Health-Related Quality of Life Among Children with Genetic Conditions and their Families: A Scoping Review

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INTRODUCTION

- Measuring health-related quality of life (HRQoL) is key to patient-centered value assessment of genomic medicine interventions, including genomic sequencing and gene-based therapies.
- HRQoL has not yet been systematically assessed across genetic conditions that manifest in childhood.

METHODS

Systematically searched PubMed, EMBASE, and grey literature for original research articles published since Jan. 2010 that reported primary data on:

1. Pediatric HRQoL for patients (<18 years of age) with pediatric-onset genetic conditions;
2. HRQoL in family members and caregivers of pediatric patients with genetic conditions;
3. Family well-being in families affected by pediatric genetic conditions.

INCLUDED CONDITIONS:

- Conditions assigned a Mendelian Inheritance in Man (MIM) number (#, phenotype description with molecular basis known)

EXCLUDED CONDITIONS:

- Multifactorial conditions
- Adult-onset conditions
- Hereditary cancer syndromes that increase risk for typically adult-onset cancers

OBJECTIVE

To describe the empirical literature on HRQoL of pediatric patients with genetic conditions and HRQoL and well-being of their family members.

OUTCOMES OF INTEREST

Instruments used to measure HRQoL in pediatric patients and their family members; other family well-being measures; thematic domains in qualitative studies; areas with gaps in evidence on HRQoL to inform future research

RESULTS

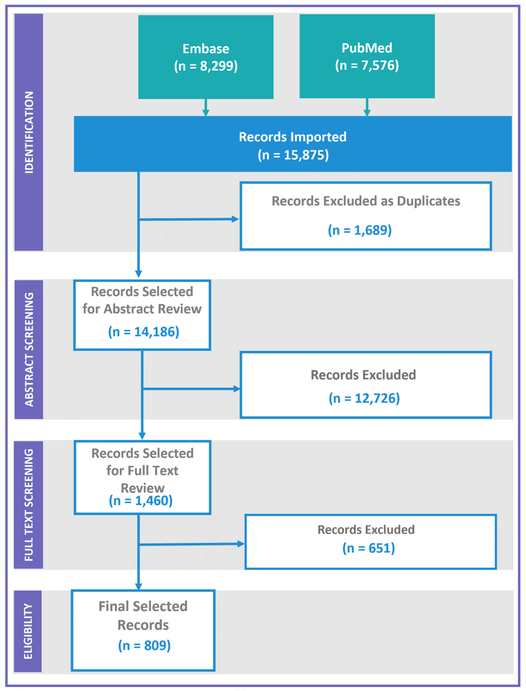

Figure 1. PRISMA diagram

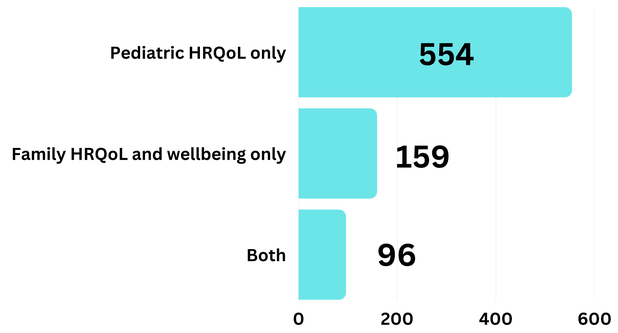

Figure 2. Topic of included articles

- Most research was led in the US (33%) or the United Kingdom (7%)
- The majority of pediatric HRQoL studies (64%) employed a cross-sectional survey study design; 16% reported data from interventional trials

- The Pediatric Quality of Life Inventory (PedsQL) was the most frequently used instrument (30%) to assess pediatric HRQoL.
- The SF-36 was the most frequently used instrument (19%) to measure HRQoL in family members.

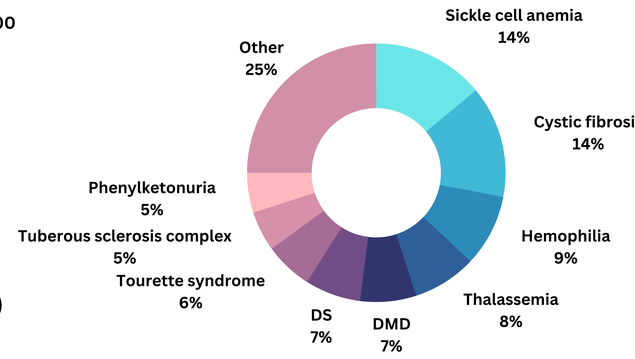

Figure 3. Conditions studied in included articles
DS, Down syndrome; DMD, Duchenne muscular dystrophy

CONCLUSION

While the heterogeneity of conditions and approaches to HRQoL assessment pose challenges for comprehensive evaluations of genomic medicine interventions, there is a growing body of empirical literature on HRQoL and well-being across a wide range of pediatric genetic conditions.

LEARN MORE

This review is registered on OSF (https://osf.io/sxk8u/).

SCAN ME

ACKNOWLEDGEMENTS

Funding: K99HG011491; U24HD107562

This project was funded in part by the Family Well-Being Research Network, a collaboration among the University of Michigan, the University of Pittsburgh, and The Harvard TH Chan School of Public Health, with funding from the Eunice Kennedy Shriver National Institute of Child Health and Human Development cooperative agreement.

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