



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 inter-connected. 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 healthcare 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 healthcare 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 well-being in both patients and families

Ethical Aspects of Care

ethical concerns arising 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	<ol style="list-style-type: none"> 1. What outcomes of care coordination are most important to patients and families? 2. Who is responsible for care coordination? <ol style="list-style-type: none"> a. with internal providers? b. with external providers? 3. How do patient and disease characteristics impact needs and outcomes of care coordination? 4. What skill sets and values are required to be a care coordinator? <ol style="list-style-type: none"> a. perspectives of providers and parents 5. What care coordination activities are needed in different settings and with varying patient needs? <ol style="list-style-type: none"> a. age, medical needs, etc. 6. What care coordination models and strategies most improve outcomes and relieve burdens for patients and families? 7. How can options and awareness of ways to sustainably pay for care coordination be raised?
Communication	<ol style="list-style-type: none"> 1. What interventions/systems can remove barriers and promote communication between healthcare providers and community resources such as service agencies and schools? 2. What resources or training can help healthcare providers and schools coordinate care and communication? 3. How can existing technologies be used more consistently and applied in new ways to improve effective communication? <ol style="list-style-type: none"> a. among healthcare providers and between healthcare providers and other community/service providers including schools 4. How do we change healthcare culture to help facilitate optimal communication among care teams, patients, and their families?
Accessing Resources and Care	<ol style="list-style-type: none"> 1. How can information about resources and services be more accessible and easily shared? <ol style="list-style-type: none"> a. resources and services include insurance, specialty providers, community services, financial resources, transportation services 2. What technologies can improve access to resources and care for families? 3. What technologies can improve access and delivery of care among providers between healthcare systems? 4. How can care and resources be provided in ways that minimize time, travel, and costs for patients and families?

Proposed Research Questions for Each Topic: Continued...

Topic	Proposed Research Questions
Impact on Family Unit	<ol style="list-style-type: none"> 1. What helps families come together in response to a rare disease diagnosis and through evolving developmental stages? 2. How is resilience (as related to experienced trauma) developed and supported in families impacted by rare disease? 3. What strategies effectively support siblings of children with rare conditions?
Community and Support in Society	<ol style="list-style-type: none"> 1. What needs to be present for children with rare diseases to have full and engaged experiences in their developmental/ educational processes? <ol style="list-style-type: none"> a. in various settings including school, daycare, community, etc. 2. What is needed to facilitate or prepare individuals/ organizations to provide appropriate accommodations for children with rare diseases? <ol style="list-style-type: none"> a. role of unconscious bias in receiving and the effectiveness of accommodations 3. What peer support activities or interventions improve health or psychosocial outcomes? <ol style="list-style-type: none"> a. role of information literacy in support group outcomes 4. How can we lower societal stigma associated with rare disease and increase acceptance of differences? <ol style="list-style-type: none"> a. tools for caregivers, advocates, places of work 5. How do we build empathy and advocacy in those not experiencing rare disease?
Mental Health and Identity	<ol style="list-style-type: none"> 1. How can mental health interventions be tailored to patients and families impacted by rare disease? 2. How can mental health be integrated into healthcare delivery to support well-being for patients and families with rare disease? <ol style="list-style-type: none"> a. training healthcare providers outside mental health specialties to address patient and family mental health needs b. creating a warm and welcoming environment to reduce stress and increase openness to mental health discussions 3. How can children and adolescents share information about their condition in ways that are comfortable and retain their identity outside the condition?

Proposed Research Questions for Each Topic: Continued...

Topic	Proposed Research Questions
Ethical Aspects of Care	<ol style="list-style-type: none"> 1. How should quality of life be evaluated? <ol style="list-style-type: none"> a. inclusion, weighting, and balancing of patient, parent, and clinician views b. inclusion, weighting, and balancing aspects of quality of life <ol style="list-style-type: none"> i. biomedical, psychosocial, etc. 2. How do clinicians balance patient and parent empowerment against their perceived best interests for the child? <ol style="list-style-type: none"> a. removing barriers to parent empowerment and advocacy b. managing expectations around influence in decision making processes 3. What disparities exist in rare diseases care based on socioeconomic and cultural factors? <ol style="list-style-type: none"> a. family resources, education, language, religious/cultural views, etc.
Uncertainty	<ol style="list-style-type: none"> 1. In what ways do children and adolescents with rare diseases experience uncertainty? 2. How can healthcare providers effectively communicate uncertainty to patients and families? 3. What coping strategies are effective for patients and families to manage uncertainty and improve emotional well-being? 4. How can healthcare providers reduce uncertainty or help patients and families manage uncertainty?

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