Supporting Families with Congenital Adrenal Hyperplasia:

Encouraging Whole Family Health

by

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Submitted in partial fulfillment of the requirements for the degree
of Doctor of Psychology in the Department of Clinical Psychology
Antioch University New England, 2014

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SUPPORTING FAMILES WITH CONGENITAL ADRENAL HYPERPLASIA: ENCOURAGING WHOLE FAMILY HEALTH

presented on June 9, 2014

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Dedication

This dissertation is dedicated to my whole family in thanks for their support, encouragement and patience.

Acknowledgements

This project evolved over many years of brainstorming, group process, and exploration of the needs of others. I would like to thank the Gender Management Service at Children's Hospital Boston for allowing me to sit with them and understand so much more about the struggles and needs of these families than I ever could have imagined. This project would not have come about without the inside perspective and support of Dr. Laura Edwards-Leeper, who helped me organize and contain my ambitious striving to meet every need at once. My dissertation committee, headed by Dr. Susan Hawes, and including Dr. Gina Pasquale, was an ever enthusiastic spark to my fire, encouraging me to reach far and wide to incorporate as much as possible. The balance of this team allowed me to find my own direction and design a well-rounded program. I thank my family and dear friends, especially H.K., E.P., and M.W., who consistently and caringly encouraged me to continue toward my goals.

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Abstract

In this dissertation, I discuss the medical and psychological needs of families with children with Congenital Adrenal Hyperplasia (CAH). Due to these needs, I have designed and described a program of social support and psychoeducation to be offered to parents and families. Specifically, I discuss the difficulty that parents have when finding out that their child has CAH, the emotional toll this takes on a parent, on their relationship, and on their family. Using a Family Systems Illness Model, I designed a program that takes into account family functioning, organization, structure, and communication when determining what would be most helpful for these families at different stages in their adaptation to this disease. I then lay out a clear plan of action for implementing this program in settings where parents would gather and connect such as children's hospitals that treat their children. This program could also be used outside of a hospital setting in many different supportive environments.

Keywords: Congenital Adrenal Hyperplasia (CAH), Family Systems Illness model, family sculpture, narrative family intervention, chronic childhood illness, quality of life

Chapter 1

This dissertation lays out a program of social support and psychoeducation for families and children with Congenital Adrenal Hyperplasia. CAH is a multifaceted and complex disorder with many facets that may affect a child and family. Families with children who experience chronic disorders are at risk for increased marital stress, isolation, and increased mental health struggles, all of which can increase the likelihood of difficulty with adherence to medical treatment. As such, it is helpful for families to feel supported and educated, as well as connected to their care providers. The seminar style program of support laid out in the following chapters will help families to feel supported, increase education around the needs of the children, and allow care providers to assess the knowledge level and needs of the families for whom they care.

Background

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive disorder that is caused by mutations of several genes that are important for the formation and processing of androgens, cortisol and aldosterone (Hsu & Rivkees, 2005). These hormones help the body deal with stress, maintain proper levels of glucose in the body and the proper balance of fluid and electrolytes, and have an impact on the development of external sex characteristics in utero (Ernst et al., 2007; Masturzo, 2001; Slijper, Drop, Molenaar, & de Munick Keizer-Schrama, 1998). Because of these genetic mutations, children born with CAH have a variety of medical needs that must be acknowledged and attended to by their parents on a daily basis. Due the potential virilization of females with CAH, a sex assignment that is counter to the child's external appearance may be necessary (Pasterski et al., 2007; Slijper et al., 1998). While there are treatment options available, there is no cure. Additionally, CAH continues to impact a child's psychosexual development throughout their life because of both the prenatal and ongoing hormonal

differences. This chronic disease affects many aspects of a child's life, and the entire family as well.

Research suggests that parents' ability to accept and understand their child's medical and psychological needs can have an impact on the consistency and care their child receives (Schwarzer, 1991). The treatment of this disorder may require daily medication and multiple surgeries, as well as knowledgeable and appropriate medical response in times of sickness or injury (King, Mitchelhill, & Fisher, 2008). CAH has potential implications for the following: learning styles and cognitive ability, Autism spectrum characteristics, gender identity, conformity to gender role, sexual orientation, sexual satisfaction, fertility, increased depression and anxiety, increased likelihood of hypertension, advanced bone age, short stature, precocious puberty, and potentially a higher risk of Multiple Sclerosis (Auyeung, 2009; Bergamaschi et al, 2004; Cohen-Bendahan, Buitelaar, van Goozen, Orlebeke, & Cohen-Kettenis, 2005; Cohen-Bendahan, van de Beek, & Berenbaum, 2005; Collaer, Brook, Conway, Hindmarsh, & Hines, 2009; Cull, 2005; Dittmann, 1990; Ehrhardt, 1981; Fausto-Sterling, 1992; Gaudiano, 2010; Hines, 2003; Hurtig, 1987; Inozemtseva, 2008; Knickmeyer, 2006; Maheu, 2008; Mueller, 2008; Pasterski et al., 2005, 2007; Puts, McDaniel, Jordan, & Breedlove, 2008; Quadagno, 1977). All this, in addition to the possibility of the child having a physical appearance that is not the norm can be overwhelming. The news of all these possibilities can be incredibly difficult for parents to accept and understand in a short period of time, and this processing and understanding may be necessary in the case of the potential medical impact of CAH. The support of a clinical psychologist, as well as social support from other parents in a similar situation, can be comforting for parents (Ell, 1996; Hughes, 2007). However, because of the history of secrecy and shame that has existed around CAH and other Disorders of Sexual Development, medical

staff have been less likely to inform parents of the potential benefits of psychological and social support (Hughes, 2007).

In 2001 Lenore Abramsky examined what medical providers reported telling parents in the United Kingdom when prenatal chromosomal analysis indicated a Disorder of Sexual Development diagnosis (Abramsky, Hall, Levitan, & Marteau, 2001). Through a semi-structured interview with medical professionals in charge of discussing results with parents, she found it "disturbing to note the haphazard nature of how parents were informed of the diagnosis, what information was given, and what was implied" (Abramsky et al., 2001, p. 465). Further, parents' responses to these discussions with medical providers suggested that their first impressions from this discussion "may affect how information presented later is interpreted or even whether it is sought" (Abramsky et al, 2001, p. 463). There are few studies that examine the reactions of parents when a child is born with a congenital disorder and what these reactions might mean for the care of the child over time. What is expected, as can be seen by examining what research has been done, is that parents will go through a period of shock and denial following discovery of the disorder. This response is likely to be followed by anger, sadness, and eventually adaptation and reorganization (Drotar, Baskiewicz, Irvin, Kennell, & Klaus, 1975). The suggested course of action for medical providers taken from this research emphasizes understanding, repetition of technical information, and connecting parents with others as a means to assist parents with the necessary adjustments to enhance bonding and attachment. Additionally, in her research into conditions other than CAH, Beverley Myers examined the success of discussions with parents regarding discovery of a child's chronic or fatal condition; she found that the characteristics and manner of the provider having the discussion had great impact on the parents' reactions as well as their ability to take in and retain information (Myers, 1983).

There is, on the other hand, a great deal of research on the impact of chronic illness on families, as a whole and each individual member (Goldberg, Janus, Washington, Simmons, MacLusky & Fowler, 1997; Lavigne & Ryan, 1979; Seiffage-Krenke, 1998; Svavarsdottir and Rayen, 2005). Although some forms of CAH can go undiagnosed until adulthood, most forms of CAH are diagnosed and treated in infancy and childhood. Because this is the case, a review of the research on chronic childhood illness is an appropriate starting point for anticipating parents' experiences of their child's diagnosis of CAH. Each individual within a family can be affected by one member's chronic illness (Williams, 1997). Additionally, the organizational structure and daily interactions of the family can be negatively affected (Rolland, 1999). Quality of life studies are just beginning to show the impact on the child of the functioning of the family and parental response to childhood chronic illness (Brinkmann, Schuetzmann, & Richter-Appelt, 2007; Rolland, 1999). In general, there are indications that family response and functioning as a whole may have a greater impact on the course of the child's illness and acceptance of the disease by the child than the severity of the disease itself (Bennet, 1994; Lavigne & Faire-Routman, 1993).

A framework is needed when working with an entire family system to understand the experiences of the family and the effects that a chronic disease can have on a system. The Family Systems-Illness Model developed by Rolland (1994) does this by classifying illnesses and the stages of complexity, which can have effects on the individual child as well as the family system. Every illness is different and places different demands and stressors on a family, however most involve onset, course, outcome, and level of incapacitation (Rolland, 1994). Additionally, each disorder has different phases; crisis, chronic, and terminal. The time of diagnosis of a child with CAH is the initial crisis phase, though this can be repeated in instances

of salt-wasting CAH with adrenal crises possible at any moment. While CAH is rarely terminal, it is still potentially possible for death to occur due to dehydration or adrenal crisis. At various times throughout the child's life, each of these phases can impact the child and family, sometimes simultaneously.

Having a child with a chronic and potentially life-threatening illness can be seen as a significant loss for a parent (Olshansky, 1962). An ongoing disorder can cause "profound sadness or chronic sorrow that [is] periodic, yet, since the child lived, enduring for the child's lifetime" (Olshansky, 1962). This ongoing chronic sorrow can affect the day-to-day quality of life for families who are living with CAH. While any aspect of the disorder or typical daily life could serve as a trigger for an episode of sorrow, parents may be particularly vulnerable to "the state of being physically overwhelmed, and feelings of invalidation, including lack of voice, isolation, feelings of unfairness, and the outcome of renormalization" (Kendall, 2005, p.48). While each episode ideally returns back to this homeostasis of 'renormalization', research suggests that social isolation increases the risk of disease, premature death and depression while, not surprisingly, social support promotes health and wellbeing (Bisschop, Kriegsman, Beekman & Deeg, 2004). This calls for a system of social and professional support for these families, which may be the most important resource they can have (Meleski, 2002).

Several variations of CAH are possible, based on the different genes that have mutated. These differing variations then have an impact on the degree of excess or deficiency in hormone production (Hsu & Rivkees, 2005). The most severe form of CAH is Classic Salt Wasting CAH, which occurs at a rate of 1 in 15,000 (Pang et al., 1988). Those with this severe form of CAH, aside from external sex differences, have difficulty with salt levels in the body, which may lead to dehydration, abnormal growth over time, and a potential adrenal crisis and death (Hsu &

Rivkees, 2005). Those with Classic CAH are required to take medication daily for their entire lives and parents must know how to supplement these medications at times of stress or injury to compensate for their child's inability to balance these physiological needs on their own (King et al., 2008). Because of the complexity of these issues, it is essential that parents feel supported and heard so that they can feel free to ask for more information and help as needed.

Psychoeducation has been shown to be an important treatment tool for both physical and mental illnesses (Campbell, 1997; Karamandidou, Weinman, & Horne, 2008; Kaslow et al., 2000; Ng et al., 2008; Pollio, North, Reid, Miletic, & McClendon, 2006; Tyerman & Booth, 2001). As a method of treatment, psychoeducation gives families more information about this disorder, as well as the tools needed to manage everyday life for everyone in the family. This knowledge can be used to change behaviors and feelings. Additionally, the model of seminar-style care increases connections between families who are also learning and managing this disorder.

Families dealing with CAH have some options for support available to them. Many clinics offer local educational programs to gather families together. Additionally, there are forums and message boards online that are active and frequently used by parents to find connections with other parents. The MAGIC (Major Aspects of Growth In Children) foundation offers newsletters, weekly emails, legal assistance, scholarships, educational programs, and an annual conference to support children and their families. Having support that is available, knowledgeable, and familiar can be a great tool for families.

Statement of the Problem

The American Academy of Pediatrics' (AAP) is a source for information concerning the outcome of children born with a Disorder of Sexual Development (DSD) such as CAH.

According to the AAP, there are three components to psychosexual development at the core of determining how best to help an infant with DSD. These are gender identity, gender role, and sexual orientation (Lee, Houk, Ahmed & Hughes, 2006). Whenever a multidisciplinary team attempts to determine the best course of action, it should be with these three components in mind. The team should strive to factor in all three of these, additionally looking at "diagnosis, genital appearance, surgical options, need for lifelong replacement therapy, potential for fertility, views of the family, and sometimes, circumstances relating to cultural practices" (Lee, Houk, Ahmed & Hughes, 2006, p. e491). With all of this hindering something as easily taken for granted by a typical population as a clear sex assignment, which is only a small piece of CAH, it is no wonder that "shortly after the diagnosis, the majority of both parents react… with shock, grief, anger, and shame" (Slijper et al., 2000, p. 11). Parents need support and information to help them process all of the different aspects of this complicated and unexpected disorder.

Study Objectives

The program of social support and education laid out in this dissertation has been designed to fit into existing medical service programs and supplement the ongoing care of a multidisciplinary team. Because parental knowledge, acceptance and care are so important to the health and well-being of the child, this program increases knowledge, feelings of tolerance and understanding, and adherence to medical treatment for parents of children with CAH. In the past, medical care was not as ongoing, and diseases such as CAH would have had greater consequences and shorter lifespans for these children (Witchel, 2010). Now that children are able to live longer lives, they continue to need care for the length of that life. Parents need greater supports to manage the medical, behavioral, and psychosocial impacts of this disorder. This program serves as an initial step in creating an environment that would become the

"medical home" for these children and families, allowing all of their care to be coordinated and simplified. This educational and supportive seminar is a beginning of ongoing connection for families as well as the providers from whom they will receive services.

Significance of the Study

Whenever a child is born, there is the immediate question: is it a boy or a girl? Imagine if you had just had a baby and had to tell people that you did not know the sex, and the doctors and nurses did not either. That moment could have a severe impact on any parent, but is often just the beginning of the impact of CAH on individuals and families. Parental acceptance and adjustment can affect medical adherence as well as a child's acceptance of their own disorder. With ongoing care of a child with CAH being needed throughout their entire life, it is sensible to assess the family structure and functioning early on in the course of the child's life. This will allow health care providers to offer a more tailored individual experience for each family. Additionally, connecting families decreases the feelings of isolation and shame that can be problematic for these families. CAH affects 1 in 15,000 children born each year. Every newborn in the United States is screened for this disorder, allowing for more children to be diagnosed at an earlier age. While this lessens the likelihood of a child having a salt-wasting adrenal crisis, it also leaves a lot of parents stymied by a disorder that they did not even know existed.

Summary

When a child is expected, most parents anticipate the care and guidance a child will need throughout their life. This in and of itself can be overwhelming, but when a child is born with a congenital disorder, one that affects outward appearance, health and lifespan, and a child's sense of identity, it is to be expected that the task in front of the parents will indeed be extremely

overwhelming and require additional supports. Parents and children with CAH will face a long road of medical intervention, the consideration of medical needs within their daily life, changes in social expectations, and the possibility of isolation and altered mental health. The goal of this dissertation was to design a program of educational support and social connection in order to give a different view on this disorder and how it may affect families' lives. As such, this program, as laid out in the following chapters, can serve as a source of life changing support to families, which may, in turn, alter the quality of life of the children born with CAH. The following chapters detail the characteristics of CAH, the likely emotional path for the families, and the value of psychoeducation as a tool of support. Next, the process of creating the seminar program is reviewed, as well as the details of the program. Finally, a discussion of ways to determine the quality and efficacy of the program will be given, allowing providers using the program a means for evaluation.

Chapter 2: Literature Review

Congenital Adrenal Hyperplasia is a chronic, life-altering disorder that can be diagnosed prenatally, at birth, or later in life. As there are ongoing medical as well as social implications of the disorder; CAH is an all-encompassing disorder that affects the lives of not only the children with the disorder, but their entire family as well. This literature review will discuss the details and ramifications of CAH itself. Additionally covered will be the relevant literature discussing impact on family and a model of understanding this impact, the Family Systems Illness model. As there is much to learn for families when they are faced with CAH, psychoeducation will also be an important part of the seminar, and its efficacy will be discussed herein.

The included relevant topics for discussion of literature are the types of CAH that exist, the medical and psychological ramifications of these various types, and the psychological effects of chronic disease on both children and families. I have chosen these topics for review in order to examine what CAH looks like for parents and children as well as what impact that may have on the physical course of the disease and the impact on the family.

What Is Congenital Adrenal Hyperplasia?

Overview of Disorder

Congenital Adrenal Hyperplasia (CAH) is an inherited genetic disorder that affects hormone production in the adrenal glands. The physical differences result in an excess of androgen and deficiencies of cortisol and aldosterone. The effects of these imbalances are detrimental both pre and postnatally and can lead to both illness and growth problems (King et al., 2008). With CAH, the body cannot produce cortisol. When the pituitary gland sends out Corticotropin Releasing Factor (CRF), the brain receives the message to produce Adrenocorticotropin Hormone (ACTH), which in turn sends a message to the adrenal glands to

produce the needed cortisol (Hsu & Rivkees, 2005). In a typically functioning body, this message would be received by the adrenal glands, cortisol would be produced, and the ACTH would cease being produced. With CAH, however, the adrenal glands cannot produce cortisol, though they keep trying. This causes the glands to grow overly large in an attempt to compensate, thus the "hyperplasia." In addition to the improper production of cortisol, the kidney functioning seen in CAH leads to a lack of production of aldosterone. Because there is no production of cortisol or aldosterone, the kidneys overproduce androgens (Hsu & Rivkees, 2005; Wilson, 2010).

Lack of aldosterone. Aldosterone is the hormone that is responsible for maintaining a proper electrolyte balance in the body. This is done by releasing potassium from individual cells and retaining sodium. If there is no aldosterone, the sodium (salt) does not stay in the cells and the potassium does. Without enough salt in the cells, fluid levels are affected and circulation and blood pressure can become dangerously low. In extreme situations, blood circulation can stop altogether and this 'shock' can lead to death. In addition to difficulty with blood circulation, the lack of salt in the cells can lead to a condition called hyponatremia, which can lead to seizures. Too much potassium in the cells leads to a condition called hyperkalemia, which can cause an irregular heart beat which could also cause shock and even death (Hsu & Rivkees, 2005; Wilson, 2010).

Lack of cortisol. Cortisol is important to help the body react to illnesses and stressful situations. It is responsible for maintaining homeostasis and returning the body to homeostasis following a stressor. It is also responsible for maintaining blood sugar levels, which in turn is essential to prevent mental confusion and seizures as a result of hypoglycemia. Cortisol is additionally responsible for maintaining proper blood pressure, and a deficiency, like with an

aldosterone deficiency, can lead to poor circulation and shock (Hsu & Rivkees, 2005; Wilson, 2010).

Overproduction of androgen. In utero, the overproduction of the male hormone androgens can lead to virilization of female babies. This can cause some virilized female babies to be mistaken for male at birth (Wilson, 2010). Postnatally, an excess of androgens can lead to such symptoms as rapid physical development and growth as seen by excessive height compared to same-age peers. Signs of early puberty can also be seen, such as body odor, armpit hair, pubic hair and acne as young as age four or five (Hsu & Rivkees, 2005). Excessive androgen can also calcify growth plates earlier than what is typical, which may cause a shortened adult stature. Later in life, CAH can cause problems with infertility; in men due to testicular adrenal rest tumors (TART) and in women due to infrequent or a complete lack of menstrual cycles (Claahsen-van der Grinten, Hermus, & Otten, 2009; Hsu & Rivkees, 2005). Other cosmetic problems such as severe acne, hirsutism, and male pattern baldness can also be seen (Hsu & Rivkees, 2005).

Types of Congenital Adrenal Hyperplasia

There are three distinct types of CAH. The first, Classical Salt-Wasting CAH, is the most severe and is an early onset version, presenting at birth. The most apparent sign of severe classical CAH is virilization. This is usually present at birth in affected females, but is delayed until the third or fourth year of life in boys (Hughes, 1986). In female babies there can be noticeable abnormality of the genitals, although the internal reproductive organs are typically developed. For male babies the external genitalia usually appear typically developed, however they can be enlarged or have differences in pigmentation (Hughes, 1986; Merke & Bornstein, 2005; Wilson, 2010). The question of surgical correction of the genitals is one that can be

considered for females with CAH (Mendonca, Domenice, Arnhold, & Costa, 2009). This is often not a consideration for males however because development of external sex organs proceeds in a typical manner for males with CAH.

About 75% of people with classical CAH have the severe "salt-wasting" form (Lekarev, Parsa, Nimkarn, Lin-Su, & New, 2010). The severity of the disorder can be life threatening as the fluid and electrolytes are out of balance in the body. Also, malnutrition and failure to thrive can be an issue as some symptoms include increased urination and poor feeding which can lead to vomiting, dehydration and cardiac arrhythmia (Merke & Bornstein, 2005). The likelihood of death in the first few weeks of life is high due to this. Children who survive the first few weeks of life and maintain stable physical functioning still risk rapid growth, precocious puberty, and premature skeletal maturation (Merke & Bornstein, 2005). A typical presentation for a young child with CAH is extreme height compared to same age peers. However, because of the changes in bone growth, these children are likely to achieve full height much earlier than other children and ultimately have a shorter than average stature.

Treatment of salt-wasting CAH includes taking lifelong steroids. Females with CAH have normal internal genital structures and can experience normal menstruation, potential fertility, and successful pregnancy. Because of the irregularity of hormones, CAH can cause hirsutism, and this could be treated as well (Merke & Bornstein, 2005).

A less severe form of CAH is non-classical, virilizing CAH. In these cases, adrenal insufficiency is prevented due to the typical amount of steroids being produced (Wilson, 2010). At birth, a child with this form of CAH will appear to have typically formed genitals for their sex, however they are at risk for early puberty, excess hair growth, and there is risk of clitoral enlargement later (Merke & Bornstein, 2005). Although children with this form of CAH do not

have the issues of salt wasting seen in classical CAH, they are still at risk of accelerated growth as well as early bone maturation and growth plate closures because of the excess of androgens over time (Wilson, 2010).

An additional form of non-classical CAH can present in the late teens and early twenties. This form is often called Late-Onset CAH because it can go unseen until symptoms begin to appear later in development. In this form, female teens will have menstrual irregularities, severe acne, excessive facial hair growth, and increased risk of infertility (Hughes, Nihoul- Fékété, Thomas, & Cohen-Kettenis, 2007). Additionally, they may show the beginning of male pattern baldness and could be diagnosed with Polycystic Ovarian Syndrome (Claahsen-van der Grinten, Stikkelbroeck, Sweep, Hermus, & Otten, 2006; Hagenfeldt, Janson, Holmdah, Falhammar, Filipsson, Frisén, et al. 2008; Merke & Bornstein, 2005). Both males and females with Late-Onset CAH may show premature development of body hair, body odor, rapid growth spurt, oily hair and skin, severe acne, mood swings, and infertility (Claahsen-van der Grinten et al., 2006; Hagenfeldt et al., 2008; Merke & Bornstein, 2005). Males may show early beard growth, enlarged penis with small testes, and a low sperm count (Claahsen-van der Grinten et al., 2006; Hagenfeldt et al., 2008; Merke & Bornstein, 2005).

Clinical presentation in males. Unlike with virilized female infants, CAH in males is not readily identified directly after birth because the child appears to have developed typically when examined externally (Merke & Bornstein, 2005). If the CAH is the severe salt-wasting form, babies may present at 1-4 weeks with failure to thrive, recurrent vomiting, dehydration, hypotension, hyponatremia, hyperkalemia, and shock (Merke & Bornstein, 2005). This can be a dangerous time if the infant is misdiagnosed with pyloric stenosis or gastroenteritis and treatment with glucocorticoids is delayed. If the child has the less severe form of CAH, they may not be

diagnosed until later in childhood. Signs of CAH in later childhood would include early development of pubic hair, phallic enlargement, extreme height for age, and advanced skeletal maturation, as seen in bone growth analysis (Merke & Bornstein, 2005; Warne, 1992). In a small percentage of males with less common forms of CAH, ambiguous genitalia or female genitalia may be seen due to a lack of production of testosterone during the first trimester of pregnancy (Merke & Bornstein, 2005).

Etiology

For a child to have the recessive disorder CAH, both parents must be carriers of the affected gene. CAH is caused by the lack of a specific enzyme. There are several types of possible deficiencies, though three are the most likely. Ninety-five percent of cases of CAH are caused by 21-hydroxylase deficiency (Hsu & Rivkees, 2005). It is essential for a specific diagnosis to be made due to the differing needs of different types of CAH, as the specific missing enzyme give clues as to the likely response to treatment.

21-hydroxylase deficiency. This is the most common form of CAH. The gene affected in this form of CAH is called CYP21A2, which is key to the production of the enzyme 21-hydroxylase (Merke & Bornstein, 2005; Witchel & Azziz, 2011). This enzyme is an essential part of the adrenal production pathway for cortisol, as well as for the conversion of 17-hydroxyprogesterone to 11-deoxycortisol. When 21-hydroxylase is blocked, the 17-hydroprogesterone builds up without conversion and the body increases production of testosterone in response. This type of CAH is most likely to result in virilizing symptoms due to the overproduction of androgens. Additionally, this type is most likely to cause salt wasting symptoms due to the lack of salt-retaining hormones, which can in turn cause problems with low blood pressure (Hsu & Rivkees, 2005).

11-beta hydroxylase deficiency. In about five percent of cases of CAH, aldosterone and cortisol production are affected due to the deficiency of this enzyme (Hsu & Rivkees, 2005). In these cases, however, the result is not usually as severe as salt-wasting. This is because individuals with this form of CAH produce enough other salt-retaining hormones to compensate for the lack of aldosterone and prevent salt-wasting (Hsu & Rivkees, 2005). 11-OH deficiency does increase production of androgens and can cause virilization, however in this case it causes difficulties with high blood pressure rather than the low blood pressure of 21-OH deficiency (Hsu & Rivkees, 2005).

3-beta hydroxysteroid dehydrogenase deficiency. In less than three percent of cases of CAH, the cause is a 3-beta deficiency. Like the other forms of CAH, this does cause impairment in cortisol and aldosterone, but unlike the other forms, it causes a lack of androgen production. This, in turn, causes under-masculinization rather than over-masculinization in males with CAH (Hsu & Rivkees, 2005). This form of CAH can cause ambiguous genitalia in either sex.

Prevalence

Congenital Adrenal Hyperplasia is an autosomal recessive disorder caused by a genetic mutation. Since a recessive gene causes all forms of CAH, both the mother and father must be recessive carriers of CAH for a child to have CAH. In ninety to ninety-five percent of cases, CAH is caused by a mutation or deletion of CYP21A, which results in 21-hydroxylase deficiency (Hsu & Rivkees, 2005). This genetic difference can be a mutation or partial deletion and is fairly common in selected populations, occurring in as many as 1 in 3 Ashkenazi Jews, or 1 in 7 individuals in New York City (New, 2010). Those with this deletion can be unknown carriers of the disorder, rather than affected themselves. The estimated prevalence of carriers is 1 in 60 in the general population with CAH appearing in 1 per 15,000 (Hsu & Rivkees, 2005).

CAH caused by 21-hydroxylase deficiency is found in all populations, it is not specific to one race or ethnic group. 11-beta-hydroxylase deficiency is more common in persons of Moroccan or Iranian-Jewish descent (New, 2010). The prevalence of 3-beta deficiency is unknown as it is extremely rare.

Course of the Disorder

At birth, CAH may be determined due to virilization of females, or salt loss in either sex (Hughes et al., 2007). In a child with salt-wasting CAH, an adrenal crisis in the first month of life may be the first sign of CAH. Because of the deficiencies in production of cortisol and aldosterone, a newborn affected by CAH will experience decline in health, resulting in extremely low blood sugar, poor blood circulation and electrolyte abnormalities (Hsu & Rivkees, 2005). This may then lead to shock with the potential for loss of consciousness and possibly sudden death. Warning signs in a newborn include "dehydration, vomiting, reflux, irritability, and lethargy" as well as poor weight gain or possible weight loss and prolonged jaundice (Hsu & Rivkees, 2005, p. 61).

Those neonates with CAH who are not salt-wasters may still present with abnormal genitalia. While a neonate with abnormal genitalia may be the result of several disorders of sex development, CAH can easily be diagnosed through hormone testing (Hsu & Rivkees, 2005). When a child is born with atypical genitalia, doctors use a system called the "Prader scale" to differentiate the various levels of masculinization. A female child who is slightly masculinized, as determined by degree of labioscrotal fusion and the position and length of the phallus will be diagnosed as "Prader 1" whereas a female child who appears as a typical male would be diagnosed as "Prader 5" (Wolfe-Christensen et al., 2012).

Some children with CAH are born with typically developed genitalia, thereby not giving the immediate outward appearance of the disorder. Non-virilized and non salt-wasting females and males may present to a pediatrician in early childhood years with extreme height for age, precocious puberty, early acne and body odor, or early hair growth (Hsu & Rivkees, 2005). Nonclassic CAH is generally recognized at or after puberty because of oligomenorrhea or virilizing signs in females (Merke & Bornstein, 2005).

Medical Ramifications

In addition to the signs and symptoms that can lead to diagnosis and determination of CAH, individuals with CAH are at an increased risk for short stature as adults, even though they may be extremely tall for age in early childhood (Merke & Bornstein, 2005; Warne, 1992). The growth plates of children with CAH close prematurely due to the high concentrations of sex steroids (Merke & Bornstein, 2005). The treatments available for CAH, glucocorticoids, also suppress growth (Merke & Bornstein, 2005). Studies have shown that early detection and treatment during the early years of life and throughout puberty are most likely to positively affect adult height (New, Gertner, Speiser, & del Balzo, 1988; Young & Hughes, 1990). Additionally, children with CAH struggle with their weight due to steroid treatments and hormone irregularity (Merke & Bornstein, 2005). Obesity is a likely symptom of CAH, and the rates of obesity are higher in individuals with CAH than in an unaffected population (Merke & Bornstein, 2005). There is some speculative research being conducted to examine the potential risk of multiple sclerosis among individuals with CAH (Bergamaschi et al, 2004).

Mortality/morbidity. At birth, most girls with severe CAH are noticed due to their external genital differences. However, CAH in boys is not as likely to be discovered because the outward genital appearance is typical. Because of this, these children may not be diagnosed and

treated until they present in the first weeks or months of life with an adrenal crisis (Merke & Bornstein, 2005). Because it is unclear what occurs medically for a newborn in adrenal crisis, this can be fatal if treatment is not begun right away. Adherence to medical treatment in the first few years is also reported to be difficult for parents due to the unpredictability and novelty of the medical need, and "about eight percent of patients have been reported to experience hypoglycemia" (Merke & Bornstein, 2005, p. 2130) within the first few years of life. While CAH is not inherently fatal, mismanagement of treatment or lack of diagnosis may cause life-threatening symptoms.

Fertility. Males with CAH have decreased fertility compared to non-affected males due to testicular adrenal rest tumors (Claahsen-van der Grinten et al., 2006). For females with CAH, the difficulty with fertility will depend on the type of CAH present. Severe salt-wasting CAH can inhibit fertility by decreasing ovulation and menstruation. In less severe forms of CAH, an adult female can struggle with ovarian hyperandrogenism (polycystic ovary syndrome), ovarian adrenal rest tumours, and previous genital surgery (Claahsen-van der Grinten et al., 2006; Hagenfeldt et al., 2008).

Behavioral Ramifications

CAH affects every child differently. However, there are some potential areas of difference that are likely to be seen in most children with CAH. These include differences in gender-based play, social interactions, learning, sexuality and gender identity, and mood. While some of these are minor and not always present, some, such as the differences in play behaviors of young children with CAH, and sexual orientation of adults are seen as hallmark traits of individuals with CAH.

Gender-based play. Young children with CAH have long been seen to show differences in gender-based play behaviors. Girls with CAH have shown increased male-typical toy, playmate, and activity preferences (Ehrhardt & Meyer-Bahlburg, 1981; Hines, Ahmed, & Hughes, 2003, Hines, 2004; Pasterski et al., 2005). Some studies report that this difference may be caused by hormonal treatment after birth (Fausto-Sterling, 1992; Quadagno, Briscoe, & Quadagno, 1977), however others report that it may be due to disease characteristics rather than prenatal or postnatal exposure to hormone treatments (Auyeung et al., 2009). Girls with CAH also tend to show elevated male traits such as assertiveness, aggression, and rough play (Berenbaum & Hines, 1992; Berenbaum & Resnick,1997; Cohen-Bendahan, Buitelaar, van Goozen, Orlebeke, & Cohen-Kettenis, 2005; Ehrhardt, Epstein et al.,1968; Ehrhardt & Meyer-Bahlburg1981; Hampson, Rovet, & Altmann,1998; Zucker et al., 1996). Girls with CAH may also show increased preferences for stereotypical boy toys and male playmates (Berenbaum & Hines, 1992; Berenbaum & Snyder, 1995; Dittmann et al., 1990). Additionally, they often show less interest in play with dolls or domestic imaginative play and less interest in makeup and clothing than other girls their age (Dittmann et al., 1990; Ehrhardt & Meyer-Bahlburg, 1981; Leveroni & Berenbaum, 1998). Girls with CAH have been found to be less tender-minded on the sensitivity scale of personality inventories than girls without CAH (Matthews, 2009). Additionally, they have been found to have greater physical aggression and less interest in infants than other females (Matthews, 2009). Male children with CAH were more similar to typically developing males in interest in infants, though they were found to be less dominant, more tender-minded, and less physically aggressive than unaffected males (Matthews, 2009).

Girls with CAH, when compared to their unaffected sisters, were more interested in male-typical toys. It has been speculated that this may be the result of parents treating their

daughters with CAH differently from their other children due to the ambiguity of their physical genital appearance. However, some research has shown that parents often encourage sex-typical play in their daughters with CAH more than their other children, and yet these girls continue to choose typical boy type toys (Pasterski et al., 2005; Pasterski et al., 2007).

Social interactions. Girls with CAH have been found to have some difficulty with social interactions. When tested on the Autism Spectrum Quotient (AQ), girls with CAH were found to score significantly higher on autistic traits than girls without CAH, specifically related to social skills and imagination (Knickmeyer et al., 2006).

Learning and performance difficulties. In addition to difficulties with social interactions and differences in play behaviors, children with CAH show deficits in motivation for voluntary action, in this case, lack of motivation toward action based on monetary reward (Mueller et al., 2013). They also have increased spatial relational skills compared to girls without CAH, more similar to boys with or without CAH (Mueller et al., 2008). Girls with CAH often show reduced fine motor skills, though they exhibit increased grip strength and greater targeting skills when compared to girls without CAH (Collaer et al., 2009). Boys with CAH showed reduced grip strength that is believed to be due to their generally smaller stature (Collaer et al., 2009). Girls with CAH also show verbal and reading related learning disabilities, including lower performance in repetition and expression tasks, decreased right-left comprehensions, and lowered verbal semantic fluency (Inozemtseva, 2008). Some studies have additionally shown memory deficits in children with CAH (Maheu et al., 2008). Research continues to be needed to explore potential cognitive functioning differences, as it is not yet known what long-term hormonal differences and treatment with synthetic hormones may cause.

Sexuality and gender. Most girls with CAH will grow to have a female gender identity and a heterosexual orientation. However, there is a higher percentage of women with CAH who report a dissatisfaction with their gender, a wish to change to a male identity, or a homosexual orientation than the unaffected population (Dessens, Slijper, & Drop, 2005; Gupta et al., 1998; Meyer-Bahlburg, Dolezal, Baker, Ehrhardt, & New, 2006; Meyer-Bahlburg et al., 1996; Slijper, Drop, Molenaar, & Keizer-Schrama, 1998). Women with CAH also report difficulty with sexual satisfaction, decreased libido, lower rates of sexual activity, and difficulty achieving orgasm and sexual pleasure (Dittmann, Kappes, & Kappes, 1992; Dittmann et al., 1990; Hines, Brook, & Conway, 2004; Kuhnle & Bullinger, 1997; Meyer-Bahlburg, 2001; Meyer-Bahlburg, Dolezal, Baker & New, 2008; Mulaikal, Migeon, & Rock, 1987; Wisniewski, Malouf, & Gearhart, 2004; Zucker et al., 1996).

Women with CAH also often show a higher level of bodily concerns. Genital surgery done at a young age and without consent of the child can cause some individuals to feel as though their body is not their own (Cull, 2005). These girls are also shown to delay dating and sexual relationships, possibly due to such concerns about their bodies as well as difficulty with social interactions (Hurtig, 1987). Additionally, CAH and medication both cause increased weight, which may add to these difficulties.

Mood. Women with CAH have increased levels of depression and stress, potentially caused by weight problems, body issues, sexual difficulties, and challenges with relationships as well as the impact of hormones on mood. Additionally, the stigma of having atypical genitalia and an atypical gender expression may increase these struggles (Cull, 2005). Even children with CAH often report mood problems, such as depression, anxiety and distress (Johannsen, Ripa, Mortensen, & Main, 2006; Kuhnle, Bullinger, & Schwarz, 1995).

Treatment Options

Children. Children with CAH show various symptoms depending on the severity and type of CAH that they have. Females with CAH may have the severe form and virilized genitalia. Others may have less severe CAH that does not include virilized genitalia. Most forms of CAH found in childhood will require medical treatment of some sort.

Virilized females are the typically discussed population of children with CAH when reviewing treatment options. Eighty-five percent of female children with CAH are recommended to have surgery to alter their enlarged genitalia (Crouch & Creighton, 2007; Crouch, Laio, Woodhouse, Conway, & Creighton, 2008; May, Boyle, & Grant, 1996; Mulaikal et al., 1987; Stikkelbroeck, Hermus et al., 2003; Stikkelbroeck, Oyen et al., 2003). There is debate over timing of surgeries, with some researchers recommending as early as 2-6 months of age, and others recommending waiting until the child is old enough to understand and choose for themselves (Clayton et al., 2002; Crouch & Creighton, 2004; Nihoul-Fékété, 2008). Even after two or more surgeries to create more typical looking genitalia, many girls will need to have further surgeries at a later time to continue this process, or to correct problems from earlier surgeries, such as vaginal stenosis, fistulas, and incontinence (Crouch & Creighton, 2007; Crouch et al., 2008; Jordan-Young, 2012; May, Boyle, & Grant, 1996; Mulaikal et al., 1987; Stikkelbroeck, Hermus et al., 2003; Stikkelbroeck, Oyen et al., 2003). When surgery is done early in life, it is recommended that further surgeries be delayed until adolescence "to achieve better cosmetic and functional results." (Gollu, 2007, p. 843).

Historically, these feminizing surgeries have attempted to create more typical looking genitals, however the procedures have not always been successful in terms of functionality.

"Data suggest that all genital surgeries leave most women with nerve damage, impairing sensitivity and orgasmic capability, sometimes severely" (Jordan-Young, 2012, p. 1741).

More recently, however, surgical procedures have improved and doctors are attempting to leave their patients with options for the future. They are now attempting to do tissue-preserving feminizing clitoroplasty that will leave the option open for the patient to choose to reverse the surgery and identify as male with corresponding genitalia (Chia, 2007). This takes into consideration that some females with CAH will prefer to identify as male. Culturally, parents may choose to raise their child as male due to the external presentation of genitalia. In countries where it is more beneficial to be male, a higher number of children (seventy-four percent) with CAH are reared as male (Kulkarni, Panigrahi, Das, Kaur, & Marwaha, 2008).

Because most children with CAH have had genital surgery, continued monitoring throughout childhood is necessary. Doctors are required to inspect the genitals following surgery and during hormonal treatment. For some girls, vaginal dilation is required to lengthen the vagina or keep it open after surgery. In addition, observation of the clitoris is required because excessive growth can be a sign of poor hormonal treatment (Jordan-Young, 2012). These follow up medical visits can be difficult for children and their parents as it can feel "intrusive and dehumanizing" (Karkazis, Bostwick, & Martin, 2008, p. 265).

Children with CAH who have typical genitalia still require treatment with hormones to balance the excess of androgen and absence of cortisol. Children are treated with glucocorticoids to obtain optimal levels of hormones to prevent adrenal crisis and to allow for the child to reach an appropriate adult height (Claahsen-van der Grinten et al., 2011). In children the most frequent medication is daily hydrocortisone. This medication protocol requires determining and maintaining the proper dose of medication that will approximate natural levels

of hormones. This is different for each child, and imbalance can result in hyperandrogenism, Cushing's syndrome, atypical puberty, and reduced height in adulthood (Dörr, 2007). Additionally, as adults a person with CAH will require continued treatment in order to increase the likelihood of fertility, prevent hirsuitism, prevent testicular adrenal rest tumors, decrease the risk of osteoporosis and cardiovascular disease (Claahsen-van der Grinten et al., 2011).

In addition to daily treatment with synthetic glucocorticoid replacement, individuals with CAH sometimes require an additional dose of medication, called a stress dose of their medicine to mimic the naturally occurring increase in cortisol during stress (Shulman, Palmert, & Kempt, 2007). These medical applications can be in the form of pills, intramuscular injection, or rectal administration (Nickels & Moore, 1989). These stress doses may be necessary when an individual has a headache, cold, fever, infection, any form of illness, trauma or surgery (Claahsen-van der Grinten et al., 2011).

Adults. Treatment of CAH for adults, when it is not a continuation of childhood treatment, is generally for non-classical CAH, which may not be found until a woman has difficulty conceiving a child. At that time, women can be treated with ovulation inducers, or hydrocortisone treatment to assist them with becoming pregnant and continuing a viable pregnancy to term (Bidet et al., 2010). When a woman is found to have CAH that is inhibiting her fertility, her partner should have genetic testing as well so as to determine if their potential child would be at risk of having CAH as well (Bidet et al., 2010).

Prenatal diagnosis. Due to the genetic etiology of CAH, a family with a child who has already been diagnosed with CAH has the option of doing prenatal testing to determine if a child not yet born has CAH as well. Additionally, family members may choose to have genetic testing done to determine carrier status prior to planning future pregnancies. A fetus can be examined

through ultrasound for an increase in nuchal translucency (Masturzo et al., 2001). Also, amniotic fluid analysis can be done for women who have had a child with salt-wasting CAH (Hughes, Dyas, Riad-Fahmy & Laurence, 1987). Additionally, chorionic villi sampling can culture the cells to determine specifically the genetic type and severity of CAH (Nimkarn & New, 2007). When a woman who already has a child with CAH becomes pregnant again, she has the option of beginning prenatal treatment with dexamethasone (Nimkarn & New, 2007). This treatment, when begun before nine weeks gestation, can prevent the virilization of female fetuses with CAH. By 9-11 weeks gestation, testing can determine if the fetus has CAH and whether treatment can continue or be discontinued (Nimkarn & New, 2007). With proper dosing and continuation throughout pregnancy, the affected females will not have virilization of their genitals, though they will still have CAH and need to be treated after birth.

Prenatal treatment with dexamethasone is an extremely controversial topic. Some studies have shown that this treatment may affect future learning and performance as well as self-perception, though others have shown that there are no major behavioral problems, long term memory or IQ difficulties (Brabham et al., 2000; DeKosky, Nonneman, & Scheff, 1982; Lajic, Nordenström, & Hirvikoski, 2011; Mueller et al., 2008).

The Effects of Chronic Childhood Illness

There has been a fair amount of research done examining the physical and psychological effects of Congenital Adrenal Hyperplasia on children; however, not much research can be found examining how the disorder affects the system of the family and other members of the family. The ongoing physical care needs for children with this disorder, as well as the psychological ramifications of CAH, require care and concern from parents and family members. A chronic childhood illness is “an illness that can last for an extended period, at least months, often for life,

and cannot be cured" (Kenny, 1994, p. 1). CAH falls into this category because it is a congenital disorder with continuous care needs and there is no cure. Therefore, examining the literature on chronic childhood illness as a whole is necessary in order to gain an understanding of the ramifications that such an illness has on a family system and individual family members.

Effects on the Child

While the known physical and psychological effects of CAH have been discussed earlier, the literature does not give a full picture of what life is like for children with this disorder on a day-to-day basis. There has been a great deal of research on other chronic childhood disorders, however, and examining this research so as to form a more cohesive picture of the psychological and social ramifications of chronic childhood disorders is essential.

Research has examined the effects of chronic childhood medical disorders on the psychological well being of the child. Several studies show that children with a chronic medical disorder are more at risk for behavioral problems, emotional problems, or problems at school as compared to children without a chronic disorder (Goldberg et al,, 1997; Pless & Nolan, 1991; Silver, Westbrook, & Stein, 1998). This can even be seen in children with minor chronic disorders. For example, Dennis, Rostill, Reed and Gill (2006) found that, compared to the general population, children with atopic eczema had increased levels of internalizing behaviors.

A typically developing child does not have the same concerns and anxieties as a child with a chronic disorder. Additionally, a noticeable disorder such as CAH, where a child is required to wear a medical alert bracelet, and may have a different external appearance than their peers, increases the anxiety around what others may think. Harbeck-Weber, Fisher and Dittner (2003) highlighted this fact, reporting that this anxiety "has implications for the development of a maladaptive cycle characterized by less social interaction, increased peer perceptions of

withdrawal, and fewer opportunities to learn and practice skills related to making and keeping friends" (p. 109). A chronic and noticeable disorder may disrupt or complicate normal social development because of this anxiety. The above authors point out that ill children often are anxious about how their peers will react to them and may therefore not involve themselves in peer relationships with the same intensity or comfort level as they would if they were healthy.

The environment of secrecy that typically surrounds disorders of sex development may continue to lead to withdrawal and lowered self-esteem in children with CAH. Varni, Rubenfeld, Talbot, and Setoguchi (1989) examined variables that predicted self-esteem in children with limb deformities. The authors found that children's perceptions of classmates' social support was the variable that most highly predicted self-esteem in children with deformities. Children who perceived their peers as supportive adapted better than children who perceived their peers to be ostracizing. Children who hid their differences from their peers may be less likely to feel supported by those peers (Varni, et al., 1989). Aditionally, Varjas et al., (2008) found that children with a sexual minority status are more likely to be physically or verbally bullied in their schools. A child who experiences such status due to the virilization effects of CAH may have fear and anxiety surrounding the possibility of peers becoming aware of their differences.

The presence of a chronic childhood illness may not only limit a child's ability to interact with peers, but may result in more negative interactions as well. Negative peer interactions over time can affect many aspects of a child's future life, such as their sense of self, self-confidence, self-competence, and self-esteem. In sum, a chronic childhood illness not only affects children physically, but also socially, both of which can have huge ramifications on their psychological health.

Effects on the Sibling of an Ill Child

In a meta-analysis of published research, Vermaes, van Susante, and van Bakel (2012, p. 172), found that siblings of a child with a chronic illness are likely to have "more internalizing problems and less positive self-attributes" when compared to peers without a chronically ill sibling. Their review of relevant research found that older siblings and siblings of children with potentially fatal chronic illness were more likely to be affected negatively. In a family with a child with a chronic illness, it is likely that the typically developing children will have less of the parents' attention due to the heightened needs of the ill child. This analysis suggested that these children are more at risk for internalized difficulties due to the parents need to focus on the ill child in the family. The researchers hypothesized that siblings may be more likely to suppress their emotions, and less likely to go to their parents for help with difficulties, as they may have found this to be unsuccessful in the past due to parents needing to spend more time caring for an ill sibling.

Effects on the Parents

There has been much research that has examined the effects of parenting a child with a chronic illness. While not every parent will be negatively effected, the general findings of this body of research shows that parenting a child with a chronic illness increases the likelihood of mental health problems in parents. Parents with chronically ill children are more likely to experience depression and marital discord as compared to parents without a chronically ill child (Quittner et al., 1998). Additionally, Cadman, Rosenbaum, Boyce and Oxford (1991) stated that, compared to parents of healthy children, parents with a chronically ill child were significantly more likely to be involved with some form of mental health treatment.

A child with CAH may have visible physical differences, though these may only be seen by those close to the child as they often involve genital differences. A study of parents of children with upper limb differences found that worry about relationships was common for these parents (Murray, Kelley-Soderholm, & Murray, 2007). These parents often worried about their child's relationships with siblings and extended families. Additionally, parents often worried about how their relationships with their partners may be affected by these differences.

Some research has explored the idea that mothers are more adversely affected by a child's illness than fathers. Svavarsdottir and Rayens (2005), in their study of families with children with asthma, compared measures of well being, vitality and general health for mothers and fathers and found that mothers scored much lower in these areas. Generally, mothers are responsible for more day to day care of children and families, and as such, the majority of research studies have examined the effects on mothers rather than on fathers or parents in general. However, when speaking of disorders of sex development specifically, and ambiguous genitalia which may lead to a change in understanding of a child's sex, Slijper , Drop, Molenaar, & de Muinck Keizer-Schrama (1998) report that while it is difficult for parents in general, fathers more than mothers responded to a change by mourning the loss of the son they thought they were going to have. Research on this topic does tend more toward the mother's experience.

Because mothers traditionally tend to do more of the day-to-day work with children, and have the expected role of primary caretaker, the expectation of many families is that the mother will do more of the care and support of a chronically ill child. Due to this, there may be greater strain on mothers, as can be seen in Quittner, DiGirolamo, Michel and Eigen's (1992) study of mothers' depression and role strain. This study examined the stressors of doing hands-on care and coordination of providers on a daily basis. It may be that the initial stress of caring for an ill

child will diminish over time. Kovacs et al. (1985) looked at mothers of children with diabetes and found that their levels of stress related to caring for their ill children diminished as they became more competent in their ability to handle the illness. The stress of medical care likely is a large part of the stress of a chronically ill child.

Brody and Simmons (2007) explored the changes in stressors for fathers of children with cancer. They found that fathers went through several stages of change throughout the time of their child's illness. These changes both impacted and were impacted by the fathers' responsibilities to provide financially for the family. Additionally, they found that fathers often took on the role of communication with others, and as such felt that communication about diagnosis and treatment were especially important to fathers. They also explored the father's role in reassuring their child, which they felt tied into the importance of communication for fathers. When a parent has a child with a chronic illness, the roles they take may be different and this may account for the differences in levels and types of stress for mothers and fathers.

The parental relationship is also impacted by chronic childhood illness. Marital discord was found to be higher in couples with a chronically ill child than in those without a chronically ill child (Kenny, 1994). In a case study of two families, Pearce (2008) found that those parents were more likely to focus only on their child and the illness, to the exclusion of focusing on their relationship and social interaction. These families were less likely to socialize with others and risked becoming isolated and not experiencing typical family life.

Marital relationship is a key point of interest for many researchers, such as Berge, Patterson & Reuter (2006) who researched the relationships of parents of children with chronic illnesses. They found that mothers and fathers had different responses to the adjustments that resulted from their child's illness. Fathers, according to their study, felt satisfaction or

dissatisfaction in their relationship, not as a product of the changes as a result of the illness, but as a continuation of the state in which the relationship had been prior to the illness of the child. If a father perceived his marital relationship to have been strong prior to the illness, he continued to feel that it was a strong relationship. If there were weaknesses in his relationship, he continued to view these weaknesses and lessened satisfaction throughout the course of the child's illness. Additionally, the research showed that father's rates of depression were related to this sense of satisfaction in their relationship, and the strength and closeness of the relationship prior to the illness. Mothers, on the other hand, related their sense of marital satisfaction to their own perception of how the child's illness had impacted the family. A mother who was more stressed by her child's illness was less likely to feel satisfaction in her relationship and more likely to show symptoms of depression.

Lavee & Mey-Dan (2003) explored the impact of adult's relationships by interviewing couples with children who were being treated for cancer. They explored ten facets of these relationships and found several negative consequences of childhood illness on the marriage. These parents reported that their sexual relationship had changed, and mothers reported that their relationships with extended family was negatively affected. However, they noted that fathers felt that their relationships with extended family were positively influenced. The researchers noted that couples reported positive changes in some areas of the relationship, for example, communication, conflict resolution and trust, but these positive changes did not seem to continue over time. Families with a child who had been ill for less than a year noted positive changes in their marital relationship, but families with children who had been ill for more than three years showed fewer positive changes, and families with children ill more than four years showed none

of these positive changes. Thus, while the strain of a child's illness may influence a relationship in the positive direction initially, this change does not continue over time.

For parents of children with CAH and other disorders that may cause ambiguous genitalia, research has shown that they are likely to feel shame and guilt over their child's disorder, and most especially over the ambiguity of their child's physical sex and gender. In interviews of these parents, Warne (1992) found that these parents often went through a period of grief and bereavement such as might be expected when one loses a child. These parents also feel "bewilderment about the medical implications of the child's condition" (p.245), and they may feel confused, threatened, or angry about the implication of a genetic origin to the disorder. In the immediate time period following diagnosis and determination of ambiguous genitalia, parents were found to respond with "shock, grief, anger, and shame and in the mothers … with guilt" (Slijper et al., 2000, p. 9). Another study of parental response found that parents of girls with CAH and overmasculinized genitalia were less likely to feel symptoms of depression than parents of male children with undermasculinized genitalia (Wolfe-Christensen et al., 2012).

Effects on the Family System as a Whole

When a child has a chronic illness, the entire family is affected. While the individual members are affected, and the parents' relationship with each other is affected, the system of the family as a whole is affected as well. Day to day family functioning may be required to change, especially in cases where a child needs to take medication at precise times of day. These areas of functioning may include daily routines, school performance, sibling relationships, parenting methods, career choices, financial decisions, transitions from one stage of development to the next, and friendships within and outside the family (Stein, 1983). In a study of insulin-dependent children with diabetes, Seiffage-Krenke (1998) followed adolescents for four years. The author

found multiple changes within the family systems over time. It was reported that organization and structure increased in these families over time and that they became less stimulating and warm. When a child in a family requires daily medication, and at a certain time of day, as is the case with some children with CAH, it seems likely that an increase in structure and organization would be likely. This increase in structure may make it easier to maintain the medical care necessary to keep stress levels at bay for these children. Wamboldt and Wamboldt (2000), however, found that an increase in structure and rigidity of organization may cause a decrease in a family's ability to be spontaneous and warm toward other family members. Structure and organization are necessary for the medical health of the child, however, spontaneity and warmth are necessary in a family as well. A family dealing with a child with a chronic illness must constantly renegotiate and work to find the balance of medical care for the child as well as the other aspects of family life that require continuous care and effort. The variables that may assist with this process will be important to examine and learn about in order to help the family adjust and maintain a healthy balance of medical care and family life.

Importance of Including the Family in Treatment

Health-related quality of life, the "physical, psychological, social, cognitive, functional and behavioral dimensions of well-being and functioning as perceived by the person concerned" (Brutt et al., 2009), can be impacted by both family functioning and social experience. As such, it is important for the family to be cohesive and to adjust to an illness such as CAH. Studies have shown that in a family with a child with a chronic illness it is not the illness itself or the severity that determines how a child and the family will adjust to the illness, but rather it is the health of the family system (Bennet, 1994; Lavigne & Faire-Routman, 1993). Varni et al. (1989) explored the self-esteem of children with limb deformities and found that the children

with more highly organized families had higher levels of self-esteem. Aditionally, life stressors of family members were found by both Bennet (1994) and Labvigne and Faire-Routman (1993) to be important, along with variables of the parents, in how the illness affected the child. Children with atopic eczema were found to be more well-adjusted when they reported that their family emphasized independence and organization (Gil et al., 1987). In another study of children with atopic eczema, Dennis et al. (2006) found that children with this illness were better able to adapt to the disorder when their family was supportive and had more positive psychological health themselves. These factors had more impact than the medical course of the disorder. Other studies have shown that family functioning (Kenny, 1994), and the support of family members (Martire, 2005) play an essential role in a child's adjustment to and acceptance of a chronic illness.

Interpersonal relationships within the family can affect this adjustment. Bristol, Gallagher and Schopler (1988) reported that the parental relationship was key in the adaptation to a child's illness. The couples who reported feeling supported by their partners also reported less difficulty in adapting to their child's illness as compared to couples who reported not feeling supported by their partners. Families with fathers who were rated as high in involvement with their families also reported better maternal adjustment as well as higher engagement by both parents, better functioning as a whole family, and less negative impact on the family by the illness (Gavin & Wysocki, 2006). Additionally, social support seems to be a protective factor for parents. Kazak et al. (1997) found that parents with greater perceived social support were less likely to rate themselves as having psychological difficulties. Finally, Martire, Lustig, Schultz, Miller and Helgeson (2004) reported in a review of literature that interventions that involved all family

members had a positive impact on the ill child and the entire family, decreasing depression and anxiety in all family members involved.

The well being and health related quality of life of a child with a chronic illness is clearly impacted by the entire family. Research has shown that the functioning of the family system, as well as the relationships within the family both have a clear impact on the ill child. When treating a child with a chronic illness, it is beneficial to all involved to treat the entire family in order to increase the benefits to all family members.

It is important for children and their families to have peer support in accepting and dealing with a chronic illness. In a statement of best practice for the management of Disorders of Sex Development, Nihoul-Fékété (2008) discussed the need for early establishment of this peer support. When dealing with a disorder that is well known, it is much easier to adjust and have support than when dealing with a disorder in which little is known and carries a stigma of sexual anomaly. "Peer support ends the isolation and stigma; support groups can help the family and contribute to finding the best quality care; children who form relationships with peers and other affected adults early in their lives benefit from a feeling of normalcy early on, well before adolescence" (Nihoul-Fékété, 2008). Offering support and connection to parents and children early in life is the best way to help ensure the continuation of these attachments and support throughout life.

Family Systems-Illness Model

A child's chronic illness clearly can have a large impact on the entire family. The family system as a whole is impacted by the illness, which can, in turn, impact the child's prognosis and quality of life. A systemic approach to treatment, involving the entire family, would therefore be a logical intervention to improve the quality of the family system as well as the prognosis of the

child and the quality of life of the child. In his book, *Families, Illness, and Disability: An Integrative Treatment Model*, John Rolland (1994) does just this as he discusses the systemic perspective approach to treatment of chronic illness. He takes into account multiple systemic factors that can change and influence the way a family copes with and adapts to the physical illness of a family member.

Familial Structure and Communication

Rolland (1994) believed that clinicians who are working with children ought to have a good understanding of family structure in order to understand each family's functioning with illness. He explored the basis of family life in order to really see how a family is functioning at any given time. He believed that family organization and structure as well as how a family communicates, would give those working with the family a greater understanding of that particular family's level of functioning.

At the outset, Rolland (1994) believed that one must understand how a family is structured and organized. In order to examine this, he explored cohesion and adaptability as two of the most important aspects of structure and organization. Adaptability is defined as the "extent to which the family system is flexible and has the ability to change" (Maynard & Olsen, 1987, p. 502). Adaptability is essential to a family system in order for those within the family to be able to be flexible or make changes to things like leadership style, relationship rules and roles within the relationship, especially in times of stress which demands such changes (Olsen, 2000).

Another important factor to family functioning is that of communication within the family. According to Rolland, there are two specific ways that communication is important when dealing with a chronic illness. There is the first and foremost importance of the ability to communicate with medical staff and to understand medications, dosages, and response to illness

or injury. Secondly, it is important that the members of the family be able to communicate about their feelings and with each other, as families who are able to turn to each other for support fare better in times of stress (Olsen, 2000; Olsen & Gorall, 2003).

Another important factor in the structure and makeup of the family is the multigenerational experiences that a family or each individual member has had with illness and loss. All families have had loss experiences, and this can affect the way that each member as well as the system as a whole responds to this particular illness and child. Rolland suggested that one way to learn about each family would be for a clinician to make a genogram with the family in order to explore past loss, illness or stress experiences with the family system. This can be helpful in determining past responses, but also in exploring current stressors and functioning of the family.

Not to be forgotten, Rolland (1999) believed that worldview and belief systems are essential information when working with anyone. Illness can bring out various beliefs about different aspects of life and family members may share beliefs or have differing beliefs. In order for a clinician to work with a family, that clinician should have a simple understanding of the family's belief system. This may include religious beliefs, beliefs about death, perceptions of illness in general, or beliefs about this specific illness. Additionally, CAH is a genetically inherited condition and each family's beliefs and knowledge about the origins of the disorder may be helpful for understanding the family as well.

Psychosocial Typology of Illness

Once a clinician has explored a family's communication and organization with regard to the illness, they can better develop appropriate interventions to assist the family in functioning in a healthier manner. Rolland's (1994) discussion of families and illnesses, however, does

acknowledge that different illnesses require different forms of organization. He developed a system of categorizing an illness to help determine what would be most useful for a family at that particular time. These variables are onset, course, outcome, and incapacitation; and will be discussed in relation to CAH.

The onset stage of an illness refers to the way the illness first manifested symptoms. This is not a reference to the biological development of the illness, but rather the way the individual or the family first came to understand and know about the illness. This may be either a gradual development, or an acute realization of illness. In the case of CAH, a newborn with acute salt-wasting CAH with ambiguous genitalia most likely would have had an acute onset as the family would likely have become aware of the child's disorder at the time of birth. While there may have been a short period of time where the medical providers worked to determine what exactly was happening for the child, this time was probably as little as a few hours or days. For some children with CAH, however, the development of outwardly apparent symptoms may have happened over the course of weeks, months, or years, depending on the type, severity and presentation of their particular form of CAH. A male child with typically developed genitalia may go home from the hospital at birth with no suggestion of any disorder. Over the next month or two he may have difficulty feeding, sleeping, and a failure to grow that can be of unknown origin for some time. He would be at danger of adrenal crisis, shock, and even death at any moment until he is found to have CAH and treated properly. Additionally, some children who do not have salt-wasting CAH may develop relatively normally for the first several years of life and it may not be until they begin to go through puberty at five or six that parents and doctors begin to wonder if something is wrong. Thus it is important for a clinician to discuss the onset of CAH with the individual and the family as there are many different ways that CAH can present.

After the onset of the illness, the illness takes one of three courses: (a) progressive, (b) constant, or (c) relapsing/episodic. In the case of a progressive illness, the person will always have symptoms and these symptoms will get worse over time. A constant course illness will have some sort of event that marks the beginning of the illness, however following this event the symptoms will stabilize and not get better or worse over time. An episodic or relapsing illness is characterized by relative stability over time and the possibility of an absence of symptoms combined with periods of symptom flare ups. CAH can be categorized as constant, or episodic in nature. Some individuals with CAH will be at risk for adrenal crisis, so while they may be stable day to day with medication, there will always be the risk that they could have a poor reaction to stress or illness and they will need an extra dose of medication as a result. This would be considered an episodic/relapsing illness. Others with CAH are not at risk for adrenal crisis and, while their understanding of the disorder and the impact it may have on their life will change, the progression of the disorder will be stable and constant over time.

A family's ability to adjust to the disorder can be impacted a great deal by the course of disorder. Those families with a child with the constant potential for adrenal crisis will constantly need to adapt to each relapse and stressor. These can come at any moment and are unpredictable and potentially fatal. A family whose child does not have salt-wasting CAH, on the other hand, will be able to adjust to the disorder and have the progression of the disorder remain fairly stable over time, allowing them to stabilize their own understanding and organization around the disorder. In addition to the hormonal aspect of CAH, children with atypical genitalia may also require surgical intervention, most likely several times throughout their lives. Each surgical event can be considered a relapse in the stress, weight, and importance of the illness in that family. Each presentation of the disorder brings about new stressors and new uncertainty for the

individual and the family. All aspects of cohesion, adaptability, and communication will be impacted by different courses and events in the life of the disorder.

The outcome of an illness refers to the fatality of the illness, or the potential for a shortened lifespan. Certain diseases that are known to end in death such as ALS, Huntington's disease or the like and categorized as fatal. Others, such as epilepsy do not result in death and are therefore non-fatal. The last category of illnesses are those that are less predictable in their course. Diseases such as heart disease could potentially end in a shortened life span or sudden death. CAH is usually non-fatal, though the severe salt-wasting form can be unpredictable, threaten death, and occasionally be fatal if not treated properly.

Illness can also be categorized by degree of incapacitation. While this could refer simply to physical incapacitation, Rolland (1994) expands the definition to include "mental functioning, sensation, movement energy production or the extent to which it causes disfigurement or other physical causes of social stigma" (p. 31). Level of incapacitation can vary from none, to mild, moderate, or severe. This format of thinking about illness emphasizes looking at the psychological and social impact of a disorder in addition to simple physical incapacitation. Even when a person with CAH is relatively healthy or symptom free, they may still have the knowledge that they have atypical genitalia and this could result in a much higher level of psychological or social incapacitation for them. Additionally, those with salt-wasting CAH may have to wear a med-alert bracelet and carry a stress dose of medicine with them at all times, reminding them that they could have a relapse or need medical attention at any moment. Because of these variables, CAH can vary along the continuum of incapacitation depending on each individual's perception of their illness.

According to Rolland (1994), illness can have four different variables of categorization. Each of these variables can have a different effect on the family system. An illness with a gradual onset and a constant course may require a simple adjustment for a family with a relatively stable continuation over time. By contrast, a manifestation of CAH with an acute onset and potential for relapse over a lifetime can require a family to make continuous and variable changes, potentially without warning, and over their child's lifetime. The levels of cohesion and adaptability required by a family to deal with the demands of the varying illness courses will be different for each family.

Phases of Illness

In addition to the typology of the illness, this model takes into consideration the phases of illnesses. Rolland (1999) explored three phases of illness: (a) the crisis phase, (b) the chronic phase and (c) the terminal phase. The initial phase is the *crisis* phase. This begins when symptoms are first noticed through to initial adjustment to illness. At this point in the illness, individuals, or families in the case of children, may have the knowledge that something is wrong, but they do not know yet what that is. This also can include knowing the diagnosis but not having much information about the illness yet, or understanding various facets of the disorder. At this stage, individuals or families typically feel that there is more that they need to know about this disorder and treatment. At this stage, the task is to find a doctor, determine a diagnosis, and learn as much as possible about symptoms, course, and treatment options. These tasks help individuals or families adjust to illness. At this point, families must also learn about triggers for symptoms, and ways of recognizing danger in the illness. For example, a family with a newborn with CAH will need to learn about signs of stress and what to do in case of fever or illness in their child. Families will also need to learn about how to deal with other family issues that may

occur simultaneous to the needs of the illness and child with CAH. This phase may take years, or only weeks, depending on the individual family.

Once the family moves through the initial phase and completes the tasks of that phase, they begin to move into the *chronic* phase of the disorder. This is "the time span between the initial diagnosis and readjustment and the terminal phase" (Rolland, 1994, p. 48). In this phase, the family has a good idea of what to expect, how to treat it, and the providers that will be there to assist. The family is able to "maintain the semblance of a normal life under the 'abnormal' conditions presented by a chronic illness and heightened uncertainty" (Rolland, 1994, p. 48). A family who was not expecting a chronic disorder must now figure out how to live in a new type of "normal." They must accept the disorder and the changes that they have had to make. This phase returns to the earlier mentioned course of the disorder, as it can be progressive, constant, or episodic. The family will have to learn to respond in different ways to each of these courses. An illness such as CAH will most likely follow a constant course that requires initial adjustments by the family, but maintains a form of status quo throughout the child's life. While there will be new challenges based upon the age and stage of the child's development, CAH is rarely fatal. A fatal or even life-threatening disorder would require a family to be prepared for, or to go through, the *terminal* phase, as defined by Rolland. This phase is simply when the death of the affected person is imminent and most likely to occur. At this time the family has to organize their lives around impending death and loss. The family must adjust from anticipating the loss, through the loss itself, and then through the grief that remains following the death of the individual. When CAH is fatal, it is likely to be a very fast death due to shock and adrenal crisis. This often is the case for very young infants or small children. Thus the terminal phase, with its advanced planning for death and loss, is not likely something that will apply to many families with a child

with CAH. The post-death needs, such as planning a funeral and telling family and friends about the loss would apply in the case of such a loss, however.

There are unique tasks for each phase of a chronic illness. A family must adapt and change as they go through each phase, and even several times within the phase, in order to function successfully. The structural makeup of the family may change; the organization of the family may change as well. A family may have various levels of adaptability and cohesion throughout each phase in order to adjust to the various demands and changes as the member with CAH grows and changes. In order to maintain a well-functioning family, communication is vital. A family must have effective communication in order to ease these transitions.

Psychoeducation

As a program designed to help families with an ill child work through the various phases of a disorder to increase family functioning, psychoeducation will be necessary as a format that will combine "elements of education, support and problem solving" (Pollio et al., 2006, p 31). Historically, there are a variety of formats for psychoeducation, such as individual meetings, frequent seminars, or weekly meetings over the course of several months (Campbell, 1997; Karamandidou et al., 2008; Kaslow, et al., 2000; Ng et al., 2008; Pollio et al., 2006; Tyerman & Booth, 2001). Some forms of psychoeducation include groups, which may include multiple family groups (Cykrowicz & Joiner Jr., 2007; Cummings, Faircloth, Mitchell, Cummings & Schermerhorn, 2008; Pollio et al., 2006; Weinstein, Chenkin & Faust, 1997). Psychoeducation is proposed to provide knowledge and skills for participants to make changes in their lives (Gearing, 2008). It differs from psychotherapy in this way. There are several formats for psychoeducation, some of which have been created to use manuals and prescribe a set method for conveying the information in the psychoeducation sessions. The general purpose of group-

facilitated psychoeducation has become to help participants form social support networks, to receive current information about the topic of the session, and to establish coping and problem solving skills (Gearing, 2008). Psychoeducation can be geared toward individuals or families, and can be a part of group or individual therapy.

In the last 20 years, psychoeducation has shifted from the original purpose, which was to decrease expressed emotion in families with a member suffering from schizophrenia (Solomon, Draine, Mannion & Meisel, 1997). More recently, psychoeducation has focused on many more topics. Solomon et al. noted that there has been a recent increase in family-based psychoeducation, generally known as family education. Psychoeducation and family education are very similar, with one main difference. Family education is often facilitated by someone who has a family member with the same struggles as those in the group. The focus of the group is often on coping with illness rather than making changes in behaviors and family dynamics. Trained mental health providers, on the other hand, facilitate Psychoeducation. Both modalities provide coping skills and education to families and those with the struggles on which the seminar focuses. They are different, however, in the expectation of change. Family education addresses coping with struggles, whereas psychoeducation addresses ways to change with regard to the struggles. The facilitators of these types of groups also differ.

Efficacy of Psychoeducation

Many researchers have studied psychoeducation and adult mental health issues. In an early study of psychoeducation, Hogarty et al. (1986) examined psychoeducation interventions used with families and individuals with schizophrenia. Their goal was to discover what could help prevent relapse, and so they studied 103 individuals who they randomly assigned to one of four different groups. The four groups were a social skills group, a family psychoeducation

group, a mixed social skills and family psychoeducation, and a control group. The participants in this study were residents of an inpatient mental health hospital. After one year, only fourteen percent of the patients in the family treatment condition experienced symptom relapse compared to 53% of patients in the social skills group without the family psychoeducation condition. There have been many studies of schizophrenia and treatment involving families, and these studies show that family treatment, including family psychoeducation and social support, increases symptom reduction, decreases rates of relapse, decreases hospitalization, and also increases the social support and interactions of family members (Falloon et al., 1985; Leff, Kuipers, Berkowitz, & Sturgeon, 1985; Schooler et al.,1997; Xiong et al., 1994). These studies show that while individual treatment and medication are helpful for individuals with schizophrenia, family treatment that includes education and social support were found to not only reduce negative effects of the disorder, but also to increase positive supports for the families. Schizophrenia is a severe and challenging disorder, and as psychoeducation and family support is beneficial in this population, it is likely that it would be an effective and useful treatment option for other less severe illnesses as well.

In addition to schizophrenia, psychoeducation has been found to be helpful for other populations as well. Colom et al. (2003) researched psychoeducation as a group treatment option for individuals with Bipolar Disorder. The participants in this study were in remission from their diagnosed Bipolar Disorder, meaning that they were not showing any active manic or depressive symptoms. This blind study with a randomized experimental and control groups provided the participants in both groups with medication and psychiatric care in addition to the weekly groups that the study offered. The control group met weekly with a psychologist without preplanned topics to discuss. The experimental grout met for weekly educational sessions designed to learn

about the illness, treatment compliance, early detection of symptoms and recurrences, and maintenance of a regular lifestyle. Because the control group also had a weekly support group, the study controlled for the potential effects of increased social support. The study continued for a year, and following the treatment the experimental group showed reduced rates of relapse, longer episodes of health and fewer hospitalizations than the group that was solely for social support.

More recently, the turn has been toward studying ways to increase medical treatment compliance. Karamanidou et al. (2008) explored the possibility of psychoeducation increasing the understanding of renal patients as to their medication's function. They also wanted to increase patients' awareness of their own beliefs about their medication and determine what would help them. The education in this study was a one session intervention using both written material and discussion as well as a demonstration. Also covered in this session was a discussion of incorrect beliefs of patients about their medication. The goal of the study was to change the behavior of the patients (i.e., taking medication as prescribed). In order to determine the efficacy of the intervention, the researchers surveyed the participants both before and after the session, as well as four months later. They found that those patients who had participated in the educational session had better knowledge of their medication than those who had not. They were more effective at balancing their diet and medication, and their beliefs about their medication continued to show positive change after the educational session. This suggests that a one time educational session can have lasting effects.

Another medical treatment study was carried out by Weinstein et al. (1997). This case study of one family examined the use of family psychoeducation to treat a child with asthma. Over a two week period, the authors met with the family several times in an attempt to explore

the efficacy of an educational intervention for a child with asthma as well as severe behavioral problems. The child in this study had very severe asthma and needed to be hospitalized several times a year. In their program, the authors attempted to teach the family how to control asthma in order to prevent further attacks. The educational component of the intervention included information about asthma medication, the use of medication prior to discipline, and the exploration of family dynamics that could be affecting the child, both in medical and behavioral ways. The case study showed that the parents learned techniques that continued to be beneficial even a year after the intervention. The child's asthma attacks decreased in number and severity, and behavioral outbursts were lessened as well. Thus is appears that a family dealing with a chronic childhood illness can be helped by psychoeducational intervention.

These studies show that psychoeducation within a group can be beneficial for both families and patients. Information about disorders can be increased through the use of these sessions. Additionally, families can increase their knowledge about dealing with chronically ill family members on a daily basis. Psychoeducation is an effective form of treatment for both individuals and families.

Multi-Family Psychoeducation

Multi-family psychoeducation combines multiple families in one session of psychoeducation. In this way, the intervention provides an immediate source of social support for participants and they can learn from each other's experiences. Tyerman and Booth (2001) showed how this would happen in a study they ran, showing their multi-family group psychoeducation for families with a member struggling with a traumatic brain injury. The families and facilitators met bimonthly for educational workshops. The findings from this study

were that participants felt they learned from the facilitators as well as by talk "with others in a similar situation" (p. 62). They also reported feeling that they were not alone in their experience.

Short-Term Psychoeducation

Psychoeducation is an effective intervention that can be used to assist family members who are dealing with a chronically ill child. While studies have shown this, there has been little research done into the optimal or most effective length of intervention. As stated previously, Karamanidou et al. (2008) found that a simple one-hour session was effective in helping patients on kidney medicine.

Pollio et al. (2006) ran a one day psychoeducational seminar for families of adults with severe and persistent mental illness. This study shows the effectiveness of a short-term seminar. Following the one-day seminar, family members reported an increase in their knowledge of their family members' mental illness. They also reported an increase in their ability to cope with the mental illness, an increase in feelings of control, and a decrease in levels of guilt.

Short- term psychoeducation has been shown to be helpful for parents and children adjusting to a change. Frieman, Garon and Garon (2000) conducted research with divorced parents to help them assist their children in adjusting to the divorce. They conducted an educational seminar over two three-hour sessions held over two weeks. They collected data before the seminars, after, and at six months and 1 year after the follow-up. They examined the parents' ability to parent effectively following their divorce. The intervention focused especially on helping parents learn to work together with the child's other parent. The parents in the seminar continued to report assistance and knowledge gained from the program one year later. They reported more confidence and ability to parent together even at the one-year post-test.

Short-term psychoeducation can be very helpful for families and have long lasting benefits. Psychoeducation with family members requires the coordination of many schedules and different people. As such, it is important that facilitators are able to convey the information in a short, succinct, and clear presentation that is time and situation sensitive. This allows for more families to participate and benefit from the intervention.

Format of Psychoeducation

Facilitators. The facilitators of a group can have a large impact on the outcome and effectiveness of the group (Zimmerman, Heinrichs, & Baucom, 2007). A meta-analysis by Zimmerman et al. (2007) suggests that when leading a psychoeducation group using Cognitive Behavioral Therapy, the effect size was higher when the facilitator was a psychologist. Conversely, in-group interventions for women with breast cancer, the efficacy of the group was greater when the facilitator was a medical professional. While there have not been any studies studying directly at the effects of different types of facilitators on the efficacy of psychoeducation, it appears from this meta-analysis that facilitators' training and background can correlate with outcomes, and that facilitators should be knowledgeable about the content of the psychoeducation as well as the techniques that are being used within the sessions.

Techniques. Psychoeducational interventions can use a variety of techniques. Often facilitators will use CBT techniques to teach coping skills or problem solving. Additionally, many of the aforementioned studies used family therapy within the interventions as a method of changing the day-to-day life of a family. As with any form of therapeutic intervention, techniques from many areas of psychology can be beneficial as long as they follow the tenets of psychoeducational intervention to increase knowledge, promote coping skills, problem solving, and affecting change.

When considering creating an intervention for families with children with CAH, it is important to think about the specific needs of the families as well as the children. As CAH is a disorder that is likely to remain in the chronic phase of illness, a child and a family with CAH will be maintaining a fairly normal life alongside the disorder, as long as the child is able to see themselves as a typically developing individual as well as being an individual who has a chronic congenital disorder. In order to develop a sense of meaning in their world, the use of narrative therapy, as developed by Michael White and David Epston (1989), could help both the family and the individual to separate the child from the disorder. Narrative therapy uses language to determine meaning from experience (Cashin, 2008). The context of a person or family's worldview can add or subtract from a person's individual sense of difficulty with their disorder.

For example, in the Dominican Republic, there is a village so remote that inhabitants rarely leave the village. In this village two percent of the population were found to have a rare genetic mutation that led to another Disorder of Sex Development, 5-alpha reductase (Cai et al., 1996). With this mutation, the male fetus is unable to produce a form of testosterone known as DHT. Because of this, the male fetus is born appearing female and remains so until the testosterone surge of puberty. At this time, the body develops into a typical male body and the adult is confirmed to be male (Sanghavi, 2008). Because such a large portion of the population in this village had the same disorder, the village accepted that some of their little girls grow up to be men. It is not seen as a strange occurrence (Wooten-Blanks, 2012). This, however, is not the typical expectation for children in the rest of the world. As such, a child with CAH, whose genitals are likely to be atypical, and whose attitudes and behaviors may be different from other children will have to come to their own understanding of their personal form of the disorder and their life.

Narrative therapy can be beneficial for these children and families, because it does not see the child as having something wrong with them, but rather introduces the idea that a child creates a story about themselves and the world around them in order to adapt and live in the world around them (Cashin, 2008). By changing a person's story and understanding of themselves and the world around them, the child and the family are able to look at their situation differently and ideally with a better understanding of the choices that they have in shaping their own life stories (Weber, Davis, & McPhie, 2006).

In narrative therapy, the technique of externalizing the problem is often used. Changing the language that a person may use when discussing themselves and their disorder allows this to occur. This will include the parents, in the case of the children participating in this intervention. To externalize CAH, and separate it from the child, the facilitator may ask questions about CAH as though it is a separate entity. Once the character of CAH has been created, the facilitator and the family can work together to discuss how CAH changes and influences the lives of the members of the family (Weber et al., 2006). For example, a child with CAH may feel as though they are physically different from other children. The facilitator can help them learn to discuss what it is like when "CAH" tries to convince them that they are different, or when other people notice their difference and tease or exclude them. The facilitator can help the child and the family to think of CAH as having its own voice, that can be silenced. In this way, CAH becomes something that is separate from the child; it becomes its own entity.

To assist the process of separating CAH from the child, narrative therapists have used artwork to strengthen the understanding and belief in a separation between the child and CAH (Keeling & Bermudez, 2006). In a study by Keeling and Bermudez, the therapists used sculpture as an intervention. They found "externalization of … problems through tangible objectification

(sculpture) to be helpful… and continual interaction with the sculpted figures… appeared to have compelled participants to deal with problems rather than to avoid them" (p. 415). By creating and continuing to interact with a sculpture, the patients were able to strengthen their understanding of the separation and continue with that narrative. In the case of a disorder such as CAH, which causes such severe physical differences in a child, this intervention could be a helpful way to build a sense of self for a child, which does not solely revolve around the disorder.

In addition to an individual sculpture of CAH, a family participating in this intervention may benefit from a form of therapy known as family sculpture (Woods & Martin, 1984). This form of experiential psychology, originally created by Virginia Satir, focuses on the here and now of the moment, exploring action and interaction as the most important form of expression, rather than thought and feeling (Costa, 1991; Hernandez, 1998). Because this intervention will include potentially young children, a form of expression that does not include verbal expression may help some of the younger members of a family to be included. By using an indirect method of expression, the family may learn more about each other that cannot be said with words and discussion (Kaye, Dichter, & Keith, 1986). This exploration of family dynamics does not include discussion as the primary goal, but rather action and physical expression of beliefs and feelings.

In this particular form of intervention, the family participating would create a small scene of the family, placing individuals in settings and situations that mimic their beliefs and expectations about their particular family. For example, a family with parents who frequently argue or do not get along may place the parents in separate areas of the scene, or looking away from each other. The goal is to create a scene that represents the relationships within the family.

The family may choose if they want to include multiple generations of family members, or whomever they believe constitutes their family. In this way, they will be better able to focus on themes and patterns within the family rather than on the individual and particular events or occurrences (Lesage-Higgins (1999). Participants can also use inanimate objects to represent things that they feel have an influence on the family such as the sculpture of CAH, or money to represent financial difficulties, etc. This family sculpture can represent the present moment of the family rather than the thoughts and feelings that may have become the family's story.

Size of Intervention

A psychoeducational intervention can be of any size. The studies discussed above ranged in size from one person to groups that were intended to be open to any size (Frieman et al., 2000; Griffiths & Barker-Collo, 2008; Karamanidou et al., 2008; Kaslow et al., 2000; Tyerman & Booth, 2001).

From comparisons of other studies, it has been found that there is an ideal size of group, a group must be large enough, but not too large. In a multi-family psychoeducational group, for example, there must be enough families to allow for open debate and to increase the likelihood that families will find other families with similarities. Size is also important because the facilitator does not want the group to feel too large and prevent the families from taking in the material as presented. It is important for the facilitator to carefully consider the type of activities and structure the design of interventions so as to find the best number of participants for the session.

Chapter 3: Methodology

Research Design

The current study consists of a review of the literature for the purpose of developing a new treatment intervention for families in which a child has been diagnosed with Congenital Adrenal Hyperplasia. I reviewed the following topics: congenital adrenal hyperplasia, the psychological impact of CAH, the impact of childhood chronic illness, the Family-Systems Illness Model, and psychoeducation. I used several databases; these were PsychInfo, PsycArticles, Medline, PubMed and the Gender Studies Database. Several on-line websites were also used in data collection. These websites were, www.nadf.us, http://www.congenitaladrenalhyperplasia.org/, http://caresfoundation.org.

I used the following search terms: congenital adrenal hyperplasia, CAH, disorder of sex development, adrenal crisis, illness, chronic illness, chronic sorrow, ambiguous genitalia, intersex, children, family, family dynamics, family functioning, Family-System Illness Model, cohesion, adaptability, communication, psychoeducation, multi-family psychoeducation, narrative therapy, externalization, experiential family therapy, family sculpting, and family sculpture.

I analyzed and organized the articles for potential applicability, relevance, and limitations. This resulted in the literature review (Chapter 2), which in turn led to the design of a theory of usefulness and the basis of understanding I needed to begin structuring a psychoeducational social seminar for families with children with congenital adrenal hyperplasia.

I created a questionnaire to assess the Phase of Disorder, based on the model created by Rolland (1994); see the questionnaire in Appendix B. In the Family-Systems Illness Model, there is no set time for each phase of a disorder. Each family will process and move through the

phases in their own time. Therefore, a method of exploring a family's current phase of disorder was needed. When a family moves from the crisis phase to the chronic phase, there is an increase in stability and a decrease of tension. One goal of this program was to identify a family's current phase in order to assist them in understanding their current situation and move toward a more stable phase. In order to move to the chronic phase, a family must complete certain goals laid out by Rolland. The adapted questionnaire asked families if they feel they have met these goals, such as comfort with medication administration, identifying a trusted doctor, and learning to recognize signs of adrenal stress in their child. The use of this questionnaire was intended for parents and caregivers of children with CAH to help them determine which stage their family was in while also helping to clarify the areas on which they may need to focus more time and attention.

Two lists of stress reducers were developed for the seminar. One list was made for children and adolescents (Appendix D). The other was created for adult participants (Appendix C). These lists, which would be posted on the wall, provide a variety of activities to remind each group what they can do to help to keep them calm or increase positive feelings in times of stress. The educational element of the seminar consists of a presentation of information on family functioning, the course and phases of CAH, and how CAH can affect a family system. The presentation educates families on the various phases of living with a child with a chronic disorder with the goal of helping families recognize and identify these phases as they come and go.

Evaluation of the Seminar

One goal of this seminar is to increase the knowledge that parents have of their child's illness. In order to address this question, participants will be asked to complete pre- and

post-seminar questionnaires. After registering for the seminar, parents will be sent the pre-seminar questionnaires to complete ahead of the day of the seminar. Parent participants will be asked to fill out the Congenital Adrenal Hyperplasia Knowledge Assessment Questionnaire (CAHKAQ). The CAHKAQ is a small, little known measure, without the strong backup of measures of external validity. However, the originators of this measure were able to test it for internal consistency and content validity using a Dephi technique with three rounds of evaluation by an expert panel leading to unanimous agreement upon 80% of initial items, which led to the 22 final questions (King et al., 2007). The Chronbach coefficient alpha for the CAHKAQ was found to be 0.67, with notation that removal of three particular items would have increased this coefficient to 0.70, however the developers decided that these three items contained necessary knowledge for families such that they determined a lower Chronbach alpha to be acceptable (King et al., 2007). It has been used by facilitators of educational seminars for parents (Schaeffer et al., 2010) to explore the efficacy of their seminar and conclude which parents may be in need of additional supports.

Another aim of this seminar is to change the dynamics of a family structure into a more cohesive, stable one. To assess whether this occurs over the course of the seminar, parents will be given the Family Adaptability and Cohesion Evaluation Scales-IV (FACES-IV). The FACES-IV is a moderately long, though easily completed measure that explores cohesion and adaptability within family structure. As these aspects of a family can have great impact on how a child copes with illness, they are important to explore. The FACES-IV is both valid and reliable and has been shown to distinguish clinically at risk populations (Olsen, 2011). Because of the length of time that this measure can take, parents will be asked to complete it at home prior to

attending the seminar as well as one week and three months after the seminar to measure for change and stability over time.

Parents will also be asked, at the beginning of the seminar day, to fill out the Coping Skills Questionnaire, which they will then be asked to fill out again with the post-seminar questionnaires that will be mailed to them at home.

As one demonstration of the seminar's effectiveness, parent participants should have higher scores on this questionnaire after the seminar than they did before the seminar. The questionnaires will be analyzed using a repeated measures t-test method comparing pre-seminar scores with post-seminar scores for each individual and family.

Consumer satisfaction is an additionally important area to examine. It is essential that the seminar be useful to the participants and that they feel that they have learned something from it. It is also important to make sure that the seminar flows easily and is engaging for families. A feedback questionnaire following the first presentation of this seminar will be helpful in determining the ease, engagement, and efficacy of the program.

Over time, the seminar will need to continually be updated in small ways to keep the information presented as up to date as possible. A feedback questionnaire following each presentation will help the facilitators to know what is useful and what needs strengthening to improve the program. Additionally, over time there may be medical advances in treatment that will need to be assessed and incorporated into the seminar. The impact of family systems and dynamics, quality of life, and the impact of culture on CAH and families will also be an area of continued growth and change for facilitators and families. Keeping up with these changes will allow the seminar to remain relevant and useful for families and providers alike.

A feedback questionnaire was designed in order to help assess the effectiveness of the seminar (Appendix E). This questionnaire would be given to participants at the end of the seminar in order to gather their impressions of how well the seminar was run and how closely it met their needs.

Chapter 4: Results

A Psychoeducational Seminar for Families Dealing with Congenital Adrenal Hyperplasia

The purpose of this dissertation was to design a clinical intervention that would help families and children with CAH adjust to this illness and the potential changes in their lives as a result. As the literature review showed, a child's adjustment to chronic illness and their quality of life can be greatly affected by the health of their family system. For that reason, this intervention would include all of the members of a family in which one or more children have CAH. The seminar includes a variety of activities and learning opportunities, including community and social support establishment, skill building, education about CAH and family dynamcs, and a chance to think about CAH in new ways.

Seminar Activities

The seminar would include several modules with a variety of activities and learning opportunities. It would begin with psychoeducation on the impact of CAH on both physical and behavioral health, introducing the idea of the family as a system within which each member impacts the others, and these interactions may impact health. The seminar will also introduce the idea of CAH as its own entity, using a narrative therapy tool to externalize the disease from the child. Following these discussions, families will, together, explore the roles and perceptions of those within the family with the goal of increasing communication within the family. The seminar additionally would provide the opportunity for families to connect with others who understand their experiences. This chapter provides the framework and design of the seminar.

Psychoeducation

The literature review discussed the efficacy of psychoeducation as a valuable tool to assist families coping with illness. As CAH has impacts on both physical and behavioral health, psychoeducation has been shown to be valid tool to provide families with the knowledge and skills needed to help their family adjust to illness (Barlow & Ellard, 2004). According to the Family Systems-Illness Model, a family must adjust their levels of communication, cohesion, and adaptability in order to fully adjust to the presence of a chronic illness in their lives. Because psychoeducation offers strategies and information, it should be an ideal intervention for these families.

Psychoeducation is not just a means of providing information. Facilitators work with families to give them the information about problem-solving and copings skills, but also give them opportunities to practice these skills and utilize them in a safe environment in order to increase the likelihood that they will be able to continue the use of these skills in every day life. Psychoeducation also keeps the goal in mind of enabling families to increase their problem solving skills and ways of coping. These are both essential skills needed by families when dealing with a child with CAH. As CAH may potentially have a negative impact on the child with the disorder, as well as the entire family, it would be essential for the families to increase their coping skills and problem solving to adapt to their child's unique presentation and struggles. The psychoeducational design for this seminar should allow parents and children an opportunity to learn positive ways to deal with stressors and the unexpected.

Skills Development

In addition to learning skills to cope with the unexpected stressors of CAH itself, the seminar would equip participants with problem-solving and coping skills to deal with potential

negative emotions and situations that may arise. Research indicates that the emotional and social ramifications of CAH, or any chronic childhood illness, may have a negative impact on all family members. As such, it will be necessary for each family to have problem-solving skills and coping mechanisms in place to manage these possibilities. This seminar would include training in problem-solving as well as coping skills, geared toward the individual needs of each participating family unit.

Social Support Networking

Finally, the seminar would bring together families and allow them to begin to establish a social support network of other families that have had similar experiences. As shown in the review of literature, social support helps individuals and families cope with chronic childhood illness. As this seminar would include many families, this would allow them to connect with and support other families with children with CAH. Because CAH is not a common disorder, and has a history and continued aspect of secrecy, many families may not have met other families who are dealing with CAH. While housed within the national MAGIC conference, there would be many families attending the seminar that may be dealing with other disorders in their families. This seminar would give families the opportunity to identify other families who were also learning how to live with CAH.

Format of the Seminar

Participants

This seminar is designed for families with children from infancy up to age 12 who have been diagnosed with CAH, though the break out groups are for children ages 8 and up (siblings may be older than 12). Families with younger children would be able to participate in the seminar, however their children would participate in group childcare on site, provided by the

organizers of this seminar. The rationale for this decision was that children under age 8 might not be able to separate from their parents, follow instructions, and participate in group activities in a way that is a meaningful and a productive use of time. However, families with children under 8 are most likely to need assistance with the early stages of dealing with CAH and understanding the disorder as well as feeling connected to social supports. Research suggests that cohesion and adaptability, two main variables that affect family functioning and therefore a large focus of this seminar, naturally change when children become adolescents. As such, it stands to reason that the impact of CAH would be different for children and families at a younger age than in the teen years. While it is likely very important to connect with families of adolescents, and to offer supports for teens as they go through adolescence, this is a topic that would require an additional grouping and set of materials. As this seminar was only designed to be a day long, it would not be possible to cover that amount of material in one seminar. Therefore, only one age group could be focused on for this seminar. Because many children are diagnosed with CAH either at birth or in early childhood, this was the age that would be focused upon. Additionally, it is essential for the content of the seminar that the children involved know about and understand their CAH to a certain degree. The communication of the families will need to be considered and families informed ahead of the seminar that the content of the seminar includes clear discussion of the effects of CAH, physically, emotionally, and behaviorally. Parents would be informed that their children should have a fair understanding of CAH and its impact in order to participate.

Research has shown that short-term psychoeducation can have a real and lasting impact on families. It is not probable that a longer, more lasting, intervention would be practical for most families, nor would such an intervention be possible for families who do not live in areas

with a great number of other families dealing with CAH. Additionally, this seminar is intended for families with young children (mostly under 12), and as such, these families may not have the time or inclination to attend a longer-term intervention. Families that would have to travel long distances to congregate together and with providers would certainly be excluded by practicality. In order to reach the largest number of families at once, offering a short term intervention at a national conference would allow families to come together in a way that may not be possible in their home areas.

Facilitators

The seminar requires four presenters. As stated in the literature review, psychoeducation is more appropriate and useful when presented by trained professionals. It is important for the facilitators to be well versed and knowledgeable in all topics discussed. In the case of CAH, it is useful to have trained mental health practitioners as well as a medical social worker or nurse on the team of facilitators. Presenters should be experienced in working with children and families as well as have a working knowledge of family systems theory, to give them a background understanding in the underlying ideas behind the scope of the seminar. While the goal of the seminar is to be mental health related rather than medical health, it is likely that questions would arise that would touch upon the medical aspects of CAH. As such, presenters should all have a general knowledge of the etiology and manifestation of CAH as well as a basic understanding of the treatment options available. Additionally, with a medical social worker or nurse on the team of presenters, families would feel that they had someone they could go to with questions of a medical nature. In order to have a general knowledge of CAH, presenters would be required to read the Literature Review section of this clinical research project. Presenters would also meet

and become known to each other prior to the presentation of the seminar so that they could review and plan for the seminar and each of their specific parts.

Seminar Design

While the seminar has some segments that are designed for the family as a whole, there are segments that are broken down by sub-groups. As stated above, children under age 8 would be cared for near their parents to allow parents time and space to participate as needed while maintaining close contact with their youngest children. Separate group activities would also be presented for parents and caregivers, affected children, and siblings, each in their own groupings. Since the sibling-groups would likely be the largest groups, as well as the most diverse in age, these groups would be split into two groups, by age, with older children in one group and younger children in another. As there is not cutoff age for siblings, there is not a cutoff specific age for each group, though the general goal is for the younger children's group to be ages five through nine and the older children ten and above. Having these separate groups would allow facilitators to adjust the needs of the group to what is developmentally appropriate for that age. Using separate segments for each group would allow participants to speak openly and honestly without worry or concern for members of other groups. Parents will be free to speak without worry about speaking in front of their children, and siblings will be given this free space as well.

Size of the seminar. The seminar would be expected to be large enough to allow for a variety of experiences, but small enough to allow for bonding amongst group members. As such, it is expected that each group, such as the sibling group, or the affected child group, would have at least 8 but no more than 12 participants. This was designed to allow for instruction on a more personal level as well as varied discussion and participant socialization. A group that was too large could make it difficult for group members to feel connected and invested, while a group

that was too small could make open discussions more difficult. Based on these projections, the seminar is designed to reach 32 to 48 participants each day it would be offered.

Content of the seminar. The seminar would run a large portion of the day, during an event such as the annual MAGIC Foundation Conference that is held in July. This conference brings together large numbers of families whose children have disorders that affect growth, including CAH, and thus provides an optimal opportunity for this type of presentation. It would begin at 9:00 am and run through a social lunch until concluding at 2:15 pm. The seminar was designed to begin with everyone gathering together. Following introduction of the facilitators, there would be a brief introduction to the goals of the seminar: to help families learn to adjust to life with CAH. At this time, families would have the opportunity to speak and offer small introductions, including the names and ages of family members, when they began to deal with CAH, and what they were hoping to gain from the seminar. Following this period of introductions, participants would be split into the following groups: parents, caregivers, children under 8, children with CAH, young siblings, and older siblings. The children under 8 will be slightly separated from their parents and provided with entertaining activities not on the topic of dealing with CAH. The introductory segment of the seminar is designed to take thirty minutes and run from 9:00 am to 9:30 am.

Individual Section for Parents and Caregivers

Parents and caregivers would have one clinician to lead their portion of the seminar. This facilitator could be the medical social worker, or a mental health clinician. In this segment, parents would go around the group to give another introduction of themselves. At this time, they also would be asked to add a brief description of their experiences with CAH. This gives parents the opportunity to speak openly about their experiences without concern for distressing their

children. This also serves as a beginning to cohesion within the group, as it is expected that there would be some similar experiences shared. Allowing each participant to speak would begin to help them to feel comfortable talking publicly about CAH and how it has affected them and their family. This segment is planned to run for 30 minutes, from 9:30 am to 10:00 am.

The second segment of the parent and caregiver portion of the seminar consists of a brief, information presentation. This presentation, with a printout to allow for ease in taking notes if wanted, provides an introduction and education about the family system elements of cohesion, adaptability, and communication. Also covered would be the course and expected phase of CAH and how each of these can affect cohesion, adaptability, and communication. Just before this presentation, participants would complete the Phase of Disorder Questionnaire and the CAHKAQ (Appendix B), which would then be discussed after the presentation. The presentation also discusses ways of improving cohesion and adaptability in the interest of the families' health and functioning using methods of communication. This segment is intended to identify and elaborate methods of positive change within family systems. This section is intended to last 45 minutes, running from 10:00 to 10:45 am.

Following this presentation, participants would have a fifteen-minute break to use the restroom and have a snack or check in on their children under age 8. Upon return, the facilitator would begin a discussion on ways to problem solve levels of cohesion and communication in the family that are specific to the child's particular course and phase of CAH. To begin, the facilitator would review the Phase of Disorder Questionnaire in order to help each family determine which phase they are currently in, as well as what would support their child's progress to the next phase. For example, Rolland (1994) states that in order to move into the chronic phase, a family must have met these goals: found a doctor to treat the child, decided on treatment

options, learned how to administer treatments, begun to learn about the illness, and started to adjust to life changes that may have come with the disorder. The questionnaire asks the family to rate themselves on a five-point scale for each goal. If a family scores three-or-below on a goal, this suggests that they need to work on that goal in order to move to the chronic stage. During this use of the questionnaire, the facilitator would begin a discussion with preset questions in order to start a conversation about how families can work to alter levels of cohesion and adaptability.

A list of preset questions is part of the organization of the seminar (Appendix A), however the facilitator would also be able to allow the conversation to flow in a more natural, conversational manner. These questions would be included in the notes given to the families at the beginning of the informative presentation to allow them to continue conversations outside of the seminar. The purpose of this portion of the seminar is to give parents and caregivers the opportunity to attempt to use some of the information that they have just received through the presentation. This practical application of the information will help the participants to retain the information for a longer term way to help them problem solve and make changes to their family system. This also allows the families to begin to talk about family dynamics, which is a first step to changing those dynamics. By discussing family dynamics together, the group continues to bond and support each other, increasing the cohesion of the group. Participants can learn from others' experiences, increasing their ability to problem solve situations in the future. This segment would run 45 minutes, from 11:00 to 11:45 am.

The final segment of the parent and caregiver section focuses on helping the adults to learn healthy coping skills. As coping with stress is vitally important when parenting a child with a chronic illness, and as stress reduction is vital in CAH specifically, this is an important

segment of the seminar. The facilitator would begin with a discussion amongst the participants about healthy ways to cope and tolerate distress. This includes methods of seeking support as well as self-care and healthy choices. This discussion could be led by following a preset list of questions (Appendix A), or could evolve more freely. A list of potential coping skills would also be handed out to participants (Appendix C). The purpose of this segment is to increase the parent and caregiver's ability to cope with the negative aspects of CAH. This section would last 30 minutes, from 11:45 to 12:15.

Individual Section for Children with CAH

One clinician would facilitate the group for children with CAH. This begins with another round of introductions. As the seminar was designed to use narrative techniques to externalize CAH from the children, the facilitator keeps this framework in mind throughout every segment of the seminar. Narrative therapy focuses on allowing someone to retell his or her story in a less negative way, a way that will be less harmful to that individual. The use of externalization allows children to begin to place some separation between themselves and CAH. Using this technique allows children to begin to form an identity and sense of self that is separate from having this disorder. The goal of this is to decrease the psychological ramifications and incapacitation of an illness. By giving the children some space between themselves and their disorder, it is hoped that this will allow the children to see their struggles in a more neutral manner, thereby making it easier to look for and find solutions. The facilitator would ask the children to introduce themselves to the group by stating their name, the age at which CAH first came to visit and their favorite color. By asking when CAH first came to visit, the facilitator is beginning to allow externalization of CAH. By asking about the child's favorite color, the facilitator acknowledges that there is more to the child than just CAH, and that they are

individuals, with individual likes and dislikes. These introductions last about 15 minutes, running from 9:30 to 9:45 am.

Following the introductions, the facilitator would move fluidly into the next segment of the seminar, a craft project. A script is provided for the facilitator (Appendix A) that states that the children would be seeing their CAH in a new way. The facilitator would give each child a plain muslin doll and invite the children to decorate the doll so that it would look like their CAH. Children will be given art supplies to support a variety of expressions of CAH, such as paint, markers, felt, glue, feathers, pipe cleaners, yarn, glitter, paper, ribbons, beads and sequins. The goal of this group is to use art to allow the children to externalize and separate from CAH. Art allows them to stay engaged and focused on the task. This activity lasts 20 minutes, from 9:45 to 10:05. Following this activity, children are allowed a break for the bathroom and snack.

When the next segment begins, children are asked to sit in a circle and talk about their projects. Each child in the group would introduce their CAH doll and discuss why they decorated it the way that they did. They would be encouraged to be as descriptive as possible when talking about their CAH, what it sounds like, how much space it takes up, etc. This would create more of an identity for their CAH, and encourage children to think of CAH as an entity separate from them. Once each child has had the opportunity to speak, the facilitator begins to lead a discussion of how CAH has affected each child's life. This could follow a preset list of questions (Appendix A) or flow more naturally from the earlier discussions. This continues to be conducted in narrative style with questions such as: does CAH ever make you think bad things about yourself? The purpose of this section is to help children begin to think of themselves as separate from CAH and to start to communicate about the ways that CAH has affected their lives and their family. This segment runs about 45 minutes, from 10:20 to 11:05.

Next, the children will have a second short break for the bathroom or talking to each other. The children's problem-solving portion of the seminar follows this break. Using narrative language, the facilitator would ask the children about the ways that CAH causes trouble for them. Children are allowed to speak freely about negative effects of CAH, such as feeling badly about themselves, feeling sick, or being excluded from activities due to their medical concerns. The facilitator then encourages the children to discuss with each other the ways that they have solved or thought about solving these problems. When someone has difficulty with solutions, the other children will be encouraged to help that child to think of options for solving these struggles. For example, a child may not have been able to play competitive sports at school due to concerns about salt-wasting, dehydration, and stress levels. The children will be encouraged to come up with ideas to deal with this such as finding other social activities at school that are allowed, having a less strenuous role on the team, or talking with their parents and doctors about their interest in sports and trying to find a way that they can play. The children are encouraged to brainstorm different ideas together as well as discuss what has and has not worked for them in the past. The children would then be broken up into two groups and each group will be given a role play situation of a child with CAH being left out of something they want to do (Appendix A). The groups would be asked to play out the scene complete with solution to the problem ending the scene. This gives children practice using the skills they will need to solve ongoing problems faced by children with CAH. They are able to learn from each other's experiences. This segment runs 45 minutes, from 11:15 to 12:00.

The final segment for children focuses on identifying healthy coping skills. The facilitator discusses with the children the ways that they help themselves to feel better when CAH causes trouble for them. A handout of coping skills and healthy activities will be given to

the children to reference when they are upset (Appendix D). The goal of this segment is to increase distress tolerance and self care for the children. It runs 15 minutes, finishing at 12:15.

Individual Section for Siblings

The sibling sections are broken into two smaller groups, based on age. Because the experiences of children and siblings will vary so vastly by age, it is essential that the children be separated to allow the best possible experience to be had by all. It is expected that a 14-year-old sibling would have different challenges than a 9-year-old sibling. Therefore, while the groups are structurally the same, they are developmentally geared for different age groupings. As the families would be required to register ahead of the conference, the facilitator would know information about the siblings ages ahead of time and could adjust materials to fit those ages.

One facilitator presents the sections. As with the other groups, these segments begin with introductions, asking each child to state their name, age, family member with CAH, and favorite food. This is designed to help the group begin to bond as a coherent grouping. Narrative language would be used to allow siblings more comfort in discussing the effects of CAH on them and their family. Introductions would last 15 minutes, running until 9:45.

Following introductions, the facilitator introduces the first activity, a drawing activity (Appendix A). Each child is given a large piece of paper with three sections. The facilitator explains the activity, which is to draw three separate pictures on the paper, detailing a picture of their family before CAH came to visit, now that CAH has arrived, and what they hope their family will be like in the future, with CAH. Similar art materials to those used by the other children's groups are used to give the siblings a method of non-verbal communication around the effects of CAH on the family. This activity also allows for forward thinking about life in the

future, even with CAH still present in the family. This activity runs for 30 minutes, until 10:15, followed by a 15 minute break for snack and bathroom.

Following the break would be the discussion segment for this group. Each child would be asked to show their pictures and discuss what their picture means to them and why they chose to depict these elements of family life. The facilitator would work to point out similarities amongst the pictures in the group as a method of normalizing experience and continuing to bond the group. Using a preset list of narrative questions, the facilitator allows the siblings to begin to discuss ways that CAH has affected their family in more detail. These questions (Appendix A) could lead to a free flowing discussion such as: does CAH ever cause trouble in your family? This continues to normalize experiences had by siblings of a child with a chronic disorder and increases the children's ability to communicate about the effects of CAH. This discussion would last 40 minutes, until 11:10.

Siblings would then be given a ten-minute break to talk freely with each other. The problem solving section of the seminar follows this break. Continuing the discussion of the troubles that CAH may have caused within the family, the facilitator would discuss these troubles in more detail with the goal of finding solutions for these problems. The facilitator asks the children for suggestions as to how these problems could be solved but also offers suggestions if needed. As with the other children's group, the siblings are broken into two groups and given a role-play activity to attempt to solve problems involved with CAH in the family. These groups discuss the problem and then return to act out their scene, with a solution to the problem at the end. This gives the siblings an opportunity to think about some of the problems that arise, as well as to practice potential solutions. This segment is planned to last an estimated 40 minutes, until 12:00.

The final segment of the sibling seminar focuses on coping skills and healthy living. The facilitator asks the children to discuss things they can do when they are stressed and ways they can make themselves feel better when they are upset. A list of coping skills and healthy activities is handed out (Appendix D). The goal of this segment is to increase their ability to handle negative emotions and stressful situations. This section is planned to last 15 minutes, until 12:15.

Family Section

At this time in the seminar, families would gather back together again and the entire group comes together for lunch. Participants would be encouraged to talk and socialize with each other during lunch to build networks of mutual social support. Lunch is planned to last 30 minutes and families are asked to gather again at 12:45 for the next segment of the seminar, family sculpting. As stated earlier, family sculpting is a non-verbal communication that expresses family dynamics that might otherwise not be discussed. This is done by creating a sculpture of the family, using the family members themselves as the sculpture. Well-known family therapist Virginia Satir's family sculpting, carefully moving and posing each family member, can be useful for any age family member regardless of their verbal ability or ability to communicate emotion and experience using words. Because the goal of family sculpture is to explore underlying family dynamics including roles, responsibilities, emotional connection, cohesion and potential discord, it will be important for the facilitators to be aware of the possible need for additional supports both during and after the seminar. Facilitators should be prepared to offer crisis management supports in the moment as well as connecting families with referrals to providers with whom they could connect following the seminar.

All four facilitators would be a part of this segment. After one facilitator explains how to go about creating a family sculpture, they move around throughout the room, helping families as needed. One important instruction for this family sculpture is that each family be sure to use the doll created to represent CAH. Each member of the family would be given an opportunity to create a family sculpture. A handout list of discussion questions would be given out for each family to discuss after each sculpture (Appendix A). This would help to increase verbal communication within the family and about the impact of CAH on the family. This activity is planned to last 45 minutes, until 1:30.

Following the family sculpture activity, all families would gather in one large circle. The facilitator discusses the experience of creating a family sculpture and asks the families to discuss what it was like for them to create these expressions of family experience. Again attempting to normalize common experience, facilitators would make note of common themes and bring the group together on those points. A preset list of questions is available (Appendix A) such as: did CAH get in the way of your family sculpture, or was anyone surprised to see what CAH was up to in the family? Natural flow of conversation is also allowed. This segment would run between 1:30 and 2:00.

At the conclusion of this discussion the seminar would be nearing the end. Families would then be asked to fill out a Feedback Questionnaire (Appendix E) and reminded that they would be sent follow up questionnaires (Appendix E) in the mail in a few weeks and again in a few months. They would be encouraged to continue to use their communication and problem solving skills throughout the following months. Facilitators would provide contact information in case of questions or comments. Families would be encouraged to give each other contact information and to make future plans for the remainder of the conference or beyond if desired.

Families would also be given a handout of discussion topics to take home with them. These questions are geared toward encouraging the families to continue discussing their experiences with CAH, and to continue to increase their communication. This is designed and explained as an opportunity to continue to adjust levels of cohesion and adaptability within the family as needed. This segment was designed to run 15 minutes, completing the seminar at 2:15.

Implementation of the Seminar

The seminar could be piloted in full, during a conference such as the annual MAGIC Foundation Conference that is held in July. This conference brings together large numbers of families whose children have disorders that affect growth, including CAH, and thus provides an optimal opportunity for this type of presentation. In order to offer this seminar to the greatest number of families, it would be offered twice over two days of this four day conference. Registration for the seminar would be required before the participants' arrival at the conference. This allows the facilitators to separate the families into groups that are created by age of the child. The facilitator would put families with children under 8 in a separate group from those with children in elementary school or teens, for example. In this way, the facilitator is able to tailor the materials to the age of the child in the family. By registering ahead of time, the facilitators are able to guarantee that the groups are an appropriate size to maintain the optimal amount of learning and experience. If space existed in either group at the time of the conference, additional families would be allowed to participate on the day of the seminar without pre-registration.

In addition to its applicability to a national conference, this seminar can also be implemented in support groups through hospitals or communities in smaller areas and groups of families. Because the seminar was designed to be facilitated by four presenters at one time, it

may be difficult to carry out in this manner. However, offering the seminar in four separate sessions with the same facilitator (?) over a period of time may also be worth exploring as an option for smaller settings. Additionally, a facilitator may choose to only present one or two segments of the entire seminar, as needed. While the seminar is designed to be most useful altogether, there is merit and value to each individual section.

Chapter 5: Discussion

CAH is mostly unknown, and poorly understood by those who first learn about it when a family member is diagnosed. When a family begins to learn about the disorder and cope with the changes brought about by CAH, they may feel confused, frustrated, scared and unsure of what to do or how to help. This seminar was designed to be a support in the early days of a diagnosis, as well as a potential for increasing support for families throughout the life of a child with CAH. Additionally, it is meant to connect families with other families who are in a similar situation, and may help connect families with providers who can assist them in finding a medical home, a place for their child to continue to be seen over their lifetime, by connecting families to known providers in their home area who have experience with CAH. Depending on the facility providing the seminar, there might be different goals and possibilities for the families participating.

The seminar has been designed using a narrative family systems model. Because family support is so meaningful and necessary for children as well as siblings, it is important to involve everyone in the family in this seminar to assist them in understanding their family member with CAH as well as understanding the role and impact they also have on the family as a whole. The narrative model allows children to make sense of what has occurred in their family as well as normalizing the experience and allowing them to see their family member with CAH (or themselves) as a person outside of the disorder.

This seminar additionally explores the knowledge parents have about CAH, through the use of the CAHKAQ. As knowledge about a disorder helps with adherence to treatment, an understanding of where a family is at this point in time will begin to facilitate discussion of what may not yet be known. Additionally, by using the FACES-IV to measure the families levels of

cohesion and communication both before and after the seminar, facilitators will be able to assess the utility of the seminar and begin to explore expansion or design changes as they are possible over time. Having the families fill out this paperwork ahead of the seminar sessions will allow the facilitators to emphasize topics for discussion which may be more needed for particular familes. For example, if multiple families are noted to struggle with cohesion, the facilitators could focus more on skills necessary for increasing cohesion, whereas if a larger portion seem to struggle with adaptability, the facilitator could emphasize skills needed to increase adaptability.

Clinical Implications of the Seminar

This seminar is designed to be helpful for families of a child with CAH, however it is possible that it could additionally be useful for families with children with other chronic disorders, with some alterations. The processes of family adaptation and strengthening would be the same for families dealing with other disorders, and as such the seminar may be able to be adapted for greater applicability. The information presented would need to be altered, and the family interaction portions may need to be explored in differing ways for different diseases. The facilitators of the seminar would need to be educated in etiology, presentation, treatment, and common concerns for each of the disorders for which they wished to implement the seminar. These changes would be relatively easy to put in place, however, and the overall structure and activities of the seminar could remain the same.

Children and families with disorders such as CAH are followed and cared for by medical personnel in hospitals or community clinics. Providers who work with patients with CAH may also benefit from the seminar as the knowledge presented about the impact of family dynamics on the course of the disorder and well being of the child and family as a whole would be beneficial to the providers as they plan and implement treatments and interventions for the

families. The activities, while most useful presented as a whole, and within a group, could be used in smaller settings individually. Thus the providers could learn about them by attending and participating in the seminar, offered for professionals, and then take this information and activities back with them to their own settings. While providers would likely not need the section that is an overview of CAH, they may not have had training on the impact of such disorders on the family and as such the information about family cohesion and adaptability would be beneficial to them.

Limitations

This study is limited because it is only a design of the seminar. Because this study lays out the plan for a seminar rather than actual implementation of such a seminar, there is no way to assess the actual efficacy or utility of the seminar. A review of published literature has not shown any known family-based treatment for families dealing with CAH. As such, the formation of a seminar such as this is the first step in providing families with efficacious treatment. Furthermore, no research was found that examined the effects of CAH on the family system. Because of this, the data has had to be extrapolated from studies examining other chronic childhood illnesses. Therefore, this seminar might not address all of the concerns and difficulties that these families face. In order to counter this limitation, a feedback questionnaire will be designed so that the seminar could be improved on a continuous basis.

Areas for Future Research

In addition to this seminar, another area of potential research might be facilitating focus groups, to get a sense directly from the families and children with CAH of what they might find useful in a group setting. This could be done as the beginning of an ongoing social support and

therapeutic group, or as a stand-alone temporary group brought together solely to increase understanding of families' needs and desires.

Facilitators of this seminar may be psychologists, social workers, medical personnel or other educated providers who work with families who have a child with CAH. As such, they may be interested in exploring further ways to work over time with these families, offering greater supports and options for families who are able and interested in longer term support. The format of the seminar is also fairly easy to work with and adapt, and as such, facilitators may be able to use it to work with families with children with other Disorders of Sex Development, or even other chronic disorders that affect children. Over time, it would be interesting to follow further research on health related quality of life and potentially explore the utility of a seminar and family involvement such as this and the impact that these interventions may have on individuals with any chronic disorder first diagnosed in childhood.

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Appendix A

PSYCHOEDUCATIONAL SEMINAR

Structure of the Seminar

PART I- 9-9:30
Welcome All Families to the Seminar
Introduction of Facilitators and Participants

PART II- 9:30-12:15
Break into Individual Groups
Individual Sections of the Seminar

PART III- 12:15- 12:45
Lunch and Free Discussion Among the Families

PART IV-12:45-1:30
Family Sculpting Exercise

PART V- 1:30 -2:00
Discussion of Family Sculpting Exercise Among Families.

PART VI- 2:00 -2:15
Feedback Questionnaire
Contact Details Handout
Goodbyes

Individual Section for Parents and Caregivers

9:30-10:00: Introductions

- Each participant will introduce themselves and speak briefly about their experience with CAH.

10:00-10:45: Presentation of Information on Family Dynamics

- Fill out Phase of Disorder Questionnaire
- Family Dynamics Information presentation

10:45-11:00: Break and Snacks

11:00-11:45: Problem-Solving Discussion

- Review of Phase of Disorder Questionnaire
- Discussion Questions:
  - What type of CAH does your child have?
  - What phase of disorder is your family in?
  - Has the family gotten closer, drifted farther apart, or stayed the same since your child's diagnosis?
  - Have certain relationships changed while others stayed the same?
  - Are you more protective of your children? How do you think this may impact the family?
  - Have routines, schedules, or responsibilities changed? If so, how does this impact the family?
  - Is your family's level of cohesion and adaptability the best fit for the course and phase you're currently dealing with?
  - What changes may you have to make as the course and phase of CAH change?

11:45-12:15: Coping Skills Discussion

- Introduction of Coping Skills Discussion:
  - "In the beginning, we talked about the health of the family as a whole. Now it time to talk about how to keep you, as parents, feeling emotionally healthy."
  - What are ways that you can help yourself feel better?
  - What are ways that you can get support from others?
- Coping Skills Handout

Individual Section for Children with CAH

9:30-9:45: Introductions:

- Participants will introduce themselves, stating their name, age, when CAH first came to visit and favorite food

9:45-10:05: Craft Exercise:

- "Today we're going to get the chance to put a face to your CAH. Using these (doll), each one of you is going to have the chance to decorate them however you'd like in order to make them look like your CAH."

10:05-10:20: Break

- Participants are given the chance to use the bathroom and have a snack.

10:20-11:05 Discussion Exercise:

- Discussion of craft projects
- Discussion of other aspects of CAH
  - "First, let's learn a little more about our CAH. What does it sound like? If it big or small? Does it take up a lot of room when it comes into a room? Does it talk really loudly or whispers like a little mouse?"

- Discussion of how CAH has impacted their life
  - "Now that we know what everyone's CAH looks like, it's time to get to know it a little better. We're all very different people, but the one thing everyone has in common is that CAH knocked on your door one day. We're going to talk a little bit about what it's like when CAH comes to visit. For some people, CAH came a long time ago and is still here. For others, CAH just came recently. What has life been like since CAH come to stay with you?"

  - What was it like when CAH came?
  - How did things change when CAH came?
  - What trouble does CAH make for you, your family, your friends?
  - Did CAH bring anyone else with it, like sadness or embarrassment?
  - How did you feel when CAH came to visit?
  - What were the bad things that CAH brought?
  - What were the good things that CAH brought?
  - Does CAH ever try to make you do things you wouldn't normally want to do, like be shy, angry or yell?
  - Does CAH ever make you think bad things about yourself or others?
  - Has CAH made people in your family act differently?
  - Has CAH made some people in your family closer or gotten in the way of other people?
  - Does CAH ever try to make you keep secrets? Does CAH demand a lot of attention for anyone in your family?
  - Has CAH ever ruined an event or activity for you or your family? What would you like other people in your family to know about how you think CAH has affected you?

11:05-11:15: Break

11:15-12:00: Problem Solving

- ❖ Discussion of difficulties that CAH brings.
- "We talked a little bit before about the trouble that CAH can make for you. Sometimes people whisper, or seem uncomfortable. Right now we're going to talk about ways to help deal with those problems. First, let's start by talking about what troubles CAH causes."
- ❖ Discussion of problem-solving techniques
- "OK, now let's talk about ways to deal with these problems."
- ❖ Role Plays
- "Alright, now that we have some idea, let's practice!"
- Role Play 1) All your friends are starting sports teams but you cant play because of CAH. What can you do?
- Role Play 2) You are in class and your teacher lines everyone up by height. You're always the tallest and you don't like it. What can you do?

12:00-12:15: Coping Skills Discussion

- ❖ "Sometimes your CAH can be really annoying and make a big fuss. And sometimes when this happens it can make you feel sad, angry, alone, and just bad in general, no matter how well we handle the problems. When CAH gives you these feelings, what can you do to help yourself feel better?"
- ❖ Coping Skills Handout

Individual Section for Siblings

9:30-9:45- Introductions

Participants will introduce themselves by stating their name, age, the member of their family with CAH and their favorite food.

9:45-10:15: Drawing Exercise

- ❖ Introduction of Drawing Exercise
- "I know that no one in this room has CAH, but CAH still has a major impact on your family and on your lives. Right now we're going to do some drawings that show how CAH has affected your family."

10:15-10:30: Break

- ❖ Participants are given the chance to use the bathroom and have a snack.

10:30-11:10: Discussion Exercise

- ❖ Discussion of each participant's drawing
- ❖ Discussion of how CAH has impacted their family
- "We've seen through your pictures how CAH has affected your families. Now we're going to talk some more about how CAH changed things when it came into your family. Some of these changes may have been bad. But some may have been good. This is a place for you to talk about those changes with people who will really understand, because they may have been through similar experiences due to CAH being in their family."

- What was it like when CAH came into your family?
- How did things change when CAH came?
- What were the bad things that CAH brought?
- What were the good things that CAH brought?
- What trouble does CAH cause for you, your family, your sibling?
- Did CAH bring anyone else with it, like sadness or embarrassment?
- How did you feel when CAH came to visit?
- Does CAH ever make you think bad things about yourself or others?
- Has CAH made people in your family act differently?
- Has CAH made some people in your family closer or gotten in the way of other people?
- Does CAH ever try to make you keep secrets?
- Does CAH demand a lot of attention from anyone in your family?
- What's it like to see your sibling struggling with CAH?
- Has CAH ever ruined an event or activity for you or your family?
- What would you like other people in your family to know about how you think CAH has affected you?

11:10-11:20: Break

11:20-12:00: Problem-Solving Exercise

- ❖ Discussion of problems that CAH brings

- ❖ Discussion of problem-solving techniques
- "OK, now let's talk about ways to deal with these problems."
- ❖ Role Plays
- "Alright, now that we have some idea, let's practice!"
- Role Play 1) You are at the playground with your family and a group of children start to make fun of your brother/sister who has CAH. What do you do?
- Role Play 2) You want to go to summer camp, but Mom/Dad say that you can't because your brother/sister with CAH couldn't also go. What can you do?

12:00-12:15: Coping Skills Discussion

- ❖ "Even with the best problem solving skills in the world, there are still going to be times when CAH makes you feel sad, angry, jealous, nervous and upset. When this happens, what are things you can do to make yourself feel better?"
- ❖ Coping Skills Handout

Family Section

12:45-1:30: Family Sculpting Exercise

- ❖ "When we were all in our separate groups, we *talked* a lot about how CAH has impacted our families. In this part of the seminar, we're going to *show* each other how CAH has affected our families. Let's pretend that each of our family members is a piece of clay. They can be molded and moved anyway that we want them to be. In this way, we can make a sculpture of our families by using each other. We can put people who get along well right next to each other, maybe even hugging. Or we could put people who don't get along far away from each other so that they're not facing each other. If Mom is super bossy, you can have her put her hand on her hip and point her finger as if she's telling you what to do. Or if you brother is super funny you can have him make a funny face. And we can use the wonderfully decorated CAH Dolls that were made in the last part of the seminar to represent CAH. Since CAH is now a part of the family, it should also be in the sculpture. Each family member is going to get a chance to direct the family in making a family sculpture. If you have any questions please ask one of the presenters."
- ❖ Questions to discuss after each sculpture:
- Who was close together in the sculpture?
- Who was far away from each other?
- Where did CAH fit into the sculpture?
- Did CAH get in the way of people in the sculpture? If so, how did it impact how these people got along?

1:30-2:00: Between Family Discussion

- ❖ Discussion of families' experiences of the Family Sculpting Exercise
- What about the sculptures surprised you?
- Where was CAH placed in the sculptures?
- Did different people put the CAH in different places?
- Did CAH divide or get in the way of the family at all?

2:00-2:15: Goodbyes

- Feedback Questionnaire
- Handout of Contact Details
- Time for families to say goodbye and exchange contact details
- Homework Handout Questions:

Appendix B

Phase of Disorder Questionnaire

Please rate how strongly you agree or disagree with the statements below using the following scale:

1= I strongly agree **4 = I disagree**

2=I agree **5= I strongly disagree**

3=I neither agree nor disagree

A) I feel confident with my overall understanding of what CAH is.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

B) I have found a doctor that I trust to treat my child.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

C) I feel that I have a good understanding of the treatment options available to my child.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

D) There is a family member within the household that feels comfortable administering my child's treatment on a daily basis.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

E) My family has adjusted to the changes that have come with my child's CAH.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

Coping Skills Questionnaire

Please rate how strongly you agree or disagree with the statements below using the following scale:

1= I strongly agree **4 = I disagree**

2=I agree **5= I strongly disagree**

3=I neither agree nor disagree

A) When I feel stressed, I talk to someone for support.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

B) When I feel stressed, I do something physical (work out, play a sport, go for a walk).

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

C) When I feel stressed, I do something nice for myself (take a bath, get a massage)

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

D) When I feel stressed, I work on a hobby (painting, writing, gardening).

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

E) When I feel stressed, I want to be entertained (tv, movies, funny jokes).

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

Appendix C

Parents Coping Skills List

WAYS I CAN TAKE CARE OF MYSELF

Deep breaths

Count backwards from 10

Take a shower/bath

Talk to a friend or family member

Go for a run

Lift weights

Read a book

Play a sport you love

Watch a favorite TV show

Write in a journal

Garden

Cook something delicious

Order take-out so you don’t have to cook

Take a walk or hike

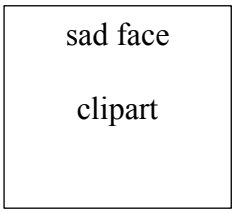
Listen to music

Ride a bike

Have fun with my kids

Appendix D

Coping Skills List for Children and Siblings



WHEN I'M UPSET I CAN…

-take slow deep breaths

-count backwards from 10

-squeeze a stuffed animal or stress ball

-talk to an adult about how I feel

-spend time with friends who make me feel good about myself

- do 10 jumping jacks

-ask to take a break

-think about a happy time

-think about all the things I'm really good at

-cry, sometimes it makes you feel better

-draw or paint

-write in a journal or diary

-think about something funny

-punch a pillow

-run as fast as I can

Appendix E

Feedback Questionnaire:

Please rate how strongly you agree or disagree with the statements below using the following scale:

1= I strongly agree

2=I agree

3=I neither agree nor disagree

4 = I disagree

5= I strongly disagree

The seminar increased my understanding of how I can help my family adjust to CAH.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

The seminar gave me practical tools that I could use to help my family adjust to CAH.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

The information in the seminar was presented in a clear manner that was easy to understand.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

The family activities were enjoyable for all members of the family.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

I would recommend this seminar to other families dealing with CAH.

(I strongly agree) 1 2 3 4 5 (I strongly disagree)

The best part of the seminar was:

The worst part of the seminar was:

In order to improve the seminar I would:

Appendix F

Homework Handout

Today you learned A LOT about CAH and how it impacts you and your family. In order to make sure that CAH doesn't take up too much space in your family, you have to practice the skills you learned today. Below are some questions that your family can talk about together to help you all keep CAH from hurting you and your family. Talk about these questions tonight after the seminar, and then on a weekly basis.

Questions:

What does everyone want their family to know about how CAH has affected them?

Did CAH get in the way of you doing something you would have liked today?

Do you think CAH made your family closer or more distant?

Does CAH ever impact the way your family acts with each other? Does it make you spend more/less time with each other? Talk more/less with each other?

What would you, as an individual, like to tell CAH today?

What, as a family, do you want CAH to know?

When CAH tries to do bad things to your family, what tools can you use to keep that from happening? Humor? Talking? Hugging? Seeing friends?

Has CAH kept you from seeing your friends, or doing things with people outside the family? What do you think about that?

Has CAH made people take on different or new jobs in the family? What do you think about that?

If you had to make another family sculpture, where would everyone put CAH? Would it be in the way of anything? How is this different than the last sculpture?

Has CAH changed at all? If so, how does our family have to change in order to make sure it doesn't start making trouble for us?

Appendix G

Congenital Adrenal Hyperplasia Knowledge Assessment Questionnaire

This questionnaire is confidential. Please assist us by answering all the questions below

Only complete this section if you care for a child with CAH

Your Name: _____ Relationship to child: _____

Child's Name: _____ Child's Date of birth: ___/___/___

Child's gender: male / female (please circle)

Today's Date: _____

1) What year was your child diagnosed with CAH? _____

2) What medications does your child take each day? _____

3) What type of CAH does your child have?
   Classical (salt losing)
   Non-classical (non salt losing)
   Unsure

OR

Only complete this section if YOU have CAH

Your Name: _____ Date of birth: ___/___/___

Gender: male / female (please circle)

Today's Date: _____

1) How old were you when you knew you had CAH? _____

2) What medications do you take each day for CAH?_____

3) What type of CAH does your child have?
   Classical (salt losing)
   Non-classical (non-salt losing)
   Unsure

Test Your Congenital Adrenal Hyperplasia Knowledge

For each question, circle one answer only.

1. CAH is a condition of the:
- a) pituitary gland
- b) adrenal gland
- c) ovaries or testes
- d) unsure

2. In CAH the body makes:
- a) too much cortisol and too much androgen
- b) too little cortisol and not enough androgen
- c) too little cortisol and too much androgen
- d) unsure

3. CAH is caused by:
- a) a virus
- b) a faulty gene
- c) a harmful environmental agent
- d) unsure

4. The chance that CAH will recur in a family is:
- a) one in four with every pregnancy
- b) with every pregnancy
- c) never
- d) unsure

5. To have CAH the child must inherit the gene from:
- a) both the mother and father
- b) the mother only
- c) the father only
- d) unsure

6. Cortisol is:
- a) an essential vitamin
- b) essential for decreasing blood sugar
- c) a hormone essential for life
- d) unsure

7. Too much androgen may:
a) slow growth
b) cause rapid growth and early puberty
c) delay pubertal development
d) unsure

8. Fludrocortisone *(Florinef)* helps regulate:
a) salt balance
b) blood sugar levels
c) growth
d) unsure

9. The main reason for treating CAH is to:
a) promote normal growth and development
b) prevent an adrenal crisis
c) both a) and b)
d) unsure

10. Treatment for CAH is required:
a) until you grow out of it
b) until adulthood
c) lifelong
d) unsure

11. In an Adrenal Crisis someone urgently needs:
a) Paracetamol (*Panadol*)
b) salt, sugar and rest
c) a hydrocortisone injection and fluids via a drip
d) unsure

12. Not taking hydrocortisone causes:
a) low blood sugar & blood pressure
b) lack of energy & drowsiness
c) both a) and b)
d) unsure

13. Treatment of salt-losing CAH <u>usually</u> includes:
a) hydrocortisone *(Hysone)* and fludrocortisone *(Florinef)* tablets
b) hydrocortisone *(Hysone)* tablets only
c) fludrocortisone *(Florinef)* tablets only
d) unsure

14. If a person with CAH misses a medication dose they should:
a) give the dose immediately
b) double the dose next time
c) do nothing & give normal dose next time
d) unsure

15. If a person with CAH is sick with a high fever, you should:
a) put them to bed until they feel better
b) give extra salt and sugar and contact the doctor
c) give extra hydrocortisone orally and contact the doctor
d) unsure

16. If a person with CAH has a slight cold but is otherwise well, you should:
a) increase their hydrocortisone dose
b) decrease their hydrocortisone dose
c) monitor temperature and watch closely
d) unsure

17. If a person with CAH is pale and extremely drowsy, they need to have:
a) an extra oral hydrocortisone dose
b) an injection of hydrocortisone and go to hospital
c) sweet salty fluid to drink
d) unsure

18. A person with CAH with persistent diarrhoea and vomiting should:
a) repeat the oral hydrocortisone dose
b) have an injection of hydrocortisone and go to hospital
c) drink sweet salty fluid
d) unsure

19. A person with CAH is at risk of an Adrenal Crisis when they:
a) play sport
b) have a general anaesthetic
c) sit for school exams
d) unsure

20. A hydrocortisone injection should be given:
a) into a vein in the arm
b) into a muscle of the upper arm or leg
c) under the skin of the upper arm or leg
d) unsure

21. Medical follow up is generally recommended for children and adolescents:

a) once a year
b) twice a year
c) more than twice a year
d) unsure

22. The <u>main</u> reason for wearing a medical alert pendant or bracelet is because it:

a) gives your name & address
b) states the diagnosis & emergency treatment
c) indicates your next of kin
d) unsure

Answers

1. **b)** *In CAH, the hormones produced by the outer part (cortex) of the adrenal glands are affected. The adrenal glands produce 3 main hormones – cortisol, androgen and aldosterone. Although the adrenal glands are controlled by the pituitary gland, CAH is a condition of the adrenal glands.*

2. **c)** *The adrenal gland cannot make cortisol due to an enzyme deficiency and produces too much androgen (male hormone) in response. Sometimes the production of the hormone aldosterone is low as well. This combination is known as "salt-losing" CAH.*

3. **b**) *CAH is an inherited disorder caused by a fault (or defect) in a gene. Genes are tiny hereditary units which determine a person's characteristics.*

4. **a)** *A couple with a child known to have CAH faces a one in four chance in every subsequent pregnancy that the baby will have CAH. There is a fifty percent chance that the baby will be a girl and therefore the chance of the baby being a girl with CAH, who may need genital surgery, is one in eight.*

5. **a)** *CAH results when 2 faulty genes, one carried by each parent, are inherited by a child, ie. an affected child inherits 1 faulty gene from each parent.*

6. **c)** *Cortisol is essential for life. It plays a role in regulating blood pressure, blood sugar and helps the body combats stress caused by infection or injury. Cortisol deficiency cause lethargy, pallor, a fall in blood pressure, inability to maintain blood sugar, electrolyte imbalance and eventually collapse.*

7. **b)** *Androgens are hormones produced by the adrenal gland in both males and females and are also produced by the testes in males.*
Androgen levels are normally low until just prior to puberty. Too much of this hormone stimulates the body to grow fast and puberty to occur early. Adequate control with replacement hydrocortisone prevents this problem. Doses need to be adjusted as the child develops.

8. **a)** *In salt losing CAH, the salt regulating hormone aldosterone is missing. Fludrocortisone is the medication used to replace this hormone.*

9. **c)** *The aim of treatment is to normalise hormone balance by replacing the hormones that are low. This promotes normal growth and development and protects the body from an adrenal crisis.*

10. **c)** *Treatment is required life long. If treatment is stopped in adulthood this can lead to infertility issues in both males and females as well as leading to the risk of life threatening adrenal crisis.*

11. **c)** *An adrenal crisis is a life threatening situation requiring immediate treatment. It occurs when there is extreme cortisol insufficiency, and causes the body to go into a state of physical collapse and severe shock.*
The immediate treatment is a hydrocortisone injection given by parents or medical staff. This is followed by intravenous fluids which are required for electrolyte imbalance.

12. **c)** *Hydrocortisone is needed to maintain normal blood sugar levels, blood pressure and physical well being. Cortisol deficiency leads to low blood sugar and blood pressure, a lack of energy and drowsiness.*

13. **a)** *CAH treatment involves replacing the missing hormones. Hydrocortisone (Hysone) tablets are used to replace the missing hormone cortisol. Adolescents and adults use the tablet Dexamethasone (Dex). If the person has "salt-losing" CAH, they also need to replace the missing aldosterone with Fludrocortisone (Florinef) tablets.*

14. **a)** *The missed medication dose should be given immediately, and then the normal regimen followed, unless the person is unwell.*

15. **c)** *When a patient has a high fever, it places the body under stress and the body's cortisol requirement increase. Doses of oral hydrocortisone need to be increased to meet these requirements. Extra doses should be given until the person is well again and advice sought from the doctor.*

16. **c)** *If the person has a slight cold but is otherwise well, this does not have a major impact on the stress system of the body, and therefore cortisone requirements remain stable. However if a fever develops, the cortisol requirements will increase and extra medication may be required. Therefore it is important to monitor the temperature and watch the person closely in case this occurs.*

17. **b)** *Signs of pallor and drowsiness indicate a patient is at risk of an adrenal crisis. Such situations require immediate treatment of an injection of hydrocortisone and transfer to hospital. It is essential that parents and carers understand that this is life threatening, and they need to act immediately.*

18. **b)** *Diarrhoea and vomiting causes loss of fluid from the body and electrolyte imbalance, and failure of medications (hydrocortisone and fludrocortisone) to be absorbed. The person requires immediate treatment. however because of the vomiting a) and c) are not appropriate answers. The person should have an injection of Hydrocortisone and go to the hospital for fluid management.*

19. **b)** *Procedures under a general anaesthetic may cause an increased need for cortisol in the body, placing the person at risk of an adrenal crisis. As precautionary management, it is standard practice to use extra Hydrocorti-sone at induction of anaesthetic, and for the period of recovery. Sitting for school exams or playing sport may put some stress on the body, however this is not at a level that would usually put someone at risk of an adrenal crisis.*

20. **b)** *The safest and most efficient place for parents to inject Hydrocortisone is into the large muscle of the thigh or the upper arm. Hydrocortisone injections given under the skin are inadequately absorbed. Only doctors should inject hydrocortisone into a vein and this can be difficult in an adrenal crisis. Intramuscular injections should be the first line of management.*

21. **c)** *Growing children and adolescents require regular follow ups three to four times per year to assess the progress of normal growth and development, management issues and education needs. Treatment of CAH requires regular medication adjustments according to need, and this is assessed by growth and development, bone age and regular blood tests.*

22. **b)** *It is recommended that any person with a medical condition with serious consequences wear a Medical Alert pendant or bracelet. If an adrenal crisis occurs, it will alert ambulance officers and doctors to the diagnosis of CAH and the need for a specific life-saving medication such as Cortisol.*

References

1. Warne GL (1989) *Your Child with Congenital Adrenal Hyperplasia*. Educational Resource Centre. Royal Children's Hospital Melbourne.
2. APEG (2000) *Congenital Adrenal Hyperplasia: Hormones and Me*. Serono Symposia Australasia.

Acknowledgement for CAHKAQ

CAHKAQ Developed by:
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Reference*: King, J., Mitchelhill,I, Fisher M. (2008). "Development of a congenital adrenal hyperplasia knowledge assessment questionnaire (CAHKAQ)." Journal of Clinical Nursing. 17(3): 1689-1696.*

Appendix H: Written Permission to use CAHKAQ

From: **Irene Mitchelhill (SCHN)** <irene.mitchelhill@health.nsw.gov.au>
Date: Thu, May 1, 2014 at 12:53 AM
Subject: RE: CAHKAQ
To: Kathryn Kraft <kkraft@antioch.edu>
Cc: "jenmking@nsccahs.health.nsw.gov.au" <jenmking@nsccahs.health.nsw.gov.au>

Dear Kathryn,

Thank you for your interest in the CAHKAQ. We would be very happy to have you use it in your dissertation.

Did you get to look at the ***CahPepTalk.com*** website? Please let me know what you think., the passcode for the English version is **"caheng".** It is designed to be a comprehensive education program for families with CAH as well as medical staff & medical & nursing students.

I am actually coming to the USA in June to present about its development, at the Endocrine Nurses Society Conference & International Congress of Endocrinology in Chicago.

I wish you well with your dissertation. What has been your involvement with families with a child with CAH?

Irene

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From: Kathryn Kraft [mailto:kkraft@antioch.edu]
Sent: Thursday, 1 May 2014 02:54
To: jenmking@nsccahs.health.nsw.gov.au; Irene Mitchelhill (SCHN)
Subject: CAHKAQ

Hello,
I am interested in using your CAHKAQ in my dissertation on family health and well being related to families with a child with CAH. I found this website and am more than happy to agree to the conditions listed. I am creating a seminar to be used within a hospital setting. Though the dissertation does not include implementing the program, I would be more than happy to send you a copy when it is completed and published.

Please see the attached form requesting use of the CAHKAQ. Thank you, Kathryn Kraft