HHS Public Access

Author manuscript

J Genet Couns. Author manuscript; available in PMC 2017 August 01.

Published in final edited form as:

J Genet Couns. 2017 August ; 26(4): 829–840. doi:10.1007/s10897-016-0060-9.

Factors Associated with Parental Adaptation to Children with an Undiagnosed Medical Condition

Tatiane Yanes[1,2,3], **Linda Humphreys**[1], **Aideen McInerney-Leo**[4,5], and **Barbara Biesecker**[6]

[1]School of Medicine, Griffith University, Gold Coast, Australia

[2]School Psychiatry, Faculty of Medicine, University of New South Wales, Sydney, Australia

[3]Psychosocial Research Group, Prince of Wales Clinical School, Faculty of Medicine, University of New South Wales, Sydney, NSW 2052, Australia

[4]University of Queensland Diamantina Institute, University of Queensland, Brisbane, Australia

[5]Institute of Health and Biomedical, Innovation (IHBI), Queensland University of Technology (QUT), Brisbane, QLD, Australia

[6]Social and Behavioral Research Branch, National Human Genome Research Institute, Bethesda, MD, USA

Abstract

Little is known about the adaptive process and experiences of parents raising a child with an undiagnosed medical condition. The present study aims to assess how uncertainty, hope, social support, and coping efficacy contributes to adaptation among parents of children with an undiagnosed medical condition. Sixty-two parents of child affected by an undiagnosed medical condition for at least two years completed an electronically self-administered survey. Descriptive analysis suggested parents in this population had significantly lower adaptation scores when compared to other parents of children with undiagnosed medical conditions, and parents of children with a diagnosed intellectual and/or physical disability. Similarly, parents in this population had significantly lower hope, perceived social support and coping efficacy when compared to parents of children with a diagnosed medical condition. Multiple linear regression was used to identify relationships between independent variables and domains of adaptation. Positive stress response was negatively associated with emotional support (B = −0.045, $p \leq 0.05$), and positively associated with coping efficacy (B = 0.009, $p \leq 0.05$). Adaptive self-esteem was negatively associated with uncertainty towards one's social support (B = −0.248, $p \leq 0.05$), and positively associated with coping efficacy (B = 0.007, $p \leq 0.05$). Adaptive social integration was negatively associated with uncertainty towards one's social support (B-0.273, $p \leq 0.05$), and positively associated with uncertainty towards child's health (B = 0.323, $p \leq 0.001$), and

Tatiane Yanes t.yanes@student.unsw.edu.au.

Compliance with Ethical Standards Conflict of Interest Author TY, AML, LW and BB declare that they have no conflict of interest.

Human Studies and Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.'

Animal Studies No animal studies were carried out by the authors for this article.

affectionate support (B = 0.110, $p \leq 0.001$). Finally, adaptive spiritual wellbeing was negatively associated with uncertainty towards one's family (B = −0.221, $p \leq 0.05$). Findings from this study have highlighted the areas where parents believed additional support was required, and provided insight into factors that contribute to parental adaptation.

Keywords

Adaptation; Undiagnosed; Parents; Uncertainty; Hope; Social support; Coping efficacy

Introduction

Collectively rare diseases account for 6–10% of human disease, this equates to approximately 2 million people in Australia and 25 million people in the United States (Guillem et al. 2008; Kirby 2012; Zurynski et al. 2008). Despite advances in genomic medicine and technology, many individuals affected with rare diseases remain undiagnosed for many years and some never receive a diagnosis. For parents who do not have a diagnosis for their child's medical condition, the uncertainty surrounding the lack of a diagnosis permeates many aspects of their lives, leaving parents with limited treatment options and an uncertain future (Graungaard and Skov 2007; Lewis et al. 2010; Rosenthal et al. 2001). Parents have reported a number of practical and psychosocial benefits to a diagnosis for their child's illness including: providing direction and clarification over their child's treatment needs, clarity over prognosis, a label to which describe their child's condition, estimating recurrence risks, acceptance by others, and access to established support networks (Carmichael et al. 2014; Graungaard and Skov 2007; Kerr and Haas 2014; Lewis et al. 2010; Makela et al. 2009; Rosenthal et al. 2001).

Mishel's Uncertainty in Illness Theory defines uncertainty as “the inability to structure the meaning of illness related events” (Mishel 1988). This occurs when an individual is unable to assign definite values to objects and events, or is unable to accurately predict outcomes due to a lack of sufficient cues. Research has identified uncertainty as a major psychological stressor for patients and their families. For parents, consequences of uncertainty in a child's illness includes: psychological distress and anxiety, depression, and cognitive disturbances, all of which have significant health costs (Jessop and Stein 1985; Stewart and Mishel 2000; Tackett et al. 2016).

Lazarus and Folkman's Transactional Model of Stress and Coping (TMSC) (Lazarus and Folkman 1984) provides a conceptual framework for understanding the process of adapting to stressful life events such as an undiagnosed condition (Lazarus and Folkman 1987; Lazarus and Folkman 1984). When faced with a stressor, such as parenting a child with an undiagnosed medical condition, the individual evaluates the personal significance of the event to determine whether it is a threat or opportunity (primary appraisal), as well as evaluating what can be done about the situation (secondary appraisal). Together these appraisals influence coping which mediates the relationship between appraisals and adaptation. The TMSC predicts that effective coping leads to adaptation over time, with greater adaptation associated with psychological well-being (Folkman and Greer 2000;

Lazarus and Folkman 1987). Adaptation to a chronic illness or disability is a multidimensional and dynamic process that represents the long-term outcome of a health threat (Biesecker and Erby 2008).

Madeo et al. (2012) investigated factors that influence the uncertainty parents experience related to their child's undiag-nosed condition. Results indicate that parents with higher levels of uncertainty more often report lower perceptions of control, less optimism, and greater severity in their child's condition. The investigators hypothesized that these parents may use less effective coping strategies and subsequently have lower adaptation to their child's condition. More recently, results from a study of mothers of children with developmental and intellectual disabilities identified an increase in mothers' quality of life following diagnostic clarification, providing further insight into the impact of uncertainty on maternal well-being (Lingen et al. 2016).

Hope has been suggested as a secondary appraisal that is likely to affect parental adaptation (Truitt et al. 2012). Snyder et al. (1991) define hope as a goal driven behavior, derived by a sense of successful agency (perception that one can achieve their goals) and pathways (planning ways to meet one's goals). Snyder and colleagues hypothesized that individuals with greater hope will not only formulate plans they are confident about, but also have alternative methods of achieving their goals, having the flexibility to alter their plans if necessary. Previous research identified hope to be positively associated with greater quality of life, life satisfaction, and psychological well-being (Bailey et al. 2007; Folkman and Greer 2000). Caregivers of children with Down syndrome and autism have described feeling hopeful following the initial period of distress, and emphasized the importance of redefining goals after the perceived loss of the desired healthy child (King et al. 2006; Poehlmann et al. 2005; Truitt et al. 2012). Further analyses identified a high correlation among hope, uncertainty, and adaptation in caregivers of children with Down syndrome (Truitt et al. 2012). Truitt et al. found that individuals who had greater uncertainty reported less hope and lower adaptation, suggesting hope is an important factor in adapting to the uncertainty related to a child's condition.

Social support is widely recognized as an important adaptive resource for parents of children with disabilities and chronic illness, with parents who report lower levels of support having higher levels of psychological distress (Bromley et al. 2004; Patterson et al. 1997; Sloper and Turner 1992). However, for parents of children with an undiagnosed medical condition, accessing appropriate support is often challenging as the absence of a diagnosis precludes them from identifying appropriate services. Parents have expressed frustration over difficulties accessing social support, with many believing it would be more accessible if their child had a diagnosis (Lewis et al. 2010; Rosenthal et al. 2001).

Coping efficacy, also known as coping self-efficacy, is defined as one's perceived ability to successfully cope with a given situation (Bandura 1997). Research has demonstrated that high levels of coping efficacy can enhance personal accomplishments and adaptation, as individuals with higher perceived coping efficacy approach difficult tasks as challenges to be mastered, rather than threats to be avoided (Cudré-Mauroux 2011). Previous research has identified four sources of influence in ones coping efficacy: mastery experiences (previous

experiences), vicarious experiences (seeing others similar to oneself succeed), positive social persuasions, and psychological state (Bandura 1997). Although coping efficacy has not been assessed in parents of children with undiagnosed medical condition, research in other populations suggests that higher coping efficacy is likely to be associated with greater parental adaptation.

Currently, there is limited research exploring the experience of parents raising a child with an undiagnosed medical condition. This study aims to assess the relationships among uncertainty, hope, social support, and coping efficacy to adaptation as informed by the TMSC (Lazarus and Folkman 1984). We hypothesize that i) greater adaptation is associated with increased hope, social support and coping efficacy; and ii) increased uncertainty is associated with lower adaptation.

Methods

Participants

Individuals were eligible to participate if they were 18 years of age or older and the parent (biological or adoptive) of a child or children affected by an undiagnosed medical condition. An undiagnosed medical condition was defined as including two or more abnormalities that appeared to be genetic in nature for which an underlying cause or diagnosis had not been identified.

The study was approved by the Griffith University Human Research Ethics Committee (BPS/16/12/HREC).

Participants were recruited via advocacy and patient support groups, online message boards and parent blogs. Groups were identified through online searches using keywords such as “undiagnosed”, “no diagnosis”, and “syndrome without a name”. Online message boards and blogs were reviewed for their appropriateness before information about the study was posted. Participants were recruited from groups based in Australia, New Zealand and the United Kingdom. Eligible participants were invited to complete a de-identified online survey. Recruitment occurred between June 2013 and December 2013.

Instruments

Demographic Characteristics—Participants reported their age, sex, relationship status, total number of children, and total number of children with an undiagnosed medical condition. Additionally, data was collected on the child's age, order of birth for undiagnosed child, and age the parent's first noticed their child had a health problem (prenatally, at birth, less than 5 years, 6 or older). Participants also provided a brief summary of their child's health problems.

Parental Adaptation—“The 20-item Psychological Adaptation Scale (PAS) measures four constructs of adaptation: self-esteem, positive stress response, social integration and spiritual/existential wellbeing (Biesecker et al. 2013). Participants rate their level of agreement with each item on a five point scale from 1 (strongly disagree) to 5 (strongly agree). For example, “Being a parent of a child with an undiagnosed medical condition

has….” “helped me accept the way things work out” (positive stress response), “helped me learn to handle difficult time” (self-esteem), “helped me know who I can count on in times of trouble” (social integration), and helped me learn my life is more meaningful” (spiritual wellbeing). Further information on the development of the scale can be found in the original scale publication(Biesecker et al. 2013). The score for each subscale and total scale are the mean response to each item, yielding scores from one to five, with higher scores indication greater adaptation. The PAS has been used to measure parental adaptation in a number of settings, including parents of children with Down syndrome (Truitt et al. 2012), muscular dystrophy (Peay et al. 2016), Rett syndrome (Lamb et al. 2016), and undiagnosed medical conditions (Madeo, A. C., Bernhardt, B. A., & Biesecker, B. (Unpublished Manuscript). Parental Adaptation to an Undiagnosed Medical Condition in their Child). The mean adaptation score for these studies has ranged from 3.5 to 4.23. In this population the Cronbach's alpha was 0.92.”

Uncertainty—Parent's uncertainty was measured using the novel Parental Uncertainty about a Child's Health scale (PUCHs). The 22 item scale was developed based on the findings from a literature review and prior research (Madeo et al. 2012). The PUCHs measures four dimensions of uncertainty among parents of children with undiagnosed medical conditions: child, reproductive, family and social. The PUCHs was designed to deconstruct the dimensions of health-related uncertainty and weight the dimensions by importance. Participants are asked to rate the degree to which they agree with each item from one (strongly agree) to five (strongly disagree), for example: “not having a diagnosis for my child's limitations leaves me”… “not knowing how to think about my child's condition”; “unsure where to go for treatment for my child's condition”; and “lacking information to make decisions about having more children”. Participants then rank the importance of each item from one (not important) to five (very important). The final score is calculated by weighing the uncertainty items by their importance and then diving by the sum of the importance items. Scores range from one to five with higher scores indicating higher levels of uncertainty. In this population the Cronbach's alpha for the PUCHs was 0.90.

Hope—The 12-item Adult Hope Scale (AHS), which is based on Snyder's cognitive model of hope, assesses two domains of hope: agency and pathway thinking (Snyder et al. 1991). Participants are asked to respond to each item on an eight point scale ranging from one (definitely false) to eight (definitely true). The subscale and total scale are scored by calculating the mean response for each item, with higher scores indicating greater levels of hope. The Cronbach's alpha as measured in this population was 0.89.

Social Support

The Medical Outcome Study-Social Support Scale (MOS-SSS) is a 19-item scale that assesses four dimensions of social support: emotional (expression of feelings, guidance or advice), tangible (provision of material aid or assistance), affectionate (expression of love and affection) and positive social interaction (availability of other people whom to do fun things with) (Sherbourne and Stewart 1991). The score for each subscale is determined by summing the relevant items, and a total score calculated by summing all items. The total

scores range from 19 to 95, with higher scores indicating greater levels social support. In this population the Cronbach's alpha was 0.96.

In addition to the MOS-SSS scale participants were also asked rate the level of support they had received while caring for a child with an undiagnosed medical condition from 1 (no support), 2 (some support), and 3 (well supported).

Coping-Efficacy

The Coping Self-Efficacy scale (CSE) was developed to measure confidence in one's ability to cope with challenges or threats (Chesney et al. 2006). In this 26-item scale participants are asked: “when things aren't going well for you, how certain are you that you can…”. Participants then rate the extent to which they believe they could perform the behaviors from 0 (cannot do at all), 5 (moderately certain can do) to 10 (certainly can do). A total score for coping efficacy is calculated by summing all scale items. Scores range from 26 to 260, with higher scores indicating higher levels of coping efficacy. The Cronbach's alpha as measured in this population for the CSE was 0.94.

Importance of Diagnosis

Respondents were asked to rate how important it was for them to have a diagnosis for their child's medical condition from 1 (not important at all) to 5 (very important).

Open Ended Questions

To better understand participants' support needs, open-ended questions were included in the questionnaire. Participants were asked: “please describe the support you have received while raising a child with an undiagnosed medical condition”, “are there any areas you feel additional support was needed”, and “who has been your greatest source of support”. Participants also had the opportunity to provide additional comments at the end of the survey if they wished.

Data Analysis

Quantitative Analysis—Statistical analysis was conducted using Statistical Package for the Social Sciences 23.0 (SPSS). Each scale was scored according to published instructions. The primary outcome was adaptation to being a parent of child with an undiagnosed medical condition. The relationship status and importance of diagnosis variables that were initially categorical were subsequently dichotomized due to the disproportionate distribution across the categories. The variables were dichotomized as follows: relationship status (committed relationship vs not in a committed relationship), and importance of diagnosis (important vs not important). The remaining demographic variables (respondent's age, total number of children, age parent first suspected a medical condition, and birth order for eldest undiagnosed child) were kept in their original categorical values. Additionally, because almost all respondents were female (53/54), and had one undiagnosed child (51/54), gender and number of undiagnosed children were excluded from the analysis.

The key independent variables (uncertainty, hope, social support and coping efficacy) remained as continuous variables in the analyses. Bivariate relationships between all

domains of adaptation and each variable were analyzed using Pearson's correlation or ANOVA as appropriate, with $p < 0.05$ considered as significant.

Multiple linear regression was performed to determine the strength of relationship between the independent variables and domains of adaptation. Variables were included in this model if they had a significant bivariate relationship to a domain of adaptation at $p \leq 0.2$. The regression model was constructed by entering variables and then using backwards elimination with $p \leq 0.05$ as the cut-off for inclusion in the model.

In order to maximize the amount of data in each analysis, partially completed surveys were used in the analysis. Requirements were set for each quantitative measure so that at least 75% of each scale was completed, with the exact cut-off depended on the number of questions. Missing data were imputed once this cut-off was met. If a participant filled out only certain scales and met the completion requirements for those scales, the data were included in the appropriate analysis.

Qualitative Analysis—Responses to the open ended questions were managed using NVivo 10. A preliminary code book was developed based on previous research (Lewis et al. 2010; Madeo et al. 2012; Rosenthal et al. 2001) and early responses to the survey. The original codebook included the following themes: support need, uncertainty, and self-care. The data was coded by the first author (TY) and reviewed by all authors. Two emerging codes, communicating with others, and experience with health care professional, were added as appropriate codes were not identified. One of the original themes (uncertainty) was removed as no data was identified for this. Through this analysis a final codebook was developed. Participant's responses were generally short and covered a limited range of topics. The literal nature of the responses precluded the need for a second coder.

Results

Participants

Out of the 108 individuals who started the survey, two indicated they were ineligible, two indicated s/he was eligible but the response to an open ended question indicated otherwise, and 42 indicated they were eligible but completed none of the survey. Thus responses from 62 participants were included in the final analysis. Of these, only 53 completed the entire questionnaire.

Participants' Characteristics

Table 1 shows the sociodemographic data on the sample. Participants were primarily female (52/53), aged between 30 to 49 years (41/53), with two children (24/53) and were in a committed relationship (45/53). Participants' undiagnosed children were primarily aged five years or less (21/53). Only two participants had two or more children with an undiagnosed medical condition.

The vast majority of participants (45/53) described their child as having intellectual and physical symptoms, primarily global developmental delay, severe intellectual disabilities, multiple congenital anomalies and various physical symptoms. Some participants (4/53)

described their child as having physical symptoms only; these parents described multisystemic symptoms affecting cardiac, respiratory and immunological systems. Additionally, some of participants (4/53) described the presence of an undiagnosed neurological degenerative disease.

Descriptive Results

The distributions of responses to the psychological variables are shown in Table 2. With the exception of the PUCHs, all measured used in this study have been previously reported in similar populations. Results from a one sample t-test comparing the mean adaptation, hope, social support and coping efficacy scores with those from parents of children with a diagnosed or undiagnosed medical condition are shown in Table 3.

When asked to rate how important a diagnosis for their child's medical condition was for them, the majority of respondents (44/54) regarded a diagnosis as important or very important. Only 11.1% of respondents (6/54) felt a diagnosis was not important for them and 7.5% (4/54) were unsure. Additionally, when asked what level of support received while caring for their child, 32% (17/53) of respondents felt they had received no support, 54% (29/53) felt they had received some support and 13% (7/10) felt they had been well supported.

Bivariate Analyses

Analysis of Pearson's correlation coefficients identified significant associations between independent variables and domains of adaptation (see Table 4). Positive stress response was negatively correlated with emotional ($p = 0.041$) and tangible support ($p = 0.003$). Higher adaptive self-esteem was correlated with greater hope agency ($p = 0.038$) and coping efficacy ($p = 0.006$), and lower family ($p = 0.025$) and social uncertainty ($p = 0.004$). Adaptive social integration was positively correlated with two domains of social support, emotional ($p = 0.029$), and affectionate ($p = 0.002$), and positively correlated with child uncertainty ($p = 0.029$). A significant negative correlation between adaptive spiritual well-being and family uncertainty was also detected ($p = 0.017$). Only one demographic variable, relationship status ($p = 0.048$), had a significant relationship to a domain of adaptation, spiritual wellbeing. The remaining demographic variables, had a significance value of $p < 0.2$ and therefore not included in further multivariate analysis.

We found a statistically significant association between the two independent variables of hope and uncertainty. Child uncertainty was significantly associated with both domains of hope agency ($p < 0.001$) and pathway ($p > 0.001$). Similarly, the family domain of uncertainty was strongly associated with both domains of hope ($p = 0.002$), and pathway ($p = 0.002$). Finally a weak association between social uncertainty and the agency domain of hope was also identified ($p = 0.051$).

Multivariate Regression Analyses

Positive Stress Response—In this population individuals with less emotional support and higher coping efficacy, reported significantly greater positive stress response (Table 5). Together these two variables explained 20% of the total variance in positive stress response

(adjusted R^2). Multicollinearity was not identified in this analysis between coping efficacy and positive stress response (VIF = 1.077, tolerance = 0.929) (Menard 1995; Myers 1990).

Adaptive Self-Esteem—Individuals who had greater uncertainty in their social support and lower levels of coping efficacy reported lower adaptive self-esteem (Table 5). Together these two variables explained 21% of variance in adaptive self-esteem (adjusted R^2).

Adaptive Social Integration—Individuals who reported greater affectionate support, higher uncertainty towards their child's health and less uncertainty about their social support were found to have greater adaptive social integration. In this analysis 33% (adjusted R^2) of the variation in social integration scores could be accounted for by these three variables (Table 4).

Adaptive Spiritual Wellbeing—In the final model, uncertainty towards one's family and respondent's relationship status explained 13% (adjusted R^2) of the variance in spiritual wellbeing (Table 5). Greater familial uncertainty was significantly associated with decreased spiritual wellbeing. However, in this model relationship status was not statistically significant ($p = 0.074$).

Qualitative Analysis

Fifty-nine participants provided responses to the open ended questions regarding their experiences raising a child with an undiagnosed medical condition. Four themes emerged from the analysis, specifically: need for support, self-care, communicating with others, and experience with healthcare professionals.

Support need included areas where participants felt additional support was required in the areas of education, emotional, financial, respite, social services and specific support groups. Participants described feeling a “loss of support” due to not having a diagnosis for their child. As one mother stated:

> “If you cannot answer the question what is the diagnosis then people/government agencies switch off… you must fit in a box to gain support”.

Some parents advocated for the need for a care coordinator to assist them navigate the system, with some participants describing support services as a “*maze*”. A care coordinator was seen as someone who would relieve parents of some of the uncertainty they felt about the services they were eligible to access due to not having a diagnosis for their child's medical condition. As one parent described:

> “If you don't know what's wrong, how do you know what assistance your missing?”

Self-care related to the various strategies parents used to cope and adapt to their child's undiagnosed medical condition. Participants detailed a range of strategies included: positive thinking, acknowledging when they need help, and learning ways to advocate for their child. For example:

> “When people around you believe there is not a problem, and you know there is, you tend to stop looking for support and just get in and support the child by

> yourself. Choose the important battles to get the essential services the child needs and try to fill the gaps for her yourself”

One parent described the importance of hope when raising a child disability. However, for this participant the absence of a diagnosis for their child's medical condition impacted their ability to feel hopeful and brought uncertainties about the future:

> “When you don't have a diagnosis, you don't have a pathway towards resolution or support. When you don't have the pathway, you are left floundering. If you have support and a way forward, you have hope. Without a diagnosis or clear treatment plan, there is a real lack of hope that things will be okay. It's hard to visualize a good outcome when everyone around you, including professionals, are unable to point you in the right direction. Lack of funding is also a huge barrier to accessing services - no diagnosis means no funding.”

Communicating with others included not knowing how describe their child's medical condition and experience with family and friends. Some parents described family and friends refusing to believe their child's health needs as they did not have a diagnosis to which describe their child's medical condition, for example:

> “The most difficult thing is getting others to believe you. When there is no diagnosis, people assume there is no problem and that the parent must be neurotic or incompetent”.

When describing their interactions with health care professionals', parents often described negative experiences such lack of empathy and support:

> “Pediatricians and gp's just don't care”.

This often resulted in parents feeling as though their concerns were dismissed or taken seriously. Parents also felt that not enough being done to diagnose their child and became frustrated by a need to constantly advocate for their child with often limited support.

> “I don't think anyone has tried very hard to diagnose my son's condition. I think social services prefer it that way, if he was diagnosed with a physical disability we would be able to claim more support, likewise if he was diagnosed with ASD” (autism spectrum disorder).

Furthermore, parents expressed frustrations over the lack of knowledge by health care professionals regarding their child's medical needs, as one parent described:

> “Professionals quite often don't seem to even understand the basic issues with your child and seem reluctant to read their notes which is frustrating when your child is medically complex and you do not have an umbrella term to roughly explain their condition overall”

Discussion

Adaptation to a health condition refers to the process of coming to terms with the implications of the health threat and the observable outcomes of that process (Biesecker and Erby 2008; Lazarus and Folkman 1984). This study measured variables identified from the

literature likely to affect the adaptive process among parents of children with an undiagnosed medical condition. When compared to other parents of children with undiagnosed medical conditions (Madeo, A. C., Bernhardt, B. A., & Biesecker, B. (Unpublished Manuscript). Parental Adaptation to an Undiagnosed Medical Condition in their Child) and parents of children with a diagnosed intellectual and/or physical disability (Lamb et al. 2016; Peay et al. 2016; Truitt et al. 2012), parents in this population had significantly lower adaptation scores. Similarly, parents in this population had significantly lower hope, perceived social support and coping efficacy when compared to parents of children with a diagnosed medical condition (Lamb et al. 2016; Manuel et al. 2003; Peay et al. 2016; Truitt et al. 2012).

In the multivariate analysis, uncertainty regarding one's social support, family, and child's health were significantly associated with specific domains of adaptation, however, not all relationships were in the direction hypothesized. In contrast to our prediction, parents who perceived greater uncertainty towards their child's medical condition had greater adaptive social integration. This is similar to a finding from (Madeo, A. C., Bernhardt, B. A., & Biesecker, B. (Unpublished Manuscript). Parental Adaptation to an Undiagnosed Medical Condition in their Child), who identified strengthening of parental relationships with friends and family in the face of perceived uncertainty about their child's health. In line with our hypothesis, parents with greater perceptions of uncertainty related to availability of social support had poorer adaptive self-esteem and social integration. Finally, parents with higher perceptions of uncertainty about their family had poorer adaptive spiritual wellbeing. These findings demonstrate the importance of genetic counselors identifying sources of uncertainty for parents and developing strategies to reduce their uncertainty and improve adaptation. For example, genetic counselors can provide parents with information regarding support services available to parents and explore family relationships in order to identify family members who may be able to provide additional support.

Contrary to our hypothesis, neither total hope scores, nor its domains were associated with adaptation in the multivariate analysis. This finding is inconsistent with those from Truitt et al. (2012) who identified a positive association between hope and adaptation among parents of children with Down syndrome. It may be that our relatively small sample size resulted in insufficient power to detect a relationship. It is important to note that an association between uncertainty and hope was identified in this study, thus it may also be possible that in this population hope is associated with uncertainty, but related to different outcomes such as life satisfaction and well-being (Folkman and Greer 2000; Lloyd and Hastings 2009). Future research should aim to include a larger sample size and investigate alternative outcomes.

Establishing support systems and re-engaging in social encounters has been shown to be an important part of successful adaptation to a child's chronic illness (Canam 1993; Taylor 1983). In the multivariate analysis, adaptive social integration was associated with one domain of social support, “affectionate”. This was a positive association indicating that as parents receive more affection, they experience greater confidence in their ability to successfully re-integrate into the lives of family and friends. Affectionate support is defined as expressions of love and affection, including behavioral manifestations of love such as hugging someone (Sherbourne and Stewart 1991). Affectionate support has been widely

reported to improve a number of psychological outcomes such adaptation, quality of life and stress response (Floyd et al. 2010; Han et al. 2014; Leung et al. 2014). Overall our findings suggest that genetic counsellors can facilitate the adaptive process in parents of children without a diagnosis, by urging parents to identify sources of love and affection. For example, a genetic counsellor can ask parents to identify those in their lives who they feel comfortable drawing affection from such as hugging, or someone who makes them feel loved and supported.

In this study parents with greater coping efficacy were found to have increased positive stress response and self-esteem. It is important to note that coping-efficacy and self-esteem are related but distinct concepts. Coping efficacy refers to ones perceived capability to achieve goals, while self-esteem is defined as ones overall perceptions of self-worth and value (Bandura 1997; Folkman 1984; Folkman and Greer 2000; Taylor 1983). This finding suggests that genetic counsellors should explore the coping efficacy beliefs of parents raising a child with an undiagnosed medical condition. For example genetic counsellors could invite parents to explore times in which they successfully coped with a challenging situation (mastery experiences) and provide positive reinforcements that parents have the abilities to achieve their goals related to their child's condition (verbal persuasions) (Bandura (Bandura 1997). Additionally, there are numerous studies that describe interventions and strategies to improve one's feeling of coping efficacy including Chesney et al. (2003), Folkman and Greer (2000), and Sofronoff and Farbotko (2002) whose one day workshop has been shown to increase coping efficacy beliefs among parents of children with Asperger's syndrome.

In this study participants overwhelmingly rated the receipt of a diagnosis for their child as important or most important. Previous qualitative studies have reported that parents desire to find a diagnosis lessened over time as parents grew to accept their child's condition and refocused their attention on daily activates (Lewis et al. 2010; Rosenthal et al. 2001). Measures in this study do not allow for further exploration to determine whether the desire to receive a diagnosis is associated with length of time without a diagnosis. Additionally, reasons for wanting a diagnosis were not directly explored in this study; however, some parents provided insight into this in their responses to the open ended questions. Parents in this study described a diagnosis as a label which allowed them to explain their child's problems to other. A label was seen as a way to establish proof that their child did suffer from a medical condition, and hence validated the needs of both the parent and child. Furthermore, parents in this study described a diagnosis as a way to gain clarity over treatment and care for their child. In addition, parents felt uncertain about which services they were eligible to access and believed the absence of a diagnosis hindered their ability to make informed treatment decisions. These findings correlate to previously published studies including those of Rosenthal et al. (2001), Lewis et al. (2010) and Makela et al. (2009).

One of the many roles of genetic counsellors is to assist their patients' access appropriate services and support. Findings from this study have highlighted the areas where parents believed additional support was required, many of which have been previously described in the literature including educational, emotional, financial, respite, social services and specific support groups (Lewis et al. 2010; Madeo et al. 2012; Makela et al. 2009; Rosenthal et al. 2001). In this study, parents also expressed frustration navigating the disability, healthcare

and educational systems, and advocated for the addition of a counsellor or care coordinator to assist them navigate these systems. Previous research from the UK has highlighted the importance of care coordinators for parents of children with disabilities and chronic illnesses (Lewis et al. 2010). This study found that parents who had access to professional support had a less traumatic experience accessing services than those who did not.

Dissatisfaction with healthcare professionals was widely expressed by many parents in this study, including poor communication, lack of adequate support, dismissal of parental concerns, and lack of knowledge. Parents felt a responsibility in advocating for their child and managing their treatment needs, and therefore wanted to be acknowledged as an authority on their child's health. Additionally, some parents did not believe healthcare providers were making sufficient effort to diagnose their child. These negative experiences resulted in some parents feeling frustrated and placed mistrust in the healthcare professionals. Graungaard and Skov (2007) described the importance of a positive relationship between parents and health care professionals during the diagnostic process. In their study, Graungaard and Skov (2007) found that parents who felt healthcare professionals displayed empathy and were able to co-operate in the diagnostic process, were more satisfied with the information provided, even if the diagnosis was more serious. Findings from this study further highlight the importance for health care professionals to display empathy and utilize effective communication strategies when dealing with parents of children with chronic illnesses and disabilities. It is important for healthcare professionals to be attuned to the needs of the parents and ensure they support their patients, even when a diagnosis is not available.

Limitations

This study was limited by the small sample size and sociodemographic variability, which restricted our ability to explore the association between adaptation and variables measured. It was also not possible to calculate an accurate response rate as the total number of individuals who had access to the web link but chose not to participate is not known.

Participants in this study were self-selecting and therefore may not be a true reflection of all parents in this population. In addition, recruitment occurred through support groups and parenting forums related to issues associated with raising a child with a disability or chronic illness, which could have introduced biases in the sample population. Parents who do not belong to such support groups or are not actively seeking a diagnosis may have different insight into the issues presented.

Additionally, the majority of parents with undiagnosed children described them as being affected with both intellectual and physical disability, leaving no opportunity for statistical comparisons between types of conditions and adaptation scores. It is well known that parental needs vary according to their child's medical condition. For example, it is possible that for parents whose children are affected more severely or affected by progressive symptoms adaptation may be lower, but also cyclical. Future studies should assess how a child's medical needs affect parental adaptation in the undiagnosed disease population.

Practice Implications

It is important for genetic counsellors to understand factors that influence parents' adaptive process as this will enhance their ability to support their clients and implement interventions to facilitate coping and ultimately, adaptation. The results of this study indicate that different appraisals are associated with specific domains of adaptation. Thus, interventions should include an assessment of different domains of adaptation and identify strategies to improve that adaptive domain. For example, parents with lower adaptive self-esteem would benefit from strategies to improve their coping efficacy beliefs and identify sources of support. Additionally, genetic counsellor could assist parents' with lower adaptive social integration by identifying sources of affectionate support, exploring uncertainties towards the health of their child, and facilitating access to additional support. Genetic counselors can further support their patients by assisting them to navigate support services and where possible, advocating for parents and their child to ensure the appropriate services are accessed based on needs, rather than diagnosis. In doing so, it is hoped that genetic counselors will assists the parental adaptive process.

Research Recommendations

More thought needs to be given regarding the relationship between independent variables and domains of adaptation, perhaps through longitudinal studies and with a larger sample size. Future studies could consider utilizing additional sources of recruitment, in particular genetic healthcare services. Studies could also explore the relationship between uncertainty and hope, and the role of additional outcomes such as quality of life. It would also be worthwhile confirming whether the value of a diagnosis remains high in other populations of parents of children without a medical diagnosis, and whether this is related to the length of time without a diagnosis.

Acknowledgements

We thank all the participants who took the time to complete our survey and share their experiences. Dr. Biesecker's effort was supported by the Intramural Research Program of the National Human Genome Research Institute. Aideen McInerney-Leo is supported by a University of Queensland (UQ) postgraduate scholarship (APA).

References

Bailey TC, Eng W, Frisch MB, Snyder CR. Hope and optimism as related to life satisfaction. The Journal of Positive Psychology. 2007; 2(3):168–175.

Bandura, A. Self-efficacy: the exercise of control. Worth Publishers; New York: 1997.

Biesecker B, Erby L. Adaptation to living with a genetic condition or risk: mini-review. Clinical Genetics. 2008; 75(5):401–407.

Biesecker B, Erby LH, Woolford S, Adcock JY, Cohen JS, Lamb A, et al. Development and validation of the psychological adaptation scale (PAS): use in six studies of adaptation to a health condition or risk. Patient Education and Counseling. 2013; 93(2):248–254. [PubMed: 23993396]

Bromley J, Hare DJ, Davison K, Emerson E. Mothers supporting children with autistic spectrum disorders: social support, mental health status and satisfaction with services. Autism. 2004; 8(4): 409–423. [PubMed: 15556959]

Canam C. Common adaptive tasks facing parents of children with chronic conditions. Journal of Advanced Nursing. 1993; 18(1):46–53. [PubMed: 8429167]

Carmichael N, Tsipis J, Windmueller G, Mandel L, Estrella E. “Is it going to hurt?”: the impact of the diagnostic odyssey on children and their families. Journal of Genetic Counseling. 2014; 24(2):325–335. [PubMed: 25277096]

Chesney MA, Chambers DB, Taylor JM, Johnson L, Folkman S. Coping effectiveness training for men living with HIV: results from a randomized clinical trial testing a group-based intervention. Psychosomatic Medicine. 2003; 65(6):1038–1046. [PubMed: 14645783]

Chesney MA, Neilands TB, Chambers DB, Taylor JM, Folkman S. A validity and reliability study of the coping self-efficacy scale. British Journal of Health Psychology. 2006; 11(Pt 3):421–437. [PubMed: 16870053]

Cudré-Mauroux A. Self-efficacy and stress of staff managing challenging behaviours of people with learning disabilities. British Journal of Learning Disabilities. 2011; 39(3):181–189.

Floyd K, Pauley PM, Hesse C. State and trait affectionate communication buffer adults' stress reactions. Communication Monographs. 2010; 77(4):618–636.

Folkman S. Personal control and stress and coping process: a Theorethical analysis. Journal of Personality and Social Psychology. 1984; 46(4):839–852. [PubMed: 6737195]

Folkman S, Greer S. Promoting psychological well-being in the face of serious illness: when theory, research and practice Infrom each other. Psycho-Oncology. 2000; 9:11–19. [PubMed: 10668055]

Graungaard AH, Skov L. Why do we need a diagnosis? A qualitative study of parents' experiences, coping and needs, when the newborn child is severely disabled. Child: Care, Health and Development. 2007; 33(3):296–307.

Guillem P, Cans C, Robert-Gnansia E, Aymé S, Jouk PS. Rare diseases in disabled children: an epidemiological survey. Archives of Disease in Childhood. 2008; 93(2):115–118. [PubMed: 17942587]

Han JW, Jeong H, Park JY, Kim TH, Lee DY, Lee DW, et al. Effects of social supports on burden in caregivers of people with dementia. International Psychogeriatrics. 2014; 26(10):1639–1648. [PubMed: 25006855]

Jessop D, Stein R. Uncertainty and its relation to the psychological and social correlates of chronic illness in children. Social Science & Medicine. 1985; 20(10):993–999. [PubMed: 3160116]

Kerr AM, Haas SM. Parental uncertainty in illness: managing uncertainty surrounding an “orphan” illness. Journal of Pediatric Nursing. 2014; 29(5):393–400. [PubMed: 24524955]

King GA, Zwaigenbaum L, King S, Baxter D, Rosenbaum P, Bates A. A qualitative investigation of changes in the belief systems of families of children with autism or down syndrome. Child: Care, Health and Development. 2006; 32(3):353–369.

Kirby T. Australia makes up for lost time on rare diseases. The Lancet. 2012; 379:1689–1690.

Lamb AE, Biesecker BB, Umstead KL, Muratori M, Biesecker LG, Erby LH. Family functioning mediates adaptation in caregivers of individuals with Rett syndrome. Patient Education and Counseling. 2016; 99(11):1873–1879. [PubMed: 27373960]

Lazarus, RS., Folkman, S. Stress, appraisal, and coping. Springer Publishing Company; 1984.

Lazarus R, Folkman S. Transactional theory and research on emotions and coping. European Journal of Personality. 1987; 1:141–169.

Leung J, Pachana NA, McLaughlin D. Social support and health-related quality of life in women with breast cancer: a longitudinal study. Psychooncology. 2014; 23(9):1014–1020. [PubMed: 24700668]

Lewis C, Skirton H, Jones R. Living without a diagnosis: the parental experience. Genetic Testing and Molecular Biomarkers. 2010; 14(6):807–815. [PubMed: 20939735]

Lingen M, Albers L, Borchers M, Haass S, Gärtner J, Schröder S, et al. Obtaining a genetic diagnosis in a child with disability: impact on parental quality of life. Clinical Genetics. 2016; 89(2):258–266. [PubMed: 26084449]

Lloyd TJ, Hastings R. Hope as a psychological resilience factor in mothers and fathers of children with intellectual disabilities. Journal of Intellectual Disability Research. 2009; 53(12):957–968. [PubMed: 19744261]

Madeo AC, O'Brien KE, Bernhardt BA, Biesecker BB. Factors associated with perceived uncertainty among parents of children with undiagnosed medical conditions. American Journal of Medical Genetics Part A. 2012; 158A(8):1877–1884. [PubMed: 22711240]

Madeo, AC., Bernhardt, BA., Biesecker, B. Parental Adaptation to an Undiagnosed Medical Condition in their Child. Unpublished Manuscript

Makela NL, Birch PH, Friedman JM, Marra CA. Parental perceived value of a diagnosis for intellectual disability (ID): a qualitative comparison of families with and without a diagnosis for their child's ID. American Journal of Medical Genetics Part A. 2009; 149A(11):2393–2402. [PubMed: 19842198]

Manuel J, Naughton MJ, Balkrishnan R, Paterson Smith B, Koman LA. Stress and adaptation in mothers of children with cerebral palsy. Journal of Pediatric Psychology. 2003; 28(3):197–201. [PubMed: 12654945]

Menard, S. Applied logistic regression analysis Sage university paper series on quantitative applications in the social sciences. Sage; Thousand Okas: 1995.

Mishel MH. Uncertainty in illness. Journal of Nursing Scholarship. 1988; 20(4):225–232.

Myers, R. Classical and modern regression with applications. 2nd ed.. Duxbury; Boston: 1990.

Patterson J, Garwick A, Bennett F, Blum R. Social support in families of children with chronic conditions: supportive and nonsupportive behaviours. Developmental and behavioural pediatrics. 1997; 18(6):383–391.

Peay HL, Meiser B, Kinnett K, Furlong P, Porter K, Tibben A. Mothers' psychological adaptation to Duchenne/Becker muscular dystrophy. European Journal of Human Genetics. 2016; 24(5):633–637. [PubMed: 26306645]

Poehlmann J, Clements M, Abbeduto L, Farsad V, Ferguson D. Family experiences associated with a Child's diagnosis of fragile X or down syndrome: evidence for disruption and resilience. Mental Retardation. 2005; 43(4):255–267. [PubMed: 16000026]

Rosenthal ET, Biesecker LG, Biesecker BB. Parental attitudes toward a diagnosis in children with unidentified multiple congenital anomaly syndromes. American Journal of Medical Genetics. 2001; 103(2):106–114. [PubMed: 11568915]

Sherbourne CD, Stewart AL. The MOS social support survey. Social Science & Medicine. 1991; 32(6):705–714. [PubMed: 2035047]

Sloper P, Turner S. Service needs of families of children with severe physical disability. Child: Care, Health and Development. 1992; 18(5):259–282.

Snyder CR, Harris C, Anderson JR, Holleran SA, Irving LM, Sigmon ST, et al. The will and the ways: development and validation of an individual-differences measure of hope. Journal of Personality and Social Psychology. 1991; 60(4):570–585. [PubMed: 2037968]

Sofronoff K, Farbotko M. The effectiveness of parent management training to increase self-efficacy in parents of children with Asperger syndrome. Autism. 2002; 6(3):271–286. [PubMed: 12212918]

Stewart J, Mishel M. Uncertainty in childhood illness: a synthesis of the parent and child literature. Scholarly Inquiry for Nursing Practice. 2000; 14(4):299–319. [PubMed: 11372189]

Tackett AP, Cushing CC, Suorsa KI, Mullins AJ, Gamwell KL, Mayes S, et al. Illness uncertainty, global psychological distress, and posttraumatic stress in pediatric cancer: a preliminary examination using a path analysis approach. Journal of Pediatric Psychology. 2016; 41(3):309–318. [PubMed: 26423322]

Taylor S. Adjustment to threatening events: a theory of cognitive adaptation. American Psychologist. 1983; 38(11):1983.

Truitt M, Biesecker B, Capone G, Bailey T, Erby L. The role of hope in adaptation to uncertainty: the experience of caregivers of children with down syndrome. Patient Education and Counseling. 2012; 87(2):233–238. [PubMed: 21937189]

Zurynski Y, Frith K, Leonard H, Elliott E. Rare childhood diseases: how should we respond? Archives of Disease in Childhood. 2008; 93(12):1071–1074. [PubMed: 18684747]

Table 1

Demographic description of final sample (n = 54)

Variable	n	%
Sex		
Female	52	98.4
Male	1	1.6
Age		
18–29 years	4	7.5
30–39 years	21	39.6
40–49 years	20	37.7
50–59 years	7	13.2
60 and above	1	1.9
Relationship Status		
Married	40	75.5
De-facto relationship	5	9.4
Single/Never Married	4	7.5
Separated/Divorced	3	5.7
Widowed	1	1.9
Total number of Children		
1	10	18.9
2	24	45.3
3	14	26.4
4 or more	5	9.4
Number of undiagnosed children		
1	50	94.3
2	1	1.9
3	2	3.8
Age of oldest undiagnosed child		
0–5 years	21	39.6
6–10 years	15	28.3
11–15 years	8	15.1
16–20 years	5	9.4
21 years and above	4	7.5
Order of birth for oldest undiagnosed child		
Oldest	23	43.4
Middle	7	13.2
Youngest	23	43.4
Age parents first suspected a medical condition		
Prior to birth	5	10.2
At birth	16	32.7
0–5 years	24	49.0
6–10 years	4	8.2

Variable	n	%
Undiagnosed Condition		
Intellectual and physical symptoms	45	85
Physical symptoms only	4	7.5
Progressive symptoms	4	7.5

Table 2

Means, range, standard deviations of independent variables and primary outcomes

Variable (scale range)	n	Mean ± sd.	Response Range
Adaptation (1–5)	62	2.899 ± 0.790	1.00–4.75
Uncertainty (1–5)	54	3.600 ± 0.990	1.07–5.00
Hope (1–8)	58	5.312 ± 1.262	2.38–8.00
Social Support (19–95)	59	58.17 ± 17.972	19–95
Coping Efficacy (26–260)	53	124.471 ± 40.128	45.00–226.00

sd. Standard deviation

Table 3

One sample t-test comparing the mean adaptation, hope, social support and coping efficacy scores with those from parents of children with a diagnosed or undiagnosed medical condition

Variable	Published Scores ± sd	One Sample t test
Adaptation (PAS)	4.23 ± 0.593[a]	t(61) = −13.252, p = ≤0.001
	3.74 ± 0.760[b]	t(61) = −8.372, p = ≤0.001
	3.5 ± 0.9[c]	t(61) = −5.983, p = ≤0.001
	3.804 ± 0.78[d]	t(61) = −9.010, p = ≤0.001
Hope (AHS)	6.63 ± 0.875[a]	t(57) = −7.948, p = ≤0.001
Social Support (MOS-SSS)	73.01 ± 20.62[e]	t(58) = −6.342, p = ≤0.001
Coping Self Efficacy (CSE)	156.5 ± 51.6[c]	t(52) = −5.811, p = ≤0.001

sd. Standard deviation

[a]Parents of children with Down Syndrome (Truitt et al. 2012);

[b]Parents of children with undiagnosed medical conditions (Madeo, A. C., Bernhardt, B. A., & Biesecker, B. (Unpublished Manuscript). Parental Adaptation to an Undiagnosed Medical Condition in their Child);

[c]Mothers of children with muscular dystrophy (Peay et al. 2016);

[d]Parents of children with Rett syndome (Lamb et al. 2016);

[e]Mothers of children with cerebral palsy (Manuel et al. 2003)

Table 4

Bivariate associations between primary outcomes, independent variables and sociodemographic variable

		Outcome Variable Parental Adaptation			
		Positive Stress Response	**Self Esteem**	**Social Integration**	**Spiritual Wellbeing**
Pearson's correlation					
Uncertainty Child's Health (*n* = 54)	r	–	–	**0.298**	–
	p-value	–	–	**0.029**	–
Uncertainty Family (*n* = 54)	r	−0.213	**−0.304**	–	**−0.326**
	p-value	0.122	**0.025**	–	**0.016**
Uncertainty Social Support (*n* = 54)	r	–	**−0.388**	0.195	–
	p-value	–	**0.004**	0.158	–
Hope Agency (*n* = 58)	r	–	**0.273**	–	0.185
	p-value	–	**0.038**	–	0.165
Emotional Support (*n* = 59)	r	**−0.267**	–	**0.279**	0.168
	p-value	**0.041**	–	**0.032**	0.198
Tangible Support (*n* = 59)	r	**−0.382**	–	0.245	–
	p-value	**0.003**	–	0.066	–
Affectionate Support (*n* = 59)	r	–	–	**0.392**	–
	p-value	–	–	**0.002**	–
Positive Social Interactions (*n* = 59)	r	−0.202		0.228	–
	p-value	0.125		0.083	–
Coping Efficacy (*n* = 53)	r	0.266	**0.370**	–	–
	p-value	0.055	**0.006**	–	–
ANOVA					
Relationship status (committed relationship vs not committed relationship)	F (d.f)	1.771 (1)	–	–	**3.652 (1)**
	p-value	0.189	–	–	**0.048**
Importance diagnosis (important vs not important)	F (d.f)	–	–	–	1.875 (1)
	p-value	–	–	–	0.164

Only variables with $p < 0.2$ are shown; statistically significant associations ($p < 0.05$) are in bold

Table 5

Multiple linear regression predicting adaptation

Variable	B	CI	P
Positive Stress Response			
$R^2 = 0.197$ $p = 0.002$			
Emotional Support	−0.045	−0.074 – −0.017	0.002
Coping Efficacy	0.009	0.003–0.015	0.005
Self Esteem			
$R^2 = 0.212$ $p = 0.001$			
Uncertainty Social Support	−0.248	−0.437– −0.059	0.011
Coping Efficacy	0.007	0.001–0.013	0.019
Social Integration			
$R^2 = 0.328$ p = <0.001			
Uncertainty Child's Health	0.323	0.129–0.517	0.002
Uncertainty Social Support	−0.273	−0.468 – −0.079	0.007
Affectionate Support	0.110	0.045–0.175	0.001
Spiritual Wellbeing			
$R^2 = 0.129$ $p = 0.011$			
Uncertainty Family	−0.221	−0.412 – −0.030	0.014
Relationship Status	0.607	−0.062 – 1.276	0.074

Only variables with $p < 0.2$ in the bivariate analysis were included in the model. Table includes all predictor variables that remained in the model after backwards step elimination. B unstandardized regression coefficient, CI 95% confidence interval