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

Objective

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
    Participants (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
    Participants (03): “So, you know there’s the challenges once you get diagnosed, but there’s 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
    Participants (16): “A lot of people want to know, ‘When is, you know, when is my child gonna die?’… And just the unknowns about that.”

- Managing the child’s major life transitions
  - Example quotes
  - Early childcare and development
    Participant (08): “…when you’re also dealing with 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): “…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.”

- 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.