CURRÍCULUM VITÆ

NAME: Sérgio Abílio Teixeira Bernardo de Sousa

DATE OF BIRTH: 31/12/1977

NATIONALITY: Portuguese

PROFESSIONAL ADDRESS:
Serviço de Genética Médica,
Hospital Pediátrico de Coimbra – CHUC EPE,
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ACADEMIC DEGREES

MD, Medical Doctor — 1995-2001
Medical School of the University of Coimbra, Portugal

MSc, Master in Molecular Medicine and Oncology — 2005-2008
Medical School of the University of Porto, Portugal
Research project concerning the *GLA* gene molecular analysis and its allelic variants in Fabry disease.
Supervision: Filipa Carvalho; João Paulo Oliveira;

PhD — 2009-2013
Institute of Child Health, University College London
Project entitled: Clinical and molecular characterization of genetic syndromes without known genetic aetiology
Supervision: Gudrun Moore. Awarded in 2014

PROFESSIONAL ACTIVITIES

Post-graduate Medical Internship ("Internato Geral") — Jan 2002-Jun 2003
University of Coimbra's Hospital, Portugal

Specific medical training ("Médico eventual") — Jul - Dec 2003
Dermatology Service, University of Coimbra's Hospitals, Portugal

Medical genetics residency (Five years program) — Jan 2004 – Jan 2009
Medical Genetics Department, Paediatrics Hospital of Coimbra, Portugal
Including training at the *Clinical Genetics Unit, Great Ormond Street Hospital for Children, London, UK,* under supervision of Professor Raoul Hennekam; at the *Service de Génétique Médicale, Hôpital Necker-enfants Malades, Paris, France,* under supervision of Professor Valérie Cormier-Daire.

Medical / Clinical Genetics Specialist — Fev 2009 – Present
Positions:
"Assistente Hospitalar": Fev 2009 – May 2021
"Assistente Hospitalar Graduado": May 2021 – presente
(Consultant degree /*Grau de Consultor* obtained in 20/05/2021)
Medical Genetics Department, Paediatrics Hospital of Coimbra

CURRÍCULUM VITÆ

TEACHING

Teaching assistant ("Assistente convidado") of the discipline "Histology and Embryology I" of the 1st year of the 6-year Veterinary Medicine Course *Escola Univesitária Vasco da Gama, Coimbra, Portugal*	2001-2002
Teaching assistant ("Assistente convidado") of the discipline "Medical Genetics" of the 2nd year of the 6th years Medical Doctor Degree *Medical School of the University of Coimbra, Portugal*	2008 – 2011
Assistant Professor ("Professor auxiliar convidado"), "Medical Genetics course" of the 4th year of the 6th years Medical Doctor Degree, part-time, *Medical School of the University of Beira Interior, Covilhã, Portugal*	2014 - 2017
Assistant Professor ("Professor auxiliar convidado") of the discipline "Medical Genetics" of the 2nd year of the 6th years Medical Doctor Degree *Medical School of the University of Coimbra, Portugal*	2017 - present

AWARDS

International

- **Isabelle Oberlé Award** (Young Investigator Award) for outstanding Research on Genetics of Mental Retardation, for the oral presentation "Nicolaides–Baraitser Syndrome: Delineation of the Phenotype". **European Human Genetics Conference 2009,** Viena, Austria, 23 a 26 de Maio de 2009 (first and presenting author).

- **John M. Opitz Young Investigator Award 2010** for the paper published at AJMG: "Nicolaides–Baraitser Syndrome: Delineation of the Phenotype" (first author).

Portuguese

- Award *"Prémio Prof. Doutor Henrique Oliveira"* and Roche Foundation Scholarship for having the best classification of the medical students graduated in 2001 at the University of Coimbra Medical School.
- Award Dr. A. Torrado da Silva – Best Communication (Poster) of the III Meeting on Perinatology of Centre Portugal, Coimbra, 2004, given by the scientific committee of the III Meeting on Perinatology of Centre Portugal (co-author, presenting).
- First Prize for the Best Oral Communication at "12ª Semana do Médico Interno do Centro Hospitalar de Coimbra", given by its scientific committee, 2006 (first and presenting author)
- First Prize for the Best Oral Communication at the 10th International Symposium of the *Sociedade Portuguesa Doenças Metabólicas* (SPDM), given by its scientific committee, 2014 (first and presenting author).
- Award "*Prémio de Investigação Clinica*" for the best clinical research oral communication at the 19ª annual meeting of the *Sociedade Portuguesa de Genética Humana* (SPGH), 2015, given by its scientific committee, entitled: "Etiological investigation of sensorineural hearing loss: high diagnostic yield and invaluable benefit to patients and families." (co-author).
- Award "*Prémio Amândio Tavares*" at the 20.ª Reunião Anual da Sociedade Portuguesa de Genética Humana, 2016, given by its scientific committee, for the oral communication entitled: "Diagnosis of mendelian disorders using a comprehensive 4813 genes next generation sequencing panel – review of 92 cases." (co-author)
- Award "*Prémio Eufémia Ribeiro*" for the best oral communication at *Congresso Nacional da Associação Portuguesa de Diagnóstico Pré-Natal*, 2017, given by its scientific committee, entitled: "Utilização de um painel alargado de genes ou sequenciação exómica total em contexto pré-natal – experiência de 13 casos" (co-author).

- Award *"Prémio de Investigação Clínica"* for the best clinical research oral communication presented at the 21st Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), 2018, given by its scientific committee, entitled "Clinical and radiological characterization of *EXTL3*- related skeletal phenotype." (first and presenting author).
- Award *"Prémio de Investigação Clínica"* for the best clinical research oral communication presented at the 22nd Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), 2018, entitled "A new mutation in RPL10 associated with X-linked syndromic intellectual disability in two families and literature review." (senior author)
- Award "*Prémio Amândio Tavares*" at the 22nd Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), 2018, given by its scientific committee, for the oral communication entitled: "National Study on osteogenesis imperfecta – genotype and phenotype in 150 Portuguese patients." (shared senior author)
- Award "*Menção Honrosa em Caso Clínico"* at the 23rd Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), 2019, given by its scientific committee, for the clinical case oral communication entitled: "Pachydysostosis of the fibula in a case of Gardner Syndrome – a case report" (senior author)
- Award "*Prémio Melhor Comunicação Oral*" for the best clinical oral communication presented at the 3rd Meeting on Rare Bone Diseases 2022, entitled "Achondroplasia case series: experience of a paediatric multidisciplinary clinic" (senior author)
- Award "*Menção Honrosa em Comunicação Oral*" at 3rd Meeting on Rare Bone Diseases 2022, given by its scientific committee, for the oral communication entitled: "*COL1A2* multiexon deletion in Osteogenesis Imperfecta type 2 – clinical case." (senior author)
- Award "Clinical case" for the best clinical case communication presented at the 26th Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), 2022, entitled "Two case report of a new recognizable syndrome – DEGCAGS (Developmental delay with gastrointestinal, cardiovascular, genitourinary, and skeletal abnormalities)" (senior author).
- Award "*Prémio Melhor Comunicação Oral*" for the best oral communication presented at the 4th Meeting on Rare Bone Diseases 2023, entitled "RMRP-related spectrum – clinical and molecular characterization of ten Portuguese patients". (senior author)
- Award "*Menção Honrosa em Comunicação Oral"* for the oral communication presented at the 4th Meeting on Rare Bone Diseases 2023, entitled "Polyostotic fibrous dysplasia and the role of NaF PET/CT– A case report". (senior author)
- Award "*Menção Honrosa em Comunicação Oral"* for the oral communication presented at the 4th Meeting on Rare Bone Diseases 2023, entitled "RPL13-related spondyloepimetaphyseal dysplasia– expanding the phenotypic spectrum"- (senior author).

SCHOLARSHIPS

– Doctoral Grant – present PhD research project at the Institute of Child Health, London (2009-2013, 4 years), given by the *Fundação para a Ciência e Tecnologia*, Portugal - SFRH/BD/46778/2008.

– Grant for partly supporting the realization of three months of training at the Département de Génétique et Unité INSERM U393, Centre de Reférence des Maladies Osseuses Constitutionelles, Hôpital Necker-Enfants Malades, Paris, given by the Caloust Gulbenkian Foundation, 2008.

– Grant for partly supporting the realization of four months of training at the Clinical and Molecular Genetics Unit, Great Ormond Street Hospital / Institute of Child Health, London, given by the Caloust Gulbenkian Foundation, 2007.

– Scholarship funding for attending the "46th Annual Short Course in Medical and Experimental Mammalian Genetics", given by the organization (The Jackson laboratory and Johns Hopkins University), 2005.

CURRÍCULUM VITÆ

POSTGRADUATED INTERNACIONAL COURSES

5th Course in Genetic Counselling in practice — May 2-7, 2004
European School of Genetic Medicine,
Bertinoro, Italy

46th Annual Short Course in Medical and Experimental Mammalian Genetics — July 17-29, 2005
The Jackson Laboratory and Johns Hopkins University,
Bar Harbour, Maine, USA

V Course of Foetal Medicine – "Diplôme Universitaire de Medicine Fetal - Professeur Yves DUMEZ" — Octob 10-14, 2005; Nov 21-25, 2005; Feb 13-17, 2006
Gabinete de Estudos da Maternidade Bissaya-Barreto and
Université René Descartes Paris V– Faculté Necker Enfants Malades,
Coimbra, Portugal

Second European Course in Clinical Dysmorphology "What I know best" — March 28-29, 2008
Istituto di Genetica Medica, Università Cattolica del Sacro Cuore, Policlínico Universitário Agostino Gemelli, Rome, Italy

"Promoting Bone Health in MPS VI: Framing new therapies" — Octob 7-8, 2008
Children's Hospital & Research Center Oakland, California, EUA, and The Department of Pediatrics, University of Padova — San Francisco, USA

PUBLICATIONS

Book chapter:

1. **Centros e Redes de Referência.** Sérgio B. Sousa. In book: "Livro branco das doenças raras e dos medicamentos órfãos em Portugal / coord. Francisco Batel Marques, Joaquim Marques ; P-Bio, Associação Portuguesa de Bioindústrias, D.L. 2019. ISBN 978-989-20-9744-2, apresentado a 28.02.2020 (http://p-bio.org/pt/livrobranco-das-doencas-raras-e-dos-medicamentos-orfaos/)

Peer-review articles:

My bibliography link:

https://www.ncbi.nlm.nih.gov/sites/myncbi/1fIr8O40--hko/bibliography/48095243/public/?sort=date&direction=descending

Senior author and correspondent author

1. **Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome.** Thomas AC, Williams H, Setó-Salvia N, Bacchelli C, Jenkins D, O'Sullivan M, Mengrelis K, Ishida M, Ocaka L, Chanudet E, James C, Lescai F, Anderson G, Morrogh D, Ryten M, Duncan AJ, Pai YJ, Saraiva JM, Ramos F, Farren B, Saunders D, Vernay B, Gissen P, Straatmaan-Iwanowska A, Baas F, Wood NW, Hersheson J, Houlden H, Hurst J, Scott R, Bitner-Glindzicz M, Moore GE, Sousa SB*, Stanier P*. **Am J Hum Genet.** 2014 Nov 6;95(5):611-21

First author and correspondent author

1. **Phenotype and genotype in Nicolaides-Baraitser syndrome.** Sousa SB, Hennekam RC; Nicolaides-Baraitser Syndrome International Consortium. **Am J Med Genet C Semin Med Genet.** 2014 Sep;166C(3):302-14.
2. **Gain-of-function mutations in the phosphatidylserine synthase 1 (PTDSS1) gene cause Lenz-Majewski syndrome**. Sousa SB, Jenkins D, Chanudet E, Tasseva G, Ishida M, Anderson G, Docker J, Ryten M, Sa J, Saraiva JM, Barnicoat A, Scott R, Calder A, Wattanasirichaigoon D, Chrzanowska K, Simandlová M, Van Maldergem L, Stanier P, Beales PL, Vance JE, Moore GE. ***Nat Genet***. 2014 Jan;46(1):70-6.
3. **Intellectual disability, coarse face, relative macrocephaly, and cerebellar hypotrophy in two sisters**. *Sousa SB*, Ramos F, Garcia P, Pais RP, Paiva C, Beales PL, Moore GE, Saraiva JM, Hennekam RC. ***Am J Med Genet A***. 2014 Jan;164A(1):10-4
4. **Intellectual disability, unusual facial morphology and hand anomalies in sibs**. Sousa SB, Venâncio M, Chanudet E, Palmer R, Ramos L, Beales PL, Moore GE, Saraiva JM, Hennekam RC. ***Am J Med Genet A***. 2013 Oct;161(10):2401-6
5. **Tetra-amelia and lung agenesis syndrome – case report and review.** Sérgio B. Sousa, Raquel Pina, Lina Ramos, Naigel Pereira, Martin Krahn, Wiktor Borozdin, Jürgen Kohlhase, Marta Amorim, Katia Gonnet, Nicolas Lévy, Isabel M. Carreira, Ana Bela Couceiro, and Jorge M. Saraiva. ***Am J Med Genet A***. 2008 Nov 1;146A(21):2799-803.
6. **Síndrome de Noonan – Reavaliação e Estudo Molecular de 16 casos** Sérgio B. Sousa, Margarida Venâncio, Helena Gabriel, Lina Ramos, Isabel Santos, Sebastian Beck, Marta Jorge, Luisa Simão, Purificação Tavares, Jorge M. Saraiva. ***Acta Pediátrica Portuguesa***. 2006:37(4):145-53.
(http://www.spp.pt/Userfiles/File/App/Artigos/38/20121219174918_artigo_original_145.pdf

First author (non correspondent)

1. **Heterozygous missense mutations in *SMARCA2* cause Nicolaides-Baraitser syndrome**. Van Houdt JK, Nowakowska BA, Sousa SB, van Schaik BD, Seuntjens E, Avonce N, Sifrim A, Abdul-Rahman OA, van den Boogaard MJ, Bottani A, Castori M, Cormier-Daire V, Deardorff MA, Filges I, Fryer A, Fryns JP, Gana S, Garavelli L, Gillessen-Kaesbach G, Hall BD, Horn D, Huylebroeck D, Klapecki J, Krajewska-Walasek M, Kuechler A, Lines MA, Maas S, Macdermot KD, McKee S, Magee A, de Man SA, Moreau Y, Morice-Picard F, Obersztyn E, Pilch J, Rosser E, Shannon N, Stolte-Dijkstra I, Van Dijck P, Vilain C, Vogels A, Wakeling E, Wieczorek D, Wilson L, Zuffardi O, van Kampen AH, Devriendt K, Hennekam R, Vermeesch JR. ***Nat Genet***. 2012 Feb 26;44(4):445-9 (shared first authorship)
2. **Expanding the skeletal phenotype of Loeys-Dietz syndrome.** Sousa SB, Lambot-Juhan K, Rio M, Baujat G, Topouchian V, Hanna N, Le Merrer M, Brunelle F, Munnich A, Boileau C, Cormier-Daire V. ***Am J Med Genet A.*** 2011 May;155A(5):1178-83
3. **Postnatal growth retardation, facial dysmorphism, spondylocarpal synostosis, cardiac defect, and inner ear malformation (cardiospondylocarpofacial syndrome?)- a distinct syndrome?** Sousa SB, Baujat G, Abadie V, Bonnet D, Sidi D, Munnich A, Krakow D, Cormier-Daire V. ***Am J Med Genet A.*** 2010 Mar;152A(3):539-46.
4. **Nicolaides–Baraitser Syndrome: Delineation of the Phenotype**. Sérgio B. Sousa, Omar A. Abdul-Rahman, Armand Bottani, Valérie Cormier-Daire, Alan Fryer, Gabriele Gillessen-Kaesbach, Denise Horn, Dragana Josifova, Alma Kuechler, Melissa Lees, Kay MacDermot, Alex Magee, Fanny Morice-Picard, Elizabeth Rosser, Ajoy Sarkar, Nora Shannon, Irene Stolte-Dijkstra, Alain Verloes, Emma Wakeling, Louise Wilson, and Raoul C.M. Hennekam. ***Am J Med Genet A***. 2009 Aug*;* 149A:1628–1640.
5. **Further Delineation of Spondylometaphyseal Dysplasia With Cone-Rod dystrophy**. Sérgio B Sousa, Isabelle Russell-Eggitt, Christine Hall, Bryan Hall and Raoul CM Hennekam. ***Am J Med Genet A***. 2008 Dec 15;146A(24):3186-94.

Co-author

1. **Once-weekly TransCon CNP (navepegritide) in children with achondroplasia (ACcomplisH): a phase 2, multicentre, randomised, double-blind, placebo-controlled, dose-escalation trial.** Savarirayan R, Hoernschemeyer DG, Ljungberg M, Zarate YA, Bacino CA, Bober MB, Legare JM, Högler W, Quattrin T, Abuzzahab MJ, Hofman PL, White KK, Ma NS, Schnabel D, Sousa SB, Mao M, Smith A, Chakraborty M, Giwa A, Winding B, Volck B, Shu AD, McDonnell C. **EClinicalMedicine**. 2023 Oct 2;65:102258. doi: 10.1016/j.eclinm.2023.102258. eCollection 2023 Nov. PMID: 37823031
2. **Evolution of clinical and radiological presentations of spondyloepimetaphyseal dysplasia, RPL13-related: Description of 11 further cases.** Francisca Díaz-González, Manuel Parrón-Pajares, Elsa Lucas-Castro, Silvia Modamio-Høybjør, Lucia Sentchordi-Montané, Verónica Seidel, Pablo Prieto, Guillermo Tarraso-Urios, Marta Codina-Sola, Anna M. Cueto-González, Mary J. Ballesta-Martínez, Fernando Santos-Simarro, Sergio B. Sousa, Karen E. Heath. Clin Genet. 2023 Jul;104(1):100-106. doi: 10.1111/cge.14351. Epub 2023 Apr 30.
3. **Optimising the diagnosis and referral of achondroplasia in Europe: European Achondroplasia Forum best practice recommendations.** Cormier-Daire V, AlSayed M, Alves I, Bengoa J, Ben-Omran T, Boero S, Fredwall S, Garel C, Guillen-Navarro E, Irving M, Lampe C, Maghnie M, Mortier G, Sousa SB, Mohnike K. **Orphanet J Rare Dis**. 2022 Jul 27;17(1):293. doi: 10.1186/s13023-022-02442-2. PubMed PMID: 35897040; PubMed Central PMCID: PMC9327303.
4. **Albright's hereditary osteodystrophy: an entity to recognize.** Maduro AI, Pinto Saraiva A, Pimenta Rodrigues O, Marques M, B Sousa S, Malcata A, Perez de Nanclares G, Serra S. **Rheumatology** (Oxford). 2022 Nov 2;61(11):e356-e357. doi: 10.1093/rheumatology/keac277. PMID: 35551352
5. **ANKRD11 pathogenic variants and 16q24.3 microdeletions share an altered DNA methylation signature in patients with KBG syndrome.** Awamleh Z, Choufani S, Cytrynbaum C, Alkuraya F, Scherer S, Fernandes S, Rosas C, Louro P, Dias P, Neves M, Sousa S, Weksberg R. Hum Mol Genet. 2023 Apr 20;32(9):1429-1438. doi: 10.1093/hmg/ddac289.
6. **Optimising care and follow-up of adults with achondroplasia**. Fredwall S, Allum Y, AlSayed M, Alves I, Ben-Omran T, Boero S, Cormier-Daire V, Guillen-Navarro E, Irving M, Lampe C, Maghnie M, Mohnike K, Mortier G, Sousa SB, Wright M. . **Orphanet J Rare Dis.** 2022 Aug 20;17(1):318. doi: 10.1186/s13023-022-02479-3. PubMed PMID: 35987833; PubMed Central PMCID: PMC9392284.
7. **Biallelic Variants in the Ectonucleotidase ENTPD1 Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia.** Calame DG, Herman I, Maroofian R, Marshall AE, Donis KC, Fatih JM, Mitani T, Du H, Grochowski CM, Sousa SB, Gijavanekar C, Bakhtiari S, Ito YA, Rocca C, Hunter JV, Sutton VR, Emrick LT, Boycott KM, Lossos A, Fellig Y, Prus E, Kalish Y, Meiner V, Suerink M, Ruivenkamp C, Muirhead K, Saadi NW, Zaki MS, Bouman A, Barakat TS, Skidmore DL, Osmond M, Silva TO, Murphy D, Karimiani EG, Jamshidi Y, Jaddoa AG, Tajsharghi H, Jin SC, Abbaszadegan MR, Ebrahimzadeh-Vesal R, Hosseini S, Alavi S, Bahreini A, Zarean E, Salehi MM, Al-Sannaa NA, Zifarelli G, Bauer P, Robson SC, Coban-Akdemir Z, Travaglini L, Nicita F, Jhangiani SN, Gibbs RA, Posey JE, Kruer MC, Kernohan KD, Morales Saute JA, Houlden H, Vanderver A, Elsea SH, Pehlivan D, Marafi D, Lupski JR. **Ann Neurol**. 2022 Aug;92(2):304-321. doi: 10.1002/ana.26381. Epub 2022 May 28.
8. **The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder.** Kumble S, Levy AM, Punetha J, Gao H, Ah Mew N, Anyane-Yeboa K, Benke PJ, Berger SM, Bjerglund L, Campos-Xavier B, Ciliberto M, Cohen JS, Comi AM, Curry C, Damaj L, Denommé-Pichon AS, Emrick L, Faivre L, Fasano MB, Fiévet A, Finkel RS, García-Miñaúr S, Gerard A, Gomez-Puertas P, Guillen Sacoto MJ, Hoffman TL, Howard L, Iglesias AD, Izumi K, Larson A, Leiber A, Lozano R, Marcos-Alcalde I, Mintz CS, Mullegama SV, Møller RS, Odent S, Oppermann H, Ostergaard E, Pacio-Míguez M, Palomares-Bralo M, Parikh S, Paulson AM, Platzer K, Posey JE, Potocki L, Revah-Politi A, Rio M, Ritter AL, Robinson S, Rosenfeld JA, Santos-Simarro F, Sousa SB; Undiagnosed Diseases Network, Wéber M, Xie Y, Chung WK, Brown NJ, Tümer Z. **Hum Mutat**. 2022 Feb;43(2):266-282. doi: 10.1002/humu.24308. Epub 2021 Dec 11. PubMed PMID:

34859529.

9. **Congenital cutaneous ossification.** Gomes TF, Kieselová K, Santiago F, Cardoso JC, Cunha F, Sousa SB, Perez de Nanclares G. **J Paediatr Child Health**. 2022 Jul;58(7):1262-1264. doi: 10.1111/jpc.15814. Epub 2021 Oct 27. PubMed PMID: 34706128.

10. **Vitamin D-Dependent Rickets Type 1A in Two Siblings with a Hypomorphic CYP27B1 Variant Frequent in the African Population.** Joana de Brito Chagas, Carolina Cordinhã, Carmen do Carmo, Cristina Alves, Karen E. Heath, Sérgio B. Sousa, Clara Gomes. **J Pediatr Genet.** 25 October 2021 (online) DOI: 10.1055/s-0041-1736559

11. **Achondroplasia in the First Years of Life – Importance of Early Referral to Pneumology.** Raquel Penteado, Gustavo Bento Soares, Sérgio Sousa, Alice Mirante, Núria Madureira. **Port J Pediat.** 2021;52:187-92. https://doi.org/10.25754/pjp.2021.21466

12. **The first European consensus on principles of management for achondroplasia.** Valerie Cormier-Daire, Moeenaldeen AlSayed, Tawfeg Ben-Omran, Sérgio Bernardo de Sousa, Silvio Boero, Svein O. Fredwall, Encarna Guillen-Navarro, Melita Irving, Christian Lampe, Mohamad Maghnie, Geert Mortier, Zagorka Peijin, and Klaus Mohnike. **Orphanet J Rare Dis**. 2021; 16: 333. Published online 2021 Jul 31. doi: 10.1186/s13023-021-01971-6

13. **Value-based decision-making for orphan drugs with multiple criteria decision analysis: burosumab for the treatment of X-linked hypophosphatemia-** Björn Vandewalle, Miguel Amorim, Diogo Ramos, Sofia Azevedo, Inês Alves, Telma Francisco, Helena Pinto, Sérgio Sousa. **Curr Med Res Opin** . 2021 Jun;37(6):1021-1030. doi: 10.1080/03007995.2021.1904861. Epub 2021 Apr 1.

14. **Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases**. Melo US, Schöpflin R, Acuna-Hidalgo R, Mensah MA, Fischer-Zirnsak B, Holtgrewe M, Klever MK, Türkmen S, Heinrich V, Pluym ID, Matoso E, Bernardo de Sousa S, Louro P, Hülsemann W, Cohen M, Dufke A, Latos-Bieleńska A, Vingron M, Kalscheuer V, Quintero-Rivera F, Spielmann M, Mundlos S. **Am J Hum Genet.** 2020 Jun 4;106(6):872-884.

15. **Complex movement disorder in a patient with heterozygous YY1 mutation (Gabriele-de Vries syndrome).** Carminho-Rodrigues MT, Steel D, Sousa SB, Brandt G, Guipponi M, Laurent S, Fokstuen S, Moren A, Zacharia A, Dirren E, Oliveira R, Kurian MA, Burkhard PR, Bally JF. **Am J Med Genet A**. 2020 Sep;182(9):2129-2132. doi: 10.1002/ajmg.a.61731. Epub 2020 Jul 6. PMID: 32627382

16. **De novo *SMARCA2* variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides–Baraitser syndrome**. Cappuccio G, Sayou C, Tanno PL, Tisserant E, Bruel AL, Kennani SE, Sá J, Low KJ, Dias C, Havlovicová M, Hančárová M, Eichler EE, Devillard F, Moutton S, Van-Gils J, Dubourg C, Odent S, Gerard B, Piton A, Yamamoto T, Okamoto N, Firth H, Metcalfe K, Moh A, Chapman KA, Aref-Eshghi E, Kerkhof J, Torella A, Nigro V, Perrin L, Piard J, Le Guyader G, Jouan T, Thauvin-Robinet C, Duffourd Y, George-Abraham JK, Buchanan CA, Williams D, Kini U, Wilson K; Telethon Undiagnosed Diseases Program, Sousa SB, Hennekam RCM, Sadikovic B, Thevenon J, Govin J, Vitobello A, Brunetti-Pierri N. **Genet Med** (2020). Nov;22(11):1838-1850. doi: 10.1038/s41436-020-0898-y. Epub 2020 Jul 22. PMID: 32694869.

17. **Autosomal-Recessive Mutations in *MESD* Cause Osteogenesis Imperfecta**. Moosa S, Yamamoto GL, Garbes L, Keupp K, Beleza-Meireles A, Moreno CA, Valadares ER, de Sousa SB, Maia S, Saraiva J, Honjo RS, Kim CA, Cabral de Menezes H, Lausch E, Lorini PV, Lamounier A Jr, Carniero TCB, Giunta C, Rohrbach M, Janner M, Semler O, Beleggia F, Li Y, Yigit G, Reintjes N, Altmüller J, Nürnberg P, Cavalcanti DP, Zabel B, Warman ML, Bertola DR, Wollnik B, Netzer C. **Am J Hum Genet**. 2019 Oct 3;105(4):836-843. doi: 10.1016/j.ajhg.2019.08.008. Epub 2019 Sep 26. PubMed PMID: 31564437; PubMed Central PMCID: PMC6817720.

18. **The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene.** Peter VG, Quinodoz M, Pinto-Basto J, Sousa SB, Di Gioia SA, Soares G, Ferraz Leal G, Silva ED, Pescini Gobert R, Miyake N, Matsumoto N, Engle EC, Unger S, Shapiro F, Superti-Furga A, Rivolta C, Campos-Xavier B. **Genet Med.** 2019 Jul 2;. doi: 10.1038/s41436-019-0595-x. [Epub ahead of print] PubMed PMID: 31263216.

19. **RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6.** Szenker-Ravi E, Altunoglu U, Leushacke M, Bosso-Lefèvre C, Khatoo M,

Thi Tran H, Naert T, Noelanders R, Hajamohideen A, Beneteau C, de Sousa SB, Karaman B, Latypova X, Başaran S, Yücel EB, Tan TT, Vlaminck L, Nayak SS, Shukla A, Girisha KM, Le Caignec C, Soshnikova N, Uyguner ZO, Vleminckx K, Barker N, Kayserili H, Reversade B. ***Nature.*** 2018 May;557(7706):564-569. doi: 10.1038/s41586-018-0118-y. Epub 2018 May 16. Erratum in: Nature. 2018 Jul 5;:. PubMed PMID: 29769720.

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CONFERENCES/TALKS, BY INVITATION *(selected – update 2022)*

1. **Acondroplasia e outras displasias ósseas.** Sérgio. B Sousa. Integrada em Mesa redonda "A importância de equipas multidisciplinares em doenças raras". 29º Encontro de Pediatria. 10-11/01/2022, Coimbra

2. **Displasias ósseas – o estado de arte.** Sérgio. B Sousa. Simpósio corporativo "Displasias esqueléticas – Novas Abordagens", Reunião da Sociedade de Endocrinologia e Diabetologia Pediátrica da Sociedade Portuguesa de Pediatria, Vila Nova Gaia, 03/06/2022

3. **Progresso na equipa multidisciplinar de displasias ósseas do CHUC, na ERN-BOND, nos ensaios clínicos e novos tratamentos. Mapeamento das equipas portuguesa.** Sérgio Sousa, 3as Jornadas de Doenças Ósseas Raras, Coimbra, 6-7/06/2022

4. **A baixa estatura desproporcionada -o que pensar e como abordar?** Sérgio B. Sousa. Curso Formação Contínua - da Sociedade Portuguesa de Pediatrica, Módulo XVIII - ENDOCRINOLOGIA, 09/06/2022

5. **Hereditariedade e Opções Reprodutivas em Diferentes Displasias.** Sérgio B. Sousa. "5º Encontro Associação Nacional de Displasias Ósseas", Guimarães, 25-26/06/2022

6. **WP5 CPMS activities.** Plenary Meeting European Reference Network on Rare Bone Diseases. Online. 26/09/2022

7. **Healthcare experience on Vosorotide administration. Reference hospitals - CHUC** Sérgio B. Sousa. VII International Congresso on ASDD(Achondroplasia and other skeletal dysplasia with Dwarfism). Gijon, Spain. 9-11/10/2022

8. **Trip to the diagnosis of ASDD.** Sérgio B. Sousa. VII International Congresso on ASDD(Achondroplasia and other skeletal dysplasia with Dwarfism). Gijon, Spain. 9-11/10/2022

9. **Taking the decision to start therapy.** Sérgio B. Sousa. VOXZOGO®q (vosoritide) Experience Networks (V.E.NN) Virtual Meeting, 12 October 2022

10. **Serviços clínicos** Sérgio B. Sousa Conferência PT_MedGen Estratégia Nacional para a Medicina Genómica, Lisboa, 03/11/2022

11. **Displasias ósseas - Visão geral e abordagem clínica.** Sérgio Sousa. 1º Seminário de Displasias ósseas. Funchal, 04/11/2022

12. **Seguimento multidisciplinar em displasias ósseas – experiência do CHUC e possibilidades de colaboração.** Sérgio B. Sousa. 1º Seminário de Displasias ósseas. Funchal, 04/11/2022

13. **Variability in assessing Anthropometric Parameters" Sérgio B. Sousa. Webinar on Anthropometric Parameters in Achondroplasia, 25/11/2022 Displasias ósseas.** Sérgio B. Sousa. 49º Congresso Português de Neonatologia. 8 e 9 de Abril de 2021. Figueira da

Foz

14. **SMARCA2-related syndromes.** Sérgio B. Sousa. webinar and discussion with Nicolaides-Baraitser Syndrome Support Group and Nicolaides Baraitser Syndrome worldwide Foundation", 01/08/2020.

15. **Projeto In2Genome - Resultados globais e perspetiva hospitalar**. Sérgio Sousa. Workshop "WES na prática clínica em Portugal", Coimbra, 16/11/2019.

16. **Brief ERNs introduction**. Sérgio Sousa. 23ª Reunião Anual da Sociedade Portuguesa de Genética Humana, Coimbra, 14/11/2019.

17. **Displasias Ósseas.** Sérgio Sousa. XII Curso Teórico-Prático de Endocrinologia Pediátrica - Parte 2 - Curso Crescimento e Puberdade, Coimbra, 7-8/11/2019.

18. **Equipa Multidisciplinar de Doenças Ósseas Raras do Centro Hospitalar e Universitário de Coimbra – Atualização das atividades da equipa e Rede Europeia de Referência BOND.** Sérgio Sousa. 1º Congresso Nacional de Doenças Raras, Núcleo de Doenças Raras da Sociedade Portuguesa de Medicina Interna, Sintra, 31 de outubro a 2/11/2019.

19. **The development of new enzyme replacement therapies: the example of Hypophosphatasia.** Sérgio Sousa. 15th International SPDM Symposium - Portuguese Society of Metabolic Diseases, Coimbra, Portugal, 14-16/03/2019.

20. **Clinical Genetics of Skeletal Dysplasias.** Sérgio Sousa. 6th International ALPE Congress on Achondroplasia & Other Dysplasias, 12-14.10.2018, Gijon, Spain

21. **Diagnóstico Prénatal de Doenças Ósseas Raras – Diagnóstico Etiológico.** Sérgio Sousa. XI Congresso de Osteoporose da Sociedade Portuguesa de Osteoporose e Doenças Ósseas Metabólicas, 13-15.05.2018, Coimbra, Portugal

22. **Equipa Multidisciplinar de Doenças Ósseas Raras do Centro Hospitalar e Universitário de Coimbra (ERN-BOND-CHUC) – Apresentação do centro, equipa, organigrama e fluxo de utentes.** Sérgio Sousa. 1as Jornadas de Doenças Ósseas Raras. Hotel D. Luis, Coimbra, Portugal, 16-17.02.2018

23. **Acondroplasia e Outras Displasias Ósseas.** Sérgio B. Sousa, Curso Pré-Congresso de Manifestações Músculo-Esqueléticas de Doenças Sistémicas, 5º Congresso Nacional e XXII Jornadas de Ortopedia Infantil, Lisboa, 23 a 25 de março de 2017.

24. **O que é a Hipofosfatasia?** Sérgio B. Sousa, 1ª Reunião de Hipofosfatasia, Sociedade Portuguesa de Osteoporose e Doenças Ósseas Metabólicas (SPODOM), Coimbra, 18/03/2017.

25. **Skeletal dysplasias - what is new and future perspectives.** Sérgio Sousa. Curso de Atualização em Doenças Genéticas do Esqueleto - Curso Pré-congresso - 4º Congresso Nacional e XXI Jornadas de Ortopedia Infantil, Braga, 10/03/2016.

26. **Metabolic defects in phospholipid metabolism II: The phosphocholine, phosphatidyl serine pathway: Choline kinase, Phosphocholine cytidylyl transferase, phosphatidylserine synthase 1**. "Classification and diagnostic approach of IEM affecting the synthesis and remodelling of complex lipids" -Course organized by Recordati Rare Diseases Fondation. Paris. 24-26/06/2015

27. **Workshop- chondrodysplasia.** Sérgio B. Sousa, Valérie Cormier-Daire. "Classification and diagnostic approach of IEM affecting the synthesis and remodelling of complex lipids" - Course organized by Recordati Rare Diseases Fondation. Paris. 24-26/06/2015

28. **Next Generation Sequencing techniques.** 16º Congresso Nacional de Pediatria. Albufeira, 22-24/10/2015.

29. **Skeletal dysplasias.** Mesa "Interface Endocrinologia e Genética". 5.ª Reunião Annual da Sociedade Portuguesa de Endocrinologia e Diabetologia Pediátrica (SPEDP)

30. **Achondroplasia – new perspectives on the treatment of genetic disorders.** 23º Encontro de Pediatria do Hospital Pediátrico de Coimbra, 19/01/2015

31. **Skeletal dysplasias – after the diagnosis**. (conferência). V Congreso Internacional - Acondroplasia y otras Displasias, Fundación ALPE, Gijón, Asturias, Espanha, 11/10/2014

32. **Nicolaides-Baraitser syndrome and related disorders involving components of the BAF complex**. Autumn Spanish Dysmorphology Meeting, Hospital Universitario de La Paz, Madrid, Espanha, 8/10/2014.

33. **Lenz-Majewski Syndrome - disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia.** Sérgio B. Sousa. Birth Defects Research Centre Seminar, UCL Institute of Child Health, Londres, Reino Unido, 24 de janeiro de 2014

ORAL PRESENTATIONS *(only oral communications as first/presenting or senior author were included – update 2022)*

1. **Acondroplasia: experiência da consulta multidisciplinar de uma equipa pediátrica de referência para as displasias ósseas.** Luana Silva & Flávia Belinha, Núria Madureira, Iolanda Veiros, Pedro Figueiredo, Joana Ribeiro, Gustavo B. Soares, Beatriz Ramada, João E. Moura, João Cabral, Ana B. Roseiro, Francisco Vale, Alice Mirante, Sérgio B. Sousa. 29º Encontro de Pediatria. 10-11/01/2022, Coimbra
2. **Autosomal-Recessive Mutations in MESD – a Clinical Case of Severe Osteogenesis Imperfecta.** Vera MF Santos, Ana Beleza-Meireles, Lina Ramos, Jorge Seabra, Pedro Cardoso, Luísa Diogo, Shahida Moosa, Bernd Wollnik, Jorge M Saraiva, Sérgio B Sousa. 3as Jornadas de Doenças Ósseas Raras, Coimbra, 6 e 7 de junho.
3. **CANT1-related skeletal dysplasia: case report of two unrelated Portuguese patients with Desbuquois dysplasia, Kim variant / MED7.** Ariana C. Mendes, Alice Mirante, João Cabral, Núria Madureira, Pedro Figueiredo, João E. Moura, Sofia Maia, Silvia Modamio-Hoybjor, Jorge M. Saraiva, Karen E. Heath, Sérgio B. Sousa. 3as Jornadas de Doenças Ósseas Raras, Coimbra, 6 e 7 de junho.
4. **Recessive multiple epiphyseal dysplasia case series: same SLC26A2 variant, different clinical features.** Marta Marques, Daniela Oliveira, Sofia Maia, João Cabral, Inês Balacó, Cristina Alves, Janet Pereira, Karen E. Heath, Silvia Modamio-Hoybjor, Alice Mirante, Sérgio B. Sousa, Jorge M. Saraiva, 3rd Meeting on Rare Bone Diseases, Coimbra, 6 e 7 de junho.
5. **Caffey disease: clinical case with striking neonatal presentation.** Peixoto D, Correia L, Almeida S; Figueiredo S, Tarquini O, Estanqueiro P, Modamio-Hoybjor S, Heath KE, Sousa SB. 3rd Meeting on Rare Bone Diseases, Coimbra, 6-7/06/2023
6. **Achondroplasia case series: experience of a paediatric multidisciplinary clinic.** Luana Silva* & Flávia Belinha*, Núria Madureira, Iolanda Veiros, Pedro Figueiredo, Joana Ribeiro, Gustavo B. Soares, Beatriz Ramada, João E. Moura, João Cabral, Ana B. Roseiro, Alice Mirante, Sérgio B. Sousa. 3rd Meeting on Rare Bone Diseases, Coimbra, 6-7/06/2023
7. **Two case report of a new recognizable syndrome – DEGCAGS (developmental delay with gastrointestinal, cardiovascular, genitourinary, and skeletal abnormalities.** Mafalda Santos, Joana Azevedo, Ana L. Carvalho, Sérgio B. Sousa. 26th Annual Meeting,

Sociedade Portuguesa de Genética Humana, Coimbra, 17 a 19 de novembro.

8. **Pachydysostosis of the fibula in a case of Gardner syndrome – a case report.** Daniela Oliveira, Sofia Maia, Inês Balacó, Paulo Coelho, Susana Almeida, Margarida Venâncio, Jorge Saraiva, Gen Nishimura, Sérgio B. Sousa. 23ª Reunião Anual da Sociedade Portuguesa de Genética Humana, Coimbra, 14 -16/11/ 2019.
9. **Genotype and phenotype in 201 Portuguese patients with Osteogenesis Imperfecta - Unusual molecular results challenge genetic counselling"** André M. Travessa*, Patrícia Dias*, Miriam Aza-Carmona*, Joana Rosmaninho-Salgado, Teresa Saraiva, Ana Grangeia, Marta Amorim, Miguel Gonçalves-Rocha, Graça Araújo, Heloísa Santos, Márcia Rodrigues, Ana Medeira, Isabel Cordeiro, Juliette Dupont, Oana Moldovan, Ana Beleza, Joaquim Sá, Jorge M. Saraiva, Lina Ramos, Margarida Venâncio, Sofia Maia, Sofia Fernandes, Gabriela Soares, João P. Freixo, Francisca Díaz, Céu Barreiros, Carolina de la Torre, Anabela Bandeira, João Campagnolo, Fátima Godinho, M. Cassiano-Neves, Viviana Tavares, Filomena Teixeira, Teresa Kay, Renata Oliveira, Ana Fortuna, Sérgio B. Sousa#, Ana Berta Sousa#, Karen E. Heath#. 14th International Skeletal Dysplasia Society Meeting, Oslo, Norway, 11 a 14/09/2019 and in 22ª Reunião Anual da Sociedade Portuguesa de Genética Humana, Porto, 15-17/11/2018. [selected for the "Prémio Amândio Tavares" award).
10. **Myhre Syndrome – a report of four unrelated cases.** Sara Moreira Ribeiro, Teresa Carminho Rodrigues, Pedro Louro, Fabiana Ramos, Janet Pereira, Valerie Cormier-Daire, Jorge M. Saraiva, Alice Mirante, Margarida Venâncio, Sérgio B. Sousa. 2ª Jornadas de Doenças Ósseas Raras, Coimbra, 15/02/2019.
11. **Spondyloenchondrodysplasia and SLE – report on three unrelated cases**. Joana Rosmaninho-Salgado, Renata Oliveira, Joana Caetano Serra, Alice Mirante, Manuel Salgado, Paula Estanqueiro, Cátia Duarte, Luis Inês, Margarida Venâncio, Belinda Xavier, Sheila Unger, Andrea Superti-Furga, Jorge M. Saraiva, Sérgio B. Sousa. 2ª Jornadas de Doenças Ósseas Raras, Coimbra, 15/02/2019.
12. **A new mutation in RPL10 associated with X-linked syndromic intellectual disability in two families and literature review**. Laço M, Venâncio M, Grote L, Friez M, Chanudet E, Bachelli C, Williams H, Sousa S. 22ª Reunião Anual da Sociedade Portuguesa de Genética Humana, Porto, 15-17/11/2018.
13. **Wiedemann-Steiner Syndrome – clínical and molecular characterization of 9 patients from four national hospital centers.** Fernandes S, Sá MS, Carvalho I, Rodrigues M, Oliveira R, Louro P, Sá J, Dias P, Soares C, Soares G, Saraiva JM, Sousa SB. 22ª Reunião Anual da Sociedade Portuguesa de Genética Humana, Porto, 15-17/11/2018.
14. **Opsismodysplasia – a phosphoinositides biosynthesis and remodelling delfect: three unrelated Portuguese cases.** Almeida PM, Santos H, Saraiva JM, Seabra J, Reis C, Venâncio M, Heath K, Cormier-Daire V, Sousa SB, 1ª Jornadas de Doenças Ósseas Raras, Coimbra, 16 e 17/02/2018. And in 14th International SPDM (Sociedade Portuguesa de Doenças Metabólicas) Symposium. Porto, 15 a 17/03/2018
15. **Hypophosphatasia: report on two Portuguese cases**. Rosmaninho-Salgado J, Beleza A, Matos G, Seabra J, Saraiva JM, Sousa SB. 1ª Jornadas de Doenças Ósseas Raras, Coimbra, 16-17/02/2018.
16. **Familial case of spondyloepiphyseal dysplasia, Kimberley Type, with a novel splicing mutation in Aggrecan gene**. MN, Dinis I, Saraiva JM, Sentchordi-Montané L, Heath KE, Sousa SB. 1ª Jornadas de Doenças Ósseas Raras, Coimbra, 16-17/02/2018.
17. **The type II collagenopathies spectrum – 3 case reports: spondyloepiphyseal dysplasia congenita; spondyloepimetaphyseal dysplasia, Strudwick type; and mild spondyloepiphyseal dysplasia with early onset osteoarthritis.** Sousa SB, Heath KE, Rosmaninho-Salgado J, Pereira J, Saraiva JM. 1ª Jornadas de Doenças Ósseas Raras, Coimbra, 16-17/02/2018.
18. **National study on cleidocarnial dysplasia – clinical and molecular characterization of 15 Portuguese patients**. Fernandes S, Machado C, Soares AR, Aza-Carmona M, Oliveira R, Moura CP, Louro P, Ramos L, Fortuna AM, Dupont J, Saraiva JM, Heath KE,

Sousa SB. 1ª Jornadas de Doenças Ósseas Raras, Coimbra, 16- 17/02/2018. And in 21ª Reunião Anual da Sociedade Portuguesa de Genética Humana. Caparica, 16 a 18/11/2017.

19. **Microdeleção 16p11.2: a cromossomopatia mais frequentemente associada a obesidade.** Joana Rosmaninho-Salgado. Luís M. Pires. Susana Ferreira, Joana B. Melo, Isabel M. Carreira, Joaquim Sá, Jorge M. Saraiva, Sérgio B. Sousa. 69ª Reunião Anual da SPEDM. Vilamoura, 1-4/02/2018.
20. **Clinical and Radiological Characterization of EXTL3-related skeletal phenotype.** Sousa S.B., Hall C.M., Mendoza-Londono R., Dias P., Davies E.G., Oud M.M., Tuijnenburg P., Hempel M., Stabej P., Lessel D., Arts H., Kuijpers T. **13th International Skeletal Dysplasia Society Meeting 2017**, 20th-23rd September 2017, Bruges, Belgium; and in

    21th Annual Meeting of the Portuguese Society of Human Genetics 2017, November 16-18, Almada, Portugal. [selected for the clinical research award).
21. **Expanding the MEF2C happloinsuficiency syndrome: report of a new MEF2C mutation.** Joana Rosmaninho-Salgado, Lina Ramos, Jorge M Saraiva, Sérgio B Sousa. XLVI Conferências de Genética Doutor Jacinto Magalhães: Neurogenética Pediátrica, Porto, 3/02/2017.
22. **X-Linked Intellectual Disability Syndrome type Nascimento, caused by UBE2A mutations – MRXS30 - Report of two affected brothers and literature."** Sofia Fernandes, Jorge Saraiva, Sérgio Sousa. 20ª Reunião da Sociedade Portuguesa Genética Humana, Coimbra, 10-12/11/2016.
23. **X-linked intellectual disability caused by a novel PAK3 mutation in a large pedigree".** J Rosmaninho-Salgado, H.Williams, M. Harakalova, G.Haaften, F. Ramos, JM Saraiva, G Moore, SB Sousa. 20ª Reunião da Sociedade Portuguesa Genética Humana, Coimbra, 10-12/11/2016.
24. **Genotype and phenotype in Lenz-Majewski Syndrome.** Sérgio B. Sousa. Dagan Jenkins, Estelle Chanudet, Guergana Tasseva, Emily Bliss, Miho Ishida, Joaquim Sá, Jorge Saraiva, Angela Barnicoat, Richard Scott, Alistair Calder, Duangrurdee Wattanasirichaigoon, Krystyna Chrzanowska, Martina Simandlová, Lionel Van Maldergem, Anne Hing, Margherita Silengo, Glenn Anderson, James Docker, Mina Ryten, Janet Pereira, Kevin Mills, Peter Clayton, Philip Stanier, Philip Beales, Jean Vance and Gudrun E. Moore. **12th International Skeletal Dysplasia Society Meeting 2015**, 29th July - 1st August, Istanbul, Turkey
25. **Identification of a novel gene causing a recognizable and distinct autosomal recessive intellectual disability and ataxia syndrome with cerebellar atrophy, relative macrocephaly, and coarse facial features.** Sérgio B. Sousa, Anna C Thomas, Hywel Williams, Núria Setó-Salvia, Chiara Bacchelli, Estelle Chanudet, Dagan Jenkins, Mary O'Sullivan, Louise Ocaka, Konstantinos Mengrelis, Miho Ishida, Glen Anderson, Deborah Morough, Mina Rytan, Jorge M Saraiva, Fabiana Ramos, Bernadette Farren, Dawn Saunders, Paul Gissen, Ania Straatmaan-Iwanowska, Frank Baas, Nicholas Wood, Robert Robinson, Joshua Hersheson, Henry Houlden, Raoul Hennekam, Jane Hurst, Richard Scott, Maria Bitner-Glindzicz, Gudrun E Moore, Philip Stanier. **16th Manchester Dysmorphology Conference**, Manchester, UK, 13th November 2014.
26. **Lenz-Majewski syndrome: disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia.** Sérgio B. Sousa, Dagan Jenkins, Estelle Chanudet, Guergana Tasseva, Emily Bliss, Miho Ishida, Joaquim Sá, Jorge Saraiva, Angela Barnicoat, Richard Scott, Alistair Calder, Duangrurdee Wattanasirichaigoon, Krystyna Chrzanowska, Martina Simandlová, Lionel Van Maldergem, Anne Hing, Margherita Silengo, Glenn Anderson, James Docker, Mina Ryten, Janet Pereira, Kevin Mills, Peter Clayton, Philip Stanier, PhilipBeales, Jean Vance and Gudrun E. Moore. **European Conference of Human Genetics 2014**, May 31 – June 1, Milan, June , 2014
27. **Lenz-Majewski Syndrome – Lenz-Majewski syndrome: disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia.** Sérgio B Sousa, Dagan Jenkins, Estelle Chanudet, Guergana Tasseva, Emily Bliss, Miho Ishida, Joaquim Sá, Jorge M Saraiva, Angela Barnicoat, Richard Scott, Alistair Calder, Duangrurdee Wattanasirichaigoon, Krystyna

Chrzanowska, Martina Simandlová, Lionel Van Maldergem, Anne Hing, Margherita Silengo, Glenn Anderson, James Docker, Mina Ryten, Janet Pereira, Kevin Mills, Peter Clayton, Philip Stanier, Philip L Beales, Jean E Vance, Gudrun E Moore. 10th International Sociedade Portuguesa Doenças Metabólicas (SPDM) Symposium. Cascais, Portugal – 20-21/03/2014 [Best Oral Communication Award].

28. **Nicolaides-Baraitser syndrome: Delineation of the phenotype**. Sousa SB, Abdul-Rahman OA, Bottani A, Cormier-Daire V, Fryer A, Gillessen-Kaesbach G, Horn D, Josifova D, Kuechler A, Lees M, MacDermot K, Magee A, Morice-Picard F, Rosser E, Sarkar A, Shannon N, Stolte-Dijkstra I, Verloes A, Wakeling E, Wilson L, Hennekam RC. European Human Genetics Conference 2009, Viena, Austria, 23 –26/05/2009 [Isabelle Oberlé Award].
29. **Pitt-Hopkins Syndrome – clinical report.** Sérgio Sousa, Alexandra Cabral, Christiane Zweier, Margarida Venâncio, Anita Raunch, Jorge Saraiva. British Human Genetics Conference (BSHG) 2007; York, United Kingdom – 17-19/09/2007.
30. **Structural chromosomal rearrangements – Difficulties in prenatal genetic counselling.** VI Meeting of the Portuguese Prenatal Diagnose Centres, Vidago, Portugal – July 1-3, 2004.
31. **Noonan syndrome – clinical evaluation and molecular analysis of 11 cases.** 10ª Semana do Médico Interno do Centro Hospitalar de Coimbra. November 15-19, 2004.
32. **From prenatal detection of adrenal mass to appropriate follow-up**. II International Meeting on Neonatology e XXXIV Jornadas da Secção de Neonatologia da Sociedade Portuguesa de Pediatria, Lisboa, November, 16-18.
33. **Analysis of the Medical Termination of Pregnancies at the Bissaya-Barreto Maternity between 2000 and 2005.** 12ª Semana do Médico Interno do Centro Hospitalar de Coimbra, Coimbra, Portugal - 11-15/12/2006 [1º Award].

RESEARCH AND DEVELOPMENT (R&D) PROJECTS

- In2Genome project (2017-2020). Implementation of diagnostic whole-exome sequencing (WES) and building a genomic interpretation team in a public hospital setting with regional collaborations. Partnership between Serviço de Genética do Centro Hospitalar e Universitário de Coimbra, Coimbra Genomis and Genoinseq (Biocant). Funding: Projeto nº 17800; Projeto de Investigação e Desenvolvimento Tecnológico em copromoção, Centro2020.

CLINICAL TRIALS / OBSERVATIONAL STUDIES

- Principal Investigator in clinical trial Accomplish (TCC-201; Ascendis**) "**A Dose Escalation Trial Evaluating Safety, Efficacy, and Pharmacokinetics of TransCon CNP Administered Once Weekly in Prepubertal Children With Achondroplasia"; since August 2021

- **Sub-Investigator** in clinical trial "A Study Of Safety, Tolerability And Effectiveness Of Recifercept In Children With Achondroplasia" (C4181005; Pfizer); May 2021 -2022.

- Principal Investigator in observacional study ACHieve (TCC-NHS-01; Ascendis) "A Multi-center, Longitudinal, Observational Study of Children With Achondroplasia"; since Sept 2020.

- **Principal Investigator** in observacional study Dreambird (TA46-02/C4181001; Therachon/Pfizer); "Observational Study Investigating Clinical & Anthropometric Characteristics of Children With Achondroplasia" since July 2019.

- Sub-Investigator in clinical trial MO-Ped (PVO-2A-201; Clementia/Ipsen)"A Phase 2, Randomized, Double-Blind, Placebo-Controlled Efficacy and Safety Study of Palovarotene in Subjects with Multiple Osteochondromas."; since 2018.

CURRÍCULUM VITÆ

SCIENTIFIC SOCIETIES

- Member of: Portuguese Society of Human Genetics; European Society of Human Genetics; International Skeletal Dysplasia Society.
- **Secretary** of the Portuguese Society of the Human Genetics in 2016, member of its executive board 2015-2017.
- **Scientific Committee member** of the Portuguese Society of the Human Genetics (2018-2021)
- **Board member** of European Society of Human Genetics (2019-2023)
- Member of **Steering Committee** of the European Achondroplasia Forum

OTHER ITEMS

Coordination roles:

- **Coordinator of the Centro Hospitalar e Universitário de Coimbra skeletal dysplasia multidisciplinary team** (2014-present), **founding member of the European Reference Network on Rare Bone Disorders** (ERN-BOND; 2017-present).
- **Co-Leader of WP4 - CPMS and Healthcare group - of European Reference Network on Rare Bone Disorders**
- Organization of the Coimbra annual meeting on Rare Bone Disorders (***Jornadas de Doenças Ósseas Raras***): 1st in 16-17/02/2018 (https://asic.pt/images/congressos/doencasosseas.pdf); 2nd in 15/02/2019 (https://asic.pt/images/congressos/doencasosseas_2019.pdf); 3rd in 6-7/06/2022 (https://www.asic.pt/index.php/congressos-asic/460-3as-jornadas-de-doencas-osseas-raras); 4th in 29-30/06/2022 (https://mail.asic.pt/index.php/congressos-asic/483-4as-jornadas-de-doencas-osseas-raras)
- **Coordinator of the Portuguese Study Group on Genetic Skeletal Disorders**, GruPEDGE, group affiliated to the Portuguese Society of Human Genetics (2018-present)
- **Coordinator of the CHUC Genomic Interpretation Unit as part of the Project In2Genome** (2017-2020, see above)

Reviewer:

- *American Journal of Human Genetics, American Journal of Medical Genetics*, *European Journal of Human Genetics*, *European Journal of Medical Genetics*, and *Acta Pediátrica Portuguesa*.

Patient advocacy

- participation and talks in multiple meetings of: Nicolaides-Baraitser Syndrome support group; ALPE foundation; Associação Nacional de Displasias Ósseas ANDO Portugal.

Consultancy

- related mainly to skeletal dysplasias (achondroplasia; X-linked hypophosphatemia, etc)
- member of the European Achondroplasia Forum
- consultancy and/or advisory boards for Ascendis, Exigo consultores/Kiowa Kirin, Biomarin.

Other:

- **Languages: Advanced Level: Portuguese** (mother tongue), **English (**Certificate in Advanced English**,** *University of Cambridge (Council of Europe Level C1),* December 2004Classification: Grade B), **French (**Diplôme Pratique de Langue Française, *Alliance Française,* June 1994, Classification: 14,4/20.
- **Goodenough College Alumni 2009-2013**, London
- **Erasmus program** – the fifth year of the M.D. degree (1999-2000) was done at the *Lyon-Sud Medical School, University Claude Bernard Lyon ,* Lyon, France.