

Decentralised convenience: Digitised clinical assessment for impaired ambulation

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INTRODUCTION

Lysosomal storage disorders (LSDs) are characterised by their disruption of metabolic processes due to the build up of undigested substrate within the lysosome.

Some of the symptoms observed in patients impair ambulatory capabilities and general mobility across different types of LSDs such as sphingolipidoses

1. Acute bone pain¹
2. Ataxia¹⁻⁴
3. Frequent and uncontrolled muscle contractions¹
4. General neurological decline¹
5. Muscle atrophy¹
6. Neurological pain²
7. Seizures³⁻⁴
8. Spasticity²

It is important that healthcare approaches for LSD patients are not only clinically robust but are also considerate of the stresses they already face.

Digital, smartphone based technology can provide LSD patients with a more accommodating model of care through decentralised and remote monitoring.

METHODS

Completed clinical studies involved patient groups diagnosed with various sphingolipidoses.

SPHINGOLIPIDOSIS TYPE	PATIENT POPULATION
Gaucher disease (GD) (types 1 and neuronopathic (nGD))	Paediatric and adult
Late Onset Tay Sachs (LOTS)	Adult
Niemann Pick disease - type C (NP-C)	Paediatric

Table 1. Sphingolipidosis type and patient population involved in completed clinical studies

Ambulatory capabilities were assessed remotely with remote smartphone based technology using the following measurements:

AMBULATION MEASUREMENT	FORMULA
Average daily steps (ADS)	Total number of steps on active days / total number of days in a month
Average daily maximum (ADM)	Mean maximum number of steps per 30 minute epoch per day / active days in a month
Average daily steps per 30 minute epoch (ADE)	Total number of steps in active day / number of active epochs Mean steps per epoch / active days in a month
Average daily steps (ADS)	Total number of steps on active days / total number of days in a month

Table 2. Ambulation metrics collated by wearable device

The data collected from the ambulation measurements was compared against the conventional Six Minute Walk Test (6MWT) which patients had to complete.

RESULTS

The following was observed across studies:

- Most patients used the wearable device at baseline
- Longitudinal adherence to wearable device was highly variable across studies

SPHINGOLIPIDOSIS TYPE	% PATIENT USE OF WEARABLE DEVICE	PATIENT SAMPLE SIZE (n)
Gaucher disease - types 1	100	5
Neuronopathic Gaucher disease	62.5	16
Late Onset Tay Sachs (LOTS)	100	8
Niemann Pick disease - type C (NP-C)	50	8

Table 3. Proportion of patients who used wearable devices across sphingolipidoses studies

This approach allows for the collection of real world data which will provide a more thorough outlook on the condition of patients' through providing:

- Granular data sources with an accurate representation of patients' ambulatory capabilities
- Continuous longitudinal data capture: this may allow for tracking of disease progression or effect of a therapeutic intervention
- Patient specific data capture: as a result treatment can be tailored accordingly, moving away from the "one size fits all" model of care

Remote, decentralised clinical studies allow patients and carers to enrol and take part from the comfort of their own homes, consequently providing them with convenience and safety by:

- Offsetting the risk of inducing further harm to patients: traveling to a clinical centre may be emotionally and physically taxing, particularly for patients with impaired ambulation and mobility.
- Decentralised clinical assessment may also be more economical for patients and carers.

CONCLUSION

Completed clinical studies on sphingolipidoses have shown the viability of digital smartphone technology in aggregating useful data on ambulatory capabilities.

Data sets may prove to be useful in informing clinical practice and general treatment of LSD patients. The technology can provide patients and carers with convenience and safety which may also improve the efficiency of clinical studies. This can facilitate the adoption of more holistic, patient centric approaches within healthcare.

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