

BONE DYSPLASIAS

2026

SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

21 a 23 Maio

Comunicações Orais - Casuísticas e Casos Clínicos II | Oral Communications - Case Series and Clinical Cases II

Skeletal dysplasias with major epiphyseal involvement (total duration: 35 mins; discussion: 13 mins)

1. Clinical Spectrum and Therapeutic Challenges in Adult COMP-related Dysplasias (6 mins) - *Rodrigues, Mariana Mendes (Rheumatology Department, Unidade Local de Saúde de Coimbra (Coimbra Local Health Unit))*
2. Phenotypic Spectrum Associated with Variants in the COL2A1 Gene: a Retrospective Study of Cases Diagnosed at Two Hospitals (6 mins) - *Torres, Katerine (Serviço de Genética Médica, Departamento de Pediatria, Unidade Local de Saúde de Santa Maria)*
3. Type II Collagenopathy: The Milder end of the Spectrum (4 mins) - *Panoeiro, Jonathan (Serviço de Genética Médica, Centro de Genética Médica Dr. Jacinto Magalhães, Unidade Local de Saúde Santo António)*
4. Same Name, Different Disease: Genotype-Phenotype Correlations in Adult Patients with Multiple Epiphyseal Dysplasia in a Tertiary Centre (6 mins) - *Costa, Sara Alves (Rheumatology Department, Unidade Local de Saúde de Coimbra (Coimbra Local Health Unit))*

Discussion: 13 mins

Skeletal dysplasias in pregnancy (total duration: 15 mins)

5. Pregnancy in Women with Skeletal Dysplasia – Follow-up and Prognosis - Preliminary Findings (6 mins) - *Rodrigues da Silva, Concha (Instituto de Ciências Biomédicas Abel Salazar - UP)*
6. TANGO1 Loss-of-Function Underlies Prenatal Lethal Skeletal Dysplasia with Absent Bone Mineralisation (4 mins) - *Pinho, Sara (Serviço de Genética Médica, Departamento de Pediatria, Unidade Local de Saúde de Santa Maria)*

Discussion: 5 mins
