that some mothers expressed distress after lip closure because they had to readapt to the baby's new facial features and expressions. These findings as well as the themes identified from this current study suggest that health care professionals should alert families to the possible distress associated with readapting to their baby's face after a cleft lip repair. It is possible that health care professionals would not recognize this possibility when it is assumed that the surgery to repair a cleft "normalizes" the "defect."

"Coping", a prenatal theme derived from the current analysis, which participants described as a benefit of prenatal diagnosis, was consistent with reports in the existing literature. Davalbhakta and Hall (2000) found that 85% of individuals in their study who had prenatal diagnosis felt it psychologically prepared them for the birth of a child with a cleft. Similarly, a study by Sagi et al., (1992) found that the available option of prenatal diagnosis seems to reduce uncertainty and restore parents' sense of personal control over their lives. Additionally, the majority of participants in these studies were glad they received their diagnosis prenatally. Prenatal diagnosis appeared to be psychologically beneficial to these particular parents. For instance, Rey-Bellet and Hohlfeld (2004) found that 28 out of 29 couples thought that prenatal diagnosis had given them enough time to work through their feelings and accept the child at birth. The

“loss of the perfect baby” subtheme identified in this study is supported by the cleft literature. Forth (2003) states “the painful process of coming to terms with the reality of having a child with an abnormality can be likened to the bereavement process, as they progress through the various stages of grieving, struggling to come to terms with their unique loss, that is the loss of a perfect child”.

In the current study, the breakdown of the “coping” theme into subthemes may provide additional insight into psychological preparation. For instance, the discussion of religion and the impact of faith on coping mechanisms by multiple participants in this study have not been addressed in the literature related to clefting, although the concept of spirituality is discussed within the scope of the genetics literature. In his review of genetic counseling literature, Saal (2002) discusses culture and religion and their potential role in prenatal decision-making. He states “management decisions that are made depend on the religious and moral background of the patient and their understanding of the implications of such decisions”. Some of the participants in this study seemed to feel that their religious beliefs allowed them to better cope with their child’s diagnosis of a cleft. Future studies could be conducted that closely examine the role of religion in the coping mechanisms of parents undergoing prenatal diagnosis of a cleft.

“Preparation” was the other theme that emerged from the interview transcripts in the prenatal group. This theme was divided into subthemes to better capture the participants’ experiences. Some of these subthemes, such as “planning for the baby’s needs,” “telling family and friends,” and “additional testing” have been previously discussed in the literature. In regards to the preparation theme, Rey-Bellet and Hohlfeld (2004) reported that 97% of the parents in their

study had a good understanding of their child's future operations and their timing. Another study by Matthews et al. (1998) found that seven of nine families had the opportunity to consult with the cleft team before the child's birth. In the Matthews study, all families who consulted with the cleft team before the birth of their child felt it was helpful in understanding the situation. Additionally, all of the parents who had the opportunity to consult with the cleft team felt that the experience was valuable and would recommend it to a friend (Matthews et al., 1998). Another study that supports this theme of planning for the baby's needs found that parents who received a prenatal diagnosis understood the "special nursing and feeding requirements of a cleft baby and were able to adapt to the special needs easily" (Davalbhakta & Hall, 2000). The "additional testing" theme elicited in this study has been previously identified in the literature as well. Johnson and Sandy (2003) cite the opportunity for more investigations for other structural anomalies and chromosomal abnormalities as a distinct advantage of prenatal diagnosis.

An additional subtheme identified within the "preparation" theme was "knowledge is empowering". Some patients described feeling empowered when they have adequate, appropriate information, which enables them to actively understand and influence their health care. This concept has been examined in the literature, though not specifically in the cleft-palate literature. D'Alessandro and Dosa (2001) state, "Without information, children and families cannot engage in meaningful discussions or make thoughtful decisions regarding medical care." Feeling empowered was closely linked in this study to participants' use of the Internet, since many participants gained information from clefting websites. Future studies may further examine the use of the Internet and its role in providing information to parents of children with clefts.

The “emotional disruption of the pregnancy” theme elicited from interview transcripts in the current study is supported, and was initially derived, by previous research (Matthews et al., 1998). Matthews et al. (1998) cite the emotional disturbance of a placid pregnancy as a reason given in opposition to the use of prenatal diagnosis for clefts. Higher levels of anxiety in women who have been told they have a higher chance of carrying a baby with an abnormality has been previously described (Forth, 2003). For example, in their study designed to investigate the potential relationship between psychological outcome and associated variables in women attending a tertiary referral center for prenatal diagnosis, Leithner, Maar, Fischer-Kern, Hilger, Loffler-Stastka, & Ponocny-Seliger et al (2004) found that the mood and anxiety scores of the participants were comparable to those of patients with a major depressive episode. This demonstrated that women facing a prenatal diagnosis experienced considerable psychological distress. Distress was not limited to the women with proven fetal malformations, rather all the women in the Leithner et al. study (2004) experienced acute distress, even those referred with the diagnosis of a sonographic sign. They concluded that the sole suspicion of having any prenatal problem may have a tremendous emotional impact on a pregnant woman (Leithner et al., 2004). This theme was also addressed in the cleft literature by Matthews et al. (1998) who found that three individuals out of nine felt that the anxiety produced by the prenatal diagnosis was a disadvantage. In another investigation in which all parents were offered professional psychiatric counsel following a prenatal diagnosis of a cleft, 7% contacted the referral specialist while 17% regretted afterwards not having done so (Rey-Bellet & Hohlfeld, 2004). These results, in combination with the supporting evidence from the current investigation, may demonstrate the

need to assess the psychological status of the parents and offer supportive services throughout their pregnancy.

In contrast, some parents in the present study could not identify any disadvantages associated with a prenatal diagnosis. Similar findings have been identified in other research (Sagi et al., 1992; Matthews et al., 1998). A study by Sagi et al. (1992), found that 82% of parents felt there were no disadvantages in performing prenatal diagnosis of a cleft. However, the population studied in that investigation did not undergo prenatal diagnosis and therefore were not speaking from personal experience. In the Matthews et al. study (1998) two-thirds of families (six out of nine) who underwent prenatal diagnosis of their child's cleft felt there were no disadvantages. This theme of "no disadvantages" may indicate that the medical team is providing more appropriate support to these families in comparison to the recent past. Future studies should be conducted to further examine this issue.

Interestingly, participants in the postnatal group of the current study were divided on their views of prenatal diagnosis. Some parents would have liked to have had a prenatal diagnosis, whereas others were satisfied with the diagnosis being made in the delivery room. These results appear to be somewhat consistent with previous research, which has examined perception of prenatal diagnosis in parents who received their child's cleft diagnosis at birth (Maes, Demey & Appelboom-Fondu, 1998; Davalbhakta & Hall, 2000; Wyszynski, Perandones & Bennun, 2003; Berk, Marazita & Cooper, 1999; and Sagi et al., 1992). The percentage of parents in the existing literature who would not have preferred prenatal diagnosis ranged from 9% to 41% (Sagi et al., 1992; Berk et al., 1999). There were also a significant proportion of parents who were not sure if

they were interested in prenatal diagnosis, ranging from 13% to 22% (Sagi et al., 1992; Wyszynski et al., 2003). Only one study found that all participants who received their child's diagnosis at birth would have rather had a prenatal diagnosis (Davalbhakta & Hall, 2000). In the future it is likely that the sensitivity of ultrasound in detecting clefts will increase. Therefore, more clefts will be diagnosed prenatally. These results may indicate that not all parents are interested in receiving a prenatal diagnosis. Although detailed ultrasounds in the second trimester of pregnancy have become more routine, these results may provide evidence that parents should be counseled about the possibility of detecting an anomaly before they undergo the procedure. Parents might be asked to consider whether they would like to be aware of that information before they have the ultrasound.

The topic of pregnancy termination was included in the current study in an attempt to clarify the perceptions parents have regarding termination based on the diagnosis of an isolated CL+P. The existing literature is somewhat contradictory surrounding this issue. The incidence of terminations based on the finding of an isolated cleft ranges from 0% (zero out of nine) in the Matthews et al. study (1998) to 92% (11 out of 12) in the Blumenfeld et al. study (1999). The high percentage of terminations reported in the Blumenfeld study, conducted in Israel, has been debated in the existing literature. Possible reasons cited for this high frequency of terminations include the fact that these clefts were diagnosed early in pregnancy by transvaginal ultrasound, the particular cleft team involved may have a greater perception of burden, and cultural differences may exist in how the anomaly is perceived (Johnson & Sandy, 2003). Parental opinion on terminating a pregnancy may be highly dependent upon cultural and religious backgrounds. Many individuals in the current study discussed God in their explanation of why

they did not consider pregnancy termination. In support of this, a study by Wyszynski et al. (2003) performed in Argentina; found that zero individuals out of 146 would consider termination. In Argentina 90% of the population is Roman Catholic and abortion is illegal. Regardless of the reasons why participants do or do not consider pregnancy termination, the current study seems to support the existing literature that an individual's desire to undergo prenatal diagnosis does not mean they would decide to have an abortion upon identification of an anomaly. In fact, the current study showed quite the opposite; all individuals in the prenatal group were glad they received the diagnosis prenatally, but none of them considered terminating the pregnancy.

There is a paucity of information in the literature regarding parental opinions of predictive genetic testing for isolated cleft lip and palate. For nonsyndromic CL+P “we are still far from having the genetic knowledge that is necessary to allow testing. Much progress has been made in identifying genetic loci that are potentially involved in the development of the anomalies, but much work remains” (Berk et al., 1999). Although these types of tests have not yet been developed for clinical use, it is important to understand the interest of parents in order to be prepared for the future. This study found that parental views are mixed, but the majority of participants expressed interest in this type of testing. On the other hand, some parents were not convinced of the utility of such testing since results would most likely be reported in the form of a risk percentage. However, other prenatal tests, such as the maternal serum screening and first trimester screening tests are reported as percentages and are widely used throughout the US population. Additional studies are needed that further address this issue.

Questions related to Internet use were included in the present study because of the lack of information in the literature examining this aspect of prenatal diagnosis. Most participants in this study felt that the worst- case scenarios were posted on the Internet. There was a mixture of reactions from the participants ranging from those who did not want to search the Internet at all to those who visited specific sites daily to participate in cleft chat rooms. The participants appeared to be satisfied with the list of sites they were given by the clinic. Providing anticipatory guidance for all families regarding the Internet may be beneficial. The only study in the clefting literature which has also examined this issue found that 17% of parents felt the Internet was a good way to obtain information (Rey-Bellet & Hohlfeld, 2004). This study also reported handing out a list of useful websites with accurate information to their families. Perhaps this is a practice which could be established in other cleft clinics. It would be interesting to perform follow-up analysis which examined Internet use in more detail.

A potential limitation to this study was the absence of a control population, such as a group of parents who have a child with another type of birth defect. This would have added an additional layer of comparison to the analysis. For example, what are the differences in experiences between parents of a child with an oral cleft in comparison to the parents of a child with an internal abnormality, such as a congenital heart defect? Another possible limitation is the fact that this study included only parents of a child with an isolated cleft. The experience of parents whose children have a facial cleft as part of a genetic syndrome or chromosome abnormality was not studied. Their views may be different than that of the participants in this study and their perspectives may provide useful information for health care professionals. Finally, methods of triangulation (combining multiple observers, theories, methods or data sources) and member

checking (review of the data by the study participants) were not utilized in the current study. It is possible that the use of these approaches would have uncovered additional information.

## 6. CONCLUSION

This investigation revealed a number of themes related to the parental experience of receiving a diagnosis of their child's cleft prenatally or at the time of birth. From the qualitative analysis of interview transcripts, there appeared to be more similarities between the two groups of participants than differences. Parents in both groups spent a great deal of time discussing the experience of initially receiving the diagnosis. They seemed to be affected more by *how* they were informed than the timing of the diagnosis.

In general, parents who received their child's diagnosis prenatally were satisfied with knowing about the cleft before the birth of their child. On the other hand, a number of participants in the prenatal group explained that the diagnosis negatively affected their pregnancy. These findings suggest that although prenatal diagnosis of a cleft may have a negative impact on the pregnancy, parents may still want to know in advance.

An interesting finding from this investigation was that some parents in the postnatal group seemed to have preferred to receive the diagnosis at the time of birth rather than prenatally. Although this may be the preference of some parents, it is likely that in the future a greater proportion of clefts will be identified prenatally by ultrasound. In the United States, if an abnormality is identified on ultrasound, the finding is explained to the patient. Therefore, there may be parents who receive a prenatal diagnosis that would have preferred to find out about their child's cleft at the time of their child's birth. This study may afford health care professionals an

understanding of the parental reaction to receiving such news prenatally. In the future, greater awareness of the parental experience of a prenatal diagnosis may assist health care professionals in caring for patients who might have rather received the diagnosis at birth. Additionally, parents should be made aware of the possibility of identifying an abnormality on ultrasound and perhaps this issue should be part of an informed consent process prior to undergoing an ultrasound.

This study may have implications for genetic counselors as well as health care professionals involved with craniofacial patient care. This investigation may enable genetic counselors to understand the experience of receiving a prenatal diagnosis of a cleft and therefore better meet patients' needs. "Delineating clients' considerations about the new diagnostic possibilities is important for genetic counselors, who are called on to help counselees make decisions in accordance with their own values" (Fraser 1974). Additionally, an understanding of this experience may allow genetic counselors and other professionals involved in craniofacial care to provide anticipatory guidance to their patients.

In conclusion, high-resolution ultrasound has become the standard of care for many pregnant women in this country. This study contributes to the understanding of the experience of receiving a prenatal or postnatal diagnosis of an oral cleft. Understanding this as well as the consequences and impact of prenatal diagnosis is an important contribution to the field of public health.

**APPENDIX A**

**INSTITUTIONAL REVIEW BOARD LETTER OF APPROVAL**



## University of Pittsburgh

### *Institutional Review Board*

3500 Fifth Avenue  
Ground Level  
Pittsburgh, PA 15213  
(412) 383-1480  
(412) 383-1146 (fax)

#### MEMORANDUM:

TO: Mary L. Marazita, Ph.D.

FROM: Christopher Ryan, Ph.D., Vice Chair *CR/SV*

DATE: August 6, 2004

SUBJECT: IRB# 0407052: Assessing the Impact of Prenatal Diagnosis of Clefting on the Maternal/Paternal Birth Experience

The above-referenced proposal has received expedited review and approval from the Institutional Review Board under 45 CFR 46.110 (6,7).

if applicable, please include the following information in the upper right-hand corner of all pages of the consent form:

Approval Date: August 4, 2004  
Renewal Date: August 3, 2005  
University of Pittsburgh  
Institutional Review Board  
IRB# 0407052

Adverse events which occur during the course of the research study must be reported to the IRB Office. Please call the IRB Adverse Event Coordinator at 412-383-1519 for the current policy and forms.

The protocol and consent forms, along with a brief progress report must be resubmitted at least **one month** prior to the expiration date noted above for annual renewal as required by FWA00006790 (University of Pittsburgh), FWA00006735 (University of Pittsburgh Medical Center) and FWA00000600 (Children's Hospital of Pittsburgh).

**Please be advised that your research study may be audited periodically by the University of Pittsburgh Research Conduct and Compliance Office.**

CR:ky

## **APPENDIX B**

### **PARTICIPATION REQUEST SCRIPT**

I am asking for your assistance with a research study that is being conducted by the Cleft-Craniofacial Center and the University of Pittsburgh. The purpose of this research is to study the impact of prenatal diagnosis of cleft lip with or without cleft palate (CL+P) on the maternal and paternal birth experience. We are asking parents who learned of their child's cleft at the time of delivery and also those parents who were informed through prenatal diagnosis by ultrasound to participate. Specifically, we hope to develop a better understanding of how prenatal diagnosis of CL+P affects the birth experience for parents compared to the birth experience for parents whose children were diagnosed with CL+P at birth. We hope to determine what is most useful and important to parents when they meet with the doctors and staff at the Cleft-Craniofacial Center.

In order to conduct the study an interview will take place with each parent at the CCC at Children's Hospital in Pittsburgh. If you are not able to travel to the Cleft-Craniofacial Center the interview may take place over the phone. The interview will be tape-recorded and will take between 30 minutes and one hour to complete. The interview will focus on your thoughts and experiences as a parent in the time period surrounding the birth of your child. Some questions will be multiple choice-type format and others will be open-ended where you will be able to

describe your experience in as much or little detail as you wish. You will have the option to refuse to answer any question. Individual interview responses will be kept strictly confidential.

We would appreciate your help with this important project. Your decision to participate or not is completely voluntary.

## **APPENDIX C**

### **SCRIPT FOR FOLLOW-UP CALL**

Hi, may I speak with one of the parents of \_\_\_\_\_? My name is Rachel Malinowski, I am calling from the Craniofacial Cleft Palate Center at Children's Hospital in Pittsburgh in regards to the prenatal diagnosis study you were recently contacted about. Is this a good time for you to talk? I believe Dr. Carla Weidman, the psychologist at the clinic, regarding a research study we are conducting, contacted you recently? Are you still interested in participating?

I'm calling to introduce myself; I will be the person conducting all of the interviews. I wanted to let you know that I will be sending some packets in the mail with an informed consent document. The informed consent says that you agree to participate and it gives an overview of the study. It tells you that we will be conducting an interview either over the phone or in person that will be taped and we will also be sending a multiple-choice questionnaire in the mail for participants to fill out. We'll be sending two copies of the consent, one to sign and send back, and one to keep for your records. You'll need to initial each page of the document, sign the second to last page and then send it back in the self-addressed stamped envelope. We will be sending a separate envelope for each parent. I'll give you a call next week to review the informed consent with you and to see if you have any questions regarding the consent. We can set up the interview at that time.

Can I get your first name and the name of \_\_\_\_\_'s father/mother?

Do you have the same last name?

Do you have the same address and can I confirm the address with you?

If you have any questions you can feel free to call our toll free number and leave a message for

Rachel: 1-866-501-6999

## APPENDIX D

### LETTER PACKAGED WITH INFORMED CONSENT



University of Pittsburgh

*School of Dental Medicine  
Division of Oral Biology*

Suite 500, Cellomics Building  
100 Technology Drive  
Pittsburgh, PA 15219  
412-648-8381  
Fax: 412-648-8779

Date  
Participant name  
Address

Dear Ms./Mr. \_\_\_\_\_:

As you know, you are being invited to participate in the research study entitled “Assessing the Impact of Prenatal Diagnosis of Clefting on the Maternal/Paternal Birth Experience” because you have a child who was born with cleft lip with or without cleft palate. This study is a collaboration between the Cleft-Craniofacial Center at Children’s Hospital of Pittsburgh and the School of Dental Medicine at the University of Pittsburgh. Enclosed you will find two copies of the informed consent document for this research study. By initialing each page of the informed consent form and providing your signature on page 6, you agree to act as a participant in this research study. If you decide to take part in this study you will participate in an interview which will be tape-recorded and will last approximately 30 minutes. Additionally, you will be asked to

complete a multiple-choice questionnaire containing approximately 25 questions. I have enclosed a pre-paid envelope for you to return the consent to me, after we have reviewed it and if you agree to participate. Please keep the other copy for your records.

I will call you within the next two weeks to review the informed consent with you and answer any questions you have. Please call our toll-free number and leave a message for me if you have questions or if I should contact you at an alternative phone number: 1-866-501-6999. I look forward to speaking with you soon. Thank you again for considering this important research project.

Sincerely,

Rachel Malinowski, B.S.

Research Co-Investigator

Cleft-Craniofacial Center

## APPENDIX E

### INTERVIEW GUIDE FOR PRENATAL GROUP

Date: \_\_\_/\_\_\_/\_\_\_

Study participant's name: \_\_\_\_\_

Interview method: \_\_\_ Phone \_\_\_ In-person

Parent: \_\_\_ Mom \_\_\_ Dad

Parental age: \_\_\_\_\_

Education level of parent: High school degree \_\_\_ Associate's degree \_\_\_ Bachelor's  
degree \_\_\_ Master's degree \_\_\_ Doctoral degree \_\_\_  
Other \_\_\_\_\_

Child's diagnosis: \_\_\_ unilateral cleft lip  
\_\_\_ bilateral cleft lip  
\_\_\_ cleft lip and palate, unilateral  
\_\_\_ cleft lip and palate, bilateral  
\_\_\_ other \_\_\_\_\_

Was [child] your first-born child? \_\_\_\_\_

How old is [child]? \_\_\_\_\_

1. Tell me the story of how you first found out about your child's cleft.
2. Was she/he born with other health problems in addition to the cleft?

3. Which week in pregnancy were you informed of [child]'s cleft?
4. Who informed you of [child]'s cleft and what were you thinking when you heard of the diagnosis?
5. What were you thinking about the way the doctor told you about the diagnosis?
6. What about how the doctor told you made you feel comfortable?
7. What do you think would be the ideal way for a doctor to tell his patient about a cleft on an ultrasound?
8. Going in to the ultrasound had you thought about the possibility that something might be seen by the doctor?
9. How soon after the diagnosis or birth did someone on the cleft team counsel you?
10. How did the prenatal diagnosis affect the rest of the pregnancy?
11. Did you meet with a genetic counselor to discuss your child's cleft? What did you find valuable from this encounter?
12. How prepared were you in general for your baby's arrival at the time of birth?
13. Describe your (thoughts) feelings during the first week of [child]'s life.
14. Describe your (thoughts) feelings after [child] had his/her first surgery performed.
15. People have many ideas about what causes a cleft lip. What are your thoughts about what may have caused your child's cleft lip?
16. When parents are given a diagnosis that their child has a cleft lip, many thoughts about the pregnancy and raising a child who will need multiple surgeries crosses their mind. Some parents consider ending the pregnancy and some parents don't. Could you tell me your thoughts about this issue?
17. If you were to have another child how would you feel about utilizing prenatal diagnosis?

18. What do you feel are the advantages and disadvantages of prenatal diagnosis?
19. Do you believe that prenatal diagnosis made the adjustment to [child]'s diagnosis of cleft lip easier than if the diagnosis were given at the time of birth? How?
20. How did having a prenatal diagnosis help you explain to your family that your baby would be born with a cleft lip?
21. What were the roles or responsibilities that you and your husband (wife) took on surrounding the time of your child's diagnosis? How were decisions made for your family at that time? (involvement of husband/wife)
22. Did you tell friends in addition to family members about the cleft before your child was born?
23. If you accessed the Internet to obtain information regarding cleft lip and/or palate, was the information you found helpful? How do you feel this information helped you understand more about cleft lip and/or palate?
24. If a genetic test were to become available in the future would you utilize the test to determine your risk for having a child with a cleft?
25. What would be your recommendations to the cleft team on the process of telling parents about their child's cleft? (who should provide that information, how many visits should the parents have with the cleft team and who should be involved with each meeting, was the written information helpful, was there too much or too little information given)
26. Should parents meet with another parent who has had a child born with a cleft?

## APPENDIX F

### INTERVIEW GUIDE FOR POSTNATAL GROUP

Date: \_\_\_/\_\_\_/\_\_\_

Study participant's name: \_\_\_\_\_

Interview method: \_\_\_ Phone \_\_\_ In-person

Parent: \_\_\_ Mom \_\_\_ Dad

Parental age: \_\_\_\_\_

Education level of parent: High school degree \_\_\_ Associate's degree \_\_\_ Bachelor's  
degree \_\_\_ Master's degree \_\_\_ Doctoral degree \_\_\_  
Other \_\_\_\_\_

Child's diagnosis: \_\_\_ unilateral cleft lip  
\_\_\_ bilateral cleft lip  
\_\_\_ cleft lip and palate, unilateral  
\_\_\_ cleft lip and palate, bilateral  
\_\_\_ other \_\_\_\_\_

Was [child] your first-born child? \_\_\_\_\_

How old is [child]? \_\_\_\_\_

1. Tell me the story of how you first found out about your child's cleft.
2. When were you informed of your child's cleft?

3. Was your child born with other health problems in addition to the cleft?
4. Who informed you of your child's cleft and could you describe your feelings (and thoughts) at that time?
5. How did you feel about the way the doctor told you about the diagnosis?
6. What about how the doctor told you made you feel comfortable?
7. What do you think would have been the ideal way for your doctor to tell you about the diagnosis?
8. How prepared were you in general for your baby's arrival at the time of birth?
9. How soon after the birth did someone on the cleft team counsel you?
10. Did you meet with a genetic counselor to discuss your child's cleft? What was valuable from this encounter?
11. Describe your feelings (thoughts) during the first week of your child's life.
12. Tell me about your child's first surgery.
13. If you were to have another child would you be interested in prenatal diagnosis? Why?
14. What do you feel are the advantages and disadvantages of prenatal diagnosis?
15. People have many ideas about what causes a cleft lip. What are your thoughts about what may caused your child's cleft lip?
16. If you accessed the Internet to obtain information regarding cleft lip and/or palate, was the information you found helpful? How do you feel this information helped you understand more about cleft lip and/or palate?
17. If a genetic test were to become available in the future would you utilize the test to determine your risk for having a child with a cleft?

18. Consider this hypothetical question: When parents are given a prenatal diagnosis by ultrasound that their child has a cleft lip, many thoughts about the pregnancy and raising a child who will need multiple surgeries crosses their mind. One thing that some parents consider is terminating the pregnancy. Some parents consider ending the pregnancy and some parents don't. Can you tell me your thoughts on this issue.
19. What would be your recommendations to the cleft team on the process of telling parents about their child's cleft? (who should provide that information, how many visits should the parents have with the cleft team and who should be involved with each meeting, was the written information provided helpful, was there too much or too little information)
20. Should parents meet with another parent who has had a child born with a cleft?

## APPENDIX G

### DEMOGRAPHICS OF PARTICIPANTS

<b>Participant ID</b>	<b>Timing of diagnosis</b>	<b>Sex of child</b>	<b>Child age (in months)</b>	<b>Diagnosis</b>	<b>Interview method</b>	<b>Mom age (years)</b>	<b>Dad age (years)</b>
PNS1	prenatal	M	8	CL+P unilateral	Phone	39	NA
PNS2	prenatal	M	8	CL+P unilateral	Phone	24	28
PNS3	at birth	F	14.5	CL unilateral	Phone	27	26
PNS4	prenatal	M	3	CL+P bilateral	Phone	28	39
PNS6	at birth	M	5.5	CL+P unilateral	Phone	29	31
PNS7	prenatal	F	4	CL+P unilateral	In person	27	30
PNS8	prenatal	M	3	CL+P unilateral	Phone	35	NA
PNS9	prenatal	M	5	CL unilateral	Phone	40	33
PNS11	prenatal	M	9.5	CL unilateral	Phone	37	NA
PNS12	prenatal	M	14	CL+P unilateral	Phone	34	NA
PNS13	at birth	F	18	CL unilateral	Phone	33	NA
PNS14	at birth	M	12	CL unilateral	Phone	24	NA
PNS16	at birth	M	15	CL+P unilateral	Phone	35	36

Abbreviations: M (male), F (female), CL+P (cleft lip and palate), NA (did not participate in study)

## BIBLIOGRAPHY

- Abramowicz, S., Cooper, M.E., Bardi, K., Weyant, R.J., & Marazita, M.L. (2003). Demographic and prenatal factors of patients with cleft lip and cleft palate. *JADA*, 134, 1371-1376.
- Alkazaleh, F., Thomas, M., Grebenyuk, J., Glaude, L., Savage, D., Johannesen, J., et al. (2004). What women want: women's preferences of caregiver behavior when prenatal sonography findings are abnormal. *Ultrasound Obstet Gynecol*, 23, 56-62.
- Beeson, D. (1997). Nuance, complexity, and content: qualitative methods in genetic counseling research. *Journal of Genetic Counseling*, 6, 21-43.
- Bender, P.L. (2000). Genetics of Cleft Lip and Palate. *Journal of Pediatric Nursing*, 15, 242-248.
- Berk, N., Marazita, M.L., & Cooper, M.E. (1999). Medical genetics on the cleft palate-craniofacial team: understanding parental preference. *Cleft Palate-Craniofacial Journal*, 36, 30-35.
- Blumenfeld, Z., Blumenfeld, I., & Bronshtein, M. (1999). The early prenatal diagnosis of cleft lip and the decision-making process. *Cleft Palate-Craniofacial Journal*, 36, 105-109.
- Boyatzis, R.E. (1998). *Transforming qualitative information: Thematic analysis and code development*. Thousand Oaks: Sage Publications.
- Boyd, P.A., Chamberlain, P., Hicks, N.R. (1998). Six-year experience of prenatal diagnosis in an unselected population in Oxford UK. *Lancet*, 352, 1577-1581.
- Bryman, A. (2004). *Social Research Methods*. Oxford: Oxford University Press.
- Byrne, M.M. (2001). Evaluating the findings of qualitative research. *Association of periOperative Registered Nurses*, 73, 703-704.
- Byrnes, A.L., Berk, N.W., Cooper, M.E., & Marazita, M.L. (2003). Parental evaluation of informing interviews for cleft lip and/or palate. *Pediatrics*, 112, 308-313.
- Charmaz, K. (2000). Grounded theory: Objectivist and constructivist methods. In N. K. Denzin & Y. S. Lincoln (Eds.), *Handbook of Qualitative Research* (pp. 509-529). Thousand Oaks: Sage Publications.
- Chitty, L.S., Hunt, G.H., Moore, J., & Lobb, M.O. (1991). Effectiveness of routine ultrasonography in detecting fetal structural abnormalities in a low risk population. *BMJ*, 303, 165.

- Climenti, M., Tenconi, R., Bianchi, F., & Stoll, C. (2000) Evaluation of prenatal diagnosis of cleft lip with and without cleft palate by ultrasound; experience from 20 European registries. *Diagnosis Prenatal*, 20, 870-875.
- D'Alessandro, D.M. & Dosa, N.P. (2001). Empowering children and families with information technology. *Arch Pediatr Adolesc Med*, 155, 1131-1136.
- Davalbhakta, A. & Hall, P.N. (2000). The impact of antenatal diagnosis on the effectiveness and timing of counseling for cleft lip and palate. *Br J Plast Surg*, 53, 298-301.
- DeSantis, L. & Ugarriza, D.L. (2000). The concept of theme as used in qualitative nursing research. *Western Journal of Nursing Research*, 22, 351-372.
- Eysenbach, G. (2003). The impact of the Internet on cancer outcomes. *CA Cancer J Clin*, 53, 356-371.
- Forth, S. (2003). Antenatal diagnosis of cleft lip and palate: Supporting the parents. *Journal of Neonatal Nursing*, 9, 57-61.
- Fossey, E., Harvey, C., McDermott, F., & Davidson, L. (2002). Understanding and evaluating qualitative research. *Australian and New Zealand Journal of Psychiatry*, 36, 717-732.
- Fraser, F.C. (1974). Genetic counseling. *Am J Hum Genet*, 26, 636-661.
- Grandjean, H., Larroque, D., Levi, S., & Eurofetus Study Group (1999). The performance of routine ultrasonographic screening of pregnancies in the Eurofetus study. *Am J Obstet Gynecol*, 181, 446-454.
- Hafner, E., Sterniste, W., Scholler, J., Schuchter, K., & Philipp, K. (1997). Prenatal diagnosis of facial malformations. *Prenatal Diagnosis*, 17, 51-58.
- Johnson, N., & Sandy, J.R. (2003). Prenatal diagnosis of cleft lip and palate. *Cleft Palate-Craniofacial Journal*, 40, 186-189.
- Jones, M. (1999). Prenatal diagnosis of cleft lip and palate: experiences in southern California. *Cleft Palate-Craniofacial Journal*, 36, 107-109.
- Jones, M. (2002). Prenatal diagnosis of cleft lip and palate: detection rates, accuracy of ultrasonography, associated anomalies, and strategies for counseling. *Cleft Palate-Craniofacial Journal*, 39, 169-173.
- Leininger, M.M. (1985). *Qualitative Research Methods in Nursing*. Orlando,: Grune and Stratton, Inc.
- Leithner, K., Maar, A., Fischer-Kern, M., Hilger, E., Loffler-Stastka, H., & Ponocny-Seliger, E. (2004). Affective state of women following a prenatal diagnosis: predictors of a negative psychological outcome. *Ultrasound Obstet Gynecol*, 23, 240-246.

- Levi, S., Hyjazi, Y., Schaaps, J.P., Defoort, P., Coulon, R., & Buekens, P. (1991). Sensitivity and specificity of routine antenatal screening for congenital anomalies by ultrasound: the Belgian multicentric study. *Ultrasound Obstet Gynecol*, 1, 102-110.
- Lippman, A. (1999). Embodied knowledge and making sense of prenatal diagnosis. *Journal of Genetic Counseling*, 8, 255-274.
- Lopez, K.A. & Willis, D.G. (2004). Descriptive versus interpretive phenomenology: their contributions to nursing knowledge. *Qualitative Health Research*, 14, 726-735.
- Maes, S., Demey, A., & Appelboom-Fondu., J. (1998). Impact of ultrasound screening for facial cleft on mother-child relationships. *Annals of the New York Academy of Sciences*, 847, 249-251.
- Mason, J. (2002). *Qualitative researching*. Thousand Oaks: Sage Publications.
- Matthews, M.S., Cohen, M., Viglione, M., and Brown, A. (1998). Prenatal counseling for cleft lip and palate. *Plastic and Reconstructive Surgery*, 101, 1-5.
- Merriam-Webster Online. Retrieved April 5, 2005, from <http://www.m-w.com/>.
- Miles, M.B. & Huberman, A.M. (1994). *Qualitative data analysis: an expanded sourcebook*. Thousand Oaks, CA: Sage Publications.
- Miller, W.L. & Crabtree, B.F. Clinical research. In N.K. Denzin, & Y.S. Lincoln (Eds.), (1998). *Strategies of Qualitative Research*. Thousand Oaks, CA: Sage Publications (pp. 292-314).
- Nicholson, J. (2002) Parents' perspective on cleft lip and palate. In D.F. Wyszynski (Ed.), *Cleft lip and palate: from origin to treatment* (pp.474-480). Oxford: Oxford University Press.
- Patton, M.Q. (2002). *Qualitative research and evaluation methods*. Thousand Oaks: Sage Publications.
- Rey-Bellet, C. & Hohlfeld, J. (2004). Prenatal diagnosis of facial clefts: evaluation of a specialised counseling. *Swiss Med Wkly*, 134, 640-644.
- Ritchie, J., & Lewis, J. (2003). *Qualitative research practice: a guide for social science students and researchers*. London: Sage Publications.
- Rubin, H.J., & Rubin, I.S. (1995). *Qualitative Interviewing: The Art of Hearing Data*. Thousand Oaks: Sage Publications.
- Ryan, G.W., & Bernard, H. R. (2000). Data management and analysis methods. In N. K. Denzin & Y. S. Lincoln (Eds.), *Handbook of Qualitative Research* (pp. 645-668). Thousand Oaks: Sage Publications.
- Saal, H.M. (2002). Prenatal diagnosis: when the clinician disagrees with the patient's decision. *Cleft Palate-Craniofacial Journal*, 39, 174-178.

- Sagi, M., Shiloh, S., & Cohen, T. (1992). Application of the health belief model in a study on parents' intentions to utilize prenatal diagnosis of cleft lip and/or palate. *American Journal of Medical Genetics*, 44, 326-333.
- Sandelowski, M. (2000). Focus on research methods: Whatever happened to qualitative description? *Research in Nursing and Health*, 23, 334-340.
- Schuth, W., Karck, U., Wilhelm, C., & Reisch, S. (1994). Parents' needs after ultrasound diagnosis of a fetal malformation: an empirical deficit analysis. *Ultrasound in Obstetrics and Gynecology*, 4, 124-129.
- Shaikh, D., Mercer, N.S., Sohan, K., & Soothill, P. (2001). Prenatal diagnosis of cleft lip and palate. *British Journal of Plastic Surgery*, 54, 288-289.
- Sohan, K., Freer, M., Mercer, N., Soothill, P., & Kyle, P. (2001). Prenatal detection of facial clefts. *Fetal Diagn Ther*, 16, 196-199.
- Soltis, J.F. (1984). On the nature of educational research. *Educational Researcher*, 13, 5-10.
- Stadter, C.S. (2002). Genetic counseling and interpretation of risk figures. In D.F. Wyszynski (Ed.), *Cleft lip and palate: from origin to treatment* (pp.408-411). Oxford: Oxford University Press.
- Stoll, C., Alembik, Y., Dott, B., & Roth, M.P. (2000a). Associated malformations in cases with oral clefts. *Cleft Palate Craniofac J*, 37, 41-47.
- Stoll, C., Dott, B., Alembik, Y., & Roth, M. (2000b). Evaluation of prenatal diagnosis of cleft lip/palate by fetal ultrasonographic evaluation. *Ann Genet*, 43, 11-14.
- Strauss, R.P., Sharp, M.C., Lorch, S.C., & Kachalia, B. (1995). Physicians and the communication of "bad news": parent experiences of being informed of their child's cleft lip and/or palate. *Pediatrics*, 96(1 Pt 1), 82-9.
- Wyszynski, D.F., Perandones, C., & Bennun, R.D. (2003). Attitudes toward prenatal diagnosis, termination of pregnancy and reproduction by parents of syndrome with nonsyndromic oral clefts in Argentina. *Prenat Diagn*, 23, 722-727.