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ORIGINAL RESEARCH

Young Adults with MSUD and Their Transition to Adulthood: Psychosocial Issues

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Abstract Maple Syrup Urine Disease (MSUD) is an autosomal recessive condition with an incidence of 1 in 185,000 births worldwide. Regardless of the type of MSUD, treatment includes immediate and lifelong dietary restriction of isoleucine, leucine and valine. There is little known about the psychosocial impact of MSUD on the developmental milestones of emerging adulthood. We used a qualitative case study approach to explore the human experiences of MSUD on young adults ($n=8$) and parents ($n=8$). All participants were administered a semi-structured, qualitative interview as well as quantitative measures. Six core themes emerged: 1) lifelong strain of dietary management; 2) social isolation from peers and impact on dating; 3) impact of MSUD on academics and employment; 4) medical experiences and transition to adult care; 5) impact on family functioning; and 6) positive effects and growth. The results of this investigation highlight and expand awareness of the psychological and social needs of young adults with MSUD. This study calls for a collaborative, multidisciplinary effort in the treatment of these patients and their families.

Keywords Maple syrup urine disease (MSUD) · Psychosocial · Coping · Genetic counseling · Emerging adulthood

Introduction

Emerging adulthood, the period from the late teens through the twenties, is “characterized by dramatic changes and explorations”—changes in career interests, world views, and romantic partnering (Arnett 2000; Zukerman et al. 2011).The transition from dependence to independence can be a challenging process for any adolescent and is defined through various domains including, cognitive, emotional, and behavioral. The experience is unique to each individual and is influenced by one’s physical and mental ability to navigate these domains. Adolescents with chronic illness and disability have additional concerns, rendering them especially vulnerable as they progress into adulthood. While there are aspects of all chronic disease that commonly influence all afflicted individuals, there are also “disease-specific” aspects of chronic diseases that can influence an individual in a disease specific manner (Tansella 1995). For example, dietary restrictions, invasive medical procedures, and physical abnormalities may interfere with the development of self-esteem. There is little known about the impact of a chronic illness such as Maple Syrup Urine Disease (MSUD) on the milestones of emerging adulthood. Such studies are important because characteristics of MSUD may pose barriers to achieving various academic, vocational, and social milestones (Simon et al. 2007; Zukerman et al. 2011).

Maple Syrup Urine Disease is an autosomal recessive condition with an incidence of 1 in 185,000 births worldwide. Due to a founder mutation observed in certain Mennonite populations, the disease incidence is as high as one in

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380 live births (Strauss et al. 2009). Maple Syrup Urine Disease is caused by a deficiency in the activity of the branched-chain α-keto acid dehydrogenase (BCKAD) complex, an enzyme that is responsible for the catabolism of three branched chain amino acids (BCAA), isoleucine, leucine and valine. Impairment of BCKAD leads to accumulation of BCAAs in plasma and body tissues resulting in neurotoxicity and consequent effects on cognitive, adaptive, and psychosocial functioning.

The clinical phenotypes of MSUD can be divided into four categories based mainly on BCKAD enzyme activity. The four types of MSUD include classical, intermediate, intermittent and thiamine responsive (Dancis 1974; Strauss et al. 2009; Wilcox and Cederbaum 2007). The classical form is the most severe form with neonatal onset. Newborns affected with classical MSUD have 0–2% of normal BCKAD enzyme activity. They are normal at birth but develop symptoms soon thereafter. If left untreated, seizures and coma ensue, ultimately leading to death. Those who do survive, exhibit brain injury, spasticity and profound mental retardation if left untreated. The intermediate form is less severe than the classical form and displays variable age of onset. There is a milder neonatal course and approximately 3–30% of normal enzyme activity. When ill, they can present like a child with classical MSUD. Individuals with the intermittent form of MSUD have approximately 5–20% of normal enzyme activity and show normal early development. Symptoms observed in the classical form are usually brought about by illness or infection. They are asymptomatic between episodes. Individuals with the thiamine responsive form have 2–40% BCKAD activity and do not exhibit neonatal illness. They may present later with a phenotype similar to the intermediate form. As the name implies, treatment for thiamine-responsive MSUD children includes addition of thiamine to their dietary regimen.

Regardless of the type of MSUD, treatment includes immediate and lifelong dietary restriction of isoleucine, leucine and valine. The goal of dietary treatment is to maintain branched chain amino acids (BCAAs), particularly leucine, within normal limits while ensuring adequate calories and protein and avoiding nutritional deficiencies as studies have shown a correlation between cognitive decline and elevated blood leucine levels (Jurecki 2006; Wilcox and Cederbaum 2007). While this treatment markedly reduces mortality it has a variable influence on morbidity. Studies attempting to identify risk factors associated with cognitive and neurological disturbances have identified time of diagnosis, maintenance of dietary control and levels of leucine and its derivative levels in the blood (Kaplan et al. 1991; Nord et al. 1991) as important factors that influence morbidity. A few studies have documented improved cognitive and neurological outcomes in early-treated and optimally managed children with MSUD (Hilliges et al. 1993). Thus, immediate and continued treatment greatly decreases the severity of injury and leads to a more favorable outcome.

Purpose of the Present Study

This study focuses on young adults with MSUD as they transition into adulthood. The study's goal was to assess the psychosocial concerns of young adults with MSUD as well as to enhance our understanding, sensitivity, and awareness of the rich human experiences of those who live with a chronic illness. This study utilized a mixed methodology framework to provide descriptive cognitive and psychosocial information of young adults, and information on the impact of MSUD on young adults and on their families. In addition, we used qualitative methods to describe the illness experience of young adults with MSUD. Researchers are increasingly acknowledging the importance of adopting a qualitative perspective to understanding young adults' illness experiences (Woodgate 1998).

Methods

Participants and Procedure

Participants included young adults with MSUD (16–27 years of age) and one of their primary caregivers. A total of 16 participants were recruited for this study, eight young adults with MSUD and eight parents of young adults with MSUD. Six of the eight participant pairs (a pair consisting of a young adult and their parent) were recruited from the MSUD Support Group's biannual meeting after a presentation of the study's purpose and goal. Two of the eight participant pairs, recruited from the University of California, San Francisco (UCSF) Neurometabolic/Biochemical Clinic, were contacted by letter and informed of the study. All participants underwent informed consent prior to starting the study. The study was approved by the Institutional Review Board at Palo Alto University.

Interviews (by first and second authors), were conducted either at the MSUD Support Group's biannual meeting or at UCSF. The interviews were, on average, 2 h in length for the young adults and, on average, 2 h in length for the parents. The young adults were interviewed separately from their parents. All interviews were tape-recorded and transcribed to ensure accuracy.

Assessment Measures

For the patients, the assessment measures consisted of a semi-structured interview and the Wechsler Abbreviated Scales of Intelligence (WASI). Parents completed a semi-structured interview as well as the Pediatric Quality of Life Inventory, Family Impact Module

Semi-structured Interviews

The MSUD Young Adult Interview consisted of 139 open-ended questions and was designed to provide an understanding of the challenges and experiences of young adults with MSUD. The Parent Interview consisted of 120 open-ended questions and was designed to provide an understanding of the stresses imposed by the occurrence of MSUD in a young adult on the family as well as the experience of the primary caregiver of a young adult with MSUD. Both interviews included demographic questions such as age, education, marital status, ethnicity, and occupation. The open-ended questions were categorized into several topics: life experiences with respect to MSUD; relationships with peers and family; academic and employment experiences; romantic relationships (dating, marriage and children); medical experiences and transition to adult care; impact of MSUD on family functioning; and positive effects.

Questions about life experiences with respect to MSUD are conceptualized as a perceived impact on the self. Items in this domain along with questions about relationships with family and peers, are based in part on the work of Mellin and her colleagues (Mellin et al. 2004) in their study of parents' experiences of parenting an adolescent daughter with type 1 diabetes. In addition, these questions were drawn from semi-structured interviews used to examine the experiences of patients with other inherited metabolic disorders including Neimann Pick B (NPD type B) (Henderson et al. 2009) Gaucher disease (Packman et al. 2006) as well as from surveys from parents of children with MSUD (Packman et al. 2007).

The domains of schooling and employment capture the construct of transition to adulthood. These questions are drawn from Packman et al.'s studies mentioned above as well as from the literature that describes the transition to adulthood in young people with chronic illnesses (Arnett and Taber 1994; Hartman et al. 2000; Palmer and Boisen 2002; Tong et al. 1998). The domain of medical experiences asks questions about the individual's experience with their health-care team, and elicits suggestions for improving the care of individuals with MSUD. The items for the MSUD interviews were reviewed by a panel of experts consisting of one geneticist, two genetic counselors and one pediatric psychologist and their suggestions were incorporated into the final version.

Objective Measures

Wechsler Abbreviated Scale of Intelligence (WASI) The Wechsler Abbreviated Scale of Intelligence (WASI) measures an individual's verbal, nonverbal, and general cognitive functioning. The WASI is nationally standardized and yields a Verbal, Performance, and Full Scale IQ score. Standardization data have been collected from a large nationally representative sample of children and adults from 6 to 89 years. A prorated IQ score was obtained by administering the Vocabulary and Matrix Reasoning subtests from the WASI.

The Vocabulary Subtest provides a measure of word knowledge and fund of information (Kaufman 1994; Sattler 1992) and has been identified as the best measure of general and verbal cognitive ability. The Matrix Reasoning subtest provides a measure of nonverbal reasoning ability and analysis and synthesis of visuospatial material (Sattler 1992) and has been identified as the best measure of general nonverbal ability.

The Pediatric Quality of Life Inventory (PedsQL) Family Impact Module The PedsQL, Family Impact Module (Varni et al. 2004) was used to measure the impact of the illness on parents and the family. The 36-item PedsQL Family Impact Module encompasses six scales measuring parent self-reported physical, emotional, social, and cognitive functioning, communication, and worry. The Module also measures parent-reported family daily activities and family relationships. Varni et al.'s (2004) preliminary reliability and validity study suggests strong internal consistency (Total Scale Score, alpha=.97; Parent HRQOL Summary Score, alpha=.96; Family Functioning Summary Score, alpha=.90; and Module Scales, average alpha=.90, range=.82–.97).

Data Analysis

All responses to the MSUD Young Adult and Parent Interviews were transcribed. A qualitative case study methodology was used to explore the human experiences of MSUD patients and families. A case study examines phenomena in a real-life context when the boundaries between the phenomena and the context are not clearly defined and multiple sources of data are used Yin (1993). Such an approach allows an investigator to retain the holistic and meaningful characteristics of real life events.

In the current study, the experiences of psychosocial adjustment occurred within the life context of young adults and parents living with MSUD, a chronic illness. The case study methodology facilitated the identification of factors that affect these patients and families and their unique responses. Interviews with young adults and their parents elicited information regarding life experiences, relationships, and medical experiences in the context of MSUD.

The authors examined and coded responses provided by the young adults and their parents. A list of themes, experiences, and concerns were compared across individuals (between young adults with MSUD and between primary caregivers to young adults with MSUD). Discrepancies in coding were discussed and reconciled. Themes were identified and illustrative quotes were selected to explicate our findings.

Results

Description of Sample

A total of 10 families, including young adult patients and their parents, expressed interest in the study. Of these, 2 were excluded for the following reasons: patient was less than 16 years of age ($n=1$); patient was not available to complete the study ($n=1$). The final sample of 16 participants included 8 young adults and one of their parents. Table 1 summarizes the demographic and background data for the young adults with MSUD and parents evaluated in this study. The average full scale IQ of the young adults was 90.88 +/− 25.53 which is consistent with past studies of young adults with MSUD (Hilliges et al.1993; Kaplan et al. 1991).

Qualitative Data

The qualitative data generated from the interviews with young adults and parents are presented below. The interview was organized into 6 interview categories and an attempt has been made to present the major themes within each interview category that have influenced the various domains mentioned above. The six core themes include: 1) the lifelong strain of dietary management; 2) social isolation from peers and impact on dating; 3) impact of MSUD on academics and employment; 4) medical experiences and transition to adult care; 5) impact on family functioning; and 6) positive effects and growth. Supporting quotations from various participants (young adults and parents) are presented at the end of the results section to illustrate the major points (Table 2).

Life Experiences with Respect to MSUD: Lifelong Strain of Dietary Management

The majority of young adults stated that the hardest part of having MSUD is the diet. When asked what they don't like about having MSUD, the consensus answer was "formula" and the "inability to eat a variety of foods." A majority of the young adults indicated that there have been times when they did not follow the diet even though it was harmful to them. According to one young woman, who commented about her own inability to follow the diet,

> The biggest mistake that people with MSUD make is not sticking with the diet. They don't stick with their diet and they feel kind of shorted. They feel deprived—they want everything that other people have.

To cope with the demands of MSUD and the stringent diet, one young man stated that he uses positive self-talk: "I just say it's another day and nothing is going to bring me down."

When asked how they describe MSUD to others, 6/8 stated that they give incomplete information: "People I don't know I just say I'm allergic to protein… People I know a lot more, I say I don't have an enzyme to break down certain amino acids." When asked to describe their condition in more detail, they get frustrated regarding others' lack of knowledge about their medical condition, treatment and diet.

Young adults responded with a range of answers about the hardest age to deal with MSUD. Some responded with ages up to 5 years old stating reasons such as medical fragility and symptom manifestations. Many others responded with the "teen years" because of peer pressure, the desire for independence, and the need to be responsible for their diets. When parents were asked a similar question, a majority of them reported the newborn, infant, toddler years to be the most difficult years due to lack of information about the conditions, frequent hospitalizations, food compliance and diet management issues.

In a related vein, some young adults and parents indicated there were economic burdens due to inadequate or no health insurance. Notably, many necessary items were not covered by insurance, such as specialty foods and therapies.

Impact of MSUD on Relationships

Social Isolation: Relationship with Peers A developmental challenge for adolescents includes separating effectively from parents and establishing a self-identity that is capable of acting independently (Woodgate 1998). Many of the young adults recall social activities being limited in high school. Suris et al. (2004) found that the development of peer relationships and self-image issues may be in conflict with the demands of chronic illness. The authors note that some young adults with diabetes find it unacceptable to take lunchtime injections or those with cystic fibrosis may refuse to carry inhalers. These young adults prioritize social time with peers (i.e., "fitting in") over disease control. Similarly, many participants in the current study chose not to drink formula in front of other people or only drank their formula at home. A majority of participants stated that they faced a dilemma of disclosure when they were asked to describe MSUD to their peers or others. They stated that they were afraid of being socially isolated from their peers if their

Table 1 Demographic and background information—young adult patients with MSUD and parents

Variable		Young Adult Patients (n=8)	
Gender			
Males		3	
Females		5	
Current Age			
Mean		21.90 years	
Range		16–27 years	
Educational Status			
Attending High school		1	
Attending Community College/Taking post-high school course		5	
Attending College		1	
College Graduate		1	
		Parents (n=8)	
		Number	
Educational Background			
High school diploma or equivalent		1	
1–3 years of college or equivalent		5	
2 or more years of graduate school		2	
Marital Status			
First marriage		4	
Divorced		1	
Remarried		3	
Total Yearly Income			
\$26,000–\$40,000		1	
\$41,000–\$60,000		1	
\$61,000–\$80,000		1	
\$81,000–\$100,000		1	
\$101,000–\$200,000		1	
More than \$200,000		1	
Do not recall		2	
WASI	Full Scale IQ	Percentile	Confidence Interval
Participant 1	59 (Extremely Low)	0.3	55–66
Participant 2	60 (Extremely Low)	0.4	56–67
Participant 3	88 (Low Average)	21	83–94
Participant 4	90 (Average)	25	85–96
Participant 5	90 (Average)	25	85–96
Participant 6	92 (Average)	30	87–98
Participant 7	124 (Superior)	92	115–126
Participant 8	127 (Very Superior)	96	120–132
Summary	Mean=90.88, Standard Deviation=25.53		

WASI Full Scale IQ has a mean of 100 and standard deviation of 15

friends found out they had MSUD. They stated that the teenage years were especially hard—they were aware of the medical fragility of their condition and yet, feared peer rejection. They expressed frustration about following a stringent diet and not being able to fit in with their friends.

Overall, a majority of the participants experienced social isolation and expressed frustration about having the disease. Most of them felt following the diet with the restrictions it imposed on them was very difficult. They felt isolated from their peers as they were not able to enjoy the same foods.

Relationships with Immediate Family When asked how MSUD influenced their relationship with their parents, young adults responded with a range of answers including feeling dependent and frustrated to having no effect on the relationship or even feeling closer. The main thing the

Table 2 Supporting illustrative quotations: MSUD young adults and parents

Interview Category: *Theme*	
Life Experiences: Strain of Dietary Management	
Frustration with others lack of knowledge	"Some people think that you can't do things like they can… like you're not as smart as them, and you are. You can be just as smart as any other person, even with MSUD."—27 year-old woman
What age is the hardest in dealing with MSUD?	"When you start reaching the end of your teens, it becomes a burden after that. After that, you're actually on your own. You've got to know to do things yourself. Your parents really support you so much. But after that, you have to try and do everything yourself.... Watching your diet, managing your diet, making sure how much of your products you've got, making sure you can actually put in a prescription order before you run out of stock, making sure you do blood tests, the list goes on. Those are things you need to take into consideration, but there's other things you've got to learn to maintain, your diet- you've got to learn to be in charge of it."—21 year-old woman
Impact of MSUD on Relationships	
Social Isolation: Relationship with Peers	"I didn't like that I couldn't eat like all my friends I did. Everybody's like 'ohhh ice cream's so great' you know? And 'pizza's so amazing' you know?"
Dating Experiences:	*Message to young adults that they date*
	"Just be yourself. I want someone to like me for me."
	—27 year-old man
	"It shouldn't get in the way."—27 year-old man
	"To be straight with the person they're dating, and if they don't accept it, just don't pay attention to it. There will be other people who will understand and will accept them for who they are"—21 year-old woman
Impact of MSUD on Academics:	
School Experiences	*Advice to those with MSUD*
	"Keep a positive attitude and keep your head up high. Don't let it control your life."—27 year-old man
	"You can be just as normal as everybody else."
	—19 year-old man
	"I would say get involved with something, don't be a loner."—17 year-old woman
	"Just try and be patient, try and get along with people. Ignore all the comments that people make about MSUD because they don't understand."—21 year-old woman
Impact of MSUD on Family Functioning	"Every time he is sick, every time the phone rings in the middle of the night, I feared that he would get sick and die. That's the bottom line."—parent of young adult with MSUD
	"MSUD definitely takes time away from the family. In the younger years, I was too busy with the kids and we had no time for each other."—parent of young adult with MSUD
	"It is difficult because I can't do the things that I would want to do without taking into consideration how my child is going to manage, whose going to look after her, or is she going to be okay?"—parent of young adult with MSUD
Medical Experiences:	
Transition to adult care—being seen in a children's hospital	"Felt weird. I walk in and there are a bunch of kids there."—27 year-old man
	"It's a little embarrassing because last time I went and got a blood draw I drove there by myself and signed papers."
	—19 year-old man
	"Well, now I kind of feel stupid because I'm the oldest one there. I feel too old. I think the oldest person next to me was a 10 year old, and like yeah, there's seven years between me and him!"—17 year-old woman
	"No! I want to go to an adult hospital. I'm an adult, not a little kid."—27 year-old woman
Positive Effects and Growth:	"Even if you have struggles in life …you're just as smart, or just as good as anybody else. Yeah, so you can't look down on others because of things that they have in their life. So I guess it taught me not to look down on others now."—27 year-old man
What MSUD has taught me:	"Having MSUD has taught me that no matter how rare a condition is, you just keep going. That's all I can say. Just keep doing what you can or what your body can do. That's what I want to keep saying. What your body can do."
	—19 year-old man
	"Has taught me that what I have is not a huge thing to worry about because there's more worse cases out there in the world. Hopefully, someday there will be a cure."—21 year-old woman

young adults report is that their parents have been the most helpful by providing support, guidance and help with the diet. Young adults with MSUD who had other siblings with MSUD ($n=3$) appreciate having someone else with whom to share similar experiences.

Romantic Relationship Experiences (Dating, Marriage, Children) Establishment of intimacy is among the developmental tasks that occur during the young adult years. Intimacy involves establishing emotional closeness to other people as a basis for enduring relationships (Muuss 1988). To better understand how MSUD might influence romantic relationships, questions addressing dating and marriage were asked. Most young adults worried about acceptance and the reaction of their dates to MSUD yet reported that the person they were dating did not stop dating them after they told them that they had MSUD. The young adults initially explained that they had eating restrictions (e.g. vegetarian or lactose intolerant) and most said they worked around dinner dates by eating salad, fries and/or vegetables. None of the young adults have allowed their dates to taste their formulas.

Four out of eight of the participants wished to marry someday. One young man said he would not marry, and one participant mentioned he was not at that stage yet. Two young women mentioned they did not think about it and are not worried about marriage and having MSUD.

Young adults were asked to consider the possibility of having children similarly affected as themselves with MSUD. Half of the young adults wanted to know if their future partner is a carrier for MSUD. A range of responses were provided—no concerns, not wanting to give a child MSUD, and not wanting the added responsibilities of having a child with MSUD.

However, if the young adults were to have a child with MSUD, they would manage their child's diet in the following ways, "the way my parent did… take away food not allowed," "track protein according to doctor's orders," "allow them to cheat so that they can learn consequences." Ultimately, most young adults said they would love and accept their child if they were found to have MSUD and try to normalize their child's life.

Impact of MSUD on Academic and Employment Milestones

School Experiences Chronic illness and the demands of treatment regimens may have a significant impact on school functioning, school attendance, and educational achievement (Storch et al. 2008). Among the challenges for individuals with MSUD is the absolute requirement of taking their formula. When asked about their memories regarding grade school and formula some recalled that they would drink it at home while others would bring it to school and drink it at school with the help and support of a teacher, nurse or school dietician. In the academic realm, some young adults reported learning disabilities that required special classes or extended time to take tests and complete work. Some advice young adults with MSUD offer for making the grade school years easier included: sitting in the front of the class to take notes, "don't make a big deal out of it," and attend a special needs school before attending a mainstream school.

During the high school years there is a new set of challenges in the physical, emotional, social, and educational domains for any adolescent. When specifically asked how they coped with social situations in high school a majority of young adults mentioned their condition limited their social activities. Two of them stated that they were not allowed to go out with friends and three others stated that they drank their formula and ate food at home before going out.

Memories parents recalled about grade school included delayed academic development, self-esteem and behavioral issues, and second guessing educational placement (mainstream school vs. home schooling). In terms of managing the diet during the grade school years, parents recall constantly communicating with the school nurse about the diet and food logbook, sending extra things to school for teachers for class parties, instructing the child to not throw anything they hadn't eaten away but bring it home to determine protein intake, packing formula and lunches so that the child learned what s/he could and could not have. During the high school years the parents reported that their child was frequently fatigued and ill and they were often unable to participate in school activities. Parents described feeling "scared" that their child would be taken advantage of or concerned that their child felt isolated. Some parents who had children with both ADHD and MSUD experienced behavioral and academic problems. Another parent had no issues because of the special education placement that separated their child from same-aged peers. The collective advice parents offer to other parents of adolescents with MSUD in high school is "do the best for your child, keep life normal, set goals, stay involved with the school and keep in mind that every child is different."

The college years present new challenges and beginnings and are often the first time the young adult is making decisions. There is a challenge of dependence vs. independence as a young adult takes over responsibility for the management of their chronic illness from his or her parents (Tong et al. 1998). By parent report, factors such as developmental delays, hospitalizations and school absences due to illness interfered with the young adults' post high school education. At the time of this study, one of the eight young adults had graduated from college. The majority (6/8) of the young adults had taken post high school courses (i.e., started some course work in college, special education courses at community colleges) while the remaining young adult was still in high school. The young adults studied a variety of subjects including computer science, basic functioning skills (self–care, transportation, money exchange), multimedia, physical therapy, arts and crafts.

Work Experiences and Vocational Achievement Developing a realistic vocational goal is another challenge for young adults. Among the eight participants, half had a part-time job and two were looking for jobs. They worked in a variety of jobs that ranged from shoe sales, stocking shelves, odd jobs in a fast-food restaurant to computer labeling and sorting mail. The young adults stated that their employers were aware of their medical condition and diet restrictions. When they were asked what they would tell someone with MSUD about having a job two of them said not to disclose their medical condition at work, while two others said bring your formula to work as it helps keep the energy level up. One of the participants said—"just make sure that whereever you work, there's something you can eat." It is clear from these responses that the diet imposes restrictions on how much and how long they can work.

While the major concerns for young adults were fatigue on the job and dietary issues and restrictions (i.e., trying to find something to eat everyday), parents were also concerned about their child's inability to do work because of fatigue, going too long without eating, and becoming less able to communicate when levels are off.

Medical Experiences and Transition to Adult Care

Medical care for individuals with MSUD is generally monitored by healthcare providers familiar with inborn errors of metabolism (IEM). Most young adults with MSUD rated the medical care they received highly (9–10 with 10 being best). "Ten because the doctors are very knowledgeable and if I say I'm slightly out of whack…they are very helpful." Another response was—"Because they tried to do everything to get my levels down and they always tried to manage my diet—changing my proteins from time to time." While some of the parents also rated the medical care highly some did not mainly due to frustrations on many levels including inconsistency with care, poor communication, mixed level of expertise (especially with dieticians), and lack of understanding in what parents perceived to be a crisis situation.

The young adults were asked if they were treated at a children's hospital and their impressions of receiving care at a children's hospital. A majority of the participants received their care at a children's hospital and although they were very happy with their medical care none of them liked going to a children's hospital because it "was embarrassing or made them feel weird." Similarly, some parents reported concern regarding their young adult child with MSUD continuing to receive their care in a pediatric setting but most reported being satisfied with care mostly because there is no other alternative.

Impact of MSUD on Family Functioning: Quantitative Findings from the Family Impact Module

The PedQL Family Impact Module (Varni et al. 2004) measures the impact of a chronic illness on parents and the family. It assesses domains of parent functioning (physical, emotional, social, cognitive, communication, worry) and family functioning (daily activities, and family relationships) and provides a total Family Impact Score, Family Functioning Score, and Parent HRQoL score. When our sample of parents of young adults with MSUD were compared with an inpatient normative sample of medically fragile children who reside in a long term care (LTC) facility (Varni et al. 2004), there was a measurable impact on the families of MSUD patients in emotional, social, and cognitive functioning, worry, daily activities and family relationships as well as in the 3 summary scores (Parent HRQoL, Family Functioning, Total Impact) (See Table 3).

In the area of parent functioning, the lowest scores were in the emotional and worry domains. These results are consistent with and complement our findings from interviews with parents. When asked to report the hardest thing they face with respect to MSUD, parents reported the following concerns: fear of death, worry about the future, hospitalizations and illnesses, and concerns about managing the diet. One parent noted that cultural stigma, shame, societal ignorance, and fear result in isolation of a child with MSUD. Parents cope with these issue in a variety of ways including avoidance, using support systems (such as family, friends,/therapist), and "having emotional breakdowns" at times. Parents reported that they find the MSUD Support Group helpful because it provides education on MSUD and creates a feeling of normality because they meet others that are similar.

In the area of family functioning, the lowest scores were in family relationships. This is also consistent with interview findings in which parents stated that they must take on additional roles and responsibilities because of the special health and medical needs of their children. While most parents said that having a child with MSUD has not influenced their relationship with their spouse, two parents said that it did have an impact because it took time away from marriage, and created frustration and conflicts with their spouse because of differences in parenting styles. Most all reported that relationships with extended family members were affected. Reasons for the affected relationship included feelings of isolation, desensitization of severity of MSUD by family members due to frequent hospitalizations, differential treatment and issues of blame. Parents also stated that

Table 3 Pediatric quality of life inventory version 2.0 family impact module

	Young Adults with MSUD *N*=8	LTC *N*=12	Home *N*=12
Domain			
Physical	78.85 (16.45)	82.99 (17.36)	53.03 (22.83)
Emotional	63.75 (12.46)	78.33 (18.26)	64.48 (26.59)
Social	75.00 (21.91)	85.42 (17.34)	61.93 (25.99)
Cognitive	77.19 (21.19)	88.75 (12.81)	74.09 (18.95)
Communication	77.08 (19.80)	73.61 (24.58)	52.15 (24.67)
Worry	59.38 (20.08)	69.17 (21.09)	56.82 (25.52)
Daily Activities	78.13 (20.38)	85.14 (24.75)	51.89 (31.48)
Family Relationships	65.62 (17.41)	83.75 (23.07	78.95 (27.62)
Parent HRQL	73.33 (16.54)	83.75 (15.55)	62.94 (19.83)
Family Functioning	70.31 (16.28)	84.27 (20.47)	68.81 (24.11)
Total Score	71.27 (15.11)	81.0 (17.06)	62.49 (17.26)

Scale=0–100. Standard deviation presented in parentheses. Higher scores indicate better functioning. LTC=Medically fragile children who reside in a long term care facility; Home= Medically fragile children who reside at home

sibling relationships were influenced because the non affected sibling would feel "embarrassed" because of their affected sibling. According to parental reports, the MSUD affected child had some adjustment issues and experienced jealousy. Parents with more than one child with MSUD reported feeling less anxiety because of their previous experience with a child with MSUD. Most parents reported more support and understanding from friends than from extended family.

Positive Effects and Growth

While it is clear that MSUD places an emotional burden on patients and families, the participants also reported that MSUD has had a positive influence on their lives, leading to a shift in priorities and a world view that is more compassionate and caring. As noted by one parent:

> It's changed my whole life.... It has opened my eyes to a lot of things. It has made me a more quality person—but I really think I've always been compassionate but I think I am more compassionate toward other kids.

All young adults and parents reported that having MSUD or a child with MSUD, respectively, has given them advantages such as the instillation of hope, the ability to cope, compassion, tolerance, personal growth, and empathy.

Notably, most of the young adults did not report negative effects specific to their relationships with family or with their social life in general. According to Martire and Schulz (2007), support from and interactions with close family members have positive effects on the emotional well-being of individuals with chronic illness. Participants indicated that having MSUD deepened their family relationships and that the family felt protective.

Discussion

The goal of this study was to investigate psychosocial issues of young adults with MSUD as they transition into adulthood. The participants expressed many psychosocial concerns and recurrent themes among participants were revealed. Specific concerns experienced by young adults included: 1) lifelong strain of dietary management; 2) social isolation from peers and impact on dating; 3) challenges in academics and in employment; 4) medical experiences including transitioning to adult care; 5) impact on family functioning; and 6) positive effects and growth.

We need to be mindful of the many demands and psychosocial issues that families of young adults with MSUD face and what is *unique* about MSUD *as a chronic illness*. MSUD (and other inborn errors of intermediary metabolism) (Cederbaum et al. 2001) are characterized by: acute metabolic decompensation(s); the possibility of neurodevelopmental delays; treatment by nutritional manipulations and supplements; and, ultimate treatment by liver transplant. There is a lifelong strain of dietary management as well as cognitive disabilities. The patients face ongoing metabolic decompensation and a risk of death with decompensation. There are multiple visits to hospitals and physicians. The diagnosis can be in the newborn period or very early infancy; therefore, it is a life-long process, as opposed to cancer, diabetes, or even other inborn errors (Packman et al. 2006), which may strike an adult. Conventional treatment does not cure the disease and the treatment may not be effective. There is a lack of control over the treatment parameters when the child gets older.

A majority of the young adults felt frustrated with having MSUD, had difficulty following the diet, felt socially isolated and not able to fit in with their friends. In the view of the young adults, the teenage years were the hardest in terms of dealing with MSUD because of "peer pressure" along with the need to be responsible for their diet. The young adults had multiple challenges in school, work, and in dating. In grade school, the greatest challenge (for young adults and parents) was the requirement of taking formula. This was a major strain for young adults and a source of considerable stress for parents. In high school, young adults continued to experience restrictions related to eating and participating in social activities with friends. In the vocational arena, the dietary restrictions continued to play a major role in how much and how long the young adults can work. Dating and romantic relationships were also a challenge as most young adults worried about acceptance by their dates. Findings from the interviews suggest that a majority of the young adults with MSUD have a normal desire to date and marry. In general, the desire to have a child similarly affected as themselves was not desirable but acceptable should it occur.

With respect to medical experiences, as children with MSUD continue to survive into adulthood the necessity for transition into adult care needs to be addressed. In general, young adults with MSUD reported feeling "weird," "upset," and "embarrassed" with continuing to obtain their medical care in a pediatric setting. While some of the parents reported concerns regarding their young adult child with MSUD continuing to receive their care in a pediatric setting most reported being satisfied with care.

A paper published by Lee (2002) addresses the difficulty faced by individuals with chronic conditions as they transition from childhood to adulthood. The authors state that having any metabolic disorder impinges on everyday life and people with any IEM have multiple physical, learning, psychological and social problems. In addition, the treatments of these disorders are complicated and difficult to administer. Hence, many adults are still seen within pediatric settings. In fact, only very few medical centers offer special medical care for adults with IEM (Simon et al. 2007). Therefore, further investigation into adult care facilities for these patients may be beneficial.

In the case of MSUD, as with other IEM, there are economic and psychosocial barriers to transition. The first barrier is access to insurance coverage (i.e., young adults with chronic illnesses such as MSUD can have more difficulty obtaining insurance due to insurance policies that exclude coverage of their genetic disease) (Callahan and Cooper 2007; Wiener et al. 2011). Psychosocial barriers include developmental readiness. Youth with cognitive or neuropsychological deficits may especially be at risk for difficulty with transition (Wiener et al. 2011). The IQ scores of young adults with MSUD tend to fall in the low average to average range with some individuals classified as extremely low (FSIQ less than 70). Cognitive and/or neuropsychological deficits can effect academic functioning, complicate diet adherence, the ability to communicate healthcare needs, and decision-making (Wiener et al. 2011; Malee et al. 2008).

Finally, the present findings suggest it is difficult to locate adult providers who are as skilled as pediatric providers in addressing all aspects of MSUD. In a related vein, Wiener et al. (2011) reported that HIV+adolescents identified barriers for transition from pediatric to adult care. These barriers included a perceived lack of competency among adult providers in regards to the specific medical and psychosocial needs of HIV+adolescents.

Research Recommendations

As more adolescents with MSUD survive longer and enter adulthood, the issues of transition from the pediatric setting to an adult setting is becoming an important topic to address. In one study of young adults with PKU transitioning from pediatric to adult care, an interdisciplinary consultation-hour of pediatric and adult specialists was highly recommended (Mutze et al. 2011). In addition, it was suggested that young patients be involved in the transition process. Patients with PKU have also suggested that they would not prefer transfer at the same time as other life events (e.g., graduation from school). It may be beneficial for future studies to develop and examine transition models for patients with MSUD. Helping young adults with MSUD in their transition would serve to increase their sense of independence in all aspects of their lives, especially those aspects affected by their illness (Freed and Hudson 2006). Another area warranting future study involves training and educational opportunities for health professionals, patients, and families. Adult providers may require more education on how to care for younger patients with MSUD. Likewise, pediatric providers may require education on how to meet the needs of older patients. In addition, patients and families may require training to address what to expect in transition to adult services.

Study Limitations and Practice Implications

Some methodological issues should be noted. The sample used in this investigation consisted of all Caucasian individuals. Therefore, it is hard to predict whether the experience of this sample is similar to that of a different ethnic cohort of MSUD patients and families. Another limitation concerns the small sample and predominantly qualitative nature of the findings. Qualitative data are not intended to be generalized to the population of interest. A possible confound may be due to the retrospective reporting that was necessary to obtain data regarding development and diagnosis. The unreliability of retrospective memory may impact the assessment of patients' and families' memories of growing up with MSUD. Finally, the majority of participants in this study were drawn from a support group, and therefore may not reflect the full spectrum of the disease in terms of its medical manifestations or emotional, social, and developmental impact. On the other hand, given the rarity of the disorder, it is reasonable to consider that many, if not most families would be affiliated with this support group.

The results of this investigation further awareness of the psychosocial impact of living with MSUD and provide a valuable contribution to the genetic counseling literature. This study calls for a collaborative, multidisciplinary effort in the treatment of these unique patients and their families. It is essential to involve geneticists, genetic counselors, nutritionists, psychologists, and individuals who have expertise in insurance coverage and social work. While MSUD clearly places a major burden on young adults and parents, participants reported that it can also have a positive impact and allow one to be more caring and compassionate.

This was the first investigation of psychological issues faced by young adults with MSUD transitioning into adulthood. The data suggest that young adults experience psychosocial stressors throughout childhood, adolescence, and young adulthood. As MSUD patients are confronted with challenging medical conditions on a daily basis, providing these individuals with adequate support and care in all areas of their physical and psychological being is essential.

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