

RELATÓRIO DE EXECUÇÃO

FINAL REPORT

BONE DYSPLASIAS

2026

SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

**BONE
DYSPLASIAS**
2026
SIMPÓSIO DE
DISPLASIAS
ÓSSEAS

RELATÓRIO DE EXECUÇÃO
FINAL REPORT

BONE DYSPLASIAS

Bone Dysplasias have genic origin and comprise a very heterogeneous group of rare diseases that mainly affect the development, structure, and composition of bones, cartilage, and dentin. They result in short stature, bone fragility or deformities, and frequently compromise the life and mobility of individuals.

Bone Dysplasias are an extremely important area within bone pathologies, but it has been poorly developed and recognized, leading to a lack of support and implementation of adequate measures, particularly in adult patients.



Hundreds of bone dysplasias are currently identified, both non-lethal and lethal, divided into several groups according to 4 criteria: phenotypic, radiological, biochemical, and genetic.

Diagnosis continues to be, in most cases, a challenge for professionals, with a global prevalence of 1 case per 5,000 births. Although it can be diagnosed during pregnancy, most are only identified after birth and postnatally, and it is common for the exact identification of the pathology to take years to occur.

Bone Dysplasias

IDENTIFIED BONE
DYSPLASIAS

771

CLASSIFIED IN
VARIOUS GROUPS

41

DIVIDED ACCORDING
TO SPECIFIC CRITERIA

4

PEOPLE LIVING WITH
BO IN PORTUGAL

1500



Bone Dysplasias

Walking Together

Considering the complexity and specificity of these diseases, any action in this area requires complex multidisciplinary and differentiation, involving patient representatives and multiple medical specialties and subspecialties - including medical genetics, rheumatology and pediatric rheumatology, endocrinology and pediatric endocrinology, physical medicine and rehabilitation, orthopedics, nephrology and pediatric nephrology, otorhinolaryngology, ophthalmology, cardiology and pediatric cardiology, pulmonology and pediatric pulmonology, radiology, stomatology, obstetrics and neonatology, as well as psychology professionals, social services, and senior diagnostic and therapeutic technicians.

In this regard, the Portuguese Association of Osteogenesis Imperfecta (APOI) and the Multidisciplinary Team for Bone Dysplasias at ULS Santa Maria, a member of the European Reference Network for Rare Bone Diseases (ERN-BOND), together with its affiliated centers (namely ULS Almada Seixal, ULS S José, and Hospital CUF Descobertas), join efforts to promote another edition of the UNbreakable Alliance Project.

Aiming to expand and strengthen this *UNbreakable* Alliance, this year we invited a new partner, the National Association of Bone Dysplasias (ANDO Portugal), to organize the Symposium. Furthermore, Hospital CUF Descobertas plays a prominent role as host of the scientific activities, welcoming national and international experts



much more than a project

The “ALIANÇA INQUEBRÁVEL” (UNbreakable Alliance) project was created in 2016 by the Portuguese Association of Osteogenesis Imperfecta – APOI, due to the Association's awareness of the difficulties in access and equality to healthcare in Osteogenesis Imperfecta (OI) and other Rare Skeletal Diseases. Its social and scientific responsibility, as well as the emerging need to implement measures that raise awareness for strategic action for 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, with hospital institutions, Scientific Societies, Patient Associations, Academic Institutions, the pharmaceutical and medical device industry, and Official Bodies playing a fundamental role for these issues

Thus, this innovative partnership project was born, in which its main and sole goal is the "improvement of quality of life for people with OI and other bone dysplasias". For the effect, it is underlined with development of various activities for scientific updates, health literacy, and fellowship.



An Alliance of multiple Partners

A project that involves the scientific and non-scientific Bone Dysplasias Community in a joint purpose where patients and professionals come together to reflect on the latest developments in rare bone diseases and define the best practices for the future.

Bone Dysplasias 2026: Bone Dysplasias 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.

PARTNER INSTITUTIONS



AFFILIATED CENTRES



COMMON GOALS

1. Create a space for reflection and debate at a national level focused on:

Development of excelling outcomes for professionals in the field, facilitating the implementation of scientific advances including:

- Debate on best practices in the diagnosis and multidisciplinary treatment of bone dysplasias and other rare bone diseases;
- Participation of international speakers and exchange of experience with other multidisciplinary reference centers, particularly for the Portuguese and Iberian reality, as well as improving care in this group of pathologies;
- Presentation of Portuguese clinical cases, case series, and national studies.

2. Health literacy, empowerment and capacity building for patients and families::

- Updates on the most recent treatments in Bone Dysplasias (BD);
- Capacity building for health decision-making;
- Capacity building strategies on how to deal with daily challenges related to Osteogenesis Imperfecta;

3. Present updates on ERN-BOND and the Portuguese teams as national reference centres for rare bone diseases;

4. Raise awareness among policymakers and other stakeholders about issues related to rare bone diseases.



BONE DYSPLASIAS

2026

**SIMPÓSIO DE
DISPLASIAS
ÓSSEAS**

Bone Dysplasias 2026: Bone Dysplasias Symposium brings together the entire community to establish a true global Alliance for rare skeletal diseases under the umbrella of the *UN*breakable Alliance. Many other partners have been invited to join us for this project that will promote a space of opportunities for specialists and patients to meet, discuss, work together, share experiences, and develop key strategies to implement new advances.



When

21st to 23rd may 2026

Working with a scientific committee composed of Portuguese doctors and scientists and patient representatives, this **UMbreakable Alliance** has created an inspiring agenda to bring to our country the most recent updates and developments on Bone Dysplasias.



Where

CUF Descobertas Hospital welcomed participants for the first two days (May 21 and 22), whose target audience was predominantly healthcare professionals from all specialty areas dealing with bone dysplasias and other rare bone diseases – including geneticists, orthopedists, pediatricians, physiatrists, physiotherapists, endocrinologists, rheumatologists, radiologists, internists, dentists, nurses, psychologists, etc.

The third day (May 23) was particularly dedicated to people with Bone Dysplasias and their families and took place at the Lisbon Oceanarium.

Both auditoriums are located in one of the city's most prestigious areas, Parque das Nações, which with its recreational and leisure spaces, additionally allowed for socialization among participants.



21st and 22nd May

Hospital CUF Descobertas

Auditório



23rd May

Oceanário de Lisboa

Sala

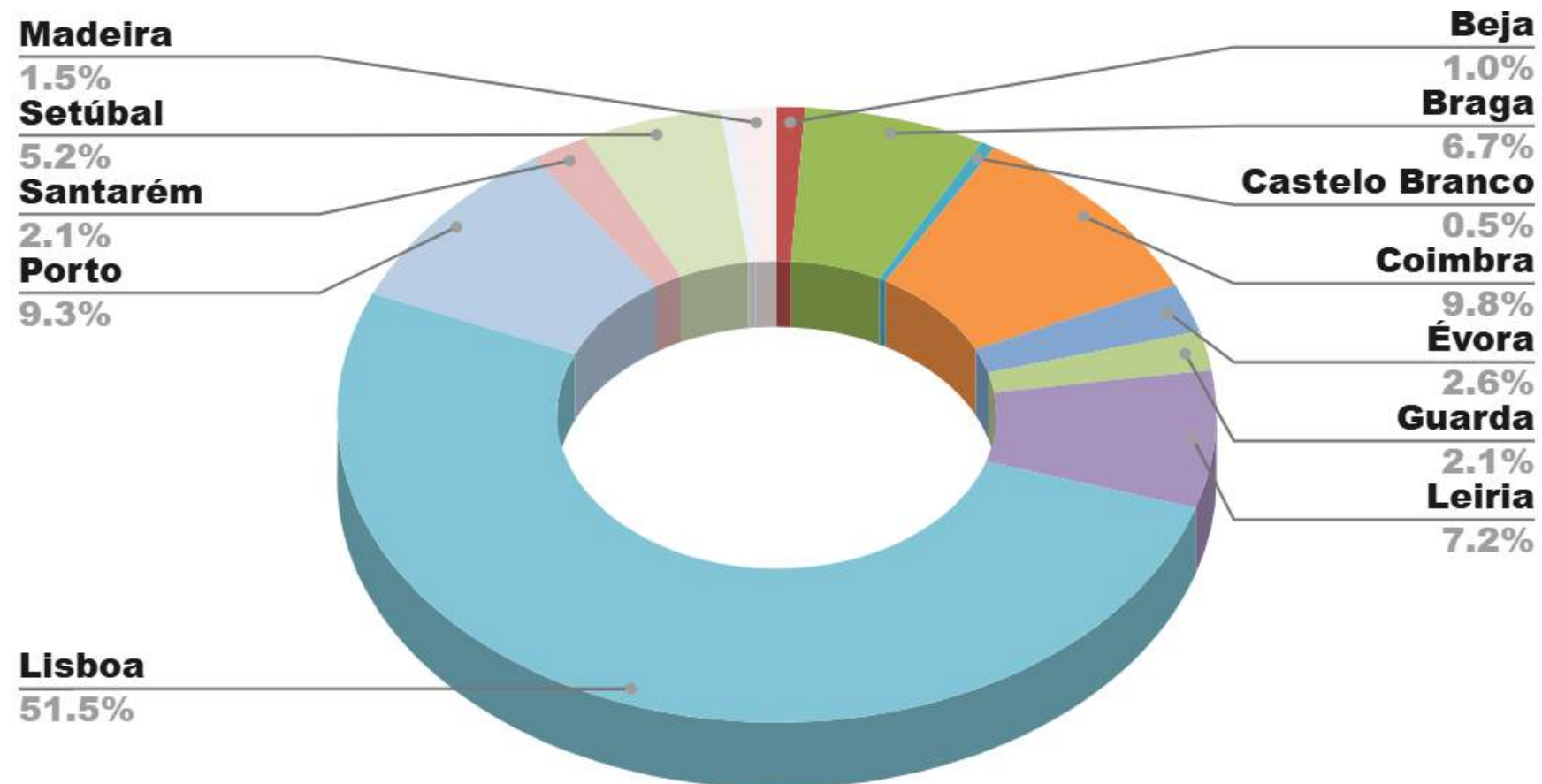


Who

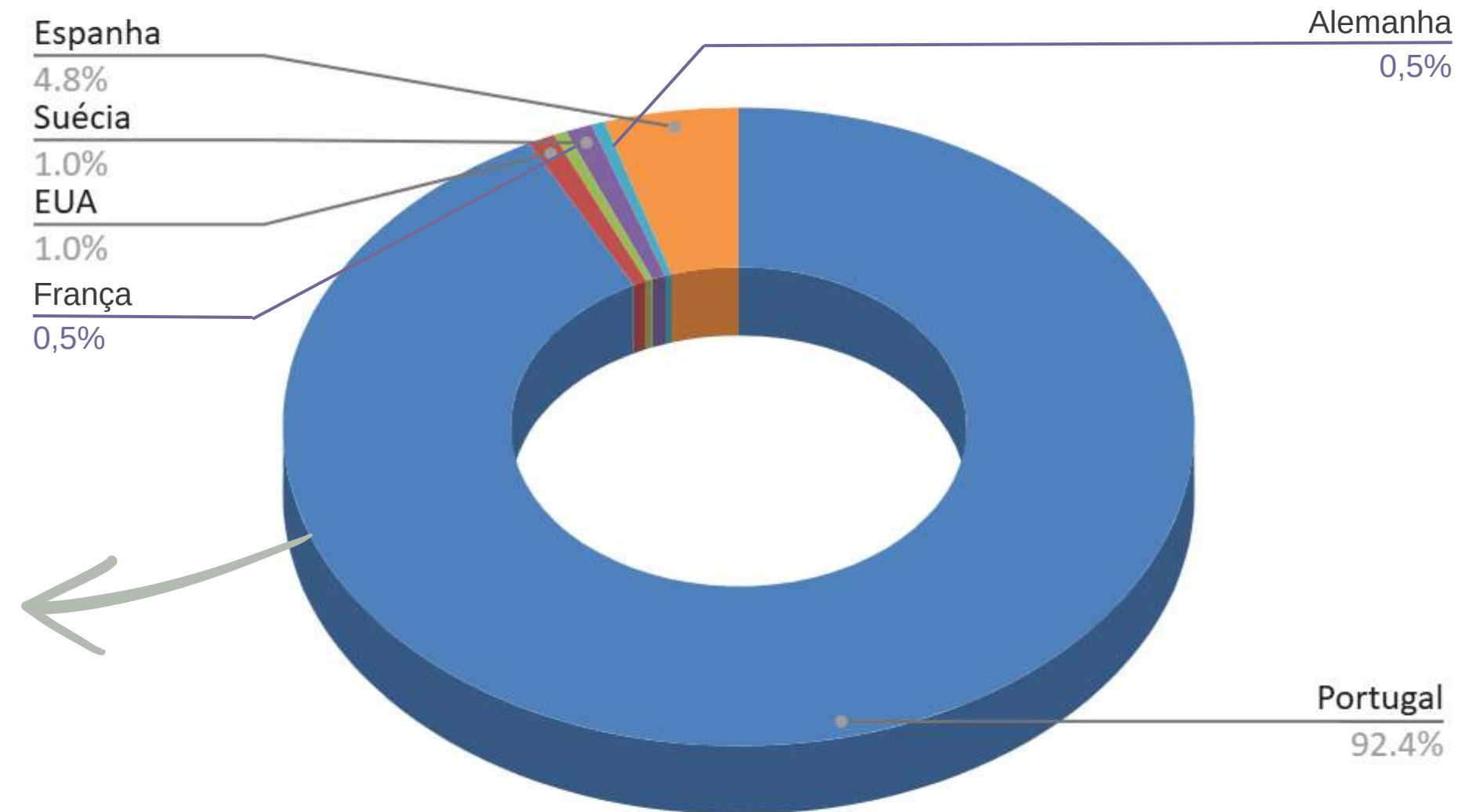
The project had national and international scope and involved **210 participants**.

With multiple renowned and prestigious guests from all parts of the country and the islands, it also included several foreign participants, namely from the United States of America, Germany, Sweden, France, and Spain.

PORTUGUESE DISTRICT OF ORIGIN



COUNTRY OF ORIGIN



BONE 2024 DYSPLASIAS

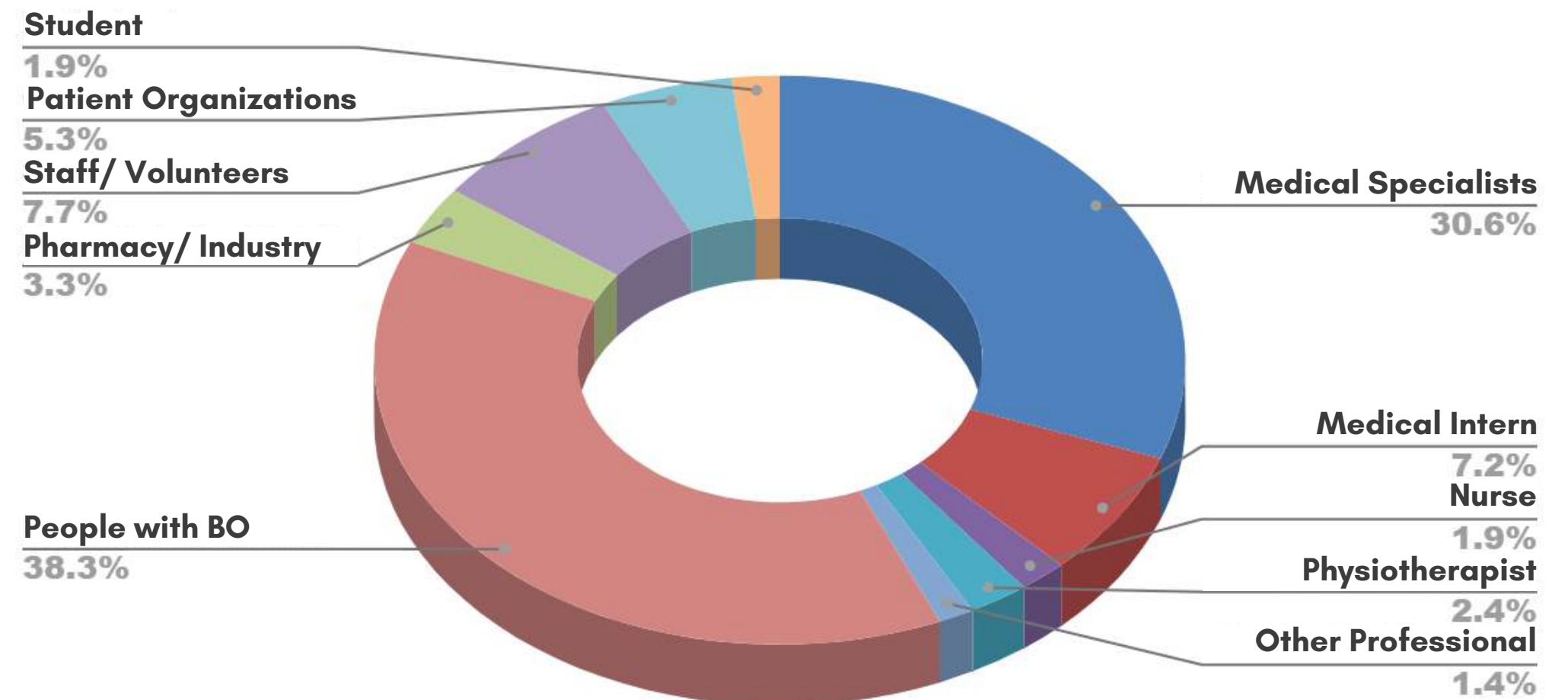
SIMPÓSIO DE DISPLASIAS ÓSSEAS

An excellent opportunity to establish connections, update knowledge and develop the network of contacts among the various types of participants.

The enormous participation of people with bone diseases and their families stands out, highlighting the importance of literacy for the development of better health and quality of life.

Who

PARTICIPATION CATEGORIES



ORGANIZING COMMITTEE

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



PARTNERSHIPS AND SUPPORT

Bone Dysplasias 2026 establishes itself as a scientific milestone of excellence with profound recognition at national and international levels. More than a medical meeting, the event stands out as an inspiring model of multidisciplinary cooperation and joint work between healthcare professionals and patients, with the latter being the main drivers and co-promoters of the meeting – a unique involvement that ensures, in practical terms, patient-centre approaches on all fronts. The relevance and impact are firmly validated by the institutional sponsorship of multiple globally recognized organizations in the field of rare diseases and, particularly, rare bone diseases, as well as by the scientific sponsorship of the main medical societies from the various specialties involved.



INSTITUTIONAL PARTNERS



Oceanário de Lisboa
Sempre diferente.



com o apoio da Freguesia Parque das Nações





SCIENTIFIC PARTNERS



SPNP
Sociedade Portuguesa
de Nefrologia Pediátrica



PARTNERSHIPS AND SUPPORT

As a genuinely solidarity-driven project, the event commits to involving the whole community and establish a true global alliance, It ensures the inclusive participation of patients, families, students and healthcare professionals through a social responsibility program that, thanks to the commitment of sponsors and social partners, enabled financial support. This provided a significant reduction of logistical costs and collection of in-kind donations, allowing its implementation with minimal financial impact for the association and a symbolic cost for participants.



PREMIUM PARTNERS





SUPPORT AND PATRONAGE

Gold

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Martins**

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≈ desde 1837 ≈



Grupo CaixaBank

VOLUNTARIADO

SUPPORT AND PATRONAGE



SUPPORT AND PATRONAGE



SUPPORT AND PATRONAGE



SUPPORT AND PATRONAGE



CASH & CARRY

EM CONTEXTOS HOSPITALARES E LARES,
CADA REFEIÇÃO CONTA.

Soluções Prontas

Conheça as nossas
Soluções Alimentares aqui:



Solicite uma proposta através de
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PENNE COM
ESPINAFRES E RICOTA
Soluções Prontas
RECHEIO



DELÍCIA DE CHOCO
prontas



ARROZ DE PATO





PUBLICITY

In a joint effort on behalf of people with bone dysplasias, several entities joined this Alliance contributing to the publicity of the event:



ANDO PORTUGAL | DISPLASIAS ÓSSEAS | PONTES | COLBIOS | NOTÍCIAS | EVENTOS | RECURSOS | [FAÇA PARTE](#)



BONE DYSPLASIAS 2026
21-23 Maio | May
Lisboa

Bone Dysplasias 2026: Simpósio Displasias Ósseas 21-23 maio

[Home](#)[Eventos](#) > [Eventos Anteriores](#) Bone Dysplasias 2026: Simpósio Displasias Ósseas 21-23 maio

O Bone Dysplasias 2026 | Simpósio de Displasias Ósseas, vai realizar-se em Lisboa entre 21 e 23 de maio de 2026. Trata-se de um evento de referência dedicado às **displasias ósseas** que reunirá profissionais de saúde, investigadores, estudantes das áreas de saúde, associações, pessoas com displasias ósseas e as suas famílias, num espaço de partilha, atualização científica e cooperação multidisciplinar.



PARQUE DAS NAÇÕES
JUNTA DE FREGUESIA LISBOA

[INÍCIO](#) [FREGUESIA](#)

Simpósio sobre Displasias Ósseas no PN

17-ABR-2026





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EVENTOS E CONGRESSOS

 maio 2026

Bone Dysplasias 2026 – Simpósio de Displasias Ósseas

De Quinta-feira, 21 maio 2026
Até Sábado, 23 maio 2026

Bone Dysplasias 2026 – Simpósio de Displasias Ósseas



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CALENDÁRIO

Bone Dysplasias 2026 Simpósio de Displasias Ósseas
Local: Lisboa
Dia: 21 de Maio de 2026 a 23 de Maio de 2026
Organização: APOI / Equipa de Displasias Ósseas da ULS Santa Maria / ANDO Portugal
Temática: Displasias Ósseas

D	S	T	Q	Q	S	S
	1	2	3	4	5	6
7	8	9	10	11	12	13
14	15	16	17	18	19	20
21	22	23	24	25	26	27
28	29	30				



CANTIC
CRITIC Amadora

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« **Todos os Eventos**

Este evento já decorreu.

Bone Dysplasias 2026: Simpósio de Displasias Ósseas



aA | | Português

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BONE DYSPLASIAS 2026 PRAZOS PROLONGADOS

Atualizado : 16/04/2026

BONE DYSPLASIAS 2026 - PRAZOS PROLONGADOS

SÓ ATÉ 26 ABRIL

As inscrições para o Bone Dysplasias 2026 continuam abertas, e só até 26 de abril é possível garantir a sua participação por um valor reduzido.

VOLUNTEERING

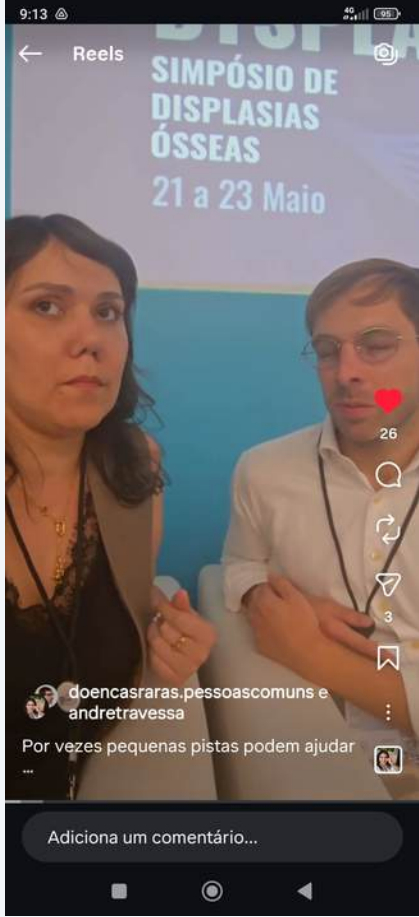
The strong sense of social responsibility was one of the most remarkable pillars of Bone Dysplasias 2026, reflected in the collective commitment to making the project accessible, inclusive and profoundly human. This purpose only came to life thanks to the exceptional and tireless work of the volunteers, who demonstrated admirable dedication in all phases of its execution, ensuring that every logistical and operational detail ran perfectly. Within this spirit of commitment, we express a very special thanks to the fabulous group of volunteers from Banco BPI. They ensured with immense professionalism and care essential tasks at the secretariat, catering support and close assistance to all participants. Their presence and support throughout the three days of the Symposium were absolutely vital to the success and to the welcoming atmosphere that characterized the event.



VOLUNTARIADO



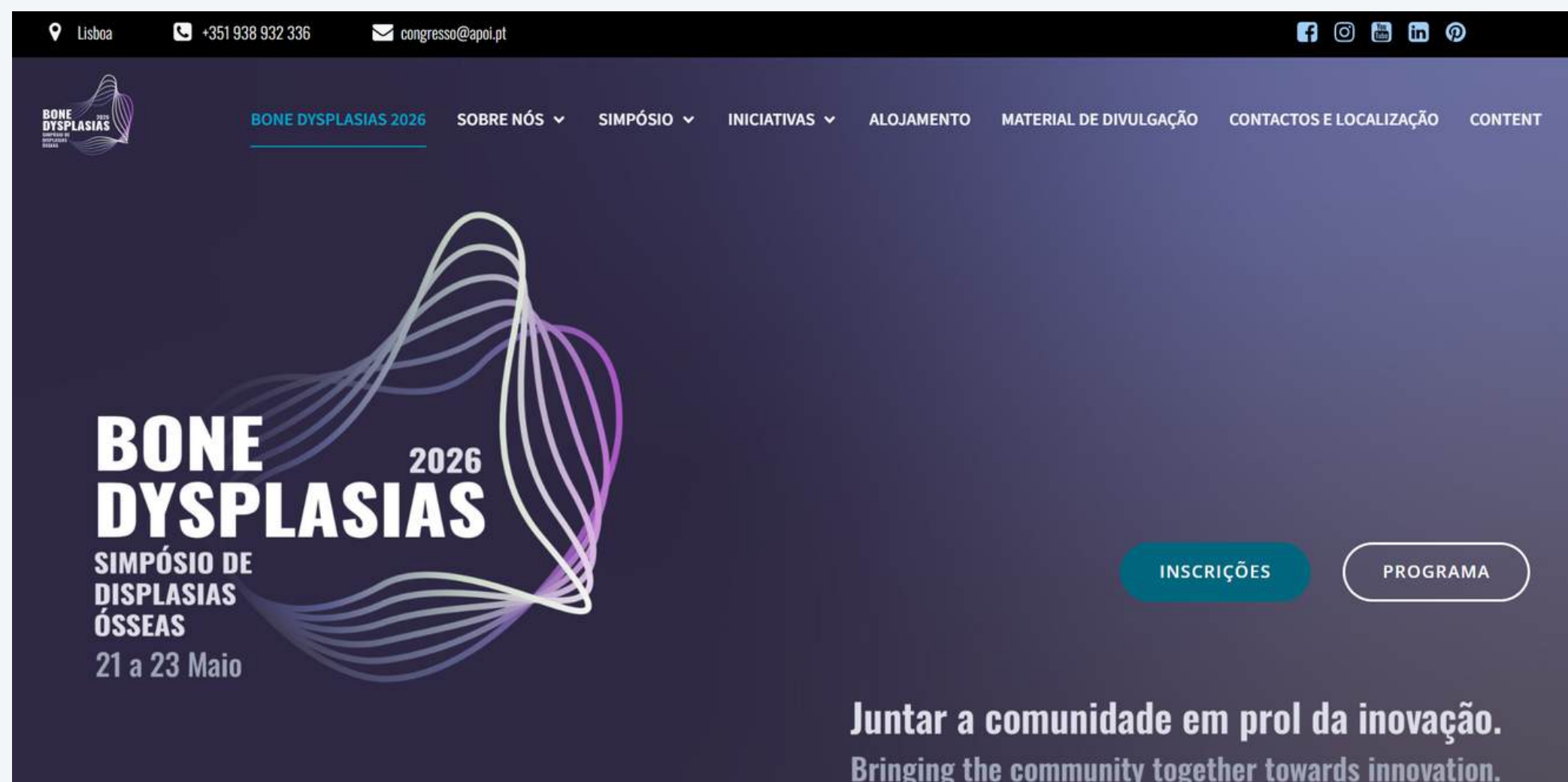
PUBLICITY



ACTIVITIES AND PROGRAM

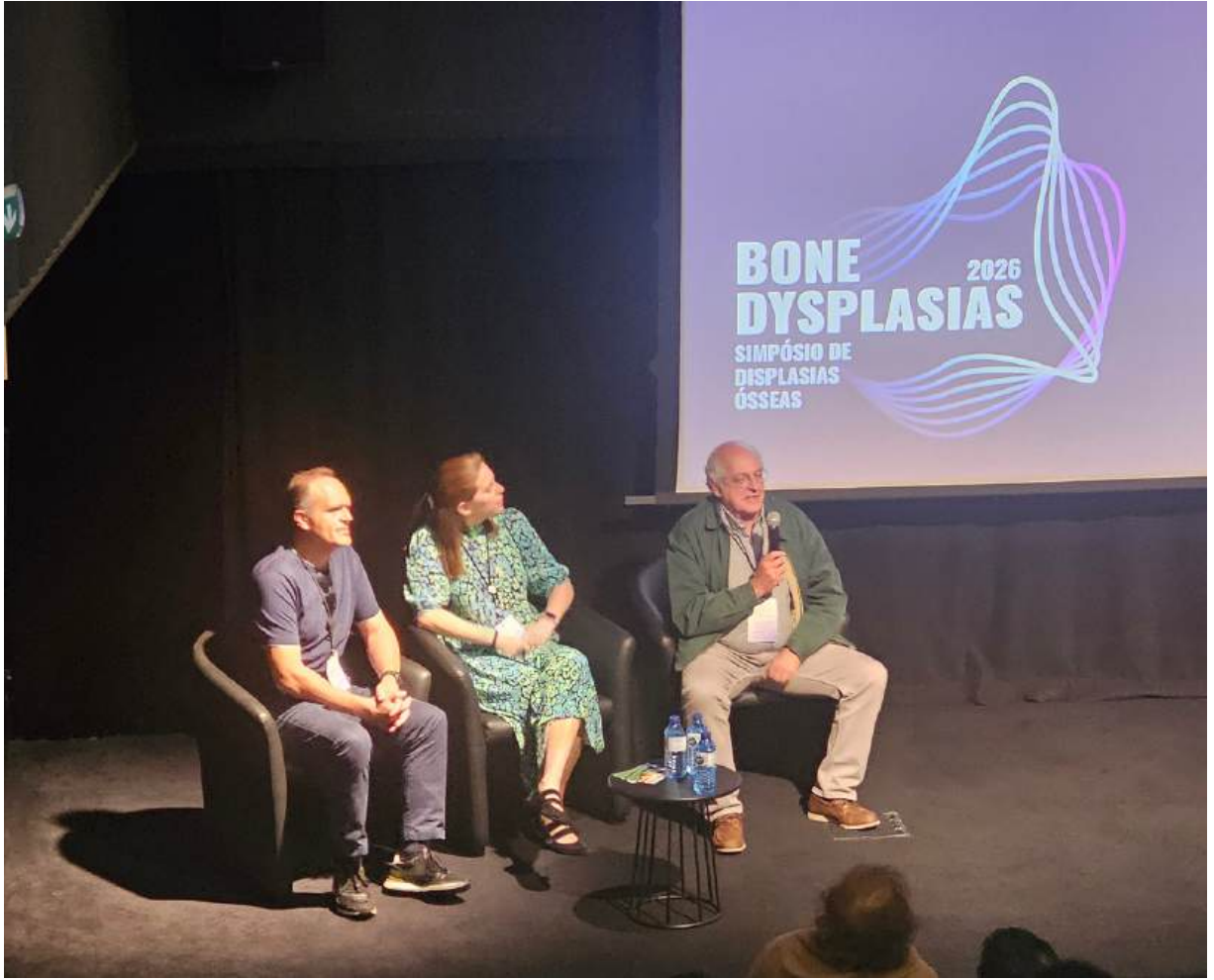
Working with a scientific committee composed of Portuguese doctors and scientists and patient representatives, this *UN*breakable Alliance composed an inspiring agenda to bring to our country the most recent updates and developments on Bone Dysplasias.

The immense environmental concern and responsibility led us to opt for predominantly digital publicity and making information available through a dedicated event website.



bonedysplasias.apoi.pt





PROGRAMA | PROGRAMME

Quinta-feira, 21 de maio de 2026

14:00-14:15	Abertura <i>Welcome</i> 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	<u>Comunicações orais – Casuística e Casos Clínicos I Oral communications – case series and clinical cases </u> Moderadores: Patrícia Almeida Dias; Ricardo Henriques
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 <i>Human Growth Foundation Courtesy Session</i> 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 <i>Practical Aspects for Osteogenesis Imperfecta</i> Palestrante: Miguel Rodriguez Molina - Fisioterapia em Adultos com Acondroplasia : Estratégias Funcionais e Gestão de Energia Baseadas na Evidência <i>Physiotherapy for Adults with Achondroplasia: Evidence-Based Functional Strategies and Energy Management</i> Palestrante: Cláudia Aguiar
12:30-12:50	Prémio Dedicção – 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

 Click the Link!

14:00-14:50	Ortopedia nas Displasias Esqueléticas (Pediatria e Adultos) <i>Orthopedics in Skeletal Dysplasias (Pediatrics & Adults)</i> Moderador: Ricardo Maia - Alongamento Osseo na Era Intramedular <i>Limb Lengthening in the Intramedullary Era</i> Palestrante: Tah Pu Ling - Aumento Osseo na Osteogénese Imperfeita <i>Bone Augmentation in Osteogenesis Imperfecta</i> Palestrante: Zagorka Pejcin - Manejo das Deformidades da Coluna <i>Management of Spinal Deformities</i> Palestrante: Jorge Mineiro
14:50-15:40	<u>Comunicações orais – Casuísticas e Casos Clínicos II</u> <u>Oral communications – case series and clinical cases II</u> Moderadores: Anabela Bandeira, João Campagnolo
16:00-17:05	<u>Comunicações Orais – Casuísticas e Casos Clínicos III</u> / <u>Oral Communications – Case Series and Clinical Cases III</u> Moderadores: Renata d'Oliveira; Andreia Martins
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



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)
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



MAIO MÊS DE SENSIBILIZAÇÃO
OSTEOGÉNESE IMPERFEITA

6 MAIO
hoje celebramos,

DIA INTERNACIONAL DA
OSTEOGÉNESE
IMPERFEITA



associação portuguesa de
osteogénese imperfeita

#WISHBONEDAY

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)



SOLIDARITY INITIATIVES

With a strong sense of social responsibility, the *UNbreakable* Alliance Project Team extended its action to the implementation of solidarity activities to support the patient community.

The Campaigns for the Allocation of 1% of Income Tax to the Portuguese Osteogenesis Imperfecta Association (**Tax ID 508 902 924**) were reinforced, as well as the promotion of awareness-raising actions for Osteogenesis Imperfecta, through the campaign "**May - Osteogenesis Imperfecta Awareness Month**".



IRS SOLIDÁRIO
Há um quadro mágico que torna sonhos em realidade!

apo
associação portuguesa de osteogénese imperfeita

AJUDAR MUITO, SEM GASTAR NADA

DEPOIS DE VALIDAR AS SUAS FATURAS NO PORTAL DAS FINANÇAS, AJUDE-NOS A MELHORAR A QUALIDADE DE VIDA DAS PESSOAS COM OSTEOGÉNESE IMPERFEITA

508 902 924

MODELO 3 **CAMPO 11**

INSTITUIÇÕES DE PARTICULARIDADE SOCIAL

DIA 6 DE MAIO
DIA INTERNACIONAL DA OSTEOGÉNESE IMPERFEITA

VESTE AMARELO,
USA UMA FLOR AMARELA NO CABELO
USA UMA GRAVATA AMARELA
USA UM LENÇO AMARELO

—
CELEBRA A VIDA

—
PORQUE DESPERTAR CONSCIÊNCIAS,
FAZ A DIFERENÇA

MAIO
MÊS DE
SENSIBILIZAÇÃO
OSTEOGÉNESE IMPERFEITA



apo
associação portuguesa de osteogénese imperfeita

#WISHBONEDAY
#1Voice4OI

AWARD

BD2026

With the aim of stimulating scientific production in the area of dysplasias and rare bone diseases and recognizing the merit of the 24 papers presented, the Organization of **Bone Dysplasias 2026: Bone Dysplasias Symposium** implemented the **Bone Dysplasias 2026 Award** for the best paper. Five **Honorable Mentions** were also awarded.



PRÉMIO MELHOR TRABALHO

Vencedor

Katerine Torres

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



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



DEDICATION AWARD

20 YEARS

Associação Portuguesa de Osteogénese Imperfeita

Throughout its 20 years of existence, APOI has had the honor of counting on unparalleled contributions of tireless dedication. The Dedication Award represents a gesture of sincere gratitude and underlines the impact of these individuals' work in our community. They are the link of prestige and recognition between our history and the impact it projects on society, and they embody the values and mission of the association, bringing with them a moral and institutional authority that validates the work developed by the institution.

By honoring the individuals who have accompanied and contributed exceptionally to our Cause, the association is not only preserving its own legacy, but also strengthening its network of influence and credibility with partners and the community.

Our honorary members are the beating heart of our history and the living example of the solidarity that unites us. More than a title of distinction, being an honorary member of our association is a profound recognition of those who, with generosity and dedication, devoted their time and care to looking after others. These special people are not just pillars of support; they are beacons of hope that illuminate our path and inspire us to continue our mission.

Having their presence and their name linked to our cause is an honour that reminds us daily that no journey is made alone and that love for others is the most powerful force we have to transform lives.

Thank you for embracing this MISSION 🧡



Margarida Custódio dos Santos



Manuel Cassiano Neves



ANNUAL CLINICAL RESEARCH AWARD IN THE AREA OF SKELETAL DYSPLASIAS

The Organization also supported the promotion and award ceremony of the **Heloísa Santos Award** (Annual Clinical Research Award in the Area of Skeletal Dysplasias), which took place during the awards ceremony.

PRÊMIO ANUAL DE INVESTIGAÇÃO CLÍNICA NA ÁREA DAS DISPLASIAS ESQUELÉTICAS

HELOÍSA SANTOS 2026

Vencedor

André Travessa

Molecular
Syndromology

Research Article

Mol Syndromol
DOI: 10.1159/000547923

Received: February 10, 2025
Accepted: August 7, 2025
Published online: August 20, 2025

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

André M. Travessa^{a,b,c} José Carlos Romeu^d Teresa Mirco^e
Carolina Vaz-de-Macedo^f Maria João Palma^f Silvia Modamio-Hoybjør^{g,h,i}
Céu Barreiros^{j,k,l} Andreia Magalhães^{k,l} Rafael Correia Barão^m
Karen E. Heath^{g,h,i} Ana Berta Sousa^{a,b}



RESULT ANALYSIS

The Bone Dysplasias Symposium has come to an end, leaving a profound mark at the academic, scientific and social levels.

Now, it's time to have a look at the results!

For APOI and the Organizing Committee, listening to those who were with us is the key to understanding whether we exceeded your expectations and to doing even better at future events. Therefore, we challenged participants to share their opinion through a Satisfaction Questionnaire focused on organization, hospitality, the scientific program and the independence of the sessions.

A very public thank you to all who collaborated with us in this evaluation!



BD2026 - Questionário de Satisfação

Caro(a) Participante,

Vimos, antes de mais, agradecer a sua participação no Simpósio Bone Dysplasias 2026, a qual foi fundamental para o sucesso do mesmo!

Terminado o Simpósio, é agora o momento de refletir e fazer a sua avaliação.

É muito importante para a APOI, para a Equipa Multidisciplinar de Displasias Ósseas da ULS Santa Maria e para a restante Organização, saber a sua opinião sobre o evento, para:

- Podermos avaliar em que medida satisfizemos as suas expetativas;
- Melhorarmos os futuros eventos.

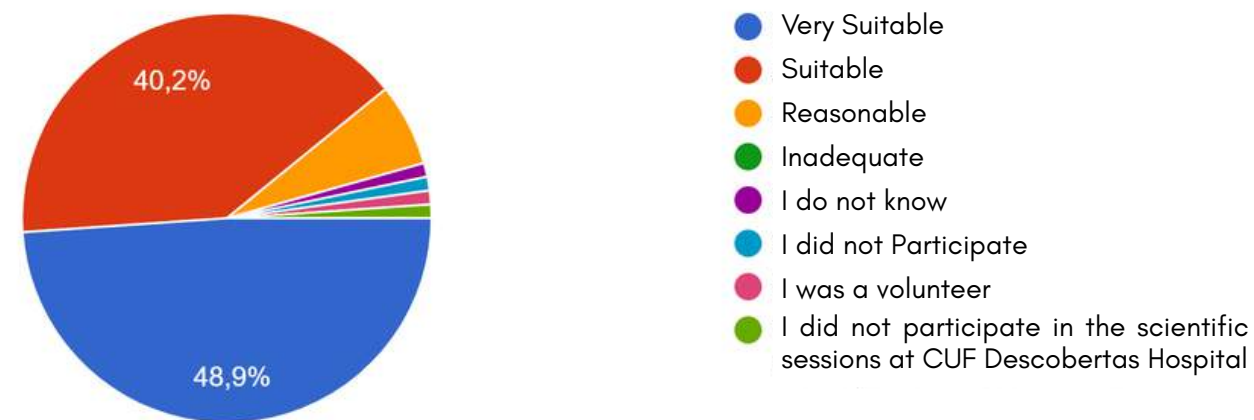
Como participante do BD2026 solicitamos que nos forneça a sua opinião sobre a organização, acolhimento, refeições, programa científico, adequação das sessões e independência científica do simpósio através do preenchimento deste formulário

WHAT PARTICIPANTS SAY - SESSIONS OF MAY 21 AND 22

Suitability of the format and sessions

THE FORMAT AND METHODOLOGY USED FOR THE SESSIONS AND ORAL COMUNICATIONS OF BONE DYSPLASIAS 2026 SYMPOSIUM WAS:

92 answers



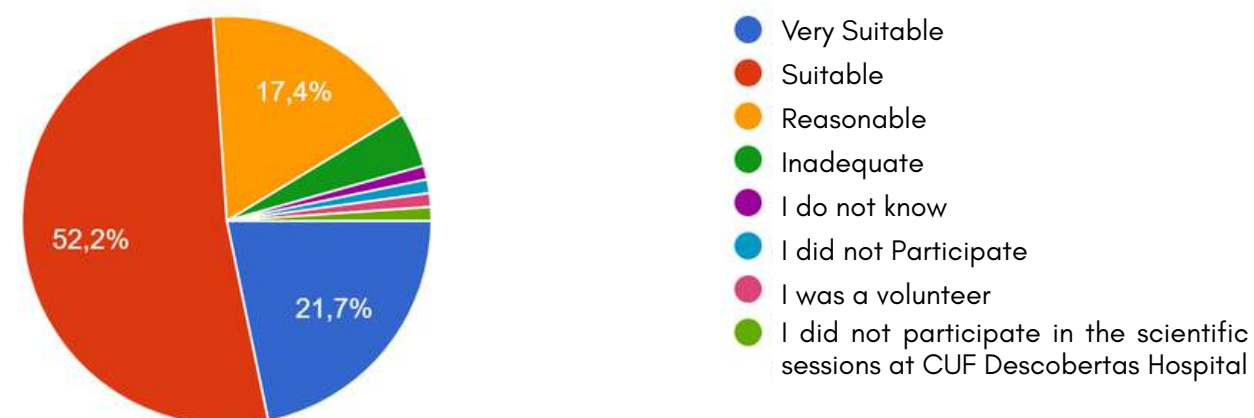
In general, the DURATION OF THE SESSIONS OF BONE DYSPLASIAS 2026 SYMPOSIUM WAS:

92 answers



The time provided for QUESTIONS AND DISCUSSION was:

92 answers



In general, did the SPEAKERS AND MODERATORS of the Symposium demonstrate scientific quality, clarity, and good communication skills?

92 answers



WHAT PARTICIPANTS SAY - SESSIONS OF MAY 21 AND 22

Suitability of the format and sessions

Did the EDUCATIONAL GOALS of Bone Dysplasias 2026 Symposium meet your expectations?

92 answers



Did your participation in Bone Dysplasias 2026 Symposium allow you to ACQUIRE NEW KNOWLEDGE?

92 answers



Was the Symposium USEFUL for your professional activity (in case you are an healthcare professional) or for your knowledge of the disease and its impact (in case you have a personal relation with the disease)?

92 answers



What is your overall assessment of the SCIENTIFIC PROGRAM of the Symposium?

92 answers

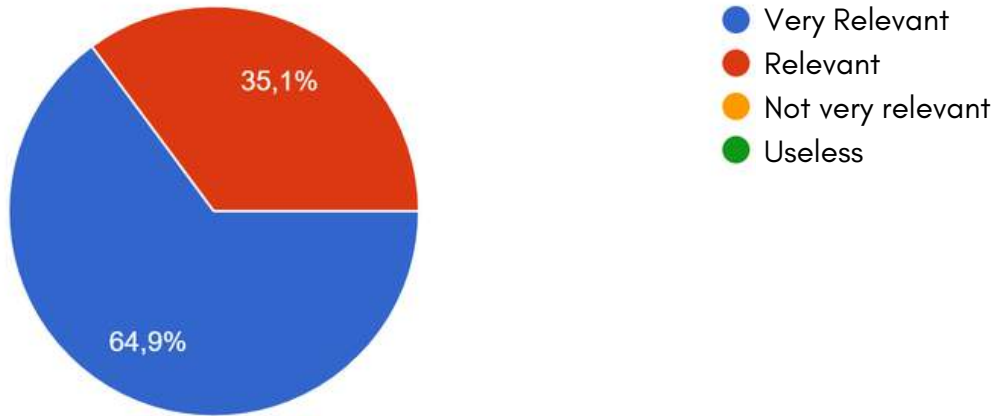


WHAT PARTICIPANTS SAY - SESSIONS OF MAY 23

Suitability of the format and sessions

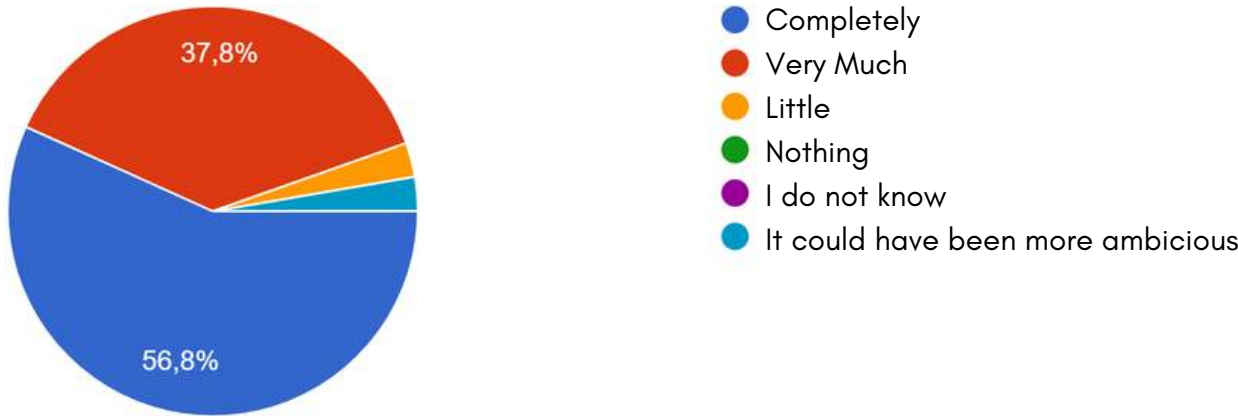
Sessions in Health Literacy in the Lisbon Oceanarium. What is the general RELEVANCE of the topics covered?

37 answers



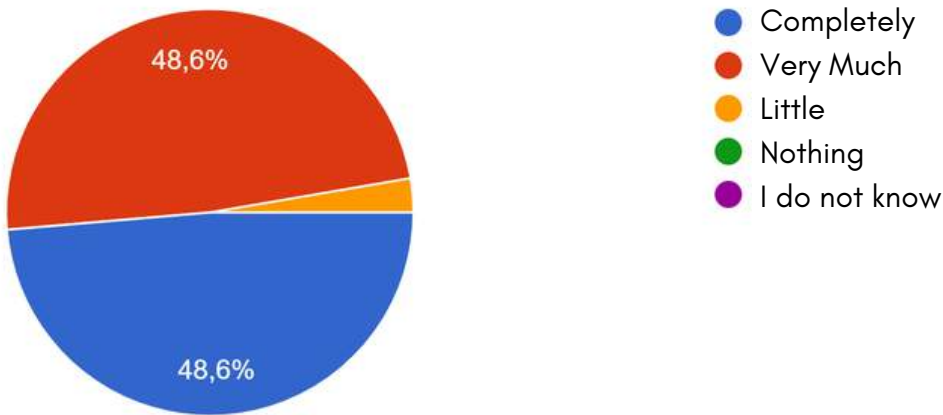
Did the EDUCATIONAL GOALS of Bone Dysplasias 2026 Symposium meet your expectations?

92 answers



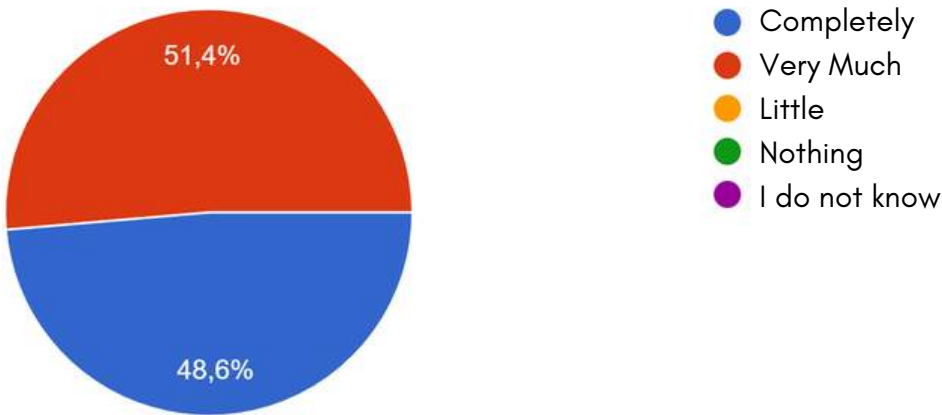
Do you consider that the ADEQUACY OF THE PROGRAM'S CONTENT corresponded to the proposed objectives?

37 answers



In general, was the INFORMATION PRESENTED at Bone Dysplasias 2026 well-balanced and consistently supported by valid scientific evidence?

37 answers

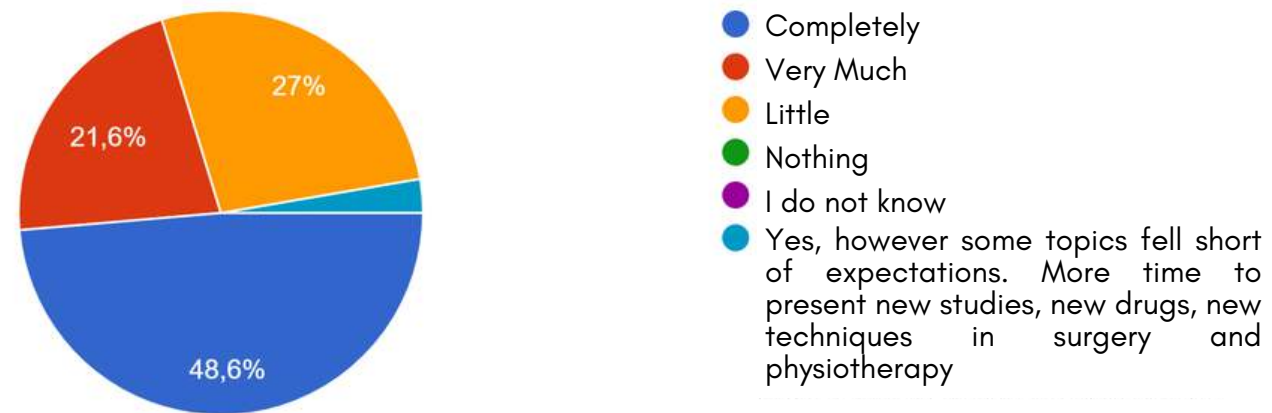


WHAT PARTICIPANTS SAY - SESSIONS OF MAY 23

Suitability of the format and sessions

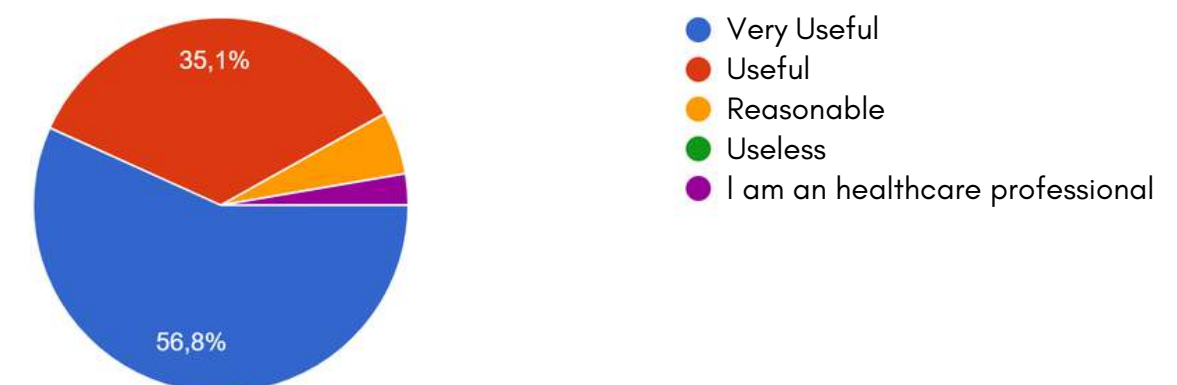
Did your participation in Bone Dysplasias 2026 Symposium allow you to ACQUIRE NEW KNOWLEDGE? ?

37 answers



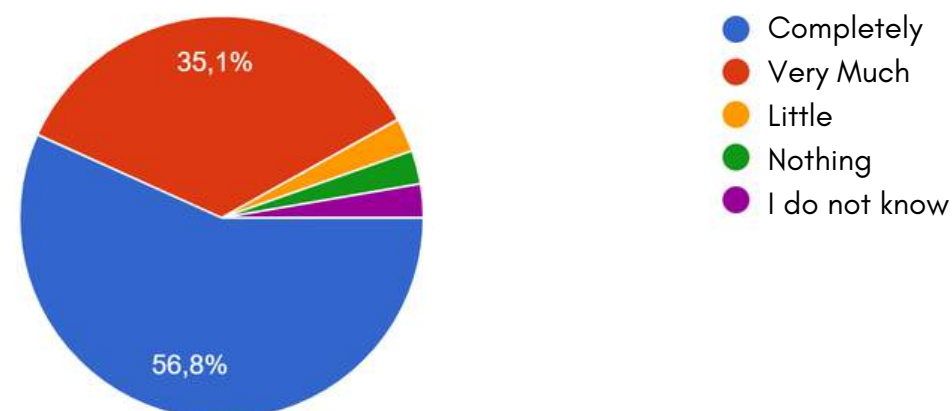
Was the Symposium USEFUL for your knowledge of the disease and its impact (in case you have a personal relation with the disease)?

37 answers



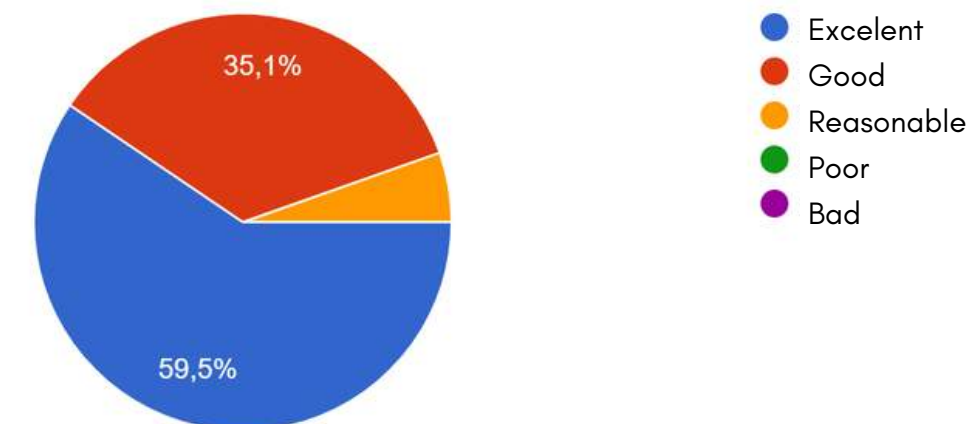
Ensure impartiality and scientific rigor it is one of our priorities. Do you consider the information presented and discussed kept scientific impartiality and was FREE OF COMERCIAL BIASES?

37 answers



What is your overall assessment of the HEALTH LITERACY PROGRAM of the Symposium?

37 answers

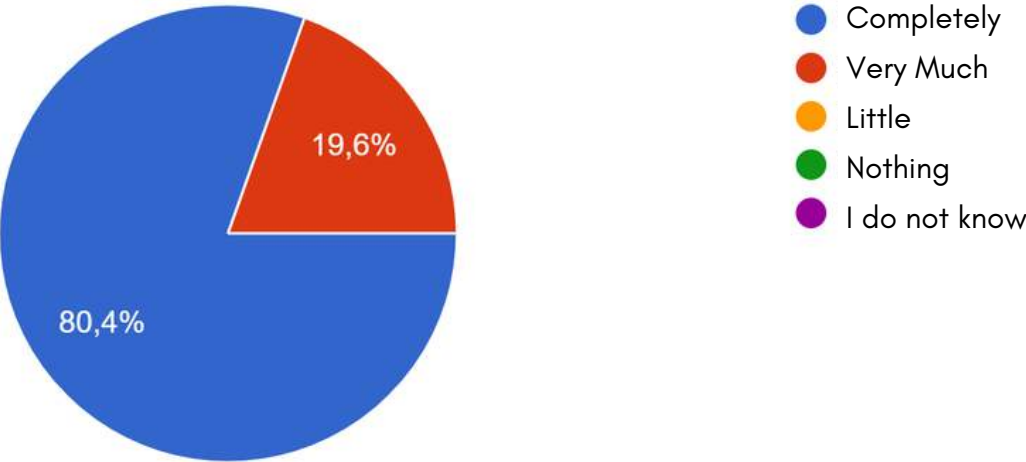


WHAT PARTICIPANTS SAY

Administrative Support and Local Reception

Overall, were STAFF and VOLUNTEERS easy to identify, approachable and helpful?

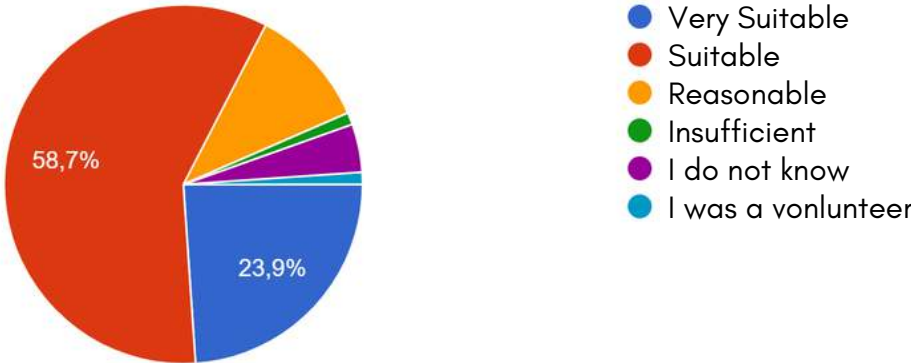
92 answers



We are deeply grateful to all the **VOLUNTEERS** who supported this project through all its stages

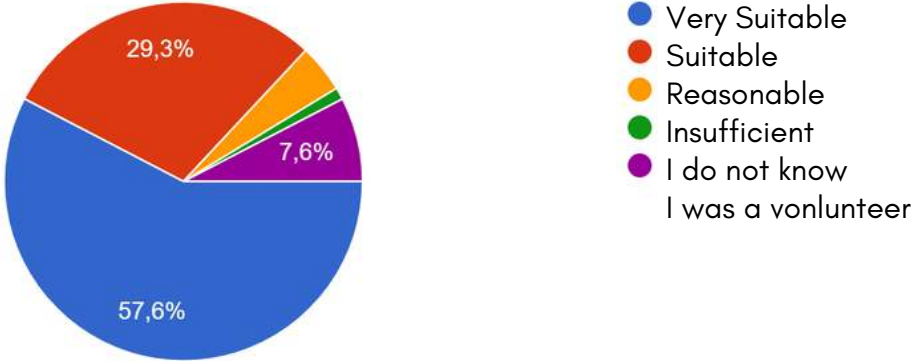
The PUBLICITY of the Symposium was:

92 answers



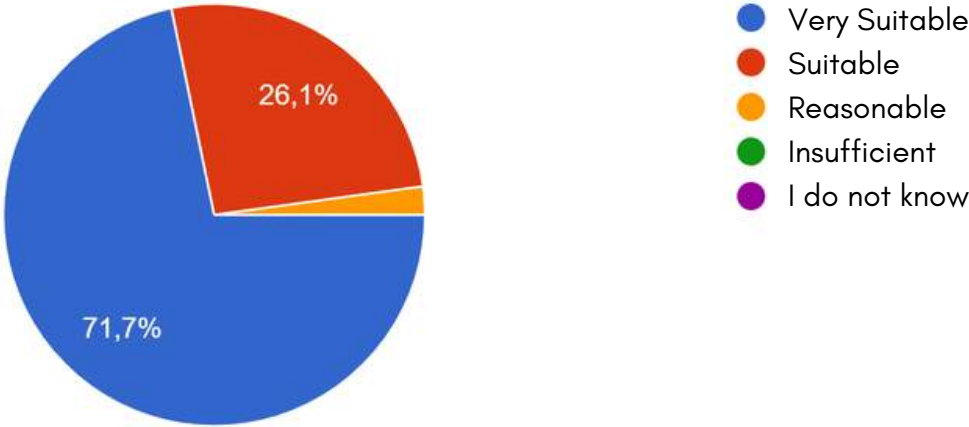
The ADMINISTRATIVE SUPPORT previous to the event and need to communicate via email or telephone was:

92 answers



The ADMINISTRATIVE SUPPORT and LOCAL RECEPTION during the event was:

92 answers



WHAT PARTICIPANTS SAY

Administrative Support and Local Reception

Catering and hospitality offerings for participants at the event venues were secured through donations from civil society companies, which, with their generosity and strong sense of social responsibility, joined this UNbreakable Alliance.

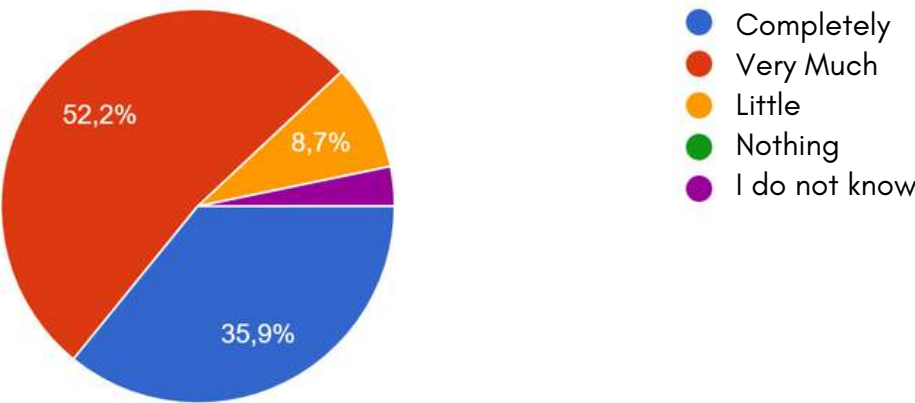
Thank you all!



Surplus: distributed among families and volunteers.

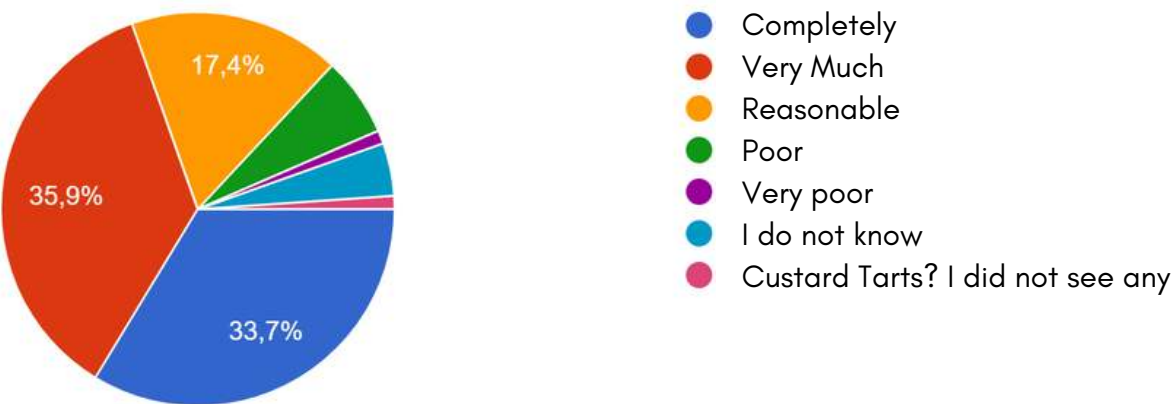
Were BREAKS long and welcoming enough?

92 answers



Was the CATERING OF COFFEE BREAKS sufficient, varied and easily accessible?

92 answers



Overall, how do you assess the quality presented by SOLUÇÕES PRONTAS - RECHEIO?

92 answers

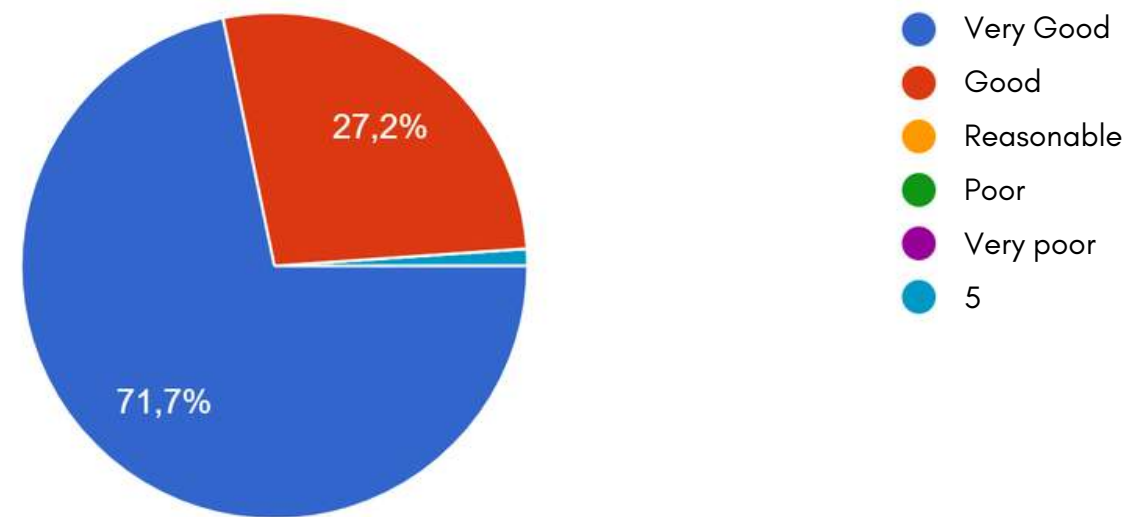


WHAT PARTICIPANTS SAY

Global Assessment of the Symposium

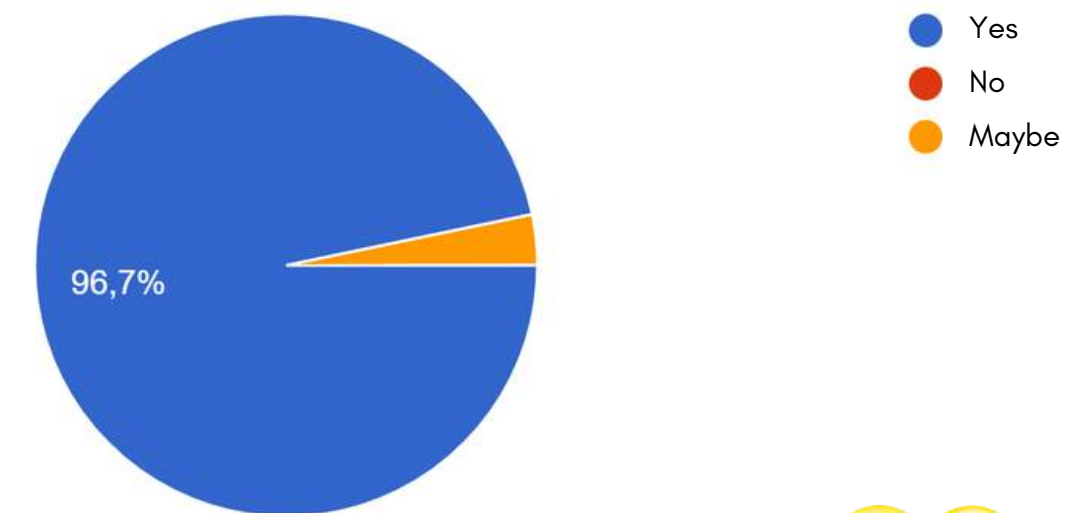
Overall, how do you evaluate the Symposium?

92 answers

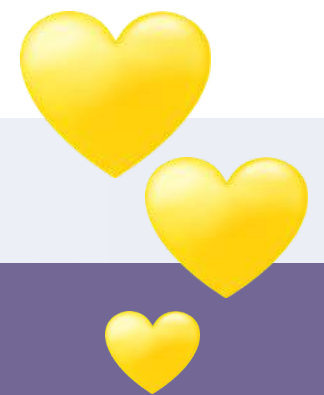


Would you recommend BD2026 to friends and colleagues?

92 answers



... and we are **SO PROUD**... thank you for this evaluation and for trusting our work!



RESULTS

More than an event, this new edition of the UNbreakable Alliance project became, once again, an unforgettable milestone due to the number of people it mobilized, the borders it crossed, and the enormous relevance of the topics addressed.

During Bone Dysplasias 2026, essential bonds were strengthened and the global network of contacts in bone dysplasias expanded, with the Unbreakable Alliance playing a central role in reinforcing this shared vision.

Exceeding what we had projected, the success achieved at various levels converges in a single goal: to transform for the better the quality of life of those living with this condition. We believe in the legacy of this gathering, which stimulated scientific advancement and united Academia, Health services, and families in unprecedented synergies. Most importantly, by working on literacy, capacity building, and patient empowerment, we gave voice and autonomy to people with bone dysplasia so they can be protagonists of their own health.

While it is true that the direct impact focuses on the health ecosystem, the indirect impact on civil society will be inevitable and profound. By equipping patients and professionals with better tools, we are designing a future with greater inclusion in school and at work, less absenteeism, and an end to the social stigma that so often limits the full potential of people with disabilities.



THANKS

It is with enormous pride that we share the success of this initiative, whose results have far exceeded our best expectations.

We publicly express our deepest gratitude to each participant and to all supporting entities, whose vote of confidence was vital for us to achieve the goals we set out to accomplish. We are convinced that by joining forces in a spirit of a true UNbreakable Alliance — bringing together patients, healthcare professionals, industry, and civil society — we will be able to go further in achieving better conditions and a higher quality of life for people with bone dysplasias.

To all those who, directly or indirectly, made this project possible,

our most sincere thank you!

ALIANÇA  NQUEBRÁVEL





Juntar a comunidade em prol da inovação.
Bringing the community together towards innovation.

BONE DYSPLASIAS

**SIMPÓSIO DE
DISPLASIAS
ÓSSEAS**

21 a 23 Maio

2026



associação portuguesa de
osteogenese imperfeita

JUNTE-SE A NÓS

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