Project Report

PKU Adult Care Community

Take me to the
PKU Adult Care Community

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Table of Contents

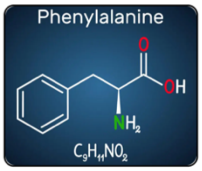
Executive Summary

As part of a Dietetic Internship (DI) program, this project was envisioned, developed and deployed over a one-month time period as a way to provide resources to the adult phenylketonuria (PKU) community. The goal of the project is to determine short and long-term solutions to bridge the care gap needs of individuals with PKU.

This project followed the standard community assessment process and relied on subject matter expertise from two community stakeholders: Kendra Bjoraker and Shiela Ryan. Informal and formal assessment pointed to the need to develop a framework for PKU care – a Care Map (or PKU Journey Management Map). A care map is a tool that individuals can use to document, describe, visualize and characterize their own life. By creating a care map, individuals can be more empowered and equipped to address care barriers such as access to available resources: food, formula, clinics, insurance and mental health care.

Creating a care map community for PKU adults required the following steps:

- Gather community needs from key stakeholders
- Conduct a care map investigation
    - Review formal literature
    - Investigate tools, resources and examples
    - Document candidate care map categories for PKU
- Conduct a formal survey
- Assess survey needs
- Prioritize possible solutions
- Implement a solution
    - Gather care map resources
    - Tailor care map categories to PKU community needs
    - Build a website as a repository for examples and resources
- Document next steps and possible follow-on activities



PKU Adult Care Community

Welcome to the PKU Adult Care Community & Support Website! This is a place where we have collected and organized resources to help you on your life-long PKU journey. Using these examples you can create your own care map, a personal resource to help you focus on critical needs and navigate care. Spending the time to develop your unique story on paper can provide a foundation for self care, empowerment and personal growth.

As this project evolves look for updates on care map workshops, facilitated zoom meetings by licensed practitioners and other tools to hep you on your journey.

PKU Resources

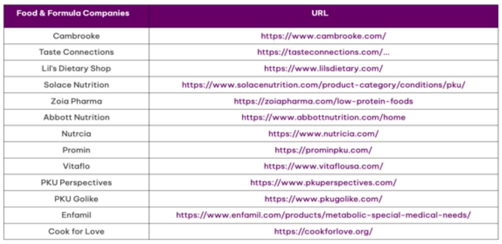
- » *Your Unique Story*
- » *Daily Life*
- » *Health Insurance & Financial Assistance*
- » *Medical Foods & Formula*
- » *Clinical Support*
- » *Career & Education*
- » *Research & Treatment Advances*

Key results of the survey were:

- Strong Desire for Community and Support
- Gaps exist in Adult PKU Care and Clinic Experiences
- Barriers to Care: Insurance, Medical Food Coverage, and Holistic Treatment Approaches

A PKU Community website was created in order to publish, free to the community, example care maps and links to resources https://foodsciencebykc.com/community-experience-copy/.

Medical Foods & Formula



Food & Formula Companies	URL
Cambrooke	https://www.cambrooke.com/
Taste Connections	https://tasteconnections.com/...
Lil's Dietary Shop	https://www.lilsdietary.com/
Solace Nutrition	https://www.solacenutrition.com/product-category/conditions/pku/
Zoia Pharma	https://zoiapharma.com/low-protein-foods
Abbott Nutrition	https://www.abbottnutrition.com/home
Nutrcia	https://www.nutricia.com/
Promin	https://prominpku.com/
Vitaflo	https://www.vitaflousa.com/
PKU Perspectives	https://www.pkuperspectives.com/
PKU Golike	https://www.pkugolike.com/
Enfamil	https://www.enfamil.com/products/metabolic-special-medical-needs/
Cook for Love	https://cookforlove.org/

Two next steps in this project are to explore communication channels to promote the care map concept (FLOK, NPKUA, regional PKU organizations) and to coordinate with existing community programs (e.g. summer camps & adult retreats) to facilitate group sessions for individuals to document their own care map. It will be important to identify funding sources to maintain the website, continue development of tools, and mine resources to keep the information relevant and updated.

Introduction

Phenylketonuria (PKU) is a rare metabolic genetic disorder affecting approximately 17,500 adults, age 18 and older living in the United States. In the last 50 years because of advancements in newborn screening and medical nutrition therapy to restrict protein, individuals with PKU can expect to have normal physical and cognitive growth. However, there are at least 7,800 adults in the US living with PKU that are not receiving metabolic care or following a specialized diet.

The reasons for the care gaps are multi-faceted and complex. Due to uncertainties about treatment needs, the first generation of children diagnosed with PKU (born approximately 1968 – 1980) were taken off diet at age five. The most commonly used PKU diet, low protein foods and formula, is difficult to return to and adhere to due to taste, smell and access. There is a lack of universal insurance coverage for medical foods and there are no adult PKU clinics– so adults must receive treatment at pediatric clinics. The effects of PKU on individual cognition also contribute to the care gap. In particular individuals who are considered "off diet/treatment" are known to have impacts to executive function and decision making. This creates a cascading cause and effect relationship between the need for care and increased difficulty navigating care further amplified by the effects of barriers to care.

What is PKU?

- PKU is a genetic disorder where the body can't process phenylalanine, an amino acid found in protein.
- Without treatment, phenylalanine builds up in the blood, potentially causing physical, neurocognitive, and psychological issues.
- PKU is diagnosed through newborn screening

Treatment: The PKU Diet

- **Low-phenylalanine diet:** The cornerstone of PKU treatment is a special diet that limits phenylalanine intake.
- **Special formulas:** Individuals with PKU drink special phenylalanine-free formulas.
- **Restricted foods:** High-protein foods like meat, fish, eggs, nuts, beans, and dairy products are avoided.
- **Allowed foods:** Low-protein foods like fruits, vegetables, some grains and other specially formulated medical foods are part of the PKU diet.
- **Aspartame avoidance:** The artificial sweetener aspartame, which contains phenylalanine, must be avoided.
- **Supplements:** Children with PKU may need vitamin and mineral supplements, if they do not get it from their formula, to ensure proper growth and development.

Treatment: Medications

- **Sapropterin (Kuvan):** A medication that helps some people with PKU process phenylalanine more efficiently.
- **Pegvaliase (Palynziq):** A medication that can help lower phenylalanine levels.

Monitoring and Testing

- **Regular blood tests:** Blood phenylalanine levels are monitored regularly to ensure they stay within a safe range.
- **Frequent testing:** Newborns with PKU may need testing as often as once a week or more often for the first year of life.
- **Ongoing monitoring:** Testing may continue throughout childhood and adulthood.

PKU Care Team

- **Primary care physician:** The primary care medical provider for PKU patients.
- **Metabolic specialist:** A doctor specializing in metabolic disorders like PKU.
- **Genetic specialists**: A doctor specializing in diagnosing and treating genetic disorders.
- **Registered dietitian:** Provides medical nutrition therapy. Develops and educates patients on the PKU diet to include meal plans, access to specialized formulas, food sources and recipes.
- **Neuropsychologists:** A specialist in brain function and how it impacts an individual’s thinking, behavior and emotions. They conduct evaluations to help improve and optimize all aspects of daily living and mental health.
- **Psychologist/counselor:** Provides emotional support and guidance to the family.
- **Social worker:** Helps with navigating the healthcare system and accessing resources.
- **Patient Advocacy Specialist:** pharmaceutical representatives that help navigate prescribed therapies
- **Patient Advocacy Groups (PAG):** Organizations that represent and support patients and their families who are dealing with specific health conditions or diseases.

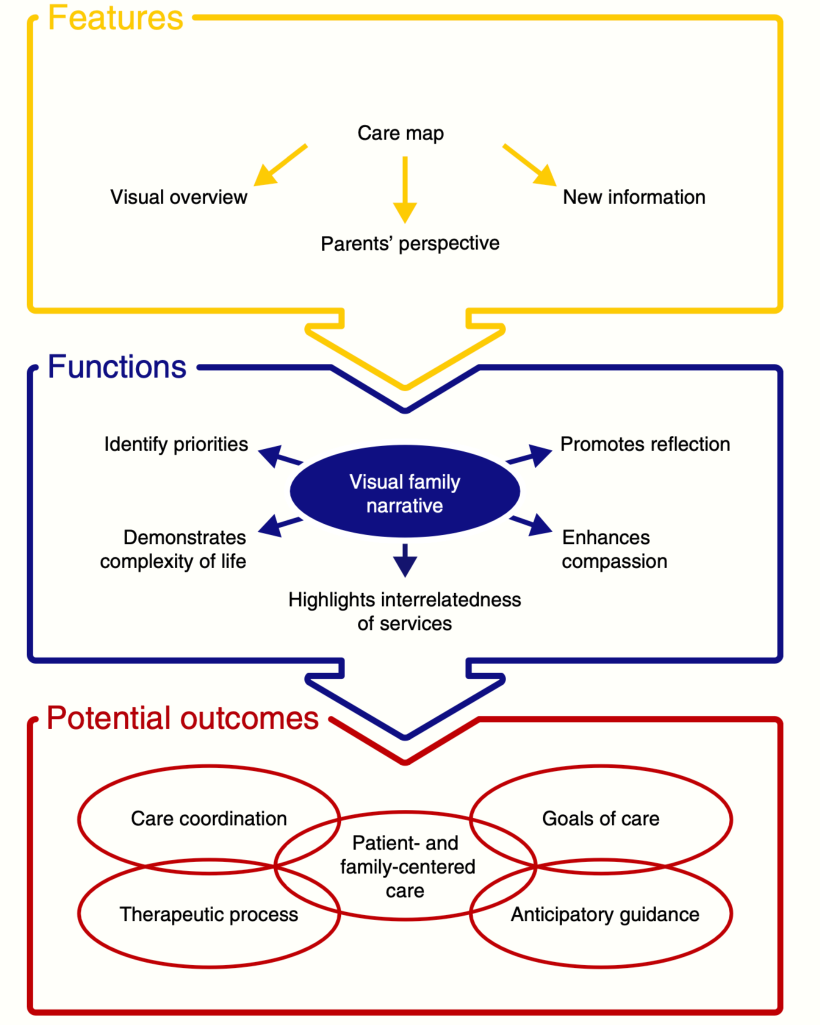
Importance of Lifelong Management

- **Access to care**: clinic appointments: medication prescriptions, and monitoring, laboratory testing
- **Continued diet:** The PKU diet is followed throughout life.
- **Pregnancy:** Women with PKU need to strictly follow the diet both before and during pregnancy to prevent complications for the fetus.
- **Psychosocial support:** The challenges of managing PKU can have a significant impact on the individual's psychological and emotional well-being, so access to counseling and support groups is important.

Elements of the Care Map

Placing the Individual at the Center of Their Story

The concept of a care map was originally developed by a parent of a special needs child as a way to put on paper all of the different aspects of daily life – medical team, school, social impacts, treatments, insurance, hobbies, family and community support. As shown in the graphic below, features of a care map are documented that place the individual in the center of care leading to improved outcomes such as focused care coordination, revised/updated goals for care, and new therapeutic processes.



Creating a holistic view of PKU care, management and wellness that is simple to understand and easy to tailor for each individual requires careful consideration of key questions and categories of information to consider.

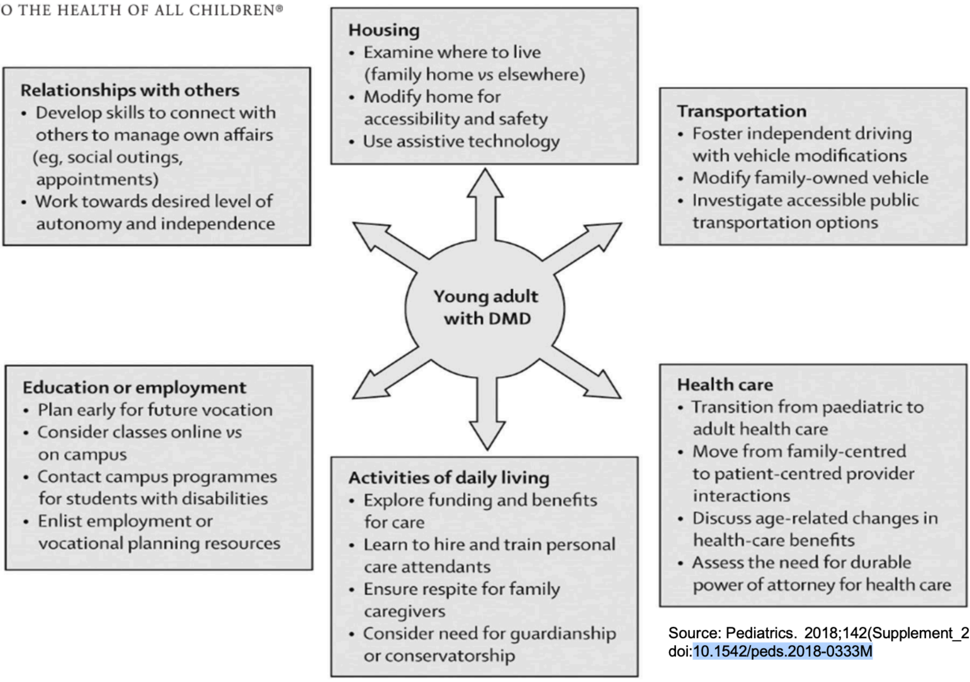
Care Map Examples

Most examples of care maps have been developed by families of patients with specialized, unique or complex pediatric care needs.

American Academy
of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®

Journal Article: *A Transition Toolkit for Duchenne Muscular Dystrophy*



Young adult with DMD

Housing
- Examine where to live (family home *vs* elsewhere)
- Modify home for accessibility and safety
- Use assistive technology

Relationships with others
- Develop skills to connect with others to manage own affairs (eg, social outings, appointments)
- Work towards desired level of autonomy and independence

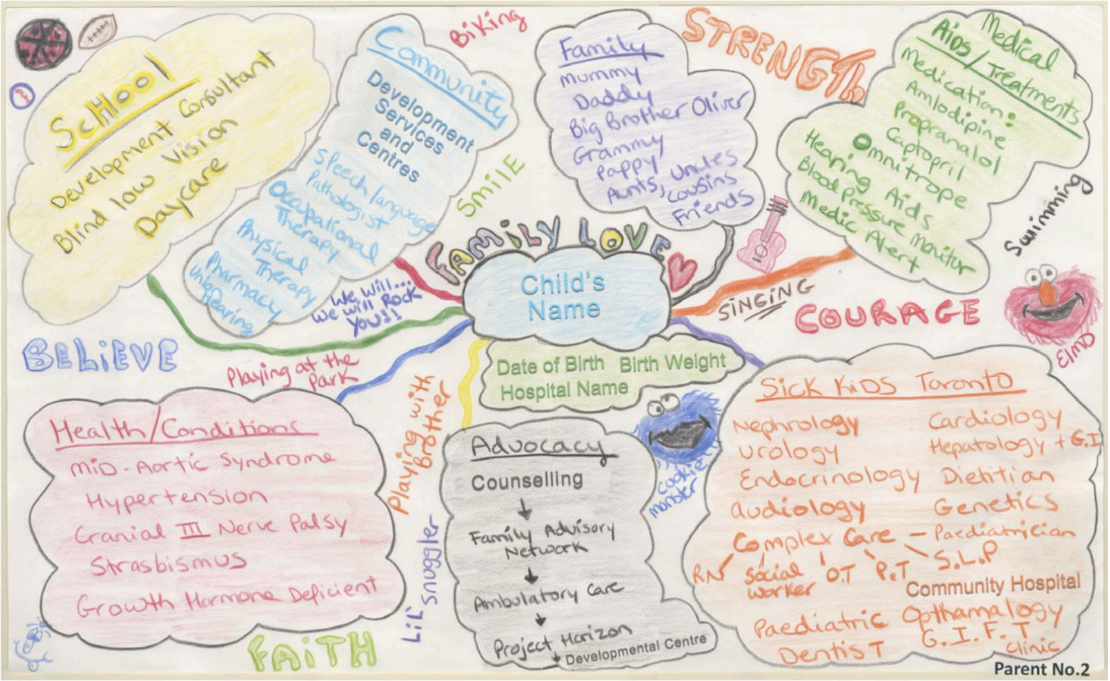
Transportation
- Foster independent driving with vehicle modifications
- Modify family-owned vehicle
- Investigate accessible public transportation options

Education or employment
- Plan early for future vocation
- Consider classes online *vs* on campus
- Contact campus programmes for students with disabilities
- Enlist employment or vocational planning resources

Activities of daily living
- Explore funding and benefits for care
- Learn to hire and train personal care attendants
- Ensure respite for family caregivers
- Consider need for guardianship or conservatorship

Health care
- Transition from paediatric to adult health care
- Move from family-centred to patient-centred provider interactions
- Discuss age-related changes in health-care benefits
- Assess the need for durable power of attorney for health care

Source: Pediatrics. 2018;142(Supplement_2):S110-S117. doi:10.1542/peds.2018-0333M



Parent No.2

Tailored PKU Care Map Categories

Documenting a care map and creating a community with a corresponding website is a way to build a sense of belonging as well as establishing a mechanism for shared stories/lived experience experiences: celebrating progress, finding a role model, developing stronger resilience, coping skills and increasing general wellness.

The support community is central to the care map concept.

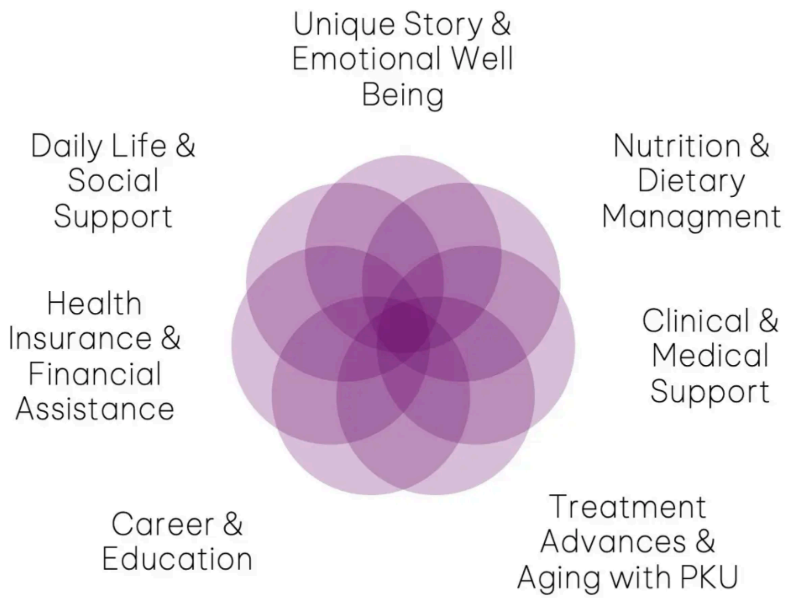
- Family: Parents, siblings, other members
- Peer networks: mentorship, online communities, advocacy groups
- Relationship to PKU care team (clinicians, dietitians, psychologists)
- Role of PKU organizations and patient groups in fostering belonging

The list below of seven categories is a starting point to develop a care map. They are not boundaries or fixed categories. Instead, these categories are meant to create ideas, stimulate thoughts so that each individual will create their own personal, unique map to inform and guide their own care.

1. Your Unique Story & Emotional Well Being

- Personal journey with PKU: early experiences, challenges and milestones
    1. Identify beyond PKU: Passions, relationships, personal vision and life goals
    2. Lived experiences: how PKU has shaped personal resilience and adaptability
    3. Self-compassion, confidence and general outlook
- Managing PKU-related anxiety & stress
    1. Accessing mental health counseling/support groups
    2. Overcoming PKU burnout & treatment fatigue
    3. Self-care & stress management techniques
    4. Ability to manage feelings and stress related to living with a chronic condition
    5. Self-care resources developed by a Neuropsychologist

Your Life Journey Care Map



Resource	URL
Care Map Examples	Care Map Examples
Vision Board	Link (Under Construction)
Goal Planning	Link (Under Construction)

2. Daily Life & Social Support

- Housing & living situation
- Transportation
- Time management for PKU care
- Peer mentorship & PKU community involvement
- Educating family, friends, and coworkers about PKU
- Advocating for PKU awareness in schools & workplaces

Daily Life & Social Support

Resource	URL
PKU Binder	https://www.npkua.org/wp-content/uploads/...
Clinic Listing	https://www.npkua.org/resources/find-a-clinic/
Mentor program	https://www.npkua.org/support/mentor/
Conference & Events	https://www.npkua.org/npkua-events/
Regional Affiliate Organizations	https://www.npkua.org/affiliates/
FLOK App	https://flok.org/nutrition/...
NPKUA	https://www.npkua.org/

3. Nutrition & Dietary Management

- Low-protein diet adherence
- Access to PKU-friendly food options
- Understanding food labels & protein tracking
- Meal planning & cooking strategies
- Geographic access to PKU specialists
- Transportation challenges for clinic visits
- Affordability & availability of medical foods
- Overcoming stigma & misinformation about PKU Access to food
- Navigating diet (social, travel, dining out, holidays, special occasions

Nutrition & Dietary Management

Food & Formula Companies	URL
Cambrooke	https://www.cambrooke.com/
Taste Connections	https://tasteconnections.com/...
Lil's Dietary Shop	https://www.lilsdietary.com/
Solace Nutrition	https://www.solacenutrition.com/product-category/conditions/pku/
Zoia Pharma	https://zoiapharma.com/low-protein-foods
Abbott Nutrition	https://www.abbottnutrition.com/home
Nutrcia	https://www.nutricia.com/
Promin	https://prominpku.com/
Vitaflo	https://www.vitaflousa.com/
PKU Perspectives	https://www.pkuperspectives.com/
PKU Golike	https://www.pkugolike.com/
Enfamil	https://www.enfamil.com/products/metabolic-special-medical-needs/
Cook for Love	https://cookforlove.org/

4. Health Insurance & Financial Assistance

- Understanding insurance coverage for medical foods
- Financial assistance programs & grants
- Employer benefits & workplace accommodations
- Budgeting for PKU-related expenses

Health Insurance & Financial Assistance

Health Insurance Resources	URL
General Insurance Coverage	https://www.npkua.org/wp-content/uploads/...
State Statutes	https://ciswh.org/resources/dietary-treatment-statutes/
InsuranceTerminology	https://www.npkua.org/resources/insurance-coverage/
Letter of Medical Necessity Example	https://acrobat.adobe.com/id/...
Letter of Medical Exclusion PKU	https://acrobat.adobe.com/id/...

Financial Assistance Resources	URL
Abbott Pathway Plus	https://www.pathwayreimbursement.com/
BioMarin Rare connections	https://www.biomarin-rareconnections.com/
CAMBROOKEcare	https://www.cambrooke.com/support/reimbursement/
Coram Healthcare	https://www.coramhc.com/
Cycle Vita	https://cyclevita.life/
KUVAN Patient Assistance Program (K-PAP)	https://www.kuvan.com/support-resources/
Mead Johnson Helping Hands	https://hcp.meadjohnson.com/s/insurance-assistance...
Medicaid	https://www.medicaid.gov
Patient Advocacy Foundation	https://patienthelpline.org/...
Vitaflo Formula4Success	https://www.vitaflousa.com/formula4success
Eaton Pharmaceuticals	https://www.etonpharma.com/
Rx Assistance - Patient Assistance	https://www.rxassist.org/patients

5. Clinical & Medical Support

- Existence of a PKU Care team (doctors, dietitians, psychologists, other providers)
- Care partnerships for shared decision making with providers
- Access to laboratory for testing blood Phe levels
- Prescription & access to medical foods/formula
- Frequency of clinic visits and Phe monitoring
- Place to learn about treatment options
- Cognitive function and strategies for maintaining brain health
- Access point for other specialized care (family planning, mental health, neurological testing, etc.)
- Use of telehealth and digital tools for better information, faster

Clinical & Medical Support

Clinical Support Resources	URL
Clinic Finder	https://www.npkua.org/resources/find-a-clinic/

Rare Disease Resources	URL
Rare Disease Resources	https://raredisease.powellcenter.med.ufl.edu/...
Rare Disease Podcasts	https://raredisease.powellcenter.med.ufl.edu/...

6. Career & Education

- School accommodations (504 plans, IEPs)
- Transitioning from pediatric to adult PKU care
- Workplace support for PKU dietary needs
- Career choices & managing PKU in the workplace
- Managing PKU while in college or professional training
- Workplace accommodations and self-advocacy
- Impact of PKU on cognitive function and work performance

Career & Education

Resource	URL
Unique to Each Individual	Link (Under Construction)
Career Planning	Link (Under Construction)
Scholarship Programs	Link (Under Construction)

7. Treatment Advances & Aging with PKU

- Research & emerging treatments for PKU
- Engaging in clinical trials & advocacy efforts
- Long term health implications
- Cognitive health & aging considerations
- Managing PKU through life transitions (college, parenthood, aging)

Treatment Advances & Aging with PKU

Research & Treatments Resource	URL
NPKUA Research Grants	https://www.npkua.org/wp-content/uploads/...
Clinical Trials	https://clinicaltrials.gov/search?cond=PKU
PCT Therapeutics	https://pubmed.ncbi.nlm.nih.gov/39368841/
Gene Therapy	https://www.youtube.com/watch?v=5ChXI6cSQs0

Electronic Survey

The purpose of the survey was to understand barriers to care that could be addressed by forming a community, defining a PKU care pathway, creating new resources (toolkit, handouts, care guide, care facilitators) or improving communication and access to treatments.

Method: Create a concise list of questions to be able to quantify and prioritize ways to improve outcome, measured by number of adults who return to care at a metabolic clinic.

Survey Questions

Consent Agreement – Yes.

1. Do you believe that regular care from a licensed metabolic practitioner is necessary to maintain your overall health?
    a. Yes, absolutely required
    b. Yes, a good idea
    c. Unsure either way
    d. No, optional
    e. No, Not Needed at all

2. At what age do you believe that specialized PKU medical treatment is required for optimal wellness?
    a. Age 0-5 years old
    b. Age 5-18 years old
    c. Age 19 – 50 years old
    d. Age 50-75 years old
    e. Lifetime of care is needed.
3. Prioritize the reasons why PKU treatment and care is needed:
    a. Prevent Brain fog
    b. Improve memory and organizational abilities (Executive Function)
    c. Improve attention
    d. Prevent physical effects (tremors, skin rashes, brittle hair, paralysis)
    e. Provide for care during pregnancy/maternal health
4. What is your preferred method of receiving care for PKU?
    a. Phone
    b. Video
    c. Email
    d. In person
5. How frequently do you <u>want to interact</u> with your PKU/Metabolic clinic for information or treatment?
    a. Monthly
    b. 2 times per year
    c. 1 time per year
    d. Every 5 years
    e. I prefer not to interact with a clinic
6. What type of information would you like to receive at your clinic visits
    a. Results of recent blood tests results
    b. Taste test new formulas or foods
    c. Learn about new treatment options
    d. Learn about maternal PKU care
    e. other _____).
7. Are you interested in participating in community events, activities or informal care networks? (Y/N)
    a. If yes, what types of <u>group</u> resources or events are you interested in
        i. Online events (Zoom) focused on <u>PKU related</u> topics: (nutrition, cooking, new treatments and care)
        ii. Online events (Zoom) focused <u>non PKU activities</u> (cooking class, art class, book club, travel)
        iii. Face to face events (conferences, group dinners, sports events, outdoor activities, camp)
        iv. None
        v. Other ______).
    b. If yes, what types of <u>private</u> resources or events are you interested in:

        i. Videos and podcasts related to PKU care
        ii. Online applications and websites – e.g., FLOK, NPKUA
        iii. Personal coaching by phone/video, with a licensed provider.
        iv. Anonymous online coaching or chat, with a licensed provider.
        v. None
        vi. Other ______).

8. Who do you talk to about your experience living with PKU?
    a. Parent or family members
    b. Other adults/friends with PKU
    c. Other adults/friends without PKU
    d. Health provider (PCP, clinic)
    e. PKU is not something I typically talk about
9. How do you feel about having PKU, and your ability to manage your care? (can select multiple answers)
    a. Proud
    b. Self-confident
    c. In-control
    d. Neutral
    e. Anxious
    f. Ashamed
    g. Depressed
    h. Other (explain in 1 word) _________.
10. Would you be interested in participating in a phone/Zoom interview to further discuss PKU community care needs? Y/N

Survey Results

The survey was sent out on 3/25/25 to 25 Adults with PKU, and 11 responses were returned by 3/27/25. Open ended verbal comments were received in the NPKUA Community Mentor Meeting 3/26/25.

Top 3 Key Takeaways

1. **Strong Desire for Community and Support**
   Individuals with PKU highly value both virtual and in-person opportunities for education, treatment discussions, and peer support. Group activities, whether online or in-person, motivate them to manage their PKU more effectively. Additionally, they want private access to PKU-specific resources.
2. **Gaps exist in Adult PKU Care and Clinic Experiences**
   Many adults feel that PKU care remains overly focused on childhood, leading to frustration with pediatric-centered clinics. They express a need for adult-focused care, more personalized meal planning, and proactive discussions about treatment management. Standardizing PKU care across clinics and improving patient-provider communication (using patient-centered language) are seen as essential improvements.
3. **Barriers to Care: Insurance, Medical Food Coverage, and Holistic Treatment Approaches**
   Insurance challenges, lack of medical food coverage, and the high cost of low-protein foods create significant obstacles to PKU management. Patients also feel that clinics often focus

narrowly on Phe levels rather than addressing broader concerns like mental health, neurological symptoms, and lifestyle-specific dietary needs.

Survey Raw Data

1. 100% show moderate to high interest in participating in virtual group nutrition education, cooking classes or treatment discussions.
2. 100% show moderate to high interest in participating in group In Person events (conferences, sporting events, outdoor activities)
3. 100% show moderate to high interest in private access to PKU community support resources (websites, FLOK, NPKUA)
4. 100% talk about their experience with PKU with a parent or family member
5. 100% want the results of blood tests during clinic visits
6. 91% prefer In Person Care
7. 91% want to learn about new treatment options during clinic visits
8. 82% believe regular care from a licensed metabolic practitioner is necessary for health.
9. Top three reasons for treatment
    - 82% Prevent Brain Fog
    - 82% Improve memory and executive function
    - 73% Prevent physical effects
10. 64% want to interact with their clinic 2x per year
11. Key Comments
    - CLINIC
        - Want to learn about new low protein foods at clinic
        - Would like to work with dietitian on meal planning:
            - Palynziq – high protein choices
            - Maternal Gestational diabetes – low sugar choices
            - Personalized meal plans
            - Meal plans for athletes
            - Meal plans for weight loss
            - Meal plans that take into account restrictions/lifestyle
    - Group Activities
        - Any group activity motivates me to manage my PKU to the best of my ability
        - Any group where multiple PKU individuals are present
    - Feelings about PKU [Not a great designed question, as participants could select multiple answers]
        - 64% In control
        - 27% Anxious, depressed
    - 8 responded positively to a follow up Zoom Interview
    - Open Comments:

- "I know this is a widespread issue not easily solved, but being seen in a pediatrics clinic is frustrating. Every time I check in, they ask where my child is. It's just another reminder every time of the laser focus on childhood with PKU in the community as a whole. "

- “Insurance has been a barrier to the entire PKU community at some point in their lives”
- "Lack of adult PKU clinics (as a 46-year-old man, my needs/concerns/nutrition requirements are quite a bit different than a 6 year old boy or a 16 year old). One size fits all diet advice ("eat more fruits and vegetables" Cost of low protein foods and lack of insurance coverage for PKU medical formulas. Clinical focus on keeping phe levels "in range" without considering other aspects (my levels are "good" but my clinic is completely unconcerned about weight gain, anxiety/depression, neurological symptoms, brain fog, etc.)"
- "The lack of medical food coverage is a significant barrier to PKU care. I also think clinic experience is significant. I’ve had poor interactions with geneticists that don’t listen to my goals and push their agenda and I know this isn’t an isolated occurrence as I’ve heard this throughout the community. Providers need more education on patient centered language and collaboration with the patient on achieving treatment goals. I know how important going to clinic is to maintain my PKU management but I know others who stop going to clinic because of poor interactions with providers and clinicians. I also believe there is a need for standardized practice across clinics. The CF community has standardized guidelines and practices and this model could be implemented within the PKU and other IEM communities for consistent care."
- "Would love to have proactive conversations about how to effectively manage the diet with Palynziq ie. assistance with meal planning for high protein options."

Informal Zoom Interview Focus Group

Following the Care Map categories outlined above, a focus group session will occur to experiment with both the value of creating a care map and the process steps for facilitating the development of a care map.

Purpose: Understand personal reasons associated with obtaining care, motivations, barriers and concepts for care/community formation.

Gain input on SPECIFIC resources/links to add into the Care Map such as Formula companies, treatments, wellness resources, coping strategies. Gain ideas for care coordination needs, roles, tools, methods, etc. with the goal of getting adults into care.

1. What is your biggest challenge in managing PKU as an adult, and how has it impacted your daily life?
    a. Sticking to the low-protein diet and formula consistently
    b. Balancing PKU management with work, school. Or social life
    c. Dealing with the emotional or mental health impact of PKU
    d. Lack of motivation or feeling that PKU care doesn’t fit into my life
    e. Other: explain
2. If you have ever stopped attending clinic or struggled to stay engaged with care, what factors contributed to that decision?
    a. Time constraints or difficulty scheduling appointments
    b. Feeling like clinic visits don’t provide helpful or new information
    c. Negative past experiences with healthcare providers
    d. Cost, insurance, or transportation barriers
    e. Other: explain
3. What emotions do you associate with going to clinic – whether positive or negative?

    a. Anxiety or stress – I worry about judgement or bad news
    b. Indifference – I don’t feel like it makes a big difference in my life
    c. Motivation – I feel more accountable and encouraged to manage my PKU
    d. Frustration – I wish there were better resources or support for adults
    e. Other: explain
4. Do you currently feel judged or misunderstood by healthcare providers when discussing your PKU management? If so, what would make that experience better? If so, what would make that experience better?
    a. Yes, I’ve felt judged – less pressure and more understanding would help
    b. Sometimes – I wish providers would ask about my challenges rather than assume things
    c. No, my provider listens to me and understands my experience
    d. I don’t attend clinic often enough to say
    e. Other: explain
5. What are the biggest barriers (time, cost, access, feeling overwhelmed, lack of trust, feeling shamed, words and language they use, etc.) that make it difficult for you to stay connected to PKU care?
    a. Time – I’m too busy to keep up with appointments and diet
    b. Cost – insurance, formula coverage, or clinic visits are a financial burden
    c. Emotional burnout – I feel overwhelmed or disconnected from my care
    d. Lack of access – there are no convenient clinic options or telehealth services
    e. Other: explain
6. How do you feel PKU fits into your identity – do you see it as a central part of who you are, or something in the background?
    a. PKU is a big part of who I am and influences my daily life
    b. It’s important, but I don’t let it define me
    c. I don’t think about PKU much unless I have to manage it
    d. I try to ignore it because it feels overwhelming or frustrating
    e. Other: explain
7. What type of support – whether from family, peers, or healthcare providers – would make it easier for you to engage with your PKU management?
    a. More understanding and involvement from family and friends
    b. Better communication and encouragement from my healthcare team
    c. A strong peer community of adults with PKU who relate to my experience
    d. More flexible or personalized treatment options
    e. Other: explain
8. If the clinic experience could be redesigned in a way that worked better for you, what would that look like?
    a. More virtual or telehealth options instead of in-person visits
    b. Shorter, more focused appointments with on the necessary tests and check-ins
    c. A more adult-centered approach that acknowledges different life stages and challenges
    d. A more welcoming, supportive environment instead of just medical discussions
    e. Other: explain
9. Are there any things that would increase your satisfaction with clinic visits?
    a. No, I am getting personalized, useful guidance for my specific needs

    b. Yes, if I could learn about new treatment options or research that could change my management
    c. Yes, if the clinic experience were more flexible and less time-consuming
    d. Yes, If I had a stronger connection with my healthcare team and felt truly supported
    e. Other: explain

10. If you could give one piece of advice to healthcare providers about working with adults with PKU, what would it be?
    a. Listen without judgment – everyone's PKU journey is different
    b. Make care feel relevant – help me understand why it matters for my future
    c. Offer more flexibility – acknowledge that adult life is busy and complex
    d. Focus on real-life solutions, not just numbers and lab results
    e. Other: explain

Additional open-ended questions:

- How has PKU impacted your emotional well-being, and what support or coping mechanisms have helped you the most?
- Who in your life has been the most supportive in your PKU journey, and what kind of support do you wish you had more of?
- How would you describe your relationship with your PKU care team, and what could make your healthcare experience better?
- Have you ever faced challenges with PKU in your education or career, and how have you navigated them?
- What concerns, if any, do you have about managing PKU as you get older, and what would help you feel more confident about the future?

Forming a Community & Next Steps

Forming a community of adults is a difficult task – particularly when a combination of socio-economic, access to information and cognitive barriers exist. The first step in the process was to create a single website where information could be gathered into a central place.

Social media could be used to create self-service care guides, identify care map facilitators or success stories from people who have used a care map. One of the most important next steps could be connecting to existing PKU clinics to reach a broader audience.

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