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# Promoting psychological well-being in women with phenylketonuria: Pregnancy-related stresses, coping strategies and supports



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## ABSTRACT

**Objective:** To explore the pregnancy-related stresses anticipated and experienced by women with phenylketonuria (PKU) and the coping strategies and supports utilised or anticipated to be beneficial during pregnancy.

**Methods:** Thematic analysis of interview data from eight women with PKU in a cross-sectional, qualitative study. Five of the participants had never had a pregnancy but were planning to in the future, two participants had children, and one participant was pregnant.

**Results:** The central concern regarding pregnancy was achieving and maintaining the essential low Phe levels, in the context of the devastating effects of high levels. The Transactional Model of Stress and Coping was utilised to understand the coping strategies and supports utilised or anticipated to be beneficial during pregnancy. Similarities and differences between the women who had experienced pregnancy, and those who were planning a pregnancy in the future were evident in key coping strategies, with knowledge seeking, positive reappraisal, and reassurance seeking reported. Support from health professionals and other mothers with PKU was key for all women. Psychological support was identified as a

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resource perceived to be beneficial to promote psychological well-being during pregnancy but not yet provided.

**Conclusion:** Pregnancy is associated with significant stresses for women with PKU. Clinical implications of the findings include provision of psychological support.

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## 1. Introduction

Significant planning is required for pregnancies for women with phenylketonuria (PKU) as elevated levels of blood phenylalanine (Phe) during pregnancy causes negative birth outcomes such as pregnancy losses, congenital heart disease, and other developmental problems such as intellectual disability [18,25]. These adverse outcomes are common, for example Lenke and Levy [14] reported that in women with Phe levels >20 mg per decilitre, 73% had microcephaly and 92% of children had intellectual disabilities. To prevent such outcomes and allow normal development of the foetus, pregnancies must be planned and a strict low-phenylalanine diet must be maintained prior to and throughout pregnancy [8,21]. Phe levels of under 240  $\mu\text{mol/l}$  are considered to be safe in pregnancy [13] and at the time the women in the current study were pregnant, clinical practice in South Australia was to aim for Phe levels prior to and during pregnancy of 75 to 150  $\mu\text{mol/l}$ . This compares with safe Phe levels of 120 to 360  $\mu\text{mol/l}$  for women with PKU who are not pregnant [24], and Phe levels of between 20 and 70  $\mu\text{mol/l}$  in women without PKU during pregnancy (e.g., [17]).

The strict low Phe diet required during pregnancy is a vegetarian diet, with large quantities of low protein foods developed specifically for people with PKU, and large quantities of a medical formula supplementing the vitamins and minerals that the body cannot synthesise [21]. As this formula has a strong, bile-like odour, a taste that many find unpleasant, and a viscous consistency, women often find this difficult to consume during pregnancy, particularly when affected by pregnancy-related illness, such as morning sickness [2,9]. During pregnancy, Phe requirements increase, and complications such as hyperemesis make obtaining control more difficult.

Extensive monitoring is required throughout pregnancy, with weekly medical appointments and twice weekly blood tests to ensure that Phe levels are in the safe range for pregnancy [21]. As the required Phe levels during pregnancy to prevent negative birth outcomes are significantly lower than usual acceptable levels [21], significant diet and lifestyle changes are necessary to achieve and maintain the restricted Phe levels [2]. As such it would be expected that pregnancy would be a particularly stressful period for women with PKU.

There has been some consideration of the experiences and needs of women with PKU prior to and during pregnancy. For example, Waisbren et al. [25] reported a study of young women who were neither pregnant nor planning a pregnancy, finding social support for birth control and positive attitudes towards birth control to best predict birth control use. They also noted that most women in the study saw having children as a “natural sequel to finding a husband” (p. 303) with few acknowledging that an unplanned pregnancy may occur. These findings raised questions regarding the best way to support women with PKU as the first generation of women with treated PKU were reaching child bearing age.

An evaluation of a programme designed to provide support to women with PKU during their pregnancy found some promising effects for a peer support programme [21]. Women with a child with PKU were trained to provide social support and enhance positive attitudes in women with PKU who were pregnant or planning pregnancy. However, there were no differences in metabolic control or pregnancy outcomes between the women who participated in the programme and those who did not, suggesting that further work is needed to understand the issues faced, and the best way to provide support.

To date, research has not yet focussed on the experience of women with PKU during pregnancy or the factors that impact on this, such as stresses, coping strategies, and available supports. The concerns of women with PKU when they are considering a pregnancy and the strategies and supports that they believe will assist them to cope during pregnancy will also be considered in the current study; information which will provide valuable information to guide clinical practice.

## 2. Methods

### 2.1. Participants

The participants in this study were eight women (mean age 29.9 years, SD 7.9 years, range 21–42 years) with PKU living in South Australia. All but one of the participants ( $n = 7$ ) were currently maintaining the PKU diet ('on diet') and were taking a supplement (medical formula). Six of these women were having regular blood tests with the frequency of tests ranging from weekly to once every four to six weeks. These women were all early-treated (treated for PKU from soon after birth), and all had tested IQs in the normal range (not intellectually impaired).

Five of the participants had never had a pregnancy but were planning to in the future, two participants had children and were not planning any further pregnancies, and one participant was pregnant (third trimester). The women with children had 3 and 5 children respectively, all of whom had normal intellectual and health outcomes. Two participants had experienced pregnancy complications, including miscarriages, which were unrelated to the management of PKU. Two participants had a history of major depressive disorder, with one of these participants also experiencing recurrent bulimia nervosa. Another participant had a history of bipolar affective disorder, with a current depressive episode. As such, 37.5% of the participants in this study reported experiencing significant psychological disorders in their lifetime. For a summary of further participant characteristics (relationship status, education background, current study, and employment status) see [Table 1](#).

### 2.2. Interview procedure and materials

The study was approved by the Human Research Ethics Committee of the Children, Youth, and Women's Health Service. Participants were recruited through the Women's and Children's Hospital with all women with PKU living in South Australia invited to participate ( $n = 20$ ). Eight women expressed interest in participating in the study (40% response rate). Interviews took place in a location convenient to the participant and were conducted by TM.

Demographic information was collected verbally at the beginning of the interview. The M.I.N.I. (English Version 6.0.0) was then administered. The M.I.N.I. is a brief, verbally administered questionnaire assessing lifetime experience of psychological disorders [22]. The M.I.N.I. follows the diagnostic criteria for the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR) and the International Classification of Diseases (ICD-10) for 15 major categories of Axis 1 psychiatric disorders, suicidality and anti-social personality disorder (this section was not utilised within this study). Administration time ranges from

**Table 1**  
Participant characteristics.

Demographic variable	n
Relationship status	
Married	3
De facto <sup>a</sup>	2
Single	3
Employment	
Full-time	4
Part-time	2
Casual	1
Highest level of education	
Year 11	1
Year 12	3
Vocational qualification	2
Bachelor degree	2
Current study	
Full-time university level study	1
Part-time university level study	1
Full-time vocational study	1

<sup>a</sup> In a couple, who although not legally married to each other, live together on a genuine domestic basis as husband and wife.

15 min to approximately 30 min depending upon the quantity of symptoms endorsed by the participant. This diagnostic tool has received extensive reliability and validity testing and is widely utilised in both research and clinical settings [12,23]. This structured measure was utilised to gain insight into participants' experience of both diagnosed and undiagnosed psychological disorders.

The semi-structured interview then followed. The interview was designed based on the relevant literature by the research team to ensure the study aims were achieved. This interview schedule was utilised as a base to generate discussion with probing to gain further detail occurring throughout. The initial questions of the interviews regarded the impact of PKU on the participant's life from childhood. The following areas were then explored, dependent on the participant's circumstances:

- For women with children: their experiences during pregnancy, including difficulties they encountered, positive aspects of their pregnancies, coping strategies and supports utilised, and other factors, such as supports, that they would have liked to have received during pregnancy.
- For women who had not yet had a pregnancy: their perception of pregnancy, including concerns, factors and supports that they believe would assist them to cope during pregnancy.

The length of the complete interviews ranged from 30 to 90 min. All interviews were audio taped and transcribed verbatim. Transcriptions were analysed qualitatively utilising the guidelines for thematic analysis as described by Braun and Clarke [1].

### 3. Results

Several themes emerged related to pregnancy-related stresses and coping. The issues reported by women with and without children generally matched closely so are presented and discussed together. Comment is made on areas where there were differences between the women who had experienced pregnancy, and those who were planning a pregnancy in the future. Illustrative quotes for each theme are presented in Tables 2 and 3.

#### 3.1. Anticipated and experienced stresses

Women reported anticipating and experiencing a number of key pregnancy-related stresses, related to the unique experience of managing PKU during pregnancy. These were 1) achieving and maintaining the low Phe levels required prior to, and throughout, pregnancy, 2) the time consuming nature of managing PKU throughout pregnancy, and 3) the impact on their social interaction.

##### 3.1.1. Achieving and maintaining levels prior to and throughout pregnancy

The core concern for the women was achieving and maintaining the low Phe levels required prior to, and throughout, pregnancy. Women who had not yet had a pregnancy tended to express this concern

**Table 2**

Anticipated and experienced stresses during pregnancy: illustrative quotes.

- 
- |  |   |
|--|---|
| 1) Achieving and maintaining the low Phe levels required prior to, and throughout, pregnancy | <p>"I particularly worry about is just trying to stick to the diet, that such strict diet and just not knowing what I can actually eat and if I'll be satisfied and I won't binge and stuff like that."</p> <p>"My concern was I wouldn't be able to maintain low enough levels to have a normal baby; that was always in the back of my head."</p> <p>"Well, ultimately the, you know, if anything were to go wrong to effect the, the baby's development or in-utero or after, I suppose, that you'd be directly responsible for it. And that would be really horrible. You'd feel kind of guilty for the rest of your life."</p> <p>"(I was) very sick (with morning sickness) and I would drink my medicine and then I'd spew it all up... And I was panicking. The mental stress that I had gone through, just the fact that I'd drunk all this medicine, a litre, and I just vomited it all out."</p> |
| 2) The time consuming nature of managing PKU throughout pregnancy                            | <p>"It's a lot of time, it's very time consuming, because you've gotta do everything right. And measure exactly this much food."</p>  |
| 3) The impact on women's social interaction  | <p>"The going out stuff is hard, you just have to not go or tend to eat like biscuity things and stuff like that."</p> <p>"We'll go to the football and I'll take my scales with me so I can weigh some chips so I can work out how much they are, so and you'd be sitting there weighing everything and people'll be looking at you and think, oh God. Sitting in the football stand weighing your food before you eat it."</p>  |
-

through discussion about their ability to maintain the strict diet to, in turn, maintain levels. These concerns were supported by the mothers, with all describing this as the core worry and source of stress throughout their pregnancies. A number of women reported that they were so concerned about this that they had once believed that they would never have children for this reason. For most women, their concerns about maintaining levels were described in the context of their concerns about the impact of elevated Phe levels on their baby's development.

The mothers emphasised that there are many factors during pregnancy that lead to an increase in Phe levels separate from diet management, and that the experience of these factors lead to increased stress during pregnancy. Such factors include pregnancy-related illness (such as morning sickness), being unwell (such as a cold), and weight loss. Two of the mothers had struggled with weight loss and were hospitalised due to this during one pregnancy each. One of these mothers reported that, whilst experiencing the weight loss was very stressful, admission to hospital was a positive experience as it allowed her the time, space, and support to focus on her pregnancy. These two mothers also reported experiencing pregnancy-related illness and that this had also lead to difficulties in their ability to consume and keep down the supplement and maintain levels. Only one woman who had not had a pregnancy commented that she was concerned about the impact of pregnancy-related illness on managing PKU during pregnancy. This indicates that this issue is one that is not generally of concern in the consideration of pregnancy; however it is of great concern during pregnancy.

### 3.1.2. Time consuming nature of managing PKU through pregnancy

Issues regarding the time consuming nature of managing PKU throughout pregnancy were raised mainly by mothers, with only one woman who hadn't had a pregnancy raising concerns about this. As such, this does not appear to be of central concern in the consideration of pregnancy; however an issue that becomes apparent during preparation for a pregnancy.

The combination of weekly dietician or medical visits, twice weekly blood tests, and constant measuring and recording of food appeared to be stressful for all mothers. Other specific areas of concern differed depending on the women's life circumstances at the time. Women with other children found issues such as cooking for themselves and the family, and getting blood tests in on time, difficult. Women

**Table 3**

Anticipated and experienced coping during pregnancy: illustrative quotes.

#### 1) Access to supports

"But the only thing that keeps you positive is the fact that you have that continual contact with the Women's and Children's [hospital]."

"... it was really, it's truly one on one when I was pregnant with all of my kids with [dietician]. As to every single thing I was putting in my mouth."

"I'd say support of my family and my husband basically. That's what's got me through as well as the medical (support)."

"During the pregnancy to have someone who's been there and done it, or is going through it with you, or something like that, if you're lucky enough to have that."

"I'd probably need a little more than that [general hospital and dietician support]. Like psychological help or something."

#### 2) Problem-focussed coping skills including developing relevant skills and seeking knowledge

"I'd need the practice and make it regular and make it so that every week or fortnight your blood levels are below whatever, 200 or 150. You know, etc, etc, to make sure that I can do this for a sustained amount of time."

"If they just did an information session where everything was all laid out to you on the table and you could raise any concerns you might have then, before you actually go ahead, I think would be beneficial prior to. Maybe not for everyone, but at least have that option there."

"... shall I eat beforehand? Shall I take the chance that there'll be stuff there for me to eat? And, or shall I take snacks as well? So it's a matter of adjusting your lifestyle and taking, being prepared I suppose."

#### 3) Emotion focussed coping skills including positive reappraisal and reassurance seeking

"But I mean it's a sacrifice you make for a short time."

"... just count down."

(focus on) "the big picture" (the birth of a healthy baby).

"There might be times when you think, this is difficult, so difficult. I don't know how I'm going to do this for 9 months. But then, once you compose yourself, get yourself together, get everything under control and then think, I'm doing this for a reason, there's going to be something beautiful at the end of it, and umm... it gets you there."

"... or if I've got any concerns, if I'm feeling unwell, I'll just ring them (hospital staff) and say, hey I'm not feeling the best... So they're really good, they're really on the ball and quite supportive with everything and always ask how you're feeling and... So that reassurance sort of keeps you at ease with everything..."

working full-time found weekly appointments to be difficult, whilst women living in rural areas were concerned about how their pregnancy might be managed from afar.

### 3.1.3. *The social impact*

The impact of managing PKU during pregnancy on the women's social life was only raised by mothers. This again appears to be an issue which is not of central concern in the consideration of pregnancy, but has a significant impact during the pregnancy.

Mothers reported that they found it difficult to go out to social gatherings which involved food as they generally could not eat the food served. The mothers reported that they had to take their scales to weigh their food anywhere that they may eat and have their supplement with them.

## 3.2. *Anticipated and experienced coping*

A number of different ways were identified through which the anticipated and experienced stress and worry associated with managing PKU during pregnancy could be managed, including 1) access to supports, 2) problem-focussed coping skills including developing relevant skills and seeking knowledge, and 3) emotion focussed coping skills including positive reappraisal and reassurance seeking.

### 3.2.1. *Supports*

The importance of a number of different supports during pregnancy was emphasised by both mothers and those who had not had a pregnancy. The general support from the hospital, through aspects such as regular appointments, frequent blood tests, and advice over the phone, was key for the majority of the women. A number of women, both mothers and women who had not had a pregnancy, identified the importance of the support of the dietician specifically. The support of their partner and family was also central to coping during pregnancy.

The majority of the women who had not had a pregnancy and one mother reported that they would like the support of other mothers with PKU during their pregnancy. A mother identified that such support is not currently offered as an option through the hospital and the majority of women reported that they knew few other women with PKU to seek such support from. The women emphasised the significance of gaining support and advice from someone who had coped through similar experiences as theirs. The women made suggestions such as having support sessions with another mother with PKU, being able to contact another mother over the phone when required, or having a support line that they could utilise.

A number of women who had not yet had a pregnancy commented that they felt that access to psychological services would assist in supporting them through their pregnancy. Whilst no mothers reported having received psychological assistance during their pregnancy, one mother identified that she felt that the stress and worry were overwhelming during one of her pregnancies. One woman who had not yet had a pregnancy suggested that a brief psychological intervention early in the pregnancy as a preventative measure may be beneficial, followed by access to psychology on an 'as needs' basis throughout the remainder of the pregnancy.

### 3.2.2. *Problem-focussed coping strategies*

Problem-focussed coping refers to action taken either within the environment (for example, preparing financially) or within oneself (for example, learning new skills) to alter the stressful situation [10,11]. Several problem-focussed coping strategies were perceived as either helpful or potentially helpful for coping during pregnancy.

The majority of women who had not had children emphasised the importance of the preparation time prior to pregnancy to cope with the pregnancy. They reported that, within this period, they would acquire the necessary skills (such as measuring food and counting protein), knowledge (such as appropriate foods), and confidence to be able to manage and cope with the pregnancy. Many women also reported that obtaining knowledge would be helpful in coping with a pregnancy. Some women, including a mother, reported that they would like, or would have liked, knowledge and an understanding about PKU and pregnancy (including the process, diet, and necessary skills) in their late teenage years or early twenties even though they were not yet considering pregnancy at that time. These same women and others reported that obtaining a comprehensive understanding of the management of PKU during pregnancy shortly prior to the beginning of the preparation period would be beneficial to coping.

Mothers described adjustments that they made to their lives to cope with changes during pregnancy, such as eating before going out, and taking scales, snacks and supplement when going out. Adapting to the circumstances meant that these mothers were still able to attend and be involved in social functions.

### 3.2.3. *Emotion-focussed coping strategies*

Emotion-focussed coping strategies are utilised to alter the emotion attached to a situation rather than changing any aspect of the situation itself. This is achieved by either reappraising the problem to change the relational meaning of the situation (for example, emphasising the positive of the situation) or altering the way in which the situation is attended to (for example, avoidance) [4,10]. Whilst mothers reported utilising emotion-focussed strategies such as positive re-appraisal and reassurance seeking during their pregnancies, these were not discussed by women who had not had a pregnancy.

Mothers described coping with stresses by reminding themselves that this was a short-term difficulty with the aim of supporting the birth of a healthy baby. Mothers also emphasised the importance of frequent reassurance from medical professionals to cope. They described obtaining this reassurance through the regular appointments and regular blood test results, "... me hearing my Guthrie results was a way of letting me know my baby's ok" and through phone calls to the hospital when stressed or unsure about something.

## 4. Discussion

This qualitative study explored the key pregnancy-related stresses anticipated or experienced by women with PKU, and the strategies and supports they either utilised or anticipated to be beneficial to coping during pregnancy. Whilst much of the literature in this area does not have a consistent theoretical focus, a number of studies focussing on women with high-risk pregnancy have utilised the theoretical model, the Transactional Model of Stress and Coping (see [6,15]). This model provides a viable framework through which the relationship between high-risk pregnancy and maternal psychological distress, and factors that influence this relationship, can be understood. This model suggests that coping with a high-risk pregnancy is a complex interaction of cognitive appraisals, coping strategies and coping resources [4,11]. Particular patterns of thinking and responding, plus access to resources such as social support, can reduce the impact of high-risk pregnancy on psychological well-being [6,15].

Appraisal of the situation as within their control is associated with increased psychological well-being for women with a high-risk pregnancy, whilst appraisal of threat and uncertainty is associated with increased psychological distress ([6,15,19,27]). Positive reappraisal is an effective emotion-focussed coping strategy for women with a high-risk pregnancy, whilst avoidance is associated with increased psychological distress ([6,26,27]). The efficacy of problem-focussed coping strategies, such as preparing for motherhood, is unclear, potentially due to the uncertain nature of high-risk pregnancy [15,19,26]. Whilst general social support is beneficial for women with high-risk pregnancy, support from medical professionals is of particular importance for women with a high-risk pregnancy due to a chronic condition [15,19].

The Transactional Model of Stress and Coping [11] was used to guide understanding of the factors utilised or anticipated to be utilised by the women in the current study during pregnancy to cope, or anticipated to be beneficial to coping, during pregnancy. As expected, key stresses were specific to the experience of pregnancy for women with PKU. Coping strategies and supports utilised or anticipated as beneficial were generally similar to that within the literature in the area of high-risk pregnancy and coping [6]. However, some supports not yet considered within the high-risk pregnancy literature were identified. As literature in the area of PKU has not yet considered the issues explored here, this study makes a novel contribution to understanding pregnancy-related stresses and coping for women with PKU.

This study revealed that the core concern for both mothers and women who had not yet had a pregnancy is achieving and maintaining the low Phe levels required prior to and during pregnancy. This finding is consistent with research identifying that women with PKU find attaining and maintaining metabolic control during pregnancy to be challenging [9,21]. Considering the significant negative impact of elevated Phe levels on the developing foetus, it is unsurprising that this is the core stress regarding pregnancy for these women. The issues of the time consuming nature of managing PKU during pregnancy and the social impact were raised almost exclusively by the mothers in the sample. This indicates that these are issues that are not pertinent when women are considering pregnancy; however have a significant impact during pregnancy. These findings are



consistent with research which indicates that practical issues, such as the significant diet and lifestyle changes required, are barriers to dietary control for women with PKU during pregnancy [2].

Awareness and understanding of the pregnancy-related stresses anticipated and experienced by women with PKU are of importance for clinicians involved in the care of these women. Awareness allows clinicians to be sensitive to such stresses, educate women regarding potential stresses, and to normalise such worries and stresses.

A number of coping strategies and supports were identified. Women who had not yet had a pregnancy tended to focus upon supports and problem-focussed coping strategies, such as knowledge seeking and skill building. Whilst mothers also discussed such strategies and supports, they also expressed utilising emotion-focussed strategies, such as positive reappraisal and reassurance seeking. This is consistent with Lazarus [10] who argues that, as coping is a process, problem-focussed strategies are often utilised early in the stressful situation, with an increase in the use of emotion-focussed strategies over time. Emotion-focussed coping strategies may be utilised in the midst of a stressful period, but may not be considered as a potential coping strategy in the preparation for such a time.

Knowledge seeking was an important problem-focussed coping strategy identified by all women. The importance of knowledge about pregnancy, including factors such as the process, diet, and expectations, as a young woman was emphasised, in addition to having a full, comprehensive understanding prior to preparation for pregnancy. An optional information session prior to preparation for pregnancy was suggested, within which comprehensive information about pregnancy and expectations is provided. Research has shown that education regarding the effects of PKU on pregnancy is important in preventing unplanned pregnancies and improving metabolic control prior to and during pregnancy [9,25]. However, the findings of this study suggest that comprehensive information regarding pregnancy, including the process and skills required may assist women in coping prior to and during pregnancy. The development and implementation of pregnancy-specific information sessions, and potentially take-home information packs, for young women and potential new mothers is one way in which clinicians could further assist women with PKU in coping prior to, and during, pregnancy.

The importance of the support of health professionals prior to and during pregnancy was emphasised by all women. Frequent appointments, advice and blood tests were key to coping with a pregnancy for many of the women, with an emphasis on the support of the dietician. Levy-Shiff et al. [15] found that support from medical professionals was particularly important for women with a high-risk pregnancy due to a chronic condition, as is PKU. Mothers described utilising the emotion-focussed strategy of seeking reassurance from medical professionals when they were feeling stressed and anxious. This finding is consistent with O'Brien et al. [19] who emphasised the importance of seeking reassurance from medical professionals for women with high-risk pregnancy in managing stress and anxiety. Awareness of the importance of their support and reassurance can allow medical professionals to ensure that sensitive and responsive care is provided throughout the pregnancy.

The women in this study emphasised the importance of receiving support from another mother with PKU to assist them in coping during pregnancy, yet indicated that they did not know other women to seek such support from informally. Rohr et al. [21] found that the support of another mother with PKU assisted women to attain metabolic control for pregnancy much earlier. Together, this provides evidence that such support is beneficial to women with PKU. Such support could be provided informally by willing mothers with PKU, either through meeting or over the phone, or through a more formal means, such as the development of a national support line. Telephone support is currently utilised for patients with other medical conditions and services facilitating this support indicate that the provision of support in this manner has proved invaluable for patients [20]. Additionally, given the accessibility of online social media in recent times, development of formal points of contact through social networking outlets may also be one way in which contact between women with PKU could be facilitated. Research has indicated that such use of social networking has led to an increase in social support for patients and increased quality of life, whilst the inclusion of medical professionals in the facilitation of such groups may also increase the positive benefits achieved [7]. As such, it is suggested that the development of support communities, including medical professionals, through online social networking be explored for women with PKU, in addition to the provision of telephone support.

Support from partners was also central to coping for women. Services should consider how best to include and support the women's partners in care during pregnancy.

It is likely that some of the stressors experienced by women during pregnancy may differ depending on whether it is a first pregnancy, or whether women already have children. Women may learn from their previous experiences, and may cope better in subsequent pregnancies. On the other hand, there may be additional stressors when women need to also care for children during pregnancy.

Increased exposure to stressors during pregnancy increases the risk of the development of psychological disorders during high-risk pregnancies [15,16]. In the current study, women expressed concerns about psychological well-being and coping during pregnancy. In addition, a significant proportion of the women in this sample had experienced significant psychological disorders within their lifetime, consistent with research indicating that this population is at higher risk of developing psychiatric symptoms [3,5]. As such, psychological support may assist in alleviating stress and anxiety during pregnancy and reduce the risk of the development of psychological disorders. The provision of a brief psychological intervention in the preparation for the pregnancy and in the early stages, focussing on strengthening coping skills and psycho-education regarding common psychological disorders such as depression and anxiety, may be beneficial as a preventative measure and assist in the promotion of psychological well-being. It is recommended that this then be followed by access to psychological support as required for the remainder of the pregnancy.

A limitation of this study is the self-selected nature of the sample. The women who participated were open to discussing their condition, pregnancy and psychological well-being and, likely, those who were not as willing to discuss such matters were underrepresented. Additionally, the majority of women within this study were maintaining the PKU diet. Research suggests that, whilst it is now recommended that the diet is maintained throughout the lifespan, many women cease the diet in adulthood [9,21]. Extensive research evidences the emotional and cognitive effects of cessation of the diet in adulthood, with increased rates of internalising symptoms such as depressive and anxious symptoms [3,5]. As such, the experiences of this sample may not be reflective of the population.

The retrospective, self-report nature of the study is a further limitation. Due to the small number of women with PKU in South Australia, only one woman was pregnant at the time of the study and all other mothers retrospectively reported their pregnancy experiences. As stress and coping changes across the pregnancy, assessment at multiple periods during pregnancy would also provide a deeper understanding of women with PKU's experiences. In addition, five women were discussing their perceptions of what a pregnancy would be like for them, but had never been pregnant. It is likely that their perceptions would be different once they had experienced a pregnancy. A further limitation of this study is that women with PKU treated within one centre alone are considered. Treatment through other centres may result in different experiences. As such, research collaboration across centres should be considered.

## 5. Conclusions

The findings of this study provide some critical insight into the pregnancy-related stresses anticipated and experienced by women with PKU and the coping strategies and supports they utilise or believe are beneficial during pregnancy. Promotion of maternal well-being is key to achieving positive outcomes for both the mother and child. Awareness of the unique pregnancy-related stresses for women with PKU assists health professionals to provide sensitive and responsive care. Provision of comprehensive information is essential in supporting women in their decision to have a child. Developing pathways for contact between pregnant women and mothers with PKU may assist the promotion of psychological well-being during pregnancy, as may provision of psychological support.

## Conflicts of interest

None.

## References

- [1] V. Braun, V. Clarke, Using thematic analysis in psychology, *Qual. Res. Psychol.* 3 (2006) 77–101, <http://dx.doi.org/10.1191/1478088706qp0630a>.
- [2] A.S. Brown, P.M. Fernhoff, S.E. Waisbren, D.M. Frazier, R. Singh, F. Rohr, S.A. Rasmussen, Barriers to successful dietary control among pregnant women with phenylketonuria, *Genet. Med.* 4 (2002) 84–89, <http://dx.doi.org/10.1097/00125817-200203000-00006>.

- [3] V.L. Brumm, D. Bilder, S.E. Waisbren, Psychiatric symptoms and disorders in phenylketonuria, *Mol. Genet. Metab.* 99 (2010) S59–S63, <http://dx.doi.org/10.1016/j.ymgme.2009.10.182>.
- [4] S. Folkman, R.S. Lazarus, If it changes it must be a process: study of emotion and coping during three stages of a college examination, *J. Pers. Soc. Psychol.* 48 (1985) 150–170, <http://dx.doi.org/10.1037/0022-3514.48.1.150>.
- [5] J.K. Gentile, A.E. Ten Hoedt, A.M. Bosch, Psychosocial aspects of PKU: hidden disabilities – a review, *Mol. Genet. Metab.* 99 (2010) S64–S67, <http://dx.doi.org/10.1016/j.ymgme.2009.10.183>.
- [6] C. Giurgescu, S. Penckofer, M.C. Maurer, F.B. Bryant, Impact of uncertainty, social support, and prenatal coping on the psychological well-being of high-risk pregnant women, *Nurs. Res.* 55 (2006) 356–365, <http://dx.doi.org/10.1097/00006199-200609000-00008>.
- [7] S. Idriss, J. Kvedar, A. Watson, The role of online support communities benefits of expanded social networks to patients with psoriasis, *Arch. Dermatol.* 145 (2009) 46–51, <http://dx.doi.org/10.1001/archdermatol.2008.529>.
- [8] R. Koch, E. Friedman, C. Azen, W. Hanley, H. Levy, R. Matalon, F. De la Cruz, The international collaborative study of maternal phenylketonuria status report 1998, *Ment. Retard. Dev. Disabil. Res. Rev.* 5 (1999) 117–121, [http://dx.doi.org/10.1002/\(SICI\)1098-2779\(1999\)5:2<117::AID-MRDD4-3.0.CO;2-W](http://dx.doi.org/10.1002/(SICI)1098-2779(1999)5:2<117::AID-MRDD4-3.0.CO;2-W).
- [9] R. Koch, F. Trefz, S. Waisbren, Psychosocial issues and outcomes in maternal PKU, *Mol. Genet. Metab.* 99 (2010) S68–S74, <http://dx.doi.org/10.1016/j.ymgme.2009.10.014>.
- [10] R.S. Lazarus, Coping theory and research: past, present and future, *Psychosom. Med.* 55 (1993) 234–247 (doi: 10.1.1.115.9665).
- [11] R.S. Lazarus, S. Folkman, *Stress, Appraisal, and Coping*, Springer, New York, 1984.
- [12] Y. Lecrubier, D.V. Sheehan, E. Weiller, P. Amorim, I. Bonora, K. Harnett Sheehan, G.C. Dunbar, The Mini International Neuropsychiatric Interview (M.I.N.I.). A short diagnostic structured interview: reliability and validity according to the CIDI, *Eur. Psychiatry* 12 (1997) 224–231, [http://dx.doi.org/10.1016/S0924-9338\(97\)83296-8](http://dx.doi.org/10.1016/S0924-9338(97)83296-8).
- [13] P.J. Lee, D. Ridout, J.H. Walter, F. Cockburn, Maternal phenylketonuria: report from the United Kingdom Registry 1978–97, *Mol. Genet. Metab.* 71 (2005) 233–239.
- [14] R.R. Lenke, H.L. Levy, Maternal phenylketonuria and hyperphenylalaninemia: an international survey of the outcome of untreated and treated pregnancies, *N. Engl. J. Med.* 303 (1980) 1202–1208.
- [15] R. Levy-Shiff, M. Lerman, D. Har-Evan, M. Hod, Maternal adjustment and infant outcome in medically defined high-risk pregnancy, *Dev. Psychol.* 38 (2002) 93–103, <http://dx.doi.org/10.1037/0012-1649.38.1.93>.
- [16] M. Lobel, Pregnancy and mental health, in: H. Friedman (Ed.), *Encyclopedia of Mental Health*, Academic Press, San Diego, 1998, pp. 229–238.
- [17] K. Matalon, P.B. Acosta, L. Castiglioni, et al., Protocol for Nutrition Support of Maternal PKU, The National Institute of Child Health and Human Development, Bethesda, MD, 1998. 1–51.
- [18] T.W. Ng, A. Rae, H. Wright, D. Gurry, J. Wray, Maternal phenylketonuria in Western Australia: pregnancy outcomes and developmental outcomes in offspring, *J. Paediatr. Child Health* 39 (2003) 358–363, <http://dx.doi.org/10.1046/j.1440-1754.2003.00174.x>.
- [19] E.T. O'Brien, S. Quenby, T. Lavender, Women's views of high-risk pregnancy under threat of preterm birth, *Sex. Reprod. Healthc.* 1 (2010) 79–84, <http://dx.doi.org/10.1016/j.srhc.2010.05.001>.
- [20] Redkite, Community Based Support, Retrieved June 23, 2012, from <http://www.redkite.org.au/community-based-support> 2009.
- [21] F. Rohr, A. Munier, D. Sullivan, I. Bailey, M. Gennaccaro, H. Levy, S. Waisbren, The resource mothers study of maternal phenylketonuria: preliminary findings, *J. Inherit. Metab. Disord.* 27 (2004) 145–155, <http://dx.doi.org/10.1023/B:BOLL.0000028785.20901.d9>.
- [22] B.V. Sheehan, Y. Lecrubier, K. Harnett Sheehan, P. Amorim, J. Janvas, E. Weiller, G.C. Dunbar, The Mini International Neuropsychiatric Interview (M.I.N.I.): the development and validation of structured diagnostic psychiatric interview for DSM-IV and ICD-10, *J. Clin. Psychiatry* 59 (1998) 22–33.
- [23] B.V. Sheehan, Y. Lecrubier, K. Harnett Sheehan, J. Janvas, E. Weiller, A. Keskiner, G.C. Dunbar, The validity of the Mini International Neuropsychiatric Interview (M.I.N.I.) according to the SCID-P and its reliability, *Eur. Psychiatry* 12 (1997) 232–241, [http://dx.doi.org/10.1016/S0924-9338\(97\)83297-X](http://dx.doi.org/10.1016/S0924-9338(97)83297-X).
- [24] J. Vockley, H.C. Andersson, K.M. Antshel, N.E. Braverman, B.K. Burton, D.M. Frazier, S.A. Berry, Phenylalanine hydroxylase deficiency: diagnosis and management guideline, *Genet. Med.* (2014), <http://dx.doi.org/10.1038/gim.2013.157> (Advance online publication 2 January 2014).
- [25] S.E. Waisbren, S. Shiloh, P. St. James, H.L. Levy, Psychosocial factors in maternal phenylketonuria: prevention of unplanned pregnancies, *Am. J. Public Health* 81 (1991) 299–304, <http://dx.doi.org/10.2105/AJPH.81.3.299>.
- [26] A.M. Yali, M. Lobel, Coping and distress in pregnancy: an investigation of medically high risk women, *J. Psychosom. Obstet. Gynaecol.* 20 (1999) 39–52, <http://dx.doi.org/10.1037/a0013242>.
- [27] M. Lobel, A.M. Yali, W. Zhu, C.J. DeVincent, B.A. Meyer, The impact of optimistic disposition on emotional distress during high-risk pregnancy, *Psychol. Health* 17 (1) (2002) 77–95.