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ORIGINAL RESEARCH

Quality of Life and Autonomy in Emerging Adults with Early-Onset Neuromuscular Disorders

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Abstract Emerging adulthood is an important period in the development of one's identity and autonomy. The ways in which identity and autonomy are viewed by emerging adults and how they impact quality of life (QoL) in individuals with early-onset neuromuscular conditions is not yet known. This study focused on understanding and exploring relationships between self-perceptions of emerging adulthood, autonomy, and QoL. Five previously validated measures were incorporated into an online survey and distributed to young adults with early-onset neuromuscular conditions and unaffected controls. Topics explored included individuals' views regarding their overall QoL, disease-specific QoL, components of emerging adulthood, and autonomy. We found that a sense of higher disease impact was associated with a lower Overall General QoL. Additionally, perceptions of key autonomy factors "negativity" and "instability" were uniquely associated with Overall General QoL in the case group as compared to controls, whereas "attitudinal autonomy" (attaining the ability to plan and follow through with goals) was important to this age group regardless of health status. The specific factors of emerging adulthood and autonomy that were significantly correlated with Overall General QoL can be used for developing targeted counseling and interventions to improve QoL for individuals and their families.

Keywords Quality of life · Health-related QoL · Autonomy · Emerging adulthood · Neuromuscular conditions · Self-perceptions · Genetic counseling

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Introduction

The criteria by which we measure health outcomes and therapeutic success are based on both the biological and functional outcomes of medical care and the psychological and social experiences of an individual's life. Assessing individuals' views of their life and health status helps health-care professionals understand what functional, psychological, and social areas of life significantly impact their clients' wellbeing. Such self-evaluations measure a person's quality of life (QoL). Although QoL is a multidimensional concept, commonly studied areas within *general* QoL

assessments include individuals' perceptions of their physical, social, and emotional involvement in the world around them. The development of *health-related* QoL measures, which also consider disease-specific experiences, makes it possible to gauge individuals' personal assessment of their health status. Health-care professionals who work with individuals with genetic diseases or chronic conditions need to understand both their clients' general QoL as well as their health-related QoL to direct discussions and treatments toward the specific domains deemed most relevant to clients' overall health and well-being. In fact, in a letter of the first edition of the journal, *Quality of Life Research,* the editors postulate that quality of life may be the most important parameter to consider when assessing treatment in clients with chronic disease (Editors 1992, p. 3). While the editors did not advocate for any specific QoL measures, they asserted the substantial value of including this often previously ignored construct in health-related research. Thus, dedicated efforts to better understand quality of life issues in clients with chronic conditions, such as early-onset neuromuscular disorders, would be predicted to positively impact health outcomes in individuals with those conditions.

An important framework in which to assess clients' QoL and to conduct genetic counseling is a family systems perspective (Rolland and Williams 2005; Uhlmann et al. 2009), where clients are viewed within the context of a dynamic system involving their biopsychosocial needs and experiences, family beliefs and legacies, and the interactions between individuals within their family system. In addition, given the needs of individuals differ throughout the lifespan and depend, at least in part, on illness progression, a family systems approach that takes developmental stages of life into account becomes especially valuable (Rolland and Williams 2005). Given the importance of being cognizant of and responsive to a client's stage of life, it is encouraging that some genetic counseling and clinical genetics studies (e.g., Hamilton et al. 2009) are beginning to investigate the time of life termed "emerging adulthood," a phase that bridges the gap between adolescence and adulthood (Arnett 2000).

Emerging adulthood denotes a unique phase of life from age 18 to 29 and encompasses a time when individuals are taking on some, but not all, responsibilities of becoming an adult. This developmental stage applies to many races, ethnicities and abilities within developed countries (Arnett 2003; Reifman et al. 2007). It centers on "not simply a brief period of transition into adult roles but a distinct period of life course, characterized by change and exploration of possible life directions" (Arnett 2000, p. 469). It is distinguished as a time of experimentation, identity exploration, instability, self-focus, and feeling "in-between" adolescence and adulthood (Arnett 2000). As Blomquist (2007, p. 297) noted, "this concept of emerging adulthood is important when looking at realistic outcomes for employment and living independently for all youth, and especially youth with disabilities."

Central to emerging adulthood is a search for autonomy, which is the development of self-determination. Autonomy allows individuals to both experiment with and explore different identities, to focus on their sense of self and well-being, and to experience the changes in relationships, residences, and jobs that are typical of this developmental stage. Autonomy increases as emerging adults rationally evaluate choices, feel more confident in their beliefs and decisions, and gain control over their ability to achieve goals (Nelson and Barry 2005). Autonomy, therefore, is critical in the principle of informed choice, and therefore a valued component of the genetic counseling process (Köpke et al. 2009).

Young adults with certain genetic illnesses may have less autonomy due to the physical, psychological, or cognitive constraints of their disorders and the subsequent necessity (or perceived necessity) of substantial family assistance. Such individuals may not be able to proceed easily with the development of autonomy if disabilities affect their independence or if their sense of family duty outweighs their personal choice in the decision-making process (Anderson et al. 1994). Even if emerging adults with genetic or chronic conditions have developed critical thinking and independent decision-making skills, if they are not physically able to execute certain goals because of their condition, they may not feel confident in their independent abilities. Therefore, the development of autonomy may be particularly challenging for individuals with neuromuscular disorders that negatively impact their mobility because they may require assistance for many everyday tasks. In progressive neuromuscular disorders, disease advancement intersects with several different stages across the lifespan, often including emerging adulthood, thereby necessitating that genetic counselors be particularly attuned to the developmental perspective advocated by family systems theorists.

In research drawing upon Arnett's (2000) theory of emerging adulthood, Galambos et al. (2007) investigated the transition to adulthood for individuals with neuromuscular disorders by measuring their "psychosocial maturity," a construct involving self-perceptions of autonomy and identity development—two components integral to emerging adulthood. They found that 20- to 30-year-olds with neuromuscular illnesses reported lower perceived autonomy than those without such disorders, leading the researchers to advocate for more studies on the transition through emerging adulthood for individuals with neuromuscular disorders (Galambos et al. 2008). Galambos et al. (2007, 2008) brought to light the importance of emerging adulthood for those with motor disabilities, yet they did not measure their participants' quality of life—a factor that some view as

essential to assess in individuals with neuromuscular disease (Carter et al. 2007).

One other study in genetic counseling has discussed emerging adulthood as a period in which autonomy and identity development are especially important (Hamilton et al. 2009), though without assessing individuals' sense of emerging adulthood or perceptions of autonomy in the study. Therefore, to gain a deeper understanding of how the developmental period of emerging adulthood may relate to how individuals experience genetic illness and quality of life, we incorporated measures of both emerging adulthood and autonomy in our current study of individuals with early-onset neuromuscular disorders. Our inclusion of an unaffected comparison group of individuals in the same age range helped us to identify those emerging adulthood and autonomy characteristics that are specific to individuals with these disorders and those that are shared across health status in this age group. As Galambos et al. (2007) asserted, "it is important to learn how the transition to adulthood is similar and different for individuals with and without motor disabilities so that education, intervention, and transition planning for emerging adults with disabilities can be effectively designed and implemented to maximize their potential" (p. 827). Furthermore, Carter et al. (2007) stressed the importance of assessing health-related QoL in individuals with neuromuscular disorders.

Based on the theoretical and empirical work on emerging adulthood and autonomy, we anticipated that certain aspects of emerging adulthood and autonomy would differ between groups. We hypothesized that individuals with early-onset neuromuscular disorders would score lower than the unaffected comparison group on some measures of emerging adulthood, such as experimentation, because of potentially limited possibilities for individuals with chronic illness in traditional realms (e.g., living situation, work, and transportation). Because lessened mobility often results in the need to rely on others for everyday tasks, we also predicted that individuals with neuromuscular disorders would demonstrate lower scores on developmental components of emerging adulthood related to feeling independent and having freedom. We further hypothesized that the two groups would differ in their sense of autonomy. In addition, we sought to understand which, if any, relationships between autonomy, emerging adulthood, and quality of life might be specific to individuals with these conditions and which may be common to emerging adults regardless of health status. Given the paucity of previous research on this topic, we felt an exploratory investigative approach was necessary. Ultimately, armed with an increased understanding, we hope to better inform genetic counselors and other health-care professionals of the issues most salient to their emerging adult clients with early-onset neuromuscular disorders.

Methods

Participants

Case Group

The case sample included 24 English-speaking individuals with self-reported congenital myopathies and congenital muscular dystrophies (CM/CMDs) and other early-onset neuromuscular conditions in the emerging adulthood phase (from age 18 to 29). We recruited participants using two separate methods approved by a University of Michigan Institutional Review Board. First, we recruited individuals through the Congenital Muscular Dystrophy International Registry (CMDIR), an online organization created to identify and support individuals with CM/CMDs. A similar announcement was displayed on the research and disease specific pages of the Muscular Dystrophy Association (MDA), www.mda.org. Clinicians affiliated with genetics clinics at our University also informed any appropriate participants about the survey. All interested individuals were given information about how to access the website or contact the research team to participate in the survey. Details of participants in the case group can be found in Table 1.

Comparison Group

The comparison group was a convenience sample of individuals similar to the case population in age, gender, and living situation (e.g., residing with parents) who were recruited from a commuter campus of the University of Michigan. The comparison sample initially consisted of 151 English-speaking individuals; however, we excluded those who self-reported a neuromuscular condition and those who differed from the case group in terms of racial/ethnic background due to reported physical and psychological health disparities among certain racial groups (Brondolo et al. 2009). This left a final comparison group of 93 individuals.

Measures

General Quality of Life

We used the Quality of Life: Adolescent Version (Raphael et al. 1998) to measure how individuals conceptualize their life in nine domains: Physical Being, Psychological Being, Spiritual Being, Physical Belonging, Social Belonging, Community Belonging, Practical Becoming, Leisure Becoming, and Growth Becoming, described in Table 2. This instrument was developed and validated for 10- to 25-year-old individuals; therefore, it was appropriately constructed for most emerging adults (ages 18 to 29). This measure has

Table 1 Frequencies and description of diagnoses for individuals in the case sample

Diagnosis		n (%)	Common Clinical Features	Inheritance (bold=most common)	Known Genes Involved (bold=most common)	Typical Age of Onset in Literature	Age of Onset in Case Sample
Congenital Muscular Dystrophies	Collagen VI myopathies	5 (20.8%)	• proximal joint laxity; • distal contractures; • rigid spine; • respiratory problems	Autosomal Dominant Autosomal Recessive	*COL6A1, COL6A2, COL6A3*	Congenital-young adulthood	Birth-5 y
	Dystrogylcanopathies	4 (16.7%)	• mild to profound proximal muscle weakness; • potentially significant cognitive impairment; • variable cardiac and respiratory involvement	Autosomal Recessive	*FKRP, Fukutin, LARGE, POMGNT1, POMT1, POMT2*	Congenital-young adulthood	6 mos-5 y
	Laminopathy (Lamin A/C)	2 (8.2%)	• proximal muscle weakness and wasting; • contractures; • cardiac arrhythmias; • dilated cardiomyopathy	Autosomal Dominant	*LMNA*	Congenital-young adulthood	Birth-2 y
Congenital Myopathies	Centronuclear myopathy	3 (12.5%)	• hypotonia; • proximal muscle weakness; • respiratory difficulties	X-Linked Autosomal Dominant Autosomal Recessive	***MTM1***, *DNM2, BIN1, RYR1*	Congenital-young adulthood	Birth-2 y
	Nemaline myopathy	2 (8.2%)	• lower facial weakness; • bulbar weakness; • respiratory problems	Autosomal Dominant Autosomal Recessive	*ACTA1, NEB, TPM3, TPM2, TNNT1, and CFL2*	Congenital-young adulthood	Birth-2 y
	Congenital myopathy, other	2 (8.2%)	• hypotonia; • contractures; • respiratory problems	Autosomal Recessive	-	Congenital-young adulthood	Birth-2 y
	Central Core disease	1 (4.2%)	• hypotonia; • mild to severe proximal muscle weakness	Autosomal Dominant Autosomal Recessive	***RYR1***	Congenital-young adulthood	Birth-2 y
Other	[a] FSHD	3 (12.5%)	• upper limb and facial weakness and wasting; • weakness can progress to hips and legs	Autosomal Dominant	*D4Z4*	Childhood-young adulthood	Birth-5 y
	[a] LGMD type 2A (Calpain deficiency)	1 (4.2%)	• proximal muscle weakness and wasting; • contractures	Autosomal Recessive	*Calpain3*	8-15 years	Birth-2 y
	[a] Charcot-Marie-Tooth	1 (4.2%)	• peripheral neuropathy causing muscle weakness and loss of sensations in lower extremities; • foot abnormalities (pes cavus or hammer toes);	Autosomal Dominant Autosomal Recessive	*PMP22, EPZ, LITAF, EGR2, NEFL, MFN2, KIF1B, RAB7A, LMNA, BSCL2, GARS, NEFL, HSPB1, MPZ, GDAP1, HSPB8, DNM2, MTMR2, SBF2,*	Congenital-adulthood	Birth-2 y

Table 1 (continued)

Diagnosis	n (%)	Common Clinical Features	Inheritance (bold=most common)	Known Genes Involved (bold=most common)	Typical Age of Onset in Literature	Age of Onset in Case Sample
		• gait difficulties; • weakness can progress to upper extremities	X-Linked	*SH3TC2, NDRG1, EGR2, PRX, FGD4, FIG4, YARS, GJB1, and PRPS1*		

[a] while these conditions are not classified as CM/CMDs, we included these participants due to similarities of symptom presentation and course of disease progression indicated from survey questions not otherwise used in analyses

shown reliable internal consistency for each domain and reliable association with their validation measures (Raphael et al. 1996). Using Raphael et al.'s scoring rubric, importance and satisfaction scores were combined for each item. Scores on the subscales were then added and divided by three so that each participant received an average Overall General QoL score.

Health-Related Quality of Life (HR QoL)

To assess the impact of the neuromuscular condition on each person's life (in the case group), we used the Individualized Neuromuscular Quality of Life Questionnaire (INQoL©; Vincent et al. 2007). This is a validated measure developed by the Mapi Research Trust Education Information Dissemination group (Rose et al. 2006; Rose et al. 2005) and has shown test-retest reliability in practice (Vincent et al. 2007). This group provided a scoring system to calculate an Overall HR QoL score by combining the responses of importance within each life domain. In accordance with the scoring system, domain scores were then added together, divided by 180, and multiplied by 100 to obtain a percentage score for Overall HR QoL. Higher scores indicate a greater impact of the disorder on an individual's life. Cronbach's alpha coefficients showed that internal consistencies were greater than .70 for four out of the five subscales, with the Cronbach's alpha coefficient for the Independence subscale at .66.

Emerging Adulthood

To assess emerging adulthood, we administered the Inventory of the Dimensions of Emerging Adulthood (IDEA; Reifman et al. 2007). The instrument contains 28 statements describing this period in a respondent's life for five separate subscales, as seen in Table 2. The IDEA measure also includes an Other-Focused subscale (3 items), which does not measure one of the five components of emerging adulthood. Rather, it identifies when individuals take on greater responsibilities for others in their lives—essentially the opposite of being Self-Focused. Therefore, we did not use this subscale in analyses. Reifman et al. reported strong convergent validity for the measure and good test-retest reliability for four of the subscales. Cronbach's alpha coefficients showed that the internal reliabilities of the subscales were all greater than .70 for the current sample.

Autonomy

To measure personal feelings of independence and self-determination, we employed the Adolescent Autonomy Questionnaire (Noom et al. 2001), described in Table 2. This measure has been empirically tested and validated in the original Dutch version and is now being tested in English

Table 2 Measures used in online survey

Area of study	Measure	Authors	Components	# of Items	Meaning	Evaluation of results
Overall general QoL	Quality of Life: Adolescent Version	Raphael et al. 1998	Physical Being	12	"Being—who you are as a person"	Participants addressed the importance of and then again their satisfaction with 54 separate items to assess how they perceive their life to be in these areas. All items used a 5-point Likert scale, with 1 indicating *Not At All Important* (or *Not At All Satisfied*) and 5 indicating *Extremely Important* (or *Extremely Satisfied*).
			Psychological Being	12		
			Spiritual Being	12		
			Physical Belonging	12	"Belonging—how you fit in with people and places"	
			Social Belonging	12		
			Community Belonging	12		
			Practical Becoming	12	"Becoming—things you do in your life that define you"	
			Leisure Becoming	12		
			Growth Becoming	12		
Overall Health-related QoL[a]	Individualized Neuromuscular Quality of Life Questionnaire (INQoL©)	Vincent et al. 2007	Activities	2	How a person views daily work and leisure activities	The questions ask how important the specific domain is in the participant's life, using a 7-point Likert scale, with 0 indicating *Not At All Important* and 6 indicating *Extremely Important*. Higher scores indicate a higher impact of disease.
			Independence	2	How a person considers their freedom to choose and carry out activities	
			Social Relationships	6	How a person views their interactions with friends, family, acquaintances, and strangers	
			Emotions	2	How a person's muscle condition makes them feel emotionally	
			Body Image	2	How a person's muscle condition affects the way they view their personal appearance	
Emerging adulthood	Inventory of the Dimensions of Emerging Adulthood	Reifman et al. 2007	Identity Exploration	7	Trying to find one's personal identity, increase self-awareness, through defining one's beliefs and values	Viewpoints were assessed on a 4-point Likert scale ranging from 1 (*Strongly Disagree*) to 4 (*Strongly Agree*). Higher scores on the subscales indicate a greater association with the emerging adulthood stage.
			Negativity/Instability	7	Feelings that emerging adulthood is a time of stress and uncertainty	
			Experimentation/ Possibilities	5	Individuals' sense that there are many different possibilities available to them that they can explore	
			Feeling "In-Between"	3	Feeling in many emerging adults that they have moved away from childhood and adolescence, but have not quite become a full adult yet	
			Self-Focused	6	Individuals being more independent and responsible for themselves, yet also feeling a sense of freedom	

Table 2 (continued)

Area of study	Measure	Authors	Components	# of Items	Meaning	Evaluation of results
Autonomy	Adolescent Autonomy Questionnaire	Noom et al. 2001	Attitudinal Autonomy	5	Individuals' goal-setting and decision-making	Participants were asked to rate, using a 5-point Likert scale, how 15 different statements described them, with 1 indicating that the statement is a *Very Bad Description* and 5 indicating that the statement is a *Very Good Description*. Seven questions were later reverse-coded so that higher scores on all three subscales indicate greater perceived autonomy.
			Emotional Autonomy	5	Emotional independence from others, such as resisting peer pressure and accepting responsibility for one's self	
			Functional	5	Ability of individuals to plan how to achieve their goals	
	Worthington Autonomy Scale	Anderson et al. 1994	Family Loyalty Autonomy	10	How an individual feels in terms of feeling independent of family beliefs and practices, sometimes stated as feeling free of family or parental "binding"	Ten statements about familial situations were rated on a 4-point Likert-type scale, with 1 indicating *Strongly Disagree* and 4 indicating *Strongly Agree*. Higher scores indicate greater perceived autonomy.

[a] This measure was only administered to the case group

versions as well (Noom, M.J, personal communication, September 23, 2010). Cronbach's alpha coefficients showed that internal consistencies were greater than .70 for all three subscales.

In accordance with family systems theory, we also measured Family Loyalty Autonomy by using a subscale from the Worthington Autonomy Scale (Anderson et al. 1994), described in Table 2. Anderson et al. reported strong internal reliability of the measure, as well as good predictive validity. Cronbach's alpha showed good internal consistency of the measure (α=.81) for the current sample.

Demographics

We asked all participants to indicate their age, gender, race/ethnicity, education level, and parental income level. Those in the case group were also asked to indicate their specific neuromuscular disorder.

Procedure

All study procedures were approved by the Institutional Review Board at the University of Michigan (IRB: HUM00042688). The survey was conducted online and participants were able to download a PDF of the consent document or print the online version. A waiver of documentation of consent was granted because names and birthdates were not recorded. For the one individual who completed the survey by telephone with a study team member reading each question and subsequently filling out the online form, the consent form was also read, and the option of having it sent by email was given.

The survey was created and distributed through Qualtrics™, an online survey system. The order of the measures was chosen randomly by the study team to alternate between long and short sets of questions, in an effort to hold the participants' interest. All participants completed the survey in the same order: demographics, Inventory of the Dimensions of Emerging Adulthood, Quality of Life: Adolescent Version, Adolescent Autonomy Questionnaire, Worthington Autonomy Scale, and Individualized Neuromuscular Quality of Life Questionnaire (INQoL©).

Statistical Analyses

All data were analyzed using Predictive Analysis Software (PASW-formerly SPSS) Version 18. Analysis of covariance (ANCOVA) and multivariate analysis of covariance (MANCOVA) were used to compare measures between groups using age as a covariate. Pearson's partial correlations were used to identify associations between subscales of individual measures with Overall General and Overall HR QoL scores, with age as a covariate.

Results

Sample Characteristics

For both groups, ages ranged from 18 to 29 years. The average age of the case sample was 23.5 years and of the comparison group was 20.2 years, a statistically significant difference, $t(115)=5.06$, $p=.0001$. In the case group there were 15 women and 9 men, and in the comparison group there were 55 women and 39 men. Four racial/ethnic groups were represented in the case and comparison groups, respectively: Asian/Asian American ($n=2$, $n=11$), Hispanic ($n=3$, $n=4$), Multiracial/Multiethnic ($n=2$, $n=4$), and White/Non-Hispanic ($n=17$, $n=74$). Yearly parental income level did not significantly differ between groups, $t(94)=-.94$, $p=.35$, with both groups reporting an average income between $40,000 and $70,000. Additionally, there was no significant difference between the case and comparison group in terms of education level, $t(115)=-0.25$, $p=0.80$, with an average education level of attendance or completion of college/university.

In terms of diagnosis, the individuals in the case sample represented many CMs and CMDs as well as a few other neuromuscular conditions (see Table 1). Because there are only a few individuals represented with each condition, there was not enough statistical power to perform separate analyses for each specific diagnosis.

Group Comparisons

We performed initial analyses to see if significant differences existed between the two groups for any of the measures, while taking into account the covariate of age. For all analyses alpha was set at .05. For the multivariate analyses of covariance (MANCOVA) the effect size was measured using partial eta squared.

Overall General QoL

An analysis of covariance (ANCOVA) was conducted to compare the two groups on Overall General QoL. Because of the significant age difference between groups, age was entered as a covariate. The ANCOVA showed that the case group ($M=.57$, $SD=1.01$) and comparison group ($M=.65$, $SD=.64$) did not significantly differ in Overall General QoL, $F(1, 113)=.03$, $p=.87$. There was also no effect of age, $F(1, 133)=.41$, $p=.52$.

Emerging Adulthood Subscales

A MANCOVA was used to see if the two groups differed in the six subscales of emerging adulthood, while taking into account the effect of age. The overall MANCOVA was significant for age, Wilks's Lambda=.83, $F(5, 110)=4.54$, $p=.001$, $\eta^2=.17$. Follow-up univariate analyses showed that age was significantly related to Experimentation/Possibilities [$F(1, 114)=10.10$, $p=.002$, $\eta^2=.08$] and to Feeling In-Between, $F(1, 114)=5.62$, $p=.02$, $\eta^2=.05$. Younger age was associated with higher scores on both subscales.

After controlling for the effect of age, the MANCOVA also showed an overall group effect, Wilks's Lambda=.88, $F(5, 110)=2.92$, $p=.02$, $\eta^2=.12$. Univariate analyses revealed a significant difference between the case and comparison group in the Identity Exploration subscale, with the comparison group ($M=3.30$, $SD=.41$) scoring higher than the case group ($M=3.04$, $SD=0.58$), $F(1, 114)=6.29$, $p=.01$, $\eta^2=.05$. In addition, the comparison group ($M=3.24$, $SD=0.36$) scored significantly higher than the case group ($M=2.95$, $SD=.64$) on the Self-Focused subscale, $F(1, 114)=8.38$, $p=.005$, $\eta^2=.07$.

Autonomy Subscales

To determine whether the groups differed in their sense of autonomy, a MANCOVA was computed with the four subscales of autonomy as the dependent variables. Age was entered as a covariate. The overall MANCOVA was not significant for age [Wilks's Lambda=.95, $F(4, 108)=1.67$, $p=.16$, $\eta^2=.06$] or for group, Wilks's Lambda=.96, $F(4, 108)=1.25$, $p=.29$, $\eta^2=.04$.

Overall General QoL: Correlation with Components of Emerging Adulthood and Autonomy

To investigate what specific components of emerging adulthood and autonomy may relate to Overall General QoL, we computed a series of Pearson partial correlations, while controlling for age. Correlations were run on the case sample and comparison sample separately. In this way, we could discover if different patterns emerged for the two groups (see Table 3).

In terms of components of emerging adulthood, correlational analyses performed on the case group showed that Overall General QoL was significantly inversely correlated with Negativity/Instability and was marginally associated with both Experimentation/Possibilities and Self-Focused. For the comparison group, Overall General QoL was significantly positively correlated with the Experimentation/Possibilities and Self-Focused subscales.

As for autonomy, Overall General QoL in the case group was significantly positively correlated with Attitudinal Autonomy and Functional Autonomy, and marginally correlated with Family Loyalty Autonomy. For the comparison group, Overall General QoL was significantly positively correlated with Functional Autonomy and Family Loyalty Autonomy.

Table 3 Overall general QoL and overall HR QoL: their correlations with subscales of emerging adulthood and autonomy

Subscales	Overall general QoL		Overall HR QoL
	Case group	Comparison group	Case group
Identity Exploration	$r_{ab.c}$=.17 (p=.456)	$r_{ab.c}$=.08 (p=.440)	$r_{ab.c}$=–.28 (p=.228)
Experimentation/Possibilities	$r_{ab.c}$=.39^{+} (p=.073)	$r_{ab.c}$=.28* (p=.008)	$r_{ab.c}$=–.62* (p=.003)
Negativity/Instability	$r_{ab.c}$=–.58* (p=.005)	$r_{ab.c}$=–.15 (p=.146)	$r_{ab.c}$=.77* (p=.0001)
Self-Focused	$r_{ab.c}$=.41^{+} (p=.061)	$r_{ab.c}$=.24* (p=.020)	$r_{ab.c}$=–.67* (p=.001)
Feeling In-Between	$r_{ab.c}$=.02 (p=.944)	$r_{ab.c}$=.07 (p=.517)	$r_{ab.c}$=–.18 (p=.436)
Attitudinal Autonomy	$r_{ab.c}$=.69* (p=.001)	$r_{ab.c}$=.15 (p=.152)	$r_{ab.c}$=–.51* (p=.021)
Emotional Autonomy	$r_{ab.c}$=.30 (p=.193)	$r_{ab.c}$=.04 (p=.721)	$r_{ab.c}$=–.14 (p=.559)
Functional Autonomy	$r_{ab.c}$=.64* (p=.002)	$r_{ab.c}$=.29* (p=.005)	$r_{ab.c}$=–.52* (p=.020)
Family Loyalty Autonomy	$r_{ab.c}$=.39^{+} (p=.078)	$r_{ab.c}$=.36* (p=.0001)	$r_{ab.c}$=–.56* (p=.010)

To control for the effects of age, Pearson partial correlations ($r_{ab.c}$) were computed

* = statistically significant correlation

$^{+}$ = marginally significant correlation

Overall HR QoL: Correlation with Overall General QoL and with Components of Emerging Adulthood and Autonomy

Overall HR QoL and Overall General QoL were highly correlated (r=–.80, p=.0001). The greater the impact of disease, the lower the overall general quality of life for individuals with early-onset neuromuscular conditions.

We computed Pearson partial correlations to discover the relationships between Overall HR QoL and individuals' sense of emerging adulthood and autonomy, while controlling for age (see Table 3). Analyses revealed a similar pattern of results as was found with Overall General QoL. Overall HR QoL was significantly positively correlated with Negativity/Instability and negatively correlated with Experimentation/Possibilities and Self-Focused. As anticipated, lower impact of disease related to increased feelings of experimentation and possibilities, as well as feelings of freedom and self-responsibility. Higher impact of disease was associated with greater feelings of uncertainty. The analyses also showed that Overall HR QoL was significantly negatively correlated with Attitudinal Autonomy, Functional Autonomy, and Family Loyalty Autonomy. The greater the impact of disease, the lower these three forms of autonomy. All statistically significant correlations between Overall General QoL and either emerging adulthood or autonomy were also statistically significant for Overall HR QoL. Therefore, results will only be discussed for Overall General QoL although they can be applied to Overall HR QoL as well.

Discussion

The primary aim of this study was to identify factors important to a population of emerging adults with early-onset neuromuscular disorders so that genetic counselors and other health care professionals might better address unique psychosocial issues in a client population undergoing multiple life transitions. Using several different measures in our analysis, as summarized in Table 2, we found many significant correlations in the case group between the QoL measures and the factors of emerging adulthood and autonomy. We also found a number of significant correlations that are salient to this age group regardless of health status, making the application of our results relevant for genetic counselors in many specialties.

Group Differences in Subscales of Emerging Adulthood and Autonomy

Supporting our hypothesis, the case group scored lower than the comparison group on the Self-Focused subscale of emerging adulthood, which measures how individuals view their personal independence from others and responsibility to themselves (instead of to others). This finding indicates that individuals with early-onset neuromuscular disorders are less likely to have a sense of freedom and self-responsibility. It is possible that increased time spent with parents or caregivers, due to increased dependency, can delay an individual's opportunities for developing these skills. Interestingly, however, we did not see a group significant difference for any of the autonomy scales, which also deal with freedoms and independence. This suggests that the specific questions in the Self-Focused subscale related to focusing on oneself and optimism, as opposed to those questions related to independence (i.e., autonomy), are likely driving observed group differences for sense of freedom and self-responsibility.

The case group scored lower than the comparison group on the subscale of Identity Exploration as well, indicating that individuals with these conditions are less likely than their unaffected peers to feel able to find their individual identity or increase their self-awareness at this time of life. However, the accuracy of this interpretation must be tempered as some of the questions in the Identity Exploration subscale that led to this conclusion ask about separating from parents and planning for the future, which may be more limited for affected individuals who may be dependent

on others, most likely their parents, for care and assistance. While this result was not originally predicted, it suggests that in contrast to their unaffected peers, affected individuals in this age group may not be as concerned with, may not place a high importance on, and/or may not have the opportunity for defining their personal identity at this time in their lives.

We had also hypothesized that individuals in the case group would score lower on the emerging adulthood subscale of Experimentation/Possibilities due to possible limited options for living situation, work and transportation. The results for this subscale were significantly associated with age, because this covariate was taken into account during analyses. Interestingly, we did not find a significant difference between the case and comparison groups in this subscale, suggesting that individuals with and without neuromuscular conditions feel similarly about this time in their life as a period of experimentation and possibilities. Grootenhuis et al. (2007) explain that many individuals with progressive diseases experience a "response shift" to life experiences, in which they regularly change the way they internally evaluate themselves and their experiences in accordance with their disease course. This "response shift" may explain why the case group did not actually differ in their views of Experimentation/Possibilities: Perhaps their life-long adaptation to disease has caused them to redefine these concepts with respect to their life with a chronic condition.

As mentioned previously, the two groups did not differ in their sense of Attitudinal, Functional, Emotional, or Family Loyalty Autonomy. We found these results surprising in light of the differing results for the groups on the Self-Focused subscale of emerging adulthood, which measures independence and freedom—key concepts of autonomy. Given that individuals with chronic diseases often have increased dependence and spend more time with family, who help manage their condition (Rolland 1994), it seemed reasonable to postulate that individuals within the case group would have lower Family Loyalty Autonomy because they could have more to lose if they act or speak against family views. Such reasoning is consistent with family systems theory, where positive dynamic interactions with family are essential in maintaining existing relationships (Rolland and Williams 2005). However, if individuals with these conditions are concentrating less on Identity Exploration and Self-Focus, as described above, our results could reflect that they may not *perceive* that they have lower autonomy than their unaffected peers. Because our study focused on *perceived* autonomy, our results may not reflect an accurate measure of *actual* autonomy. This finding can be seen as a positive result as it suggests that the desired or perceived autonomy for emerging adults is not negatively impacted by disease status.

Relationships Between Overall General QoL, Emerging Adulthood, and Autonomy

Our investigation of correlations between Overall General QoL, autonomy scales, and dimensions of emerging adulthood allowed us to discern those issues associated with QoL that may be specific to those with neuromuscular disorders in the emerging adulthood stage.

Scores on the Experimentation/Possibilities and Self-Focus subscales were marginally to significantly positively correlated with Overall General QoL for both groups, suggesting that the impact of these factors on quality of life is not unique to individuals with early-onset neuromuscular conditions. This same pattern of results was also found for the Functional Autonomy and Family Loyalty Autonomy measures, again indicating that these autonomy issues are important for all individuals in emerging adulthood. Understanding that these aspects of emerging adulthood and autonomy are important for most individuals at this stage of life may be helpful for individuals with these conditions, as well as their families, because they may recognize that these concerns are common for peers in the emerging adulthood stage. This may help normalize these issues of autonomy for individuals and families living with early onset neuromuscular disorders. Recognizing the importance of individuals' stage in life is integral to family systems theory as well as to the practice of genetic counseling (Rolland and Williams 2005; Uhlmann et al. 2009).

In contrast, scores on the Negatively/Instability subscale were inversely correlated with Overall General QoL for the case group only. Feelings of negativity and instability may be more prominent in individuals with disabilities or genetic conditions because they have to manage their condition, adapt to progression of their disease, address limitations associated with their disease on a daily basis, and take responsibility in navigating their own health care. Changes in any of these health-related areas on top of other life changes may foster negative feelings about one's self, one's abilities, and one's future stability. In addition, because the neuromuscular conditions of the respondents in our study are rare and progressive, individuals may have a sense of isolation, furthering negative thoughts. These feelings may be particularly prominent when one is trying to take responsibility for additional aspects of one's life in the transition to adulthood. Thus, intentional exploration of issues related to negativity and instability with emerging adults who have neuromuscular conditions by genetic counselors and other health care providers may be important. Specific discussions to enhance awareness about these issues and to proactively address any relevant interventions may improve quality of life for affected individuals and their families.

Overall General QoL positively correlated with Attitudinal Autonomy for the case group, but did not for the comparison group. This is one of the most striking differences in the

pattern of correlations between the two groups. Although our correlational analyses do not afford us the ability to assess causal direction, these results may indicate that feelings of autonomy in decision-making and goal-setting (i.e., Attitudinal Autonomy) play a larger role for individuals with early-onset neuromuscular conditions when they evaluate their general quality of life than for unaffected emerging adults.

The literature on young adults with other life-long medical conditions (Westwood et al. 1999) can help elucidate why this type of autonomy may be particularly important for the QoL of individuals with chronic illnesses at this stage of life. While investigating the transition from pediatric care to adult health care systems, one study (Westwood et al. 1999) found that 77% of adolescents (M=16 years old) and 75% of adults (M= 27 years old) with cystic fibrosis felt that making their own decisions about treatment was "extremely important." In addition, 61% of adolescents and 87% of adults felt it was "extremely important" to take charge of their own lives (Westwood et al. 1999). Therefore, most individuals in this age group who acknowledge the progressive nature of their condition also realize the importance for their continued involvement in their health care. Because pediatric clinics often function in a more paternalistic fashion to help guide clients and families through complex systems, Attitudinal Autonomy may not be promoted. However, when individuals transition to adult care services, they are expected to cognitively evaluate choices and make their own decisions; therefore, it would be beneficial to promote and facilitate autonomy in decision-making before this transition occurs (Bailey et al. 2003). Our case group may have recently transitioned to adult care or be in the process of transitioning to adult care, where the expectations of autonomy in decision-making are much more evident and critical. This may be why this Attitudinal Autonomy was correlated with Overall General QoL; because it is an area of life that is highly salient and necessary for successful navigation of the adult health care system, especially for informed choice and decision-making.

Practice Implications

Given our study design, implications discussed herein directly relate to dealing with clients with early-onset neuromuscular conditions and other chronic conditions, but they are likely also relevant to individuals in the emerging adulthood phase, regardless of health status.

In relation to the subscales of emerging adulthood, genetic counselors and other health-care professionals can directly discuss issues of experimentation and possibilities with clients and assure them that such concerns and concepts are typical for those in this time of life. Normalizing specific areas of concern can help individuals with chronic conditions relate to their unaffected peers.

In addition to normalizing concerns about autonomy, we may also turn to our scope of practice for ways to address Functional and Family Loyalty Autonomy. Part of a genetic counselor's job is to help "identify and access local, regional, and national resources and services" that can benefit clients and families (National Society of Genetic Counselors 2007). Directing someone to resources such as disability or career services may provide avenues for independence that were previously unknown to a client, which may help them find ways of achieving goals (Functional Autonomy). In addition, such resources may provide independence from family members or caretakers, providing an avenue of increasing Family Loyalty Autonomy as well. Increasing these kinds of autonomy may also help to increase perceptions of overall QoL as well, the ultimate goal for many health care professionals. Additionally, because these forms of autonomy were significantly associated with Overall General QoL for both groups, we know that this result is independent of health status, meaning these forms of autonomy are important areas for genetic counselors to address with individuals in a variety of clinical areas to promote individuals' ability to make informed choices and take an active role in their health care.

Our finding that individuals with early-onset neuromuscular conditions experience lower QoL and increased feelings of Negativity/Instability suggests that it could be helpful to highlight interventions that foster higher feelings of stability and, hopefully, positivity. One such intervention is to discuss the difference between illness- and non-illness-related aspects of life (Navon 1999). Distinguishing between these two factors helps identify which areas are controllable and which areas are not. While controllability is not the same thing as stability, it may help individuals feel more stable and positive if they are able to feel in control of more aspects of their life than they initially realized.

Attitudinal Autonomy was found to be particularly relevant for our case group and may be related, in part, to navigating the health care system. Helping to include clients from a young age in their health management may increase their confidence and ability to make informed decisions and empower them in their continued care. It is important to promote all forms of autonomy for the client's sake as well as for the parents/caregivers (Bailey et al. 2003; Rolland 1994). Increased dependence on family during times of expected independence can increase family strain, which can be harmful for parents/caregivers over time. Facilitating "flexibility of both role reorganization and willingness to use outside resources is crucial" for adaptation to changes in disease progression and changes in the life cycle, such as moving into emerging adulthood (Rolland 1994, p. 25). One way to address this issue is by meeting with the client alone as well as with his or her family to see how expectations of responsibility or independence of the client may differ among family members. Such conversations may help multiple members of the family

understand ways they can help the client achieve his or her desired autonomy in the future. Additionally, this may promote higher family functioning/cohesion and lower family stress, which has been shown to influence quality of life and has been linked to better overall health outcomes (Cohen and Biesecker 2010; Szyndler et al. 2005).

Limitations

While the results from this study are thought provoking and can help facilitate discussions for encouraging autonomy in emerging adults, there are a number of limitations to keep in mind as well. Our sample size for the case group was small, and may not be representative of all individuals with these neuromuscular conditions. Also, because we standardized the order in which measures were presented across participants, an unrecognized order effect may have occurred. In addition, we were neither able to identify directionality for our correlations nor discern disease-specific associations, both of which could provide useful insights. Regression analysis could have provided more specific results about predictive relationships for our findings; however, our limited sample size made this an impossible test for the current data set. Along with our limited sample, we had unequal n's in the two groups, which reduced the statistical power of our analyses. Also, because our primary measure of autonomy was originally created for adolescents and not emerging adults, it may not be the most appropriate measure of this construct for participants in emerging adulthood.

Lastly, the case group was highly heterogeneous in terms of clinical diagnoses. Although we had intended to focus only on individuals with congenital muscular dystrophies and congenital myopathies (CM/CMDs), in order to increase our sample size respondents with related neuromuscular diagnoses in terms of reported age-of-onset and symptomology were also included. Even among those with CM/CMDs, clinical presentation, access to care, and symptom management may all be varied.

Future Directions

This study was exploratory in nature and there are many areas for future work. In the future, we hope to distinguish if ambulation, ventilator support, activities, pain, and weakness accurately reflect a relationship between disease severity and QoL or autonomy. CMDIR is currently working to create a severity outcome scale that we could apply to our data. Another avenue for future research could be to recruit more individuals with CM/CMDs through clinical services to gain more power in statistical analyses and stratify the data into disease-specific groups to see if there are any differences. Lastly, it will be important to understand how our suggested clinical interventions are implemented by clinicians who provide care to emerging adults with early-onset neuromuscular diseases and to assess the impact of such interventions on these individuals' QoL.

Conclusions

In conclusion, we have found that individuals with chronic health conditions, such as neuromuscular conditions, have many concerns similar to their peers, but also some concerns that may be unique to their situation. It is important for healthcare professionals to recognize these similarities and differences and incorporate them into discussions with their clients that explore ways of maintaining a positive perception of general QoL. The practical implications described provide a way of structuring psychosocial conversations with individuals in the emerging adulthood phase. Because QoL is not a concept that can be changed in one clinical session, it is important to revisit the ideas of independence and positive outlooks on life as individuals mature. This type of discussion can also provide an avenue for individuals to express their concerns not only about the functional aspects of their medical care, but also about their emotional and psychological wellbeing.

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