Child and Family Experiences with Inborn Errors of Metabolism: A Qualitative Interview Study with Representatives of Patient Groups

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on behalf of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN)

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Context & Background

The Canadian Inherited Metabolic Diseases Research Network (CIMDRN)

- Canadian Institutes of Health Research (CIHR)-funded multidisciplinary practice-based research network (core funding 2012-2017)
- Aims to develop evidence-informed approaches to health care for rare pediatric inborn errors of metabolism (IEM)
- For IEM and other rare diseases, increasing recognition of the need for evidence regarding the experiences and needs of patients and their families

Patient-Oriented Outcomes Research

- Patient-oriented outcomes research has emerged as a priority more broadly:
  - E.g., North American initiatives: Strategy for Patient Oriented Research (SPOR, Canada), Patient Centered Outcomes Research Institute (PCORI, US), Patient-Reported Measurement Information System (PROMIS, US)
- Increasing discussion in literature of related concepts: e.g., patient-informed care (Gardiner, 2008) personalized medicine (Hamburg, 2010)
- Particular relevance to rare genetic diseases: clinical heterogeneity is typical, value of interventions is sensitive to patient preference

Patient-Centered Outcomes Research Institute www.pcori.org
Strategy for Patient-Oriented Research www.cihr-irsc.gc.ca/e/41204.html
Gardiner R. (2008) In: Bos L., et al. (eds.) Amsterdam: IOS Press
Hamburg, MA et al. (2010) *N Engl J Med.* 363:301-304. DOI: 10.1056/NEJMp1006304

Objective

- In accordance with the recommendation of patient/family members of CIMDRN's external advisory board (EAB), we sought to interview IEM-specific patient group representatives:
  - to increase understanding of patient and family experiences, needs and priorities;
  - to inform patient-centered research and care for those living with IEM.

Methods

Eligibility

- **Groups:** Rare disease patient groups with a public Internet presence and a focus on one or more IEMs.
- **Participants:** Individuals employed by, or working on a volunteer basis for, an eligible group, and who held a spokesperson and/or representative role .

Sampling frame

- Developed using suggestions from study investigators; the Canadian Directory of Genetic Support Groups (2009) and; targeted internet searches.

Participant recruitment

- First contact through e-mail or electronic web forms from group web sites.
- Respondents provided signed, informed consent to participate
- Telephone interviews were scheduled, conducted and recorded

Data collection

- Interviews conducted by two members of CIMDRN's research staff
- Semi-structured interview guide developed using a scoping review of patient-oriented outcomes relevant to IEM (Khangura et al., 2015)
  - Topics included quality of life, physical, mental and social health, health care experiences, etc.
- Interviews were transcribed and verified

Qualitative descriptive analyses

- Primarily qualitative descriptive (Sandelowski, 2010).
- Broad categories identified from the first four interview transcripts
- Specific codes subsequently identified and applied using NVivo (version 10)
- Coding reviewed for credibility and trustworthiness by a second researcher
- Regular discussion regarding thematic saturation and further recruitment

Privacy, confidentiality and ethics

- Data privacy procedures developed to protect the confidentiality of participants and the children and families they described
- Protocol approved by the Ottawa Health Sciences Network Research Ethics Board (OHSN-REB)

Canadian Association of Genetic Counsellors. (2009). http://www.lhsc.on.ca/Patients_Families_Visitors/Genetic_Support_Directory/
Khangura, S.D., Karaceper, M.D., Trakadis, Y., Mitchell, J.J., Chakraborty, P., Tingley, K., et al. (2015). *BMC Pediatrics*, 15 (7) DOI: 10.1186/s12887-015-0323-x.
Sandelowski, M. (2010). Res Nurs Health, 33 (1). doi: 10.1002/nur.20362.

Results

Participants

- Eighteen group representatives provided signed, informed consent and participated
- Twelve participants self-identified as a parent of an affected child and/or as a patient

Interviews

- Conducted between February 28, 2014, and September 17, 2014, inclusive.
- Ranged in duration from 30 to 60 minutes

Diseases/categories represented (n if >1 groups)

- alpha-1 antitrypsin deficiency
- galactosemia
- leukodystrophies
- Pompe disease
- Fabry disease
- glucose transporter type 1 (Glut1) deficiency syndrome
- mitochondrial disorders
- porphyrias
- fatty oxidation disorders
- glycoprotein storage diseases
- mucopolysaccharidosis (MPS) (2)
- rare diseases (information specific to IEM was solicited)
- Gaucher disease
- inborn errors of metabolism (general)
- phenylketonuria (PKU) (2)
- Wilson disease

Perspectives of IEM Patient Group Representatives on Child & Family Experiences

Three Overarching Themes

Despite significant heterogeneity within and across diseases and their manifestations, we identified three overarching themes that bore relevance to all of the experiences, needs and priorities described in the interviews:

Coping with uncertainty and the unknown

Example quotes

Dealing with the unknown

Participant (07): *"… I think something that exists with all of us is the unknown. I would think that that's probably one of the largest things that impact us, is the unknown."*

Uncertainty around the diagnosis

Participant (03): *"So, you know there's the challenges once you get diagnosed, but there's all those challenges before you were diagnosed, and you know, often people go through a couple of years and several specialists until they get the diagnosis."*

Unknowns about the child's prognosis and future

Participant (16): *"A lot of people want to know like, 'When is, you know, when is my child gonna die?'… And just the unknowns about that."*

Uncertainty among health care providers

Participant (09): *"… sometimes the local GP is not understanding about what's going on and admitting quite openly that they are unable really to shed any light on what is happening at the moment…"*

Strategies for coping with uncertainty

Participant (08): *"I say, you know, 'Get a plan with your pediatrician or your metabolic specialist. Go to the local hospital, talk to the ER manager ahead of time before you run into a situation. Leave them your protocol so they can flag your information'."*

Managing the child's major life transitions

Example quotes

Early childcare and development

Participant (08): *"… when you're also dealing with like daycare providers – and they've got a lot of kids they're dealing with – and how do you make sure that your child is getting what he or she needs at the daycare?"*

Transition to school

Participant (15): *"...often* [the symptoms] *our children experience are difficult for others to detect. So that's always a concern for parents... especially when someone else is filling that caregiver role you know either at school or the parents are at work and things like that."*

Transition through adolescence

Participant (12): *"I know back when they were 9 or 10 they wanted to play baseball;* [the affected participant's son was] *not playing baseball. And they're at the age where they talk really quick and...* [he's] *in a wheelchair now... they're always incredibly friendly but there's nobody who would come over and watch a movie with him."*

Participant (13): *"… you know a lot of the children… are now, as it were, the oldest children who've ever lived with these conditions… So, there's no experience of how you provide services and support for people in this position; essentially you're making it up as you go along…"*

Struggle for improved outcomes & interventions

Example quotes

Disease communities: challenges and successes

"Participant (01*): "So there's no treatment. So the challenge for these people, first of all you know, to get a diagnosis. Second of all, to get a physician who is informed about it... we have… four or five physicians that would see patients with* [the disease]."

Participant (18): *"So yeah, I just think that right now the* [disease name] *atmosphere is very, very positive… I'd probably say, it's probably more positive than negative out there!* [laugh]"

Role of the information age

Participant (16*): "Yeah, and things now with social media are so different than they were…* [when] *you couldn't Google it. You had to get this big medical textbook out. But now families are… finding families on Facebook before they're finding the* [rare disease patient group]."

Suggestions for improved health care

Participant (03): *"… I think everybody who has a child with a complex medical condition like this would benefit from having more coordination. You know, being able to go into a clinic and see several specialists in one day, for example those kinds of things… the burden of getting to all these appointments is huge."*

Strengths, Limitations & Conclusions

Strengths

A broad range of experiences from multiple children with many different IEMs and their families were represented and described.

Limitations

We sought proxy reports of child and family experiences; the experiences of those not involved with patient groups were not represented.

Conclusions

Health care providers and decision-makers can support children with IEM and their families by partnering with the children and families to help reduce uncertainty, support families as they manage their affected child's life transitions, and contribute toward the collective struggle that IEM disease communities navigate toward improved outcomes and interventions.

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