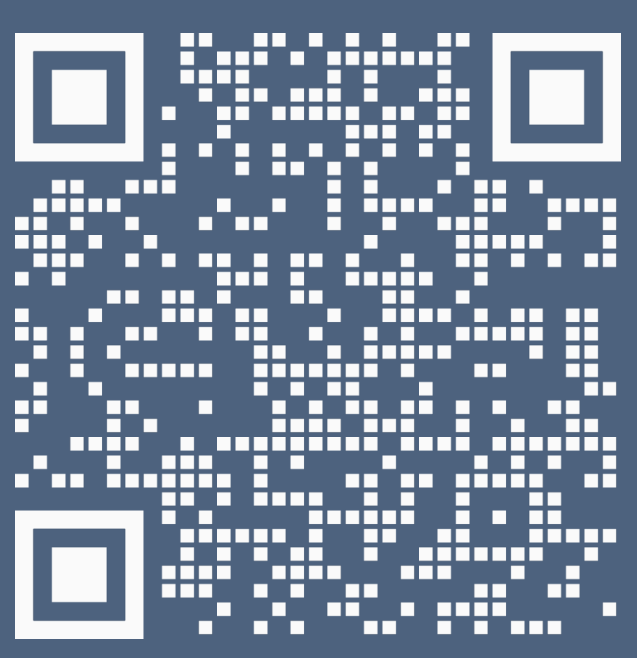


# SLEEP DISTURBANCE IS AN IMPORTANT FEATURE OF NEUROLOGICAL LYSOSOMAL DISEASE: FOR PATIENTS AND FAMILIES

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## Introduction

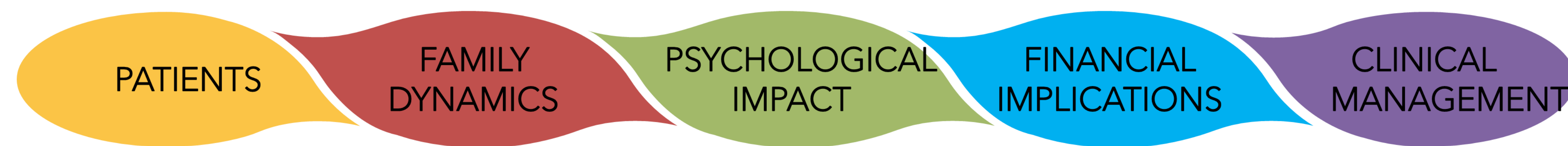
Dealing with a sick child is something that all parents will experience but the disturbance caused by dealing with symptoms is generally a short-term inconvenience.

Families living with Lysosomal Diseases with Neurological phenotypes often have to manage lifelong significant symptomatology that has consequences for both the patient and the family.

Focus Groups discussing the consequences of Niemann-Pick C and the GM Gangliosidoses were conducted to identify the most important symptoms impacting the QoL of these patients, Sleep disturbance was noted as a significant factor consistently affecting most patients AND their Families.

## Altered Sleep Patterns Implications

The impact of Gangliosidosis, such as GM1 and GM2, on sleep quality within a family is multifaceted and can significantly affect the well-being of both the affected individual and their caregivers. The following aspects illustrate the complex nature of this impact:



### Fragmented Sleep Patterns:

Neurological symptoms associated with GM, often lead to fragmented sleep. Frequent awakenings, difficulty in falling asleep, and restlessness can contribute to a disrupted sleep cycle.

### Caregiver sleep deprivation:

Attending to the needs of the affected individual during the night can result in sleep deprivation for caregivers, affecting their physical and mental health.

### Impact on sibling relationships:

Sleep disruptions may affect siblings' sleep routines and can potentially strain sibling relationships.

### Employment and financial strain:

Caregivers may face challenges in maintaining employment due to sleep disruptions and the caregiving demands associated with gangliosidosis. This can result in financial strain for the family.

### Effect of medications

Symptomatic medications may have side effects that impact sleep quality. Balancing benefits and disturbances is a crucial consideration.

## Assessment of Sleep

Assessing sleep disturbances in childhood neurological diseases, such as gangliosidosis, involves a comprehensive and multidimensional approach. Here is a general methodology that can be adapted based on the specific characteristics of the neurological disease under investigation:

<b>ANAMNESIS</b> Detailed interviews to gather information about sleep patterns. Explore any observed behaviors during sleep.	<b>SLEEP DIARIES</b> Daily record about child's sleep habits. Useful for tracking changes over time and potential triggers for sleep disturbances.	<b>ACTIGRAPHY</b> Non-invasive method to monitor sleep-wake patterns over an extended period. Provides objective data.
<b>POLYSOMNOGRAPHY</b> Overnight study in a sleep laboratory. Monitors various physiological parameters during sleep.	<b>MONITORING SEIZURE ACTIVITY</b> Implement methods to assess seizure activity during sleep. EEG may be valuable for identifying nocturnal seizures.	<b>QUESTIONNAIRES &amp; RATING SCALES</b> Validated sleep questionnaires and rating scales to assess specific aspects of sleep quality, behaviors, and potential sleep disorders.

## Patient Voice

Comprehending and mitigating the influence of NPC, GM1 and GM2 on sleep quality within the family structure is imperative for delivering holistic care. In addition to its direct impact on the affected individual, suboptimal sleep quality reverberates throughout the broader familial context. Insights gleaned from focus groups highlight the discernible detriment of inadequate sleep on the mental well-being of parents, accentuating the fatigue experienced in their caregiving role through persistent or disrupted sleep patterns

## References

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## Discussion

While the multifactorial nature of sleep quality and quantity precludes their direct utilization as clinical endpoints in studies, the evaluation of these dimensions in both the afflicted child and their family remains integral to comprehensive management in families affected by NPC, GM1 and GM2. Additional research is warranted to elucidate potential repercussions of sleep disturbances on both the patient and the parent/caregiver within the context of the diseases, thereby contributing to a deeper understanding of the overall impact on the affected individuals and their support networks.