Interactive clinical case discussions on skeletal dysplasias - Role of genetic testing in diagnosis

With Dr. Fatih Ezgü and Dr. John J. Mitchell

Wednesday, 4 September 2024

19:30 hrs: Start of buffet dinner

20:00 hrs: Start expert programme (45 min)

Hotel Vincci Porto – Room Bolsa 1+2 (ground floor)

Alameda de Basílio Teles 29

Porto, Portugal

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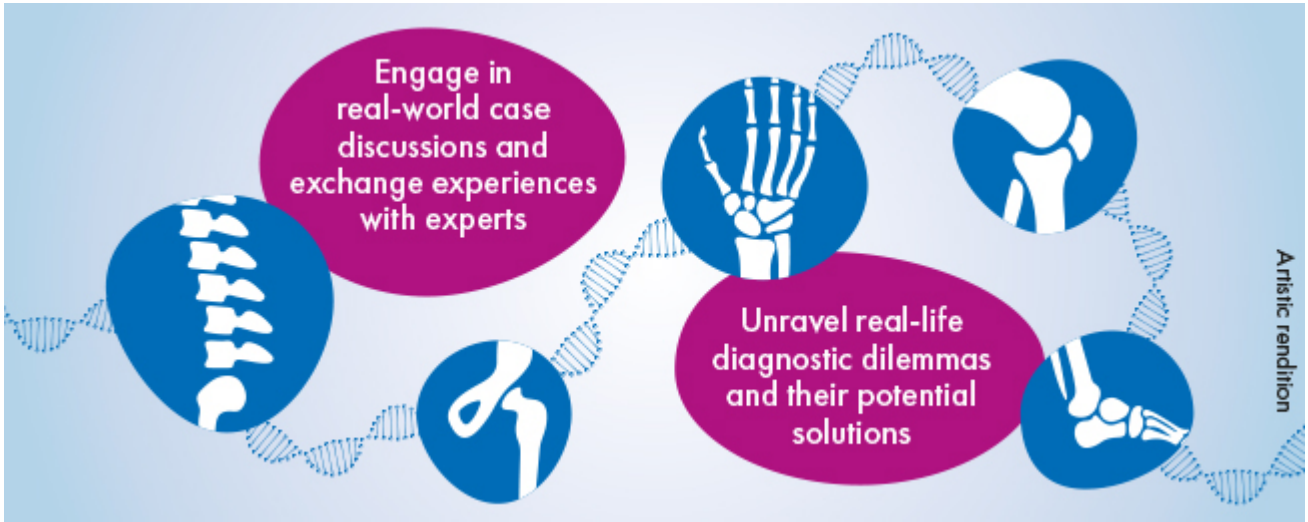

Fatih Ezgü, MD, PhD - Ankara, Türkiye

Prof. Fatih Ezgü is head of the Division of Paediatric Genetic Disorders at Gazi University, Ankara, Türkiye. He has worked in metabolic and genetic diseases for 30 years and has published over 125 articles in peer-reviewed journals. Prof. Ezgü

was trained at Ankara and Gazi University Hospitals in Türkiye as well as at the Cedars-Sinai Medical Center, Los Angeles, CA, USA, where the International Skeletal Dysplasia Registry was located. His primary interests are mucopolysaccharidoses (MPS) as well as skeletal dysplasias, with a focus on molecular diagnostics and novel treatments.

John J. Mitchell, MD - Montreal, QC, Canada

John J. Mitchell is Associate Professor at the McGill University, Montreal, QC, Canada. He practices as a Paediatric Endocrinologist and Biochemical Geneticist at the Montreal Children's Hospital. Prof. Mitchell's current research interests revolve around population screening, phenylketonuria, and lysosomal storage disorders. He also has a large MPS IV clinic and has been involved in the natural history study (MOR-001) and other clinical studies in MPS IVA. Prof. Mitchell has published numerous articles on inborn errors of metabolism. He has also been involved in national and international guideline development for the treatment of orphan diseases.

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