ENZYME REPLACEMENT THERAPY FOR STORAGE DISEASES: NEW THERAPEUTIC STRATEGIES FOR THE FABRY DISEASE.

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The project is aimed to develop new treatments based on enzyme replacement therapy for the Fabry disease. A rare X-linked disease caused by the lack of the lysosomal enzyme alpha-galactosidaseA and the progresive accumulation at the intracellular level of glycosphingolipids, particularly globotriaosylceramide (GL3). A wide variety of symptoms may characterize a Fabry patient being the liver, hart, brain and kidney the most clinically relevant altered organs. In this project we will produce the defective active enzyme by in vitro systems and different specific nanoconjugates targeted for intralysosomal delivery. Gene delivery approaches will be also considered. In vitro and in vivo models will be tested for efficacy and toxicity as well as in vivo biodistribution of designed therapeutic conjugates. Several multidisciplinary teams have been gathered to ensure the development of the project and an ambitious workplan has been set to obtain an early proof of concept.