

Impact of Raising Children With Rare Diseases on the Family: A Mixed-Methods Study

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PURPOSE: This study investigated how a child's functional ability, family cohesion, and healthcare satisfaction impact quality of life and family functioning of parents raising children with Barth syndrome (BTHS) or congenital muscular dystrophy (CMD).

DESIGN: A mixed-method design was used in this study.

METHODS: Forty parents of children with BTHS or CMD and forty parents of age-matched unaffected children participated in this study. Both groups of parents completed questionnaires providing information on their child's functional ability, family cohesion, healthcare satisfaction, parental quality of life, and family functioning. Following the completion of the questionnaires, in-depth qualitative interviews were conducted with two parents of the rare disease group. Statistical analyses were performed using general linear models to model parental quality of life and family functioning as an outcome of a child's disability. The audio-recorded qualitative interviews were transcribed verbatim and analyzed using thematic analysis.

RESULTS: Multivariable general linear model results for modeling of parental quality of life and family functioning of parents in the rare disease group was significantly lower than those in the unaffected group (-13.79 , 95% CI: $[-23.82, -3.75]$, $p = 0.0078$). Following the coding process, four main themes emerged as the primary descriptors of the parents' experiences and needs of healthcare service use: 1) difficulty getting a diagnosis 2) difficulty using healthcare services, 3) support and use of support networks, and 4) need for services.

CONCLUSIONS: This study contributed to laying the groundwork for understanding the experiences of parents raising children with rare diseases.

IMPACT STATEMENT: The findings of this study provide evidence for the need to develop effective strategies to support positive family functioning and well-being for families of children with rare diseases in the field of occupational therapy.

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