# Rare Disease Research Agenda
## Shared in Rare Project

## Purpose and Methods

The Shared in Rare project is an effort to identify research priorities to address ethical and social concerns that are shared across patients with rare diseases and their families. The agenda was developed by Rare Voices, a group of parents of children with a rare disease, teens who have a rare disease or sibling with a rare disease, and clinicians and researchers who work in rare diseases. To develop the research agenda, Rare Voices:

1. Conducted listening sessions with members of the rare disease community.
2. Synthesized viewpoints from the listening sessions into core topics.
3. Reviewed the medical literature on each topic.
4. Developed research questions based on the knowledge gaps for each topic.
5. Ranked the research topics considering both importance and potential for improvements.



## Research Priorities

The research topics are listed and defined below in order of priority. We note that Rare Voices deemed all topics important as well as interconnected. Proposed research questions for each topic are given on the following pages.

**Coordinating Care**
Communication and collaboration among all stakeholders in a child's care to improve health care and patient and family experience.

**Communication**
Communication processes that support patients and families with rare disease in all settings.

**Accessing Resources and Care**
Challenges in accessing health care and resources due to systematic barriers.

**Impact on the Family Unit**
The impact of rare disease on the immediate family.

**Community and Support in Society**
Community that can support patients and families through their journey.

**Mental Health and Identity**
Challenges to and ways to support mental health and wellbeing in both patients and families.

**Ethical Aspects of Care**
Ethical concerns that arise in the course of care for rare disease.

**Uncertainty**
The causes and need to manage uncertainties experienced as a result of rare disease.

# Proposed Research Questions for Each Topic

| Topic | Proposed Research Questions |
|---|---|
| **Coordinating Care** | 1. What outcomes of care coordination are most important to patients and families?<br>2. Who is responsible for care coordination?<br>   a. with internal providers?<br>   b. with external providers?<br>3. How do patient and disease characteristics impact needs and outcomes of care coordination?<br>4. What skill sets and values are required to be a care coordinator?<br>   a. perspectives of providers and parents<br>5. What care coordination activities are needed in different settings and with varying patient needs?<br>   a. age, medical needs, etc.<br>6. What care coordination models and strategies most improve outcomes and relieve burdens for patients and families?<br>7. How can options and awareness of ways to sustainably pay for care coordination be raised? |
| **Communication** | 1. What interventions/systems can remove barriers and promote communication between health care providers and community resources such as service agencies and schools?<br>2. What resources or training can help health care providers and schools coordinate care and communication?<br>3. How can existing technologies be used more consistently and applied in new ways to improve effective communication?<br>   a. among health care providers and between health care providers and other community/service providers including schools<br>4. How do we change health care culture to help facilitate optimal communication among care teams, patients, and their families? |
| **Accessing Resources and Care** | 1. How can information about resources and services be more accessible and easily shared?<br>   a. resources and services include insurance, specialty providers, community services, financial resources, transportation services<br>2. What technologies can improve access to resources and care for families?<br>3. What technologies can improve access and delivery of care among providers between health care systems?<br>4. How can care and resources be provided in ways that minimize time, travel, and costs for patients and families? |
| **Impact on Family Unit** | 1. What helps families come together in response to a rare disease diagnosis and through evolving developmental stages?<br>2. How is resilience (as related to experienced trauma) developed and supported in families impacted by rare disease?<br>3. What strategies effectively support siblings of children with rare conditions? |

**Discovering Cures. Transforming Care.**

**childrensmercy.org/research**

# Proposed Research Questions for Each Topic: Continued...

| Topic | Proposed Research Questions |
|---|---|
| **Community and Support in Society** | 1. What needs to be present for children with rare diseases to have full and engaged experiences in their developmental/educational processes?<br>   a. in various settings including school, daycare, community, etc.<br>2. What is needed to facilitate or prepare individuals/organizations to provide appropriate accommodations for children with rare diseases?<br>   a. role of unconscious bias in receiving and the effectiveness of accommodations<br>3. What peer support activities or interventions improve health or psychosocial outcomes?<br>   a. role of information literacy in support group outcomes<br>4. How can we lower societal stigma associated with rare disease and increase acceptance of differences?<br>   a. tools for caregivers, advocates, places of work<br>5. How do we build empathy and advocacy in those not experiencing rare disease? |
| **Mental Health and Identity** | 1. How can mental health interventions be tailored to patients and families impacted by rare disease?<br>2. How can mental health be integrated into health care delivery to support wellbeing for patients and families with rare disease?<br>   a. training health care providers outside mental health specialties to address patient and family mental health needs<br>   b. creating a warm and welcoming environment to reduce stress and increase openness to mental health discussions<br>3. How can children and adolescents share information about their condition in ways that are comfortable and retain their identity outside the condition? |
| **Ethical Aspects of Care** | 1. How should quality of life be evaluated?<br>   a. inclusion, weighting, and balancing of patient, parent, and clinician views<br>   b. inclusion, weighting, and balancing aspects of quality of life<br>      i. biomedical, psychosocial, etc.<br>2. How do clinicians balance patient and parent empowerment against their perceived best interests for the child?<br>   a. removing barriers to parent empowerment and advocacy<br>   b. managing expectations around influence in decision making processes<br>3. What disparities exist in rare diseases care based on socioeconomic and cultural factors?<br>   a. family resources, education, language, religious/cultural views, etc. |
| **Uncertainty** | 1. In what ways do children and adolescents with rare diseases experience uncertainty?<br>2. How can health care providers effectively communicate uncertainty to patients and families?<br>3. What coping strategies are effective for patients and families to manage uncertainty and improve emotional wellbeing?<br>4. How can health care providers reduce uncertainty or help patients and families manage uncertainty? |

**Discovering Cures. Transforming Care.**

**childrensmercy.org/research**