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

Rachel M. Roberts [a,*], Tamara Muller [a], Annabel Sweeney [b], Drago Bratkovic [c], Anne Gannoni [d]

[a] School of Psychology, University of Adelaide, Adelaide, Australia
[b] Department of Nutrition, Women's and Children's Hospital, Adelaide, Australia
[c] Metabolic Unit, Women's and Children's Hospital, Adelaide, Australia
[d] Department of Psychological Medicine, Women's and Children's Hospital, Adelaide, Australia

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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

* Corresponding author at: School of Psychology, University of Adelaide, North Tce, Adelaide, SA 5005, Australia.
E-mail addresses: rachel.roberts@adelaide.edu.au (R.M. Roberts), tamara.muller2@gmail.com (T. Muller), Annabel.Sweeney@health.sa.gov.au (A. Sweeney), Drago.Bratkovic@health.sa.gov.au (D. Bratkovic), Anne.Gannoni@health.sa.gov.au (A. Gannoni).

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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.

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 μmol/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 μmol/l. This compares with safe Phe levels of 120 to 360 μmol/l for women with PKU who are not pregnant [24], and Phe levels of between 20 and 70 μmol/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.