The challenges of parenting a child with phenylketonuria (PKU): Parents’ lived experiences

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What is PKU?

- Phenylketonuria (PKU) is a rare inherited metabolic disorder which can cause neurological damage if left untreated.

- PKU is identified through newborn screening in most European/Western countries and it has a prevalence of 1 in 10,000 births.

- Parents find out about diagnosis when their child is asymptomatic.

- People with PKU cannot metabolise phenylalanine, found in protein-rich foods.

- Treatment= strict diet

- Life-long adherence to this treatment regime has been recommended (National Society for PKU 2014).
The challenges of parenting a child with PKU

- Parents have to supervise and monitor the nutritional intake of their child, attend regular appointments and submit regular blood tests.

- Parents report **significant emotional challenges** early on in their child’s life, despite an absence of ill health, including **grief and trauma reactions** alongside caring for their baby (e.g., Lord, Ungerer & Wastell, 2008)

- **Challenges** are ongoing as their child develops (e.g., Medford, Hare & Wittkowski, 2017) – weaning is one major challenge.
Parenting children with I.M.Ds

Parents report:

- Significant burden on the family
- Time constraints including:
  - Stress related to preparing a special diet
  - Keeping records of blood results
- Restricted social life
- Coping with the ‘threat’ of metabolic crisis/the unknown
- Difficulties in managing life transitions
Impact on parents

- Reports of parenting stress in parents of children with PKU have been mixed.

- Parents report lower or similar levels of stress and quality of life compared to parents of healthy control children and parents of children with other IMDs (Kazak, Reber & Snitzer, 1988; ten Hoedt et al, 2011).

- In our study (Medford et al. 2017), caregivers (59%) showed high levels of significant psychological distress on a questionnaire (GHQ-12). However, the results indicated that caregivers’ ability to adhere to treatment was not affected.
Parenting a Child with Phenylketonuria (PKU): an Interpretative Phenomenological Analysis (IPA) of the Experience of Parents

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Abstract
Phenylketonuria (PKU) is a rare inherited metabolic disorder which can cause neurological damage if left untreated. PKU is identified through newborn screening in developed countries, and treatment begins immediately to prevent these severe consequences. When a child is diagnosed, parents must assume immediate responsibility for the management of PKU and prevention of neurological damage. Quantitative studies have identified significant psychosocial stressors for parents, but little is known about how the parents experience this process. This study aimed to explore the experiences of parents of children with PKU under the age of two. It is the first study to examine these experiences in this way. Seven parents were interviewed about their experiences, and interpretative phenomenological analysis was used to analyse the data. Three main themes were identified: control, striving for normality and acceptance of PKU at a transactional level. Links between the themes and processes underpinning the results were explored with reference to existing literature and theories from a clinical psychology perspective. The role of acceptance of PKU was central to the parent’s experiences. Clinical implications and suggestions for further research are discussed.

Keywords Phenylketonuria · Parents · Experience · Interpretative phenomenological analysis · Qualitative

Introduction
Phenylketonuria (PKU) is an inherited metabolic disorder (IMD) with a prevalence of 1 in 10,000 births (National Society for Phenylketonuria 2014; Williams et al. 2008). People with PKU cannot metabolise phenylalanine (phe), an amino acid found in most protein sources. Phc accumulating in the brain and blood, resulting in permanent neurological damage and concomitant intellectual disability and epilepsy. This can be averted through a strict life-long regimen of a phe restricted diet and amino acid supplements (National Society for Phenylketonuria 2014; Al Hafid and Christodoulou 2015). Management of PKU places significant demands on parents with supervision of the child’s nutritional intake, ongoing medical appointments and regular blood tests. Parents of children with IMDs report significant burden (Gramer et al. 2014) and challenges include time constraints, stress and restrictions on social life (Bijlengelhaven et al. 2013) along with emotional, mental and interpersonal stress (Cederbaum et al. 2001; Packman et al. 2007). In particular, there are stressors relating to dietary provision, the threat of metabolic crisis, coping with uncertainty, managing life transitions and struggling for improvement in care (Khanura et al. 2016; Medford et al. 2017; Read 2003; Stoch et al. 2008; Zelmer et al. 2004).
Aims of our qualitative IPA study

- Little is known about the experiences of parents caring for a child with PKU in the first two years of the child’s life.

- The aims were to explore their *lived experiences*, the processes parents went through in adjusting to and coping with these challenges.

- Interpretative Phenomenological Analysis (IPA) (Larkin, Smith & Clifton, 2006; Smith, 1999; Smith & Osborne, 2008) allows researchers to explore *how people make sense of their experiences and the significance they attach to their experiences.*
Interview

Parents were interviewed in their own home using a semi-structured interview schedule:

1. Experience of the diagnostic process
2. Processes of parenting
3. Challenges and coping
4. Support
Participiants

- 18 packs were sent out, 1 person declined and 8 did not respond. Two parents consented but could not be contacted.

- **6 mothers and 1 father consented to participate.**

- All were white British, married and living with their partner and child(ren). They were financially stable and educated, with high levels of family support.

- All parents reported good treatment adherence.
Three superordinate (and 12 subordinate) themes were identified:

1. Control
2. Striving for normality
3. Acceptance of PKU (as a continuum)
Diagnosis

CONTROL

STRIVING FOR NORMALITY

ACCEPTANCE OF PKU (As a continuum)

Acceptance
Positive attitude toward PKU and management – PKU accepted as part of child. Child seen as normal. PKU seen as manageable.
Reduced parent stress
Allows flexible control of phe-levels.
Reduced pressure on parent to ‘make child normal’

Non acceptance
Negative attitude toward PKU and management – PKU seen as something that should be gotten rid of. Child seen as different, desire child to be normal.
Maintains anxious control of phe-levels. Maintains pressure to ‘make child normal’.
Increased parent stress.

Potential future impact on child’s adjustment and attitude towards PKU and management?
Theme 1: Control

• **Control** = 1) Fears of the consequences of non-compliance
   2) Increased parental responsibilities
   3) Emotional and societal consequences of control

• “Alice: Some days would be good because you’d think “It’s fine, it’s just a diet and blah de blah”… and then you have other days when you think, “Well what happens if she doesn’t stick to her diet… and what happens if we don’t…” and you know them things that go through your head… ….. you don’t want your child to have any kind of brain damage, do you?

• Despite their own anxieties, parents adjusted to the requirements of managing their child’s PKU treatment and created family routines to facilitate this.
Theme 2: Triving for normality

• Then parents tried to **minimise the impact of PKU on their child** and develop a ‘new normal’.

• **Normality:**
  1) A different, healthy child
  2) Fear of child feeling different
  3) Effortful creation of a normal life
  4) Achievement

• **Jemima:** To watch him eat something I’ve cooked is great….. cause it’s such a major thing I think… erm… yeah.. Because it’s all I’m ever thinking about… **trying to think of new things for him and**… Things he might enjoy… then I make something and he really likes it, and I think “Yeah, I’ve cracked it. I’ve done it. I’m doing a good job.”
Theme 3: Acceptance

1) Acceptance of diagnosis and management
2) Lack of knowledge, understanding and information
3) Support from others
4) Becoming an expert
5) Gratitude

- Parents may experience denial, anger and depression (grief) first.
- The dual model of grief (Stroebe & Schut, 2010) may apply: Parents are immediately forced into primarily ‘restorative’ coping and then oscillate between *restorative and loss-oriented coping*
- Past and present appraisals of threat or stress may influence parental acceptance of PKU.
- *Emma:* If I’m out and about and I’m in a restaurant and there’s something I don’t know, I’ll put a question on Facebook, and whoever’s out there, out of the thousand people, because they’re from America and everywhere, they’ll jump in and answer my question for me. So, I’ve got constant 24-hour-support.
Summary

- Parent and child psychological factors play a role in treatment adherence.

- Training of healthcare professionals to raise awareness of how differently parents cope and that parents may require further assistance with this (including psychological support).

- Offer of peer support and clear post-diagnosis pathways.

- Psychosocial or psychological interventions to promote dietary compliance may be particularly important for parent of older children or those teenagers, as metabolic control worsens with age.
Gratitude

- As part of the process of acceptance

- Jemima: Everything is so much more special when [child] does it… because of everything that was put against him you know… you can’t help but think, 60 years ago… how different my little boy would be… only 60 years ago.
Questions?

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Relevant references


• Research Gate- Anja Wittkowski or [https://www.research.manchester.ac.uk/portal/anja.wittkowski.html](https://www.research.manchester.ac.uk/portal/anja.wittkowski.html)