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Uncertainty and Perceived Personal Control Among Parents of Children With Rare Chromosome Conditions: The Role of Genetic Counseling

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Little is known about the impact of genetic counseling on parental uncertainty or perceived control regarding the prognosis of a child with a chromosomal disorder. By exploring the parents' concerns and needs surrounding the child's diagnosis, a genetic provider can help to facilitate effective coping. This study tested the association of measures of parental uncertainty and perceived control with the perceived helpfulness of the genetic counselor. A survey was distributed to 875 members of the Chromosome Deletion Outreach (CDO) support group. We hypothesized that parents' perceptions about the helpfulness of the genetic counselor would modify the relationship between perceived uncertainty, perceived control, and coping. Among the 363 respondents, there was a significant negative correlation of the perceived helpfulness of seeing a genetic counselor with the levels of uncertainty ($rs = -0.20$, P-value < 0.001). Lower perceived helpfulness of the genetic counselor, along with less perceived personal control, less benefit of a diagnosis, and lower parental age were significant predictors of the highest perceptions of uncertainty. The Transactional Model of Stress and Coping was used as a framework for interpreting the relationships between parental uncertainty, perceived control, and outcome variables. There was a significant positive correlation between parents' perceived personal control and their reports of helpfulness of the genetic counselor ($rs = 0.20$, P-value <0.0006). Genetic counseling can be enhanced for parents faced with rare disorders by using interventions focused on reducing feelings of uncertainty and enhancing feelings of control.

KEY WORDS: uncertainty; perceived personal control; adaptation; genetic counseling; rare chromosomal disorders

How to cite this article: Lipinski SE, Lipinski MJ, Biesecker LG, Biesecker BB. 2006. Uncertainty and perceived personal control among parents of children with rare chromosome conditions: The role of genetic counseling. Am J Med Genet Part C Semin Med Genet 142C:232–240.

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This research was supported by the Intramural Research Program of the National Human Genome Research Institute, National Institutes of Health.

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DOI 10.1002/ajmg.c.30107

INTRODUCTION

In order to provide optimal counseling for parents who have children with rare disorders, it is important to assess the parents' understanding of their child's diagnosis and to facilitate such understanding to assist in adaptation [Bernhardt et al., 2000; Biesecker and Peters, 2001]. Genetics providers are often the first to disclose a chromosomal condition and provide insight about the implications of this diagnosis [Smith, 1998]. In situations where there are high levels of uncertainty, genetic counselors must address the lack of available information and then help the parent identify effective coping strategies.

For most rare chromosome disorders, there is limited information avail-

able on the natural history and prognosis because they have been reported in only a few individuals [Borgaonkar, 1997]. Parents of children with such rare disorders would be expected to have high levels of perceived uncertainty due to the limited prognostic information.

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Additionally, uncertainties about an affected child's health, cognitive limitations, and life span often persist even after the child has been given a diagnosis [Stewart and Mishel, 2000; Rosenthal et al., 2001; Lenhard et al., 2005].

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The time from the first recognition that something significant may be wrong with their child to the confirmation of a diagnosis has been identified as a particularly stressful time for parents and is characterized by heightened uncertainty [Stewart and Mishel, 2000]. Lazarus and Folkman's Transactional Model of Stress and Coping [Folkman, 1984] theorizes that the perception of stress depends on a number of subjective, cognitive judgments that arise from the dynamic interaction of a person and his or her environment. When a parent receives a diagnosis of a rare chromosome condition for his or her child, s/he assesses the impact the diagnosis will have on their life (primary appraisal). Based largely on past experiences, the parent also considers what can be done about the situation (secondary appraisal). Included in these appraisals are the uncertainty of the child's future, perceptions of the condition's severity, and the degree to which the parent feels in control of meeting the demands of the situation. These appraisals predict an individual's choice of coping strategies, and are important antecedents of an individual's adaptation to the condition [Folkman, 1984] (Fig. 1).

How an individual perceives the uncertain nature of their child's situation shapes their subjective cognitive response and their assessment of whether the situation is seen as a harm or a loss, a threat or a challenge, or some combination of these interpretations [Folkman and Greer, 2000].

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Consequences of parental uncertainty commonly include anxiety, depression, cognitive disturbances, or helplessness [Jessup and Stein, 1985; Schepp, 1991; Grootenhuis and Last, 1997]. Parents of

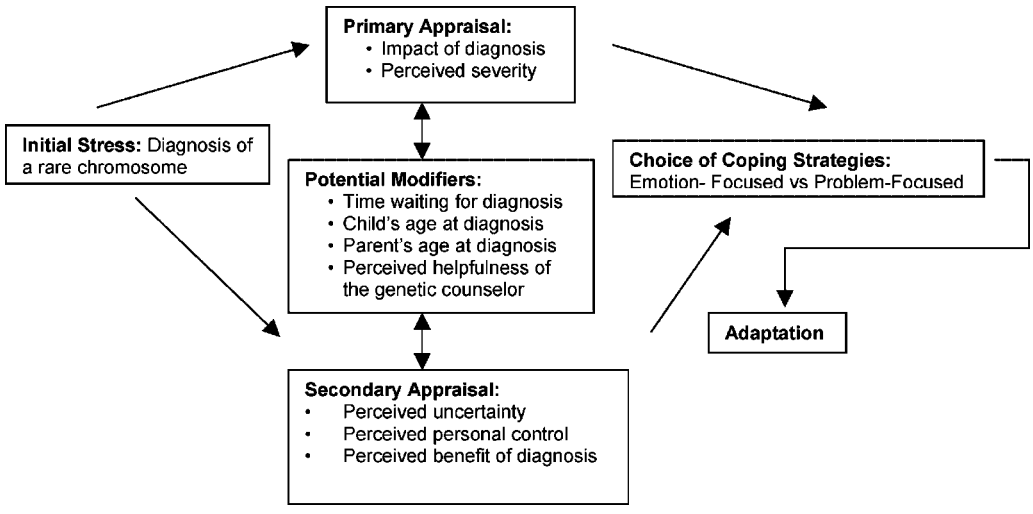

Figure 1. Transactional model of stress and coping [adapted from Lazarus and Folkman, 1984].

children diagnosed with cancer who scored high on levels of uncertainty were at an increased risk for anxiety and post-traumatic stress disorder [Santacroce, 2002]. However, some parents have reported that personal growth can be a positive outcome of uncertainty by recognizing that an uncertain outcome leaves open the possibilities for a positive outcome for their child [Cohen, 1993; Clarke-Steffen, 1993a; Rosenthal et al., 2001]. Therefore, while negative consequences of living with uncertainty have been reported, parents perceive benefits as well.

Even a minimal degree of prognostic uncertainty contributes to the emotional burden a parent must endure, perpetuating a feeling of being out of control [Mishel, 1983; Cohen, 1993; Clarke-Steffen, 1993b]. Perceived personal control is an important construct because it broadly addresses behaviors that may be amenable to intervention [Folkman, 1984; Litt, 1988]. Perceived personal control is "the belief that one has at one's disposal a response that can influence the aversiveness of an event" [Thompson, 1981].

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Perceived personal control research typically examines an individual's response to an event that is happening in one's own life, rather than in the life of one's child [Berkenstadt et al., 1999]. Perceived *parental* control has been used in studies of parents' perceptions of their control over their child's behavior [Bugental and Johnston, 2000; Guzell and Vernon-Feagans, 2004].

The present study modified the definition and assessed perceived control as it related to the control a parent felt over his or her child's overall health condition. We hypothesized that the perceived helpfulness of the genetic counselor would modify the relationships of perceived uncertainty and perceived control to coping. We used a stress and coping perspective to understand the relationships among parental uncertainty, perceived control, and the contribution of the genetic counselor to learn about the influences of the health care provider within situations of uncertainty.

MATERIALS AND METHODS

Participants for this study were recruited from the Chromosome Deletion Outreach (CDO), a support group of families, individuals, and professionals addressing rare chromosome disorders. Inclusion criteria for this study were that participants must be over 18 years old, reside in the United States, be English-speaking, and be parents who have a child with a rare chromosome disorder who was under the age of 21 years. A "rare chromosome disorder" was defined as any chromosomal condition that has not been well described and for which minimal prognostic information is available. Well-studied conditions such as Down syndrome, DiGeorge syndrome, Cri du Chat syndrome, Smith–Magenis syndrome, and Williams syndrome were therefore excluded. For this study, the upper limit population prevalence was 1/120,000 [Nussbaum, 2002 (personal communication)].

Parents in the CDO were mailed a survey and were invited to use either a paper version or a web-based version of the survey. The National Human Genome Research Institute Institutional Review Board reviewed and approved the study. The Board of the CDO support group also approved it. Surveys were collected from June 1, 2003 through September 12, 2003.

Sociodemographic and medical information was collected on the parents and their affected children. Participants were asked about the diagnosis, such as when a concern was first noted about their child, the child's age at diagnosis, whether their child's condition was detected by prenatal diagnostic testing (amniocentesis or chorionic villus sampling), and whether a genetics professional provided the diagnosis.

Ten-point Likert scales were used to evaluate several domains, including: parents' perceived benefit of diagnosis (10 = very beneficial), perceived personal control (10 = a lot of control), severity of child's condition (10 = very severe), and perceived helpfulness of the genetic counselor (10 = very helpful). The Likert scale measuring perceived personal control, previously used by Zakowski et al. [2001], was modified to assess the *parents'* perceived personal control of their child's condition. Participants were asked, "On a scale of 1 to 10 (1 = no control to 10 = a lot of control), how much control do you feel you have over your child's condition?" Open-ended questions were used to clarify parental experiences with perceived benefits.

The Parents' Perception of Uncertainty Scale, PPUS, was used to measure the parents' perceptions of uncertainty about their child's condition [Mishel, 1983]. The PPUS is a 31-item instrument (reliability coefficient 0.89) designed to examine the uncertainty parents experience related to their child's illness. Scores range from 31 to 155, with higher scores indicating greater uncertainty. The PPUS is composed of four dimensions of uncertainty consistent with theoretical and empirical predictions. The four domains include: *Ambiguity* (the absence or vagueness of cues, such as a fever or recurrent illnesses, concerning the planning or carrying out of care for the child [13 items]), *Lack of Clarity* (receiving or perceiving information about the child's treatment and the system of care [9 items]), *Lack of Information* (the absence of information concerning the diagnosis and the seriousness of condition [5 items]), and *Unpredictability* (the inability to make daily or future predictions concerning symptoms and illness outcome [4 items]). The reliability coefficients for the four sub-scales are 0.87, 0.81, 0.73,

and 0.72, respectively. Slight modifications were made to refer to the study population's circumstances, in consultation with Mishel, to maintain the scale's validity. Parental coping was assessed using the Ways of Coping Checklist-Revised (WCC-R) [Vitaliano et al., 1985].

Data were analyzed using NCSS 2001 statistical software (NCSS, Kaysville, UT). Dichotomous variables were compared using Chi-square analyses while continuous variables were compared using student *t*-test analyses and ANOVA. Some of the demographic variables with multiple response categories were dichotomized before multivariate analyses were performed. These included: the person giving the diagnosis (genetics professional vs. not a genetics professional) and the method of diagnosis delivery (in person vs. by phone, letter, or email). Stepwise multiple regression analysis was performed to determine which variables were significant predictors of the outcome of uncertainty. Analysis of variables that resulted in a *P*-value ≤ 0.05 were considered statistically significant and considered as candidates for inclusion in a multiple regression model. Canonical correlation analysis was performed to determine if there was significant correlation of two variables.

RESULTS

Surveys were mailed to 875 members of the CDO support group who were parents of a child with a rare chromosome condition. Of the 380 surveys that were returned, 360 were mailed and 20 (5.3%) were completed on the internet. Seventeen surveys were excluded for the following reasons: having a now deceased child who had a rare chromosome condition (N = 4), having a child who was over the age of 21 (N = 2), or having a child with a diagnosis of either Cri du Chat syndrome (N = 3), DiGeorge syndrome (N = 3), Williams syndrome (N = 2), or Smith-Magenis syndrome (N = 2), and having an affected grandchild rather than an affected child (N = 1). This left a total of 363 surveys that were used for data analysis, resulting in an overall response rate of 41.5% (363/875).

The population characteristics are found in Table I. Three hundred and fifty-two different rare chromosome conditions were represented in the study population. The genetic abnormalities that affected the children included autosomal deletions, duplications, trisomies, ring chromosomes, and mosaic karyotypes. Eighteen of the anomalies involved the sex chromosomes.

Parents reported being notified of their child's diagnosis from multiple sources, including a medical geneticist (47.5%), a genetic counselor (22.3%), and a pediatrician (16.2%). Other health care professionals mentioned included neurologists, neonatologists, and obstetricians. It is worth noting that some parents may have answered that they saw a genetic counselor when they actually saw a medical geneticist. This can be inferred by the qualitative responses that referred to the "genetic counselor" as "he." According to the 2002 professional status survey done by the National Society of Genetic Counselors, 94% of the professionals are female [Parrott et al., 2002]. The prevalence of the masculine pronoun suggests that the parents may have confused the professional who provided the genetic counseling.

In general, parents scored their child's condition as serious. The average

TABLE I. Demographics of Study Sample

	N = 363	%
Relation to child		
Mother (biological)	337	92.8
Father (biological)	17	4.7
Mother (by adoption)	8	2.2
Step mother	1	0.3
Gender of parent		
Female	344	95.3
Male	17	4.7
Highest education level achieved of parent		
Elementary school/junior high	1	0.3
High school/GED	53	14.6
Some college/some tech school	137	38.1
Four years of college	101	27.0
Post graduate	69	19.1
Ethnicity		
Caucasian	328	90.3
Caucasian/Hispanic	9	2.5
Hispanic or Latino	8	2.1
African American	5	1.3
African American/Caucasian	2	0.6
Not Hispanic or Latino	2	0.6
Asian	4	1.1
Other	5	1.5
Problem suspected before birth	84	23
Received prenatal diagnosis	25	7
	Age in years (SD)	
Age of responding parent	37 (7)	
Age of child	7.4 (5)	
Age of child when problem suspected	0.4 (0.7)	
Age of child when chromosome diagnosis made	2.0 (3.1)	

score on the 10-point Likert scale was 6.6 ± 2.2. Parents reported that serious aspects of their child's condition included seizures, multiple surgeries, failure to thrive, generalized hypotonia, congenital heart defects, lung abnormalities, cleft lip/palate, mental/physical delay, and autism.

The average score on the Likert-scale for parents' perceived personal control was 3.8 ± 2.4. This score suggested that, in general, parents felt they had little control over their child's condition. Furthermore, there was a positive correlation of parents' perceived personal control to their reports of helpfulness of the genetic counselor ($rs = 0.20$, $P < 0.0006$).

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This suggests that when genetic counseling is perceived as helpful, it may be related to interventions that help parents gain a sense of control over their child's condition. A negative correlation was found for parents' perceived personal control and their perceived seriousness of the child's condition suggesting that parents felt less in control when they perceived their child's diagnosis to be more serious ($P < 0.0005$).

The mean uncertainty score based on the PPUS was 82.7 ± 14.5, suggesting that participants had high levels of uncertainty. The results from the PPUS scale revealed that parents of children with rare chromosomal conditions scored higher than parents of other study samples, such as parents of children with leukemia (PPUS score 70.3 ± 14.5) and with cystic fibrosis (PPUS score 79.6 ± 16.7) [Mishel, 1997]. For parents who received a diagnosis from a non-genetic professional, those who subsequently saw a genetic counselor had significantly more certainty than parents who did not see a genetic counselor (83 ± 13 vs. 93 ± 15, P-value <0.05).

Correlation analyses determined which variables were correlated with the total PPUS score. Perceived benefit of the diagnosis (-0.25, $P < 0.0001$), perceived helpfulness of the genetic counselor (-0.20, $P < 0.0005$), perceived personal control (-0.32, $P < 0.001$), and parental age (-0.13, $P < 0.02$) negatively correlated with the total PPUS score. Perceived seriousness of the condition positively correlated with the total PPUS score (0.17, $P < 0.002$). These results suggest that parents with higher uncertainty were younger, perceived the diagnosis to be less beneficial, perceived the genetic counselor to be less helpful, perceived their child's condition to be more serious, and perceived that they had less control over their child's condition.

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Stepwise multiple regression analysis determined which variables were significant predictors of the level of uncertainty in this population. The only significant positive predictor of uncertainty was perceived seriousness of the condition (see Table II). Perceived helpfulness of the genetic counselor, perceived personal control, perceived seriousness of the condition, and perceived benefit of the diagnosis were all significant *negative* predictors of uncertainty. As seen in Table III, an ANOVA demonstrated that perceived personal control, perceived benefit of diagnosis, and perceived helpfulness of the genetic counselor were significantly

TABLE II. Stepwise Multiple Regression Analysis to Determine Which Variables Were Independent Predictors of Uncertainty (n = 229)

Variable	Regression coefficient (SE)	P-value
Perceived helpfulness of genetic counselor	−1.03 (0.32)	0.0015
Perceived personal control	−1.04 (0.38)	0.0061
Perceived seriousness of child's condition	1.05 (0.42)	0.013
Perceived benefit of diagnosis	−0.72 (0.35)	0.042
Education level	−1.87 (1.82)	0.31
Received diagnosis from genetic professional	−1.37 (1.87)	0.46
Problem suspected before birth	2.25 (3.35)	0.50
Ethnicity	−1.60 (3.28)	0.63
Time awaiting diagnosis	−0.40 (1.42)	0.78
Time since receiving diagnosis	−0.40 (1.42)	0.78
Parent's age	−0.04 (0.17)	0.80
Child's age	0.29 (1.37)	0.83
Received diagnosis in person	−0.34 (1.87)	0.86
Marital status	−0.09 (2.47)	0.97

TABLE III. Analysis of Variance Between Quartiles of Uncertainty (1 = Lowest, 4 = Highest) for Perceived Seriousness of Child's Condition, Perceived Personal Control, Perceived Benefit of Diagnosis, Perceived Helpfulness of Genetic Counselor, and Coping

Variables	Quartile 1 (mean ± SE)	Quartile 2 (mean ± SE)	Quartile 3 (mean ± SE)	Quartile 4 (Mean ± SE)	*P*-value
Perceived personal control	5.1 ± 0.2	3.5 ± 0.2	3.6 ± 0.3	3.1 ± 0.2	<0.0001
Perceived benefit of diagnosis	8.3 ± 0.3	7.7 ± 0.3	8.1 ± 0.3	6.5 ± 0.3	<0.0001
Perceived helpfulness of genetic counselor	6.3 ± 0.3	5.9 ± 0.3	4.3 ± 0.3	4.9 ± 0.3	<0.0001
Perceived seriousness of child's condition	6.2 ± 0.2	6.5 ± 0.2	6.6 ± 0.2	7.1 ± 0.2	0.07
Coping	73.3 ± 1.4	72.2 ± 1.6	75.9 ± 1.9	72.4 ± 1.5	0.43

lower in subjects in the top uncertainty quartile and higher in the lowest quartile. Interestingly, coping did not appear to be significantly different among quartiles of uncertainty. Stepwise multiple regression analysis was performed to determine which variables were significant predictors of coping. As seen in Table IV, only uncertainty, the parent's age, and perceived personal control were significant predictors of coping.

Of the 292 participants who reported seeing a genetic counselor, the average perceived helpfulness of the genetic counselor reported by the participants was 5.4 ± 2.9. In order to test our hypothesis, we also assessed which variables correlated with the perceived helpfulness of the genetic counselor. This demonstrated that the total uncertainty (PPUS) score (−0.20, $P < 0.0005$), perceived personal control (0.20, $P < 0.001$), the child's age at which the diagnosis was made (0.14, $P < 0.03$), time spent awaiting the diagnosis (0.135, $P < 0.04$), and time since receiving the diagnosis (−0.13, $P < 0.04$) were significantly correlated with the perceived helpfulness of the genetic counselor. Of the 292 parents who saw a genetic counselor, the mean uncertainty was 82.4 ± 14.6 compared with 83.3 ± 13.5 for the 62 parents who did not report seeing a genetic counselor ($P = 0.67$).

To learn more about the interactions of these parents with the genetic counselors, the following question was asked: "How could the genetic counselor have been more helpful?" Seventy-five percent (N = 221/292) of the parents who saw a genetic counselor responded to this question. One of the most prevalent suggestions for genetic counselors was to provide more information and resources.

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Interestingly, while many parents requested more information, they also frequently acknowledged that there was a paucity of information available even to health care providers. Almost three-quarters of the parents who saw a genetic counselor (71%) also wrote statements about what they thought the genetic counselor did that was helpful. Responses fit into two main categories: informational and psychological. The informational category was composed mainly of ways the genetic counselor was informative about the child's diagnosis. The description of genes and chromosomes was the most frequently addressed topic. However, while parents recognized the potential benefit of genetic counselors, they were disheartened by the services they received. Importantly, some parents commented on how they would have appreciated more hope and encouragement from their genetic counselor. Another common theme that

TABLE IV. Stepwise Multivariable Regression Analysis to Determine Which Variables Were Independent Predictors of Coping (n = 226)

Variable	Regression coefficient (SE)	*P*-value
Uncertainty	0.20 (0.07)	0.004
Parent age	−0.36 (0.17)	0.034
Perceived personal control	0.77 (0.38)	0.043
Perceived seriousness of child's condition	0.60 (0.42)	0.15
Education level	2.08 (1.80)	0.25
Received diagnosis from genetic professional	1.53 (1.85)	0.41
Child's age	1.03 (1.35)	0.45
Time awaiting diagnosis	−0.99 (1.42)	0.49
Perceived helpfulness of genetic counselor	0.19 (0.32)	0.55
Received diagnosis in person	−0.92 (1.85)	0.62
Time since receiving diagnosis	−0.60 (1.36)	0.66
Problem suspected before birth	1.11 (3.29)	0.74
Ethnicity	−0.90 (3.22)	0.78
Marital status	−0.11 (2.42)	0.96
Perceived benefit of diagnosis	−0.001 (0.35)	0.99

emerged was that parents felt they did not have an adequate amount of time during a session. Additionally, many wanted the genetic counselor to be a liaison between them and the health care team. Only 24 parents (10.3%) answered that they thought the genetic counselor was as helpful as s/he could have been.

DISCUSSION

Uncertainty is a major component of an illness experience that affects psychological adaptation. The precise nature of the effects of uncertainty, however, remains largely unknown. Having adequate knowledge about a diagnosis, of uncertainty its etiology, and its management implications imparts power to an individual to respond to their health status [Smith, 1998]. We sought to understand the role of health care providers in influencing perceived uncertainty and perceived personal control within a population where knowledge regarding management implications and prognosis is minimal. By understanding the effects a health care professional has on these psychological constructs, interventions can be designed to help patients mobilize effective coping strategies to improve their adaptation.

Having adequate knowledge about a diagnosis, its etiology, and its management implications imparts power to an individual to respond to their health status.

The negative correlation found of parents' age to uncertainty suggests that with increasing parental age (independent of years since diagnosis), parents perceive less uncertainty about their child's condition. Information offered to younger parents regarding the plan of treatment, system of care, and unpredictable nature of their child's illness was perceived by them as vague. Interventions aimed at minimizing uncertainty by highlighting what is known and what remains unknown may be more effective in meeting the needs of younger parents at the time of diagnosis or shortly thereafter. This uncertainty may stem from the limited experience younger parents typically have with healthcare systems. Therefore, education of genetic information and how to care for their child may reduce uncertainty. In a previous study of parents whose children have undiagnosed multiple congenital anomalies, parents reported frequently worrying that their child's condition would lead to early death, yet were afraid to ask health care providers whether it was likely [Rosenthal et al., 2001]. These data suggest that identifying whether a child's findings may be life threatening or not is an example of how pieces of relatively certain information can be introduced in an uncertain situation.

Further, this correlation of age to uncertainty may be also explained by the fact that older individuals have fewer reproductive plans. However, most rare chromosomal conditions are understood to be de novo and unlikely to recur in future pregnancies.

The Transactional Model of Stress and Coping predicts that perceived seriousness interacts with perceived personal control. These secondary appraisals are therefore important components to a person's overall assessment of how to cope with a situation. Participants' perceptions of the seriousness of the child's diagnosis proved to be an important correlate to their sense of personal control. Parents felt less in control when they perceived their child's diagnosis to be more serious. When perceived seriousness can be modified by information, this may help parents to gain feelings of control. But for those conditions for which the greater degree of perceived seriousness is an accurate assessment of the child's condition, modifying perceptions is not adaptive. Rather, counseling strategies should focus on ways to cope with the serious nature of the child's condition. In situations that are less amenable to personal control, emotion-focused coping strategies are more often considered adaptive [Zakowski et al., 2001].

The positive correlation that was found between parents' perceived personal control and their reports of helpfulness of the genetic counselor suggests that helpfulness is related to gains in perceived control. Although this finding is consistent with published goals of genetic counseling, we are not able to conclude from our study whether the counseling resulted in enhanced feelings of control, whether other factors contributed to enhanced control, or whether those who found genetic counseling more helpful were also the ones who also felt they had more control over their child's condition [Berkenstadt et al., 1999; Biesecker and Peters, 2001].

Genetic counselors who are aware of the possibility of heightened uncertainty and low levels of perceived personal control can devise interventions to enhance counselees' sense of control [Berkenstadt et al., 1999]. They can help parents reappraise their situation by affirming that there is minimal information available, by reassuring parents that an exhaustive search for information has been performed, and by facilitating ways for them to cope emotionally with the residual uncertainty.

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As previous data suggest [Clarke-Steffen, 1993a; Cohen, 1993; Rosenthal et al., 2001], some parents may be able to find hope in uncertainty and use this as an effective coping strategy. Studies that have used teaching interventions within the Transactional Model of Stress and Coping have reported significant improvements in coping efficacy, distress, and depression [Folkman et al., 1991; Chesney et al., 1996]. Study participants learned to accurately appraise the controllability of a stressor and to adjust their choice of coping strategies. Additional studies are needed to explore the interaction effects between counseling and perceived control and whether or not counseling can generate greater perceived control as an outcome of such interventions.

Overall, the findings from this study have implications for genetic counselors and other health care providers as participants were largely disappointed in the counseling they received for their child's chromosomal condition. The inverse correlation of perceived uncertainty and perceived helpfulness of the genetic counselor is striking. This is consistent with other data suggesting that individuals who saw a genetic counselor reported a preference for certainty in order to plan for the future [Skirton, 2001]. Parents may therefore benefit if genetic counselors initiate contact in the future, particularly if any additional information is learned regarding a particular diagnosis or prognosis.

The data also show that parents who have known about their child's condition for longer periods of time indicated that the genetic counselor was less helpful. It is unclear in what ways genetic counseling may have been less helpful for these parents. Possible explanations for this include general improvements in how genetic counseling is provided, increasing burden of caring for an affected child over-riding the significance of counseling, or that information provided by the genetic counselor was not as helpful to these parents as they had already developed strategies to cope with their child's condition. Genetic counseling is an important resource at the time of diagnosis and then periodically over time as parents adapt to their child's condition. Further research is needed to outline ongoing needs and to investigate interventions that may be helpful.

Furthermore, the answers to the open-ended questions suggest that parents were unable to distinguish between the roles of medical geneticists and genetic counselors. Parents who received a diagnosis from a medical geneticist were less likely to see a genetic counselor than those who received a diagnosis from a non-geneticist physician. The reason for this is unclear, but we speculate that there was not enough time within a clinic visit to meet with a genetic counselor, a genetic counselor was not available at the institution, the medical geneticist felt comfortable providing the counseling, or the parents were unclear about the professional who was providing the genetic counseling. These findings suggest that genetic counselors need to make their role and availability more transparent to parents, as was also suggested by findings from a prior small study of genetic counseling outcomes [Bernhardt et al., 2000]. Genetic counselors may need to explain their expertise and their intent to help parents to cope with the uncertainty in their child's condition. Counselors may also need to invite parents to make their needs more explicit. Responses to this study suggest that in the absence of prognostic information regarding their child, parents want to learn about resources (like the CDO), have more time to express their concerns about their child's condition, and hear expressions of hopefulness from their genetics providers.

One of the many roles of genetic counselors is to facilitate parents' adaptation to their children's condition by helping a parent understand their child's diagnosis [Biesecker and Peters, 2001]. Genetics providers are often the first to discuss the underlying basis for a chromosomal condition and provide insight about the implications this diagnosis may have for the health of the individual and other family members [Smith, 1998]. In situations where there are high levels of uncertainty, genetic counselors must address the lack of available information and then help the parent to identify effective coping strategies. In the case of a generally uncontrollable event, a more emotion-focused approach to coping may work best to reduce stress because one's internal state may be more amenable to change than the situation itself [Zakowski et al., 2001]. This is referred to as the goodness-of-fit concept in the stress and coping literature. Parents may initially engage in problem solving behaviors, such as information seeking, and end up frustrated with the lack of information available on their child's condition. By refocusing their coping to an emotion-focused style, the counselor may help these parents gain a greater sense of personal control. By acknowledging the parents' uncertainty regarding their child's condition, a genetic counselor may be able to help the parents come to accept that prognostic information is not available. However, it is essential that the genetics professionals reassure the parents that they are available to help with any medical problems that may arise and will notify them should further information about their child's specific condition become available. This strategy can help parents reevaluate their appraisal of the situation, lead to a heightened sense of personal control, and work toward mobilizing their use of effective coping strategies. While it is important for the parent and child to be followed by genetic professionals for their care, it is also important for the child's medical course to be followed in order to provide others prognostic information specific to their condition.

There are several limitations to this study that should be considered. We did not study the parent's personal characteristics and disposition that may have had important influences on their reactions to their child's diagnosis. Biases in ascertainment, response, and recall are likely as participants for this study originated from a national support group and the parents who agreed to participate in research may be different from other parents who do not belong to a national organization. It is also possible

that parents in the CDO may have more certainty because they are part of a support group. Parents with higher levels of uncertainty may use the support organization as a means to enhance their feelings of control. Parents may not have remembered accurately their feelings when they were given their child's diagnosis. No information was obtained for non-respondents.

Although previous literature has suggested that uncertainty can be perceived as a positive experience, the overall findings from this study suggest that the uncertainty associated with a diagnosis with minimal prognostic information is interpreted as a negative experience. Furthermore, parents who felt greater uncertainty perceived themselves to have less control over their situation. This study demonstrated that the perceived helpfulness of the genetic counselor was correlated with levels of uncertainty and perceived control. By exploring the parents' concerns and needs surrounding the child's diagnosis, genetic providers may be able to help parents implement coping strategies for future stressful events related to their child's condition. Genetic counselors and other health care professionals can work to modify the negative outcomes of uncertainty, promote lowered perceptions of uncertainty and heightened levels of perceived control, and foster coping skills that may help parents in their overall adaptation to their child's condition.

ACKNOWLEDGMENTS

The authors thank all the parents who took the time to answer our survey.

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