Several important diseases have been related with mutations that disrupt the normal splicing process leading to an abnormal and/or deficient protein production. Thus, correction of erroneous splicing is an important goal for which novel pharmacological and “molecular” therapies have begun to be designed and tested.

Lysosomal storage disorders (LSDs) are a group of inherited diseases that can result in severe and progressive pathology due to a specific lysosomal dysfunction. In large cohorts of patients with LSDs, several splicing mutations have been identified. Treatment strategies such as substrate reduction and enzyme-replacement therapy, among others, are available for conventional LSDs, yet still with some limitations. Therefore, for splicing mutations, splicing therapeutics is a potential alternative or an adjunct therapeutic strategy.

This study is aimed at identifying, firstly, potential splicing regulatory elements that are affected by a series of pathogenic mutations and next at developing therapies in order to restore the normal splicing of the pre-mRNAs involved.

Name of the disease(s), group(s) of diseases or gene(s) concerned by the project:
Mucopolysaccharidosis type II (IDS gene) ; Mucopolysaccharidosis type IIIC (HGSNAT gene)

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