

Quality of life in Brazilian patients with Gaucher disease and Mucopolysaccharidoses

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Introduction: Lysosomal diseases (LD) are rare genetic diseases caused by specific enzymatic deficiencies. There is growing interest in the literature to transform the concept of quality of life (QOL) in a quantitative measure that can be used in clinical trials and economic models, and in which the results are comparable between different populations or diseases. The SF-36 is a multidimensional questionnaire consisting of 36 items, comprising 8 scales or domains. It presents a final score from 0 to 100, in which zero represents the worst state of health and 100 the best. There are no Brazilian studies evaluating QOL in patients with LD. **Objectives:** 1) to assess the QOL of a sample of Brazilian patients with Gaucher disease (GD) and Mucopolysaccharidosis (MPS), through the application of the SF-36; 2) to compare the results between the two groups of diseases. **Material and Method:** The sample was of convenience. Patients over 12 years of age with GD and MPS, who were seen at the outpatient clinic for LD at the Medical Genetics Service, Hospital de Clínicas de Porto Alegre (HCPA), Brazil, and who are able to answer the SF-36 questionnaire, could be included in the study. The questionnaire was applied to patients at the time of their inclusion in the study (T1) and twelve months (T2) later. **Results:** Included 30 patients (GD=21 / GD Type I = 20, GD Type III = 1; MPS = 9 / MPS I = 3, MPS II = 2, MPS VI = 2, MPS IVA = 2). The mean age of patients was 23.1 years, 17 (56.6%) patients were male, 22 (73.3%) were on enzyme replacement therapy (ERT). The analysis presented is based on T1. The domain with the lowest mean for patients with GD was "the limit bound to physical appearance" = 61. For patients with MPS, the domain with the lowest mean was "vitality" = 45; of these patients, only the two patients with MPS IVA were not on ERT – their mean in the domain "general state of health" was = 37 (for patients on ERT, this value was = 49). Six patients with GD were not on ERT, being the domain "limitation of emotional aspects" (mean = 39) the domain with the lowest mean - for those on ERT, the value of this domain was = 68. **Conclusion:** This is the first time that the SF-36 is used in Brazilian patients with LD. The values found seem to be associated with the severity of the clinical disease analyzed. ERT appears to have improved the general state of health of patients. Further studies are required for these rare conditions. **Support:** FIPE / HCPA, MCT / CNPq / MSSCTIE-DECIT 37/2008 and 67/2009.