EUCLYD- A European Consortium for Lysosomal Disorders

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The EUCLYD consortium aims at developing a scientific network among outstanding communities of basic and clinical investigators in five European countries (Italy, Netherlands, UK, Sweden and Germany) to study various aspects of lysosomal storage diseases (LSD). LSDs are rare disorders (overall prevalence of 1/5000 to 1/8000), each due to a specific lysosomal enzyme deficiency, leading to intracellular storage of a variety of undegraded substrates (sphingolipids, glycosaminoglycans, glycoproteins and glycogen) in different tissues. Among the 40 to 50 LSDs presently known, EUCLYD will focus on Gaucher disease, Pompe disease, MPS VI and Multiple sulfatase deficiency, as prototypes of disorders with different stored materials in various organs and tissues outside the CNS. The issues to be investigated in the proposed project are: pathophysiology and mechanisms underlying the symptoms and leading to devastating clinical consequences; natural history; testing of novel therapeutic approaches. These issues will be addressed by patients studies and with mouse models recapitulating the phenotype of LSDs. Recent advances in the pathophysiology of LSDs point to the role of secondary processes triggered by substrate storage. A better understanding of these processes is crucial to identify new targets of therapy. Enzyme replacement and substrate reduction therapies were successfully introduced in the treatment of specific LSDs. However, pilot clinical trials should be supported by post-marketing studies to further assess the efficacy of these therapies and to develop standardized treatment protocols. To this purpose a precise knowledge of the natural history of LSDs is essential. Novel therapeutic strategies, such as enzyme enhancement and gene therapy are still under investigation in cell systems and in animal models. EUCLYD will provide further testing of these approaches in order to translate state of the art knowledge into clinical applications and reach patient's bedside.

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Contract type Small or medium-scale focused research project