



Phenylketonuria—the lived experience

Nicole Frank, Ruth Fitzgerald, Michael Legge

Abstract

Aim This study explored the lived experience of phenylketonuria (PKU) for the New Zealand adult and its relevance for issues of treatment adherence.

Method In-depth qualitative interviews were conducted with eight New Zealand adults with early-treated PKU regarding their experiences of living with PKU. The interviews were transcribed, and then analysed using grounded theory. A review of relevant medical, scientific and social science literature placed this analysis in a broader context.

Results A number of consistent themes emerged as characteristic of the participants' life experiences, including a chronic uncertainty existing on several levels, the challenges posed by the maintenance of interpersonal relationships with respect to the PKU diet, and a basic incompatibility between the PKU diet and many lifestyle demands. Social science commentary on the topics of risk management, stigma, and other types of "dieting" experiences further elucidates these themes.

Conclusion Based on the findings of this research, medical practitioners may be able to better tailor their services for, and interactions with, the adult PKU community, for example, by facilitating self-management, conveying realistic expectations of metabolic control, and increasing the volume of information directed to PKU adults.

Inherited metabolic diseases are a clinically diverse group of medical conditions which require specialist diagnosis and care. Although individually they are rare, they have a collective incidence estimated to be 1 in 1500 persons, which would indicate that many general practitioners would encounter an inherited metabolic disease in their practice at some stage.^{1,2}

Despite the poor prognosis of many of these disorders—which may result in intellectual handicap, deformities, and premature death—some may be treated or have intervention therapy to circumvent the pathological sequelae of the disease. One such inherited metabolic disease is phenylketonuria (PKU) and related forms of hyperphenylalanaemia, an autosomal recessive disorder first described in 1934. This group of inherited disorders of phenylalanine metabolism is primarily due to either a deficiency of the enzyme phenylalanine hydrolase in the classical form, or mutations in the enzyme in the variant forms.

With an incidence of 1 in 15,000 births in New Zealand, it is likely that there will be approximately 4 infants per year born with this disease (based on 60,000 births per year). Screening for this and other metabolic disorders at birth, ensures early detection and intervention under the guidance of a metabolic physician.

For PKU, the intervention therapy appears comparatively simple (although, as the results of the research will demonstrate, in practice, the treatment can be quite complicated and demanding). A restrictive diet low in phenylalanine and routine

monitoring of blood phenylalanine levels ensures that the amino acid does not increase to the point of neurotoxicity causing intellectual handicap.

Maintenance of this diet up to adolescence has been a success story in preventing the complications of this disease, and until recently, it was thought that dietary treatment of PKU could be discontinued once the individual reached adolescence with no adverse consequences. However, that assumption has since come into question.

Recent research shows the possibility of “alarming problems in cognition and social functioning” for adults who discontinue the PKU diet.³ Although there is no international consensus on the issue, many medical practitioners today, including the specialised medical team providing tertiary care for the PKU population in New Zealand, recommend life-long adherence to the PKU diet to all PKU patients.

Despite this recommendation, studies almost unanimously report low rates of treatment adherence in both paediatric and adult PKU populations,⁴⁻⁸ and adherence appears to decrease with age.⁸⁻¹¹

In this research we investigate the social impact of the restrictive diets for people living with PKU and how people living with PKU assess the constraints and consequences of dietary adherence in their own private lives outside of the medical consultation. In conducting this research we explored the lived experience of PKU for the New Zealand adult and its relevance for issues of treatment adherence in the light of contemporary medical, scientific, and social science literature around the subject.

Method

The research was conducted in two parts. First, the PKU-related literature was investigated, with articles being located through the databases Medline, Science Direct, Proquest, Factiva, InfoTrack, Nutrition and Food Sciences, and the *JAMA* website. In addition, relevant social science literature was also reviewed, including literature on the social significance of food and eating, dieting, stigma, and risk, with the last category added to the review after having been raised repeatedly by the participants during interviews.

By combining the medical and scientific literature with the social science literature, a broader understanding of individuals' accounts of living with PKU could be constructed. This multidisciplinary approach is in line with the NHC (2005) proposal for improving clinical support for people with chronic illness.¹²

The second part of this research involved qualitative research using data from interviews with eight New Zealand adults living with PKU. The goal of the interviews was to elicit data about the psychosocial aspects of the decision-making relating to choosing to adhere to or avoid the PKU diet.

The open-ended interviews lasted between 50 and 90 minutes, and covered the following topics: participant understandings of PKU, impressions of normalcy and difference in regard to PKU, identity formation, challenges of PKU and treatment adherence, coping mechanisms, change of experience over time, relationships with medical personnel, familial and social impact of PKU, emotional aspects of the experience of living with PKU, risk perception and how that relates to treatment adherence, personal agency and control, and the sensual aspects of the PKU diet.

Participants were located through a national database available to the National Metabolic Services team in Auckland. All PKU adults listed in this database (n=46) were contacted by letter inviting them to join the research project, and eight responded (the low response rate being typical of third party recruitment). However, this was a sufficient number of participants for data analysis using grounded theory,¹³ which focused on the meanings and experiences of living with PKU.

Six of those who responded were female; two were male. The ages of the participants at the time of the interview ranged from 31 to 43 years. Although this group presented a high degree of variation in areas such as marital status, socioeconomic standing, and life experiences (e.g. occupation, travel, pregnancy, and child-rearing), it should be noted that their collective perspective could vary in important ways

from those adults with PKU who did not respond to the project invitation. For example, although the older range of New Zealand adults was well represented (including one individual who was reportedly diagnosed just before routine screening began in New Zealand), the views of younger adults (in their 20s) were not represented.

The interviews were conducted by the first author (NF), transcribed verbatim, returned to the participants for checking, and then analysed by the first author (NF), with a second researcher (RF) checking the transcripts and thematic analysis for consistency of interpretation. The biomedical science information in both the interview analysis and the literature review was verified by the third author (ML).

Qualitative software was not considered necessary for this project. Ethical approval was obtained from the New Zealand Multi-Region Ethics Committee.

Results

Overview—The themes which emerged from the interviews are set out in Table 1, and their detailed analysis forms the basis of a more extensive publication¹⁴ and an unpublished Master's thesis.¹⁵ However, a number of the open coded categories were particularly relevant to patient-practitioner interactions. These were collectively identified in the axial coding as “uncertainty”, “difference” and “incompatible lifestyle demands” (shown by asterisk), and can be identified at a higher level of abstraction as forming part of the Medical, Social, and Personal Spheres of life for the interviewees.

Although a number of other themes arose within each of these spheres, for the purpose of this article, we will focus on only one theme per sphere (i.e. the theme which impacts the medical consultation most explicitly). The selected theme from each sphere of living will be discussed in tandem with the results obtained from the literature survey.

Medical Sphere: evaluating uncertainty—All interviewees raised the topic of uncertainty (functioning at the level of both future and present) in various contexts, and found this to be anxiety-provoking. Awareness of an uncertain future pervades the experience of living with PKU for this project's interviewees. Participants stressed the newness of their situation, and the fact that they are the first generation of early-treated adults with PKU, frequently noting that no-one (including the medical community) “knows what the future holds” for people with PKU.

Uncertainty was expressed in relation to how long they would live and how long they would retain their mental faculties. Phrases such as “uncharted territory” and “no-man's-land” were used to describe their situation, conveying a deeply uncertain future. Present uncertainty that people with PKU must confront is connected to the lack of medical certitude concerning PKU, and stresses ambiguity pertaining to the optimal mode of treatment. For although the New Zealand medical community appears to be consistently recommending a life-long PKU diet, the impression remains among participants that the necessity or benefit of this is still ultimately uncertain.

Participants also expressed uncertainty around the value of the ideal blood phenylalanine level, raising concerns that they received conflicting information from year to year. An additional area of uncertainty was identified relating to how to achieve the desirable levels, and the lack of apparent correlation with self-perceived behaviour.

Table 1. Overview of grounded theory coding analysis

Open Coding	Axial Coding	Selective Coding	Core Category
Extent of medical attention/intervention Experimentation (“guinea pig”) Appreciation of medical attention/intervention Dependence upon medical attention/intervention Medicalization/objectification Medical expectation (moral onus)	Medical Scrutiny	Medical Sphere Evaluation of medical information and expertise	NEGOTIATOR
Anxiety about future health and ability Inconsistent medical views on best treatment Uncertainty concerning current blood levels Unsure of present effects of condition Hypothetical present (“what if?”) Risk	Uncertainty*		
Medical expertise Usefulness of internet Information from friends, family, society Self-expertise (embodied knowledge)	Multiple Sources of Information		
Distinct food behaviour Stigma Exclusion Accommodation by others (or lack thereof) Camaraderie between individuals with PKU Normality Prevalence of alternative diets in society Others’ lack of understanding Pressure/temptation to conform Reactions of others Avoidance of potentially awkward situations	Difference*	Social Sphere Management of information, impressions and interpersonal relationships (to elicit positive social responses)	
Self-denial Limitation of culinary experience Restricted quantities of food Lack of choice, freedom, and spontaneity Inability to “treat” oneself Lack of variety Sensual aspects of food (unsatisfactory) Challenges to life pursuits (e.g. having children, travel)	Deprivation	Personal Sphere Strategic incorporation of the PKU diet into personal lives	
Labour-intensive diet Distaste for cooking PKU-related time investments Time limitations Importance of routine for dietary adherence Lack of routine in everyday life	Incompatible Lifestyle Demands*		

The asterisk indicates coded categories discussed in this article and movement from left to right across the table indicates increasingly abstracted levels of analysis. The core finding is that people living with PKU who were interviewed for this project consider the primary effect of living with their condition to be that they become “expert negotiators” in the *medical*, *social*, and *personal* spheres of their lives.

Uncertainty cannot be acted upon until it has been evaluated, and risk assessment plays a large role in this process of evaluation. Many of the observations made in the social science literature regarding this stress that both the recognition of, and the value given to, risks are subjective and contextually dependent,¹⁶ leading to varying perspectives on risk. Often, the perspective of a medical professional differs from that of a lay-person.

Petersen and Lupton observe that “the medical practitioner will tend to interpret data on risk through her or his own emotionally charged experience in working with individuals, having responsibility for their care and treatment, and being in the position of seeing patients die or avert death.”¹⁷ For these reasons, Peterson and Lupton argue that doctors will tend to view risks as more severe or negative than will patients, and will expect a higher level of adherence to recommended treatment to avoid the risk.

Lay people, however, may draw from a number of sources in addition to medical opinion to formulate their evaluations of risk, including their own experiences, the experiences of friends, the familiarity or abstractness of a particular risk, or the perception of a particular risk relative to other perceived risks— leading to a discrepancy between the risk assessments of the two parties.

Two participants gave especially good examples of the assessment of risk based on the information at hand. Interestingly, both individuals were off-diet at the time of the interview. The first participant very clearly assessed risk associated with PKU in relative terms, comparing it with other forms of risk to which she perceived herself to be susceptible. Because she saw her weight and her family history of other illnesses as additional (possibly more pressing) health risks, future harm from being off-diet was for her, “just another possibility”.

She therefore concluded:

...particularly with my other bits and pieces that I've got thrown into the mix as well...I think...that controlling blood levels is not necessarily going to make a big difference (female, age 35)

The second example of risk assessment highlights the abstract nature of risk communication. This participant explained that after reviewing some recent research demonstrating the benefit of returning to the PKU diet (for those who have been off-diet)

...it seems so far away...Like if 1 of the 200 people [*with PKU*] in New Zealand would say to me, 'Well, that happened to me,' I would probably take it more seriously...It just seems like another world (female, age 38).

Social Sphere: managing social difference—Although “difference” was experienced by interviewees to varying degrees, it was reported by all project participants. The most evident outward expression of difference was considered to be in relation to their eating habits—either when they are unable to eat what those around them are consuming, or when they consume something (such as the protein substitute) unfamiliar to others. Both instances serve to create a distinction between the individual with PKU and others. In many cases, this difference led to a lack of social acceptance, exclusion, and stigma (especially in childhood).

The interviewees in general held a deep appreciation for the social nature of food consumption, a topic that the social science literature elaborates upon in great detail.

Indeed, sharing food can be a potent symbol of community and relationship, friendship, trust, and intimacy.

Alternatively, the refusal of food can send equally strong messages indicating the refusal of such intimacy—effectively communicating “enmity and hostility”.¹⁸ Individuals with PKU wishing to maintain dietary control must continually reject food that is not permitted in their strict dietary regimen, and participants experienced this to be a very awkward social situation—desiring to maintain their diet, yet not wishing to inadvertently offend. The expectations involved with the sharing of food (and the resulting temptation and persuasion initiated by others) was reportedly one of the primary difficulties encountered in dietary adherence, and one of the most common reasons for transgression of the PKU diet.

Goffman’s work on stigma provides some interesting insights into the social nature of difference and the corresponding management of interpersonal relationships. He speaks of both information management and impression management in this regard. Goffman uses the concept of controlling or managing information in the context of difference to refer to the questions of “to display or not to display; to tell or not to tell; to let on or not to let on; to lie or not to lie; and in each case, to whom, how, when, and where.”¹⁹ These questions are certainly a matter of consideration and sometimes concern for adults with PKU, as the interviews demonstrated. Of perhaps even greater concern for many participants is Goffman’s concept of impression management.

Many examples of “impression management” were provided by the interviews, such as one participant’s method of always explaining her condition in social circumstances so as to not appear “rude” by her lack of participation in food-related events, or another participant’s offhanded, trivializing attitude toward her condition, which she found caused others to likewise regard her condition as minor and unproblematic.

In listening to participants explain their methods of managing their interpersonal relationships in the face of social difference, it is notable that emphasis is routinely placed on the feelings of others rather than the individual with PKU (as is common with experiences of stigmatisation¹⁹). This has important implications for treatment adherence. Participants consistently explained that they were most likely to transgress their PKU diet when the comfort and feelings of other people were at stake.

For instance, when asked in which situations she would find herself most likely to abandon (or make exceptions to) the diet, one participant replied:

...if I’ve been invited to someone’s place and they’re maybe someone who doesn’t know me very well, or knows me but they’re really kind of uptight about being a really good hostess, or...they’re not going to cope with me saying, “Well I can eat this, but I can’t eat that and that,” and they’d feel really bad about it...It’s usually more about the other people than me (female, age 35)

This leads to an interesting finding. Whereas some medical literature stresses the importance of a strong network of social support in adherence to the PKU diet,^{3,5,20,21} and adjustment to chronic medical conditions in general,²² participants in this project alternatively spoke of others primarily as a *challenge* to dietary adherence, and consistently reported a very high degree of self-motivation, self-discipline, and independence in their maintenance of the PKU diet.

Personal Sphere: negotiating incompatible lifestyle demands—In common with members of the general population, individuals with PKU have strong feelings concerning the lifestyles they desire and for which they feel suited.

For instance, the strong disinterest in cooking and baking expressed by interviewees represents one grave conflict between desired lifestyle and the substantial time required to prepare special PKU foods (such as baking with low-protein flour).

In addition, as society shifts towards increasing consumption of convenience foods, individuals with PKU often feel left behind, attempting to juggle the modern expectations of jobs and families, without the aid of quick meal-time options. Several other challenging lifestyle incompatibilities were also mentioned, including adherence to the PKU diet while travelling (especially for extended periods of time and in unfamiliar locations).

A review of the social science discourse around dieting reveals that this “unrealistic” nature of the strict PKU diet (in its ideal form) represents one of many similarities between people’s experiences of the PKU diet and people’s experiences on other types of diets.

Both the PKU diet and other types of diets (e.g. diets for weight-loss) are externally defined, and as such, may not take into considerations the demands of “real life”. Commenting on women’s attempts to loose weight through dieting (but equally applicable, it seems, to the situation of people on a PKU diet), Bordo argues that “...total control [*over food*] is ultimately unsustainable” and that “ ‘the diet’ is itself a precarious, unstable...state...”²³

Similarly, Orbach notes that the “ideal” presented to women on diets for the purposes of weight-loss produces “a picture that is far removed from the reality of women’s day-to-day lives”.²⁴ The parallels here are self-evident and indicative of further insight to be gained through such a comparison.

The participants in this project negotiate these competing lifestyle demands partly through strategic dietary flexibility, delineating between the “exceptions” that would cause guilt and those considered permissible. For instance, exceptions made for the benefit of others were often justified, as in the example provided in the previous section. Many also appeared to view the occasional exception as harmless, but a string of exceptions as ultimately poor adherence.

As one participant explained...

...it’s not too bad if I just do a bit here and there, but if [*one thing leads to another and*] it all ends up bad choices, then I feel really bad” (female, age 43).

Another participant mentioned that intentional indiscretion would not cause guilt...

...if I made a conscious thing that...I was going to eat something and was just going to enjoy it, then no way [*would I feel guilty*]” (female, age 35)

Additionally, many participants indicated that “special occasions” such as birthdays, special meals out, or weddings, may constitute a justification for making guiltless exceptions to the PKU diet.

Routine also proved to be a strategic element aiding in adherence to the PKU regimen for many. Due to the limitations that the diet imposes on the individual, once a particular system is found to suffice, participants often hold to that system with little alteration. It is when participants are caught up in a healthy routine that they find treatment adherence to be most feasible. This particular strategy, however, is limited in its usefulness by the unpredictability of life and the impossibility of maintaining a routine at all times.

Ultimately, every aspect of PKU treatment (from the timing of blood tests to the invention of palatable, convenient ways to prepare the medical foods) requires a great deal of organisation and planning.

Participants expressed that one of the most difficult consequences of PKU is the need to think constantly, and ceaselessly, about diet. This results in what one participant referred to as a considerable “mental burden”.

Discussion

The difficulty of managing the highly restrictive low phenylalanine diet for PKU has significant ongoing effects for the patients in all aspects of their lives and serves as a reminder that patients juggle more than their diagnosis in living with a chronic illness.

Medical consultations have become simply one more arena in which to form an independent opinion of the relevance or otherwise of the advice being offered. On the other hand, the clinician has the opportunity to offer significant support through the provision of compassionate guidance to these patients in negotiating a life in which the future is perceived to be so uncertain.

Expert clinical support is also eagerly sourced and highly valued by women with PKU during their pregnancies when the maintenance of recommended levels of phenylalanine is difficult to achieve. (The recommended PKU diet during pregnancy is stricter than at other times, permitting fewer daily exchanges of phenylalanine. Tight control of phenylalanine levels is vital during this time as high levels of phenylalanine can cause serious developmental problems in the foetus²⁰.)

Many of the suggestions made by the participants for improved care relate directly to the three aspects of living with PKU identified in this paper.

One frequent grievance raised by interviews concerned new developments in knowledge and treatment concerning PKU. This was perceived to be neither readily accessible nor conveniently distributed, and due to the overall uncertainty felt by PKU adults and their need to personally evaluate available sources of information in order to choose a course of action, this was problematic.

Healthcare professionals could reduce this uncertainty by increasing the volume of information directed to the adult PKU community, and ensuring that the means of delivery is more appropriate. (For instance, a number of participants mentioned that information online is either inconvenient if they do not have ready access to the internet, or untrustworthy, indicating that there is a need for alternative methods of information distribution.)

The independence of adults with PKU stemming from the nature of social interaction with non-PKU individuals also has interesting implications for healthcare professionals. The importance of teaching dietary independence from a young age—including the cooking skills necessary for dietary adherence—was raised, as not all participants were given such instruction during childhood.

Additionally, because their treatment is largely self-governed, interviewees stressed that the accessibility of their healthcare workers was important and highly valued. The complexities of contemporary social life should also be addressed within the clinical consultation.

To advise PKU adults to “bake” in a world of fast food chains, double income families, and renegotiated gender roles runs the risk of rendering the medical consultation invalid through its lack of relevance to contemporary lifestyles.

As we gain better knowledge of the molecular and biochemical implications of inherited metabolic diseases and design intervention strategies based on biochemical interactions, it is important to retain the perspective that treatments should not be based solely on the long-term clinical outcome but also on the life experiences of the individuals living with the disease.

PKU represents a classical “treatable” inherited metabolic disorder with a good clinical outcome; however, this research has demonstrated through exploring the lived experience of PKU, how professional support could be altered to better meet the needs of those with PKU and their families.

Competing interests: None.

Author information: Nicole Frank, Professional Research Assistant, Barbara Davis Center for Childhood Diabetes, University of Colorado Health Sciences Center, Denver, Colorado, USA; Ruth Fitzgerald, Senior Lecturer, Department of Anthropology, University of Otago, Dunedin; Michael Legge, Associate Professor, Departments of Biochemistry and Pathology; University of Otago, Dunedin

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Correspondence: Ruth Fitzgerald, Department of Anthropology, University of Otago, PO Box 56, Dunedin. Fax: (03) 479 9095; email: ruth.fitzgerald@stonebow.otago.ac.nz

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