The PKU Caregiver Study

Julie Bolduc DeFilippo, MSW, LCSW
Assistant Professor or Practice/Director of MSW Field Education, Merrimack College

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Disclosures

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Learning Objectives for Today

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- Understand the PKU Caregiver Study, its objectives and outcomes
- Translate findings of the study to one's own clinical practice

Research Questions & Objectives

The PKU Caregiver Study was conducted in 2018

Research Question: What are the experiences of caregivers to people with PKU?

- The objectives of this study:
  1. Gain an understanding of the types of need experienced by caregivers of children with PKU
  2. To understand any barriers caretakers face
  3. To learn more about services provided to families.

Background of Study

- Interest in informal caregiving focused on older adults and children with disabilities
- Parent of a child with PKU

A Look At Available Research

Studies look at QoL in Europe, the Middle East and Australia

George & Gwyther (1986)	Higher caregiver burden can result in poor health outcomes, mental health disturbance and social isolation
Okwuruska et al. (2011)	Quality of Life (QOL): describe five domains related to QOL: physical status and functional abilities, psychological status and well-being, social interactions, economic and work status and religious status.
	QoL studies on caregivers to children with PKU have been conducted in Iran, the Netherlands, France, Germany, Italy, Spain, Turkey, UK, Denmark and Australia.

QoL Studies show mixed results

- Ten Hoedt et al. (2011) found that health-related QoL (HRQoL) appeared not to differ from HRQoL in parents of healthy children. Findings also showed that HRQoL was affected by emotional support, loss of friendship, and age of their child.
- Bosch et al. (2015) looked at QoL of parents to children with PKU in France, Germany, Italy, The Netherlands, Spain, Turkey and the UK and reported overall normal HRQoL for parents of children with PKU, except in the emotional wellbeing domain

- Iran – moderate to low QoL compared to general population and high levels of stress (Mahmoudi-Gharaei et al, 2011; Irannejad et al., 2018)

- In Australia, Morawska et al. (2019) found that lower HRQoL was related to guilt due to poor adherence to treatment, parenting stress due to child emotional and behavioral challenges and over reactivity in parenting.

Studies on the psychological impact of caring show concerning results

- Several studies looked at the psychological impact of caring for a child with PKU. These studies took place in Turkey, Italy and the UK.
  - All of these studies found that parents to children with PKU have clinically significant levels of depression (Gunduz, 2015; Akkus et al., 2020; Ionio, 2018; Vetrone et al.,1989; Medford et al., 2017).
  - Inconsistent results when looking at anxiety levels

Akkus' et al. (2020) - Turkey	Gunduz et al. (2015), Ionio et al. (2018), Medford et al. (2017),
no difference in anxiety levels between parents to children with PKU and parents in the general population.	found high levels of anxiety among parents to children with PKU.

PKU can result in high costs to families

- In the UK, MacDonald et al. (2016): 19 hours/week was spent by caregivers on PKU dietary management.
  - Out-of-Pocket costs were incurred from attendance at PKU events, PKU-related equipment and extra costs for food at the holidays.
  - 22% of caregivers were forced to reduce working hours;
  - 24% left their jobs to manage their care duties.

Acceptance of diagnosis is a continuum

- Carpenter et al. (2019) found that
  - Healthcare Professionals expect parents understand their child's treatment quickly
  - Parents often struggle with worries of their child fitting in
  - Parents describe their acceptance of PKU as a continuum

Social support is an important coping technique for children with PKU

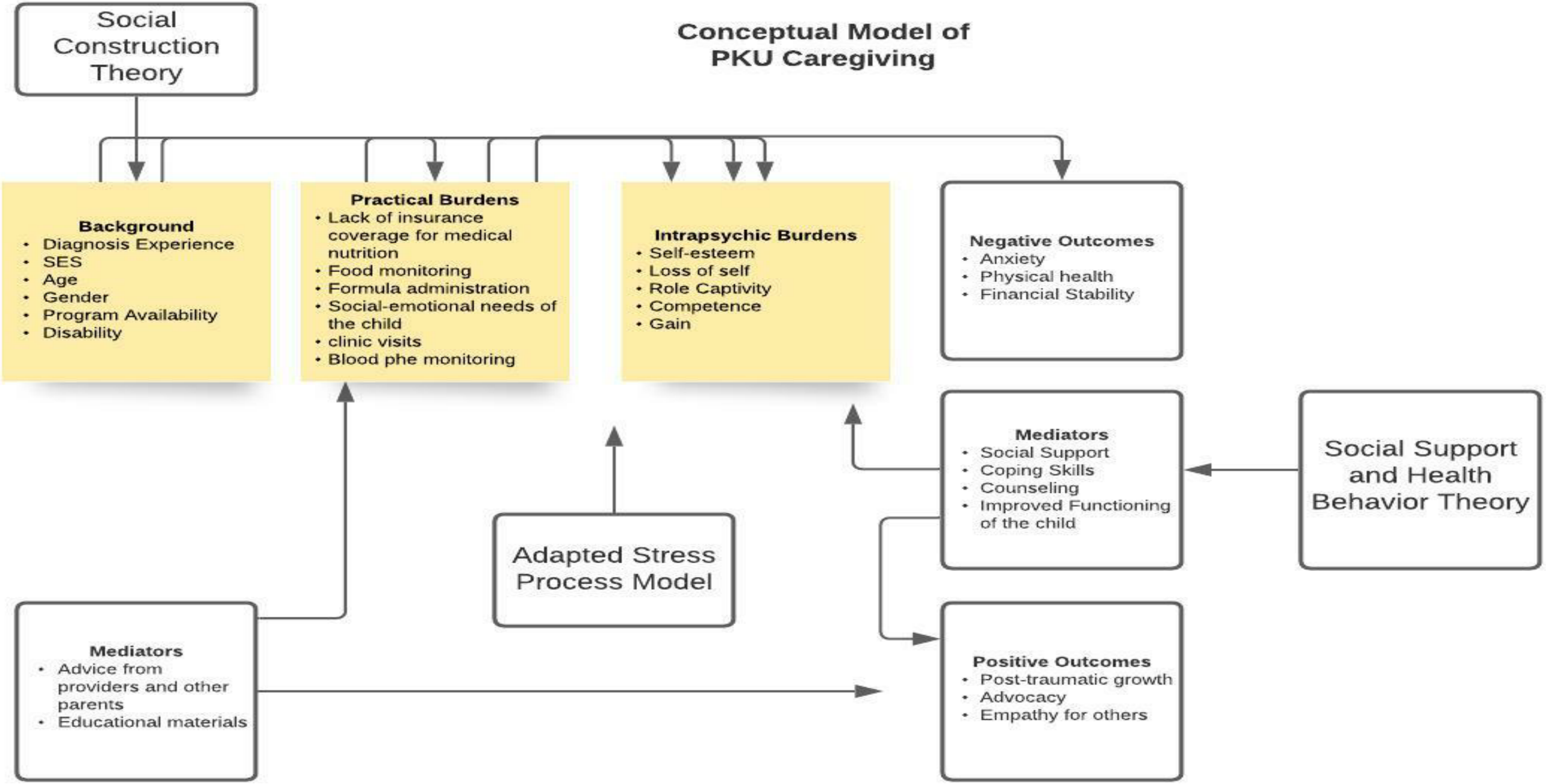
- In the U.S., Zwiesele et al. (2015) asked about the PKU child’s feelings of difference related to the disease.
  - Findings showed that involvement in the PKU community was one effective strategy to helping children cope with PKU.

Decision making varies with adherence to management

- Fehrenbach & Peterson (1989) looked at decision making of U.S. based caregivers in managing their children’s PKU.
  - Parents of children in good dietary control differed from parents whose children were in poor dietary control in their ability to integrate several behaviors into a solution.
  - Parents engaged in verbal and written problem-solving situations under conditions of both high and low time-pressure induced stress. Overall, compliant parents gave higher quality verbal and written problem-solving solutions than noncompliant parents.

The PKU Caregiver Study

Conceptual Model



Conceptual Model of PKU Caregiving

Social Construction Theory

Background
- Diagnosis Experience
- SES
- Age
- Gender
- Program Availability
- Disability

Practical Burdens
- Lack of insurance coverage for medical nutrition
- Food monitoring
- Formula administration
- Social-emotional needs of the child
- clinic visits
- Blood phe monitoring

Intrapsychic Burdens
- Self-esteem
- Loss of self
- Role Captivity
- Competence
- Gain

Negative Outcomes
- Anxiety
- Physical health
- Financial Stability

Mediators
- Social Support
- Coping Skills
- Counseling
- Improved Functioning of the child

Social Support and Health Behavior Theory

Adapted Stress Process Model

Mediators
- Advice from providers and other parents
- Educational materials

Positive Outcomes
- Post-traumatic growth
- Advocacy
- Empathy for others

Methods and Design

- A phenomenology approach in accordance with van Manen (1990) was used in this study.
- Phenomenology is an approach to qualitative research that describes the meaning of a lived experience of a phenomenon for several individuals, which in this case is the experience of caregivers to adults with PKU.
- The objective is to describe the commonalities between participants’ experiences (van Manen, 1990).

Sampling, Participants and Ethical Considerations

- Outreach on Facebook pages related to PKU and via the New England Connection for PKU and Allied Disorders listserve.
- n=8
- All mothers
- Resided in states across the country, including Massachusetts, Tennessee, California, Texas, Minnesota, Wisconsin, Oregon and Kansas
- IRB approval was obtained from Simmons University Institutional Review Board
- Informed consent was obtained from all participants

Data Collection and Analysis

- Data collected via semi structured interviews
- Moustakas (1994) modification of the Stevick-Colaizzi-Keen method was used to analyze the data.
  - Describing personal experiences with the phenomenon under study.
  - Significant statements
  - Meaning units
  - Textual descriptions
  - Structural description
  - Composite description
- NVivo software was used to code and analyze data.

Results

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Six themes emerged from PKU caregivers' experiences in this study

1. Diagnosis experience
2. Stages of care
3. Mealtime
4. Insurance challenges
5. Coping strategies
6. Positive outcomes of caregiving

Diagnosis Experience

- Newborn Screening Process
- Similar story of false positive from pediatrician only to have hope come crashing down when diagnosis was confirmed in PKU clinic.

> *“He said, "You're on my list to call. One of John’s tests came back high and we need to repeat it." I said, "Okay, is this something I need to worry about?" He said, "Nah, nobody has it. Don't worry about it." Then he called me that same afternoon at home and said, "I made an appointment with you at a geneticist for tomorrow morning at 10:00 to repeat that test." And I said, "Oh, that sounds a little more dire, are you sure everything's okay?" And he said, "yeah, don't worry about it. Nobody has it, nobody ever has it.”* (Linda).

Diagnosis Experience (cont'd)

- Misinformation given to parents by pediatrician
- "Hell", "traumatic", "hard", and "frightening".

> *"…for about the first week, every time I would talk about it, I'd cry. It was just ... with everything, we had gone through a lot to get the twins, and it was like, "I don't believe this is happening"* (Camille).

- Disruption of breastfeeding experience
- Need for information

Stages of Care: Three distinct periods

- Participants describe caretaking as occurring across three distinct periods:
  - Early childhood/middle school
  - Adolescence
  - Adult years

Stages of care: Early childhood

- Early childhood/middle childhood - All consuming
  - Management style - "specific," "neurotic," and "all out."

> *"Well, you know, we're talking about my kid's brain, right? So, I pretty much took on everything. I, for his lifetime, have always been responsible for making sure he has formula, for making all of the decisions about doctors…I had 100% responsibility. I took his blood, I called the clinics, I did it all. I did it all. I didn't want any guess work because there was no room for error." (Linda)*

Stages of care: Adolescence

- **Adolescence – Loss of control**

> *"And I think that's probably the hardest point, when they're a teen, and you know later, school-age and teenage, and starting to exert their independence, and still trying to be a caretaker and a monitor but realizing that you've got to give them some freedom and let them try it on their own. And if need be, be there to pick up the pieces."* (Stephanie).

Stages of Care: Adult years

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- ☐ **Adult years – Worry**

It's been really tough. You know like I say letting go of that control and turning it over to them and knowing that they're going to make mistakes and just the worry about the consequences of those mistakes. It's tough. I'm still struggling with that. And yeah like for my daughter who's 20, you know she's away at college now and so that was a lot of anxiety surrounding that but yeah she's really, I really have no control anymore, so I really just got to hand it over and hope you listened, and you know hope you get it. And all of that. I think she's doing a little better than she was a year or two ago but there was definitely a time where I was just sick with worry…" (Robin)

Mealtime

- Family togetherness and strategic
- Two styles
  - Family partakes inclusive of PKU child
  - Two separate meals, sometimes similar

> *"Trying to have same or similar kinds of foods. Another thing that I learned very early was if I'm having three different things on my plate, Jessica needs to have three different things on her plate. And then a mom suggested buying the Tupperware divided plates, because it forces you to put something in each one."* (Rebecca

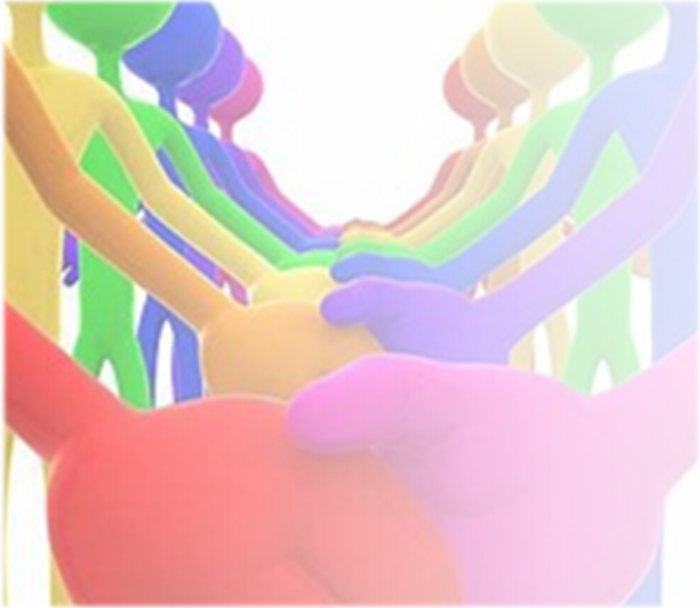
- All reported having adults in varying forms of adherence with management now. Each style seemed well suited to the family depending on its needs.

Insurance Challenges

- Policy context: There is no federal law mandating coverage of medical formula and/or food
- CA – 100% of formula covered, 80% of food
- KS – Spent $32,000 over several years
- MA (pre-coverage law) – Spent $12,000 per year
- TX - $2,500 per month

"Hell, insurance has been our biggest nemesis our entire lives. I've talked to the insurance commissioner. I have talks, I've had meetings, I've appealed my ass off and we've just gotten no where. That's okay. Thank Holy I'm educated. My husband's educated, we drive crappy cars, and we only take vacation that we can drive to so that we can afford John's milk." (Linda).

Coping strategies – what is helping caretakers



- **Faith**

> *“To me, any time you have the child that has some kind of special need, if you're there and you're doing it and doing all that you can, then you're doing a great job. I have a lot of faith. I just believe that you do what you can do. Sometimes that's all you can do.”*(Jessica).

- **Social Support**
  - Camps, conferences, parents’ groups, other families impacted by PKU, family and friends

Coping strategies – what is helping caretakers

- **Acceptance**

> ”I don't know, just accepting that that's kind of the way it was and that's the way we did it. Of course, there were times that they wanted things that other people had, but it wasn't, for the most part ... that they didn't have enough food that they liked on their own, so they didn't mind it and they just accepted it.” (Jessica).

- **Talking to others**

> “And I think I tended to do a lot of talking about PKU in general and dealing with PKU pretty much to anybody who would listen. And that was kind of my relief, in terms of coping with it. And feeling like perhaps people would at least have a little bit of a glimmer of what it was all about.” (Robin)

Positive Outcomes of Caregiving

- ☐ **Cooking**

> *"I felt like I did a lot more cooking, a lot more exploration, and not just relying on a few things. I branched out a lot more and tried a lot more things."* (Camille)

- ☐ **Empathy for Others**

> *"I think it really gives you a better awareness of how everybody else struggles and everybody's dealing with different things and even if you can't see them outwardly. And that was one thing that we really used as a lesson for our children was that things can be hard for people and maybe you don't know it. And just being empathetic to other peoples situations and being aware that other people go through things and yeah this kind of sucks but there are other people who are going through much worse things."* (Joanna).

Positive Outcomes of Caregiving

- **Advocacy**
  - Started a non-profit, advocated for state laws, wrote a book, spoke about newborn screening at March of Dimes events, founded family support groups

Take Home Messages and Opportunities for Change

Emotional support and information

- The diagnosis experience ranged from difficult to traumatic for participants. This research confirms the findings of prior studies that examine the reactions of parents to the PKU diagnosis and find it to be an emotionally stressful event (Strayer & Woten, 2018).
  - Understanding, awareness and empathy from HCPs
  - Emotional support

- Assume that parents might be coming to the diagnosis appointment having experienced difficult events and with misinformation.
  - Provide easy-to-understand information
  - Consider bite-size information and repeat at each visit.

Supporting family life

- Caretakers are strategic about how meals are presented and partaken in by families.
  - Two styles emerged in the research.
  - Support families towards choosing what is best for them
- Caregiving is lifelong. Involving children in management early might help in adulthood

Connecting with others

- Coping strategies of faith, social support, talking to others and acceptance help moms cope with guilt and management of PKU.
    - Ask your veteran PKU Caregivers if they are open to connecting with new parents or provide patients with local patient support group.
    - Social media provide opportunities to connect
- There are positives of caring for a child with PKU!

Coverage assistance support

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- “Battling” insurance companies is part of the caregiving experience.
  - Educate yourself on the coverage assistance programs available to help your patients manage insurance struggles.

What structural changes are needed?

- ☐ Best practices call for social workers to be part of the inter-professional care team and should be involved in the diagnosis event.
- ☐ Reform process of sharing news or coach doctors differently needed.
- ☐ The research underscores, quite loudly, the need for a federal law mandating coverage of medical formula and medical food for people with PKU.

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