Quality of life in patients with Niemann-Pick disease type C and their carers

Jackson Pountney - Health Psychology
PhD student at Aston University

BACKGROUND

Niemann-Pick Disease (NPC):
- NPC is a very rare lysosomal storage disorder which has an incidence rate of around 1 in 100,000
- Symptoms of NPC vary from person to person but can include: ataxia, vertical supraventricular gaze palsy, loss of motor function, psychiatric symptoms (psychosis, anxiety, depression, etc), respiratory dysfunction, hearing difficulties and many more [2,3]
- Clinical presentation and disease progression of NPC also vary greatly between a fatal condition presenting before birth (or early infancy) and a chronic neurodegenerative condition with onset occurring in adulthood [4]
- There are no curative treatments available for NPC; only Miglustat which can offer some improvement in neurological symptoms [5]
- The life-limiting and degenerative nature of NPC can negatively impact the quality of life (QoL) of both persons living with NPC and those that care for them [1]

Quality of life (QoL):
- QoL is one of the most important outcome variables in rare disease research
- Improving QoL is often the highest priority of patients living with a rare disease, as it is directly related to disease severity and can be deemed valid, i.e. capable of truly measuring QoL in NPC patients; this is called scale evaluation
- The endpoint of scale evaluation is ensuring questionnaires are reliable (able to generate consistent results) and valid (accurately measures the concept they claim to measure) in being able to assess QoL [6]

The Niemann-Pick disease type C Quality of Life Questionnaire (NPCQLQ):
- In 2019, the NPCQLQ was developed by Lydia Aston, Dr Rachel Shaw and Dr Rebecca Knibb at Aston University [7]
- Both adult and child versions of the NPCQLQ were developed
- However, these questionnaires have not yet been fully validated for wider use in the NPC population
- This means the questionnaires must first undergo rigorous statistical testing before they can be deemed valid, i.e., capable of truly measuring QoL in NPC patients; this is called scale evaluation
- The endpoint of scale evaluation is ensuring questionnaires are reliable (able to generate consistent results) and valid (accurately measures the concept they claim to measure) in being able to assess QoL [8]

WHAT THE PROJECT INVOLVES

To achieve our objectives, we will analyse patient-reported data collected by the International Niemann-Pick Disease Registry (INPD)
- Patient involvement in this stage of the research is crucial!
- NPC patients and/or their carers will be asked to complete questionnaires on the INPD patient-reported database
- Data from questionnaires concerning a variety of outcome variables will be used
- These include: patient QoL (general QoL and NPCQLQ), carer QoL, unmet needs, NPC disease/symptom severity, health service usage and more
- The NPCQLQ will undergo scale evaluation using patient-reported data from the INPD
- The reliability (consistency) and validity (accuracy) of the NPCQLQ will be tested using this patient-reported data
- Once the NPCQLQ has been validated, the patient-reported data will be used to identify other variables which have the most impact on the QoL of NPC patients, in both the short term and across time
- We can also repeat this process to look at carer QoL and QoL of NPC patients across different countries if we have enough data

BENEFITS TO NPC PATIENTS & THE COMMUNITY

- A fully validated NPCQLQ will contribute significantly towards providing patients and their carers with a much needed voice in expressing how Niemann-Pick disease affects their lives
- When validated, the NPCQLQ can be included as an endpoint in clinical trials - will be able to tell if potential new treatments in meaningful changes to QoL, experience of patients, i.e. beyond the improvement of symptoms
- This has implications for health technology assessment and drug reimbursement, drugs are more likely to be reimbursed if they are shown to improve QoL of NPC patients

REFERENCES