ENZYME REPLACEMENT THERAPY IN FABRY DISEASE: A REVIEW OF THE LITERATURE

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Abstract: Fabry disease (FD) is a storage disorder caused by deficiency in the production of the lysosomal enzyme alpha-galactosidase A (α-GAL), resulting in the accumulation of globotriaosylceramide (GL-3) in the lysosomes of endothelial cells of some organs, leading to several losses for the carrier individual. Thus, enzyme replacement therapy (ERT) enters as a specific therapy based on the incorporation of an enzyme from the extracellular environment, using it for its usual metabolism. It aims to analyze the effectiveness of ERT in Fabry disease, as well as its characteristics and methods. This is a literature review, from a bibliographic survey of studies published in Google academic, Scielo and PubMed being selected 5 articles pertinent to the theme. FD is considered a lissosomal storage disease and is an inborn error of glycosphingolipid metabolism resulting from mutations in the coding of α-GAL. Treatment, in turn, requires a multidisciplinary team, due to its multisystemic clinical presentation. Currently, two different forms of ERT are available: algasidase alpha, and algasidase beta, the former produced by human fibroblast culture and registered at a dose of 0.2 mg/kg biweekly, and the latter obtained by recombinant therapy of hamster ovaries at a dose of 1.0 mg/kg. In the overall context, clinical studies have shown a mild but significant long-term effect on the cardiovascular and renal system, considering the complication rate, which are mild and tolerable in most cases, most related to infusion. A study in which the outcome of the 10-year therapy with algasidase alfa was evaluated, showed that in 81% of the patients there were no serious clinical events during the treatment interval and 94% remain alive at the end of the study period. In addition, it was found that younger patients with less renal impairment benefited most from the therapy. Older patients with more renal impairment showed disease progression. Considering the facts analyzed, it can be concluded that ERT has proven to be a considerable form of treatment for FD and should be instituted early in order to enjoy more beneficial results. However, there are still doubts about its best regimen and limitations to be resolved, such as limited tissue penetration and the incongruence of biweekly intravenous administration during life, which will generate new optimistic fields of research in the future.

Keywords: Enzyme Replacement Therapy. Fabry Disease. Alpha-Galactosidase A Deficiency.

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