

Relationships Between Childhood Experiences and Adulthood Outcomes in Women with PKU: A Qualitative Analysis

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Abstract *Background:* The enduring impact of the childhood experiences of people with phenylketonuria (PKU) on their adulthood outcomes is equivocal. As the effect of childhood experiences on adulthood is well documented amongst the general population, the aim of this study was to explore childhood experiences considered significant by women with PKU as they relate to adult experiences and management of PKU, and psychological wellbeing.

Method: Eight women with PKU in South Australia underwent semi-structured interviews. The audio-recorded interviews were transcribed verbatim and analyzed using thematic analysis.

Results: Interviews revealed that feeling different to peers as a child, challenges with management of the condition during adolescence, parental and extended family support, and the perception of PKU as a burden during childhood were associated with adulthood experiences.

Conclusions: Thus, it is proposed that these childhood factors have a combined, long-term impact. These findings have significant clinical implications, suggesting that early psychosocial intervention relating to these identified childhood experiences has the potential to enhance positive outcomes for adults with PKU.

Successful management of phenylketonuria (PKU) can prove onerous across the lifespan (van Spronsen and Burgard 2008). Several factors influencing the experience of PKU during childhood have been identified. Family cohesion, comprising parental support (Shulman et al. 1991; MacDonald et al. 2010) and extended family support (MacDonald et al. 2010) are associated with dietary control. Conversely, children with separated or divorced parents (Olsson et al. 2007; Alaei et al. 2011), an unemployed parent (Alaei et al. 2011), or low socioeconomic status (Weglage et al. 1992) had reduced dietary control and elevated phenylalanine (Phe) levels (MacLeod and Ney 2010). Anguish and feelings of isolation have also been found to result from awareness of the differences in eating behavior between young people with PKU and their peers (Vegni et al. 2009). Adolescence can be particularly challenging. Adolescents experience less life satisfaction, restricted social experiences, and lower frustration tolerance than peers without PKU (Weglage et al. 1992). Additionally, adolescence has been associated with reduced dietary compliance (MacLeod and Ney 2010).

Despite the lifelong nature of PKU, there is little research on the influence of these early experiences on the adulthood experiences of the condition. To our knowledge, only MacDonald et al. (2010) have highlighted the important role of parental encouragement on children's

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independence and increasing responsibility in relation to dietary control and long-term adherence to the PKU diet. This is because overreliance on parents for PKU treatment may negatively impact adjustment to full responsibility in adulthood (Bosch et al. 2007).

Whilst research specific to people with PKU is limited in this area, the long-term impact of childhood experiences is well documented within the general population. For example, childhood adversity has been associated with poor health and psychological distress in adulthood (Kestilä et al. 2005), whilst dietary patterns established in childhood extend into adulthood (Mikkilä et al. 2005). As such, it is proposed that the experiences of children with PKU would impact on their adult experience of the condition, psychological wellbeing, and management of the condition.

Irrespective of childhood experiences, and despite quality of life among adults with PKU being comparable with the general population (Mütze et al. 2011), adults with PKU experience greater psychological morbidity than the general population (Ris et al. 1997), including anxiety and depression (Pietz et al. 1997). The risk of mental health problems is further increased when one is non-adherent with the PKU diet (Koch et al. 2002). Accordingly, emphasizing childhood experiences that may impact adult outcomes is of significant clinical interest – early intervention may optimize outcomes for individuals with PKU not only during childhood but also across the lifespan.

This qualitative study aimed to explore the experiences of women with PKU, gaining an account of factors considered significant during childhood from the women's own perspectives. Subsequently, tentative links between childhood experiences and adult outcomes in terms of overall experience of PKU, psychological wellbeing, and PKU management are explored.

Method

Participants

All South Australian women known to be diagnosed with PKU were invited to participate ($n = 20$). Eight women participated (40% response), aged 21–42 years ($M = 29.9$, $SD = 7.9$). Reasons given for non-participation were primarily related to insufficient time. All participants were diagnosed and treated for PKU from soon after birth and all had tested intellectual functioning in the normal range. All but one of the participants ($n = 7$) were maintaining the PKU diet at the time of the interview, and were taking a supplement (medical formula). Six women were having regular blood tests although the frequency of testing ranged from weekly to every four to six weeks. Lifetime Phe levels were estimated based on a review of case notes by a

Table 1 Participant characteristics

Participant number	Age range	Education	Diet status	Phe levels in target range (approximate % over lifetime)
1	20s to 30s	High school	Off diet	<10%
2	20s to 30s	Post high school	Strict	100%
3	20s to 30s	Post high school	Strict	100%
4	20s to 30s	High school	Strict	100%
5	20s to 30s	Post high school	Strict	100%
6	30s to 40s	Post high school	Relaxed	100%
7	30s to 40s	High school	Relaxed	100%
8	30s to 40s	Post high school	Relaxed	25%

Metabolic Physician (DB). For six participants, Phe levels were within acceptable ranges for the majority of the time, one for approximately 80% of the time, and one for less than 10% of the time (see Table 1). Five women were married/de facto and three were single. Seven women were employed, four had completed Year 11–12, two had vocational qualifications, and two had university degrees. Three participants were undertaking tertiary education. Three participants (37.5%) were assessed on the M.I.N.I. as experiencing significant psychological disorders in their lifetime (two participants had a history of major depressive disorder; one of whom also was experiencing recurrent bulimia nervosa; one participant had a history of bipolar affective disorder, with a current depressive episode).

Procedure

This study, approved by the Human Research Ethics Committee of the Children, Youth, and Women's Health Service, forms part of a study to explore women's experiences of PKU in relation to pregnancy (Roberts et al. 2014). As such, participants were all female. Experiences of PKU across the lifespan were explored within the initial study and childhood experiences were noted to be of importance in relation to outcomes in adulthood, resulting in the current research.

Semi-structured interviews (which took 30–90 min, depending on the length of participant's responses) were audio-recorded, then transcribed. Interviews began with women being asked "Can you tell me about the impact that PKU has had on you throughout your life?" (see Appendix for full list of interview questions). Participants' experiences of PKU across their lifetime were explored (reported here), followed by their perceptions and/or experiences of pregnancy (reported in Roberts et al. 2014). Interviews were analyzed using thematic analysis (Braun and Clarke 2006). Initial interviews, data coding and sorting into

themes was conducted by TR, a graduate clinical psychology student with basic training in PKU management. Reliability of coding and themes was verified through review with two psychologists/researchers with extensive experience working with people with PKU (RR, AG).

In addition, The M.I.N.I. (English Version 6.0.0) was administered. The M.I.N.I. is a brief, verbally administered questionnaire assessing lifetime experience of psychological disorders (Sheehan et al. 1998).

Results

Participants are identified by age group, education, and self-reported dietary adherence – strictly on diet, relaxed on diet or off diet (Table 1). Thematic analysis identified five themes relating to childhood experiences of PKU (Table 2) and three themes relating to adulthood experiences of PKU (Table 3). We also describe the association between childhood experiences and adulthood outcomes reported by the women.

Table 2 Childhood themes

Theme	Examples
1. Comparisons to and feeling different from peers	<p>...you see all your friends eating this and then you're like, well I can't have that and they're all buying their lunch and you can't buy nothing cos you know, it's pies and pasties and everything else. (Participant 1)</p> <p>Back then it's, oh my god what's wrong with you! You know, you can't eat that, why, what's wrong with you, is something wrong with you, you know that sort of attitude... It was hard, very difficult. (Participant 4)</p> <p>So I was sort of, almost felt an oddball, being a child growing up with PKU, or a teenager. (Participant 7)</p> <p>...there's just me with like, my you know, different dish and you know it makes it a bit hard, hard to do, but it never bothered me. (Participant 3)</p>
2. Management of PKU during adolescence	<p>... It [management of PKU] was ok probably to my teens... I sort of strayed there and just had whatever... just the chips and you know, a pasty or... not counting anything. (Participant 8)</p> <p>... high school was worse... you eventually stop eating at school... and you know, you just think, I'm not eating at school, and you slowly drift from the diet. (Participant 1)</p> <p>... I remember at 14, and for that, for like nearly a year it was really, really hard to get me to take my formula. (Participant 6)</p> <p>Girls are trying to fit in, they're going through puberty and there's boys and this and, so it's really tough. And then trying to stick to the diet. You're not always gonna do it, you're gonna fail eventually. You think, what's the point? (Participant 1)</p> <p>...back then you think you can go on without doing the right thing for your body (Participant 8)</p> <p>...a teenager's brain would not care at all about what happens in the future. (Participant 6)</p>
3. Parental support	<p>...she [Participant's Mother] went out of her way to prepare stuff for me so I did have stuff there and stuff that was, I suppose special to me, and so it didn't make me feel like I was the oddball as much... She'd make me fairly bread out of my own bread and so I'd be able to eat the fairy bread when the other kids were eating fairy bread... (Participant 7)</p> <p>...mum's always been a great cook and she kind of always adapted recipes to suit me and I think that helped a lot because you know, there's, I guess I would still be having like normal food, but with the low protein food. (Participant 3)</p> <p>...mum never experimented with me, like with the drinks and food and stuff. (Participant 1)</p> <p>Mum would get me in the kitchen and we'd bake, make all the low protein stuff... (Participant 5)</p> <p>...I guess for many, my, my mum did pretty much everything for me. (Participant 8)</p> <p>... extremely thankful to my parents for sticking to the diet so umm, so greatly and so accurately, because you know, here I am, I'm a, you know, I'm a graduate student, I've got a profession, and if it weren't for their compliance and their understanding of PKU and, and what my needs were as a child and as a baby, I wouldn't be here today. So I have them to thank for that and I'm very grateful. (Participant 6)</p>
4. Extended family support	<p>... if there was a big party or something, they'd ring mum and say, you know, what can she have. Or we've made her this, so she can have that as well... always made sure I had something so yeah, it was good. (Participant 5)</p> <p>... yeah they were supportive, they always said, if we went over there for tea, they always said, what can [participant] have, what can she eat, and all that type of thing, so yeah. They were all supportive. (Participant 7)</p>
5. Perception of PKU as a burden	<p>I wouldn't really call it more an impact, it's just, it's just I've grown up this way, it's all I know. (Participant 5)</p> <p>So, it's sort of like, ok, how it is, that's how it is and you don't know any different. (Participant 4)</p> <p>Having to eat all my low protein foods and do all that type of thing and have my supplements, take them to friends' places. Or even something as simple as going on camps and stuff like that... That would probably be the hardest thing. (Participant 7)</p>

Table 3 Adulthood themes

Themes	Examples
1. Management of PKU	I like the liberty of having a normal cookie or biscuit every now and again. (Participant 6)
2. The unique experience of having PKU	
2a. Eating in a social context	<p>...when they [restaurants] make salads, some of the salads, like Caesar salads or something, has cheese in it and say could you not have cheese, and they look at you like you're weird. . . I just feel different. . . it always brings your mood down. (Participant 1)</p> <p>...But it's just so hard, like even when you're happy, you go out for tea and, you could just imagine if you were on the diet, then what would you eat? You know, places like that don't exactly have menus for PKU people, do they? (Participant 1)</p> <p>...so really socially, it's probably been an impact and going to parties and things like that. Having functions on where umm. . . I almost have to eat before I go, I can't eat there. (Participant 7)</p> <p>It's normally fine. . . I try not to eat tons of protein during the day, so I can have like a bowl of pasta or things like that. . . I really like Asian food and Indian food which is great because it's just easy to get a pure vegetable dish. (Participant 3)</p>
2b. Effects of dietary compliance	<p>...when my levels get higher umm. . . obviously the mood swings umm you feel, you get like, you feel sort of a little bit lethargic and you don't really want to do anything and you, it takes a while to umm to process stuff and just, my thinking's just probably a little bit slower. . . (Participant 5)</p> <p>I can feel it. I just go, oh I feel like my brain's all fuzzy and I can't think straight. (Participant 6).</p> <p>... having depression as well and not being on diet really is a bad combination. (Participant 1)</p> <p>So everything was just so much better. And mood swings were probably better too. . . huge change just in how I handled day-to-day things with stress and stuff like that. (Participant 7)</p>
2c. Transition from childhood to adulthood management of the condition	<p>...like counting out properly and that type of stuff, yeah. . . she [her mother] used to make a lot of food for me, so she did it all, so umm. . . yeah I didn't really do it myself, kind of thing. (Participant 8)</p>
2d. Attitudes and perceptions	<p>I don't really worry about it, I don't think about it. . .I've had it my whole life, it's who I am, you know. (Participant 2)</p> <p>You know, it's just kind of been part of my life so I've just dealt with it and I just know exactly what I can eat and I can't eat and how much of, you know, something I can eat and that kind of thing. (Participant 3)</p> <p>...oh my god, this is so hard, I've gotta weigh everything, I can't go out, I can't do this, I have to take my own food with me, so it is a big, it is a big stress. . . (Participant 7)</p> <p>... your life revolves around your diet. . . you always think about what you're going to be eating. (Participant 6)</p>
3. Psychological wellbeing	<p>...my eating was umm. . . a problem for me. . . so I think it [PKU] might have been a part of it. Like it was all, like cuz I wasn't allowed. I didn't have a choice, I wasn't allowed to. . . (Participant with previous eating disorder)</p> <p>...you need to be working on both things at the same time. So getting back on diet and look after yourself, after your mental health, is two things that need to happen together. (Participant 1)</p>

Childhood Themes

Comparisons to and Feeling Different from Peers

A key theme noted within discussions of childhood was feelings of difference between the women and their peers, predominantly related to eating within a social context, such as at school or parties. Whilst discussed by the majority of women, the experience of such differences varied. Some women found it to be a negative experience, associated with feelings of isolation, whilst others were unconcerned and did not feel excluded or distressed. The women who found such differences to be a negative experience described mealtime away from home as challenging as they received questions regarding their restricted diet, heightening feelings of

difference and exclusion. In contrast, other women noted that they were aware of differences between themselves and peers, but this did not result in distress.

Management of PKU During Adolescence

Women noted that management of PKU was challenging during adolescence, resulting in reduced dietary compliance. The reasons noted for reduced compliance were similar. Two women attributed it to the general challenges of adolescence, with Participant 6 reporting that she lived her ". . .frustration as a teenager through my diet." Reduced compliance was also attributed to the perception of adolescence being a period associated with limited consideration of the future consequences of present actions.

Parental Support

All women spoke of the support received from their parents in childhood, highlighting the ways in which their parents assisted in the management and experience of PKU. Whilst all women emphasized the importance of this care, the nature and level of the support received varied. Several women reported receiving high levels of parental support which centered on reducing their feelings of exclusion and difference. For example, for social events with peers, parents routinely prepared and brought low-Phe food for the children similar to those foods eaten by others. These women reported that this assisted them to feel included, minimizing feelings of difference. In contrast, others indicated that the support received was more limited. Whilst Participant 1 stressed how adapting “normal” meals to be low-Phe helped to “. . .deal with the differences. . .” as a child, she reported minimal experimentation with dishes within her family. Of note, these women reported more feelings of difference from their peers.

Additionally, differences in the nature of parental support were identified. Some women reported that their parents included them in meal preparation, which assisted the development of their skills in the management of PKU in addition to providing an enjoyable experience. In contrast, one woman reported that her mother took responsibility for her diet and that she was rarely involved. She felt that this resulted in a lack of development in skills and contributed to difficulties in her adjustment to independent management of the condition as an adult. Regardless of the nature, parental support was appreciated and perceived as important.

Extended Family Support

The involvement of extended family members in the management of PKU was discussed, specifically with regard to catering at family events. When extended family members were not involved in the preparation of “PKU-friendly” meals or lacked knowledge of the condition, women felt excluded, for example, Participant 1 claimed that her extended family saw her as an outcast due to her condition.

Perception of PKU as a Burden

All women commented on the overall impact of PKU on their childhood and adolescence. Women who did not indicate that their condition was a significant burden instead appeared to have accepted the requirements for management, perceiving PKU as a way of life. In contrast, women who found PKU to be a burden as a child made reference to the difficulties associated with PKU and the sense of feeling “different” to peers.

Adulthood Themes

Themes were identified regarding PKU as it related to adulthood and current circumstances (Table 3). As noted within childhood themes, both similarities and differences were apparent in their experiences.

Management of PKU

Self-reported management of PKU formed a continuum from complete non-compliance to vigilant dietary control. Participant 1 was off diet, did not take the supplement, nor had blood tests to measure Phe levels. Participants 2, 3, and 5 were strictly on diet, took the supplement, and had regular blood tests. Participant 4 was also on diet and took the supplement but did not undertake blood tests. Participants 6, 7, and 8’s management was situated in the middle of the spectrum, all being effectively on diet and consuming the supplement, but having a somewhat relaxed approach towards dietary adherence. Participant 6 deviated from the diet occasionally and Participants 7 and 8 did not measure their Phe intake. As such, some women had elevated Phe levels.

The Unique Experience of Having PKU

Women referred to several specific aspects of having PKU as adults.

Eating in a Social Context

Food in social contexts was problematic for some women. Similar to during childhood, Participants 1, 7, and 8 were conscious of differences, still find this upsetting. Eating out socially generally continued to be perceived as difficult due to the lack of suitable meals available. Participant 8 avoided going out to dinner altogether as it “. . .was just too hard. . .” In contrast, Participant 3 did not report eating out as an adult problematic as she was able to adapt sufficiently in social eating situations.

Effects of Dietary Compliance

Several women mentioned the negative effects of being off diet, or having high Phe levels. Women reported tiredness, inattentiveness, poor memory, mood swings, and anxiety. In cases where a psychological issue was present, symptoms were reported to be exacerbated. Maintenance of Phe levels within, or close to, the recommended range was associated with improved psychological and physical wellbeing. Women reported enhanced concentration, improved coping ability, more stable mood, decreased anxiety, and higher

energy levels. These benefits of dietary compliance were a motivation for adherence.

Transition from Childhood to Adulthood Management of the Condition

Some women reported difficulty in adjusting to full responsibility for management of their condition as an adult. Contributing factors included timely access to adult services and the lack of development of appropriate knowledge and skills to achieve independent management. Participant 8, for example, noted that she had to acquire several skills required for dietary compliance in adulthood as she was not very involved in PKU management as a child. Participant 2 reported cessation of dietary compliance for a year following delays in access to adult services.

Attitudes and Perceptions

Overall, the women's attitudes towards, and perceptions of, their condition varied along a continuum. Women had either developed acceptance of, or spoke negatively about PKU. Acceptance of PKU and the requirements for its management was related to expression of relatively positive views about the condition. Women who expressed such views accepted PKU as a part of their life and experienced minimal distress related to PKU as they had adapted their lifestyle to accommodate the condition from an early age. In contrast, those with less positive attitudes towards PKU highlighted the difficulties associated with management of the condition. Additionally, poor psychological wellbeing appeared to heighten negative attitudes. One woman with depression reported, "I just really can't be bothered with my life. I just, there's no point anymore, so what's the point with the diet?"

Psychological Wellbeing

Significant differences in psychological wellbeing were noted between the women. Three women reported significant historical mental health concerns with one also experiencing current symptoms. The women who had

experienced mental health concerns all perceived PKU as contributing to, or exacerbating, their mental health condition. In reference to her depressive episodes, one woman said "It's [PKU] got everything to do with it", with another questioning whether "...PKU people are fragile or predisposed to it [depression]..." In addition, a woman with bulimia nervosa suggested that the food restriction required in PKU contributed to its onset. Hence, PKU was perceived as a risk factor for poor psychological wellbeing. One woman also stressed the negative effect of poor psychological wellbeing on dietary compliance and the overall PKU experience, highlighting the importance of positive psychological wellbeing for successful dietary management.

Connection Between Childhood Experiences and Adult Outcomes

Connections were noted between the women's childhood experiences and their outcomes as adults, including management of the condition, experiences of PKU, and their psychological wellbeing. Positive adult outcomes, such as acceptance of PKU, continued appropriate management of PKU, positive psychological wellbeing, perception of minimal difficulty eating in social contexts, and ease of transition to independent management of the condition may be related to childhood experiences such as high levels of parental and extended family support, parental support to develop skills and knowledge to manage PKU, increased feelings of inclusion, and minimal perception of PKU as a burden. For example, Participant 1 who reported poor mental health and who was not on diet discussed feeling socially excluded as a child, and reported less support than many other participants from her parents and extended family, as well as perceiving PKU as a burden. This contrasts with Participant 5 who reported good mental health, was on diet (strict) and who described extensive parental and extended family support as a child, who did not report feeling socially excluded as a child and who did not describe perceiving PKU as a burden. Table 4 highlights adulthood outcomes and potential related factors in childhood.

Table 4 Adulthood outcomes and associated childhood experiences

Adulthood outcomes	Associated childhood experiences
Eating out socially	Feelings of difference to peers
Transition to full responsibility of management	Management of PKU during adolescence
Attitude towards PKU	Parental support
Psychological wellbeing	Extended family support
Management of PKU	Perception of PKU as a burden

Discussion

This study explored childhood experiences of women with PKU as they relate to adulthood management of PKU, experiences of the condition, and psychological wellbeing. Consistent with previous literature, feelings of difference to peers due to eating behavior (Zwiesele et al. 2015; Vegni et al. 2009), increased difficulty with dietary compliance during adolescence (Weglage et al. 1992; MacLeod and Ney 2010), parental support (Shulman et al. 1991), and extended family support (MacDonald et al. 2010) were identified as key childhood experiences for women with PKU, shaping the perception of PKU during childhood. Consistent with Kestilä et al.'s (2005) and Mikkilä et al.'s (2005) findings regarding the long-term impact of early experiences, connections were noted between these childhood themes and adulthood outcomes. Adulthood outcomes ranged along a continuum dependent upon childhood experiences. Current findings suggest that the identified childhood themes collectively shape the overall experience of PKU as a child, in turn, having a combined effect on adulthood management of PKU, experiences of the condition, and psychological wellbeing.

More specifically, the experience of minimal emotional distress regarding differences with peers due to eating behavior, minimal difficulty with dietary compliance as an adolescent, and high levels of parental and extended family support together may reduce the perceived burden of PKU as a child. These factors appear to build resilience in the individual, facilitating adult outcomes characterized by effective management of PKU, minimal perception of difficulties eating in social contexts, reduced difficulty in transition to self-management, an attitude of acceptance towards PKU, and positive psychological wellbeing. This is consistent with Finkelson et al. (2001) who reported that social support, positive attitudes towards the PKU diet, and perceptions that the diet is manageable in one's daily life predicted whether adults with PKU successfully returned to being on diet.

The enduring significance of childhood experiences for individuals with PKU makes sense given the well-documented impact of early life experiences on long-term physical and psychological wellbeing amongst the general population (Kestilä et al. 2005). Distress during childhood over eating differences could lead in adulthood to deviation from the PKU diet to avoid feeling excluded, as well as negative views of the condition and poor psychological wellbeing. Difficulty with dietary compliance as an adolescent may result in relaxed or total non-adherence as an adult, with reestablishment of the diet being difficult (MacLeod and Ney 2010). Accordingly, women either off diet or with lenient dietary compliance in this study reported increased difficulty adhering during adolescence.

Parental support is important in modeling management of PKU and establishing the child's dietary patterns, potentially assisting adjustment to adulthood in terms of attitude, diet, and psychological wellbeing. Consistent with Bosch et al.'s (2007) notion that children who are overly dependent on their parents in regard to PKU management may be susceptible to difficulties in adjustment to full responsibility in adulthood, the current study revealed that parental encouragement of growing self-management is important for the ease of transition. A sense of "teamwork" provided by supportive extended family members during childhood may also influence how accepting the individual is of PKU as an adult. Accordingly, women who reported high levels of extended family support during childhood were less overwhelmed by the demands of PKU in adulthood.

The self-report, qualitative nature of this study has revealed the perspective of women with PKU regarding their childhood and adult experiences, extending the current literature, much of which has previously explored the perspective of parents when consideration is made of childhood. Although appropriate for a study of this type, the sample was limited by the number of women with PKU in South Australia who were interested in participating. Participants were open to and had sufficient time to discuss their condition and experiences whereas those who were not as willing to discuss such matters and those who had more time pressures were likely underrepresented. It is also possible that those who chose not to participate were more likely to be those who perceived fewer psychological impacts of having PKU; future work should address this. However, the authors were satisfied that saturation had been reached for the key themes described in this paper. Given these findings reflect women's experiences only, future research with both genders would provide a broader perspective and validation of the themes identified here. Further, the results provide an illustration of the experiences of women treated within one hospital. Given that treatment within other health facilities may vary, multi-site research would be useful. Finally, future work that compares the experiences of adults with PKU with adults with other conditions, particularly those that require dietary management, may be useful.

The findings of this study have significant clinical implications. Given the difficult circumstances adults with PKU can experience regardless of their childhood experiences (Ris et al. 1997; Pietz et al. 1997; Koch et al. 2002; Gassió et al. 2003), intervention as a child can prove beneficial over the life span. In conjunction with diet and Phe level monitoring, treatment strategies should also encompass a psychosocial approach. Encouragement of parental support, specifically the delivery of advice regarding best care for children with PKU (as also described by Zwiesele et al. 2015), involvement of extended family

members, and provision of PKU knowledge to school peers should be included. Additional support services for adolescents with PKU may also assist in preventing or reducing potential issues with dietary adherence as a teenager, such as hands-on meal preparation “fun” days.

Conclusion

Adult outcomes for women with PKU may be influenced by certain childhood experiences relating to the condition, in particular the feeling of difference in comparison to peers, increased difficulty with dietary compliance during adolescence, parental support, extended family support, and the overall perceived burden of PKU. Results suggest that the unique experience of these may have a combined effect on adulthood management of PKU, experiences of the condition, and psychological wellbeing. Regardless of childhood experiences, lifelong management of PKU can prove onerous for some individuals and adults with PKU are at increased risk of mental health difficulties, despite good quality of life. Adoption of a broader approach to clinical management of PKU during childhood may facilitate positive adult outcomes.

Synopsis

Childhood experiences of women with PKU include feeling different to peers, challenges during adolescence, parental and extended family support, and the perception of PKU as a burden; these have a combined effect on adulthood management of the condition and psychological wellbeing.

Compliance with Ethics Guidelines

Conflict of Interest

R. Roberts, T. Muller, A. Sweeney, D. Bratkovic, A. Gannoni, and B. Morante declare that they have no conflict of interest.

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki.

Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

All authors contributed to the planning and reporting of the work described in the article. T Muller conducted the interviews and the thematic analysis.

Interview Questions

Can you tell me about the impact that PKU has had on you throughout your life?

As you were growing up, what were your plans about having children, knowing that you had PKU?

Have your plans changed now you are an adult?

What was your understanding about how pregnancy might be different for women with PKU?

Have you had any pregnancies, how many, were they planned or unplanned?

What did (would) you consider when deciding to become pregnant (if it was a planned pregnancy)?

Questions for Women Who Have Had a Pregnancy

I'd like to hear about your experience(s) of pregnancy with PKU.

Can you tell me about what you found particularly difficult during your pregnancy?

What did you find was helpful in coping with these difficulties?

Did you find that you used different ways of coping at different times in your pregnancy (for example, 1st trimester vs 3rd trimester)?

Can you tell me about what you found particularly positive during your pregnancy?

In general, how did you feel during your pregnancy?

How did you feel during the difficult times of your pregnancy?

Can you tell me about the supports you felt were available to you during your pregnancy, for example, people in your life, medical staff, or groups you were involved in?

Which supports did you find particularly helpful during your pregnancy and why was this?

Can you tell me about some supports that you would have liked to have had during your pregnancy?

Questions for Women Who Have Had Multiple Pregnancies

Can you tell me about the differences you noticed between your pregnancies?

How do you think your previous pregnancies affected your later pregnancies experiences?

Can you tell me about any knowledge that you feel that you would've found helpful during your first pregnancy?

Questions for Women Who Have Not Had a Pregnancy, but Are Planning to in the Future

I'd like to hear about the impact you believe PKU will have on your pregnancy.

Can you tell me about the difficulties you believe you may face?

Can you tell me about what you believe may be helpful in coping with these difficulties?

What supports do you believe may be helpful during your pregnancy, for example, people in your life, medical staff, or groups?

What supports would you like to have available to you during your pregnancy?

Questions for Women Who Have Not Had a Pregnancy, and Are Not Planning to in the Future

Can you share with me about the reasons why you have chosen not to become pregnant?

What impact did PKU have on this decision?

Can you tell me about the difficulties you experienced in making this decision?

Can you tell me about what you found helpful in coping with these difficulties?

Can you tell me about the supports you felt were available to you when you were making this decision, for example, people in your life, medical staff, or groups you were involved in?

Which supports did you find particularly helpful during this time and why was this?

Can you tell me about some supports that you would have liked to have had during this time?

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