

Psychological Concomitants of Morquio A Syndrome - Longitudinal Effects of Enzyme Replacement Therapy (The MAPLE Study)

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Mucopolysaccharidosis IV, also known as MPS IV or Morquio disease, is a rare autosomal recessive genetic lysosomal storage disorder. Research thus far regarding Morquio, has primarily focused on the physical aspects of the various diseases. Less attention has been paid to the psychological toll of these diseases, whether they are direct symptoms or reactions to living with a chronic progressive disease.

Prior to 2013, there was neither a cure nor treatment (other than palliative) for Morquio disease. In the latter half of 2013, ERT became available to the broader population of patients with Morquio A disease through BioMarin's Expanded Access Program.

In a previous study, entitled "Psychological Concomitants of Morquio syndrome" the present investigator enrolled 20 adult subjects with Morquio into a pilot study to estimate a baseline incidence of psychological symptoms and overall quality of life. Subjects were all over the age of 18. Data from this study were published in 2015.

The present study extends this research into psychological health with Morquio via a comparison of psychological issues and quality of life before and after treatment (i.e. ERT). As ERT does not cross the blood-brain barrier, it would be unlikely to improve organic psychological symptoms, but may improve any reactive psychological symptoms caused by living over time with this chronic progressive genetic disease.

The present study thus seeks to follow adult patients with Morquio A disease as they begin ERT and track their psychological health every 6 months for a duration of 2 years. Adult patients with Morquio disease are invited to participate. Subjects will complete three different self-report questionnaires, the Achenbach System of Empirically Based Assessment (ASEBA) Adult Self-Report (ASR), the Short Form 36-item Health Questionnaire (SF-36), and the Brief Pain Inventory (BPI). Group aggregate data will be reported; individual questionnaire content and results will be held confidential, except as in accordance with Georgia law relating to reporting of child or elder abuse, suicidal and/or homicidal intent.