Dipartimento di Biotecnologie e Bioscienze ed Istituto Ronzoni

mercoledì 19 settembre 2018, ore 11:00, aula U3-05, edificio U3

Chemical Tools for Mucopolysaccharidosis Therapeutic Development

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Mucopolysaccharidosis (MPS) disorders are a group of lysosomal storage diseases characterised by a deficiency or lack of the enzymes required to degrade glycosaminoglycans, including heparan sulfate (HS). This results in the accumulation of undegraded HS substrate within the lysosome, which leads to significant cell, tissue and organ damage with associated progressive physical and cognitive decline and a shortened lifespan. The affected organs include the heart, bones, joints, respiratory system and central nervous system. There are no cures for MPS disorders, although some treatments are available for a subset of these disorders, principally bone marrow transplant and enzyme replacement therapies. However, the latter are limited by their extraordinary cost and the fact that they are unable to cross the blood-brain barrier or penetrate into joints. In this presentation, I will describe our efforts to develop small molecule approaches to treatment: pharmacological chaperone therapy and substrate reduction therapy. I will also describe the development of a new LC-MS/MS assay for quantitation of HS in biological samples, which is useful for diagnosis and prognosis of MPS disorders and for monitoring the efficacy of new therapies.

Host: Prof. Francesco Peri, Dott. Marco Guerrini

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