

PARENTS' EXPERIENCE HAVING A CHILD DIAGNOSED WITH
MORE THAN ONE GENETIC DISORDER

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of
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of Master of Science in Genetic Counseling

By
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CERTIFICATION OF APPROVAL

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TABLE OF CONTENTS

	PAGE
Acknowledgements.....	iv
List of Tables	vi
Abstract.....	vii
Introduction.....	1
Methods.....	7
Study Population.....	7
Data Collection	8
Data Analysis.....	9
Human Subjects Issues/Protections	10
Results.....	11
Description of Participants.....	11
Emerging Themes	12
Advice by Parents for Healthcare Providers.....	31
Advice by Parents for Other Parents.....	33
Discussion.....	36
Expected Versus Unexpected Diagnosis	38
Social Isolation.....	39
Family Centered Model	41
Novel Challenges With Two Conditions	43
Limitations	44
Future Studies	46
Conclusion	47
References.....	49
Appendices	
A. Recruitment Letter	58
B. Interview Guide	59
C. Description of Genetic Conditions.....	61

LIST OF TABLES

TABLE	PAGE
1. Participant Demographics.....	12
2. Master Themes and Subthemes	13

ABSTRACT

Genetic disorders are defined as disorders that are caused wholly or partly by genetic factors. While the effect of having a child diagnosed with a single genetic disorder has been extensively studied, to our knowledge, no published studies have described the psychosocial impact that having a child diagnosed with co-occurring genetic disorders has on parents. In the presented study, semi-structured interviews were conducted with five parents with children diagnosed with two distinct genetic disorders. These interviews explored the experiences of the parents and the challenges that arise as a result of this unique situation. Interview transcripts were analyzed using the qualitative method of interpretative phenomenological analysis (IPA). Results indicated that these parents share some experiences similar to those previously reported by parents of children with a single genetic condition, such as emotions around the diagnosis, impact on relationships with family and friends and changes in values and view on life. However, the findings also elicited some novel experiences and challenges faced by families of children with more than one genetic diagnosis. These challenges included frustration toward healthcare providers as a result of parents' opinions not being heard, delay in the initial diagnosis and difficulty balancing the medical management of two distinct conditions. Describing and analyzing the experiences of parents raising a child with multiple genetic disorders may help to provide guidance for healthcare professionals to better understand and effectively address issues faced by these families.

INTRODUCTION AND LITERATURE REVIEW

Genetic disorders are defined as conditions that are caused wholly or partly by genetic factors (Nussbaum, McInnes & Willard, 2007). These conditions may be a result of an excess or a deficiency of a whole chromosome or chromosome segments, single-gene defects or a combination of both genetic and environmental factors, otherwise known as multifactorial conditions (Nussbaum et al., 2007). Studies have estimated that approximately 5.3% of individuals will have a disease with an important genetic component by the age of 25 years (Baird et al., 1988). Nonetheless, expectant parents await the birth of a “perfect baby” and experience feelings of grief, shock, anger, shame, and fear at the time of the original diagnosis, when their expectations are replaced by the reality of the birth of a child with a disability (Ablon, 2000; Blacher, 1984; Davis et al., 2006; De Luca, Kearney, Norton & Arnold, 2011; Gravelle, 1997). There is an abundance of literature exploring parents’ experience having a child with a genetic condition. These studies range from analysis of responses to the initial diagnosis, needs during the upbringing of a child with disabilities and coping strategies for dealing with a child’s differences (Beresford, 1994; Fisher, 2001; Graungaard & Skov, 2007; Gravelle, 1997; Pain, 1999).

The time of the initial diagnosis of a genetic condition, or impact stage, is a time of high anxiety and disorganization in a family (Fortier & Wanlass, 1984). This is a shocking and difficult time for parents, with the majority experiencing a range of negative emotions, similar to those of physical loss (Marvin & Pianta, 1996). Beyond

the initial shock of a diagnosis, studies have found that transition can require significant changes in family functioning and structure, and can affect families on a behavioral, affective, interpersonal and cognitive level (Fortier & Wanlass, 1984; Heiman, 2002). It is well documented that families of children with a disability experience greater levels of stress than those without a disability. Studies exploring the causes of this increased stress have identified several contributing factors, including care-giving responsibilities (Plant & Sanders, 2007), the severity of the child's disability (Retzlaff, 2007), financial hardships (Kuhlthau, Hill, Yucel & Perrin, 2005), social isolation (Dellve, Samuelsson, Tallborn, Anders, & Hallberg, 2006) and negative societal perceptions. Parents must reorganize their lives and establish new roles in order to meet the increased needs of their child. The financial burden put on families is two-fold, with parents trying to balance the need for increased time at home to care for a child with increased health-care needs and the need for additional income to pay for medical expenses. For some families, this increased demand is matched with increased strength and collaboration, while for others, the demand exceeds the family's coping capacity, impacting parental mental health (Heiman, 2002).

A family's emotional reaction and ability to adapt to a disability has been shown to be influenced by both external factors, such as the context and nature of the child's condition, and internal factors, such as the process and situation within the family (Power & Dell Orto, 2004; Dellve et al. 2006). Regardless of these factors, however, coping with a child with a disability can be a source of anxiety,

overprotectiveness, and rigidity for families (Lardieri, Blacher & Swanson, 2000).

Various methods of effective coping have been presented in the literature, including finding ways to advocate for the child, learning from others' experience and ways of coping, and seeking information (Fisher, 2001; Graungaard & Skov, 2007; Heiman, 2002). Despite the negative feelings that accompany a diagnosis of a disability in children, studies have found that most parents have some positive feelings of love, joy and acceptance (Heiman, 2002; McAllister et al., 2007). The joys that parents experience are their child not dying, enjoyment of the little things that their children do, new perspectives on life as a result of challenges, and strength in the face of adversity (Kearney & Griffin, 2001).

While the effect of having a child diagnosed with a single genetic disorder has been extensively studied, there are far fewer studies exploring the impact that the co-occurrence of two or more disorders in an individual can have on families. With the increase in technological advancements and the decrease in mortality rates of individuals with genetic disorders, the presence of children with more than one genetic disorder is expected to increase (Roos, 2002). Over the last 50 years, there have been multiple case reports of children diagnosed with more than one genetic disorder, such as a combination of William's syndrome and Klinefelter syndrome in a young boy (Lee et al., 2006), the co-occurrence of Duchenne muscular dystrophy and X-linked Oculo-Facio-Cardio-Dental syndrome in a young girl (Jiang et al., 2009) and a young girl with Down syndrome who was subsequently diagnosed with Ehlers-Danlos syndrome-Hypermobility type (Buterbaugh, Mroczkowski, Shankar &

Visootsak, 2013). These case reports focus on the clinical features and potential future complications faced by these individuals.

While raising a child with a single genetic condition or disability is difficult for families, raising a child with a dual diagnosis can add an even greater degree of difficulty. Studies assessing the care-load on parents of children with multiple disabilities have found the multitude of problems the children face to have a great impact on families. In particular, the recurrent health problems that the children face have a great effect on the burden felt by parents (Tadema & Vlaskamp, 2009). For children diagnosed with multiple genetic conditions, addressing a child's health problems individually does not necessarily have a positive outcome on the child's health as a whole. As a result, studies have emphasized the necessity for a multidisciplinary approach to assessment and management of children with multiple disabilities. This allows for involvement of experts in different areas in order to develop an effective and more holistic management plan. The drawback to this approach is that there is often little collaboration between all professionals involved, therefore working with and coordinating multiple healthcare professionals can overwhelm the family, as they try to incorporate conflicting advice and differing opinions into taking care of their child (Cass et al., 1999). While the potential for increased stressors in the lives of parents of children with multiple disabilities has been acknowledged in the literature, few studies have explored the experience of parents raising a child with co-occurring conditions.

Giangreco et al. (1991) recognized the increased pressures that families of children with limited vision and hearing (dual sensory impairments) face as a result of working with a variety of professionals, and set out to explore the experiences and opinions of these families within the educational arena. Following interviews with these parents, four themes emerged, including wanting a “good life” for their children, fear, frustration and change. First and foremost, parents wanted a life for their children that included a safe and stable home, as well as a supportive social network. Parents indicated fear in planning for the future, due to the uncertainty surrounding their child’s future. Although parents acknowledged their own fear, they also felt that fear often governed the behavior of professionals. They felt that vision and hearing specialists were less likely to be involved in a child’s care if the child had more severe or multiple disabilities, as a result of the professional’s fear in not being an expert and not knowing all of the answers. Parents expressed frustration in working with a large number of professionals who lacked coordination and communication amongst each other. Lastly, parents indicated concern and negative feelings towards change in educational placements.

Similarly, a study assessing the needs of parents of children with a diagnosis of both autism spectrum disorder (ASD) and hearing loss found that parents had difficulties with the comfort level of professionals involved with the diagnosis and care of their children (Wiley, Gustafson & Rozniak, 2013). Families noted a challenge in finding professionals who were knowledgeable about both ASD and hearing loss. Further, parents expressed similar challenges surrounding the ability of

multiple professionals to collaborate. While the above studies provide insight into the unique challenges faced by families of children with a dual diagnosis and the needs of such families, to our knowledge, no published studies have explored the psychosocial impact that having a child diagnosed with any combination of genetic disorders has on parents. Describing and analyzing the experiences of parents raising a child with multiple genetic disorders may help to provide guidance for healthcare professionals in better understanding issues faced by these families, and help to address them more effectively.

The aim of the presented study was to explore the lived experiences of parents with children diagnosed with more than one genetic disorder, in the hopes of eliciting the psychosocial impact of this unique situation. An awareness of parents' experiences with the healthcare team and the impact that having a child with co-occurring conditions has on day-to-day life, may allow for a better understanding of the needs of these families, so that these needs can be better addressed by healthcare professionals in the future.

METHODS

Study Population

Participants in this study included four biological mothers and one biological father of four children diagnosed with more than one genetic disorder manifesting concurrently. For the purpose of this study, a genetic disorder is defined as any condition with a strong genetic component, including conditions of multifactorial and Mendelian inheritance. An attempt was made to interview both parents of an affected child, separately. However, of the four families, only one couple was interested and participated in separate interviews. Participants were not included in the study if they had more than one child diagnosed with multiple genetic conditions, or if they had a child with an unknown diagnosis.

All participants were recruited from the Metabolic and Pediatric Clinics at the University of California, San Francisco (UCSF) Medical Center, and Kaiser Permanente Medical Center, Oakland. Parents of patients of the Metabolic Clinic at UCSF had previously provided the Metabolic Clinic permission to be contacted about future research. These parents were contacted directly by the principal investigator, by telephone, to discuss the study and, if appropriate, to set a date and time for an interview. A maximum of three attempts was made to contact these families. A total of five families were identified and contacted in this way, and one of those families met the criteria and agreed to participate in the study. Additional participants were recruited for the study through referrals from genetic counselors or genetics nurse

practitioners at the UCSF Pediatric Clinic and at Kaiser Permanente Medical Center, Oakland. These potential participants were provided with a letter containing information about the purpose of the study (Appendix 1). The genetic counselors and genetic nurse practitioners gained permission from eligible parents to be contacted by the principle investigator to provide more information about the study and answer any questions. An additional five families were contacted in this way, three of whom were eligible and willing to participate in the research study. This study was approved by the University Institutional Review Board at California State University, Stanislaus on December 11th, 2014 (Protocol #: 1415-069). All participants were verbally informed about the purpose of the study and a verbal consent to audio-record the interview was obtained.

Data Collection

The eligible mothers and/or the father of children diagnosed with more than one genetic disorder were involved in one 30-45 minute semi-structured phone interview. Semi-structured interviews were used in order to allow participants to speak freely and provide rich descriptions about their experience from their perspective. It also allowed the researcher to probe interesting and important areas further. If possible, the mothers and fathers were interviewed separately. Interview questions included 12 open-ended questions exploring the overall experience of having a child with multiple genetic disorders and the effect of this experience on current and future life plans. Specific probes were used for some questions in order to facilitate discussion and allow for elaboration (See Appendix 2 for full interview

guide). All interviews were conducted by the principal investigator and audiotaped with the permission of participants. All names mentioned and other identifiable information were changed to protect confidentiality. The interview guide was pilot tested by one of the participants who was found not to be eligible to participate in the study, as a result of having a child who was formally diagnosed with only one genetic condition. Minimal changes were made to the interview guide.

Data Analysis

Recordings of interviews were transcribed verbatim by a professional transcriptionist. A thorough comparison was made between transcripts and audio-recordings in order to confirm accurate and verbatim transcription. Transcripts were then analyzed using the process of interpretative phenomenological analysis (IPA)(Smith et al., 1999). This method was chosen for this study because there is no prior knowledge of the experience of parents with children diagnosed with more than one genetic disorder and this form of analysis focuses on understanding how participants perceive and make sense of their lived experiences (Smith 2004). While the aim of IPA is to understand an individual's personal perception of an event, it requires that the researcher interpret not only the words spoken by a participant, but also the meaning and emotions behind those words.

To aid with the data analysis process, transcripts were 'assigned' into Atlas.ti, a qualitative data analysis software program. Analysis was conducted in accordance with the steps outlined by Smith et al. (1999). As the first step in data analysis, transcripts were first read through multiple times and annotated with initial

comments. Following the initial step, transcripts were re-read and notes from the initial analysis were transformed into key words or concise phrases, also known as themes, that captured the essence of the comments. Interpretations, associations and connections were made between all emergent themes from a single transcript until themes were organized into clusters and over-arching or subordinate themes were established. As a final step, quotations corresponding to each theme were re-checked to ensure that the subordinate theme accurately illustrated what was originally stated by the participant. This process was repeated for all cases. Subordinate themes from each case were then compared across cases, allowing for the development of master themes for the group of respondents, with each main theme comprising a number of subthemes. The analysis was primarily conducted by the first author, with two of the five transcripts co-analyzed by the second author. The two authors consulted before developing the final list of master themes in order to strengthen the credibility of the emerging themes.

Human Subjects Issues/Protections

All interviews were audio-recorded. In an effort to minimize risks to subjects, all recordings were destroyed at the end of the study. Further, all participant names and other identifiable information were changed in the transcripts. All data and identifiable information were kept in password-protected files, only accessible by the research team.

RESULTS

Description of Participants

With the help of geneticists and genetic counselors at the Metabolic and Pediatric Clinics at the University of California, San Francisco (UCSF) Medical Center, as well as at Kaiser Permanente Medical Center in Oakland, a total of ten families were identified and contacted. Of these ten families, two were unable to be reached by telephone - one did not have a voicemail set-up and the other did not return the researcher's phone calls. Another two of the families did not meet the eligibility criteria. One of those families had a child who did not have an official diagnosis of a second condition and one had a child with a single genetic diagnosis. Four families including four mothers and one father, of the original ten identified were eligible and agreed to participate (Table 1). All participants were parents of children diagnosed with two distinct genetic conditions, none of which were overlapping.

Table 1

Participant Demographics

Participant Number	Relationship to Child	Child's Gender	Child's Age	Child's Diagnoses
1	Mother	M	26 years	HMG CoA Lyase Deficiency (HMG) and Type I Diabetes
2	Mother	M	15 years	Ehlers-Danlos Syndrome, Hypermobility Type (EDS) and Familial Adenomatous Polyposis (FAP)
3	Mother	M	2 years	Dyskeratosis Congenita (DKC) and Mild Hemophilia A
4	Father	M	2 years	Dyskeratosis Congenita (DKC) and Mild Hemophilia A
5	Mother	M	8 years	Usher Syndrome and Methylmalonic Acidemia (MMA)

Note. Four families were interviewed. Both the mother and father of the child with DKC and Hemophilia A, indicated above as participant 3 and participant 4, were interviewed, separately. Please see Appendix 3 for a brief description of each condition.

Emerging Themes

As expected, analysis of the data revealed unique experiences for each family. However, 4 central themes, with subthemes for each, were identified through interpretive phenomenological analysis (IPA) of the semi-structured interviews (Table 2). Many themes were identified through this process, including some experiences that were similar to those reported by studies exploring the experience of parents with a child diagnosed with a single genetic condition. For the purposes of this study, the themes that are delineated reflect experiences that were described by

the majority of the parents, with emphasis on experiences that specifically addressed having a child with more than one genetic condition.

Table 2

Master Themes and Subthemes

Theme 1	Emotions Around Diagnosis
Subthemes	Emotions at the Time of Initial Diagnosis Differences Between Expected and Unexpected Conditions Feelings about a Second Diagnosis
Theme 2	“Me Against the Medical World”
Subthemes	Parents as Experts Parents Being Heard Healthcare Professionals Acknowledging Their Lack of Knowledge
Theme 3	Additional Challenges with Two Conditions
Subthemes	Delay in Initial Diagnosis Balancing Two Conditions
Theme 4	Impact on Life
Subthemes	Impact on Partner Relationship Impact on Relationships with Family and Friends Change in Life View

Note. Master themes, with subthemes for each, identified by comparing superordinate themes from each case across all cases.

Theme 1: Emotions Around Diagnosis

Although the parents interviewed described unique experiences around their child’s diagnoses, all parents expressed negative emotions of shock and sadness at the time of the initial diagnosis. For some parents, despite the initial negative emotions, they were able to identify a “bright side” to having a diagnosis. The three subthemes that fit under this theme include: 1. emotions at the time of initial diagnosis, 2.

differences between expected and unexpected conditions and 3. feelings about a second diagnosis.

Emotions at the Time of Initial Diagnosis

When asked about the time of the initial diagnosis, the majority of the parents described this time as “very emotional.” Three of the five parents expressed feeling shocked, since they had had a normal pregnancy and a normal childbirth.

“Yeah, well, it was my firstborn, and I had a normal childbirth. If anything it was quick for being the firstborn. It was like six hours, and so obviously I didn’t expect anything to be wrong with him. And he – the first four days he seemed like a normal baby. If anything, I would call friends to say hey, he’s sleeping a lot, and they’re like oh, you’re so lucky, he’s a keeper. And it turned out he was actually starting to struggle with his whatever was going on with him, so it was obviously very emotional to have your firstborn get rushed off to the hospital and then go to ICU and then because of his – babies are so tiny and their veins are so tiny, they end up having to put the IV into his brain area instead of like a subclavian or even an arm, they went through his brain. So he had birth pictures basically with an IV in his head. So it was very hard.”

Another parent described not only feeling shocked when her son was first diagnosed, but also being unsure as to what was happening to her son and why he was transferred to the NICU.

“Obviously that process was very like, what you say, it was a shock to us. Because my pregnancy was very normal. My birth was very normal. So I think it was just very mixed emotions as to like I just gave birth to baby and obviously my husband and I had no clue what’s happening to him. We have no medical knowledge whatsoever.”

Even after the doctors had explained the natural history of the genetic condition that her son was diagnosed with, the same mother expressed focusing on the worst possible outcome – death.

“The whole metabolic team sat down with us and they explained to us what was happening and I understand that on their part they have to tell you every

worst case scenario too. And he was in a very critical condition so they were supposed to tell us every worst-case condition. But all I heard was death. All I heard was he's not going to make it, he's not going to make it, he's not going to make it. All I heard was that."

One parent described the conflicting emotions that arise when one's child is diagnosed with a genetic condition.

"So I'm glad that they – I feel like I'm glad that they actually have a diagnosis. I feel relief from that. Less fear of the unknown. But I kind of feel like somebody told my fortune and I didn't want to find out my fortune."

Another parent spoke about the benefit of having a diagnosis.

"I also understand that if that is a diagnosis, that it gives us somewhere to look to. Like where are the headaches coming from and are they coming the EDS, and if that's where they're coming from what can we do to make them less or like how do you work around it. What preventative stuff can we do."

Four of the five parents interviewed explained a progression in their emotions from the time of the original diagnosis. The parent who did not feel different from the time of the original diagnosis explained this to be due to the presence of uncertainty around the prognosis of his child's condition, even with a diagnosis.

Differences Between Expected and Unexpected Conditions

For three of the five parents, one of their child's genetic conditions was a condition that had been previously diagnosed in other family members, and was therefore not a surprise when the child was diagnosed. When asked how the two diagnoses were made in their child, these parents stated one simple sentence about the diagnosis of the expected condition and an extended, more detailed explanation of the diagnosis of the unexpected condition. Not only were the statements about the inherited condition shorter in length, but parents also expressed less concern around

the expected condition. One mother expressed a lack of concern due to past experience in the family and an expectation of a possible diagnosis.

“Well hemophilia was easy because it runs in the family. My dad has it; two uncles with it. So with each kid they were tested as soon as they were born basically while we were still in the hospital. So [my son] was diagnosed with it. It's mild hemophilia. So I kind of knew what to expect seeing my dad go through it. That actually was not scary to find out since we knew we had a 50 percent chance.”

The father of the same child expressed a similar absence of fear or anxiety around his son's diagnosis of Hemophilia A. Although he had not had personal family experience with the condition, he expressed familiarity due to his wife's exposure to the condition.

“I would call it a surprise, but it really wasn't – to us, mild hemophilia was not that big a deal, because my wife had grown up with it with her father, that kinda thing. So we just learned about that condition with [our son], and overall it was pretty straightforward and it was known quantity throughout.”

Another mother who had grown up in a family with multiple family members affected with FAP expressed a similar ease of diagnosis.

“We had the FAP diagnosis done through bloodwork when he was probably four. So that one was easy, it's just a matter of does he have it or does he not.”

Beyond the differences in reactions to the initial diagnoses, these parents expressed more focus and concern around the prognosis and medical management of the condition that was diagnosed unexpectedly and described the unexpected condition to have a greater impact on reproductive decisions.

“So then once [our son] was diagnosed we kind of thought, "Well maybe we shouldn't have anymore." Since I'm a carrier of hemophilia there's still that risk if we have a boy that he has a 50 percent chance of being hemophiliac. But we've decided that that – with the first two boys we already decided that that's not a consideration. It's not a factor in our decision-making. But then

the dyskeratosis that one definitely made us think harder about it in the fact that I don't think I could handle having another child with dyskeratosis. I wouldn't want to put another child through a bone marrow transplant and any other issues going forward in life.”

This was also true for another parent whose first-born was diagnosed unexpectedly with both conditions, one that was autosomal recessive and the other multifactorial. Although both were diagnosed unexpectedly in her first-born, she expressed a lesser degree of concern around the autosomal recessive condition that has a known 25% chance of inheritance for her second-born.

Feelings About a Second Diagnosis

The parents interviewed were specifically asked about the emotions that arose when their children were diagnosed with an additional genetic condition. All of the parents expressed the news to be sad, upsetting and difficult because “of course you don't want anything wrong with your kid.” While the genetic conditions were described as having an impact on the parents, many parents talked about the implications that two conditions have on the child's life. For one parent, a second diagnosis was difficult to accept because his son is now even further limited from life experiences.

“So no, that was a super-huge blow to us. It was devastating because when it comes to your children, you want the best for them, obviously, and that starts with just wanting them to be born healthy. And it felt like not only did this kid already have the – by having hemophilia, he already can't participate in the – and he doesn't have all the other options every other child has, right? He can't go play contact sports. He can't – yeah, well, I guess contact sport's probably the biggest factor. I can't think of what else, but there's a bunch of limitations – oh, drinking. Yeah, he can't enjoy drinking the way maybe normal teenagers – or "teenagers" [laughs] – the way normal adults would, if he makes it to adulthood. I say teenagers because, again, we don't have a high expectation that he's gonna make it past 20, right? So there's just a whole

bunch of life experiences that he's already limited by. Then you throw in DKC and it's like, he just doesn't deserve this, right? And that was just a huge blow to us, where we're powerless to be able to provide him with the kind of life or lifestyle or education or support that – or give him, I guess, the kind of lifestyle that we'd love to be able to give all of our children.”

Similarly, another parent described the additional challenges that arise for the child, himself, as a result of the second condition.

“So once the diabetes was diagnosed, it definitely made our lives much more chaotic and it's hard to see such a young man go through so much in his life. But he's – I mean, he's amazing how he's handled it. Definitely matures an individual quicker than most kids his age, because he's had to deal with so much at such a young age. So it wasn't something we wanted, it would be nice to either have one or the other. Having both definitely complicated his life and his choices.”

One parent explained that having the first diagnosis actually made it easier to accept a second. While she expressed the initial, natural negative emotions that followed receiving a second diagnosis, she described the second diagnosis as another bump in the road. Since she had become used to the hurdles that arose at the time of every developmental milestone, she saw the diagnosis of a second condition as yet another hurdle to overcome.

“So I think pretty much from the birth all the way till now we got hit with things every few months. So it kind of made us that, ok, there's another bump. We have to cross this bump now. Ok. He's not crawling. We have to cross this bump now like physical therapy, motor, occupational therapy, blah, blah, blah, whatever. He's deaf. He's not talking. We got his implants when he was two years old and before that he's not talking. What do we do? Like he's not communicating. I know he can't talk but he's not communicating. So there's another bump. Ok. Let's go for speech therapy. Ok. Then there was another bump of he's not walking. He's like two years old. He's not walking yet. Increase the physical therapy. So that was like those bumps that we hit along the way of his development. So Usher was obviously a very big hurdle in a way but not that oh my god, this is not something that “Oh get a therapy and it will be fixed.” It is something that he is going to lose his vision one day. So that was obviously a very big thing and we went through – I

think at that point we didn't go through the seven or whatever the nine stages there are like denial or whatever. I think denial was way past our list. Like we didn't deny anything."

Theme 2: "Me Against the Medical World"

When asked about their experience with the healthcare team, all parents described their level of expertise around their child's conditions and expressed the importance of healthcare professionals listening to the parents' opinions. This theme is comprised of three subthemes: 1. parents as experts; 2. parents being heard; and 3. healthcare professionals acknowledging their lack of knowledge.

Parents as Experts

Parents described there to be few people who are experts on two conditions and the additional difficulties that emerge as a result of this combination. The parents themselves, however, expressed having a myriad of experiences with their child to provide direction as to how to best take care of them. The mother of an 8-year-old boy with Usher syndrome and MMA described learning by the process of elimination how best to deal with her son's episodes of severe vomiting.

"But later first like three, four, fifth time we got to know how to handle him. Don't move him. If you move him his condition gets worse. And I went back to the doctors and I told them and it wasn't like – obviously it's my theory, what's working with my child."

Drawing from these previous experiences, all of the parents interviewed stated that they had provided suggestions to healthcare professionals about how to best care for their child. When asked about her overall experience with the healthcare team, another mother described the following story.

"– So because of his hemophilia we already knew that nobody especially in

emergency could get [child]'s veins to give him factor. So after he had his biopsy, the bone marrow biopsy, they said, "If he gets a fever we have to go into emergency right away to get blood samples," to find out if it's a blood infection so that they could give him antibiotics right away. So when this happened I called – you know it always happens at nighttime of course. I called the hematologist and they wanted us to go to emergency. Since we've been to emergency numerous times for the hemophilia to get factor with zero success and hours and hours and hours of people trying to poke and prod him I suggested, "Why don't I just come in to the clinic tomorrow morning." It opens at 9:00 and they have an infusion center. So these nurses deal with giving chemo and I.V.s and draw blood all the time and they're much better at it than the E.R. nurses. They said, "Absolutely not. You have to go to emergency." So in that case it took about seven hours for them to draw one vial of blood because they kept losing the vein or not being able to find it so it would pop."

Using her experience from past instances at the hospital as an indicator, she provided a suggestion that she felt would result in the best outcome. Not only did these parents describe themselves as experts, but in one instance, the primary healthcare team that one mother and her child worked with, had told the healthcare professionals working in the emergency room about the mother's level of expertise.

"They would specifically say to the staff that was treating him at whatever ER he was at, listen to the mom, she knows what she's talking about. She's experienced this first hand."

Parents being heard

All of the parents who were interviewed discussed the importance of their opinions being heard. One mother had had a positive experience with her son's healthcare team, as they listened to the parents and respected their opinions. All other participants, however, described the frustration that's felt when healthcare providers don't listen to what the parents have to say. This was especially frustrating for parents as they felt they knew what was best for their child.

“To be honest I think I'm pretty calm. I listen. I could see it being in other situations as a parent freaking out and not really understanding what's going on but in my case since I'd gone through this so many times before I actually knew what I was talking about but nobody listened.”

Another mother described healthcare providers not listening to what she had to say about her son's difficulty with receiving stitches, as a result of his EDS, because they didn't believe her.

“And I'm like, you need to go back in your office, read the computer on his file and read what it has to say, because this is what happens. I'm not pulling your leg, I'm not trying to waste your time. It's like we have to be salespeople to stand up and make like, they don't listen.”

For one mother, the frustration ran deep, as not only was her voice not being heard, but also the voices of the metabolic team taking care of her son. The emergency letter that was formulated by the child's metabolic team, including information about the medical management of the child in emergency situations, was disregarded on a number of occasions.

“His girlfriend at the time had called and said he's got to go to the hospital, he got on the phone with me and he said I'll be okay, I have my letter, and so I didn't take my time, but I drove from Walnut Creek to Davis and it took the usual hour, hour and ten minutes to get there. And the only thing they had done for him was given him an anti-vomiting medicine. They hadn't given him the push of D50, they hadn't done anything. And he was starting to just crash and so I basically became a crazy woman and started yelling at the doctor and saying didn't you even look at the letter, and I can't believe you didn't look at the letter, this is what this is here for.”

The same mother explained that as a result of the challenges that she faced dealing with healthcare professionals, she “always felt like it was [her] against the medical world.” The frustration that the participants described feeling was not the same frustration that is felt when any friend or stranger does not listen to what you

have to say. The real root for these emotions was the fact that their child was not receiving the best care because of the lack of listening. As one mother stated, “they don’t listen to you and they do what they want when they're stitching up, then your child pays the price, so.”

Healthcare professionals acknowledging their lack of knowledge

Since the individual genetic diagnoses are rare, and the combination even more so, parents had difficulty finding experts on their child’s conditions. Four of the participants expressed the importance of healthcare professionals acknowledging the fact that they are not experts on the particular condition or problem at hand. One mother described having a positive experience, in which healthcare professionals acknowledged the gaps in their knowledge and the fact that the parents are the most knowledgeable about their child.

“Cause if this is working – ‘cause whatever we are suggesting is not working. So if this is working with him, yes, continue that. So it’s like very – “Ok. We know we are doctors. We’re professionals. But if something you know – you know your child and you know that this makes him more comfortable then yeah, do that.” So that was very good thing that one of them figured out was like “Ok. Yeah.” Even though they are professionals but they acknowledge that we know our child better.”

The other three participants, however, had a very different experience, with healthcare providers acting as experts in an area that they lacked knowledge. These parents expressed negative feelings in regard to these experiences. One mother felt that it would be beneficial if healthcare providers could acknowledge a lack in knowledge.

“You know, it's frustrating. I understand with all their years of extra schooling, I have a four-year degree and I understand that medical

professionals take great pride in giving up so much of their lives to help the community they want to serve through their medical school. But it would be helpful if they could take a step back and not get so tied up in the fact that they're – I mean, it's not true with all doctors, but some of them tend to have that god syndrome, where they think that they know it all because they've done all this schooling, and obviously with something like HMG, they don't know it all.”

Another parent expressed similar negative feelings.

“And it was very clear to us that there was a lot of, quote-unquote, experts – not necessarily on DKC, of course, but experts within [hospital A] that, rather than say "I just don't know anything about this, and I'll have to defer those questions," would give us answers that were vague and loaded with BS, and I just think that was the most insulting thing that could've happened to us.”

Theme 3: Additional Challenges with Two Conditions

There are added challenges raising children with more than one genetic condition. When asked about the initial diagnosis of their child's genetic conditions and how parents imagine life would differ if their child were diagnosed with a single genetic condition, parents described these added challenges. Two subthemes fell under this theme of additional challenges with two conditions. They are 1. delay in initial diagnosis, and 2. balancing two conditions.

Delay in Initial Diagnosis

Having more than one genetic condition is extremely rare; as a result, the diagnosis of a second genetic condition is not expected by either parents or medical providers. The majority of the parents interviewed discussed attributing the additional symptoms from the second condition to the first condition. The father of a 2 year-old boy with Hemophilia A and DKC describes his experience in the following statement.

“So we started out with conditions that we actually thought were hemophilia. So we were like, ‘Wow. These are like – I don't remember –‘ [my wife] was

like, ‘I don't remember my dad getting these huge goose eggs when he bumped his head, and bruising really, really, really easily.’”

Another mother described a similar experience of trying to connect the additional symptoms to the initial diagnosis that had been made. In her experience, some of the new symptoms overlapped with symptoms that can be seen in individuals with the original diagnosis that her son was given. While it was determined that her son is profoundly deaf, the lack of alarm around the additional “signs” resulted in a delay in determining the etiology of his deafness.

“Because one of the major things – so there were a lot of signs that we – once we heard the Usher syndrome but there were a lot of signs that could happen because of his metabolic disorder too. So it was very easy to ignore why he was deaf. Because there were a lot of signs that were overlapping with his metabolic disorder. It’s hard to pinpoint that oh he’s not – the balance is not because of metabolic disorder. It’s because of Usher because it’s hard to do that because balance is one of the main things in metabolic disorder too. The physical condition, the motor condition, that’s one of the main things in metabolic condition too. It was very hard to pinpoint that.”

While the parents described not being attuned to the cause of the additional symptoms that were seen, one parent described hesitation on the part of the medical providers to diagnose the child with a second genetic condition, due to the unlikelihood of this event occurring.

“And they're like, ‘Okay, he's got hemophilia.’ They didn't even really think that it was possible that he could have anything else. That's how rare it is to have two genetic disorders, right? So they weren't even – at first they came out, ‘Couldn't be leukemia or any of these things. That's just crazy. Nobody gets both of these.’”

Balancing two conditions

Almost all of the parents interviewed explained the extra challenges that result from trying to balance the management of the first condition with the management of

the second. One mother described the additional difficulties as “throwing all these little wrenches in there”, further complicating care. One mother described having to “re-learn” how to manage her son’s health after the diagnosis of the second condition. For him, balancing the effects of the two conditions could no longer be managed at home, meaning that he had to go to the hospital every time he fell ill. She provided a detailed description of the conflict between the two conditions.

“But with the diabetes, once you are – have a pancreas that's not producing insulin that you need and you're taking insulin to act for your pancreas, and then you vomit and you lower you blood sugar and you're already on insulin to lower your blood sugar, then it's just too tricky a slippery slope to actually control low blood sugar, normal blood sugar and also have HMG on board. So once the diabetes was diagnosed, it definitely made our lives much more chaotic and it's hard to see such a young man go through so much in his life.”

Another mother explained her experience balancing FAP and EDS, two seemingly distinct conditions.

“And with having all those scopes and stretchy skin, it's – it takes them twice as long to scope us, having an upper and lower GI, because of the Ehlers-Danlos, because it's like trying to thread a wet noodle over the top of a – your fingers, you know, and you can't see where you're going. And so it's – everything takes longer during a procedure. And God forbid you have to have stitches from having your colon worked on or whatever, then you have the whole suture rejection thing, which is prominent in our family, or stitch spitting, you know, so there's all these little things. We have issues with the anesthesia, so they have a hard time keeping us under, and then they have a hard time waking us up. So it's, um, there's a lot to balance with the two diagnoses when it comes to actually having to take care of the FAP.”

One of the parents who did not believe there to be added difficulties balancing the two conditions explained that the combination “just broadens the list [of problems] rather than making it deeper.” Interestingly, he was the participant whose wife was also interviewed, and she reported added difficulties associated with her son

having both conditions. While many parents described the added challenges that can occur from their point of view, one parent explained one situation in which the endocrinologist working with her son acknowledged the added challenges that arise as a result of the two conditions working against or along-side each other.

“When [my son] was at [hospital B] there was an endocrinologist that came and visited him and tried to basically work with the diabetes portion of it, and he was just astounded by how many differences there are between the two disorders and how tricky it is to deal with the protocol to deal with both of them at the same time, so and he's a medical professional.”

Theme 4: Impact On Life

It has been previously reported that having children with special needs has an impact on life at work and at home. Four of the five parents interviewed described that their child's conditions have had an impact on relationships and have resulted in a change in their view on life. The one parent who did not feel that there was any impact on her life described this to be a consequence of the simplicity of her son's conditions and the normalcy in her day-to-day life. Three subthemes fit into this theme of impact on life. They are 1. impact on partner relationship, 2. impact on relationship with family and friends, and 3. change in life view.

Impact on Partner Relationship

All of the parents interviewed reported that their child's genetic conditions did not have a negative impact on their marital relationship. The parents who were still married described their relationship as successful prior to their child's diagnoses and did not feel that their child's conditions impacted the strength of this bond.

“Thankfully with [my husband] things – we had a great relationship before and thankfully this has not affected it. I can only – I've heard that having a

sick, critically ill or losing a child can cause divorce and lots of marital issues but we've actually been great, very supportive of each other and helpful.”

The husband of the mother quoted above described a similar absence of an impact on his relationship with his wife. He drew parallels between his life at home and the lives of any other parents with two children.

“I actually – I am very pleasantly surprised that we have survived and thrived, or at least – I don't know if "thrived" is the right word. We're not better off because of this. But I don't feel at all encumbered by this at this point in time, other than I will say – so your question was both at home and at work. So at home, I would say we're about as normal as I would expect a family with two kids to be with this sort of side thing of this hemophilia and DKC kinda lingering out there.”

Another mother stated that not only had the conditions not had a negative impact on her relationship with her husband, but had in fact strengthened their relationship. She credited this bond to the strength and understanding that they provide for one another.

“Like this is more of we support each other and we understand that we're both going through something very close to our heart. It's our child. And obviously if I'm feeling something I'm feeling it because of him [our child] and he understands that and he gives me credit. And same thing, when he's going through something I know that he cares about him. And he loves him and that's why he's thinking about all of these things and at that point I have to be a stronger person and be like “I understand what you're going through and – but we did come a lot and he will come through that too.” So yeah. We support each other a lot. I think yeah, having a kid with special needs if anything it's strengthened our relationship.”

One parent expressed being able to understand a strain on relationships for couples that have “children with more difficult problems”, but she did not feel that her son's diagnoses of FAP and EDS were significant enough to have an impact on her relationship. One mother, who is no longer married, also did not feel that her

child's conditions had an impact on her separation from her ex-husband. She described her divorce to be a result of factors outside of her son's diagnoses.

“I can't say that the disorders have affected my relationship with my ex-husband because that was really always fractured. I mean, once you have an alcoholic in the mix, there's so many things that if they're not willing to deal with, there's nothing you can do about it. I mean, he's been sober now 14 years, so he finally did find his roots and figure out that this isn't something he can handle alone, but I don't think necessarily – I don't look at the disorder as a crutch to blame his alcoholism on, even though I think he used that for his own benefit.”

Impact on Relationship with Family and Friends

While every parent shared distinctive experiences when discussing the impact that their child's conditions have had on relationships with family and friends, four of the five parents spoke about the negative impact that this has had on relationships.

One mother described a shift from fostering relationships with friends to focusing on her immediate family, comprising of her husband and her son.

“So when it came to few years later when [our son] came we kind of grew apart because they didn't really understand what we were, what we were going through. And when I say we had to put our social life on hold it really meant that we had to like cut cords with them and be like, ok, this is not working out. And to me at that point that life didn't matter anymore. All that mattered was my husband, [my son] and I.”

All four of these parents associated the distance between themselves and friends to be a result of others not understanding just how difficult it is to care for a child that has significant health concerns. One parent felt that this lack of understanding stemmed from the fact that “people take for granted how easy it is if you have children that have absolutely nothing wrong with them.” She described this as a contributing factor to the wedge that was driven between her and her relatives.

“I have two brothers that they kind of take for granted what I go with – what I deal with on a daily basis because they have children that don’t have anything wrong with them. And so maybe that’s why we’re not as close, I don’t know. I love my brothers but I don’t talk to them very often, and they don’t talk to me. And on the anniversary of my daughter’s death, I’m surprised that they don’t reach out even with a card. I mean, I know it’s been a long time, but it’s like they take that for granted, as well.”

Another parent described family and friends as having sympathy but didn’t feel that people could truly conceive what a parent with a sick child experiences unless they have been in the situation themselves. He expressed others not only not understanding the impact that this can have on one’s life, but also not understanding the choices that these parents make.

“And I think it’s impossible for people to really understand what the impact of a sick child is and why you make certain decisions, career-based or otherwise, and the choices that you make. It’s like impossible to, I think, fully understand that unless you’ve done this and seen what the impact is.”

One mother described a similar experience of people not understanding but expressed an overall positive experience with family and friends.

“As for other family and friends I feel like some kind of just don’t get it and – but for the most part I’d actually say we’ve had great friends and family that have tried to help out as much as possible. I have one friend that took all my plants. She’s offered to actually take [my second son] for - while [my son with DKC and Hemophilia A] was in the hospital for seven weeks, which thankfully we didn’t have to do that. We’ve had family offer to come down and help.”

Due to the rarity of having a child diagnosed with two genetic conditions, one parent described the lack of understanding to be explicitly true for parents that have children with more than one genetic condition.

“So, you know it’s – yeah, I would say the parents like – especially with two genetic conditions, nobody, nobody knows what they’re going through. I might as well just say nobody. Because for them to find somebody else is

gonna be nearly impossible. So I'm just gonna say nobody understands what they're going through.”

Change in Life View

Both when asked explicitly and at their own free will, the majority of the parents explained that their child’s diagnoses have had an impact on how they live their lives every day. While slight variations existed between parents’ views on life, three of the parents expressed that fear about their child’s future and a potential shortened lifespan has made them embrace every day, like it’s the last. When asked specifically about any benefits to having a child with more than one genetic condition, one mother stated the following:

“I guess the only thing I could think of is on a day to day basis I probably cherish my kids more, realizing that anything can happen at any time. I could lose one or both of them. It makes me want to enjoy today more.”

Another parent expressed similar sentiments, describing his son’s genetic conditions as a motivating factor to live life to the fullest.

“But there's still this, I think, willingness to sort of embrace the day with both boys. So we don't give – there's no favoritism here, really. But to embrace where we can, either taking these boys out to go see something that's culturally interesting, or put them in nature; or camping might've waited a couple years before, but now – until we had maybe room to clean the equipment or store the stuff. But now we might raise the priority of that and say, "No, we've gotta go camping this year, because we want both these boys to experience that sooner rather than later." And maybe a little bit of that is motivated because of the fact that [my son] may not have too many laters. So I think that there's certainly an awareness enough now to try to sort of seize the day, if you like, with these guys, more so now than we might've if they were both – had neither condition or just one condition.”

One mother described the importance of quality time and building memories, as her son can hold onto these memories if he loses his vision one day.

“Like that’s – and I’m enjoying life now more than anything. Like every day for us is like we literally spend every day thinking that if tomorrow he is going to lose everything he’ll have the memory of today. And those memories are not going to come from buying him Legos or buying him – to me yes those are important, toys, buying him things, but spending time going on – just like we clean back yard together. This for me is a quality time even though he’s working but he loves it.”

The same mother explained that her son’s conditions have actually changed who she is as a person and have resulted in her changing her career path.

“I would say that, yes, before [my son] life pretty much revolved around me. I was a very self-centered person. I would say that I was a selfish person too. Yes. After [my son], yes, it’s just the complete opposite to who I am today. Like that’s something that I own up to that if I didn’t have a kid that has special needs I would be a totally different person today that like I wouldn’t be who I am today. Today I actually changed my major. I’m going back to school to work with special need kids.”

Advice by Parents for Healthcare Providers

Parents were asked whether they had any advice for healthcare professionals based on the past experiences that they have had working with them. Three of the five parents spoke about the importance of healthcare providers acknowledging the fact that they do not know everything there is to know about the child’s conditions, either separately or in combination, and listening to what parents have to say about their child’s conditions. One parent spoke about the internet contributing to the extent of knowledge that parents have around their child’s conditions and the importance of healthcare providers being aware of the fact that in certain circumstances, parents may know more than the doctor about the ins and outs of the child’s conditions.

“And the reality is that [my wife] and I are now probably somewhere in this top 10 percentile of people that really understand this disease and understand how it affects children, and we’re not doctors. But the reality is that we’ve had to do a ton of research; we’ve had to come up to speed on this; we understand

the circumstances around it, can recognize symptoms. And I think healthcare professionals have to realize that in the age of the Internet, and with people that certainly know how to do research and can figure this stuff out, that in rare diseases, they're gonna come across I think parents that could have quite a bit, if not more education than they have, and that we recognize – there was times at [hospital A] where we were already ahead of the curve in terms of our understanding of the disease than the person we were talking to. And we can smell it – excuse my French here, but we could smell the bullshit on these guys a mile away.”

One parent expressed having an overall positive experience with her child’s healthcare team and did not have specific advice to offer. She did mention the importance of healthcare providers listening to the parents and being available to parents when they require assistance. Further, she expressed the importance of healthcare providers normalizing the parents’ feelings and encouraging parents about the care that they are providing for their child.

“Sometimes literally I go to an appointment or sometimes there are moments when I go like “I can’t deal with that anymore” and I just pick up the call or I call his dietician or I call his doctors and they take their time to listen. Like it’s not “Oh you know what? Make an appointment. Or he’s not doing – he’s fine.” No. Nothing like that. I’ve never heard of that. It’s always that “I understand, like I know it’s hard” and it’s just always that encouragement that “I know you’re doing a good job” and that – I think at that moment that’s all I like to hear is I’m doing – ‘cause more of anything if I’m in the situation where I’m getting too emotional it’s because sometimes I think that I’m not doing a good job. And I think they understood that.”

Another parent gave perspective on how healthcare professionals can approach diagnosing individuals with genetic conditions.

“You know, I think it's just the idea that you can't stick a patient in a box and say well, you've got these five symptoms but you don't have these five. I mean, it's – it could be that those five symptoms won't develop until you're older or you had them when you were younger and now they're not present because of whatever. So we don't always fit into the box that is the diagnosis

or what it looks like to be diagnosed with something, and you just have to like, kind of stand back and say okay, if this was my mom or my son or my whatever, and these things were happening, if they can make it personal, then it makes it a little easier, it seems, to then be understanding.”

Along the same lines, one mother suggested that healthcare professionals not base their expectations of a child’s abilities on what has been previously reported for other children diagnosed with the same condition.

“The – when we were back at Duke, they had to do – they wanted to do an MRI of both of our children's brains because there was information at that time that said that HMG thins the myelin sheath, thereby reducing the patient's ability to do well at certain subjects like math, because their myelin sheath was thinner and didn’t support the brain as much, and that turned out not to be true, because I mean, [my son] is exceptional at math. And he has an exceptional brain and exceptional student. So you kind of wish that you weren’t given information that was so detrimental or doomsday prophecy type of thing, because a lot of people, if you didn’t feel educated, would buy into that and just assume that's true.”

Advice By Parents for Other Parents

As the final question, parents were asked what advice they would give to other parents who have a child diagnosed with more than one genetic condition. All of the responses provided were different and represented the unique experience of each parent; however, all represented the parents’ individual methods of coping. Two of the parents spoke about the importance of communication with the healthcare team, in terms of making sure that they are aware of the intricacies of the child’s two conditions and in making sure that the child has the “right medical staff on their team.” One of these mothers also provided advice about how to cope with the news of a child’s diagnoses by stating, “you gotta take a step back and realize it could be

worse”, meanwhile acknowledging that one’s ability to do this depends on the severity of the disability or disorder that a child has.

Another parent shared an effective method of coping that helped him get through the days when his son was ill.

“– but the truth is that, you know taking one day at a time is the only way to get through this and the only way to keep from just having the frustration create, like for instance, arguments or conflict within the relationship itself. And of course that carries over to how you treat your children if you're not careful and the whole bit. But if you focus, you know bring that focus into just getting through that day and then focus on the next day. You know you really – you can get through it. That is – I think for us minimized the amount of, again, conflict within the relationship itself. And it minimized kinda the emotional overload that occurs with this as well where you're like, oh, my god, you feel helpless. How am I gonna get through this?”

One mother expressed the importance of accepting the child’s diagnoses, meanwhile keeping hope, as it is not until this acceptance when someone can be proactive about caring for their child.

“The biggest advice I would give them is accept it. Accept it and then I think the acceptance part is the most hard part for parents because before accepting anything you cannot do anything about it. You can’t really take any further steps because you’re still in denial. And sometimes you’re not even in denial. Sometimes you’re waiting for some miracles to just come in. So that hope – and I understand. I’m not saying hope is a bad thing. Hope is the biggest thing and you have to have hope and we still have hope. But accepting in a sense that I have hope that someday something is going to – some technology is going to kick in or he won’t lose his vision or even if he might lose it they’re going to do something or he might have some vision back. Like there’s always that hope but in the same time we have accepted that he has Usher’s and we’re doing something about it. We have accepted that he has metabolic disorder and we’re doing something about it. We have accepted that he was deaf and we got his implant, went to speech therapy. Like I think the biggest thing is acceptance. Accept that your child has it. Hope is a good

thing. Keep the hopes up but don't lose the precious time because in early years that's the most precious time. Don't lose that. Accept it."

Another parent spoke about self-care and having personal goals outside of caring for one's child as a method of not focusing all thoughts on the child and his or her health.

"But I think it helps my and my husband's health not deteriorate because if you don't have energy as a parent you're not going to be able to help your child. You have to help yourself. So we've been waking up before the kids as much as – unless there's a hospital visit in the middle of the night. But every morning we wake up, do our cardio, do our weights and then are ready for the day. It's actually increased our energy level. It's made us feel better about ourselves so that even – because it can be depressing at times when things are not going well for your kids. So having your own personal goals and even though your child sometimes feel like your whole life revolves around them kind of just to get out of that and set your own goals."

DISCUSSION

Semi-structured interviews with the parents of children with more than one genetic diagnosis revealed four main themes: emotions around diagnosis, “me against the medical world”, additional challenges with two conditions and impact on life. Although the aim of this study was to address the psychosocial impact that having a child with more than one genetic disorder has on parents, several of the findings in the study support concepts previously established in the literature pertaining to the experience of parents with a child diagnosed with a single genetic condition. What this study adds is information about unique challenges and life experiences that families face as a result of the additional genetic diagnosis, as told by parents.

As has been previously described in the literature, the parents interviewed described the time of the original diagnosis as a “very emotional” time and described experiencing feelings of “shock” (Ablon, 2000; Blacher, 1984). Three of the mothers interviewed expressed their negative emotions being exacerbated by the fact that they had a normal pregnancy and birth, and did not expect their child to be diagnosed with a condition. In assessing the experience of parents with a child diagnosed with cystic fibrosis, Grob (2008) compared the reaction of parents to a diagnosis following seeing their child suffer with an unknown illness to the reaction of parents to a diagnosis that was made unexpectedly, right after birth. Similar to the feelings that were expressed by interviewees in the current study, Grob found that the parents who

had no reason to think that anything was wrong saw the diagnosis as a devastating surprise that they were unprepared to face (Grob, 2008).

Despite the negative emotions that were experienced at the time of the original diagnosis, the majority of the parents interviewed described the positive aspects of having a child with special healthcare needs. As was described by Kearney and Griffin, participants in the current study expressed the joy that comes with the little things that their children do and their newfound approach to living every day like it's the last (Kearney & Griffin, 2001). In addition, one parent openly shared the tremendous positive changes in her own personal values and lifestyle, as a result of her son's conditions.

Previous studies have found that parents experience fear and anxiety when the progression of their child's condition is unknown and clinicians are not able to provide parents with clear expectations about a child's prognosis and future life (Ablon, 2000). Although uncertainty around the prognosis of their child's co-occurring conditions was described by the majority of parents, surprisingly, only one of the parents interviewed in the current study discussed the negative emotions surrounding this unknown. While it's difficult to evaluate reasons why parents in the current study did not express emotional distress around their child's uncertain future, it's possible that this is a result of these parents' tendency to focus on the day at hand, due to fear of their child's shortened life expectancy and inevitable death.

Expected Versus Unexpected Diagnosis

Interestingly, the participants with a family history of a genetic condition, such as Hemophilia A and Familial Adenomatous Polyposis, expressed a diminished level of concern around the known condition running in the family, in comparison to the condition that was diagnosed unexpectedly. In all of these cases, the condition that was diagnosed unexpectedly also happened to be the second condition to be diagnosed. These parents described being aware of the chances of their child being diagnosed with the first condition and saw the diagnosis process as a binary outcome – either their child is affected or not. Although they were not explicitly asked, the parents interviewed in the current study did not express negative emotions of guilt and responsibility around the diagnosis of an inherited condition, as has been previously reported in the literature (James, Hadley, Holtzman & Winkelstein, 2006). In fact, one mother and her husband described greater reactions in response to the diagnosis of the unexpected condition that resulted from a new mutation in the child, in comparison to the inherited condition. As described above, this may be a result of the shock accompanying a diagnosis that is made unexpectedly (Grob, 2008). In the case of this particular family, with a son with Hemophilia A and DKC, another possible explanation for the difference in level of concern is that DKC is a more rare and severe condition, that carries with it many unknowns.

The same discrepancy in the degree of concern was seen when the parents were discussing the medical management of their child's conditions, as well as the impact that these conditions have had on reproductive decisions. For one mother,

although both of her son's conditions were diagnosed unexpectedly, she expressed greater concern for the diabetes, which was diagnosed when her son was 13 years old. After being educated about the autosomal recessive nature of her son's HMG CoA Lyase deficiency, the same mother described an awareness of a 25% chance of inheritance, and therefore an absence of concern, for future offspring. Following the diagnosis of diabetes in her son, however, she described greater worry. Although diabetes is inherited in a multifactorial manner and the mother's second child is not exhibiting any symptoms suggestive of diabetes, the mother's level of concern and worry have made her seek annual screening for diabetes for her second child. There are no previous studies assessing the reasons behind the difference that was observed in the current study, however, since managing two complex conditions is an emotionally overwhelming concept, it would be interesting to evaluate whether parents concentrate all efforts on a single condition as an unconscious means of coping.

Social Isolation

Previous studies have stressed the importance of the maintenance of relationships with family and friends as an effective method of coping (Hainsworth Eakes & Burke, 1994). However, as has been presented in previous literature, and described by four of the five parents in the present study, parents of children with disabilities and chronic illnesses perceive themselves as socially isolated (Beresford, 1994). There are many possible explanations as to why parents feel alone in raising children with special needs, however, participants described others not understanding

what it's like to raise a child with significant health problems as the primary hindering factor in the maintenance of friendships. Some of these parents described an effort on the behalf of friends and family, as they provided sympathetic responses. However, as one parent stated, "it's like impossible to, I think, fully understand that, [what it's like to raise a child with a condition], unless you've done this and seen what the impact is." The same parent spoke specifically about the added challenge of having a child with more than one genetic condition, as the rarity of the situation further hinders others from understanding.

While participants described the strain that their child's conditions have put on relationships with family and friends, all of the parents expressed an absence of a negative impact on marital relationships. Even the mother who has since divorced her ex-husband described her relationship with her partner as fractured prior to her son's diagnoses and did not blame the downfall of her relationship with her spouse on her son's conditions. There is conflicting data on the impact that having a child with special healthcare needs has on marital relationships. Some studies have found a higher rate of divorce in families with a child with a disability, in comparison to the general population (Hartley et al., 2010), while other studies have found that raising a child with a disability may exacerbate marital problems that are already present, but well-functioning couples adapt well (Benson & Gross, 1989). In the current study, parents described their partner's support and understanding as the primary reason for the strength in their martial relationship. Since these parents also described a lack of understanding from other family and friends, it is possible that these couples found

comfort in having someone who fully understood what it was like to raise a child with a rare combination of conditions. One mother expressed that her son's conditions had in fact strengthened her relationship with her husband.

Family Centered Model

The primary concept that was discussed by all parents, multiple times throughout the interviews, was that of the parents feeling like the experts on their child's conditions, as they have had many experiences with their children falling ill or being in the hospital. As a result of these experiences, the parents exhibited confidence in their knowledge and provided recommendations to healthcare providers on how to best care for their children. Previous studies have described similar sentiments from mothers who have had extended time caring for an ill child (Leff P & Walizer, 1992; Goldfarb et al., 2010). Acknowledgement that parents have valuable information, as a result of their extensive experience, has resulted in a shift towards family-centered care for children with special health care needs, as opposed to the traditional, medical model (Brewer, McPherson, Magrab & Hutchins, 1989). In contrast to the medical model that assumes the professional is the expert on the medically defined problem, this philosophy recognizes parents as experts on their child's needs and therefore promotes a partnership between parents and professionals for providing optimal care for a child (Rosenbaum et al. 1998). Despite the acknowledgement of the benefits of family-centered care and the shift towards this model over the past decade, the participants interviewed in the current study described healthcare providers who did not listen to the parents' suggestions. These

parents expressed frustration toward healthcare providers as a result of not being heard.

It is difficult to assess factors shared by the group of parents interviewed for the current study that contributed to the level of frustration expressed as a result of not being heard by healthcare providers and not being involved in the care of their child. However, a previous study exploring factors influencing the extent of parent involvement in decision making in the care of their child with cancer found that parents' knowledge of their child's condition and their confidence in this knowledge, as well as parents' perception of physician knowledge influenced degree of parent participation in the decision making process (Pyke-Grimm, Stewart, Kelly & Degner, 2006). Based on these findings, it's possible that the many experiences that these parents have had in the hospital, as well as the limited amount of physician knowledge around the combination of the children's conditions contribute to the parent's desire to play a more active role in the decisions made around their child's care.

Another contributing factor could be related to the additional difficulties that come with having more than one condition, such as the need to be followed by multiple specialists. In their study, Law et al. found that parents perceived the services that they received as more family centered when they received services from fewer places and when their child had fewer health and developmental problems (Law et al., 2003). While parents should be consulted on the degree of involvement

that they desire, the findings in this study suggest that parents of children with more than one condition may desire greater involvement in the care of their child.

Novel Challenges With Two Conditions

While a small number of studies have assessed the needs of parents of children with two specific conditions, such as, ASD and hearing loss, no studies have explored broadly the experience of parents with a child diagnosed with a combination of disorders. Interviews with the parents in the current study identified two challenges that were consistently discussed by the majority of participants: delay in diagnosis of a second condition, due to the improbability of having two conditions and difficulty of balancing the medical management of the two conditions.

Since having two rare genetic conditions is extremely infrequent, both parents and healthcare professionals were described as unsuspecting of a second genetic diagnosis when additional symptoms were observed. Two parents explained trying to associate the additional symptoms that were seen with the first diagnosis, and another parent explained hesitation on the part of the healthcare providers in making an additional diagnosis. A study exploring “take-home messages” described by physicians following a diagnostic error found three lessons learned by physicians that address this issue: 1. Look beyond the initial diagnosis, as one should never assume that only one condition explains everything, 2. Reconsider diagnosis if patient follows unexpected course, and 3. Consider uncommon conditions (Ely, Kaldijan, D’Alessandro, 2012). The experience of the parents interviewed in the current study

serves as another reminder to healthcare providers to keep an open mind to the possibility of rare circumstances.

In discussing the differences that parents would expect had their child been diagnosed with a single genetic condition, almost all of the participants described the difficulty of balancing the management of two conditions. For one of the parents, this difficulty was a result of both of her son's conditions being metabolic conditions. For two other parents, however, one of the conditions was seen as an obstacle in the management of the other condition. For example, for the boy with DKC, having hemophilia A complicated venipuncture and resulted in extended hours spent at the hospital, and for the boy with FAP, having EDS complicated having colonoscopies and receiving stitches following having an ostioma removed. While Wiley et al. did not specifically report on the difficulties of managing children with ASD and hearing loss, they too discussed the added challenge of having the two conditions when trying to appropriately evaluate the child's development (Wiley, Gustafson & Rozniak, 2013).

Limitations

There are several limitations with this study and methodology. This study is subject to self-selection bias. The participants who agreed to be interviewed for the study may have certain personality traits that make them more resilient and willing to discuss their experience. Further, since a small sample of parents was interviewed, and none of the children of these parents had conditions affecting physical appearance or mental capacity, the results are not generalizable to a larger

population. However, the goal of this qualitative study was to elicit rich, in-depth insights from the parents' perspective and did not aim to make more general claims.

Typically, IPA uses purposive sampling, using a closely defined group of participants who all share the experience under discussion. Although all of the parents in the current study had a child diagnosed with two distinct genetic conditions that affected the child's health but did not have an impact on physical appearance or mental capacity, all disease combinations were different from one another. Additionally, at the time of the interviews, all of the parents had been coping with their child's conditions for varying lengths of time. The differences between the different conditions and the different stages at which the parents were at in the coping process may have compromised the ability to effectively explore the common experience shared by all of these parents.

While one father participated in the current study, most of the parents who participated were mothers. Due to the small sample population, no conclusions could be drawn about the differences in experience between mothers and fathers. Additionally, due to time restraints, telephone interviews were conducted as opposed to face-to-face interviews. The absence of nonverbal cues may have compromised rapport building and the interpretation of responses. Lastly, as for all IPA studies, the researcher played a role in generating, analyzing and interpreting the data in this study. As a result, it is possible that a different researcher, with a different background and interests, would have developed a different interview guide, pulled different themes and threads, and interpreted the results differently.

Future Studies

Future studies interviewing parents of children with more similar conditions, such as all parents with a child diagnosed with two metabolic conditions, may aid in drawing commonalities between the experiences of families. Further, studies should be conducted assessing the experience of parents with a child diagnosed with two conditions, one of which affects physical appearance or mental capacity. This may provide insight on the differences experienced by the three different groups of parents, therefore aiding healthcare providers in providing more tailored care to these families. Since one of the primary themes elicited by the current study was that of the parents' desire for more involvement in their child's care and having their voices heard, further research in this area, exploring more directly the reasons behind parents' desire in increased involvement and the preferred degree of involvement, may be beneficial in understanding and therefore addressing the needs of families with a child diagnosed with two conditions. Given that there is little known about this unique situation, studies assessing the challenges faced by all stakeholders, including healthcare professionals, the affected children and their parents, may prove more beneficial than studies focusing on a single group's perspectives.

CONCLUSION

Raising a child with a single genetic condition or disability is difficult for families, however, there are even greater challenges faced by families raising a child with co-occurring genetic conditions. While the parents in the current study discussed some experiences similar to those previously reported by families of children with a single genetic condition, such as emotions around the diagnosis, impact on relationships with family and friends and changes in values and view on life, the findings elicited some novel experiences and challenges faced by families of children with more than one genetic diagnosis.

All of the participants spoke about the extent of knowledge that they had gained from the many experiences spent with their child in the hospital and the importance of their opinions on the care of their child being heard by healthcare professionals. The information provided by these parents should serve as a reminder for professionals to incorporate recommendations provided by parents and to involve parents in the decision making process around the care of their child. This is particularly important for this group of parents, as for some, the side effects of their child's conditions work in opposition, therefore further complicating management. Parents are then able to provide insider knowledge about the intricacies of balancing the management of the two conditions.

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APPENDICES

APPENDIX A
RECRUITMENT LETTER

Dear Sir or Madam:

You are being asked to participate in a research project by Paulina Nassab, a graduate student at CSU Stanislaus, in partial fulfillment of the requirements for a Master's degree in Genetic Counseling. Through this study, we hope to learn about the experience of parents with a child diagnosed with more than one genetic condition. The purpose of the study is to better understand, and thereby address, the social and healthcare needs of families, such as yours, in the future.

If you decide to participate in this study, you will be involved in one phone interview, which will take approximately 30-45 minutes. The interview will include 12 questions that will explore the overall experience of having a child with multiple genetic disorders and the effect of this experience on current and future life plans. In order to make sure that no comments are missed, interviews will be audio-recorded, with your permission. All audiotapes will be destroyed once this research project is over.

There is no cost to you beyond the time and effort required to complete the interview described above. Your participation is completely voluntary. Refusal to participate in this study will involve no penalty or loss of benefits. You may also withdraw from the study at any time without penalty or loss of benefits.

If you have further questions about the study or would like to participate, I may be reached at pauline.nassab@gmail.com. Interviews can be scheduled, at your convenience, in the month of January.

Thank you very much for your consideration.

Sincerely,
Paulina Nassab
Masters Candidate in Genetic Counseling
California State University, Stanislaus

APPENDIX B
INTERVIEW GUIDE

Introduction

I would like to start off by thanking you for taking the time to talk to me today.

**Refer to Informed Consent document

Discussion Questions

I. Diagnosis

1. Could you describe what the process of your child being diagnosed with --- and --- was like for you?

*Probes: What was your life like during that time?
What were your thoughts? Your feelings?*

2. Tell me about your experience with the health care team.

*Probes: What were some of the positive and/or negative aspects about the medical care that you received?
Any advice that you would give them?
What other information or resources would you have found useful?*

3. How do you compare your feelings at the time of your child's diagnoses to how you feel about your child's conditions now?

Probes: How have your feelings changed?

II. Daily Life

4. Could you reconstruct a typical day of caring for your child from the time that you wake up to the time that you fall asleep?
5. I would like to get a general sense of your experience having a child with more than one genetic condition. What are some of the challenging aspects and what are some of the rewarding/positive outcomes of having a child with a dual condition?

How would things differ if your child had only one condition?

6. How has your child's multiple diagnoses affected your life at home and at work?
7. How has your child's multiple diagnoses affected your relationships?

Probes: Relationship with partner? Relationship with child? Relationship with other family and friends? How did you explain your child's diagnoses to friends and family?

8. How has the way you look at yourself, others and the world around you changed as a result of this experience?

III. Long-term Affect

9. What were you told about the future of your child's conditions?

Probes: What aspects of the conditions were most remarkable to you?

10. What concerns, if any, do you have about your future? Future of your child?
11. In what way, if any, did your child's diagnoses affect future reproductive decisions?
12. What would you tell other parents who have recently had a child diagnosed with more than one genetic condition?

Probes: Any advice that you would give them?

Closing Remarks

1. Do you have any questions about the study or the questions that we've discussed today?
2. Is there anything else that you would like to add?

I've come to the end of my questions. Thank you very much for your participation today. I really appreciate it.

APPENDIX C

DESCRIPTION OF GENETIC CONDITIONS

Family 1

HMG-CoA Lyase Deficiency (HL) is a rare, autosomal recessive disorder that affects ketogenesis and leucine catabolism (Pie, J et al, 2007). Due to affected individuals' reduced capacity to synthesize ketone bodies, acute crises tend to occur when there is no exogenous intake of glucose or when there is excess glucose metabolism during times of infection, strenuous exercise or other types of stress (Pie, J et al, 2007). HL generally appears during the first year of life. Acute clinical episodes include vomiting, diarrhea, hypotonia, lethargy, cyanosis and apnea (Wysocki, S. J. & Hahnel, R., 1986). Long-term management includes following a high-carbohydrate, low-protein diet and avoiding hypoglycemia and long fasts (Gibson, K. M., Breuer, J. & Nyhan, W. L., 1988). If untreated, the disorder can lead to breathing problems, convulsions, coma and death.

Type I Diabetes Mellitus is a chronic disease caused by immune-mediated destruction of specialized, insulin producing cells in the pancreas called beta cells (Atkinson, Eisenbarth & Michels, 2014). Destruction of beta cells causes insulin insufficiency, which results in the inability to use glucose for energy or to control the amount of glucose in the blood (Simmons & Michels, 2015). This life-threatening hyperglycemia can result in clinic symptoms of weight loss, polyuria, polydipsia, fatigue, blurred vision and tingling or loss of feeling in the hands and feet (Simmons & Michels, 2015). Disease onset peaks between 5-7 years of age and at or near puberty. Once symptoms begin, treatment with exogenous insulin replacement is necessary (Atkinson et al, 2014).

Family 2

Ehlers-Danlos Syndrome, Hypermobility Type (EDS Type III) is a connective tissue disorder inherited in an autosomal dominant manner. Complications associated with the condition involve multiple body symptoms, including the skin, musculoskeletal, hematologic, gastrointestinal, cardiovascular, oral, ocular, neurologic and neuromuscular systems. Individuals with EDS Type III typically have soft or velvety skin that may be mildly hyperextensible, joint laxity or instability, chronic pain, easy bruising, functional bowel disorders, a high, narrow palate and autonomic dysfunction. Psychological dysfunction and emotional problems are also commonly seen (Levy, 2009).

Familial Adenomatous Polyposis (FAP) is a hereditary cancer syndrome that results in a predisposition to colon cancer. Affected individuals develop hundreds to thousands of precancerous colonic polyps, beginning at an average age of 16 years. Without a colectomy, colon cancer is inevitable, with the average age of diagnosis at 39 years. Other features present variably in affected individuals. These extracolonic features include an increased risk for associated cancers, including thyroid and pancreatic cancer, polyps of the gastric fundus and duodenum, osteomas, dental abnormalities, congenital hypertrophy of the retinal pigment epithelium (CHRPE), epidermoid cysts and fibromas and desmoid tumors. (Jasperson & Burt, 1998).

Family 3

Dyskeratosis Congenita (DKC) is a condition of abnormally short telomeres, characterized by a classic triad of dysplastic nails, lacy reticular pigmentation of the upper chest and/or neck and oral leukoplakia. Affected individuals are also at an increased risk of developing life-threatening complications, such as progressive bone marrow failure, myelodysplastic syndrome (MDS), acute myelogenous leukemia (AML), squamous cell cancer of the head and neck or anogenital region and pulmonary fibrosis. Additional symptoms may include eye and dental abnormalities. While affected individuals may present with any of the above symptoms, the onset and progression of DKC can vary considerably. Depending on the affected gene, various patterns of inheritance have been observed for DKC, including autosomal dominant, autosomal recessive and X-linked (Savage, 2009).

Hemophilia A is an X-linked, bleeding disorder that is characterized by deficiency in factor VIII, which is necessary for clotting activity. The severity and frequency of bleeding episodes can vary, depending on the level of factor VIII clotting activity. If untreated, affected individuals may experience prolonged bleeding after injuries, tooth extractions, or surgery, and renewed bleeding after an initial bleed has stopped. Individuals with severe hemophilia A may also experience spontaneous bleeds, primarily in the joints, but also in the kidneys, gastrointestinal tract and brain (Konkle, Josephson & Nakaya Fletcher, 2000).

Family 4

Methylmalonic Acidemia (MMA) refers to a group of autosomal recessive, inborn errors of metabolism characterized by elevated levels of methylmalonic acid in the blood and urine. The effects of MMA can vary from mild to severe, with affected individuals experiencing periods of health and intermittent decompensation, often during times of infection or other types of stress. Affected neonates present with vomiting, dehydration, lethargy, hypotonia, hepatomegaly and encephalopathy. Secondary complications include intellectual disability related to the decompensated state, renal insufficiency, failure to thrive, immune impairment and possible death.

Management of the condition includes following a low-protein, high-calorie diet (Manoli & Venditti, 2005)

Usher Syndrome Type I is an autosomal recessive condition characterized by profound, bilateral sensorineural hearing loss, vestibular areflexia and vision loss in adolescence as a result of an eye disease called retinitis pigmentosa (RP). RP is a progressive degeneration of rod and cone functions of the retina, presenting initially as night blindness and tunnel vision. Visual impairment worsens every year, however, the rate and degree of vision loss is variable. Due to the vestibular areflexia, affected individuals sit independently and walk later than expected (Keats & Lentz, 1999)