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ICGEB International SEMINAR PROGRAMME 2018

Friday, 6 July 2018 | 12:00 noon | ICGEB Seminar Room, W building | Padriciano, 99, Trieste, ITALY



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Alternative splicing (AS) has emerged in the post-genomic era as one of the main drivers of proteome diversity with at least 94% of multi-exon genes being alternatively spliced in humans. AS is therefore one of the main control mechanisms for cell phenotype, and a process deregulated in disease. Numerous reports describe pathogenic mutations in splice factors, splice sites or regulatory sequences; additionally, in many cases there is an abnormal proportion of splice isoforms (or novel isoforms) in disease compared to the physiological pattern, without an apparent driver mutation. It has therefore become essential to study how AS is regulated in physiology, how it contributes to pathogenesis and whether we can manipulate faulty splicing for therapeutic advantage. While the disease most commonly linked to deregulation of AS in several genes is cancer, there are many in-depth reports of pathogenic splice variants in diseases ranging from neuromuscular disorders to diabetes or cardiomyopathies. In recent years, a plethora of splice variants have been implicated in chronic kidney diseases as well. Examples of these and ideas on how to manipulate them for therapeutic benefit will be presented in this talk.

“Alternative Splicing in Chronic Kidney Diseases”

Host: E. Buratti

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More information at:

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Open event - Free entrance

