

BIONAND 2017 CONFERENCE SERIES

Using “Precision Medicine” in skeletal dysplasias: from gene discovery to personalized treatments.

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Abstract:

Biomedicine is going through a scientific revolution thanks to the concept of Precision Medicine. New advances in genomic analysis allow the association of mutations and genetic variants with pathological conditions. This results in a better diagnosis and personalized treatment of diseases that have difficult diagnosis and prognosis, such as cancer and genetic syndromes. My research uses a multidisciplinary approach to study genetic conditions that affect the skeleton such as Osteogenesis Imperfecta, Achondroplasia and Jeune syndrome. It begins with gene discovery by exome analysis, followed by the identification of the mechanisms of disease at the tissue, cell and molecular levels; and concludes with the development of new patient-specific therapies.

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