CURRÍCULUM VITÆ

NAME: Sérgio Abílio Teixeira Bernardo de Sousa

DATE OF BIRTH: 31/12/1977

NATIONALITY: Portuguese

PROFESSIONAL ADDRESS:

Portugal: Serviço de Genética Médica,
Hospital Pediátrico de Coimbra – CHUC EPE,
Av. Afonso Romão,
3000-602 Coimbra
Tel: 239480238

E-MAIL: sbsousa@chuc.min-saude.pt; sergiobsousa@gmail.com

ACADEMIC DEGREES

MD, Medical Doctor — 1995-2001
Medical School of the University of Coimbra, Portugal
Final classification: 17/20

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.
Final classification: Very Good

PhD — 2009-2013
Project entitled: Clinical and molecular characterization of genetic syndromes without known genetic aetiology
Institute of Child Health, University College London
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
Final classification: 19,8/20
Including:
- 4 months of training (Set-Dec 2007) at *the Clinical Genetics Unit, Great Ormond Street Hospital for Children, London, UK,* under supervision of Professor Raoul Hennekam and
- 3 months of training (Apr-Jun 2008) at the *Service de Génétique Médicale, Hôpital Necker-enfants Malades, Paris, France,* under supervision of Professor Valérie Cormier-Daire.

Medical / Clinical Genetics Specialist ("Assistente Hospitalar") — Fev 2009 – Present
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

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

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.

– 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 Prize for the Best Oral Communication at the 10th International Symposium of the *Sociedade Portuguesa Doenças Metabólicas* (SPDM), given by its scientific committee, 2014.

– Award *"Prémio de Investigação Clínica"* for the oral communication presented at the 21st Annual Meeting of the *Sociedade Portuguesa de Genetica Humana* (SPGH), given by its scientific committee, 2017.

CURRÍCULUM VITÆ

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.

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

CURRÍCULUM VITÆ

PUBLICATIONS

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.

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. **Cutis laxa and excessive bone growth due to de novo mutations in PTDSS1.** Piard J, Lespinasse J, Vlckova M, Mensah MA, Iurian S, Simandlova M, Malikova M, Bartsch O, Rossi M, Lenoir M, Nugues F, Mundlos S, Kornak U, Stanier P, Sousa SB, Van Maldergem L. Am J Med Genet A. 2018 Mar;176(3):668-675.

2. **Heterozygous aggrecan variants are associated with short stature and brachydactyly: Description of 16 probands and a review of the literature.** Sentchordi-Montané L, Aza-Carmona M, Benito-Sanz S, Barreda-Bonis AC, Sánchez-Garre C, Prieto-Matos P, Ruiz-Ocaña P, Lechuga-Sancho A, Carcavilla-Urquí A, Mulero-Collantes I, Martos-Moreno GA, Del Pozo A, Vallespín E, Offiah A, Parrón-Pajares M, Dinis I, Sousa SB, Ros-Pérez P, González-Casado I, Heath KE. Clin Endocrinol (Oxf). 2018 Feb 21. doi: 10.1111/cen.13581. [Epub ahead of print] PubMed PMID: 29464738.

3. **Fibroblasts derived from patients with opsismodysplasia display SHIP2-specific cell migration and adhesion defects.** Ghosh S, Huber C, Siour Q, Sousa SB, Wright M, Cormier-Daire V, Erneux C. **Human mutation.** 2017; 38(12):1731-1739.

4. **Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome.** Oud MM, Tuijnenburg P, Hempel M, van Vlies N, Ren Z, Ferdinandusse S, Jansen MH, Santer R, Johannsen J, Bacchelli C, Alders M, Li R, Davies R, Dupuis L, Cale CM, Wanders RJA, Pals ST, Ocaka L, James C, Müller I, Lehmberg K, Strom T, Engels H, Williams HJ, Beales P, Roepman R, Dias P, Brunner HG, Cobben JM, Hall C, Hartley T, Le Quesne Stabej P, Mendoza-Londono R, Davies EG, de Sousa SB, Lessel D, Arts HH, Kuijpers TW. ***Am J Hum Genet.*** 2017; 100(2):281-296.

5. **Systematic screening for *PRKAR1A* gene rearrangement in Carney complex: identification and functional characterization of a new in-frame deletion.** Guillaud Bataille M, Rhayem Y, Sousa SB, Libé R, Dambrun M, Chevalier C, Nigou M, Auzan C, North MO, Sa J, Gomes L, Salpea P, Horvath A, Stratakis CA, Hamzaoui N, Bertherat J, Clauser E. ***Eur J Endocrinol.*** 2013 Nov 29;170(1):151-60

6. **Loss of the BMP antagonist, SMOC-1, causes Ophthalmo-acromelic (Waardenburg Anophthalmia) syndrome in humans and mice.** Rainger J, van Beusekom E, Ramsay JK, McKie L, Al-Gazali L, Pallotta R, Saponari A, Branney P, Fisher M, Morrison H, Bicknell L, Gautier P, Perry P, Sokhi K, Sexton D, Bardakjian TM, Schneider AS, Elcioglu N, Ozkinay F, Koenig R, Mégarbané A, Semerci CN, Khan A, Zafar S, Hennekam R, Sousa SB, Ramos L, Garavelli L, Furga AS, Wischmeijer A, Jackson IJ, Gillessen-Kaesbach G, Brunner HG, Wieczorek D, van Bokhoven H, Fitzpatrick DR. ***PLoS Genet***. 2011 Jul;7(7):e1002114.

7. **Loss-of-function mutations in PTPN11 cause metachondromatosis, but not Ollier disease or Maffucci syndrome**. Bowen ME, Boyden ED, Holm IA, Campos-Xavier B, Bonafé L, Superti-Furga A, Ikegawa S, Cormier-Daire V, Bovée JV, Pansuriya TC, de Sousa SB, Savarirayan R, Andreucci E, Vikkula M, Garavelli L, Pottinger C, Ogino T, Sakai A, Regazzoni BM, Wuyts W, Sangiorgi L, Pedrini E, Zhu M, Kozakewich HP, Kasser JR, Seidman JG, Kurek KC, Warman ML. ***PLoS Genet.*** 2011 Apr;7(4):e1002050.
8. **Novel deletion encompassing exons 5-12 of the UBE3A gene in a girl with Angelman syndrome**. Beleza-Meireles A, Cerqueira R, Sousa SB, Palmeiro A, Ramos L. ***Eur J Med Genet.*** 2011 May-Jun;54(3):348-50.
9. **Accuracy of prenatal diagnosis in elective termination of pregnancy: 385 cases from 2000 to 2007.** Ramos F, Maia S, Branco M, Raposo J, Sá J, Sousa S, Venâncio M, Pina R, Galhano E, Ramos L, Saraiva J. ISRN Obstet Gynecol. 2011;2011:458120. doi: 10.5402/2011/458120. Epub 2010 Nov 8.
10. **Skeletal complications in mucopolysaccharidosis VI patients: Case reports**. Paula Garcia, Sérgio B. Sousa, Tah Pu Ling, Mário Conceição, Jorge Seabra, Klane K. White, Luisa Diogo *Journal of Pediatric Rehabilitation Medicine: An Interdisciplinary Approach 3* (2010) 63–69
11. **Clinical and Molecular diagnosis of the skeletal dysplasias associated with mutations in the gene encoding Fibroblast Growth Factor Receptor 3 (FGFR3) – a novel study in the Portuguese population.** Almeida MR, Campos-Xavier AB, Medeira A, Cordeiro I, Sousa AB, Lima M, Soares G, Rocha M, Saraiva J, Ramos L, Sousa S, Marcelino JP, Correia A, Santos HG. *Clin Genet*. 2009 Feb;75(2):150-6.
12. **Haploinsufficiency of TCF4 causes syndromal mental retardation with intermittent hyperventilation (Pitt-Hopkins syndrome)**. Christiane Zweier, Maarit M Peippo, Juliane Hoyer, Sérgio Sousa, Clayton-Smith, William Reardom, Jorge Saraiva, Alexandra Cabral, Ina Gohring, Koen Devriendt, Thomy de Ravel, Emília K Bijlsma, Raoul CM Hennekam, Alfredo Orrico, Monika Cohen, Alexander Dresweke, André Reis, Peter Nurnbeg, Anita Rauch. *American Journal of Human Genetics*. 2007 May;80(5):994-1001. Epub 2007 Mar 23.

CONFERENCES/TALKS, BY INVITATION *(selected)*

1. **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

2. **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

3. **From Genotype to Phenotype of monogenic disorders. Pathways and families of monogenic disorders.** Programa de Doutoramento em Ciências da Saúde, Faculdade de Medicina da Universidade de Coimbra, 17/11/2015.

4. **Mechanisms of disease. Skeletal dysplasias as na example**. Programa de Doutoramento em Ciências da Saúde, Faculdade de Medicina da Universidade de Coimbra, 17/11/2015.

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

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

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

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

9. **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.

ORAL PRESENTATIONS *(only oral communications as a first/presenting author were included; list of other oral communications as co-author and all posters available upon request)*

1. **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 as Clinical Investigation Award).

2. **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

3. **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.

4. **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

5. **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].

6. **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.

7. **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].

8. **Structural chromosomal rearrangements – Difficulties in prenatal genetic counselling.** VI Meeting of the Portuguese Prenatal Diagnose Centres, Vidago, Portugal – July 1-3, 2004.

9. **Noonan syndrome – clinical evaluation and molecular analysis of 11 cases.** 10ª Semana do Médico Interno do Centro Hospitalar de Coimbra. November 15-19, 2004.

10. **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.

11. **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].

OTHER ITEMS

Coordination roles:

- **Coordinator of the Rare Bone Disorders multidisciplinary team of the Centro Hospitalar e Universitário de Coimbra**, CHUC, (2014-present), the only Portuguese health-care provider centre member of the European Reference Network on Rare Bone Disorders (2017-present).
- **Coordinator of the Portuguese Study Group on Genetic Skeletal Disorders**, GruPEDGE, (2018-present)
- **Coordinator of the CHUC Genomic Interpretation Unit as part of the Project In2Genome** – consortium between Coimbra Genomics, Genoinseq and CHUC. (2017-present)

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.

Other:

- **Reviewer** for *American Journal of Human Genetics, American Journal of Medical Genetics*, *European Journal of Human Genetics*, European Journal of Medical

Genetics, and *Acta Pediátrica Portuguesa*.

- **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.