# Inês Girão Meireles de Sousa Cadeira da Silva

***email***: ines.meireles.sousa@gmail.com, ***linkedIN***: https://www.linkedin.com/in/ines-sousa-silva

## Experience

Jun 2019 – Present **Centro de Medicina Laboratorial Germano de Sousa**
*Molecular Genetics Specialist*

- Performed genetic variants classification, interpretation and diagnostic reports generation and revision.
- Provided scientific support to physicians, helping them to choose genetic tests and to interpret results.
- Engaged with both physicians and healthcare units.
- Participated in regulatory quality evaluation shema carried out by GenQA and EMQN for *BRCA* Variants Assessment in 2020.

Jun 2016 – May 2019 **Coimbra Genomics, S.A.**
*Genetics Specialist (2016), Healthcare Liaison (2017 to present)*

- Developed multiple *Genome Queries* for the Elsie platform (*in-house* exome/genome genetic tests developed to ask a specific medical question, with the respective production of a decision support semi-automatic report) and reviewed each decision support medical report.
- Provided scientific support to physicians, helping them to choose genetic tests and to interpret the results.
- Created targeted marketing materials for physicians and compiled clinical medical recommendations for genetic use.
- Engaged with both physicians and healthcare units during the activation process and ensured costumer success.
- Participated directly in the sales process by recruiting physicians and creating awareness for the Elsie platform.

Apr 2011 – May 2016 **Instituto de Medicina Molecular, Faculdade de Medicina da Universidade de Lisboa, Portugal.**
*PostDoc Position,*
*Genetics and genomics of primary spontaneous pneumothorax.*

Jul 2010 – Mar 2011 **Instituto Nacional de Saúde Dr. Ricardo Jorge, Portugal.**
*PostDoc Position*
*Genetics and functional genomics of Autism Spectrum Disorders.*

Jan 2006 – Jun 2010 **The Wellcome Trust Centre for Human Genetics, University of Oxford, UK**
*PhD in Clinical Medicine* (Approved with the highest mark).

Sept 2004 – Sept 2005 **Instituto Gulbenkian de Ciência, Portugal**
*Undergraduate research trainee* (Final classification – 19 out of 20).

Sept 2000 – Oct 2005 **Faculdade de Ciências, Universidade de Lisboa, Portugal**
*First degree in Microbial Biology and Genetics* (5 year degree).

## Selected Courses and Qualifications

See full list of courses taken on linkedin (https://www.linkedin.com/in/ines-sousa-silva/). Find below a list of a selected few:

- *Sales Training* – given by Nuno Correia Santos, held at Biocant Park, Cantanhede (January 2019).
- *Grant writing workshop - Making sense of the Science, a course on Effective Scientific Writing* – given by Dr. Judith A. Swan, held at IMM, Lisbon (October 2013).
- *Programming for Scientists Course* - held at IMM, Portugal (September 2012).
- *Bioinformatics Using Python for Biologists (BPB12)* – held at Instituto Gulbenkian de Ciência, Portugal (June 2012).
- *High Throughput Sequencing in Disease Studies* – Short course held at London School of Hygiene & Tropical Medicine, University of London (September 2011).
- *Statistical Course from the PhD programme in Genomic Medicine and Statistics*, held at Richard Doll Building, Oxford, UK (October/November 2008).

## Honours

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| Oct 2013 | *Prémio Robalo Cordeiro* - "I-GASP: an Integrative Genetic and Genomics Approach to Primary Spontaneous Pneumothorax susceptibility"<br>Sociedade Portuguesa de Pneumologia, Albufeira, Portugal |
| Jan 2012 | *L'Oréal - UNESCO For Women in Science* - "Genetics and genomics of primary spontaneous pneumothorax in the Portuguese population"<br>L'Oréal, FCT & UNESCO National Comitee, Lisbon, Portugal |
| May 2011 | *National Fellowship award* - "Whole-Genome CNV analysis identifies rare *ANXA1* duplications in Autism Spectrum Disorders"<br>European Society of Human Genetics, Amsterdam, Netherlands |
| Nov 2010 | *Prémio de Investigação Clínica* - "Whole-Genome CNV analysis identifies rare *ANXA1* duplications in Autism Spectrum Disorders"<br>14th meeting of *Sociedade Portuguesa de Genética Humana*, Coimbra, Portugal (sponsored by Alfagene) |
| Nov 2008 | *Prize scholarship 2008/2009*<br>Merton College, University of Oxford, UK<br>Prize with the aim of recognising and rewarding graduate students of the highest distinction. |
| Jan 2005 | *Excellency Award*<br>PT Multimédia, Portugal<br>Excellency Award for the best student of the year at the university level. |

## Selected Publications

I have to date **27 publications** in international peer-reviewed journals, with **more 4,500 citations** altogether (see full list on https://www.linkedin.com/in/ines-sousa-silva/). Find below a list of selected few (either recent publications or the most cited from 2006 to the present day):

- Masaki Takeuchi, Nobuhisa Mizuki, Akira Meguro, Michael J Ombrello, Yohei Kirino, Colleen Satorius, Julie Le, Mary Blake, Burak Erer, Tatsukata Kawagoe, Duran Ustek, Ilknur Tugal-Tutkun, Emire Seyahi, Yilmaz Ozyazgan, **Inês Sousa**, Fereydoun Davatchi, Vânia Francisco, Farhad Shahram, Bahar Sadeghi Abdollahi, Abdolhadi Nadji, Niloofar Mojarad Shafiee, Fahmida Ghaderibarmi, Shigeaki Ohno, Atsuhisa Ueda, Yoshiaki Ishigatsubo, Massimo Gadina, Sofia A Oliveira, Ahmet Gül, Daniel L Kastner, Elaine F Remmers (March 2017) Dense genotyping of immune-related loci implicates host responses to microbial exposure in Behçet's disease susceptibility. *Nature Genetics*, 49(3)438-443.
- **Inês Sousa**, Farhad Shahram, David Francisco, Fereydoun Davatchi, Bahar Sadeghi Abdollahi, Fahmida Ghaderibarmi, Abdolhadi Nadji, Niloofar Mojarad Shafiee, Joana M. Xavier, Sofia A. Oliveira (October

2015) Association of *CCR1*, *KLRC4*, *IL12A-ASN1*, *STAT4*, and *ERAP1* with Behçet's disease in Iranians. *Arthritis Rheumatol*, **67**(10):2742-8.

- **Inês Sousa**, Patrícia Abrantes, Vânia Francisco, Gilberto Teixeira, Marta Monteiro, João Neves, Ana Norte, Carlos Robalo Cordeiro, João Moura e Sá, Ernestina Reis, Patrícia Santos, Manuela Oliveira, Susana Sousa, Marta Fradinho, Filipa Malheiro, Luís Negrão, Salvato Feijó, Sofia A. Oliveira (May 2016) Multicentric genome-wide association study for primary spontaneous pneumothorax. *Plos One*, **11**(5):e0156103.
- The Autism Genome Project Consortium (July 2010) Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **466**(7304):368-72.
- The Autism Genome Project Consortium (March 2007) Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **39**(3):319–28.
- The Autism Genome Project Consortium (October 2010) A genome-wide scan for common alleles affecting risk for autism. *Hum Mol Genet*, **19**(20):4072-82.
- **I Sousa**, TG Clark, C Toma, K Kobayashi, M Choma, R Holt, NH Sykes, JA Lamb, AJ Bailey, A Battaglia, E Maestrini, AP Monaco, IMGSAC (June 2009) *MET* and autism susceptibility: family and case-control studies. *European Journal of Human Genetics*, **17**(6):749-58.

## Other Previous Experience

Organizer of the 1st Annual Joint Postdoctoral Meeting (IMM-IGC-Cedoc), at Praia Grande, Sintra, Portugal (Oct 2014).

Founding Member of the Postdoctoral Association (PDA) at the Instituto de Medicina Molecular, Lisbon, Portugal (Sep 2012-Sep 2013).

Organizer of the 1st Postdoctoral day and Scientific symposium, on behalf of the PDA at the Instituto de Medicina Molecular, Lisbon, Portugal (Oct 2013).

Volunteer Work at APPDA (*Portuguese Association for Development Disorders and Autism*) – (2004).

## Language Skills

Portuguese (mother tongue), English (fluent)