

Seminar

SKELETAL DYSPLASIAS

AZORES Portugal

09:00 **Opening**
HDES Board*
Directorate of Health*
Lucinda Pacheco (HDES Clinical Directorate)
Inês Alves (ANDO Portugal)

SKELETAL DYSPLASIAS: INTRODUCTION (moderated by Ana Luísa Rodrigues - HDES)

09:30 **Overview and Clinical Approach: CHUC Multidisciplinary Team**
Sérgio Sousa (CHUC)

09:50 **ANDO Portugal: Framework and Actions**
Inês Alves (ANDO Portugal)

10:05 **European Reference Network ERN BOND and National Members. CHULN Multidisciplinary Team**
André Travessa (CHULN)

10:25 **Prenatal Diagnosis and Coordination with Postnatal Care**
Fabiana Ramos (CHUC/HDES)

10:40 **BIOPAUSE**

MULTIDISCIPLINARY DIAGNOSIS AND FOLLOW-UP (moderated by Vânia Machado - HSEIT)

11:10 **Ciliopathies**
Nataliya Tkachenko (CHUdSA/HDES)

11:25 **Bone Fragility**
Cláudia Falcão Reis (CHUdSA/HSEIT)

11:40 **Achondroplasia**
Sérgio Sousa (CHUC)

11:55 **The Relevance of Radiology in Diagnosis**
Marta Silva (HDES)

*To be confirmed

CLINICAL CASES IN THE AZORES (moderated by Nataliya Tkachenko - CHUdSA/HDES)

12:15 **Presentation of Clinical Cases of Skeletal Dysplasia**
Case 1: ACAN MUTATION - André Travessa (CHULN)
Case 2: 3-M SYNDROME - Joana Franco (IFE MGF - USI)
Case 3: CAFFEY DISEASE - Joana Rodrigues (HDES)
Case 4: X-LINKED HYPOPHOSPHATEMIA - Maria Ponte (HDES)
Case 5: PSEUDOACHONDROPLASIA - João Calado (HDES)
Case 6: MELORHEOSTOSIS - Mariana Sebastião (HDES)
Case 7: OSTEOGENESIS IMPERFECTA - Daniela Valadão (USI Terceira)
Case 8: ACHONDROPLASIA - Nelson Meneses (HSEIT)

13:20 **LUNCH**

CLINICAL APPROACH IN INSULAR CONTEXT (moderated by Fabiana Ramos - CHUC/HDES)

14:30 **Cardiorespiratory Rehabilitation of Adults with Skeletal Dysplasia**
Tiago Teófilo (HNM-SESARAM)
Cláudia Aguiar (HNM-SESARAM)

14:50 **Multidisciplinary Insular Context in Rare Diseases**
Ana Luísa Rodrigues (HDES)

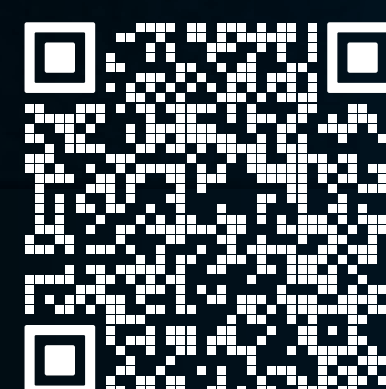
HEALTH PROFESSIONALS' PERSPECTIVE (moderated by Cláudia Falcão Reis - CHUdSA/HSEIT)

15:10 **Rare Bone Diseases in the Azores - A View from Orthopaedics**
Fernando Carneiro (HDES)

15:25 **Round Table**
Pedro Diogo Silva (HNM-SESARAM) - Physical Medicine and Rehabilitation
Ana Luísa Rodrigues (HDES) - Paediatrics
Luís Maurício Santos (HDES) - Rheumatology
Vânia Machado (HSEIT) - Nursing
Fernando Carneiro (HDES) - Orthopaedics
Ana Rodrigues (HSEIT/CHULN) - Rheumatology
Cláudia Aguiar (HNM-SESARAM) - Physiotherapy
Rui Amaral (HDES) - Radiology

16:40 **Closing Remarks**
Ana Luísa Rodrigues (HDES)

16:50 **NETWORKING COFFEE**



AÇORES
20. JAN. 2024
Hospital Divino Espírito Santo
Ponta Delgada
Inscrições em
www.andoportugal.org

