

**For Immediate Release**

**October 12, 2017**

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**Focus group captures rare disease caregiver experience, quality of life**

In a unique collaboration between the National Institutes of Health, Global Genes, Montclair State University, and the Global Foundation for Peroxisomal Disorders, researchers report preliminary results for one of the first qualitative studies examining quality of life, unmet needs, and emotional experience among caregivers for children diagnosed with Zellweger spectrum disorder (ZSD), a rare, life-limiting metabolic illness that affects 1 in 50,000 individuals. Currently, all published research in this field focuses on understanding the experience of the patient. This is the first study that evaluates the impact of ZSD on the whole family.

As there is no previously published literature studying ZSD caregivers and their experience, a qualitative research approach was used. Researchers conducted three focus groups for ZSD caregivers at the bi-annual family conference for the Global Foundation for Peroxisomal Disorders, a patient advocacy group for ZSD and related disorders.

Preliminary results found many caregivers associated their emotional experience with negative sentiments, including fear, devastation, isolation, and feeling overwhelmed. These feelings were triggered primarily by the progressive nature of the condition and uncertainty of its prognosis, the perceived lack of interest in the condition by the medical and scientific research community, and by unsatisfactory experience with medical and other institutional providers. Nonetheless, caregivers did report positive emotions associated with love, appreciation, joy and peace. This positive experience stemmed mainly from caregivers' personal connection with their child, diagnosis verification after a prolonged diagnostic pursuit, and from the availability of community support. Many caregivers described their emotional experience as a "roller coaster", illustrating the fluctuation of extreme emotions as a result of caregiving. Overall, the emotional experience of the ZSD caregiver had a considerable impact on quality of life, and there were numerous influencers on the emotional experience.

This is the first study to capture the emotional experience of the ZSD caregiver and relate it back to the factors that influence that experience as well as overall quality of life. This study has multiple implications. First, this study will be a starting point for future longitudinal studies that characterizes the changes in quality of life and emotional experience over time and identifies what contributes to those changes. Next, this study shows the utility of a focus group model in underscoring the importance of community guidance, insight, and opinion to better understand the needs in the broader rare disease community. Finally, the combined results of this and other related studies will serve as the platform for driving policy change to better serve the rare disease community as a whole.

*This study was supported by a grant from Global Genes and a Training and Career Development Award from Sterol & Isoprenoid Research Consortium (STAIR) of the Rare Disease Clinical Research Network (NIH/NICHD/NCATS).*