New therapy against rare gene defects

April 15 2014, by Marc Aderghal

On 15th April is the 1st International Pompe Disease Day, a campaign to raise awareness of this rare but severe gene defect. Pompe Disease is only one of more than 40 metabolic disorders that mainly affect children under the age of 10, often with devastating consequences. Now scientists of the European research project EUCLYD setting up new therapeutic methods to tackle these gene defects.

European scientists set up new therapeutic approaches to tackle specific Lysosomal Storage Disorders: a new drug combination and enzyme replacement therapy against Pompe disease and gene therapy against MPS VI (Mucopolysaccharidosis VI). A new step to help young patients affected by these rare diseases and fight the feeling of isolation their parents often face.

The EUCLYD consortium focuses its activity on four specific Lysosomal Storage Disorders (LSDs): Gaucher disease, Pompe disease, Mucopolysaccharidosis VI (MPS VI) and Multiple Sulfatase Deficiency (MSD). Lysosomal Storage Disorders are rare genetic diseases; each of the four diseases studied in this project has a frequency of about 1 out of 50-100.000 newborns. LSDs mainly affect children in their first decade of life, often with devastating consequences. The issues investigated in the project include the mechanisms underlying the symptoms leading to devastating clinical consequences, the natural history of the diseases, and the testing of new therapeutic approaches.

The Euclyd consortium is made of scientific partners from Italy, Sweden, the United Kingdom, Germany and the Netherlands. Through

the scientific interactions between basic and clinical investigators, EUCLYD has advanced research on these four genetic diseases. New therapeutic approaches have been set up to tackle LSDs: a new drug combination and enzyme replacement therapy against Pompe disease and gene therapy against MPS VI.

The first two strategies have already entered clinical trials, whereas the last one is very close to this step. Positive results include a measured higher efficiency of the replacement enzyme in the Pompe patients, as well as visible improvement of the mobility of MPS VI affected animals.

More information: ec.europa.eu/research/health/m … jects/euclyd_en.html

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