



# Making the choice between medical management and liver transplant: The Family Experience

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# Treatment Options for UCD

Should a patient be managed with diet and medications or should they undergo a liver transplant?

## Medical Management

- Rx drugs (Buphenyl, Ravicti)
- Highly restricted low protein diet
- Nutritional supplementation (metabolic formula) and amino acid supplements
- Does not remove risk of hyperammonemia

## Liver Transplant

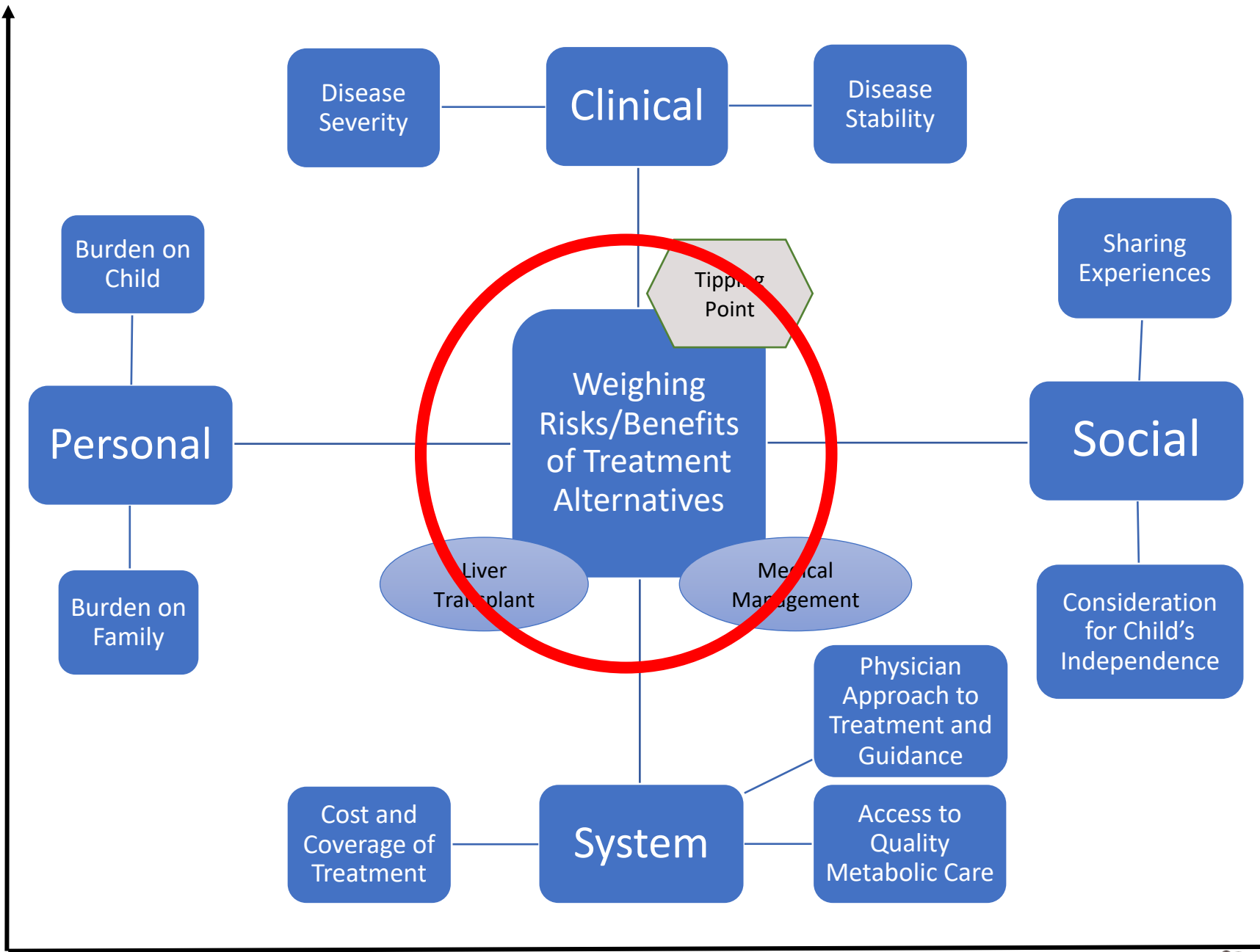
- Removes risk of hyperammonemia
- Risk of surgical complications
- Life-long immunosuppression
- Little data on long-term consequences

# About the Study

- No existing research on treatment decision-making in UCD
- Study conducted in collaboration with NUCDF, GW University, Children's National Hospital
- Examine how UCD treatment decisions are made
  - Describe the factors that influence a family's decision to continue medical management or choose liver transplant
- Hope to promote better delivery of care and information to UCD patients and their families

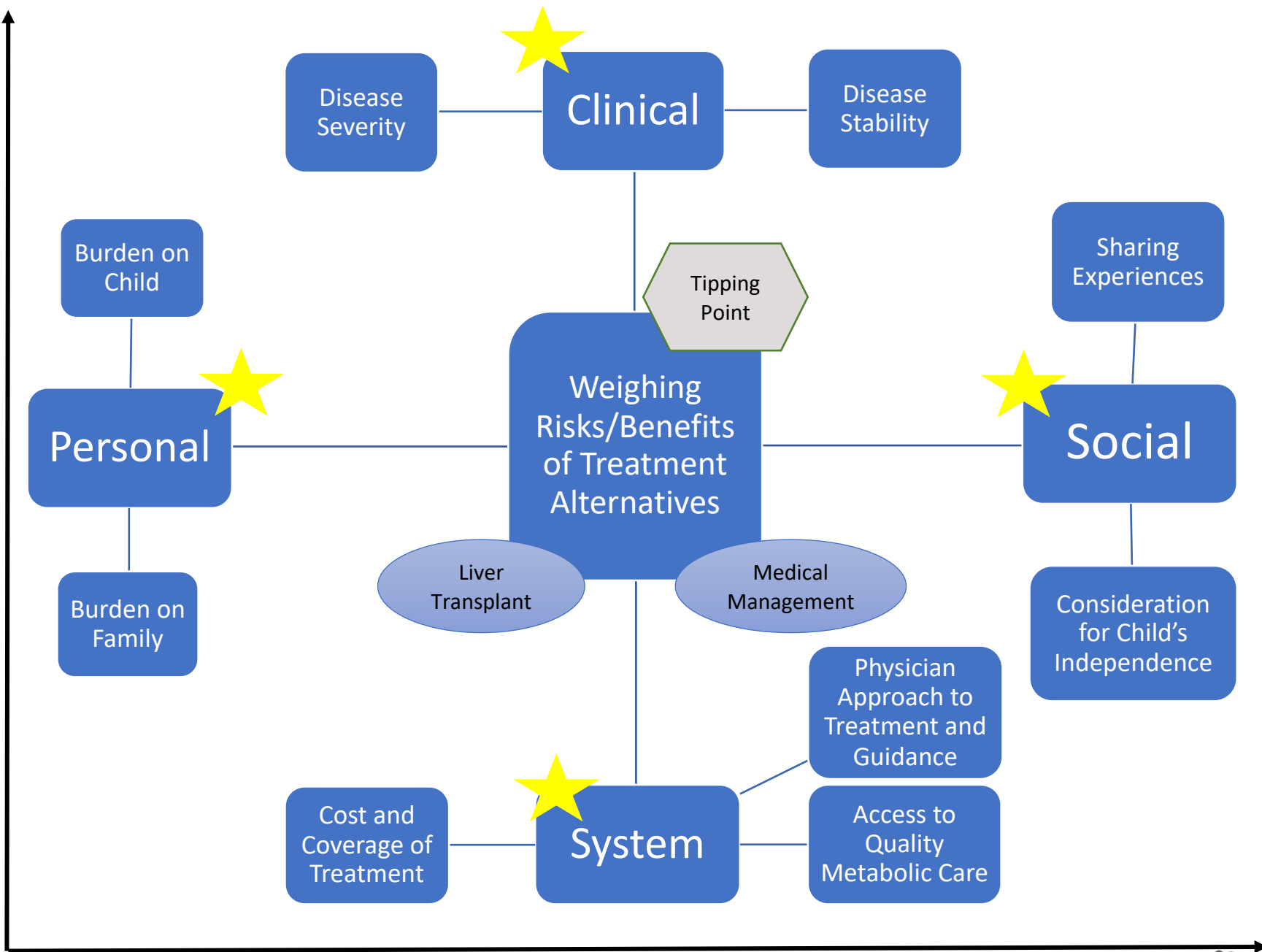
# Methods

- Phone interviews with parents of children diagnosed with UCD (N=34)
  - Neonatal and late onset
  - Medically managed and transplanted
  - Children of all ages
  - Living in different states/regions of the country
- Phone interviews with clinical providers (N=26)
  - Different types of providers on the care team
  - Different practice locations
- NUCDF hosted In-person focus groups at the NUCDF conference and web-based focus groups with parents (N=31)
- In-person and web-based focus groups with providers (N=19)



# Weighing the Risks and Benefits

- Parents put considerable effort into understanding and analyzing the relative risks and benefits of MM and LT
  - But limited data/scientific evidence to inform this decision means parents and providers often struggle to compare MM and LT
- Parents rely on experiences (their own, their providers and their peers) to guide this choice
- Clinical, personal, social, and system level issues inform each family's personal perception of risk and benefits



# Phases of Childhood and Developmental Milestones



- Major changes during key phases of child's development create new or aggravate existing challenges of MM for UCD
- Developmental phase can be a catalyst for parents to consider (or reconsider) LT as a viable treatment option
- May change patient and parent priorities, and re-frame family's perception of the risks and benefits of MM vs. LT



*“It’s just a new chapter in our lives. In order for the possibility of her [daughter] being able to maybe go to college...just be able to think about things like that...If we can’t get it under control now, I don’t know how we’re going to be able to get it under control in the next couple of years.” ~Parent*



# Disease Severity

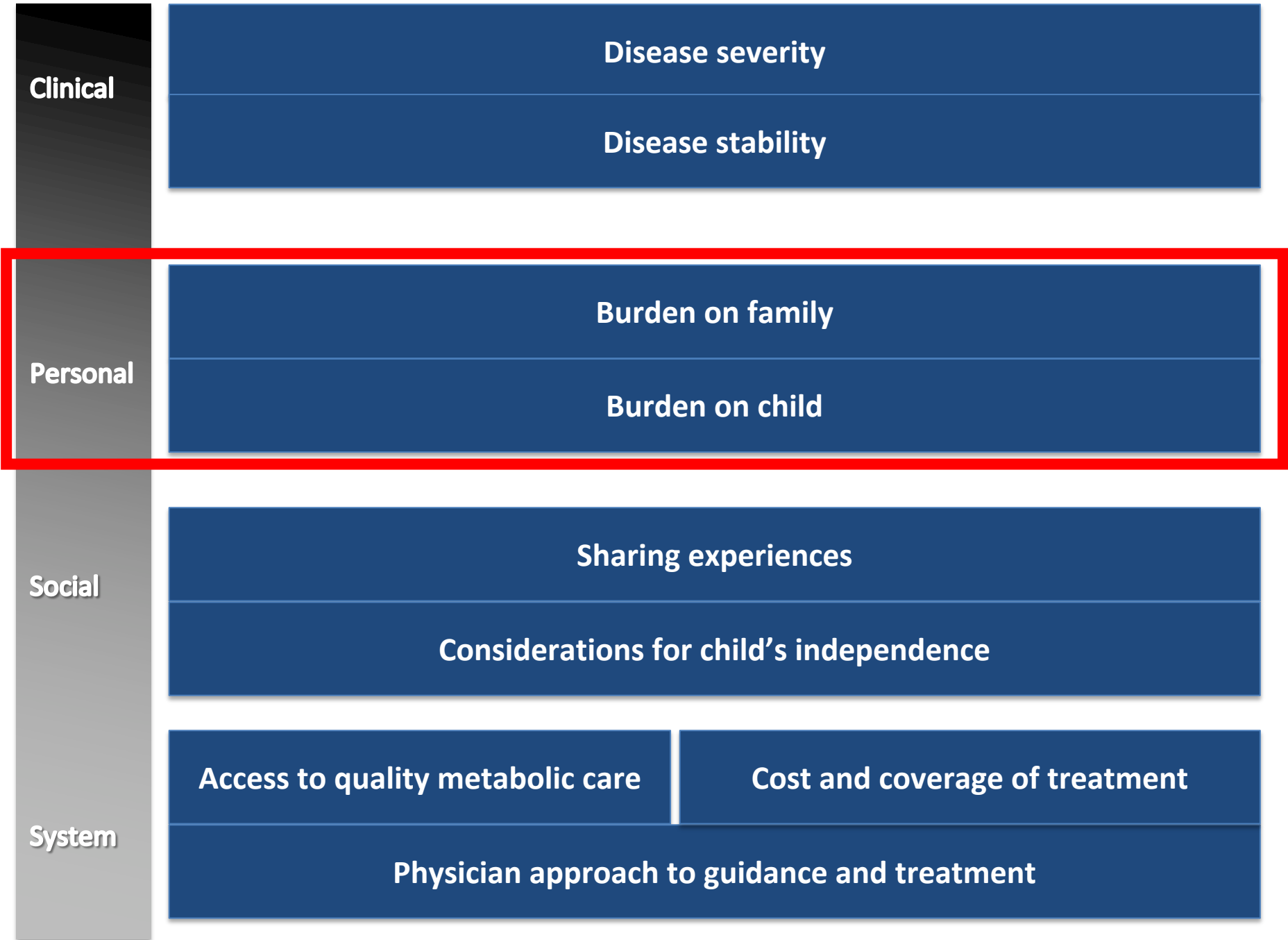
- Severity of diagnosis cited by parents and providers as key consideration in treatment choice
- Neonatal-onset and/or zero enzyme function
  - LT commonly described as child's "only option" or "best chance at long-term survival"
- Late-onset and/or partial enzyme function
  - A much more subjective and complex evaluation

# Disease Stability

- Periods of instability as a catalyst for transplant
- Transplant as a preventative measure to avoid future instability and preserve neurocognitive function
  - For some, past or current disease stability was not an indication of future disease control
  - Feared the unpredictable nature of high ammonia

*“From everything that I’ve heard, it should be for us more of a last resort scenario. My daughter’s condition, for the most part, has been pretty well-controlled... In my mind, if that is still feasible for us, why would we take on the risks that are associated with the liver transplant?” ~Parent*

*“Tons of reasons as to why we felt transplant was a better option, even though he was quite a stable kid so far, but everybody told me that might not be the case forever. That can change literally overnight, so that’s why we decided to go for the transplant.” ~Parent*



# Burden on Family

- Day-to-day challenges of managing UCD
  - 24/7 medical caregiving
  - Fear, worry, anxiety and the families emotional health
  - Altered relationship to “normal life” comforts
- Daily burdens can be a compelling reason to consider transplant
- Daily burdens may *not* drive families to pursue transplant when:
  - Parents feel mastery and comfort with child’s routine
  - Concerns about new and unfamiliar risks and burdens of transplant outweigh concerns about risks and burden of UCD

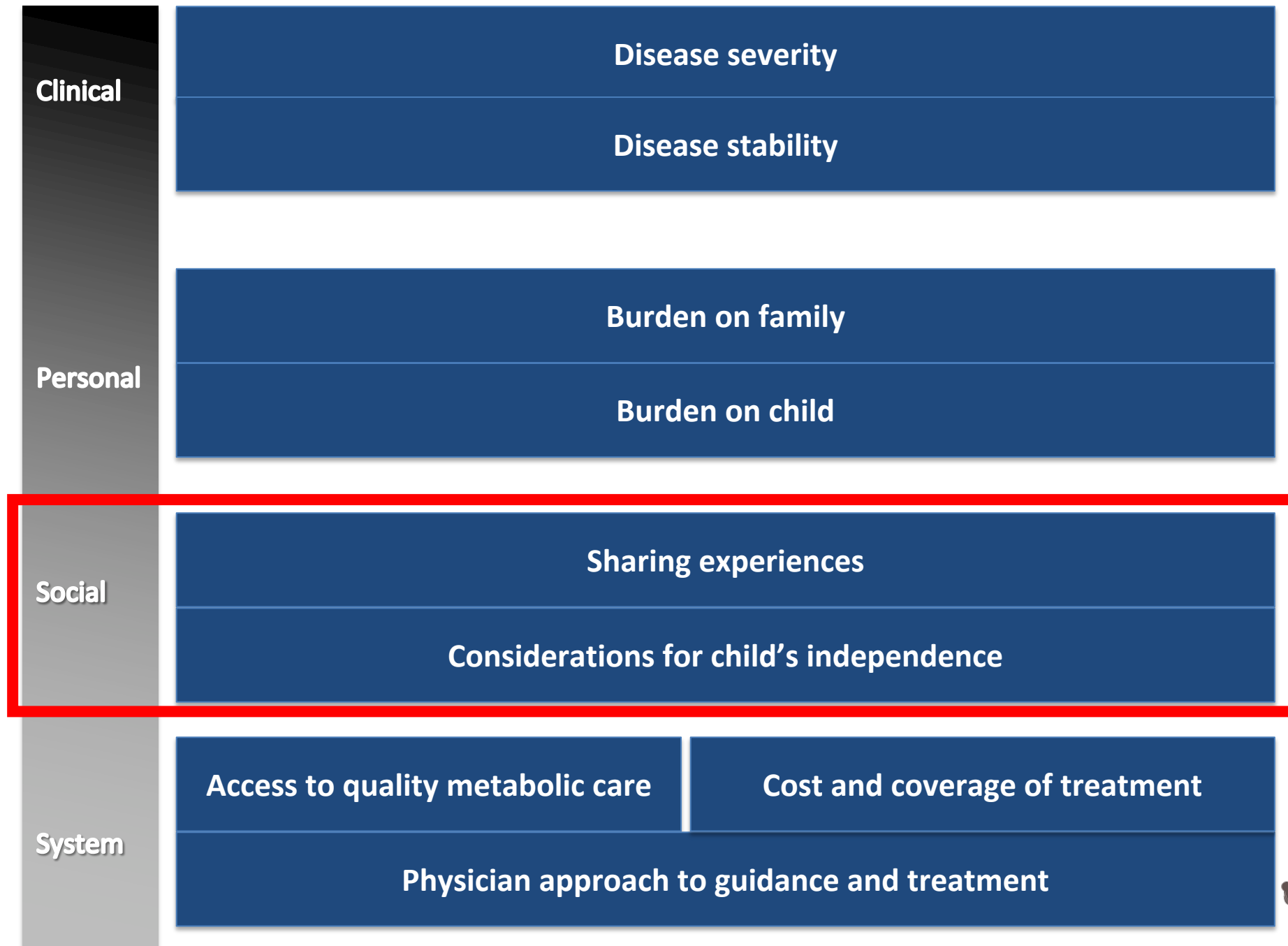
# Burden on Child

- Burden of illness on child's QOL
- Impact of UCD on child's intellectual and social development
- Parents ask- can transplant offer child an opportunity at a "normal" and better quality life?
- Parents who felt child was thriving socially and intellectually were less motivated to pursue transplant



*“That was one of the things in school. He learned how to count money, and that was a huge thing because he worked and worked at it. Then he had a high ammonia level...He remembered that he knew how to count money, but he couldn't count it anymore. We thought, oh, that quality of life's horrible...To lose that functionality was devastating for him. That played into [the decision to transplant] too.”*

*~Parent*



# Sharing Experiences

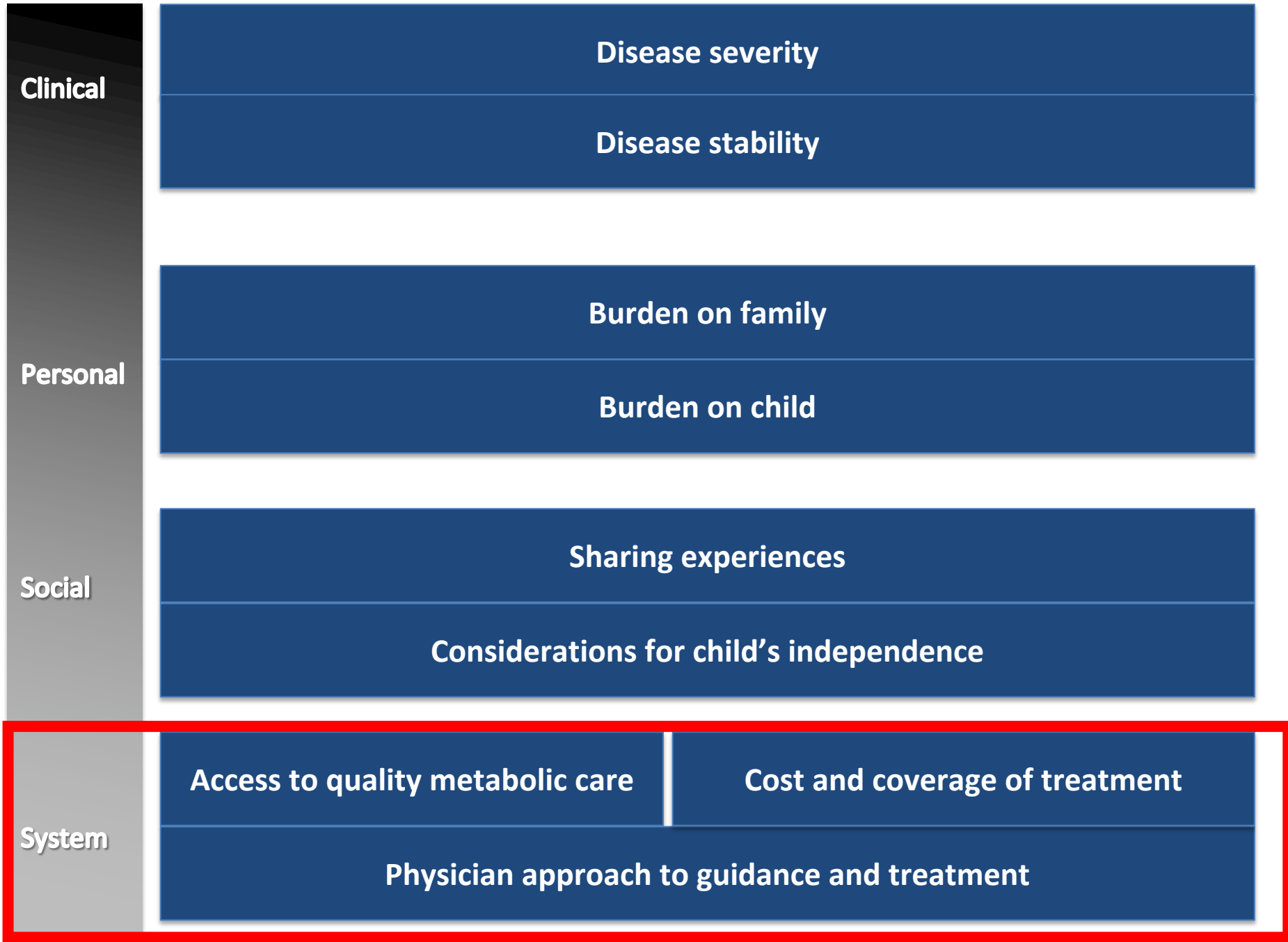
- Influence of other family experiences with MM and LT
- Motivated to transplant by:
  - Other parents' positive surgical experiences
  - Negative stories from other parents who had delayed or had foregone transplant
- Deterred from transplant/encouraged to continue MM by:
  - Stories of surgical complication
  - Families who have had success with MM

*“I probably wrote back and forth with about six, or seven, or eight different moms for a couple months. Just hearing their experiences, asking if they feel that dealing with transplant life is easier than a UCD life...A lot of their opinions really helped make my decision.”~ Parent*

# Consideration for Child's Independence

- Child's independence as a driver of treatment choice
  - Participation in preschool or grade-school programs
  - Living outside the home/attending college
- Living independently with UCD
  - Teaching child to manage their own medical needs
  - Living a safe and independent life with UCD outside the home

*“For her independence, a transplant is necessary...when her ammonia level starts to rise, she can’t make decisions on how to help herself...Living on her own and going away to college was not going to be an option.” ~Parent*



# Access to Quality Metabolic Care

- Concerns about local metabolic team and emergency medical management options
  - A consideration for transplant for some families
- Parents with confidence in metabolic team's ability to manage child's UCD often less motivated to explore alternatives like transplant



*“All these little things were adding up for us. Okay, he didn’t have a crisis for eight months, but they’re not able to quickly handle it...if we weren’t here watching like hawks all the time, would he survive?...When we went in there, my faith was that these people were going to save his life, and suddenly, I couldn’t count on them to do it right...” ~Parent*

*“I’m confident that if she does get sick that we’re in the right place. I trust the team here very much...The reason [liver transplant is] not forefront in my mind is because we are with [this] department, and we’re with [this] doctor.” ~Parent*

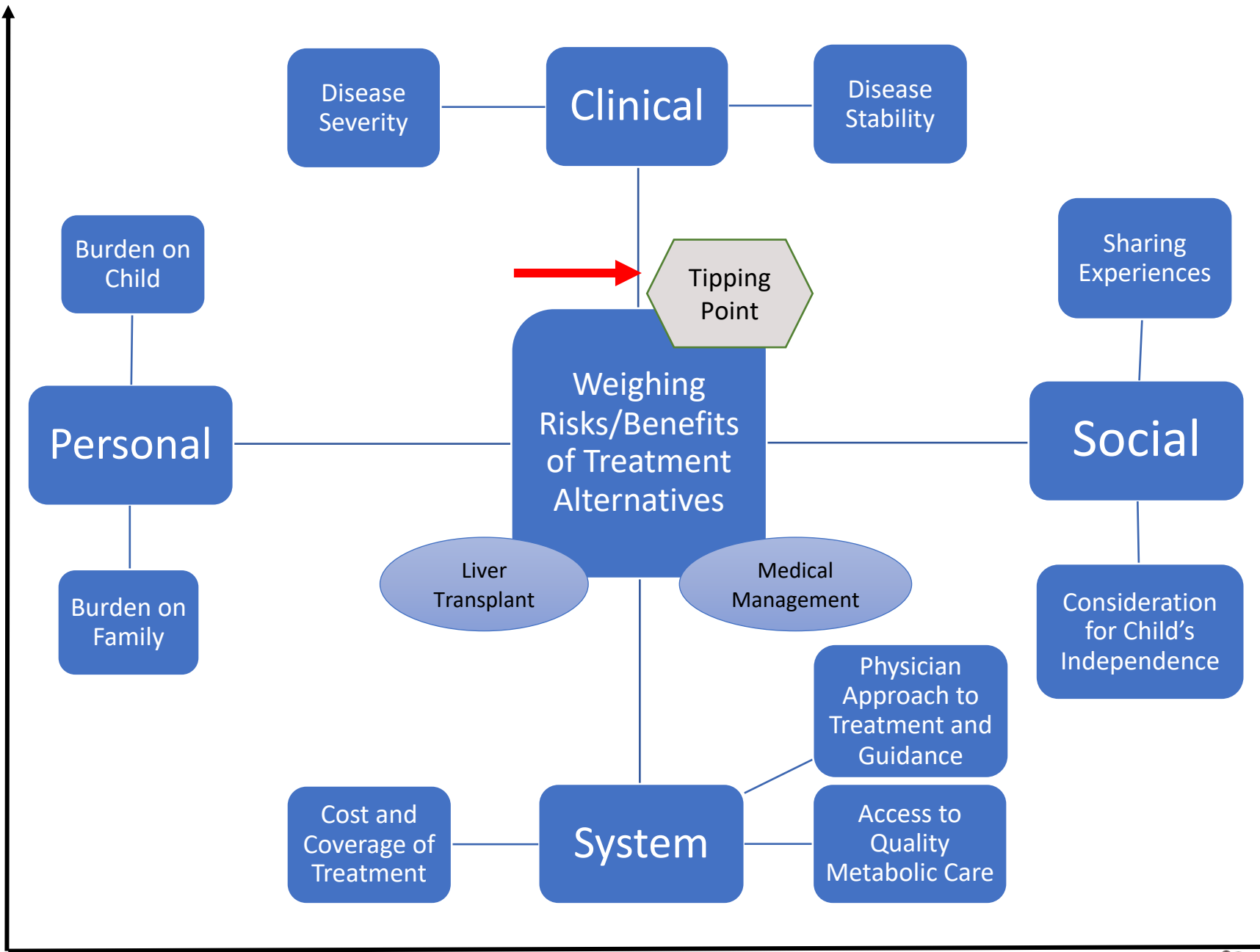
# Physician Approach to Treatment Guidance

- Varies from physician-to-physician and institution-to-institution
  - Pro-transplant
  - Transplant only for “severe” cases
  - Defer choice to family
- Physician opinion is a powerful driver of treatment choice

# Cost and Coverage of Treatment

- High costs of care and burden of navigating insurance coverage
- Differences in the cost and coverage of MM vs. LT
- MM: Disputes with insurance companies, limited coverage for metabolic formulas and medical foods
- LT often fully covered with little out of pocket cost to the family once authorized by insurer
- Financial burden of UCD contributes to overall burden of disease on family

*“It’s too hard...the struggle between insurance, and where you work, and getting covered...it’s a full-time job just to get medication...The nutrition many times is not considered medically necessary, but is medically necessary...Then you’re constantly battling with your insurance company. It’s hard enough emotionally to deal with the condition, but then to have to be bullied by insurance companies...The amount of money that we spent the first five years that [she] was diagnosed, we were living in poverty, but our income was well above poverty...we couldn’t comprehend how it could cost this much money out of our pocket to be able to keep her alive...” ~Patient*



# Implications for Practice

- Framework equips providers with evidence-based account of the patient experience
  - Adapt approach to treatment counseling
  - Anticipate patient needs and prepare for more productive patient interactions
- Framework could be considered in future guideline-development
  - Promote patient-centric approach to treatment counseling that accounts for evidence on patient and family experience

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ORIGINAL ARTICLE

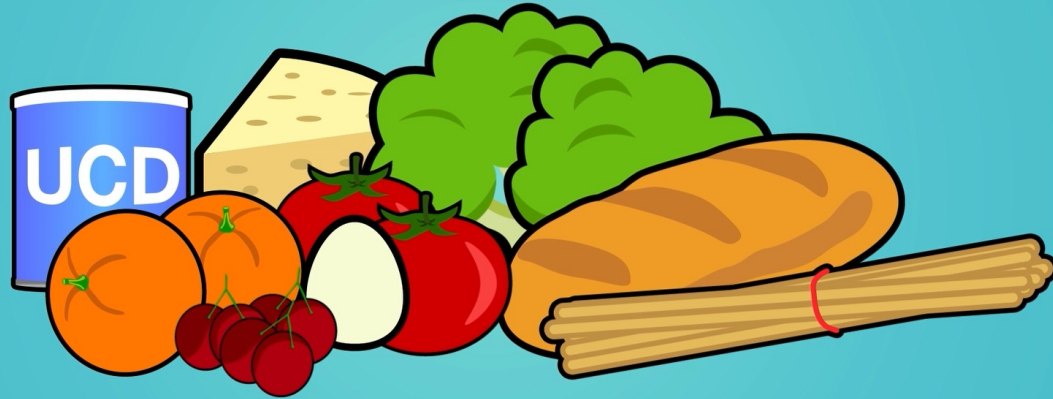


# Choosing between medical management and liver transplant in urea cycle disorders: A conceptual framework for parental treatment decision-making in rare disease

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<https://pubmed.ncbi.nlm.nih.gov/31883128/>

## MEDICAL TREATMENT OF UCD



Burden  
on Family



Daily burdens can be a compelling  
reason to consider transplant

- 24/7 medical caregiving
- Family's emotional health: fear, worry, anxiety
- Altered relationship to "normal life" comforts

Personal



## In our study, we also looked at:



UCD type



Highest  
ammonia  
level



Number of  
hospitalizations



Length of  
hospitalization



Apple  
(iPhone/iPad)



Android  
(e.g., Samsung)





## Thank You!

- GW Project Team
- NUCDF
- CNHS
- PCORI

THE FAMILIES WHO  
CONTRIBUTED  
THEIR TIME AND  
SHARED THEIR  
STORIES WITH US