No Difference in Health Related Quality of Life between Therapeutic Options for Type 1 Gaucher Disease

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Type 1 Gaucher disease (GD) is the most common lysosomal storage disorder. Previously, treatment for GD was limited to intravenous enzyme replacement therapy (ERT). ERT reduces symptoms and increases health-related quality of life (HRQoL) in people with this condition. In 2014, oral substrate reduction therapy (SRT) was approved for type 1 GD treatment. Although both therapies alleviate disease symptoms, effects of SRT on HRQoL and preferences for therapy are not well established. Electronic surveys were administered to adults with type 1 GD. HRQoL was scored with the Short Form-36 Version 2 Health Survey and descriptive statistics were used to evaluate additional survey items. No differences in physical HRQoL ($p = 0.756$) or mental HRQoL ($p = 0.650$) were observed between SRT and ERT users. SRT users most often perceived their health to be similar to when they used ERT. Additionally, SRT users expressed convenience and non-invasiveness as reasons for choosing SRT, while many ERT users cited potential side effects and satisfaction with ERT as reasons for declining SRT. There appears to be no difference in HRQoL between ERT and SRT users and no perceived change in HRQoL for SRT users that previously used ERT. Participant responses illustrate that one particular treatment may not be ideal for all patients with type 1 GD depending on perceived convenience, invasiveness, or side effects. This evidence suggests that individuals with type 1 GD be adequately counseled about the risks and benefits of both therapy options now that SRT is clinically available.

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