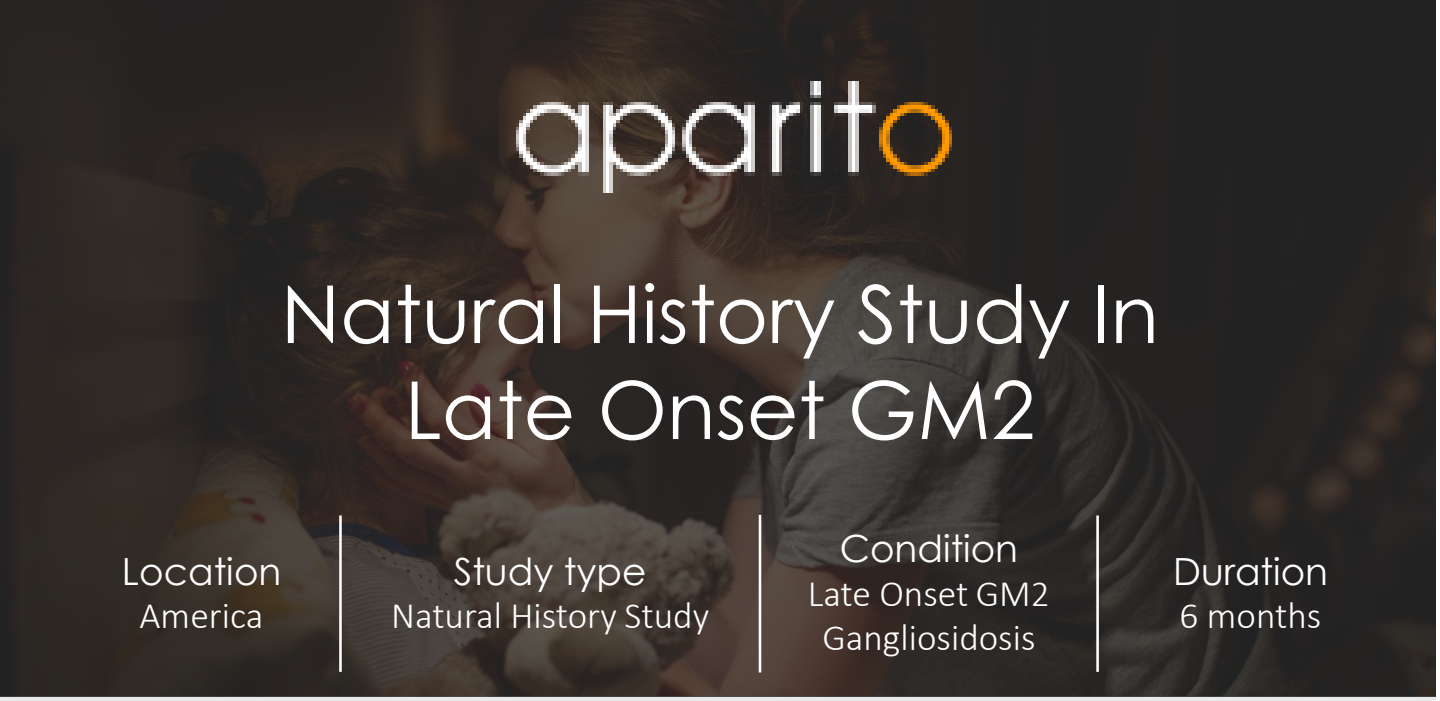

Natural History Study In Late Onset GM2

Location	Study type	Condition	Duration
America	Natural History Study	Late Onset GM2 Gangliosidosis	6 months

Overview and challenges

The GM2 gangliosidoses, Tay-Sachs (TSD) and Sandhoff (SD) diseases, are neurodegenerative disorders caused by a deficiency of the lysosomal enzyme beta-hexosaminidase A. The incidence of TSD is 1 in 320,000 and even less frequent for SD. The late-onset forms may present with ataxia, selective and progressive muscular atrophy leading to increased falls and difficulty rising from a chair or the floor, and for TSD patients, dysarthria. SD patients may have tingling, numbness or pain in their hands and feet as a presenting sign.

There are currently no disease-specific, validated tools for defining disease severity and reporting disease progression over time. **As new treatment options emerge, it is imperative to identify and validate appropriate outcome measures by which to evaluate potential therapeutic effects.**

Our approach

As part of a Natural History Study (NHS), 8 patients were recruited and monitored for 6 months, using digital health technology (including a wearable device and smartphone apps with mobile PROs) with continuous, remote monitoring and real-time capture of symptoms and **disease impact as perceived by the patients**.

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Results

Three male and 5 female patients, age 28-61 years, took part. Adherence rates for wearing the device and completing the PROs were 84% and 91% respectively. Correlations were calculated between the wearable metrics (average daily maximum (ADM), average daily steps (ADS), and average daily steps per 30-minute epoch (ADE)), the clinical parameters (6-minute walk test (6MWT), Brief Ataxia Rating Scale (BARS), and cadence from the GAITRite walking assessment) and the ten mPROs at baseline and at month 6.

Wearable metrics correlated with each other, showed positive correlation with the clinical walking assessments (6MWT and GAITRite cadence), and negative correlation with the Impact Scales. The clinical walking assessments also negatively correlated with the Impact Scales, but to a lesser extent than the wearable metrics. BARS did not show any correlation with 6MWT or GAITRite.

Conclusion

Patients were very enthusiastic and motivated to engage with the technology as demonstrated by excellent compliance. The combination of mPROs and wearables generated a feature-rich dataset, with **correlations between wearable metrics, clinical assessments and disease impact scales.**

Clinical measures did not always match patient self-perceived disease impact; for e.g. the patient with the highest reported score of Wider Impact and Tremor mPROs had one of the lowest disease impact BARS scores. However, the same patient reported the highest number of Events, and the highest number of healthcare visits. This shows that the **self-perceived impact of the disease is an important measure to consider in disease burden and may not correlate with clinical testing.**

This could be a useful and feasible way to capture remote, real-time insight into disease burden.

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