CLEIDOCRANIAL DYSPLASIA: THE LIVED EXPERIENCE

by

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ABSTRACT

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A phenomenological study was conducted to understand the lived experience of 12 individuals with Cleidocranial Dysplasia (CCD). Names were obtained from the CCD Internet website www.cleidocranialdysostosis.org. A demographic questionnaire was used to purposefully sample participants. A tape recorded open-ended interview was used to elicit qualitative information regarding: (a) participants' greatest concerns about having CCD, (b) what was most difficult about living with CCD, (c) what was most helpful in living with CCD, (d) participants' coping mechanisms, and (e) whether or not CCD was a positive or negative experience.

Results demonstrated commonalities among the 12 participants and included issues surrounding being teased as a child, having problems with teeth, passing CCD on to their children, and learning from their experiences with CCD. This study was a starting point for future qualitative research regarding CCD.

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CHAPTER ONE

Introduction

Cleidocranial Dysplasia (CCD) is a rare autosomal dominant disorder with a range of variability in expression that affects 1 in 200,000 (Feldman, 2002) to 1 in 1,000,000 live births (Jones, 1997). One-third of the cases are due to sporadic mutations, but a rare recessive form has also been noted (Goodman, Tadmor, Zaritsky, & Becker, 1975). Males and females of all ethnic groups are equally affected. Cleidocranial Dysplasia has been studied specifically in Caucasians, Hindus, Sudanese, Chinese, African Americans, Mestizos, and Jews (Siggers, 1975). Individuals with CCD are healthy, intelligent, capable of hard physical labor, and have a normal life span (Short, 1979). Phenotypically the condition is characterized by dysplastic bone formation of the face, skull, pelvis, and thoracic region. Main clinical features include hypoplastic or absent clavicles, open fontanelles and sutures, supernumerary teeth, short stature, and other skeletal anomalies. Since CCD was first described over 100 years ago, more than 500 cases have been reported in the literature (Gulati & Kabra, 2001; Jones, 1997).

Most literature about CCD focuses on the physical and genetic aspects of the disease (Cooper, Flaitz, Johnston, Lee, & Hecht, 2001; Feldman, 2002; Golan, Baumert, Held, Feuerbach, & Mubig, 2002; Gulati & Kabra, 2001; Kreiborg, Jensen, Larsen, Schleidt, & Darvann, 1999) as well as information on how to treat accompanying dental problems (Dawjee & Nikhumeleni, 2002). Within the last decade, research related to gene identification has become the focus of study (Dickman, 1997; McBride, Napierala, Chen, Zhou, & Lee, 2001; Mundlos, 1999; Rodan & Harada, 1997; Zhou, et al., 1999). However, no study focusing on the lived experience of individuals with CCD was found

in the literature. Therefore, the purpose of this study was to describe the lived experience of individuals with CCD.

Review of Literature

To understand the lived experience of individuals with CCD it is imperative to understand the physiologic conditions of the disorder. Cleidocranial Dysplasia can be traced back over 30,000 years to the Neanderthal Man (Siggers, 1975). Researchers believe the author of the Homeric epics wrote of CCD in 1250-750 BC when portraying the character Thersites (Altschuler, 2001). Beasley and Carter (Altschuler, 2001), two geneticists, independent of one another, both diagnosed Thersites with CCD because of his rounded shoulders stooping together over his caved-in chest and his "skull warped to a point" (lines 217-219 of the Iliad). In more modern times, CCD was first described by Meckel in 1760, but it was more accurately defined by Scheuthauer in 1871 (Siggers, 1975). In 1898, Marie and Sainton, two French geneticists, realized the hereditary nature of the disease and named it dysostose cleidocranienne hereditaire, or Cleidocranial Dysostosis (Short, 1979).

Cleidocranial Dysplasia was most commonly known as Cleidocranial Dysostosis until 1978 when it was changed by Rimonin to Cleidocranial Dysplasia (Atasu, Asim, & Ozbayrak, 1996). The term cleido refers to clavicle; cranial refers to head; and dysplasia means "ill formed" (Feldman, 2002).

Etiology

Cleidocranial Dysplasia is caused by a mutation in the core-binding alpha one (CBFA1) gene located on chromosome 6p21 (Golan et al., 2002; Rodan & Harada, 1997) that causes faulty ossification of endochondranous and intramembranous bones

(Feldman, 2002). Individuals with CCD have one intact copy of CBFA1 and one mutated copy (Dickman, 1997), that may be a deletion, insertion, or missense mutation. The type determines the degree of phenotypic expression and may be classified as classic, mild, or isolated primary dental anomalies (Zhou et al., 1999). Any bone can be affected, but Feldman describes the bones most affected as the classical triad of cranial, clavicular, and pelvic anomalies.

Clinical Presentation

Clinically patients present with a variety of characteristic features. The most notable defects are associated with the head and neck and include: brachycephaly, a large head with frontal bossing; small face due to small maxillary and zygomatic bones; prognathism, a jutting forward of the facial skeleton and jaw; open fontanelles; open cranial sutures; poor development of the foramen magnum; and dysplasia of the paranasal sinuses and mastoids. The eyes are wide set and the nose broad with a depressed, low bridge. The mouth and oral structures have a highly arched palate, delayed tooth eruption, tooth abnormalities, and absent or extra teeth (Feldman, 2002). Spinal abnormalities include defects such as scoliosis, kyphosis, spina bifida, syringomyelia, and spondylolysis or spondylothesis. The thorax is small and the chest is narrow with small, oblique ribs. Shoulders droop and the scapulae may be overgrown or undergrown; clavicles can be partially or fully absent. The pelvis is narrow with a wide symphysis pubis, and the extremities may have a deformity of the hip such as coxa vara or coxa valga. All joints are loose and may easily dislocate, hands have asymmetric fingers, and the feet are flat. Stature is short, but dwarfism is uncommon (Cooper et al., 2001; Feldman, 2002; Mundlos, 1999). Because of these physical characteristics, individuals

with CCD may have multiple oral, facial, or orthopedic surgeries; orthopedic complications; neurological and vascular problems (Qureshi, Lees, & Holdsworth, 1997; Short, 1979); deafness (Hawkins, Shapiro, & Petrillo, 1975; Pou, 1971; Siggers, 1975); respiratory problems, and an increased cesarean rate (Cooper et al., 2001).

The diagnosis is usually made at birth but may be delayed until adolescence (Feldman, 2002), especially if the individual has a mild form, or if the condition does not seriously affect the individual. Occasionally CCD may go undiagnosed. Three-dimensional computer tomography (3D CT) is helpful in diagnosing older children with CCD (Shen, 2000), and prenatal diagnosis with ultrasound is possible if there is a known positive family history (Hassan, Sepulveda, Teixeira, Garrett, & Fisk, 1997; Stewart, Wallerstein, Moran, & Lee, 2000).

Research Question

The research question for this phenomenological study was, “What is it like to live with Cleidocranial Dysplasia?”

CHAPTER TWO

Method

Design

A phenomenological design was used to describe what it was like to live with CCD. The literature review focusing on the lived experience was completed after the research had been collected, so the researcher obtained a pure description of the phenomenon and was not biased (Streubert & Carpenter, 1995).

Participants

Specific questions were used to both purposefully sample and gain demographic information from participants (See Appendix A). Participants were purposefully sampled from the CCD Internet website www.cleidocranialdysostosis.org to include: six males and six females of a variety of ages, some with children and some without children, and those with a new mutation and those with parents with CCD. Inclusion criteria consisted of adult men and women over age 18 who had been formally diagnosed with classic CCD, spoke and read English, lived in the United States or Canada, and were able to communicate via email. Individuals were excluded if they had major genetic disorders in addition to CCD. Individuals were not excluded because of marital status, country of origin, education level, or health problems. Participants were interviewed over the telephone at a time convenient to both the participant and the researcher.

Instruments

All those who wished to participate were initially asked screening and demographic questions by telephone (Appendix A). The screening questions included information about any other birth defects and how individuals classified their CCD

(classic, mild, or isolated and primary dental anomalies) and allowed the researcher to purposefully sample according to Denzin and Lincoln (1994).

Procedure

Data collection began after receiving approval from the Brigham Young University College of Nursing Research Committee and the Brigham Young University Institutional Review Board. The researcher contacted each individual listed on the CCD Foundation website (www.cleidocranialdysostosis.org) via email (Appendix B). Emails were sent to individuals rather than posting a general announcement on the CCD site because the webpage was designed to "connect [people] to other members of the Cleidocranial Dysplasia Foundation who [had] given their permission to release their email addresses. . .[and] to connect as many people as possible via email" (www.cleidocranialdysostosis.org). Those who were interested in participating in the study notified the researcher via email or by phone to set up times they would be available for an interview.

The informed consent (Appendix C) was emailed to all individuals wishing to participate and verbal consent was obtained at the beginning of the tape-recorded telephone interview. Screening and demographic questions were then asked of those who wished to participate (Appendix A). From this information 12 individuals were selected. The researcher then conducted personal and private telephone interviews which consisted of open-ended and appropriate follow-up questions (Appendix D) and lasted from 30 to 60 minutes each. Interviews were recorded using a telephone with voice recording capabilities and then transcribed verbatim. All identifying information was removed at the time of transcription and all transcriptions were stored in a locked cabinet. The

transcripts were compared to the original tapes to verify accuracy. The audio tapes were destroyed at the conclusion of the project.

Data Analysis

Demographic/screening data were analyzed using descriptive statistics. Audiotapes were transcribed verbatim and data were submitted to content analysis using descriptive phenomenology according to Streubert and Carpenter (1995). Trustworthiness of the data was established by consistent use of the method, use of bracketing, and by conducting member checks.

The transcribed text was read line by line and content was coded according to concepts. All coding was done by the researcher. Concepts were then organized into themes and are presented in the results section as tables and text. The numbers in the "total" column refer to the number of participants who mentioned the topic. The entire total column may not add up to 12 because some participants commented on more than one category.

CHAPTER THREE

Results and Discussion

Five themes were identified from content analysis. Each theme is represented by a table and text. The themes identified were: (a) what was most difficult about living with CCD, (b) participants' greatest concern with CCD, (c) what was most helpful in living with CCD, (d) participants' coping mechanisms, and (e) whether CCD had been a positive or negative experience.

Description of the Sample

There were six male and six female participants. The age range of the males was 22 to 54 years with the mean age of 39.3 years. The age range of the females was 21 to 53 years with the mean age of 37 years. All 12 participants were Caucasian. Seven participants were married, four were single, and one was divorced. Nine participants were from the United States and three were from Canada.

Seven participants reported an above average intelligence and five reported an average intelligence. One participant had a post-graduate degree, one had a graduate degree, two had Bachelor's degrees, seven had some college, and one was a high school graduate. Three participants worked as executives or managers, two owned their own businesses, two were college students, one worked in retail, one was a researcher, one was retired, one worked in the medical field, and one was an entertainer.

The height of the males ranged from 63-73 inches with the mean of 67.16 inches. The height of the females ranged from 58-66 inches with the mean of 60.83 inches. These demographic data show consistency with the literature because typically, those with CCD have a short to moderate stature (Jones, 1997), especially females (Jensen, 1990).

All participants were diagnosed with CCD either before birth or by the time they were 12. Five participants were new mutations and therefore had no previous family history of CCD. Five participants had mothers with CCD and two had fathers with CCD. All 12 participants had siblings; three of these siblings had CCD. Seven participants had a total of 12 children, 11 of whom had CCD (See Table 1).

Greatest Concerns

Participants' greatest concerns were passing CCD on to their children, their future health, and their appearance (See Table 2). In fact, all but two participants talked about their concern of passing on CCD to their children. Less than half were concerned about their future health or appearance.

Passing CCD on to Children

Four males and all females were concerned about passing CCD on to their children, and for most deciding to have children was a gamble. Those who understood genetics knew there would be a fifty-fifty chance of passing it on to their children. However, one participant was unaware of the genetic nature of CCD until after the birth of their first child.

Two females, who did not have children, also expressed concerns about pregnancy. They were especially worried about their physical ability to carry children and the likelihood of cesarean deliveries. In contrast, two women who had given birth reported "easy pregnancies and deliveries." One was able to deliver her first child vaginally. The other said her pregnancy was "a piece of cake" and mentioned having a cesarean section was the "only way to have a baby."

With half of the participants, concern of passing CCD on to their children did not

Table 1

Demographic Data

Age at Diagnosis:

Before birth	2
At birth	3
2 years	2
5 years	1
6 years	1
10 years	2
12 years	1

Surgeries:

Oral surgery	11
Cesarean	4
Tonsillectomy	4
Tubes in ears	3
Cosmetic	2
Knee surgery	1

Health Problems:

Dental problems	12
Breathing problems/asthma	4
Knee pain/instability	3
Back pain	3
Hearing loss	3
Multiple bone fractures	2

Table 2

Greatest Concerns

Category	Total	Males	Females
Passing CCD on to children	10	4	6
Future health	5	2	3
Appearance	4	3	1

override the desire to have children. Even the participant who was not aware of the genetic nature of CCD until after her first child was born said that information did not influence her decision to have her second child. One participant felt confident he could handle the physical problems of CCD as long as his babies had strong brains.

As a teenager and while in college, one participant never wanted to have children because she did not want to put a child through what she had gone through. As she got older, however, she decided CCD was not life threatening and that "it's always going to be something and if it's going to be CCD then I consider myself pretty lucky. I can handle that." She said children are worth it.

For participants, a major factor in deciding to have children was deciding how many children to have. Several participants expressed the need to limit the number of children they would have because they knew children with CCD would require medical and dental attention. One participant with a child who did not have CCD did not want to chance the odds of having a child with CCD and had therefore decided not to have more children.

Those who did not have children expressed concerns about passing CCD on and putting a child through what they experienced. Some planned to seek genetic counseling before making the decision. Broder and Trier (1985) found that genetic counseling in families with craniofacial anomalies was important to family planning in the 18 to 30 year old age group. Parents who were given accurate genetic information to help with decision making, lessened their chance of having another child with a craniofacial malformation.

The majority of participants with children felt guilty or responsible for passing on CCD to their children. They expressed concern about their children having to go through the surgeries and extensive dental and medical treatments. One woman worried about her son being short and her daughter having problems with her looks. She felt responsible when her son broke a bone. Others felt guilty about their children having oral surgeries.

Although most everyone was concerned about passing CCD on to their children, technological advances meant they could know before birth if their child had CCD, since prenatal diagnosis was available to some participants. Opinions regarding prenatal diagnosis of physical disabilities were largely positive with physical disabilities in general (Chen & Schiffman, 2000). One participant said that she was the first to have an alpha fetal protein test to check for CCD. She said, "we would have aborted if we would have found major birth defects" but did not consider CCD to be a major problem. The concern was that her babies had ten fingers and ten toes rather than whether or not they had CCD. Another participant said she would never consider abortion although her geneticist said that CCD was reason enough for an abortion.

Those who had children agreed things are less difficult for their children today than it was for them. Technology is more advanced and more medical resources are available so surgeries and procedures are easier. Inpatient surgeries are now done as an outpatient and many inpatient surgeries require shorter stays. For example, jaw surgeries no longer require wiring and sedation is now used for simple teeth extraction. According to some participants, surgeons today are better educated and more willing to work with families to make their experiences less traumatic. According to their parents, children

with CCD are treated better by their peers and by doctors because today they are more accepting of physical limitations.

Parents' own experiences with CCD helped them know what to expect and assisted them in obtaining proper medical interventions for their children at an early age. Several parents were more proactive in getting health care because of their experience with CCD, and they wanted to prevent "bad" experiences from happening to their children. One mother took it upon herself to gather a community of medical and dental resources around her so any issue her children developed could be properly identified and treated.

Future Health

Future health was the second most common concern expressed and was mentioned by nearly one half of the participants. The participants were concerned because they were unsure of what to expect of their future health. Osteoporosis resulting in an even shorter stature was worrisome, as was worsening of hearing loss. Progressive physical limitations resulting from back problems and arthritis were also mentioned as well as jaw bone instability due to earlier teeth extraction. Although future health was a concern, these participants failed to elaborate to the same extent they did for the concern of passing CCD on to their children.

Appearance

Appearance was the third most common concern expressed by one third of participants. Worries about their own appearance included having a small bone structure or no teeth. Other appearance-related concerns were the possibilities of a small waist, a dent in the forehead, and small shoulders. One subject expressed the concern of having to

wear braces for six years. In this study, one fourth of males and one twelfth of females with CCD, mentioned that appearance was their greatest concern. However, in the general population, one fourth of males and one third of females without CCD report some degree of dissatisfaction with their appearance (Thompson & Kent, 2001).

Most Difficult About Having CCD

Difficulties associated with having CCD were frequent and most commonly experienced during childhood and adolescence. In childhood the difficulties were physical and psychosocial problems. In adolescence the difficulties were mostly physical problems. In adulthood the difficulties were a continuation of the problems from adolescence, though often less severe, along with those difficulties associated with the aging process and some psychological issues. General difficulties, not associated with a particular time of life, were the medical community's lack of knowledge and the financial burdens associated with extensive dental care (See Table 3).

Childhood

Childhood was the time when the participants first remembered being different than their peers. Dental problems were a common difficulty of childhood that set participants apart from their peers. Most dental problems participants faced were due to baby teeth not falling out at the appropriate age. In fact, nearly all participants reported having to have some or all of their baby teeth extracted because the teeth did not fall out on their own.

Physical problems were another identifiable and major difficulty. Some physical limitations were actual limitations while others were placed on the children by parents or health care providers because they were seen as "fragile" or "breakable." Some

Table 3

Most Difficult About Having CCD

Childhood	Total	Males	Females
Physical concerns			
Dental problems	11	5	6
Short stature	6	2	4
Physical limitations	4	2	2
Psychosocial concerns			
Teased by peers	7	5	2
Problems fitting in	4	1	3
Adolescence	**Total**	**Males**	**Females**
Physical concerns			
Dental problems	7	2	5
Short stature	6	2	4
Chronic pain	3	1	2
Psychosocial concerns			
Appearance	6	4	2
Adulthood	**Total**	**Males**	**Females**
Physical concerns			
Physical limitations	6	3	3
Chronic pain	3	1	2
Dental surgeries	3	1	2
Psychosocial concerns	3	2	1
General	**Total**	**Males**	**Females**
Lack of medical knowledge			
Doctor's do not understand CCD	10	5	5
Treated like guinea pig	9	4	5
Financial concerns	4	1	3

participants had multiple fractures during childhood which added to the idea of being breakable. One female participant was limited from doing gymnastics with other children because her parents feared her "arms may come out of their sockets." Other parents were protective of their children because of their soft spots. Two participants had difficulties due to hearing loss. Another female participant had problems with her legs being different lengths. Other physical problems were decreased athletic abilities and the inability to physically keep up with other children. Adaptive physical education was the answer to this situation for one female participant

The majority of participants reported being teased by their peers because they were perceived as being different. Teasing mainly focused mostly on facial features such as a large head, wide set eyes, or a dented forehead. One subject remembered being called "Franky, after the Frankenstein character because of the shape of my head." Some participants were teased about their teeth. Others were teased because they were shorter than their peers or unable to compete athletically.

Some participants had difficulties fitting in to social settings. One participant said, "From early on, I had a sense of separation [from other kids]." Another reported he was, "kind of a freak or a circus side show." He would get attention from his peers by popping his knees in and out of joint and touching his shoulders together. Sarimski (2001), studying children with Apert's syndrome, concluded the stigma of facial deformity may explain impaired peer relations, since peers of children with facial deformities tend to be rejecting and hostile. Social withdrawal or anxiety may result when children are teased, stared at, pitied, or shunned.

Three fourths of the participants had trouble fitting in or were teased as children, while one fourth did not experience these situations. Those who grew up in small towns explained that everyone knew each other and no one was teased about their differences. Another participant was home schooled for a period in order to avoid being teased by peers.

Adolescence

During adolescence difficulties from childhood continued. Even though teasing from peers lessened, having a shorter stature than their peers became more of an issue.

The dental problems of adolescence changed from not having teeth fall out, to not having any teeth because of teeth extraction. Several participants mentioned dental problems as being most difficult because of the extensive orthodontic work, having braces for much longer than their peers, and having to endure multiple oral surgeries.

Chronic pain was also a concern for some during adolescence, especially headaches, back, and hip pain. Again, several participants expressed concern about not being able to keep up physically with their peers.

Both genders were concerned about their appearance. Females associated appearance concerns with their facial features, having a large head, or not having teeth. Males associated appearance concerns with a lack of strength, having smaller shoulders, and being short. One male subject said, “I had really flat shoulders and really skinny arms. I always wanted to be stronger.” Looking different influenced their self concept and comments from others about looking different added to the difficulty. One subject felt “a stigma” from medical professionals because they suggested there was “something wrong and different.”

Adulthood

During adulthood, physical problems continued and became more severe, while dental problems lessened. Most of the specific difficulties in adulthood were physical limitations and chronic pain. Half of the participants experienced chronic joint, neck, shoulder, back, hip, or knee pain. Others had problems lifting because shoulders dislocated easily. Without clavicles, a decrease in upper body strength continued to be a problem into adulthood.

Most participants experienced dental surgeries into their twenties, and those with their own teeth experienced mouth pain or unstable permanent teeth. Those with dentures had gum pain and weakened jaw bones.

Adulthood was also a time when the psychological effects of being different as a child surfaced. One participant explained, “I do have some baggage, but it’s not problematic to the degree where it stops me from going out.” Most adults viewed CCD as most difficult in their youth, but as adults, they thought less about it. One said, “I don’t care that I have it. It doesn’t matter. It’s like a non-issue.” Another agrees, “[today] the disease is not shaping my life. It sure did when I was fourteen but it doesn’t have any effect now.”

General

General difficulties, for all age groups included a lack of medical knowledge among health care providers and financial concerns. All but two participants believed most doctors did not understand CCD and many health care providers had either not seen CCD before or had only read about it in textbooks. One participant said, “I’ve never

talked with anyone who knows." Another said the nearest CCD specialist was 200 miles away. Most participants surveyed agreed that they "end up informing [the doctors]."

This lack of knowledge contributed to the inability of medical professionals to answer questions about CCD. For example, one male participant in his 50's was never informed of how the disorder was passed on, and another could not find a physician able to answer questions about chronic pain. Other physicians were misinformed and told their patients enough to frighten them but not enough to educate them. "You are not going to be able to deliver children vaginally," one participant was told, although her first baby was delivered vaginally. "You're not supposed to get over five [feet tall]" another participant was told, even though many participants surveyed were over five feet tall. One was told she would be crippled and wheelchair bound by adulthood; she is not. She was also informed her boys who do not have CCD could pass it on. This is not true in most cases where CCD is an autosomal dominant trait. These findings support results in a similar study using people with osteogenesis imperfecta (OI) as subjects (Claesson & Brodin, 2001), where families indicated the need for more support and empathy from their doctors. The same study indicated physicians have difficulty becoming knowledgeable about rare disorders.

In addition, most participants at one time or another felt they had been treated as guinea pigs by their physicians or dentists; most remembered feeling like objects rather than as humans. They were often asked to put their shoulders together and then they were shown to other professionals. One participant remembered "interns coming in poking and jabbing because they had never seen [CCD] before." Another remembered interns "filing

in and out of my room. I felt like I was on display." "I was treated like a lab rat," another explained.

Financial concerns were also not specific to a particular age group. Some participants were unable to receive proper dental care during childhood because parents did not have insurance and/or were unaware of how to get help. Others had insurance as children but had a difficult time obtaining dental coverage as adults. Some oral surgeries were covered by dental insurance, but other necessary orthodontic procedures were not covered because they were considered "cosmetic." Also, procedures were often more expensive because there were not specific billing codes for CCD.

Most Helpful in Living with CCD

When asked what was most helpful in living with CCD, four themes were identified: (a) having a support network, (b) having adequate medical resources, (c) gaining knowledge about CCD, and (d) maximizing physical strength (See Table 4).

Support Network

Living with CCD is easier "if you have a support network," explained one participant. A support network means being surrounded by people, including medical professionals, who understand CCD and offer emotional support. The literature also suggests a network of social support is vital to adjusting to being visibly different (Thompson & Kent, 2001). The majority of participants identified a support network as including contact with other persons with CCD, especially via the internet, which helped them "know that other people [were] dealing with the same things [they were] dealing with." Going online also helped some to see the disorder's diversity. The Internet support was also helpful for families raising children with OI who used the Internet to make

Table 4

Most Helpful in Living with CCD

Emotional Support/Support Network	Total	Males	Females
Meeting Others with CCD	8	2	6
Via Email	7	2	5
Face to Face	7	2	5
Family	6	2	4
Friends	3	2	1
Adequate Medical Resources	**Total**	**Males**	**Females**
Knowledgeable experts	7	2	5
Genetic counselors/geneticists	4	1	3
Insurance Coverage	3	0	3
Gaining Knowledge about CCD	**Total**	**Males**	**Females**
Research			
Internet	7	3	4
Medical library	3	2	1
Maximizing Physical Strength	**Total**	**Males**	**Females**
Exercise	4	2	2
Weight training	3	2	1
Physical therapy	2	2	0

international contacts with families in similar situations (Claesson & Brodin, 2001). This contact provided opportunities to discuss a variety of topics from small practical things to life changing issues. These researchers also discovered people with other disorders also have a need to connect with other people who are like them (Claesson & Brodin, 2001). In addition, meeting people face to face was "an amazing experience." People with CCD look alike even if they're not related." Another participant recommended, "if you get a chance to meet people with CCD, really try because it is helpful."

Half of the participants believed family support was also essential to living with CCD. One male participant talked about the importance of his mother's emotional support. "She instilled self-confidence and gave me support. [She always said] you're a good looking young man." Having other members with CCD in their family helped participants deal with their own CCD. "Judging from what I see in my family, [CCD] is not something I can't deal with." Thompson and Kent (2001) also found family acceptance and support was crucial in the psychosocial development of a child who looks different.

One fourth of the participants mentioned the importance of having a supportive group of friends. One participant explained, "I think it's good for people with CCD to be able to open up and tell other people about it. It is important for people around you to understand it."

Adequate Medical Resources

Over one half of the participants reported that adequate medical resources were helpful, including the importance of experts. Two participants had medical teams created by their insurance companies that included a dentist, an orthodontist, a genetic counselor,

an orthopedist, and a primary physician. OI subjects also felt it was extremely important to have a physician who listened and showed empathy, and specialist teams which linked the specialist centers and the primary health care environment (Claesson & Brodin, 2001). OI subjects were satisfied if they had access to specialists, but if they did not receive care from specialists, they were often dissatisfied (Claesson & Brodin, 2001). Many participants also found genetic counseling helpful, as did Chen and Schiffman (2000) when studying individuals with physical disabilities.

Adequate insurance coverage, especially dental coverage was another important medical resource because participants were then able to have the cost of dental work covered. Other participants mentioned the importance of medical insurance because some dental procedures were considered medical.

Gaining Knowledge About CCD

Because the lack of knowledge regarding CCD is common among health care providers, CCD participants found it helpful to educate themselves about the condition, especially through the Internet. One participant said, "with the Internet it's a whole lot easier for someone to find out what the condition is and how to deal with it." The Internet also helped one female participant understand the condition and find out that she is "not the only one" and "not a freak." One third of the participants used medical libraries to research CCD, and one participant studied anatomy and kinesiology to understand how weight training could be beneficial.

Maximizing Physical Strength

More than half of the participants found maximizing physical strength by exercise, weight training, or physical therapy was helpful. Weight training was used by

some participants to increase upper body and overall physical strength. One participant said, "[weight training] has made a world of difference as far as what I'm able to do and with appearances and everything else." This finding is supported by Scott (1988) who believes that physical therapists, who are good musculoskeletal evaluators, are in the best position to assist patients with muscle strengthening and should be part of the team of experts.

Coping Mechanisms

Half of the participants coped by accepting CCD as something they could not change and learned to live with it. Other common coping mechanisms were comparing CCD to worse disorders or forgetting about it. As one participant said, "[things] could always be worse. People have a lot worse. I have all my faculties, so I am very lucky." Less common coping mechanisms were to overcompensate by being successful, withdraw from social settings, participate in drinking and drugs, use humor and laughter, be more outgoing, and rely on spirituality or faith (See Table 5).

As Thompson and Kent (2001) explained, everyone who is visibly different develops coping strategies to deal with the disfigurement. These coping strategies are either "protection of self" or "presentation of self." With CCD, the self-protection strategies included comparing CCD to worse conditions, denial, and attributing negative behaviors of other people to the CCD. Self-presentation strategies involve dealing with others' comments, withdrawing from social settings, and concealing the disfigurement. In accepting CCD and learning to live with it, one participant said, "CCD is what you make of it." Another participant stated "you are who you are and you can't change it, so just live with it. Play the cards you are dealt." Another participant said that it was

Table 5

Coping Mechanisms

Category	Total	Males	Females
Learn to live with CCD/Acceptance	6	3	3
Make comparisons to worse disorders	3	2	1
Forget you have CCD	3	1	2
Humor/laughter	2	2	0
Be active/outgoing	2	2	0
Overcompensating	1	1	0
Withdraw from social settings	1	1	0
Drinking/smoking	1	1	0
Spirituality	1	0	1

important to be honest with oneself about CCD and to know and accept limitations.

Positive or Negative Experience

Finally, having CCD was reported as being a positive experience, a negative experience, or both a positive and negative experience. These three responses were divided evenly among participants (See Table 6).

Positives

Participants said CCD had been a positive part of their life. For example, some said CCD was helpful in their line of work. One joked that CCD helped him "squeeze into small places" in his work. Most felt CCD had improved their character, and felt they were "tougher," "better," or "stronger" for having it. Cleidocranial Dysplasia was seen by others as a "blessing" or a "silver lining." One participant said, "people have disabilities and limitations that force them to grow from hardships and to be better and stronger. It made me work harder and develop other [strengths]." It was "a negative start [with a] positive ending."

Cleidocranial Dysplasia was also a positive part of individual's lives because they learned to strengthen their own character, appreciate what they had, and treat others with respect. One participant learned "to get on with life no matter what." Another learned patience and perseverance. As they learned to live with CCD one realized that "normal is a relative term," whereas another was more appreciative of her teeth and took better care of them. Finally, CCD taught empathy for others with differences. Some participants were more understanding, sensitive, and sympathetic to other people with problems. For example, because of CCD, "I gained a great respect for other people who have more severe genetic problems and other forms of disfigurement or people who have been hurt

Table 6

Positive or Negative Experience

	Total	Males	Females
Positive	3	2	1
Negative	3	2	1
Both Positive and Negative	3	2	1
No answer	3	0	3

or burned."

Negative

The negative experiences were due to the physical pains and limitations associated with the disorder. One participant explained that CCD was physically and emotionally trying as a child and there was "nothing good about that." Another said "when you walk around with your eyeballs set far apart, you're a little bit freakish looking," and that was negative. A third said CCD was negative because if she did not have it, she could be six inches taller.

Positive and Negative

Finally, CCD was both positive and negative. Most said one way or another, "It's not a positive thing to have it, but it's not a real bad thing either." Another said, "I wish I didn't have [CCD]. I wish my son didn't have it. But if it wasn't CCD, it very well could have been something else."

CHAPTER FOUR

Conclusion

Implications for Practice

Because CCD is so rare, few specialists or experts even know CCD exists. Despite this fact, individuals with CCD have a desire to be in contact with medical professionals who are experts. The medical community needs more education in order to provide a network of medical experts. Whether through insurance companies, national organizations, or the Internet, experts can then be connected to individuals with CCD.

From this research, it is also apparent that the medical community as a whole was not perceived as sympathetic towards people with CCD. Whether it is CCD or another disorder, physicians need to be aware of how individuals who have rare disorders want to be treated; they want to be treated like everyone else. People with CCD do not mind talking with others including medical professionals about their life experiences, however experiences described need to be handled in a sensitive manner.

Another issue that arose from this research is the need for insurance companies to be aware of CCD, and perhaps cover some procedures people with CCD need. Billing codes need to be established and insurance companies also need to be aware of the conditions specific to CCD so oral surgeries and treatments can be reimbursed.

Limitations of the Study

This study has at least three limitations. One may be that the principle researcher has CCD and her life experiences may impair the study. Another is that each participant was contacted via email. This method automatically excluded those without access to a

computer and to the CCD website. Finally, this study only included English speaking people in the United States and Canada.

Recommendations for Future Research

The results of this project encourage further research on the lived experience of individuals with CCD and other rare disorders, since people with CCD want to connect with others, share their stories, and learn about others' experiences. In addition, some participants would be interested in having additional research on the pains and problems experienced by people with CCD.

Future research could focus on individuals with rare disorders and the decision of whether or not to have children. Studies could specifically focus on individuals with autosomal dominant traits and the decision process they go through when deciding to have a family.

Other areas of research could focus on the impact of support groups, particularly online support groups. The effectiveness of chat rooms, websites or email lists in coping with rare genetic disorders could be investigated.

Finally, future research could focus on how best to establish a team of experts for rare disorders. With advanced technology and the Internet, patients with any rare disorder from anywhere in the world should be able to connect with a team of experts.

Summary

The purpose of this research was to examine the lived experience of individuals with CCD. Findings indicated there was not a large variation in their life experiences, but a great deal of variation in how each interpreted and responded to those experiences. All participants appreciated being involved in the project, and indeed, whether CCD, OI,

Apert's Syndrome, or so forth (Broder & Trier, 1985; Claesson & Brodin, 2002, Sarimski, 2001) each person has a story to tell and a desire to connect with someone like them. As a researcher with CCD, my own life experience was validated as I spoke with each subject. This study confirmed the importance of qualitative research in learning about a disorder from the perspective of the individual.

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