Tackling communication difficulties in Rett syndrome

Professor Jennifer McComas, Professor of Special Education and Educational Psychology at the University of Minnesota, is an expert in intellectual disabilities, currently focused on engaging hard-to-reach families using technology. Her latest project is an intervention trial to test the effectiveness of a training system that could revolutionise the way that young people with Rett syndrome communicate.

Prof McComas has recently launched a trial using telehealth to assess an intervention model to improve communication for people with RTT.

Losing the ability to communicate is a terrifying prospect, and yet communication is a key facet of many developmental conditions that is often overlooked and under-researched. For families of people living with Rett syndrome (RTT), communication problems have an immeasurable impact on quality of life but there remains a paucity of evidence-based guidelines for improving these skills.

RETT SYNDROME (RTT)
RTT is a genetic condition affecting 1 in 10,000 girls, most commonly caused by mutations in the gene MECP2, which is vital for normal brain function. It is an X-linked disorder, affecting girls and women almost exclusively. Girls born with RTT often have a period of several months with normal development, with difficulties becoming apparent later when the effects on brain function can be severe and widespread, impacting motor control and leading to seizures.

Although strides have been made to define the genetic basis of RTT, few studies have been funded or conducted that demonstrate effective interventions for positively affecting day-to-day life, despite the devastating consequences of the condition. Girls with RTT may lose the use of their hands as well as their ability to talk, making it extremely difficult for parents to understand and respond to their child’s wants and needs. These difficulties can be a source of profound distress and frustration and yet there are few well-established strategies for parents to adopt to help their children communicate.

Professor Jennifer McComas is at the forefront of complex behavioural research, specialising in developmental disorders. A key aspect of her work is telehealth, a field of healthcare which can be carried out remotely, linking patients to healthcare professionals in real time by video connection via the internet. Prof McComas has recently launched a trial using telehealth to assess an intervention model to improve communication for people with RTT.

DEVELOPING AN INTERVENTION MODEL
Designing an intervention trial in RTT is complex given the heterogeneity in how girls and women are affected by the syndrome – a one-size-fits-all approach is doomed to fail. Prof McComas’ current trial, funded by the National Institutes of Health (NIH), is USA-wide.
The innovative trial takes a tailored approach, which allows for the wide range of abilities and ages within the research participants. The starting point of the project is to characterise the personal behaviours each participant uses to try to make themselves heard, which might include head orientation, eye gaze or reaching for objects. Fundamental to this is the central hypothesis that girls and women with RTT have a preference (i.e. smiling, looking away). Results for the other items were less clear, suggesting they were neither most nor least preferred.

After becoming accustomed to these devices, researchers can then focus on training participants to use them as aids to make requests for highly preferred items or activities identified previously.

A vital outcome of the trial is the effect on quality of life, and data to assess this are being collected during the study. Prof McComas’ premise is that the functional training will have a noticeable positive impact on quality of life for participants.

Throughout the study, researchers work from a behaviour lab on the University campus and engage remotely via video conferencing with participants in their home, using observational techniques and formalised interviews with caregivers to assess behaviours.

A huge advantage of telehealth is that it negates the barriers posed by geography to allow engagement with rural families who might otherwise struggle to access services or take part in research. Overcoming these barriers to healthcare is particularly important for people with developmental disorders such as RTT, because people with disabilities are more likely to be socially marginalised. The approach also has the advantage of being less time intensive as it takes place in the home and reduces the delay in starting the intervention compared with traditional services. Preliminary data from the study indicate that the intervention coaching results in the participants’ reliable use of the communication devices and suggest that the telehealth approach is acceptable for caregivers and is an effective way to engage with families multiple times per week.

At the end of the trial, Prof McComas and her research team hope that they will have made a major contribution to the scientific understanding of the complex communication requirements of people with RTT, as well as a blueprint for a reliable intervention. In the meantime, their commitment to overcoming the difficulties posed by this under-studied syndrome will bring hope for many families living with RTT.