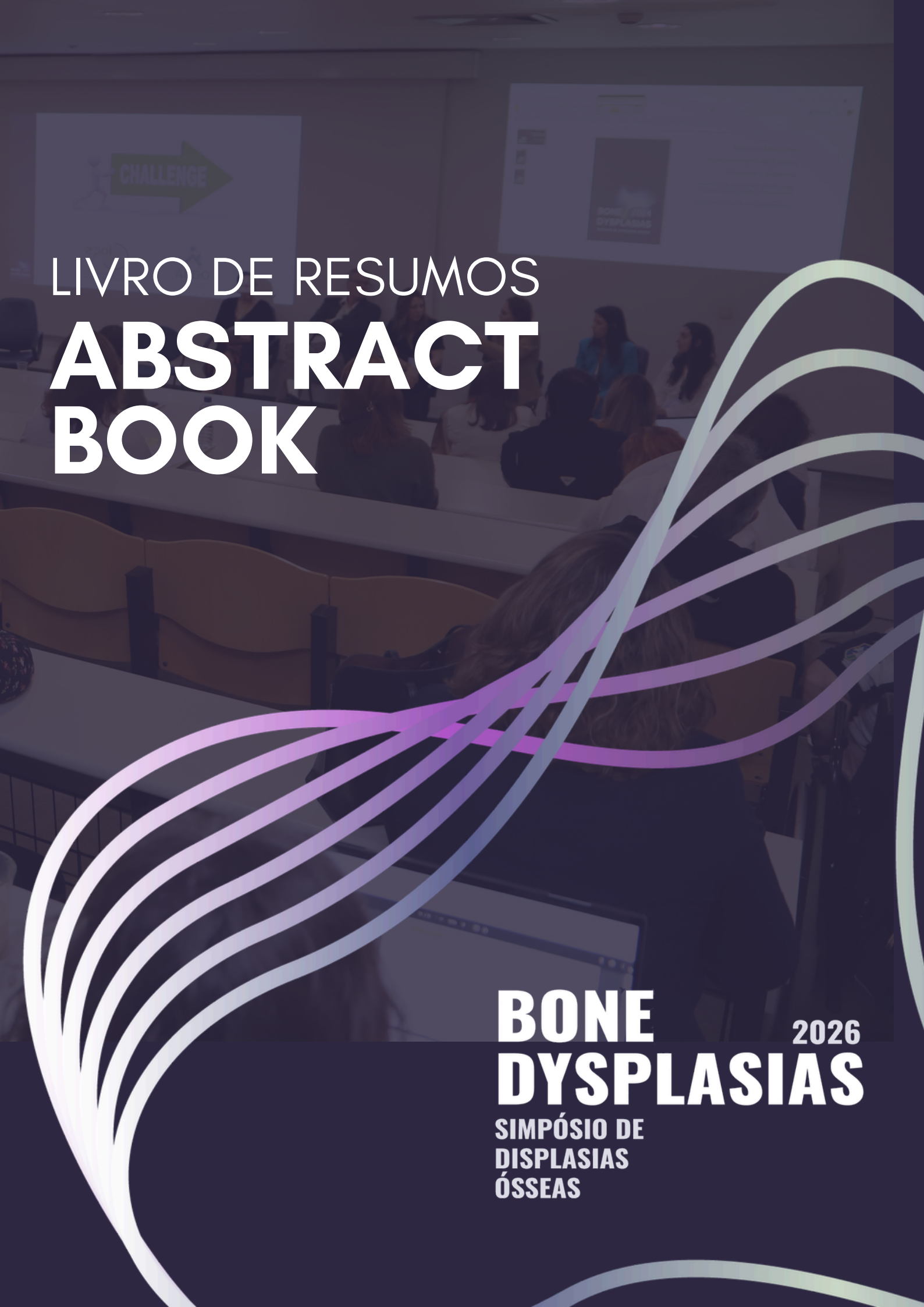

LIVRO DE RESUMOS
ABSTRACT BOOK

BONE DYSPLASIAS 2026

SIMPÓSIO DE DISPLASIAS ÓSSEAS

AS DISPLASIAS ÓSSEAS | *BONE DYSPLASIAS*

De origem genética, as **Displasias Ósseas** correspondem a um conjunto muito heterogéneo de doenças raras que afetam, maioritariamente, o desenvolvimento, a estrutura e a constituição dos ossos, da cartilagem e da dentina, refletindo-se em baixa estatura, fragilidade ou deformações ósseas e frequentemente comprometendo a vida e mobilidade das pessoas afetadas.

Estas patologias representam uma área extremamente importante no âmbito das doenças do osso, mas que tem sido pouco desenvolvida e reconhecida levando a falta de apoio e de implementação de medidas adequadas, particularmente significativa nos doentes adultos.

Nesse sentido, a **Associação Portuguesa de Osteogénese Imperfeita**, a **Equipa Multidisciplinar de Displasia Esquelética da ULS Santa Maria**, membro da **Rede Europeia de Referência sobre Doenças Ósseas Raras (ERN-BOND)**, e os seus Centros Afiliados (ULS São José, ULS Almada-Seixal e Hospital CUF Descobertas) unem esforços para implementar mais uma edição do projeto Aliança Inquebrável.

Click the Link!

Of genetic origin, Bone Dysplasias correspond to a very heterogeneous set of rare diseases that mostly affect the development, structure and constitution of bones, cartilage and dentin, resulting in short stature, fragility or bone deformations and often compromising the lives and mobility of those affected.

These pathologies represent an extremely important area within the scope of bone diseases, but have been poorly developed and recognized, leading to a lack of support and implementation of appropriate measures, particularly significant in adult patients.

*In this sense, the **Portuguese Association of Osteogenesis Imperfecta**, the **Multidisciplinary Skeletal Dysplasia Team of ULS Santa Maria**, member of the **European Reference Network on Rare Bone Diseases (ERN-BOND)**, and its Affiliated Centers (ULS São José, ULS Almada-Seixal, and Hospital CUF Descobertas) join forces to implement another edition of the **Unbreakable Alliance project**.*

Click the Link!

BONE DYSPLASIAS 2026

O Bone Dysplasias 2026: Simpósio de Displasias Ósseas assinala um marco significativo ao celebrarmos o 10.º aniversário da Aliança INquebrável, um projeto pioneiro e de continuidade da Associação Portuguesa de Osteogénese Imperfeita (**APOI**) que visa apoiar o diálogo e a cooperação nacional e internacional entre especialistas, doentes, familiares e outros stakeholders envolvidos na área das displasias ósseas.

A Conferência é organizada por uma Comissão Organizadora que reúne diversas entidades, entre as quais a Associação Portuguesa de Osteogénese Imperfeita (APOI) e a Equipa Multidisciplinar de Displasias Ósseas da ULS Santa Maria, membro da Rede Europeia de Referência em Doenças Ósseas Raras (ERN-BOND), com os seus centros afiliados, nomeadamente a ULS Almada-Seixal, a ULS São José e o Hospital CUF Descobertas.

Click the Link!

Com o objetivo de expandir e fortalecer esta Aliança INquebrável, este ano um novo parceiro, a Associação Nacional de Displasias Ósseas (ANDO Portugal), também foi convidado para a organização. Além disso, o Hospital CUF Descobertas desempenha um papel de destaque como anfitrião das atividades científicas, acolhendo especialistas nacionais e internacionais.

Para envolver toda a comunidade e estabelecer uma verdadeira Aliança Global de doenças esqueléticas raras sob a égide da Aliança INquebrável, muitos outros parceiros foram convidados a juntar-se a nós para este projeto que promoverá um espaço de oportunidades para especialistas e doentes se encontrarem, discutirem, trabalharem em conjunto, compartilharem experiências e desenvolverem estratégias-chave para implementar os novos avanços.

The Bone Dysplasias 2026: Bone Dysplasia Symposium marks a significant milestone as we celebrate the 10th anniversary of the UNbreakable Alliance, a pioneering and ongoing project of the Portuguese Association of Osteogenesis Imperfecta (APOI) that aims to support national and international dialogue and cooperation between specialists, patients, families, and other stakeholders involved in the field of bone dysplasias.

Click the Link!

The Conference is organized by an Organizing Committee that brings together several entities, including the Portuguese Association of Osteogenesis Imperfecta (APOI) and the Multidisciplinary Bone Dysplasia Team of ULS Santa Maria, member of the European Reference Network for Rare Bone Diseases (ERN-BOND), with its affiliated centers, namely ULS Almada-Seixal, ULS São José and Hospital CUF Descobertas.

With the aim of expanding and strengthening this Unbreakable Alliance, this year a new partner, the National Association of Bone Dysplasias (ANDO Portugal), was also invited to participate in the organization. Furthermore, CUF Descobertas Hospital plays a prominent role as host of the scientific activities, welcoming national and international experts.

To involve the entire community and establish a true Global Alliance for rare skeletal diseases under the aegis of the Unbreakable Alliance, many other partners have been invited to join us in this project, which will foster a space of opportunity for experts and patients to meet, discuss, work together, share experiences, and develop key strategies to implement new advances.

ALIANÇA *IN*QUEBRÁVEL | *UN*BREAKABLE ALLIANCE

O projeto "ALIANÇA INQUEBRÁVEL", criado em 2016 pela Associação Portuguesa de Osteogénese Imperfeita (APOI), nasce devido à consciencialização da associação para as dificuldades de acesso e igualdade a cuidados de saúde na Osteogénese Imperfeita (OI) e outras Doenças Esqueléticas Raras. A sua responsabilidade social e científica, bem como a necessidade emergente de iniciar medidas que tragam para a ribalta questões estratégicas subjacentes a estas comunidades - como sejam os Programas Específicos para as Doenças Raras, os Centros Especializados e as Redes Europeias de Referência, a Investigação e os Tratamentos, a Informação e Literacia e os Serviços Especializados - impelem a associação a recrutar outros parceiros para esta CAUSA, tendo as instituições hospitalares, as sociedades científicas, as associações de doentes, as instituições académicas, a indústria farmacêutica e de dispositivos médicos e os organismos oficiais um papel fundamental para a sensibilização dos seus associados e da sociedade em geral para estas temática.

Click the Link!

Nasce então este projeto de parceria inovadora, em que a APOI tem como objetivo principal e único a "melhoria da qualidade de vida aos portadores de OI e outras displasias ósseas", estando para isso subjacente o desenvolvimento de várias atividades de atualização científica, literacia em saúde e confraternização.

Um projeto que envolve a comunidade científica e não científica de Displasias Ósseas num propósito conjunto em que doentes e profissionais se reúnem para refletir sobre os últimos desenvolvimentos em doenças ósseas raras e definir melhores práticas para o futuro.

Click the Link!

The "UNBREAKABLE ALLIANCE" project, created in 2016 by the Portuguese Association of Osteogenesis Imperfecta (APOI), was born out of the association's awareness of the difficulties in accessing and achieving equal healthcare for Osteogenesis Imperfecta (OI) and other Rare Skeletal Diseases. Its social and scientific responsibility, as well as the emerging need to initiate measures that bring to the forefront strategic issues underlying these communities - such as Specific Programs for Rare Diseases, Specialized Centers and European Reference Networks, Research and Treatments, Information and Literacy, and Specialized Services - compel the association to recruit other partners for this CAUSE. Hospital institutions, scientific societies, patient associations, academic institutions, the pharmaceutical and medical device industry, and official bodies play a fundamental role in raising awareness among their members and society in general about these issues.

Thus, this innovative partnership project was born, in which APOI, whose main and sole objective is "improving the quality of life for those with OI and other bone dysplasias," is based on the development of various activities for scientific updating, health literacy, and social interaction.

A project involving the scientific and non-scientific community of Bone Dysplasias in a shared purpose where patients and professionals come together to reflect on the latest developments in rare bone diseases and define best practices for the future.

PARCERIAS E APOIOS | *PARTNERS AND SPONSORS*

Este é um Projeto Solidário que pretende envolver toda a comunidade e estabelecer uma verdadeira Aliança Global de doenças esqueléticas raras.
A nossa política é assegurar a participação de todos os interessados, nomeadamente doentes, famílias, estudantes e profissionais de todas as áreas da saúde. Para isso ser possível, agradecemos a toda a comunidade, muito em particular à indústria e empresas da sociedade civil, que se juntaram a nós contribuindo com bens, serviços ou apoios financeiros.

This is a Solidarity Project that intends to involve the whole community and establish a truly global Alliance for rare bone diseases.
Our policy is to ensure the participation of everyone that might be interested, namely patients, families, students, and health professionals of all kinds. For that to be possible we thank the whole community, in particular the industry and companies from the civil society that joined us by contribuiting with goods, services, or financial support.

PARCEIROS | *PARTNERS*

ALIANÇA INQUEBRÁVEL

apoi associação portuguesa de osteogenese imperfeita

UNIDADE LOCAL DE SAÚDE SANTA MARIA

ERN BOND EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES

CENTROS AFILIADOS

UNIDADE LOCAL DE SAÚDE SÃO JOSÉ

UNIDADE LOCAL DE SAÚDE ALMADA - SEIXAL

cuf descobertas hospital

ANDO

PARCEIROS INSTITUCIONAIS

EURORDIS RARE DISEASES EUROPE

oife

fedra FEDERAÇÃO DAS DOENÇAS RARAS DE PORTUGAL

PARQUE DAS NAÇÕES JUNTA DE FREGUESIA . LISBOA

com o apoio da Freguesia Parque das Nações

alpe Fundación ALPE Acondroplasia

HUMAN GROWTH FOUNDATION

FUNDACIÓN AHUCE osteogénesis imperfecta

SIPPT Societal Impact of Pain

PARCEIROS CIENTÍFICOS

Faculdade de Medicina

SPEDP

SPR SOCIEDADE PORTUGUESA DE REUMATOLOGIA

SPNP Sociedade Portuguesa de Nefrologia Pediátrica

APED Associação Portuguesa para o Estudo da Dor

SPGH Sociedade Portuguesa de Genética Humana

spot SOCIEDADE PORTUGUESA DE ORTOPEDIA E TRAUMATOLOGIA

SOCIEDADE PORTUGUESA DE REUMATOLOGIA PEDIÁTRICA

SPOP Sociedade Portuguesa de Ortopedia Pediátrica bySPOT

SPEDM SOCIEDADE PORTUGUESA DE ENDOCRINOLOGIA DIABETES E METABOLISMO

APOIOS E MECENATO

Gold

BIOMARIN

ascendis pharma

Outros

Vila Galé HOTÉIS

El Corte Inglés

Jerónimo Martins

Pastéis de Belém desde 1837

Delta CAFÉS

COMISSÃO ORGANIZADORA | *ORGANISING COMMITTEE*

André Travessa
Céu Barreiros
Daniela Santos
Fátima Godinho
Inês Alves
Katerine Torres
Joana Ruivo Rodrigues
Manuel Cassiano
Margarida Custódio dos Santos
Marta Rodrigues
Tiago Mendes
Susana Alberto

PROGRAMA | *PROGRAMME*

Quinta-feira, 21 de maio de 2026

14:00-14:15 Abertura | *Welcome*

Intervenientes: Cláudia Carvalho Simões (CUF); André Travessa (ULSSM); Fátima Godinho (APOI); Inês Alves (ANDO)

14:15-14:55 Atualizações em Displasias Esqueléticas | Updates in Skeletal Dysplasia:

Moderador: Ana Berta Sousa

- CPMS 2.0: Plataforma europeia para discussão de casos clínico complexos | CPMS 2.0: European Platform for the Discussion of Complex Clinical Cases
- Atualizações em Estudos Clínicos Nacionais e Iniciativas Europeias | Updates in National Clinical Studies and European Initiatives

Palestrantes: Sérgio Sousa; Karen Heath

14:55-15:15 Antropometria em Displasias Esqueléticas | Anthropometry in Skeletal Dysplasias

Moderador: Lurdes Sampaio

Palestrante: Sérgio Sousa

15:15-16:45 Para Além do Osso nas Displasias Esqueléticas | Beyond the Bone in Skeletal Dysplasias

Moderador: Alice Mirante

- Manifestações Oculares... na Osteogénese Imperfeita | Ocular Manifestations... in Osteogenesis Imperfecta

Palestrantes: Rafael Barão; João Naves

- Problemas Dentários... na Displasia Cleidocraniana | Teeth problems... in Cleidocranial dysplasia

Palestrante: Sara Fontes

- Doença Cardiovascular... na Osteogénese Imperfeita | Cardiovascular Disease... in Osteogenesis Imperfecta

Palestrante: Andreia Magalhães

- Perda Auditiva... nas Displasias Esqueléticas | Hearing Loss... in Skeletal Dysplasias

Palestrante: Mariana Correia

- Manifestações Renais... na Hipofosfatemia Ligada ao X | Kidney Problems... in X-linked Hypophosphatemia

Palestrante: Marta Pereira

17:15-17:45 Inteligência Artificial nas Displasias Esqueléticas | Artificial Intelligence in Skeletal Dysplasias

Moderador: Henrique Donato

Palestrante: Beham Javanmardi

17:45-18:30 Fazendo a Ponte entre Doenças Metabólicas e Displasias Esqueléticas – Doenças de Armazenamento Lisossomal com Envolvimento Esquelético | Bridging Metabolic Disorders and Skeletal Dysplasias – Lysosomal Storage Diseases with Skeletal Involvement

Moderador: Maria Abreu

- Avaliação Clínica e Tratamentos Atuais | Clinical Evaluation and Current Treatments

Palestrante: Patrícia Pinto

- Radiologia e Diagnóstico Diferencial | Radiology and Differential Diagnosis

Palestrante: Henrique Donato

Sexta-feira, 22 de maio de 2026

08:20-09:20 Comunicações orais - Casuística e Casos Clínicos I | Oral communications - case series and clinical cases I

Moderadores: Patrícia Almeida Dias; Ricardo Henriques

Click the Link!

09:20-09:50 Imagiologia nas Displasias Esqueléticas | Imagiology in Skeletal Dysplasias

Moderador: Joana Ruivo Rodrigues

- Haverá lugar para a radiografia convencional nas displasias ósseas? | Is there a place for conventional radiography in bone dysplasias

Palestrante: Pedro Alves

- O papel da DEXA na avaliação óssea | The role of DEXA in bone assessment

Palestrante: José Romeu

09:50-10:40 Displasias Esqueléticas na Idade Adulta | Skeletal Dysplasias in Adulthood

Sessão de Cortesia da Human Growth Foundation | *Human Growth Foundation Courtesy Session*

Moderador: André Saraiva

- Osteogénese Imperfeita | Osteogenesis Imperfecta

Palestrante: Fátima Godinho

- Acondroplasia | Achondroplasia

Palestrante: Josep Blanch Rubió

- Displasia Epifisária Múltipla | Multiple Epiphyseal Dysplasia

Palestrante: João Vitor Vieira

11:10-11:50 Construindo Pontes - Ligações entre Doentes, Profissionais de Saúde e Ciência | Building Bridges - Connections Between Patients, Healthcare Professionals and Science

Sessão de Cortesia da Associação Nacional de Displasias Ósseas | Courtesy Session of Associação Nacional de Displasias Ósseas

Moderadores: Melina Mota; Ângela AfonsoLuís Quaresma

- Promovendo o Interesse Profissional e a Educação sobre Displasias Esqueléticas | Accelerating Professional Interests and Education about Skeletal Dysplasias
- O Projeto "PonteS" | The "PonteS " Project
- SKEDYS - Qualidade de Vida em Displasias Esqueléticas | SKEDYS - Quality of Life in Skeletal dysplasias
- ColbioS - Um Biobanco Colaborativo para Displasias Esqueléticas | ColbioS - A Collaborative Biobank for Skeletal Dysplasias
- Questionários e Ferramentas para Apoiar a Investigação | Questionnaires and Tools to Support Research

Palestrantes: Céu Barreiros (APOI); Inês Alves (ANDO); Pisit (Duke) Pitukcheewanont (HGF)

11:50-12:30 Fisioterapia nas Displasias Esqueléticas | Rehabilitation for Skeletal Dysplasias

Moderador: Henrique Maia

- Aspetos Práticos na Osteogénese Imperfeita | *Practical Aspects for Osteogenesis Imperfecta*

Palestrante: *Miguel Rodriguez Molina*

- Fisioterapia em Adultos com Acondroplasia : Estratégias Funcionais e Gestão de Energia Baseadas na Evidência | *Physiotherapy for Adults with Achondroplasia: Evidence-Based Functional Strategies and Energy Management*

Palestrante: *Cláudia Aguiar*

12:30-12:50 Prémio Dedicação - 20 anos da APOI | Dedication Award - APOI's 20th anniversary

Sessão de cortesia da Associação Portuguesa de Osteogénese Imperfeita | Courtesy Session of the Associação Portuguesa de Osteogénese Imperfeita

Intervenientes: Fátima Godinho; Céu Barreiros

14:00-14:50 Ortopedia nas Displasias Esqueléticas (Pediatria e Adultos) | *Orthopedics in Skeletal Dysplasias (Pediatrics & Adults)*

Moderador: *Ricardo Maia*
- Alongamento Ósseo na Era Intramedular | *Limb Lengthening in the Intramedullary Era*

Palestrante: *Tah Pu Ling*
- Aumento Ósseo na Osteogénese Imperfeita | *Bone Augmentation in Osteogenesis Imperfecta*

Palestrante: *Zagorka Pejin*
- Manejo das Deformidades da Coluna | *Management of Spinal Deformities*

Palestrante: *Jorge Mineiro*

14:50-15:40 Comunicações orais - Casuísticas e Casos Clínicos II | Oral communications - case series and clinical cases II

Moderadores: Anabela Bandeira, João Campagnolo

Click the Link!

16:00-17:05 Comunicações Orais - Casuísticas e Casos Clínicos III / Oral Communications - Case Series and Clinical Cases III

Moderadores: Renata d'Oliveira; Andreia Martins

Click the Link!

17:05-17:30 Sessão de prémios | Award session
- Prémio Anual de Investigação Clínica na Área das Displasias Esqueléticas| Annual Clinical Research Award in the Area of Skeletal Dysplasias

Intervenientes: Heloísa Santos
- Prémio BD2026 | BD2026 Award

Intervenientes: André Travessa; Manuel Cassiano Neves; Ana Paula Barbosa

17:30-17:50 Considerações Finais| Closing Remarks

Intervenientes: Fátima Godinho; André Travessa; Manuel Cassiano Neves

MAIO MÊS DE SENSIBILIZAÇÃO
OSTEOGÉNESE IMPERFEITA

6 MAIO

hoje celebramos,

DIA INTERNACIONAL DA OSTEOGÉNESE IMPERFEITA

apoi associação portuguesa de osteogenese imperfeita

#WISHBONEDAY

Sábado, 23 de maio de 2026

09:00-09:15 Apresentação e mensagem de boas vindas

Intervenientes: Representante do Oceanário de Lisboa (a confirmar); Fátima Godinho (APOI), Inês Alves (ANDO)

Click the Link!

09:15-09:45 O que nos liga nas displasias ósseas:

- Aspetos Clínicos

Palestrante: André Travessa

- Aspetos Psicossociais

Palestrante: Micaela Bento

09:45-10:15 Opções Reprodutivas - As Verdadeiras Preocupações das Pessoas com Displasia óssea e suas Famílias:

- o que nos diz a literatura

Palestrante: Céu Barreiros

- apresentação dos resultados dos questionários

Palestrante: Inês Alves

10:45-11:15 Estratégias para Lidar com a Dor nas displasias ósseas

Palestrante: Carina Raposo

11:15-12:00 Cirurgia Ortopédica - Encontro com Especialistas (Perguntas e Respostas)

Painel: Manuel Cassiano Neves; Maria Pia Monjardino; Ricardo Maia

12:00-12:30 Cartão de pessoa com doença rara e atestado multiusos

Palestrante: João Gonçalo

12:30-13:00 O Sono nas Displasias ósseas

Palestrante: Núria Madureira

ENCONTRO DE FAMÍLIAS

09:00-10:15 - Programa Kids & Teens

PAUSA | COFFEE BREAK

10:45-13:00 - Programa Kids & Teens

ALMOÇO COMEMORATIVO - 20 ANOS DA APOI

15:00-17:00 - Visita de famílias ao Oceanário - (Apoio: Oceanário de Lisboa)

PAUSA | COFFEE BREAK

17:30-18:00 - Lanche e encerramento
Intervenientes: Fátima Godinho (APOI); Inês Alves (ANDO); André Travessa (ULSSM)

BONE DYSPLASIAS 2026
SIMPÓSIO DE DISPLASIAS ÓSSEAS

ABSTRACTS

BONE DYSPLASIAS 2026

SILVA, LEANDRO AUGUSTO

NPR2-related Short Stature from a Tertiary Center: Phenotypic Spectrum with Focus on Monoallelic Cases

Silva, Leandro Augusto (1,2); Barros Rua, Inês (2); Mirante, Alice (2,3,4); Martins, Márcia (5); Gomes, Maria Miguel (6); Monjardino, Maria Pia (4,7); Cabral, João José (4,7); Madureira, Núria (4,8); Figueiredo, Pedro (9); Veiros, Iolanda (9); Donato, Henrique (10); Campos-Barros, Angel (11,12); Modamio Høybør, Silvia (11,12); Heath, Karen E. (11,12); Saraiva Santos, Mafalda (13); Rosmaninho Salgado, Joana (13); Sousa, Sérgio Bernardo (3,4,13,14)

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3) Clinical Academic Center of Coimbra, Paediatric Hospital, ULS of Coimbra
4) Bone dysplasia multidisciplinary team (ERN BOND)
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7) Orthopedics, Paediatric Hospital, ULS of Coimbra
8) Pulmonology, Paediatric Hospital, ULS of Coimbra
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10) Radiology, ULS of Coimbra
11) Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, and Skeletal dysplasia multidisciplinary unit (UMDE-ERN BOND), Hospital Universitário La Paz, UAM, Madrid, Spain
12) Rare Diseases Biomedical Research Network (CIBERER), ISCIII, Madrid, Spain
13) Medical Genetics Department, ULS of Coimbra
14) University Clinic of Genetics, Faculty of Medicine, University of Coimbra

KeyWords: NPR2; Short Stature; Skeletal Dysplasia

Introduction:
NPR2 gene, encoding the C-type natriuretic peptide receptor (CNP), is involved in longitudinal bone growth and endochondral ossification. NPR2-variants cause a wide a spectrum from Acromesomelic Dysplasia, Maroteaux type (AMDM), usually due to biallelic loss-of-function (autosomal recessive), to milder short stature in heterozygous carriers (monoallelic/autosomal dominant; 2-6% of "isolated" short stature). Phenotypic variability, particularly among heterozygous individuals, remains incompletely characterized.

Objective/Methods:
Retrospective cohort study of paediatric patients followed at a multidisciplinary skeletal dysplasia clinic, with molecularly confirmed NPR2-related disorders. Clinical, auxological, and genetic data were collected on both monoallelic and biallelic cases. This study focusses detailed analysis on the monoallelic subgroup, also incorporating the parents.

Results:
Five paediatric index patients (3 female, median age of 7 years), were included: three AMDM patients with biallelic compound heterozygous variants; and two patients (1 female), with 10 and 15 years old, with short stature and heterozygous NPR2 variants. At first evaluation of these two cases, at 6 and 9 years, no functional limitations were reported. Observed features included fifth finger brachydactyly, short hands and broad neck, with height -3.03 and -2.19SD. Birth length was -1.89 and -2.58SD, without low birth weight. No patient had growth hormone deficiency; recombinant human growth hormone (rhGH) was not initiated. After a median follow-up of 63.7 months, height remained stable (change $\leqslant$-0.08SD). Eight heterozygous parents were identified. The mothers of the monoallelic probands were also heterozygous and had clinically significant short stature (-3.07 and -3.04SD) with fifth finger brachydactyly and dysmorphic features. Anthropometric and genotypic data from the remaining progenitors are currently being collected and will be presented.

Conclusion:
NPR2 monoallelic variants represent an underdiagnosed cause of short stature. Data on treatment response are limited, with variable outcomes reported for rhGH therapy, while vosoritide (CNP analogue) has shown promising results in clinical trials. NPR2 variant classification remains a challenge, but recent functional studies helped to clarify which ones may have an impact for the autosomal dominant form. Larger cohort studies are needed to clarify genotype-phenotype correlations and improve personalized treatment strategies.

SÁ, ROSÁLIA

Short stature with Compound Heterozygous CUL7 Variants: A possible attenuated phenotype of 3M Syndrome

Sá, Rosália (1); Saraiva Santos, Mafalda (2); Quental Sofia (3); Porto Vasconcelos Alice (1)

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2) Medical Genetics Department, Bone dysplasia multidisciplinar team (ERN BOND), Hospital Pediátrico de Coimbra, Unidade Local de Saúde de Coimbra, Coimbra, Portugal.
3) IPATIMUP Diagnostics, Unit of Medical Genetics, Rua Júlio Amaral de Carvalho, n.º 45, 4200-135 Porto, Portugal.

KeyWords: 3M syndrome

Background
3M syndrome is a rare autosomal recessive skeletal dysplasia caused by pathogenic variants in CUL7, OBSL1, or CCDC8, classically characterized by severe pre- and postnatal growth restriction (⩽-5 SD), distinctive facial features, and characteristic radiographic findings. Mild phenotypes have been rarely reported, mainly in association with OBSL1 and CCDC8.

Case Presentation
We report a 7-year-old boy with mild proportional postnatal short stature (-2.4 SD), delayed bone age, and normal head circumference, without endocrinologic or nutritional abnormalities. Subtle dysmorphic features were noted, including dolicocephaly, frontal bossing, and protruding ears. Family history was notable only for the constitutional delay of growth in the father, whose current height is above average for the Portuguese population.
Genetic testing identified two heterozygous variants in CUL7: a pathogenic frameshift variant c.3136del (p.Leu1046Trpfs*95) and a splice-altering variant c.4566A>G, classified as a variant of uncertain significance. Parental sequencing confirmed compound heterozygosity. RNA analysis through cDNA sequencing demonstrated aberrant splicing, supporting a deleterious effect, although the exact transcript consequence could not be fully determined.
Skeletal survey revealed subtle features compatible with 3M syndrome, including slender long bones, tall vertebral bodies with anterior notching, and pseudoepiphyses of the second metacarpals. However, typical metaphyseal flaring, horizontal ribs, and flaring of the iliac wings were not present. The metacarpal index was not increased.

Discussion
The integrated clinical assessment, craniofacial dysmorphisms, radiographic evaluation, and molecular testing in this case suggest an attenuated phenotype of CUL7-related 3M syndrome. The relatively preserved height (-2.4 SD vs typical ⩽-5 SD) could be explained by the presence of alternative transcripts of the CUL7 gene with residual protein function.
This phenotype poses diagnostic challenges due to the partial clinical and radiographic overlap with 3M syndrome and the inability to exclude constitutional delay of growth in childhood.

Conclusion
Recent studies challenge the classical notion that 3M syndrome has consistently extreme short stature. This work provides evidence of a broader phenotypic spectrum. Recognizing attenuated phenotypes is key to establish genotype-phenotype associations, and further functional studies may clarify the impact of different CUL7 transcripts.

ALVES, RODRIGO

Fifth Family with FLNB-related Short Stature: a New Gene for Idiopathic Short Stature with Minor Skeletal Anomalies

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KeyWords: Baixa Estatura

After comprehensive clinical, radiological, and laboratory evaluation, approximately 60-80% of children presenting with short stature (SS) are classified as having idiopathic short stature (ISS), as no definitive underlying etiology is identified. However, recent advances in molecular genetic testing have identified several genes involved in growth plate development, including ACAN, FGFR3, IHH, NPR2, and SHOX, in which pathogenic variants are responsible for a subset of ISS cases. More recently, Wang et al. described the first four families with SS associated with FLNB haploinsufficiency; variants in this gene are also implicated in Larsen syndrome.

We report the case of a 12-year-old male referred for evaluation of SS. Family history was notable for SS on the paternal side. His growth trajectory remained between the 3rd and 10th percentiles, with progressive deceleration observed after 4 years of age. He was otherwise healthy. On physical examination at 12 years and 4 months, weight was 28 kg (-2.15 SDS) and height was 134.6 cm (-2.32 SDS). He was proportionate and presented with mild joint laxity and mild pectus excavatum, without dysmorphic features. Initial laboratory and endocrine evaluations, including the clonidine stimulation test, were unremarkable. Bone age assessment was consistent with chronological age. Regarding genetic evaluation, MLPA analysis of SHOX did not identify pathogenic variants. In view of the family history, a skeletal dysplasia gene panel was subsequently performed, identifying a likely pathogenic variant in FLNB (NM_001457.4:c.3899-1G>A), not previously reported in the literature. Parental segregation studies are currently in progress.

The identification of a likely pathogenic FLNB variant in this apparently nonsyndromic presentation expands the phenotypic spectrum of filamin B-related disorders and supports its role as a rare cause of ISS or as part of a milder end of the Larsen syndrome spectrum. These findings underscore the importance of comprehensive genetic testing in children with otherwise unexplained short stature.

COHEN, PEDRO

Osteogenesis Imperfecta Type III and Femoral Neck Fracture - a Case Report

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KeyWords: Osteogenesis Imperfecta Type III, Femoral Neck Fracture, Surgical Management, Multidisciplinary Approach

Osteogenesis Imperfecta (OI) comprises a heterogeneous group of genetic disorders affecting type 1 collagen synthesis, characterised by increased bone fragility, reduced bone density, and susceptibility to fractures, even after minimal trauma.

Beyond skeletal involvement, multiple organ systems may be affected.
The severity of OI varies significantly. For that reason, OI can be classified into five main types based on clinical characteristics, with type III being the severe form.

Type III OI is marked by severe skeletal deformities, frequent fractures, and a wide spectrum of multiorgan manifestations, including hearing loss, dentinogenesis imperfecta, joint hypermobility, and craniofacial, cardiopulmonary, neurological, and ophthalmological complications.

This clinical case presents a 42-year-old female patient diagnosed with type III OI who sustained a femoral neck fracture following a fall.

The case report highlights the inherent complexity of managing femoral fractures in adults with this condition, emphasising the necessity for meticulous, individualised perioperative planning in the context of extreme bone fragility, altered anatomy, and the presence of pre-existing surgical hardware. It further underscores the value of a tailored, multidisciplinary approach that integrates surgical, pharmacological, and rehabilitative strategies to optimise outcomes and preserve quality of life.

Given the scarcity of similar reports in the literature, this case provides a meaningful contribution to the evidence base concerning surgical management in this vulnerable patient population.

PINHO, SIMÃO

Osteogenesis Imperfecta Diagnosed After an Apparent Isolated Femoral Fracture in a Newborn: Could We Have Done Better?

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KeyWords: Osteogenesis Imperfecta; Fracture; Suspicious

Introduction:
Osteogenesis imperfecta (OI) is a hereditary connective tissue disorder characterized by bone fragility and recurrent fractures. Its severity is highly variable, reflecting genetic heterogeneity and differences in collagen or bone-related pathways. In early infancy, its initial presentation may raise suspicion of non-accidental trauma, making careful clinical assessment essential for timely diagnosis and appropriate management.

Objective:
This work presents a case of OI diagnosed on the follow-up of an unusual presentation of a femoral fracture.

Methods:
A 25-day-old female infant, with history of perinatal clavicle fracture, otherwise healthy and no family history of skeletal disorders, presented to the emergency department with deformity of the left thigh after a twisting movement during feeding. Plain radiographs confirmed a femoral fracture, and treatment with a Pavlik harness was initiated. On reassessment in the orthopedic outpatient clinic, three months later, family reported no head lag. She was immediately assessed by Neuropediatrics and generalized hypotonia and developmental delay were observed. Neuroaxis magnetic resonance imaging was obtained, which revealed multiple vertebral fractures. The patient was referred to Pediatrics, where laboratory studies showed elevated creatine kinase and skeletal survey detected am healed contralateral femoral fracture. Subsequent genetic confirmed the diagnosis of OI type III (COL1A1 mutation).

Results:
The patient was managed through a multidisciplinary approach involving Metabolic Diseases, Genetics, Rehabilitation, Orthopaedics and Neuropediatrics. Bisphosphonate therapy with quarterly zoledronate was initiated. Clinical evolution has been favorable, with no further fractures reported during follow-up. At 3 years of age, she is able to walk independently and demonstrates normal psychosocial development.

Conclusion:
This case highlights that osteogenesis imperfecta may initially present as an apparently isolated fracture in early infancy, potentially mimicking non-accidental trauma. Recognition of associated clinical, radiologic, and genetic findings is essential to establish the correct diagnosis and guide multidisciplinary management. Adequate referral in the emergency department could have allowed an earlier diagnosis.

SANTOS, JOÃO

Hastes Telescópicas Fassier-Duval no Tratamento da Osteogénese Imperfeita: Experiência Clínica em Dois Casos Pediátricos

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KeyWords: Fassier-Duval

A osteogénese imperfeita (OI) é uma doença genética do tecido conjuntivo caracterizada por fragilidade óssea, deformidades progressivas e fraturas recorrentes desde a infância, frequentemente exigindo múltiplas intervenções ao longo do crescimento. O tratamento ortopédico tem como objetivos não apenas o tratamento das fraturas, mas também a prevenção de deformidades e a otimização da função.

As hastes Fassier-Duval são dispositivos intramedulares telescópicos constituídos por dois componentes (macho e fêmea) que deslizam entre si, permitindo o alongamento progressivo de forma síncrona com o crescimento ósseo. A sua colocação no canal medular dos ossos longos, assegura uma estabilização contínua, manutenção do alinhamento e redução da necessidade de reintervenções cirúrgicas.

Apresentam-se dois casos clínicos pediátricos com OI, o primeiro caso refere-se a uma criança com história de múltiplas fraturas em diferentes segmentos, inicialmente tratadas com encavilhamento elástico (TEN). Perante refraturas e deformidade progressiva, foi submetida a osteotomias de realinhamento e encavilhamento com haste telescópica Fassier-Duval do fémur bilateralmente, com evolução clínica e radiológica favorável, manutenção do alinhamento e recuperação funcional progressiva, apesar de discreta dismetria residual.

O segundo caso corresponde a uma criança com OI associada a síndrome de Ehlers-Danlos (tipo artrocalasia), apresentando fenótipo mais grave e fraturas recorrentes. Após falência de técnicas cirúrgicas prévias, foi realizada substituição por hastes Fassier-Duval em ambos os fémures, com bom resultado pós-operatório, controlo álgico adequado, consolidação eficaz e recuperação funcional satisfatória.

Em ambos os casos, a utilização de hastes telescópicas permitiu estabilização óssea eficaz, correção de deformidades e adaptação ao crescimento, reduzindo a necessidade de múltiplas reintervenções e evidenciando vantagens face a métodos convencionais, como as cavilhas elásticas.

Estes casos reforçam o papel do sistema Fassier-Duval como uma solução segura e eficaz no tratamento cirúrgico de displasias ósseas, nomeadamente OI, permitindo estabilização intramedular dinâmica, com impacto positivo na função e qualidade de vida em idade pediátrica.

CARDOSO, DIANA

COL1A1-Related Ehlers-Danlos Syndrome: Expanding Phenotypic Spectrum with Overlap Features of Osteogenesis Imperfecta

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KeyWords: Arthrochalasia Ehlers-Danlos syndrome; COL1A; Connective Tissue Disorders

Background:
The most common phenotype associated with pathogenic COL1A1 variants is osteogenesis imperfecta (OI). Rarer phenotypes include Ehlers-Danlos Syndrome (EDS) Spectrum phenotypes, characterized by severe joint hypermobility, and tissue fragility. A phenotypic overlap with OI may occur, complicating diagnosis.

Case presentation:
We present a 39-year-old female patient referred to our medical genetics consultation for evaluation of generalized joint hypermobility, recurrent subluxations, scoliosis, blue sclerae, and abnormal wound healing. Family history was notable for blue sclera and hypermobility in her father and two siblings, suggesting autosomal dominant inheritance. Her medical history included hypertension, Beighton score of 8, chronic pain associated with scoliosis, recurrent joint subluxations, muscle tears, nephrolithiasis, and gastrointestinal dysmotility. Notably, she had no history of recurrent fractures or other signs of bone frailty.
A WES-based gene panel for congenital connective tissue disorders identified a heterozygous likely pathogenic variant in COL1A1 c.572G>C p.(Gly191Ala). DXA scan revealed a low lumbar Z-score of -2.8, but normal femoral bone density and normal bone microarchitecture. Considering her phenotype, diagnoses of arthrochalasia EDS (aEDS) type 1 and overlapping EDS/OI were considered. Variants affecting this residue have been described in patients with atypically marked hyperlaxity and tissue frailty for OI standards.

Discussion:
Despite the strong association of aEDS to loss of exon 6 in COL1A1, this was the first diagnosis considered for the patient, given the marked joint hypermobility, recurrent subluxations, connective tissue fragility and absence of recurrent bone fractures. However, the presence of blue sclerae, which has been described in aEDS, highlights phenotypic overlap with OI. Furthermore, personal history of congenital hip luxation could not be ascertained. Upon literature review, most signs and symptoms associated with the COL1A1 disease spectrum can be found in patients diagnosed with both OI and aEDS, and relative severity of features informs diagnosis.

Conclusion:
This case highlights the value of broad genetic panel testing in the differential diagnosis of collagenopathies. Additionally, it provides further evidence that mainly-EDS phenotypes may not be restricted to exon 5/6 variants, and that COL1A1 disease behaves like a spectrum of disorders, rather than easily distinguishable entities.

LOPES, GRAÇA

O Crescimento de um jovem com osteogénese imperfeita

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KeyWords: Osteogénese Imperfeita

Os autores vão apresentar um caso clínico desde o RN até à idade adulta e mostrar as dificuldades no tratamento das deformidades.
A evolução das técnicas cirúrgicas até à existência de protocolos definidos do tratamento da fragilidade óssea
A evolução tecnológica dos implantes assim como dos protocolos de tratamento tem sido constante com melhoria da qualidade óssea e menor número de fraturas e com qualidade de vida que permitiu o ingresso social das crianças e jovens

RODRIGUES, MARIANA MENDES

Clinical Spectrum and Therapeutic Challenges in Adult COMP-related Dysplasias

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KeyWords: COMP mutations; Pseudoachondroplasia; Multiple Epiphyseal Dysplasia; Antioxidants

Introduction:
Mutations in the COMP gene cause skeletal dysplasias such as pseudoachondroplasia (PSACH) and multiple epiphyseal dysplasia type 1 (MED). Misfolded COMP accumulates in the chondrocyte endoplasmic reticulum (ER), triggering chronic ER stress, oxidative stress and inflammation, leading to premature chondrocyte death, growth impairment and early osteoarthritis. Management remains largely supportive, as no disease-modifying therapies are currently approved.

Objective:
To characterize the clinical spectrum of adult COMP-related dysplasias in a tertiary center and explore potential therapeutic strategies.
Methods: We conducted a retrospective analysis of adults with genetically confirmed COMP-related dysplasias evaluated at a tertiary referral hospital. A targeted literature review was also performed to identify emerging therapeutic approaches.

Results:
A total of 15 patients with pathogenic variants in the COMP gene were identified. Of these, six were in the pediatric age group (three with MED, two with PSACH, and one with an intermediate phenotype). The focus of this case series was on adult patients, among whom nine were identified: two with MED, five with PSACH, and two with an intermediate phenotype. Of these, only four patients are currently followed in adult rheumatology. The phenotypic spectrum was heterogeneous, with many patients having undergone multiple orthopedic interventions and experiencing limitations in activities of daily living, often requiring multidisciplinary management. Literature data describe reduced COMP-related pathology and suggest that anti-inflammatory and antioxidant agents, such as aspirin and resveratrol, may have potential therapeutic effects, although evidence remains preclinical.

Conclusion:
COMP-related dysplasias show marked clinical heterogeneity and significant functional impact, requiring multidisciplinary management. Treatment remains supportive, with no approved disease-modifying therapies. Further studies are needed to better define disease characteristics and evaluate emerging therapeutic strategies.

TORRES, KATERINE

Phenotypic Spectrum Associated with Variants in the COL2A1 Gene: a Retrospective Study of Cases Diagnosed at Two Hospitals

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KeyWords: COL2A1; Type II Collagenopathies

Introduction:

COL2A1 gene encodes type II collagen, a key structural component of cartilage essential for endochondral ossification, skeletal growth, joint function, and ocular and inner ear development. Pathogenic variants in COL2A1 are associated with a wide phenotypic spectrum, ranging from severe disorders such as achondrogenesis type II to milder conditions, including Stickler syndrome type I and COL2A1-related osteoarthritis.

Materials and Methods:

A retrospective observational study was conducted by reviewing medical records of patients with COL2A1-related disorders followed at medical genetics appointments in Unidade Local de Saúde Santa Maria and Unidade Local de Saúde Amadora-Sintra from April 2014 to March 2026.

Results:

A total of 34 patients from 19 families were included. Diagnoses comprised Stickler syndrome type I (n=18), achondrogenesis type II (n=5), COL2A1-related osteoarthritis (n=2), COL2A1-related Legg-Calvé-Perthes disease (n=1), spondyloepiphyseal dysplasia with metatarsal shortening (n=2), spondyloepiphyseal dysplasia congenita (n=2), Stanescu-type dysplasia (n=1), one unclassifiable family (n=2), and one 12q12-q13.11 microdeletion involving COL2A1.
Missense variants affecting protein structure were associated with more severe presentations. Achondrogenesis type II cases were linked to missense variants (glycine, proline, and tyrosine substitutions; one each) and two intronic variants. Glycine substitutions were also identified in spondyloepiphyseal dysplasia. Arginine substitutions were observed in moderate and mild phenotypes. One CNV was identified incidentally in a fetus.
Among Stickler syndrome patients, splice-site variants predominated (88.88%), supporting haploinsufficiency. All had ocular involvement, mainly high/moderate myopia (88.88%); five had hearing loss (27.77%), and four had cleft palate (22.22%). Remaining patients had missense variants, including one arginine-to-cysteine substitution and recurrent glycine-to-serine substitutions associated with milder phenotypes.

Conclusion:

The clinical spectrum of COL2A1 variants is broad, and genotype-phenotype correlations remain incompletely predictable. While splice-site variants are typically associated with milder phenotypes and missense variants, particularly glycine substitutions, with more severe forms, this is not absolute. Variability suggests additional modifying factors. Expansion to further centers is planned to include more patients and help strengthen these associations.

PANOEIRO, JONATHAN

Type II Collagenopathy: The Milder End of the Spectrum

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KeyWords: Type II Collagenopathy; COL2A1; Early-onset Osteoarthritis

Introduction:
Deleterious variants in COL2A1 are associated with a wide spectrum of clinical phenotypes, ranging from perinatal lethal disorders to milder conditions presenting from infancy to adulthood. These disorders are collectively referred to as type II collagenopathies.
This phenotypic heterogeneity poses a diagnostic challenge, particularly in distinguishing these conditions from disorders of immune, multifactorial or unknown etiology.

Case report:
We present a 16-year-old male with normal stature and polyarticular disease predominantly affecting the lower limbs, characterized by arthralgia, joint stiffness, limited range of motion, and marked gait impairment since childhood.
His mother has a history of polyarticular disease requiring orthopedic interventions by age 20, and a maternal aunt reports similar joint complaints.
Radiological evaluation at age 14 demonstrated degenerative and inflammatory changes involving the right elbow, hips, and spine, suggesting the diagnosis of juvenile idiopathic arthritis. Serological markers were negative and there was no response to immunosuppressive therapy.
Follow-up imaging at age 16 revealed marked axial involvement, including coarse sclerotic and lytic lesions, platyspondyly and cervical ankylosis. In combination with poor therapeutic response and suggestive maternal family history, these findings raised suspicion of spondyloepiphyseal dysplasia.
WES-based gene panel identified a heterozygous pathogenic variant, NM_001844.5(COL2A1):c.611G>A (p.Gly204Asp), confirming the diagnosis of osteoarthritis with mild chondrodysplasia/spondyloepiphyseal dysplasia. This variant was inherited from the affected mother. Genetic counselling for an autosomal dominant disorder was provided.

Discussion:
While classic type II collagenopathies are characterized by short stature, skeletal dysplasia, ocular abnormalities, hearing impairment and distinctive orofacial features, a subset of patients presents with atypical, milder forms of spondyloepiphyseal dysplasia manifesting as early-onset rheumatoid-like arthritis or degenerative hip disease. These milder phenotypes are important differential diagnosis from the more common autoimmune skeletal disorders and differ particularly in therapeutic approach and recurrence risk.
Our report highlights the importance of clinical suspicion and correct diagnosis in these cases, particularly since standard treatment for immune disorders may have adverse effects and is ineffective in these patients.

COSTA, SARA ALVES

Same Name, Different Disease: Genotype-Phenotype Correlations in Adult Patients with Multiple Epiphyseal Dysplasia in a Tertiary Centre

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KeyWords: Multiple Epiphyseal Dysplasia; Osteoarthritis

Introduction:
Multiple epiphyseal dysplasia (MED) is a heterogeneous condition caused by mutations in at least six genes, with dominant and recessive forms distinguished by specific clinical and radiographic features. Accurate subtype identification is essential for prognosis and genetic counseling.

Objective:
To characterize the clinical, radiographic, and genetic heterogeneity of MED in adults followed at a tertiary Rare Bone Diseases Clinic.

Methods:
Retrospective descriptive analysis of five adult patients (P1–P5) with genetically confirmed MED.

Results:
Distinct subtype profiles were observed. COMP-MED (P1, 36 years) showed proportionate short stature, joint laxity, genu varum, and early bilateral coxarthrosis requiring total hip arthroplasty (THA). COMP-MED (P2, 20 years) presented disproportionate short stature, pectus excavatum and coxarthrosis. Both had dorsolumbar scoliosis. SLC26A2-MED (P3, P4; 38 and 62 years) showed disproportionate short stature, joint contractures, brachydactyly, genu valgus and bilateral gonarthrosis and coxarthrosis (bilateral THA in P3); P4 also had facial dysmorphia and absent patella. CANT1-MED (P5, 74 years) exhibited joint laxity, genu varum, dorsolumbar scoliosis, and required knee arthroplasty.

Discussion and Conclusion:
Both COMP-MED patients presented with dorsolumbar scoliosis and coxarthrosis, but differed in stature. SLC26A2 patients shared a broadly similar phenotype, with the exception of facial dysmorphia observed in one case. The CANT1-MED patient shared features with COMP-MED, notably joint laxity and genu varum. Regardless of the underlying genetic subtype, most patients required joint replacement surgery, even at a young age.
In conclusion, despite sharing a common designation, MED encompasses biologically distinct entities with recognisably different clinical profiles. In the absence of pediatric radiographic signs, functional and articular burden becomes the dominant clinical reality, frequently culminating in joint replacement surgery even at a young age. These findings reinforce the value of specialized clinics in the long-term multidisciplinary management of MED.

RODRIGUES DA SILVA, CONCHA

Pregnancy in Women with Skeletal Dysplasia - Follow-up and Prognosis - Preliminary Findings

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KeyWords: Gestação; Displasias Ósseas

Introduction:
Skeletal dysplasias (SD) are a heterogenous group of rare disorders, with widely variable clinical manifestations.
Most SDs allow for normal fertility, but some medical challenges should be accounted for during pregnancy, since it comprises major anatomic and physiologic changes across various systems. Literature on the subject is scarce, and most often limited to occasional clinical case reports.

Methods:
This is a retrospective study of pregnancy in women with SD. Informed consent was obtained for all participants, and approval from ULSSA's ethical and access to clinical data commissions. Patients with osteogenesis imperfecta, isolated craniosinostosis or other SDs with affection limited to the skull were not included.

Results:
As of this abstract, data was obtained for 8 patients, which collectively had 10 live births, and one ongoing pregnancy. Patients diagnoses included, COL2-associated SD (2), FGFR3 SD (1), Leri-weil syndrome (1), X-linked hypophosphatemia (1), aggrecanopathy (1), 3M syndrome (1), and 1 patient awaiting molecular diagnosis. Short stature was present in 7/8 women, with adult height ranging from 118cm to 163cm. All deliveries were by c-section, with the most common indications reported being pelvic bone disease, or cephalopelvic disproportion. For most pregnancies, patients reported an absence of new symptoms (9/10 births) and stability of previously recognized symptoms during (8/10) and after (7/10) pregnancy. Most pregnancies were relatively free of complications and report successful interactions with the health system, but patient involvement in decision-making varied. The presence of complications and maternal experience do not seem to correlate with maternal height, in this cohort.

Discussion
These preliminary findings suggest pregnancy is generally safe in women with SD. Selection biases must be accounted for; patients who chose not to carry a pregnancy having been medically advised against it or due to a perceived severity of their disorder were not included. Additionally, being retrospective, this study may underscore variations in pain intensity and other symptoms. Regardless, considering the lack of evidence available in the literature for pregnancy in most skeletal dysplasias, these findings represent new and encouraging data concerning he safety of pregnancy in women with SD, and collaboration requests have been sent to expand it into a national study.

PINHO, SARA

TANGO1 Loss-of-Function Underlies Prenatal Lethal Skeletal Dysplasia with Absent Bone Mineralisation

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KeyWords: Achondrogenesis; Prenatal Lethal Skeletal Dysplasia; Absent Bone Mineralisation

Introduction:
The TANGO1 gene (also known as MIA3) has been associated with odontochondrodysplasia with hearing loss and diabetes (OMIM #619269), caused by biallelic splice-altering variants leading to exon skipping. More recently, Guillemyn et al. identified a homozygous frameshift variant in a fetus with absent bone mineralisation, suggesting a broader phenotypic spectrum related to TANGO1 deficiency.

Objective:
To report a prenatal severe skeletal dysplasia case due to a homozygous TANGO1 loss-of-function variant and highlight the associated genotype-phenotype correlation.

Methods:
A Portuguese couple of Roma descent, with two prior pregnancy terminations due to fetal micromelia suggestive of achondrogenesis, was referred at 16 weeks of gestation to our Medical Genetics Department, following abnormal obstetric ultrasound findings. Detailed prenatal imaging, postmortem examination and trio whole-exome sequencing (WES) of fetus and parents were performed.

Results:
Prenatal ultrasound demonstrated tetramicromelia, narrow thorax, increased nuchal fold, generalised edema, absent vertebral ossification, and equinovarus feet. After counselling, the couple elected to terminate the pregnancy at 16 weeks and 3 days. Postmortem physical examination and skeletal survey confirmed profound skeletal hypomineralisation, including absent cranial ossification, rhizo- and mesomelic limb shortening, brachydactyly, and generalised edema. Trio WES identified a homozygous nonsense variant (c.2479C>T, p.Arg827Ter) in TANGO1. Both parents are heterozygous carriers. This homozygous variant is predicted to result in absent or severely truncated TANGO1 protein and is associated with a severe prenatal skeletal phenotype likely incompatible with postnatal survival.

Conclusions:
This case expands the phenotypic spectrum of TANGO1-related disease and supports a genotype-phenotype correlation in which the degree of protein disruption determines skeletal involvement. TANGO1 should therefore be considered a newly recognised gene in the differential diagnosis of achondrogenesis-like lethal bone dysplasias, highlighting the importance of molecular diagnosis for genetic counselling and reproductive planning.

JESUS, SARA

TBX6-associated Hemivertebrae due to Compound Inheritance of a Truncating Variant and a Hypomorphic Haplotype: a Mild Non-syndromic Presentation

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KeyWords: TBX6; Hemivertebrae; Congenital Vertebral Malformations; Hypomorphic Allele; Spondylocostal Dysostosis; WES

Background:
Congenital vertebral malformations show marked genetic and phenotypic heterogeneity. A pseudo-dominant inheritance model has been described for TBX6-related disease, wherein the combination of a rare deleterious variant with a common hypomorphic haplotype causes a variable and often mild presentation that may appear non-syndromic.

Case Report:
We report a male patient prenatally diagnosed with thoracolumbar hemivertebrae. Postnatal imaging confirmed multiple thoracic and lumbar hemivertebrae with segmentation defects and mild kyphoscoliosis, without additional skeletal features suggestive of a specific skeletal dysplasia. Growth and neurodevelopment were within expected parameters, and no functional motor impairment was identified. Minor non-specific findings included bilateral clinodactyly of the fifth finger and a preauricular hyperpigmented lesion. Family history was non-contributory, apart from mild spinal asymmetry in distant relatives.
Karyotype and arrayCGH were normal. A skeletal dysplasia panel (WES-based) identified a heterozygous TBX6 frameshift variant (c.1264_1265dup; p.(Ala423Profs*76)), initially classified as a variant of uncertain significance and inherited from a reportedly asymptomatic parent. Subsequent targeted analysis demonstrated a TBX6 hypomorphic haplotype (T-C-A) in trans. This finding supported reclassification of the variant as likely pathogenic and established a diagnosis of TBX6-associated congenital vertebral malformations, consistent with a mild form of autosomal recessive spondylocostal dysostosis with pseudo-dominant inheritance.

Conclusions:
This case highlights the diagnostic importance of recognising compound inheritance involving hypomorphic alleles in patients presenting with apparently isolated vertebral anomalies. It further emphasises the value of comprehensive genomic analysis in informing genetic counselling and recurrence risk assessment, and delineating clinically subtle presentations within the disease spectrum.

RAPOSO, SARA

The Hidden Risk of Spinal Cord Stenosis in CHST3-Related Recessive Larsen Syndrome

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KeyWords: CHTS3, Spondyloepiphyseal Dysplasia, Myelopathy

Introduction:
CHST3-related skeletal dysplasia, also known as recessive Larsen syndrome, is characterized by ligamentous hyperlaxity, facial dysmorphism, and progressive osteoarticular abnormalities. Atlantoaxial instability but also progressive intervertebral space narrowing and endplate irregularities and kyphoscoliosis are common features.

Aim:
Case report of progressive cervical myelopathy where the multidisciplinary approach was essential to mitigate morbidity and enhance long-term quality of life.

Methods:
This report details the clinical course of a child carrying a CHST3 mutation with multiple joint dislocations. During childhood, management included serial casting and gentle manipulation for bilateral knee and foot deformities, followed by right hip and knee reconstruction (left knee reconstruction performed at 11), and a C1-C2 posterior arthrodesis to address atlantoaxial instability. Despite consistent physical therapy, the patient experienced a progressive decline in ambulation at age 10, initially associated with joint pain related to the underlying condition. However, by age 12, she presented a rapid onset of myoclonus and limb spasticity that prompted urgent investigation by Neurology. Cervical CT and complete spine MRI revealed dysplastic changes with severe cervical spinal cord stenosis, confirming cervical myelopathy. In a multidisciplinary effort with Pediatric Orthopedics and Neurosurgery, the patient underwent a complete laminectomy from C2 to C6, followed by posterior instrumented arthrodesis from C2 to T5 using autologous iliac crest bone graft.

Results:
The postoperative recovery was uneventful. At the 6-month follow-up, with sustained physical therapy, the patient exhibited significant pain relief and successfully regained independent ambulation over short distances.

Conclusion:
CHST3-related recessive Larsen syndrome is a rare entity, and its recognition may be difficult in clinical practice. A clear understanding of the condition's natural course is fundamental, and clinicians should remain vigilant for cervical spine instability but also for dysplastic changes and risk of spinal cord compression with serious neurological sequela, hidden by disease progression. Long-term, coordinated surveillance by a multidisciplinary team is essential to monitor disease progression, maximize functional capacity, and ensure early identification and management of complications.

BARREAL VEGA, CARMEN

Natural History of Spine in Achondroplasia and its Management Strategy

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KeyWords: Achondroplasia, Spine

Introduction:
Children with achondroplasia often develop thoracolumbar kyphosis during the first few months of life, a bulge in the spine (between vertebrae D12-L1) due to their generalized muscle hypotonia and macrocephaly. However, in most children, this deformity corrects itself spontaneously as the spinal muscles strengthen during development, with the onset of independent walking being the most significant stage. It can lead to permanent or progressive thoracolumbar kyphosis if preventive functional treatments are not pursued. In adulthood, neurological deficits such as paresthesia in the lower extremities, claudication, clonus, and bladder or bowel dysfunction may be more frequent as a consequence of lumbar stenosis in the lower back. It is important to have this spinal development in achondroplasia in order to obtain the treatment that best suits people with this condition.

Aims:
Study the evolution of the spine in achondroplasia from birth to adulthood, develop a strategy for the clinical evaluation of the spine in patients with achondroplasia, and implement a therapeutic intervention protocol based on the results obtained.

Methods:
A comprehensive assessment of various spinal features in achondroplasia was conducted, examining characteristics such as thoracolumbar kyphosis, lumbar hyperlordosis, scoliosis, joint mobility, and many other factors. Results. Thoracolumbar kyphosis appears in most children with achondroplasia during the first years of life. Lumbar hyperlordosis typically appears when the child begins to stand, and the kyphosis disappears. Joint mobility is good in most of the individuals studied. Scoliosis is not usually present.

Conclusion:
It is observed that the toracolumbar kyphosis present in children during the first years of life usually corrects itself once they achieve stable independent walking and their muscle tone has improved, leading in the following stage to lumbar hyperlordosis, which is characteristic of adulthood. The results will be used to develop a spinal management strategy for achondroplasia and thus provide a guide for physiotherapy treatment.

BACALHAU, MAFALDA

SLCO2A1-Related Primary Hypertrophic Osteoarthropathy: The Role of Early Diagnosis in Enabling Effective Etoricoxib Treatment

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KeyWords: Primary Hypertrophic Osteoarthropathy

Hypertrophic osteoarthropathy (HO) is a skeletal disorder characterized by abnormal proliferation of cutaneous and periosteal tissues of the extremities, with clinical manifestations including periostosis of the tubular bones, arthralgias, joint effusion, digital clubbing, and pachydermia. There are two forms of the disease: 1) primary, with a genetic etiology due to variants in two genes involved in prostaglandin E2 degradation (SLCO2A1 and HPGD); 2) secondary, which is more common and occurs as a response to serious underlying conditions, especially lung and heart diseases.

We present the case of a 24-year-old man, born to non-consanguineous and healthy parents, with a history of multiple hospitalizations for pain and inflammation of the knees, elbows, and small joints of the hands, with onset at age 17. Additionally, he presented with eyelid edema, thickening of the forehead wrinkles, and epigastric pain. An extensive etiological workout was performed, excluding secondary causes, and no clinical improvement was observed with multiple anti-inflammatory drugs. Physical examination and radiological assessment strongly suggested primary HO. Molecular analysis of the SLCO2A1 and HPGD genes was performed using a skeletal dysplasia panel, identifying a homozygous pathogenic variant in the SLCO2A1 gene, c.940+1G>A p.?. This variant affects the canonical splice site donor, is predicted to disrupt normal mRNA processing, and previous functional studies have demonstrated its pathogenicity. Monoallelic SLCO2A1 variants are mainly associated with primary HO, while biallelic variants are linked to primary HO or primary HO-enteropathy syndrome (associating, in addition, chronic nonspecific ulcers of the small intestine); however, this variant has been reported in association with both phenotypes. The patient underwent upper gastrointestinal endoscopy, which showed only duodenitis and gastritis. He was started on etoricoxib and esomeprazole, with significant and sustained clinical improvement.

This case highlights the importance of considering primary HO in young patients with chronic arthralgia, digital clubbing, and pachydermic features after exclusion of secondary causes. Given the potential association with gastrointestinal involvement, long-term digestive surveillance is warranted. Early diagnosis facilitates appropriate management, as demonstrated by the favorable clinical response to etoricoxib, and can prevent unnecessary investigations.

RODRIGUES, JOANA CORREIA

Opsismodysplasia – From Genetics to Patient Care!

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KeyWords: Opsismodysplasia; Orthopedic Surgical Strategy; Orthopedic Management; Multidisciplinary Approach

Introduction:
Opsismodysplasia is an extremely rare skeletal dysplasia, with an estimated prevalence of approximately 1 per 1 million births. It is an autosomal recessive disease, most commonly associated with mutations in the INPPL1 gene, although genetic heterogeneity has been suggested. Fewer than 30 cases have been reported worldwide, significantly limiting early diagnosis and optimal clinical and surgical management. It is characterized by a delay in bone maturation, severe disproportionate short stature and distinctive craniofacial features.

Case Report:
A six-month-old female was referred to Genetics clinic due to prenatal-onset disproportionate short stature, rhizomelic limb shortening, macrocephaly, frontal bossing, saddle nose, short neck and thorax, trident hands and mild genu varum. Molecular testing for FGFR3 and COMP was negative, excluding achondroplasia and pseudoachondroplasia. Subsequent genetic analysis identified a homozygous pathogenic variant in the INPPL1 gene (c.768_769del, p.Glu258Alafs*45), confirming the diagnosis of opsismodysplasia. She has been managed through a multidisciplinary approach, including Cardiology, Pneumology, Endocrinology, Rehabilitation and Orthopedics. Orthopedic manifestations included bilateral genu varum, coxa vara and double-curve scoliosis, the last initially managed conservatively with bracing. Due to progressive lower limb deformities, she underwent multiple surgical interventions. Initial bilateral proximal lateral tibia and trochanteric hemiepiphysiodesis at age 7 showed no clinical or radiographic improvement over the following year, with persistent deformity, leading to revision surgery at age 8. Ultimately, at age 11, due to continued lack of correction, she underwent bilateral proximal tibial and fibular osteotomies with gradual deformity correction and lengthening using a circular hexapod external fixator. Following subsequent hardware adjustments and removal at age 12, progressive bone consolidation and satisfactory alignment were achieved. At the latest follow-up (age 13), she was asymptomatic, ambulating independently and remains under regular multidisciplinary surveillance.

Conclusion:
This case report aims to raise awareness of this rare condition, which poses challenges in both diagnosis and orthopedic surgical decision-making, including the choice and timing of techniques. It also highlights the critical role of a multidisciplinary approach in optimizing patient outcomes.

MARTINS COELHO, MARTA

Hypohidrotic Ectodermal Dysplasia in Pediatric Age: Multidisciplinary Challenges with Emphasis on Early Orofacial Rehabilitation

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KeyWords: Hypohidrotic Ectodermal Dysplasia; Pediatric Dentistry; Oral Health

Introduction:
Hypohidrotic Ectodermal Dysplasia (HED) is a rare genetic disorder affecting ectoderm-derived structures, including skin, teeth, nails, and sweat glands. Its systemic involvement, and significant orofacial alterations, requires an early, coordinated, and multidisciplinary approach.

Case Report:
A 4-year-old male, third child of non-consanguineous Cape Verdean parents was diagnosed with HED at age 3. During childhood, he presented recurrent hyperthermia, infections, anemia, and failure to thrive, requiring prolonged hospitalization in Cape Verde and later in Portugal. Currently followed by multiple specialties, including infectious diseases, primary immunodeficiencies, and otorhinolaryngology. He was referred to pediatric dentistry due to feeding difficulties and delayed speech development. Clinical examination revealed typical HED features, including hypohidrosis, xerosis, pigmentary changes, and sparse, fine hair. Intraoral findings showed severe tooth agenesis consistent with anodontia, resulting in marked functional impairment and compromised oral function.

Therapeutic Intervention:
Prosthetic rehabilitation with complete removable dentures is planned to restore mastication, improve speech, and support orofacial growth. Regular follow-up will allow prosthetic adjustment according to craniofacial growth and functional needs.

Discussion:
HED has a significant systemic impact, mainly due to sweat gland dysfunction, increasing hyperthermia risk and reducing mucosal protection. Oral manifestations are nearly universal and impair quality of life. Anodontia or severe hypodontia, affects feeding, nutrition, speech and social interaction. Early prosthetic rehabilitation, preferably before school age, is recommended and should be continuously adapted within a comprehensive multidisciplinary framework.

Conclusion:
HED is a complex multisystem disorder in which dental involvement plays a central role in function and quality of life. Although not a primary bone dysplasia, it may significantly compromise craniofacial bone development, with important therapeutic and rehabilitative implications.

MENDES, ARIANA C.

From Brachydactyly to Symphalangism: Exploring Genotype-Phenotype Correlations in GDF5- and NOG-Related Digital Malformations

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KeyWords: Brachydactyly; Symphalangism

Digital malformations such as brachydactyly and symphalangism are defined by abnormalities in bone growth and joint formation, respectively. Variants in key regulatory genes, particularly GDF5 and NOG, disrupt shared pathways of skeletal and joint development, giving rise to a broad phenotypic spectrum with overlapping features that may limit the accuracy of phenotype-driven diagnostic classification.

We report four unrelated families to explore genotype-phenotype correlations within this spectrum, with emphasis on radiological findings. One case with a confirmed GDF5 pathogenic variant involves a child with bilateral brachydactyly and clinodactyly predominantly affecting digits 2-3, with familial segregation of a heterozygous frameshift variant (c.498dup; p.(Ile167Hisfs*18)). A second case describes an adult with brachydactyly associated with polyphalangism and symphalangism, with several affected relatives and a distribution pattern typical of GDF5-related conditions, currently awaiting molecular confirmation. Both families fit the clinical description of Brachydactyly type C (MIM#113100).

Similarly, NOG-associated phenotypes encompass a spectrum of joint-related anomalies. One patient presented with Multiple Synostoses Syndrome (MIM#186500): extensive symphalangism, carpal and tarsal coalitions and suspected hearing loss, associated with a heterozygous pathogenic de novo variant (c.611G>A; p.(Arg204Gln)). Another familial case, characterized by proximal symphalangism (MIM#185800), conductive hearing loss due to ossicular involvement and craniofacial features, carries a NOG variant (c.696C>A; p.(Cys232*)) initially classified as of uncertain significance and reclassified as likely pathogenic after segregation in four additional affected family members.

These cases show that, despite distinct mechanisms, GDF5 and NOG variants can produce overlapping clinical features within shared developmental pathways. Detailed radiological assessment is essential for accurate diagnosis. Across both genes, the coexistence of digital shortening, altered phalangeal patterning and joint fusion reflects a common developmental basis. Each gene is associated with a broad phenotypic continuum characterized by variable expressivity and pleiotropy, contributing to diagnostic complexity. While specific phenotypic patterns may help prioritize candidate genes, clinical evaluation alone may not reliably predict genotype, highlighting the need for an integrated diagnostic approach.

NASCIMENTO, INÊS

Acrodysostosis: Clinical Challenges and Implications for the Dental Treatment in Pediatric Patients

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KeyWords: Acrodysostosis; Pediatric Dentistry; Special Healthcare Needs; Oral Health; Quality of Life

Introduction:
Acrodysostosis is a rare genetic skeletal dysplasia characterized by brachydactyly, craniofacial abnormalities, and often cognitive impairment. It shows phenotypic overlap with pseudohypoparathyroidism type Ia and may be associated with multiple hormone resistance. Craniofacial alterations, including midface and nasal hypoplasia, may impact function and oral health, supporting the need for early, tailored medical-dental care.

Case Report:
An 11-year-old male with acrodysostosis confirmed by molecular analysis (heterozygous c.902G>A mutation in PDE4D) .The patient presents cognitive impairment and is followed by multiple specialties including speech therapy. No PTH or thyroid abnormalities were identified. Medical history includes low birth weight, neonatal hospitalization, and recent ENT surgeries.
Dental examination revealed mixed dentition, dentoalveolar discrepancy, and molar-incisor hypomineralization (MIH), with severe involvement of tooth 26. Generalized dental plaque and gingivitis were present, associated with caregivers'difficulties in oral hygiene and a cariogenic diet. Low cooperation and sensory hypersensitivity limited treatment.
Due to behavioral limitations and treatment needs, care under general anesthesia was planned and performed in November 2025. Teeth 26, 63, and 65 were extracted.
At 5-month follow-up, no new carious lesions were observed, and preventive oral care was implemented, although behavioral management difficulties persisted.

Discussion:
Acrodysostosis may increase the risk of caries and periodontal disease. Cognitive and sensory impairments further hinder preventive care and treatment. Individuals with special healthcare needs show higher oral disease burden supporting the importance of early, individualized strategies. Preventive measures such as plaque control, fissure sealants, and fluoride use are essential.
This case underscores the need for multidisciplinary management and advanced behavioral techniques, including general anesthesia, to ensure safe and effective care.

Conclusion:
Acrodysostosis presents significant challenges in pediatric dental care. Early diagnosis, preventive strategies, and individualized multidisciplinary approaches are key to improve oral health and quality of life.

SANTOS, MAFALDA SARAIVA

Acromesomelic Dysplasia, Maroteaux Type: Clinical and Molecular Characterisation of Three Cases

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KeyWords: NPR2; Acromesomelic Dysplasia, Maroteaux Type; Short Stature; Skeletal Dysplasia

Introduction:
The NPR2 gene encodes the natriuretic peptide receptor B, crucial for longitudinal bone growth and endochondral ossification. The genetic landscape of NPR2 variants includes a broad spectrum, from loss-of-function to activating changes, causing diverse phenotypes ranging from severe acromesomelic dysplasia, Maroteaux type (AMDM), to autosomal dominant short stature and, less commonly, tall stature. A full understanding of the functional consequences of several NPR2 variants and the mechanisms underlying their effects remains a challenge.

Methods:
Retrospective cohort of patients with molecularly confirmed NPR2-related AMDM, followed at a multidisciplinary skeletal dysplasia clinic. Literature review and curation of functional data and databases of reported NPR2 variants.

Results:
We report three AMDM patients with focus on detailed radiological findings. Two patients already showed prenatal shortening of long bones and short stature at birth (-3.4 SD; -2.0 SD). All developed postnatal growth deficiency prompting genetic evaluation, with molecular confirmation at a median age of 30 months. Characteristic skeletal features, such as acromesomelic shortening of the limbs and epiphyses/metaphyses irregularities, were key to the diagnosis, especially for a patient with a prior negative WES but clear clinical and radiological suspicion (after referral to our skeletal dysplasia clinic and expert consultation). Median follow-up was 42 months, without treatment. Height further declined in the two typical severe cases (-6.5 SD; -4.4 SD), reflecting the characteristic progression in AMDM. Surprisingly, a third milder case improved to -2.0 SD. Regarding genotype-phenotype correlations, this third patient harbours two missense variants, whereas the other two are heterozygous for both a missense and a nonsense/frameshift variant. Heterozygous parents' phenotype and genetic variant nature will also be discussed.

Conclusion:
This cohort highlights the phenotypic and genotypic hallmarks of recessive NPR2-related dysplasia, emphasizing the role of biallelic loss-of-function variants in severe skeletal phenotypes. Recognition of the characteristic radiological pattern is critical for diagnosis. Differential diagnosis, mainly including mucopolysaccharidoses, is crucial for accurate case evaluation. Further studies are needed to refine genotype-phenotype correlations and evaluate possible targeted therapies.

BONE DYSPLASIAS 2026
SIMPÓSIO DE DISPLASIAS ÓSSEAS

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BONE DYSPLASIAS 2026
SIMPÓSIO DE DISPLASIAS ÓSSEAS

PRÉMIO

MELHOR TRABALHO

Vencedor

Katerine Torres

Phenotypic Spectrum Associated with Variants in the COL2A1 Gene: a Retrospective Study of Cases Diagnosed at Two Hospitals

BONE DYSPLASIAS 2026
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PRÉMIO

BONE DYSPLASIAS 2026

Leandro Augusto Silva

"NPR2-related Short Stature from a Tertiary Center: Phenotypic Spectrum with Focus on Monoallelic Cases"

Simão Pinho

"Osteogenesis Imperfecta Diagnosed After an Apparent Isolated Femoral Fracture in a Newborn: Could We Have Done Better?"

Sara Alves Costa

Same Name, Different Disease: Genotype-Phenotype Correlations in Adult Patients with Multiple Epiphyseal Dysplasia in a Tertiary Centre

Ariana C. Mendes

From Brachydactyly to Symphalangism: Exploring Genotype–Phenotype Correlations in GDF5- and NOG-Related Digital Malformations

Mafalda Saraiva Santos

Acromesomelic Dysplasia, Maroteaux Type: Clinical and Molecular Characterisation of Three Cases

20 ANOS

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PRÉMIO DEDICAÇÃO

Ao longo dos seus 20 anos de existência, a APOI tem tido a honra de poder contar com contributos inigualáveis, de dedicação incansável. O Prémio Dedicação representa um gesto de agradecimento sincero esublinha o impacto do trabalho destas personalidades na nossa comunidade. Elas são o elo de prestígio e reconhecimento entre a nossa história e o impacto que ela projeta na sociedade e personificam os valores e a missão da associação, trazendo consigo uma autoridade moral e institucional que valida o trabalho desenvolvido pela instituição.

Ao distinguir as personalidades que têm acompanhado e contribuído de forma excecional para a nossa Causa, a associação está não só a preservar o seu próprio legado, mas também a fortalecer a sua rede de influência e credibilidade junto de parceiros e da comunidade.

Os nossos sócios honorários são o coração pulsante da nossa história e o exemplo vivo da solidariedade que nos une. Mais do que um título de distinção, ser sócio honorário na nossa associação é um reconhecimento profundo a quem, com generosidade e entrega, dedicou o seu tempo e carinho a cuidar do próximo. Estas pessoas especiais não são apenas pilares de apoio; são faróis de esperança que iluminam o nosso caminho e nos inspiram a continuar a nossa missão.

Ter a sua presença e o seu nome ligados à nossa causa é uma honra que nos recorda, diariamente, que nenhuma caminhada se faz sozinho e que o amor ao próximo é a força mais poderosa que temos para transformar vidas.

Obrigada por abraçarem esta MISSÃO 💛

Margarida Custódio dos Santos

Manuel Cassiano Neves

PRÉMIO
ANUAL DE INVESTIGAÇÃO CLÍNICA NA ÁREA DAS DISPLASIAS ESQUELÉTICAS

HELOÍSA SANTOS
2026

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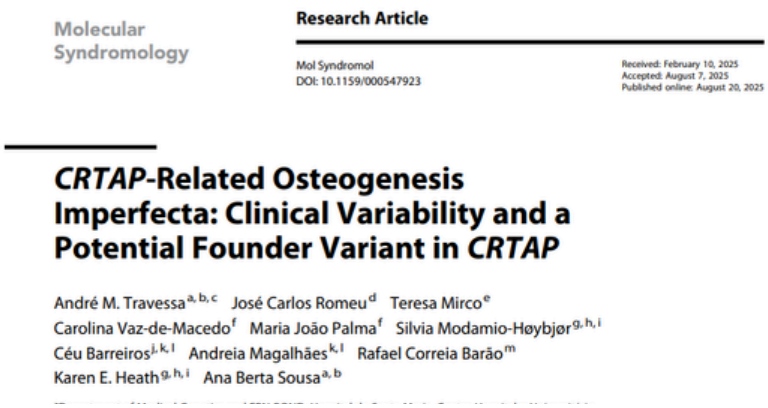

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CRTAP*-Related Osteogenesis Imperfecta: Clinical Variability and a Potential Founder Variant in *CRTAP

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BONE DYSPLASIAS 2026
SIMPÓSIO DE DISPLASIAS ÓSSEAS

COMUNIDADE DE DOENTES NO APOIO

À CIÊNCIA E INVESTIGAÇÃO

apoi associação portuguesa de osteogenese imperfeita

A **Associação Portuguesa de Osteogénese Imperfeita – APOI**, representa uma doença rara que provoca grande fragilidade óssea, levando a fraturas frequentes e deformações ósseas progressivas.

Sobre a organização

A nossa associação é uma organização nacional, voluntária e não lucrativa, reconhecida como IPSS em 2012, e inscrita como ONGPD pelo INR.IP desde 2015.

Sobre a Missão

A nossa **MISSÃO** consiste em melhorar por todos os meios possíveis a qualidade de vida dos portadores de Osteogénese Imperfeita (OI). Para isso, tentamos focar as nossas atividades no estímulo ao melhor conhecimento médico e à investigação, no ensino aos doentes e suas famílias como uma forma de os tornar parte ativa na promoção da sua própria saúde e na sensibilização da comunidade civil como uma forma de minimizar o estigma e melhorar a integração social.

Sobre a Osteogénese Imperfeita

A **Osteogénese Imperfeita (OI)**, tal como a maior partes das doenças ósseas raras, traz grandes e graves consequências para o dia-a-dia destes doentes, "arrancando-os" subitamente do seu contexto social e isolando-os por períodos de tempo prolongados, pelo que as consequências da doença vão muito além de simples fraturas e estendem-se no ponto de vista psicológico, familiar, escolar e de integração social.

uma instituição vocacionada para a *ciência* e *investigação* em prol do conhecimento médico e dos seus doentes

Apesar dos seus poucos recursos, a APOI tem promovido todos os esforços para estimular o interesse público e profissional pela OI, e tem-se afirmado como uma instituição de referência nas suas relações com a comunidade científica e com a indústria, muito em particular através do seu **Conselho Científico** e do seu **Gabinete de Apoio à Investigação**.

JUNTE-SE A NÓS

Junte-se à **Associação Portuguesa de Osteogénese Imperfeita** e beneficie de atualizações científicas em primeira mão, ficar ligado a uma rede de especialistas nacional e internacional e obter descontos e serviços nas nossas atividades e nas dos nossos parceiros.

Gabinete de Apoio à Investigação em Osteogénese Imperfeita

Gabinete Apoio à Investigação
Osteogénese Imperfeita

FAZEMOS MAIS PELOS SEUS DOENTES

FAZEMOS MAIS POR SI

COM O OBJETIVO DE MELHORAR A PARTICIPAÇÃO, QUALIDADE E QUANTIDADE DE INVESTIGAÇÃO RELACIONADA COM OI NO NOSSO PAÍS, O GAI-OI DESENVOLVE A CAPACITAÇÃO DOS DOENTES E ESTIMULA O SEU PAPEL NA INVESTIGAÇÃO, ATRAVÉS DA EDUCAÇÃO, APOIO LOGÍSTICO E ACOMPANHAMENTO LOCAL NOS HOSPITAIS E APOIA OS PROFISSIONAIS COM SECRETARIADO, MONITORIZAÇÃO DOS TRABALHOS E APOIO À FORMAÇÃO ESPECIALIZADA EM OI.

ASSOCIAÇÃO NACIONAL DE DISPLASIAS ÓSSEAS

ANDO Portugal – The National Association for Skeletal Dysplasias, founded on May 26, 2015, is a non-profit organization dedicated to supporting people living with skeletal dysplasias and their families. Our mission is to create and disseminate reliable and up-to-date information, offer guidance on socioeconomic, educational, legal, and public health matters, and contribute to medical and scientific research. ANDO is also committed to raising awareness to the challenges faced by people with skeletal dysplasias and rare diseases, promoting greater understanding and inclusion within society.

With over 300 members, ANDO Portugal has been officially recognized since 2017 by the National Institute for Rehabilitation (INR) as a Non-Governmental Organization for Persons with Disabilities (ONGPD). It is also part of an international collaborative network focused on research and advocacy, being a member of the European Patients' Academy on Therapeutic Innovation (EUPATI PT) the European Organisation for Rare Diseases (EURORDIS), and the European Reference Network on Rare Bone Diseases (ERN BOND).

Since its establishment, ANDO has developed numerous projects, initiatives, and events at both national and international levels, either independently or in partnership with other organizations. ANDO contributed to the scientific program of Bone Dysplasias 2026, reinforcing its commitment to advancing knowledge, collaboration, and patient-centered approaches in the field.

· EDUCATION · INNOVATION · RESEARCH · KNOWLEDGE

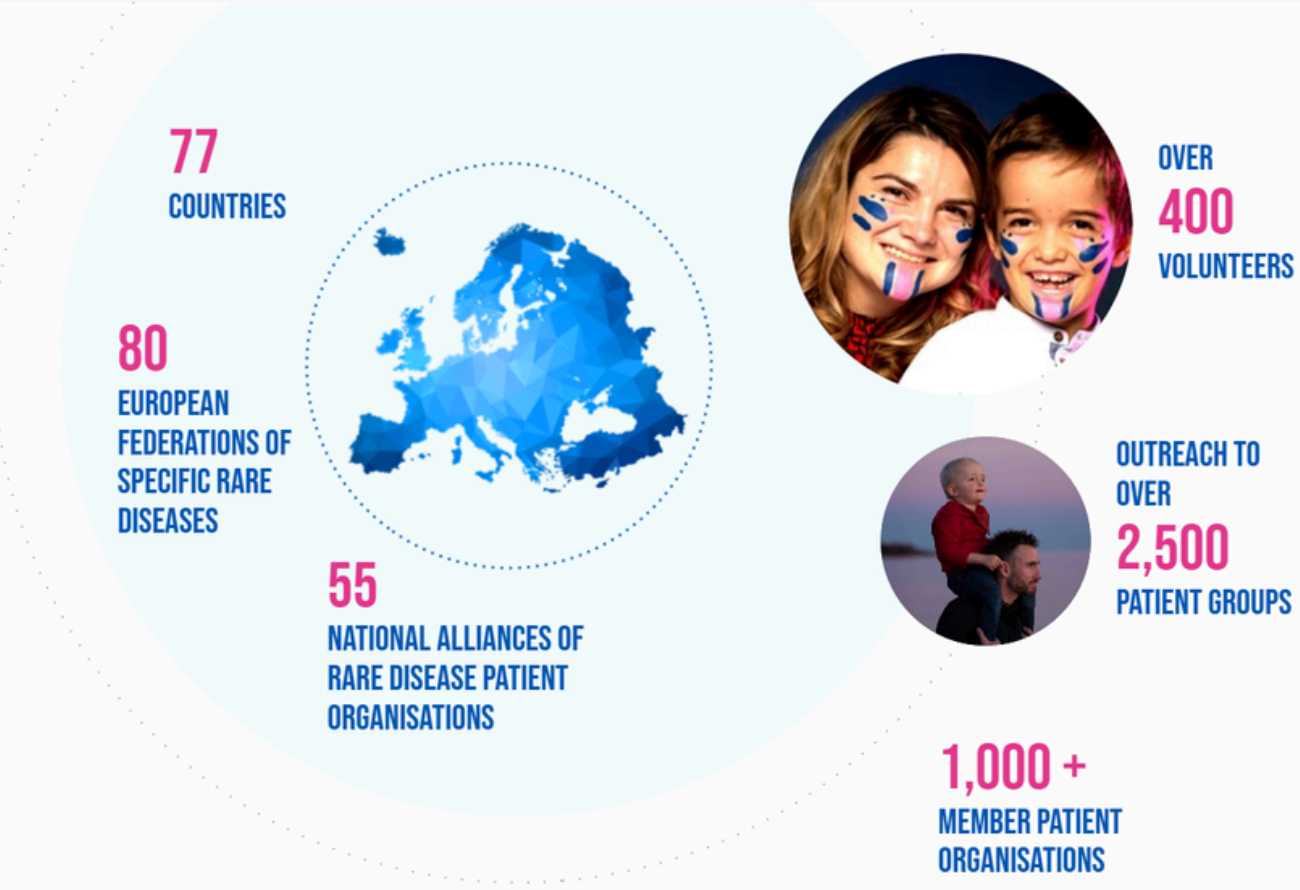
WWW.ANDOPORTUGAL.ORG

EURORDIS - Rare Diseases Europe is a unique, non-profit alliance of over 1,000 rare disease patient organisations from 77 countries that work together to improve the lives of over 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

Our mission is to work across borders and diseases to improve the lives of all people living with rare diseases to achieve their full potential, people living with a rare disease need to be:
- Recognised as equal citizens with their rights fully respected
- Diagnosed timely and accurately
- Supported with state-of-the-art medical and social care or cured
- Included in society in all aspects of life and enabled to live independently.

We are happy to support the Unbreakable Alliance Project and the Bone Dysplasia Symposium organized by our member organisation Associação Portuguesa de Osteogénese Imperfeita.



Osteogenesis Imperfecta Federation Europe

Osteogenesis Imperfecta Federation Europe (OIFE) is an umbrella association for organizations dealing with the rare genetic bone condition osteogenesis imperfecta (OI).

We are happy to support and endorse the important Simposium **Bone Dysplasias 2024**.
OIFE was first established in 1993 in the Netherlands. From 2022 we have been registered as a non profit in Belgium. Our mission is to connect and empower organizations, professionals and individuals to improve lives of people with OI.
Our ordinary member organizations are European, and our main activities are based in Europe. The Portugese OI-organization APOI is one of our valuable national member organizations. In addition to having members in Europe, we collaborate with OI-organizations around the world with the intention to exchange ideas, information and best practices. The OI-organization in Brazil (ANOI) is one of our associate member organizations, and OIFE has supported representatives from ANOI to attend this conference. We hope it will inspire new collaborations across the Atlantic!

At the moment, OIFE include 41 member organizations (20 European national organizations; 16 partner organizations - national OI-organizations outside Europe; and 5 supporting organizations).

OIFE is a member of EURORDIS - the umbrella for rare disease organizations in Europe, the European Rare Bone Forum and we recently became an affiliated member of the European Society of Endocrinology (ESE).
Our vision is children and adults with OI living active and independent lives - with access to competent healthcare and necessary social support.
Our most important projects at the moment include dissemination of results from the IMPACT survey, the Pain and OI project, patient involvement in research and development and advocating for access to better care, services and treatments for people with OI.

You can find more information about OIFE at www.oife.org

INVESTIGATOR MEETING

NOVEMBER 15TH, 2024 (ON ZOOM)
2 PM - 7 PM Central European Time
8 AM - 1 PM Eastern Standard Time

THE EVENT IS FREE
CME - the event is NOT CME accredited

GOALS OF THE EVENT

- Highlight new OI research
- Facilitate collaboration between clinical and basic researchers
- Provide a collaboration space for OI researchers in Europe and beyond
- Attract new people to OI research
- To support the younger generation of OI researchers

TARGET GROUPS

- The target group is primarily researchers and clinicians working with OI in Europe and other countries.
- Patient representatives from OI-organizations can attend if they have a special interest in research and development.
- Industry representatives are also welcome.

ABSTRACT SUBMISSION
DEADLINE 4 OCTOBER 2024

We invite your abstracts on any aspect of OI research, whether basic, translational or clinical. **A limited number of oral slots are available on the programme and priority will be given to abstracts describing novel research and hot topics related to OI.**

This is an opportunity for you to present and discuss your OI-related work with an international group of fellow OI-researchers in a relatively informal setting and without the costs and time involved in travelling to in-person meetings. The aim is to facilitate collaboration and development of research in OI.

ABSTRACT SUBMISSION FORM OPENS IN JUNE 2024

For more info:
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FEDERAÇÃO DAS DOENÇAS RARAS DE PORTUGAL

To be rare... is to be precious!

FEDRA – the Portuguese Federation of Rare Disease Associations – is a non-profit private social solidarity institution (IPSS) founded in 2008 with the aim of playing a vital role in supporting and representing people living with rare diseases in Portugal.

In 2008, a group of associations recognised the urgent need to join forces and speak with one voice in order to strengthen their impact and provide effective support to their members. Thus, FEDRA was born.

Since then, we have worked tirelessly to raise awareness of rare diseases, influence health policies, and defend the rights of those affected by these conditions. Our commitment is to foster unity and collaboration among the various associations dedicated to rare diseases, with the aim of improving quality of life and enhancing the support provided to people living with these unique conditions.

We believe that by joining forces, we can make a significant difference in the lives of those facing rare diseases.

FEDRA aspires to contribute to the building of a society founded on equal opportunities for all citizens. We believe that such a society can only be achieved through responsible and sustainable organisations recognised for the quality of their work. The effectiveness of this work is rooted in a vision centred on the active participation of the people we support, as well as on the promotion, monitoring, and defence of their social and citizenship rights.

The values and principles that underpin FEDRA are fundamental to fulfilling our mission: Authenticity, Credibility, Cooperation, Humanism, and Solidarity. These values guide our daily work and shape our commitment to the cause of rare diseases and the well-being of those affected by them.

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HUMAN GROWTH FOUNDATION

The Human Growth Foundation (HGF) is a global nonprofit organization dedicated to improving the lives of children and adults living with rare growth, bone, and endocrine conditions.

Founded in 1965, HGF works to increase awareness and advance care through research, education, patient support, and advocacy—ensuring that every individual has the opportunity to thrive, regardless of their diagnosis or circumstances.

Today, HGF serves a global community of more than 80,000 members, including patients and families, as well as physicians, nurses, researchers, and other healthcare professionals. Together, this network is united by a shared goal: to improve understanding, access to care, and quality of life for those affected by rare conditions.

In recent years, HGF has expanded its reach through a range of accessible and impactful initiatives. These include:

- Comprehensive multilingual website with trusted, evidence-based resources.
- National and international workshops, symposiums, and conferences;
- Collection of educational booklets, guides, and clinical toolkits for both families and healthcare providers;
- Growing library of video series and digital programs designed to make complex medical information easier to understand and apply.

Through these efforts, HGF provides not only reliable information but also meaningful support and a sense of community for individuals and families navigating rare conditions.

For more information, please visit www.HGFound.org.




Fundación ALPE Acondroplasia

The ALPE Achondroplasia Foundation, established in January 2000 by a group of committed families, stands as a pioneering entity in improving the quality of life for people affected by achondroplasia and other skeletal dysplasias that cause dwarfism, as well as their families.

Its fundamental mission lies in promoting the integral and harmonious development of each individual, actively driving full educational and socio-occupational inclusion.

The guiding principles that lead the work of the ALPE Achondroplasia Foundation are:

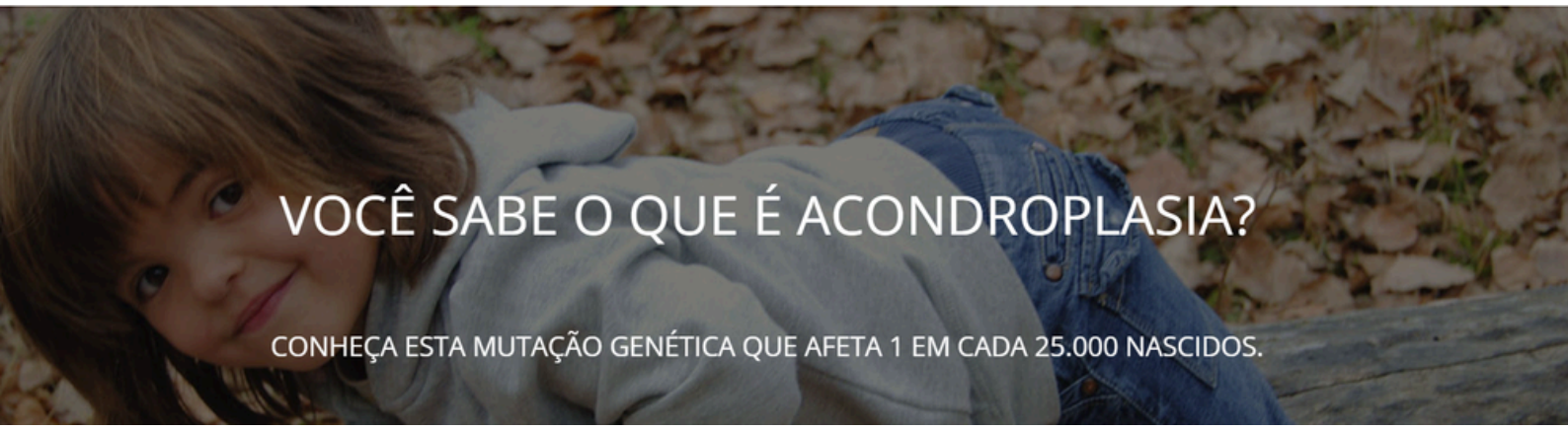
- Holistic Perspective on Health: Comprehensive approach to the physical, psychological, and social well-being of people with achondroplasia.
- Inclusive Education: Promotion of accessible and equitable educational environments that guarantee the full participation of all students.
- Promotion of Scientific and Academic Research: Impulse and support for studies in various disciplines that contribute to the knowledge and treatment of achondroplasia.
- Networking: Establishment and strengthening of strategic alliances with organizations, professionals, and families at national and international levels.

The Foundation's strategy is articulated through short, medium, and long-term objectives, covering crucial areas such as health, education, and socio-occupational fields. This multidimensional approach allows for addressing the specific needs of the community it serves in a comprehensive and sustainable manner.

The international relevance and commitment of the ALPE Achondroplasia Foundation are reflected in its status as a founding member of the Skeletal Dysplasias Alliance and its active participation in the European Reference Network on Rare Bone Diseases. It also maintains an active participation in Eurordis. These affiliations underscore its dedication to global collaboration and advancement in the field of skeletal dysplasias.

The ALPE Achondroplasia Foundation expresses its enthusiasm for joining this Unbreakable Alliance and being able to make our voices heard worldwide.

For more information: www.fundacionalpe.org



apoi

associação portuguesa de osteogenese imperfeita

JUNTE-SE A NÓS

VISITE APOI.PT

BONE DYSPLASIAS 2026

SIMPÓSIO DE DISPLASIAS ÓSSEAS

10 YEAR ALIANÇA INQUEBRÁVEL

OBRIGADO POR FAZER PARTE