Curriculum Vitae

Nádia Pinto
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1. Personal Data

Place of Birth: Bragança, Portugal
Place of current address: Porto, Portugal
Citizenship: Portuguese

2. Academic Degrees

2012
PhD in Applied Mathematics, "General algorithms for computing genetic kinship likelihoods", Faculty of Sciences of University of Porto (FCUP), Porto, Portugal

2006
MSc in Mathematical Engineering, "Construction of algorithms for reconstruction of genealogies", translated from the Portuguese: "Construção de Algoritmos de Reconstrução de Genealogias", FCUP, Porto, Portugal

2003
BSc in Mathematics – Education branch, FCUP, Porto, Portugal

3. Summary

Highly accomplished and interdisciplinary researcher with a Ph.D. in Applied Mathematics, specialized in the quantitative analysis of complex biological and health-related data. Over 15 years of experience applying and developing mathematical and statistical models, encompassing biomathematics, statistics, bioinformatics and computational biology. Expertise spans diverse areas including human and non-human genetics (population genomics, kinship analysis, forensic applications), neurosciences, and providing crucial quantitative support across various biomedical research fields. Demonstrated success in securing competitive research grants, publishing high-impact work, delivering invited lectures, supervising students, coordinating international working groups, and contributing to academic teaching, professional consulting, and the organization of scientific events.

4. Professional and Academic Activities*

03-2023 to Present
Assistant Researcher, Population Genetics and Evolution group, i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto, Portugal. Competitive position: Individual Call to Scientific Employment Stimulus - 5th Edition, Panel: Basic and Clinical Medicine, Ref. 2022.04997.CEECIND, 4th ranked candidate out of 56.

01-2019 to 01-2023
Junior Researcher, Population Genetics and Evolution group, Institute of Molecular Pathology and Immunology of the University of Porto, i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto (IPATIMUP/i3S), Portugal. Decreto-Lei nº 57/2016 de 29 de Agosto.

10-2014 to 12-2018
Post-Doctoral Fellow, Population Genetics and Evolution group, Institute of Molecular Pathology and Immunology of the University of Porto (IPATIMUP/i3S) and Center of Mathematics of the University of Porto (CMUP), Portugal. Competitive Post-Doctoral grant, "Generalized Algorithms for kinship likelihood inferred from genetic markers" (Ref SFRH/BPD/97414/2013), IPATIMUP, Portuguese Foundation for Science and Technology (FCT) and POPH - QREN.

09-2013 to 09-2014
Post-Doctoral Fellow, Genes, Population genomics and Traits group, Research Center in Biodiversity and Genetic Resources (CIBIO), Vairão, Portugal. Competitive Post-Doctoral grant, "Assessing The Whole-Genome Structure and Variation of the Tropical Adapted Zebu Cattle (Bos indicus) using Dense SNP Maps." (Ref PTDC/CVT/117851/2010), CIBIO-inBIO, FCT/MCTES (PIDDAC) and FEDER through COMPETE (POFC) Program.

01-2008 to 07-2012
PhD student, Doctoral Program in Applied Mathematics, Faculty of Sciences of the University of Porto, Portugal. Competitive PhD grant, "General algorithms for computing genetic kinship likelihoods." (Ref SFRH/BD/37261/2007), IPATIMUP, FCT and POPH - QREN.

05-2005 to 07-2013
External Collaborator and Consultant, Centre Multimedia of Porto Editora: creation, development, implementation and validation of didactical software of Mathematics, Porto, Portugal

09-2002 to 08-2008
Teacher of Mathematics at several high schools (7th to 12th grades), Portugal

*NB: Maternity Leaves from 07-2010 to 12-2010; and 02-2014 to 07-2014.

5. Teaching Activities

2024/2025

Invited Lecturer, Course: Functional Genetics, Lecture: "A multimodal approach for deepening the understanding of neurological diseases – the case of Alzheimer's disease", Doctoral Progam in Molecular and Cell Biology, Instituto de Ciências Biomédicas Abel Salazar (ICBAS), Faculdade de Ciências (FCUP), Instituto de Investigação e Inovação em Saúde da Universidade do Porto (i3S), 27th March, 2025

2023/2024

Invited Lecturer, "Tópicos de genética forense y poblacional – Evaluación de parentescos", Universidad de Antioquia, Escuela de Microbiología, Medellín, Colombia, 19th-20th March, 2024

2022/2023

Invited Assistant Professor, Course: "DNA Analysis in Kinship Identification and Traceability", Master in Applications in Biotechnology and Synthetic Biology, Master in Bioinformatics and Computational Biology Faculty of Sciences of the University of Porto (FCUP), Porto, Portugal; University Teaching Career Statute Art 32-A

2014/2015 to 2022/2023

Invited Assistant Professor, Course: "Molecular Markers: Recombining Genomic Portions", Master in Forensic Genetics, Master in Cell and Molecular Biology, Faculty of Sciences of the University of Porto (FCUP), and GABBA - Doctoral Program in Areas of Basic and Applied Biology (2016/2017 and 2018/2019), University of Porto, Porto, Portugal; University Teaching Career Statute Art 32-A; University Teaching Career Statute Art 32-A

2015/2016

Invited Professor, Course: "Instrumental Methods in Biology" (45 hours, 3 credits), Postgraduate Program in Biosciences, Biomedical Center, Roberto Alcantara Gomes Biology Institute, University of the State of Rio de Janeiro, Brazil

2009/2010 to 2012/2013

Invited Lecturer, Curricular Unit: Molecular Markers: Recombining Genomic Portions, FCUP, Portugal

2002/2003 to 2007/2008

Teacher of Mathematics at several high schools (7th to 12th grades), Portugal

6. Consultant Activities

2012 to 2017 and since 2023: Consultant at IPATIMUP Diagnostics for the interpretation and statistical evaluation of identification, paternity and complex kinship analyses.

7. Publications

Author of **76 scientific works**, including **research papers (48), scientific dissemination books (1), book chapters (6), and Scopus-indexed conference proceedings (≥ 2 pages, 21)**. At time of publication, 43 out of the 48 research papers were published in JCR Q1 journals, serving in most as first (10), senior (13), corresponding (15) or other key authorship roles (2nd or 2nd to last, 14). Over the past five years, outputs have primarily arisen from funded national and international research projects under leadership, workplans of funded PhD students under supervision - with priority given to them as corresponding authors to foster independence, coordination of international working groups, and collaborations with other researchers involving quantitative and advanced statistical analyses. The complete list of publications is provided as Annex, with a summary of the publications 2020 ownwards.

Selected publications:
a. Macedo A et al. Risk Variants in Three Alzheimer's Disease Genes Show Association with EEG Endophenotypes. J Alzheimers Dis. 2021;80(1):209-223;

b. Xavier C, Pinto N.* Navigating the blurred boundary: Neuropathologic changes versus clinical symptoms in Alzheimer's disease, and its consequences for research in genetics. J Alzheimers Dis. 2025 Apr;104(3):611-626;

c. Faustino M et al. A mathematical framework for genetic relatedness analysis involving X chromosome aneuploidies. Forensic Sci Int Genet. 2025 Jan;74:103128; and

d. Antão-Sousa S et al. Microsatellites' mutation modeling through the analysis of the Y-chromosomal transmission: Results of a GHEP-ISFG collaborative study. Forensic Sci Int Genet. 2024 Mar;69:102999.

Book:
Amorim A, **Pinto N**, "An Introduction to Forensic Genetics for Non-Geneticists", ISBN: 9781003836599, Edition: 12-2023, Editor: CRC PRESS. doi: 10.1201/9781003266716.

8. Competitive Scientific Projects

8.1. Ongoing

09-2025 to 08-2029
CapCell - "Innovative forensic trace investigation via microfluidics and single-cell genomics" European Commision - Horizon Europe; HORIZON-CL3-2024-FCT-01-02

Beneficiaries: Universiteit Maastricht, NL (coordinator), Katholieke Universiteit Leuven, BE, i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto **(co-PI)**, Medizinische Universitat Innsbruck At, Netherlands Forensic Institute Nl, Eesti Kohtuekspertiisi Instituut Ee, Politsei- Ja Piirivalveamet Ee, Bundesministerium Fur Inneres At, Copan Italia Spa It, Nimagen Bv Nl, Voxdale Be
Budget (Total): 4,499,308.75€; Budget (i3S): 204,200€

09-2025 to 08-2029
ForMat - "Forensic methylation analysis toolsets"
European Commision - Horizon Europe; HORIZON-CL3-2024-FCT-01-02
Beneficiaries: Universidad De Santiago De Compostela ES (Coordinator), King's College London UK, i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto (**co-PI**), Medizinische Universitat Innsbruck AT, Netherlands Forensic Institute NL, Pomorski Uniwersytet Medyczny W Szczecinie PL, Uniwersytet Jagiellonski PL, Polismyndigheten Swedish Police Authority SE, Bundeskriminalamt DE, Universidade do Minho PT, Euro-Funding Eu Projects Sociedad Limitada ES
Budget (Total): 4,673,455.25€; Budget (i3S): 211,078.75€

03-2024 to 02-2028
"Natural Traces in forensic investigations - how the analysis of non-human evidence can solve crime"
European Research Executive Agency; HORIZON TMA Marie Skłodowska-Curie Actions - HORIZON-MSCA-2022-DN-01-01
Beneficiaries: Johann Wolfgang Goethe-Universitaet, Frankfurt Am Main, Germany (Coordinator, scientist in charge: Jens Amendt, Institute of Legal Medicine); Eotvos Lorand Tudomanyegyetem, Hungary; Universidad Autonoma De Madrid, Spain; Univerzita Karlova Czech Republic; Tallinna Tehnikaülikool, Estonia; Lunds Universitet, Sweden; Universita Degli Studi Di Genova, Italy; Universita Degli Studi Di Modena E Reggio Emilia, Italy; Universidade Do Porto (Associated Partner: i3S – Instituto de Investigação e Inovação em Saúde da Universidade do Porto, **PI**), Portugal
Budget (Total): 2,532,931.20€, Budget (UP/i3S): 243,403.20€

03-2023 to 02-2027
"MultiAD - A multimodal approach for deepening the understanding of Alzheimer's disease", 2022.04734.PTDC
Fundação para a Ciência e Tecnolgia, Portugal; Call for R&D Projects in All Scientific Domains – 2022, 1st ranked project in Neurosciences Panel. Clinical collaboration: ULS de Santo António, Clínica de Neurociências, Porto, PT.
Beneficiary: i3S – Instituto de Investigação e Inovação em Saúde da Universidade do Porto, **PI**.
Budget (Total, i3S): 249.843,44€

09-2023 to 08-2026
"The chronnectomic brain: uncovering the neuropathological fingerprints in dementia"

State Agency of Research, Spain; Convocatoria 2022 - «Proyectos de Generación de Conocimiento»
Beneficiary: University of Valladolid (UVA), **Team Member**.
Budget (UVA, Total): 271,851€

07-2022 to 06-2026
"Time-dependent gene expression patterns of RNA markers for wound age estimation in forensic pathology"
Alexander von Humboldt Foundation; Funding from the Research Group Linkage Programme
Beneficiary: Institute of Legal Medicine, University Hospital of Cologne (UHC), Germany, **Team Member.**
Budget (UHC, Total): 54,973 €

8.2. Approved

01-2026 to 06-2027 (Tentative)
"AgeAD - Genomic Signatures of Age-of-Onset Variation in Sporadic Alzheimer's Disease"
Fundação para a Ciência e Tecnolgia, Portugal; Call for Exploratory Research Projects in all Scientific Domains, 2024. Clinical collaboration: ULS de Santo António, Clínica de Neurociências, Porto, PT.
Beneficiary: i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto (**PI**).
Budget (Total, i3S): 59.912,50€

8.3. Concluded

09-2019 to 12-2022
"Analysis and correlation between epigenetics and brain activity to assess chronic and episodic migraine risk in women"; Original title (in Spanish): "Análisis y correlación entre la epigenética y la actividad cerebral para evaluar el riesgo de migraña crónica y episódica en mujeres", 0702_MIGRAINEE_2_E, https://gib.tel.uva.es/migrainee/?lang=en_EN
European Commision; Programa INTERREG V A España Portugal (POCTEP)
Beneficiaries: Universidad de Valladolid (Proponent Institution), IPATIMUP (**PI**), Instituto de Biologia Molecular e Celular, Fundación Instituto de Estudios Ciencias de la Salud de Castilla y León
Budget (Total): 564.373,76€; Budget (IPATIMUP): 118.366,06€

06-2017 to 12-2020
"Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease"; Original title (in Spanish): "Análisis y correlación entre el genoma completo y la actividad cerebral para la ayuda en el diagnóstico de la enfermedad de Alzheimer", 0378_AD_EEGWA_2_P, https://gib.tel.uva.es/ad-eegwa/?lang=en_EN
European Commision; Programa INTERREG V A España Portugal (POCTEP)

Beneficiaries: IPATIMUP (**Proponent Institution**, **PI**), Universidad de Valladolid, Asociación de Familiares y Amigos de Enfermos de Alzheimer y otras demencias de Zamora, Associação Portuguesa de Familiares e Amigos de Doentes de Alzheimer
Budget (Total): 543.404,48€; Budget (IPATIMUP): 164.734,43€

8.4. Under Evaluation

1. "MIGRAINET - Consolidation of the collaborative network for multimodal prediction of migraine treatment response "; Original title (in Spanish): "MIGRAINET - Consolidación de la red de colaboración para la predicción multimodal de la respuesta al tratamiento de la migraña"
   European Commision; Programa INTERREG VI A España Portugal (POCTEP)
   Beneficiaries: Universidad de Valladolid (Proponent Institution), i3S - Instituto de Investigação e Inovação em Saúde da Universidade do Porto (**PI**), Instituto de Ciências Biomédicas Abel Salazar da Universidade do Porto, Fundación Instituto de Estudios Ciencias de la Salud de Castilla y León
   Budget (Total): 1.090.899€; Budget (i3S): 230.445€
2. "Exploratory Research on the Impact of Dietary Phosphate Reduction and Phosphate Binder Therapy on Endothelial Function and Gut Microbiota in Non-Dialysis Chronic Kidney Disease Patients"
   Fundação para a Ciência e Tecnolgia, Portugal; HEALTH + SCIENCE: Call for Exploratory Research Projects 2025
   Beneficiary: Universidade do Porto - Faculdade de Medicina da Universidade do Porto (**Team member**)

9. Supervisions

9.1. PhD Students

9.1.1. Ongoing

Since 10-2022
Supervisor, Rebelo M.
"Deepening the understanding of Alzheimer's disease risk and progression: a multimodal approach", Doctoral Program in Computer Sciences, Faculty of Sciences of the University of Porto, Porto, Portugal

Since 10-2022
Co-Supervisor, Rodrigues P.
"Establishing microhaplotype panels using massively parallel sequencing as a new tool in forensic genetics", Doctoral Program in Biology, Faculty of Sciences of the University of Porto, Porto, Portugal. FCT fellow Ref. 2022.11825.BD

Since 11-2021
Supervisor, Faustino M.
"Levering the statistical assessment of X-chromosomal evidence in forensic genetics", Doctoral Program in Biology, Faculty of Sciences of the University of Porto, Porto, Portugal. FCT fellow Ref. 2021.08783.BD

Since 10-2021
Supervisor, Costa C.
"Dismantling blind-trusted Black Boxes: Testing the limits and sensitivity of forensic DNA software", Doctoral Program in Biology, Faculty of Sciences of the University of Porto, Porto, Portugal. FCT fellow Ref. 2021.05655.BD

9.1.2. Concluded

02-2016 to 11-2023
Supervisor, Garcia M.G.
"Population genetic study of Argentina for the establishment of haplotype frequencies and mutations in 15 markers of the X chromosome", Doctorate in Biomedical Sciences, Faculty of Medical Sciences, Pontificia Universidad Católica Argentina, Buenos Aires, Argentina.

09-2018 to 06-2023
Supervisor, Antão-Sousa A.
"Uncovering microsatellites' mutational mechanisms", Doctoral Program in Biology, Faculty of Sciences of the University of Porto, Porto, Portugal. FCT fellow Ref. SFRH/BD/136284/2018

9.1.3. Approved

Tentative Starting Date: 10-2025
Co-Supervisor, Bakumenko A.
"Development of Computational Methods for the Analysis of Mixed Biological Trace Evidence based on Deep Learning and Bayesian Inference", Doctoral Program in Computer Sciences (tentative), Faculty of Sciences of the University of Porto, Porto, Portugal.
Grant Information: Work Plan to be developed under the European Training Network "Natural Traces" European Commission Research Framework Programme: Horizon Europe - Marie Skłodowska-Curie Actions. Reference Number: 101120165 – Natural Traces. Marie Curie Grant Agreement Number: 101120165

9.2. MSc Students

09-2022 to 12-2023
Co-Supervisor, Alegria M.
"Characterization of STR sequence variants on the X- and Y- chromosomes", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2020 to 11-2021
Supervisor, Faustino M.
"Haplotypic polymorphisms and mutation rate estimates of Y-STRs in the Portuguese population", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2019 to 12-2020
Supervisor, Costa C.
"Quantification of the forensic genetics proof: Evaluating the impact of different statistical approaches", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2018 to 11-2019
Co-supervisor, Macedo A.
"Quantifyng the genetic predisposition to a complex disease through genome-wide association", MSc in Mathematical Engineering, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2017 to 11-2018
Supervisor, Figueiredo C.
"Comparison and validation of software for mixture analyses", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2017 to 11-2018
Co-supervisor, Ribeiro J.
"The influence of Brugada syndrome in the diagnosis of the sudden death", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2016 to 11-2017
Supervisor, Machado P.
"The influence of mutation models in kinship likelihoods.", MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2016 to 11-2017
Supervisor, Antão-Sousa S.
"Estimation of bi-allelic mutation rates at Y-STRs." MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2016 to 11-2017
Co-supervisor, Simões R.

"Distinguishing kinships beyond identity and paternity." MSc in Bioinformatics, Engineering School of the University of Minho, Braga, Portugal

09-2015 to 11-2016

Co-supervisor, Fadoni J.

"Genetic analysis of haplotypic data for 17 Y-chromosome short tandem repeat loci in the population of São Paulo, Brazil." MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2010 to 12-2011

Co-supervisor, Gomes C.

"Forensic application of the study of 12 STRs: utility in different cases of biological kinship investigation" (in Portuguese, "Aplicações forenses do estudo de 12 X-STRs: Utilidade em diferentes casos de investigação de parentesco biológico"), MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

09-2010 to 12-2011

Co-supervisor, Magalhães M.

"Insertion/deletion polymorphisms in paternity investigations involving close relatives of the real father" (in Portuguese, "Polimorfismos de inserção/delecção em investigações de paternidade envolvendo parentes próximos do verdadeiro pai"), MSc in Forensic Genetics, Faculty of Sciences of the University of Porto, Porto, Portugal

9.3. Researchers, PhD

Since 04-2023 (36 months)

Xavier C, PhD, hired under the framework of the project "A multimodal approach for deepening the understanding of Alzheimer's disease", 2022.04734.PTDC

07-2022 to 12-2022

Xavier C, PhD, hired under the framework of the project "Analysis and correlation between epigenetics and brain activity to assess chronic and episodic migraine risk in women", 0702_MIGRAINEE_2_E

05-2020 to 08-2020

Lopes A, PhD, hired under the framework of the project "Analysis and correlation between epigenetics and brain activity to assess chronic and episodic migraine risk in women", 0702_MIGRAINEE_2_E

9.4. Fellowships

9.4.1. Post-Doc Fellows

03-2018 to 04-2019

Supervisor, Gomes I, Post-Doc fellow, Hired under the framework of the project "Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease", 0378_AD_EEGWA_2_P

9.4.2. MSc Fellows

09-2021 to 12-2022

Co-Supervisor, Carvalho E, MSc fellow, hired under the framework of the project "Analysis and correlation between epigenetics and brain activity to assess chronic and episodic migraine risk in women", 0702_MIGRAINEE_2_E

07-2019 to 12-2019

Co-Supervisor, Rebelo M, MSc fellow, hired under the framework of the project "Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease", 0378_AD_EEGWA_2_P

05-2019 to 12-2019

Supervisor, Rocha R, MSc fellow, hired under the framework of the project "Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease", 0378_AD_EEGWA_2_P

9.4.3. BSc Fellows

10-2021 to 12-2022

Co-Supervisor, Felício D, BSc fellow, hired under the framework of the project "Analysis and correlation between epigenetics and brain activity to assess chronic and episodic migraine risk in women", 0702_MIGRAINEE_2_E

07-2019 to 12-2019

Co-Supervisor, Cunha R, BSc fellow, hired under the framework of the project "Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease", 0378_AD_EEGWA_2_P

05-2019 to 12-2019

Supervisor, Macedo A, BSc fellow, hired under the framework of the project "Analysis and correlation between the whole genome and brain activity to aid in the diagnosis of Alzheimer's disease", 0378_AD_EEGWA_2_P

9.5. Academic Internships

9.5.1. PhD Students

09-2025 to 12-2025 (Planned)
Supervisor, González-Bao J., Doctoral Program from Facultade de Medicina, Universidade de Santiago de Compostela, Spain

03-2023 to 07-2023
Supervisor, Minervino A., Doctoral Program in Molecular and Cell Biology, ICBAS, FCUP, Porto, Portugal

9.5.2. BSc Students

03-2023 to 07-2023
Supervisor, Pereira E., BSc in Genetics and Biotechnology, UTAD, Vila Real, Portugal

09-2020 to 07-2021
Supervisor, Nascimento M., BSc in Biology, FCUP, Porto, Portugal

10. International Working Commissions

10.1. Coordination

Since 06-2024
Working Group "Quantification of the weight of the mixtures evidence in identification problems", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG). Coordinators: **Nádia Pinto**, Lourdes Prieto (Universidad de Santiago de Compostela, Comisaría General de Policía Científica, Spain), Camila Costa (i3S, FCUP, Portugal). Senior author of the corresponding paper in final preparation (15 participating worldwide laboratories).

Since 09-2017
Working Group "Segregation on X-STRs", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG). Coordinator: **Nádia Pinto**. Senior and corresponding author of the corresponding paper in final preparation (6 participating worldwide laboratories).

09-2019 to 10-2021
Working Group "Study of mutations in Y-STRs", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG). Coordinators: **Nádia Pinto**, Antóno Amorim (IPATIMUP/i3S, FCUP) and Leonor Gusmão (UERJ, Rio de Janeiro, Brazil).

Senior and corresponding author of the resulting paper (20 participating worldwide laboratories): doi: 10.1016/j.fsigen.2023.102999

09-2018 to 10-2021

Working Group "Study of mutations on one set of 12 X-STRs - Extension", GHEP-ISFG. Coordinators: **Nádia Pinto**, Leonor Gusmão (UERJ, Rio de Janeiro, Brazil) and Gabriela Garcia (Manlab, Buenos Aires, Argentina). Senior author of the resulting paper (46 participating worldwide laboratories): doi: 10.1016/j.fsigen.2025.103232

09-2017 to 09-2019

Working Group "Study of mutations on one set of 12 X-STRs", GHEP-ISFG. Coordinators: **Nádia Pinto**, Leonor Gusmão (UERJ, Rio de Janeiro, Brazil) and Gabriela Garcia (Manlab, Buenos Aires, Argentina). First and corresponding author of the resulting paper (18 participating worldwide laboratories): doi: 10.1016/j.fsigen.2020.102258

10.2. Invited Assessor

03-2014 to 11-2016

"DNA Commission on Software Validation", International Society for Forensic Genetics (ISFG). Seven international experts invited. Co-author of the resulting paper: 10.1016/j.fsigen.2016.09.002

05-2015 to 09-2015

"Kinship Paper Challenge – Advanced Level" in the "Intercomparison Program 2015: Analysis Of Dna Polymorphisms In Bloodstains And Other Biological Samples", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG).

10.3. Participation

Since 10-2023

Working Group "DVI-3, Identificación de Victimas de una Masacre en una Fosa Común", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG).

09-2015 to 11-2016

Working Group "Expression and Reporting of DNA results", Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG). Senior author of the resulting paper doi: 10.1016/j.fsigen.2016.09.003

11. Member of Organizing and/or Scientific committees

11.1. Scientific events

2024

20th Portugaliæ Genetica, 20th to 22th of March, 2024, i3S - Instituto de Investigação e Inovação em Saúde, Porto, Portugal. Member of Organizing Committee.
https://www.i3s.up.pt/event.php?v=290

2016

17th Portugaliæ Genetica, 17th and 18th of March, 2016, IPATIMUP/i3S, Porto, Portugal. Member of Organizing and Scientific Committee.
http://www.ipatimup.up.pt/portugaliaegenetica2016/index.html

11.2. Cycles of workshops

2023

Cycle of workshops in Forensic Genetics, 23th and 24th of March, 2023, i3S - Instituto de Investigação e Inovação em Saúde, Porto, Portugal. Member of Scientific Committee. https://www.i3s.up.pt/event.php?v=250

2018

Cycle of workshops in Forensic Genetics, Spanish and Portuguese-Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG) and FCUP, 25th and 26th of October, 2018, Porto, Portugal. Member of Organizing Committee. https://ghep-isfg.org/en/workshops/porto-2018/

11.3. Dissemination of knowledge

2025

International Symposium Antonio Amorim, 21st of March, 2025, i3S - Instituto de Investigação e Inovação em Saúde, Porto, Portugal. Member of Organizing Committee. https://www.i3s.up.pt/event?v=348

12. Invited Lecturer

12.1. Workshops

1. **Workshop** of the Spanish and Portuguese-Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), Online. "Uso del software Familias para la valoración

estadística de parentescos", 4 Half-days with Lourdes Prieto, Laboratorio de ADN de la Comisaría General de Policía Científica, Madrid, Spain, 7th, 14th, 21th, 28th, Octobre 2025, https://ghep-isfg.org/pt/workshops/ghep-os/autumn2025/WS1/ - To happen

2. **Workshop** "Complex kinship analyses using both unlinked and linked markers", 1 ½ days, Section of Forensic Genetics, Copenhagen University, Denmark, 19th – 20th June, 2025.

3. **Workshop** of the Spanish and Portuguese-Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), Online. "Análisis estadístico, interpretación y comunicación en genética forense", 4 Half-days with Andrea Sala, Facultad de Farmacia y Bioquímica, Universidad de Buenos Aires, Argentina, 9th, 16th, 23th, 30th, Octobre 2024, https://ghep-isfg.org/pt/workshops/ghep-os/autumn2024/WS3/

4. **Workshop** at XXIX Meeting of the Spanish and Portuguese-Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), Salta, Argentina. "Marcadores X-STR em análises de parentesco", full-day workshop with Leonor Gusmão, State University of Rio de Janeiro, Brazil, 4th June 2024, https://ghep-isfg.org/pt/workshops/ws2s24/

5. **Workshop** at Cycle of Workshops in Forensic Genetics, Instituto de Investigação e Inovação em Saúde, Porto, Portugal, Workshop 3 | X-chromosomal markers: kinship analyses (Half-day workshop), with Daniel Kling, Department of Forensic Sciences, Forensic Genetics Research Group, Oslo University Hospital, Oslo, Norway (90 min), 24th March 2023, Pinto N: 90 min, https://www.i3s.up.pt/event.php?v=250

6. **Workshop** at XXV National Congress of Criminalistics, Goiânia, Brazil. "Statistical analyses in simple and complex genetic kinships" (in Portuguese "Análise estatística em parentescos genéticos simples e complexos"), 1st October 2019, Pinto N: 240 min, https://win.iweventos.com.br/evento/criminalistica2019/programacao/gradeatividades/25

7. **Workshop** at Cycle of Workshops in Forensic Genetics, Faculty of Sciences of the University of Porto, Portugal. Session: "Disaster Victim Identification (DVI) – Applications and Statistics ", Subsection: "Disaster Victim Identification: General Principles and Theoretical Framework", 25th October 2018, Pinto N: 75 min, https://ghep-isfg.org/usercontent/content/uploads/medios/Final-Agenda_Main-w-sponsors_2.pdf

8. **Workshop** at XXI Meeting of the Spanish and Portuguese-Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), Bayahíbe, Dominican Republic. "Familial testing, X-files, FamLinkX", full-day workshop with Leonor Gusmão, State University of Rio de Janeiro, and Thore Egeland Norwegian Universty of Life Sciences, 6th September 2016, Pinto N: 75 min, https://familias.name/ghep2016/

9. **Workshop** at Instituto Universitario de Investigación en Ciencias Policiales, Universidad de Alcalá, Alcalá de Henares (Madrid), Spain. "Workshop Interpretation of mtDNA and Sex Chromosome Results in the Forensic Field", two days workshop with Leonor Gusmão, Institute of Pathology and Molecular Immunology of the University of Porto, Walther Parson, Institute of Legal Medicine, Innsbruck Medical University, and Lourdes Prieto, Comisaría General de Policía Científica, University Institute of Research in Forensic Sciences (IUICP), Madrid, Spain, 7th – 8th September, 2011, Pinto N: 195 min.

12.2. Scientific seminars

1. **Seminar** at Cologne Center for Genomics (CCG), University of Cologne, Germany. "Beyond Autosomal and Y-Chromosomal Markers: The Role of X-Chromosome in Population and Forensic Genetics", hosted by Prof. Michael Nothnagel, head of Statistical Genetics and Bioinformatics Group (45 min), 30th April 2024

2. **Seminar** at XXV National Congress of Criminalistics (in Portuguese "Congresso Nacional de Criminalística"), Goiânia, Brazil. "Quantification of the genetic proof: challenges in the present and future perspectives" (in Portuguese "Quantificação da Prova Genética: Desafios no Presente e Perspetivas Futuras"), 2nd October 2019, Pinto N: 90 min, https://win.iweventos.com.br/evento/criminalistica2019/programacao/gradeatividades/26

3. **Seminar** at XV Portugaliae Genetica, Institute of Pathology and Molecular Immunology of the University of Porto - IPATIMUP, Porto, Portugal. "Genealogies and Genetic Kinships: Related but Dissimilar Stories", 23rd March 2012, 40 min https://www.ipatimup.pt/Site/ActivityView.aspx?EventId=5&ActualActivityId=848&ActivityId=1584

12.3. Seminars for dissemination of knowledge

1. "Investigação científica na demência", 16th September 2023, Café Memória, Casa da Juventude de Esposende, Esposende, Portugal.

2. "Analysis and correlation between genomics and eletroencephalogram measurments in Alzheimer disease", 29th November 2019, Living with dementia in our home, Alzheimer Portugal, Hospital Magalhães de Lemos, Porto, Portugal

3. "The Mathematics of Forensic Genetics", 10th October 2018, "The year of Mathematical Biology", Clube de Ciência Viva da Escola Secundária Aurélia de Sousa, Porto, Portugal.

4. "Forensic Genetics: Much more than just a human affair", 23rd November 2017, "Semana da Ciência e Tecnologia: Ciência e Cidadania", Life Sciences and Environment School, University of Trás-os-Montes and Alto Douro, Vila Real, Portugal.

5. "Validating software to estimate genetic relatedness – one small step for algebra, one giant leap for forensics", 18th April 2016, Cycle of conferences: "Retratos de Empregabilidade", Faculty of Sciences of the University of Porto, Porto, Portugal

6. "The mathematics of Forensic Genetics", 10th February 2015, "Are you Biocriative?", JorTec Biologia – Biology Study Days of Faculty of Sciences and Technology of University Nova of Lisbon, Lisbon, Portugal

13. Selected Oral Communications

* Presented by

1. Faustino M*, Gurmão L, Amorim A, Kling D, **Pinto N**, The Challenge of X Chromosome Aneuploidies in Kinship Analyses, 30th Congress of the International Society for Forensic Genetics, Santiago de Compostela, Spain, Sep 09 - 13, 2024

2. **Pinto N***, Antão-Sousa S, Amorim A, Gusmão L, Are marker-specific average mutation rates the best option for biostatistical calculations?, 12th Haploid Markers Conference, Budapest, Hungary, May 17-20, 2023

3. Antão-Sousa S, Conde-Sousa E, Gusmão L, Amorim A, **Pinto N***, How often have been X- and aut-STRs mutations equivocal parental origin assigned?, 29th Congress of the International Society for Forensic Genetics, Washington DC, USA, Aug 29 - Sep 02, 2022

4. Costa C*, Figueiredo C, Amorim A, Prieto L, Costa S, Ferreira PM, **Pinto N**, Statistical analysis tools of mixture DNA samples: When the same software provide different results, 29th Congress of the International Society for Forensic Genetics, Washington DC, USA, Aug 29 - Sep 02, 2022

5. Macedo A M*, Gomes I, Martins S, Durães L, Sousa P, Figueruelo M, Rodríguez M, Pita C, Rebelo M, Arenas M, Alvarez L, Hornero R, Gómez C, **Pinto N**, Lopes A M, Genome-wide characterization of a cohort of Alzheimer's patients from Iberia: a focus on rare variants, 22nd Annual Meeting of the Portuguese Society of Human Genetics, Bencanta, Portugal, Nov 14 – 16, 2019

6. **Pinto N**, Conde-Sousa E, Chen S, Pérez-Pardal L, Goyache F, Beja-Pereira A*, Computational tools to exploit cattle exomes. 34th Conference of International Society of Animal Genetics (ISAG), Xi'an, China, Jul 28 – Aug 1, 2014

7. Pereira V*, Tomas Mas C, **Pinto N**, Amorim A, Gusmão L, Prata MJ, Morling N, Assessing the potential application of X-chromosomal haploblocks in population genetics and forensic studies,

25th World Congress of the International Society for Forensic Genetics (ISFG), Melbourne, Australia, Sep 2 – 6, 2013

8. Magalhães M, **Pinto N***, Gomes C, Pereira R, Amorim A, Alves C, Gusmão L – When the alleged father is a close relative of the real father: the utility of insertion/deletion polymorphisms. 24th World Congress of the International Society for Forensic Genetics (ISFG), Vienna, Austria, Aug 30 – Sep 4, 2011

9. Pereira R*, Phillips C, **Pinto N**, Santos C, Santos SEB, Amorim A, Carracedo A, Gusmão L - A panel of 46 Ancestry-Informative Insertion-Deletion polymorphisms (AIM-INDELs) in a single reaction. 24th World Congress of the International Society for Forensic Genetics (ISFG), Vienna, Austria, Aug 30 – Sep 4, 2011

10. **Pinto N***, Gusmão L, Amorim A - Distinguishing kinship from genealogical likelihoods. 23rd World Congress of the International Society for Forensic Genetics (ISFG), Buenos Aires, Argentina, Sep 15 – 18, 2009

14. Travel Grants

08-2022
Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), XXVII Jornadas, Washington DC, United States of America.

01-2019
International Society for Forensic Genetics short-term fellowship (~2 weeks) – DNA Diagnostics Laboratory, State University of Rio de Janeiro, Brazil.

20-2017
Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG), XXII Jornadas, Coimbra, Portugal.

Annex: Full list of publications

Annex to Curriculum Vitae: Full list of Publications

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1. Publications

*****Corresponding author; †The authors contributed equally to the work.

1.1. Research Papers in Scopus-Indexed Journals

1.1.1. Published

1. Lobo S, Dias A, Pedro A, Ferreira M, Oliveira A, São José C, Herrera-Mullar J, **Pinto N**, Colas C, Hüneburg R, Nattermann J, Boussemart L, van Hest L, Moreira L, Horton C, Farengo Clark D, Tinschert S, Golmard L, Spier I, López-Fernández A, Oliveira D, Svrcek M, Bourgoin P, Coulet F, Delhomelle H, Davis J, Zäncker B, Lazaro C, Guerra J, Almeida M, Revilla S, Patiño García A, Gundlach P, Laszkowska M, Strong V, Teixeira M, Schrader K, Steinke-Lange V, Gullo I, Sousa S, Batista M, Aretz S, Balmaña J, Aronson M, Perazzolo Antoniazzi A, Palmero E, Mansfield P, Van der Kolk L, Cats A, van Dieren J, Castellvi-Bel S, Katona B, Karam R, Pereira P, Benusiglio P, Oliveira C*. Hereditary diffuse gastric cancer spectrum associated with germline CTNNA1 loss of function revealed by clinical and molecular data from 351 carrier families and over 37.000 non-carrier controls. Gut. 2024, **Accepted (22.08.2025)**. Summary: *Genotype-phenotype analysis showing a strong association between CTNNA1 truncating variants and an increased risk of hereditary diffuse gastric cancer. Multivariable logistic regression and other methods were applied.* ***Collab w/ C Oliveira's i3S lab.***
2. Costa C*, Pereira E, Figueiredo C, Costa S, Ferreira PM, Amorim A, Prieto L, **Pinto N.** Consequences of Version Updates in Probabilistic Genotyping Software for Forensic Genetics: A Casework-Based Evaluation. Genes; **Accepted (22.08.2025)**. Summary: *Assessment of the impact of advanced stutter modelling (back and forward stutter) in probabilistic genotyping software. Enhanced computational models in newer software versions improved quantitative fit and interpretative accuracy, especially in complex or degraded mixtures. Model updates changed statistical results in select cases, underscoring the importance of algorithmic evolution in forensic genomics.* ***Collab w/ Portuguese Scientific Laboratory of the Judiciary Police (LPC-PJ).***
3. Maturana-Candelas A*, Hornero R. Poza J, Rodríguez-González V, Pablo VG, **Pinto N**, Rebelo M, Gómez C. Effect of MAPT gene variations on the brain electrical activity: a multiplex network study. Biomedical Signal Processing and Control. 2025 December. doi:10.1016/J.BSPC.2025.108129. Summary: *Multiplex network approach the relationship between genetics and neurophysiological dynamics, specifically the effect of variations in the MAPT gene on brain electrical activity, as*

measured by EEG. ***Collab w/ Biomedical Engineering Group from Valladolid University (GiB – UVA).***

4. Costa C*, Figueiredo C, Costa S, Ferreira PM, Amorim A, Prieto L, **Pinto N.** The impact of considering different numbers of contributors in identification problems involving real casework mixture samples. Int J Legal Med. 2025 May 9. doi: 10.1007/s00414-025-03500-7. Epub ahead of print. PMID: 40343479. Summary: *Data-driven study to evaluate the impact and practical implications of estimating the number of contributors on the interpretation of complex real casework mixture samples, through analysis of the computational models used in Bayesian probabilistic genotyping.* ***Collab w/ LPC-PJ***.
5. Rodrigues P, **Pinto N**, Otterlund T, Jønck CG, Prata MJ, Børsting C, Pereira V. Enhancing the Potential of Microhaplotypes for Forensic Applications: Insights from Afghan and Somali Populations. Genes (Basel). 2025 Apr 29;16(5):532. doi: 10.3390/genes16050532. PMID: 40428354; PMCID: PMC12111283. Summary: *Study focused on enhancing the potential of microhaplotypes for forensic applications, applying and optimizing specific bioinformatics models for population-based genetic analysis to improve forensic methodologies.* ***Collab w/ Copenhagen University (DK).***
6. Xavier C, **Pinto N.*** Navigating the blurred boundary: Neuropathologic changes versus clinical symptoms in Alzheimer's disease, and its consequences for research in genetics. J Alzheimers Dis. 2025 Apr;104(3):611-626. doi: 10.1177/13872877251317543. Epub 2025 Feb 16. PMID: 39956949. Summary: *Paper that critically examines the use of increasingly large datasets in genome-wide association studies for Alzheimer's disease, highlighting the importance of data quality, diagnostic methods, and potential biases, showcasing expertise in advanced genetic research methodologies and critical bioinformatics assessment*.
7. Gusmão L, Antão-Sousa S, Faustino M, Abovich MA, Aguirre D, Alghafri R, Alves C, Amorim A, Arévalo C, Baldassarri L, Barletta-Carrillo C, Berardi G, Bobillo C, Borjas L, Braganholi DF, Brehm A, Builes JJ, Cainé L, Carvalho EF, Carvalho M, Catelli L, Cicarelli RMB, Contreras A, Corach D, Di Marco FG, Diederiche MV, Domingues P, Espinoza M, Fernandéz JM, García MG, García O, Gaviria A, Gomes I, Grattapaglia D, Henao J, Hernandez A, Ibarra AA, Lima G, Manterola IM, Marrero C, Martins JA, Mendoza L, Mosquera A, Nascimento EC, Onofri V, Pancorbo MM, Pestano JJ, Plaza G, Porto MJ, Posada YC, Rebelo ML, Riego E, Rodenbusch R, Rodríguez A, Rodríguez A, Sanchez-Diz P, Santos S, Simão F, Siza Fuentes LM, Sumita D, Tomas C, Toscanini U, Trindade-Filho A, Turchi C, Vullo C, Yurrebaso I, Pereira V, **Pinto N.*** X-chromosomal STRs: Metapopulations and mutation rates. Forensic Sci Int Genet. 2025 Mar;76:103232. doi: 10.1016/j.fsigen.2025.103232. Epub 2025 Jan 27. PMID: 39893847. Summary: *Compilation and analyses of nearly 25,000 X-STR haplotypes and 3,700+ segregation trios, providing one of the most extensive population datasets for forensic genetics (equivalent to 74% of the amount produced so far). Advanced bioinformatics and computational models enabled grouping metapopulations, assessing genetic structure, and estimating locus-specific mutation rates with parental and age effects. The work highlights differentiation among continents and the limitations of current models, emphasizing the need for more data-driven computational approaches for mutation rates and haplotype frequencies in kinship analysis.* ***Collab w/ 46 worldwide laboratories from a GHEP-ISFG working group (coordinator).***

8. Costa C*, Figueiredo C, Costa S, Ferreira PM, Amorim A, Prieto L, **Pinto N**. The impact of parameter variation in the quantification of forensic genetic evidence. Sci Rep. 2025 Jan 20;15(1):2524. doi: 10.1038/s41598-024-83841-2. PMID: 39833192; PMCID: PMC11756398. Summary: *Sensitivity analyses regarding the bioinformatic modelling of key parameters on the statistical weight of mixture DNA evidence, with compromised DNA quantity/quality*. ***Collab w/ LPC-PJ.***
9. Faustino M*, Gusmão L, Amorim A, Kling D, **Pinto N**. A mathematical framework for genetic relatedness analysis involving X chromosome aneuploidies. Forensic Sci Int Genet. 2025 Jan;74:103128. doi: 10.1016/j.fsigen.2024.103128. Epub 2024 Aug 31. PMID: 39243525. Summary: *New mathematical framework for genetic relatedness analysis involving X chromosome aneuploidies, demonstrating expertise in creating complex computational and mathematical models to solve intricate kinship problems in medical genetics and forensic science.* ***Collab w/ Oslo University Hospital, Norway.***
10. Antão-Sousa S, Gusmão L, Modesti NM, Feliziani S, Faustino M, Marcucci V, Sarapura C, Ribeiro J, Carvalho E, Pereira V, Tomas C, de Pancorbo MM, Baeta M, Alghafri R, Almheiri R, Builes JJ, Gouveia N, Burgos G, Pontes ML, Ibarra A, da Silva CV, Parveen R, Benitez M, Amorim A, **Pinto N.*** Microsatellites' mutation modeling through the analysis of the Y-chromosomal transmission: Results of a GHEP-ISFG collaborative study. Forensic Sci Int Genet. 2024 Mar;69:102999. doi: 10.1016/j.fsigen.2023.102999. Epub 2023 Dec 14. PMID: 38181588. Summary: *Comprehensive collaborative study analyzing mutation rates across 33 Y-STR markers using a massive dataset of over 467,000 paternal transmissions, including 1,863 observed mutations. Advanced computational models, including logistic regression incorporating paternal age and allele length, were used to predict mutation probabilities, revealing that longer alleles and older paternal age increase mutation rates. It emphasizes the importance of large-scale data sharing and standardized reporting to improve mutation modeling in forensic genetics and to refine the evaluation of genetic evidence*. ***Collab w/ 20 worldwide laboratories from a GHEP-ISFG working group (coordinator).***
11. Felício D, Dias A, Martins S, Carvalho E, Lopes AM, **Pinto N**, Lemos C, Santos M, Alves-Ferreira M. Non-coding variants in VAMP2 and SNAP25 affect gene expression: potential implications in migraine susceptibility. J Headache Pain. 2023 Jun 29;24(1):78. doi: 10.1186/s10194-023-01615-z. PMID: 37380951; PMCID: PMC10308609. *Summary: Effect of non-coding variants in genes related to synaptic function (VAMP2 and SNAP25) on gene expression, with potential implications for migraine susceptibility. Integration of bioinformatic prediction, biostatistical validation, and experimental functional assays to dissect the regulatory impact.* ***Collab w/ J Sequeiro's i3S lab.***
12. Antão-Sousa S*, **Pinto N**, Rende P, Amorim A, Gusmão L. The sequence of the repetitive motif influences the frequency of multistep mutations in Short Tandem Repeats. Sci Rep. 2023;13(1):10251. Published 2023 Jun 24. doi:10.1038/s41598-023-32137-y. Summary: *Microsatellites' mutation modeling and how the sequence of repetitive motifs influences mutation dynamics, particularly important as 40 neurological, neurodegenerative, and neuromuscular*

disorders are determined by repeat expansions of STRs at coding and non-coding regions. ***Collab w/ State University of Rio de Janeiro, BR***

13. Felício D, Alves-Ferreira M, Santos M, Quintas M, Lopes AM, Lemos C, **Pinto N**, Martins S. Integrating functional scoring and regulatory data to predict the effect of non-coding SNPs in a complex neurological disease. Brief Funct Genomics. 2023 May 30:elad020. doi: 10.1093/bfgp/elad020. Epub ahead of print. PMID: 37254524. Summary: *Computational workflow designed to prioritize the functional relevance of non-coding SNPs in neurological disorders.* ***Collab w/ J Sequeiro's i3S lab.***
14. Garcia-Pelaez J, Barbosa-Matos R, Lobo S, Dias A, Garrido L, Castedo S, Sousa S, Pinheiro H, Sousa L, Monteiro R, Maqueda JJ, Fernandes S, Carneiro F, **Pinto N**, Lemos C, Pinto C, Teixeira MR, Aretz S, Bajalica-Lagercrantz S, Balmaña J, Blatnik A, Benusiglio PR, Blanluet M, Bours V, Brems H, Brunet J, Calistri D, Capellá G, Carrera S, Colas C, Dahan K, de Putter R, Desseignés C, Domínguez-Garrido E, Egas C, Evans DG, Feret D, Fewings E, Fitzgerald RC, Coulet F, Garcia-Barcina M, Genuardi M, Golmard L, Hackmann K, Hanson H, Holinski-Feder E, Hüneburg R, Krajc M, Lagerstedt-Robinson K, Lázaro C, Ligtenberg MJL, Martínez-Bouzas C, Merino S, Michils G, Novaković S, Patiño-García A, Ranzani GN, Schröck E, Silva I, Silveira C, Soto JL, Spier I, Steinke-Lange V, Tedaldi G, Tejada MI, Woodward ER, Tischkowitz M, Hoogerbrugge N, Oliveira C. Genotype-first approach to identify associations between CDH1 germline variants and cancer phenotypes: a multicentre study by the European Reference Network on Genetic Tumour Risk Syndromes. Lancet Oncol. 2023 Jan;24(1):91-106. doi: 10.1016/S1470-2045(22)00643-X. Epub 2022 Nov 24. Erratum in: Lancet Oncol. 2023 Jan;24(1):e10. PMID: 36436516; PMCID: PMC9810541. *Summary: Genotype-first approach to identify associations between germline variants and cancer phenotypes, showcasing experience with large-scale genetic data analysis and bioinformatics models to understand the relationship between genotype and phenotype.* ***Collab w/ C Oliveira's i3S lab.***
15. Antão-Sousa S*, Conde-Sousa E, Gusmão L, Amorim A, **Pinto N**. Estimations of Mutation Rates Depend on Population Allele Frequency Distribution: The Case of Autosomal Microsatellites. Genes (Basel). 2022 Jul 14;13(7):1248. doi: 10.3390/genes13071248. PMID: 35886031; PMCID: PMC9323320. Summary: *Analysis of the biases in estimating autosomal microsatellite mutation rates and providing of an important computational framework and open-source, user friendly, software toolkit.*
16. Costa C*, Figueiredo C, Amorim A, Costa S, Ferreira PM, **Pinto N**. Quantification of forensic genetic evidence: Comparison of results obtained by qualitative and quantitative software for real casework samples. Forensic Sci Int Genet. 2022 Apr 26;59:102715. doi: 10.1016/j.fsigen.2022.102715. Epub ahead of print. PMID: 35490558. Summary: *Advanced bioinformatics comparison of different probabilistic genotyping software (PGS) for analyzing and interpret complex DNA mixture samples, either considering or not DNA quantity.* ***Collab w/ LPC-PJ.***
17. Carvalho E, Dias A, Sousa A, Lopes AM, Martins S, **Pinto N**, Lemos C, Alves-Ferreira M*. A High Methylation Level of a Novel −284 bp CpG Island in the RAMP1 Gene Promoter Is Potentially Associated with Migraine in Women. Brain Sciences. 2022; 12(5):526. doi:

10.3390/brainsci12050526. Summary: *Identification of a novel CpG site at −284 bp in the RAMP1 gene promoter showing significantly higher DNA methylation in female migraineurs, suggesting a potential epigenetic biomarker for migraine susceptibility. Methylation levels were quantitatively analyzed and validated through statistical tests.* ***Collab w/ J Sequeiro's i3S lab.***

18. Maturana-Candelas A*, Gómez C, Poza J, Rodríguez-González V, Pablo VG, Lopes AM, **Pinto N**, Hornero R. Influence of PICALM and CLU risk variants on beta EEG activity in Alzheimer's disease patients. Sci Rep. 2021 Oct 14;11(1):20465. doi: 10.1038/s41598-021-99589-y. PMID: 34650147; PMCID: PMC8516883. Summary: *The paper uses sLORETA source localization and non-parametric statistics to analyze EEG relative power and spatial entropy in Alzheimer's patients with PICALM and CLU risk alleles. Significant disruptions in beta-band EEG activity were found, linking these genetic variants to neural dysfunction associated with Alzheimer's disease.* ***Collab w/ Biomedical Engineering Group from Valladolid University (GiB – UVA)***.
19. Neto L†, **Pinto N**†, Proença A, Amorim A*, Conde-Sousa E. 4SpecID: Reference DNA Libraries Auditing and Annotation System for Forensic Applications. Genes (Basel). 2021 Jan 2;12(1):61. doi: 10.3390/genes12010061. PMID: 33401773; PMCID: PMC7824288. Summary: *It introduces 4SpecID, a computationally efficient software tool designed for auditing and annotating large DNA reference non-human libraries. Employing graph-based algorithms and species-cluster congruence metrics, it grades data quality based on validation from multiple independent sources and genetic clustering consistency. The tool features a user-friendly interface and optimized C++ implementation with parallel computing, enabling rapid data processing and error detection across extensive public databases like BOLD. It emphasizes the need for forensic-standard validation in DNA reference datasets, enhancing reliability in species identification workflows.* ***Collab w/ Dep. Informatics, School of Engineering, University of Minho.***
20. Macedo A, Gómez C, Rebelo MÂ, Pozad J, Gomes I, Martins S, Maturana-Candelas A, Gutiérrez-de Pablo V, Durães L, Sousa P, Figueruelo M, Rodriguez M, Pita C, Arenas M, Alvarez L, Hornero R, Lopes AM, **Pinto N***. Risk Variants in Three Alzheimer's Disease Genes Show Association with EEG Endophenotypes. J Alzheimers Dis. 2021;80(1):209-223. doi: 10.3233/JAD-200963. PMID: 33522999; PMCID: PMC8075394. *Summary: It uses genome-wide genotyping arrays combined with resting-state EEG to analyze 796 variants across sixteen AD-related genes in an Iberian cohort of AD patients, mild cognitive impairment (MCI) subjects, and controls. It employs robust statistical methods including principal component analysis for population stratification, stringent quality control for genotype data, and Kruskal-Wallis tests for genotype-EEG power associations. EEG preprocessing integrates independent component analysis and spectral power calculations across conventional frequency bands. The study identifies novel significant correlations between variants in IL1RAP, UNC5C, and NAV2 genes and slowing of brain oscillations, particularly in delta and theta bands, highlighting these genes' roles in neuroinflammation, neuronal apoptosis, and neurodevelopment pathways. The work emphasizes the value of leveraging EEG-derived quantitative endophenotypes alongside genetics to dissect AD pathogenesis, using large-scale data integration and advanced bioinformatic pipelines.*

21. González RD†, Gomes I†, Gomes C, Rocha R, Durães L, Sousa P, Figueruelo M, Rodríguez M, Pita C, Hornero R, Gómez C, Lopes AM, **Pinto N***, Martins S. APOE Variants in an Iberian Alzheimer Cohort Detected through an Optimized Sanger Sequencing Protocol. Genes (Basel). 2020 Dec 22;12(1):4. doi: 10.3390/genes12010004. PMID: 33375167; PMCID: PMC7822120. Summary: *It optimizes a Sanger sequencing protocol to accurately genotype the GC-rich APOE gene region, focusing on SNPs rs429358 and rs7412 that define the APOE alleles associated with Alzheimer's disease risk. Computational analysis involved bioinformatics alignment and variant calling, as well as statistical testing for Hardy–Weinberg equilibrium and population differentiation. Odds ratios for Alzheimer's disease risk linked to APOE variants were calculated to assess regional and sex-specific frequency variations, and refine APOE genotype-phenotype associations.*
22. Rebelo MÂ, Gómez C, Gomes I, Poza J, Martins S, Maturana-Candelas A, Ruiz-Gómez SJ, Durães L, Sousa P, Figueruelo M, Rodríguez M, Pita C, Arenas M, Álvarez L, Hornero R, **Pinto N***, Lopes AM. Genome-Wide Scan for Five Brain Oscillatory Phenotypes Identifies a New QTL Associated with Theta EEG Band. Brain Sci. 2020 Nov 18;10(11):870. doi: 10.3390/brainsci10110870. PMID: 33218114; PMCID: PMC7698967. Summary: *It applies genome-wide association analysis combined with advanced bioinformatics tools to identify genetic loci linked to brain oscillatory phenotypes measured by EEG. Using multivariable linear regression, and controlling for population stratification and disease status, significant SNP associations were detected, including a novel genome-wide significant variant in CLEC16A. Functional annotation integrated eQTL and chromatin interaction data from multiple brain tissues, enabling gene prioritization and pathway enrichment analysis. Protein interaction networks and gene ontology further elucidated the biological pathways underlying brain electrophysiology, demonstrating the power of computational models to link genetics and complex neurophysiological traits.*
23. Gutiérrez-de Pablo V, Gómez C*, Poza J, Maturana-Candelas A, Martins S, Gomes I, Lopes AM, **Pinto N**, Hornero R. Relationship between the Presence of the ApoE ε4 Allele and EEG Complexity along the Alzheimer's Disease Continuum. Sensors (Basel). 2020 Jul 10;20(14):3849. doi: 10.3390/s20143849. PMID: 32664228; PMCID: PMC7411888. Summary: *This study applied advanced signal processing and statistical models to analyze resting-state EEG complexity, measured by Lempel-Ziv complexity (LZC), across the Alzheimer's disease (AD) continuum in subjects stratified by ApoE ε4 genotype. EEG preprocessing involved artifact removal via independent component analysis and regional complexity estimation to identify spatial patterns linked to genetic risk. Statistical comparisons using non-parametric tests with false discovery rate correction revealed significant differences in EEG complexity related to ApoE ε4 status before clinical AD onset. This integrative computational approach highlights potential early biomarkers of AD by linking genotype-driven brain signal complexity alterations.* ***Collab w/ Biomedical Engineering Group from Valladolid University (GiB – UVA)***.
24. Gomes I, **Pinto N**, Antão-Sousa S, Gomes V, Gusmão L, Amorim A*. Twenty Years Later: A Comprehensive Review of the X Chromosome Use in Forensic Genetics. Front Genet. 2020 Sep 17;11:926. doi: 10.3389/fgene.2020.00926. PMID: 33093840; PMCID: PMC7527635. Summary: *It reviews the use of X chromosome markers in forensic genetics, highlighting the importance of*

advanced computational and bioinformatics models for accurate kinship analysis. It emphasizes the challenges of accounting for linkage disequilibrium, haplotype frequency estimation, and recombination rates in statistical models, and also underscores the necessity of large, population-specific databases and standardized nomenclature for robust bioinformatics analysis and interpretation.

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26. Ruiz-Gómez SJ*, Hornero R, Poza J, Maturana-Candelas A, **Pinto N**, Gómez C. Computational modeling of the effects of EEG volume conduction on functional connectivity metrics. Application to Alzheimer's disease continuum. J Neural Eng. 2019 Oct 29;16(6):066019. doi: 10.1088/1741-2552/ab4024. PMID: 31470433.
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1.1.2. Submitted – Under Review

1. Cerqueira A*, Quelhas-Santos J, Sampaio S, Oliveira M, **Pinto N**, Alencastre I, Pestana M, Circulating CCL18 Is Associated With Cardiovascular Disease in Non-Dialysis Chronic Kidney Disease Patients. Cellular and Molecular Life Sciences; 1st reviews completed.
2. Costa C*, Conde-Sousa E, Figueiredo C, Costa S, Ferreira PM, Amorim A, Prieto L, **Pinto N.** Weighing the Impact of Population-level Coancestry in Forensic Genetics' Problems Involving Mixtures. Scientific Reports; 1st reviews requested

1.1.3. In Final Preparation – To Be Submitted During September 2025

1. Faustino M*, Gusmão L, Kling D, **Pinto N**. Advancing Genetic Relatedness Testing: An Informatics Solution for X Chromosome Aneuploidies and Linked Markers.
2. Faustino M, Gusmão L, Zarrabeitia M, Castañeda-Fernandez M, Pinheiro MF, Porto MJ, Brito P, Abovich M, Berardi G, Toscanini U, Cicarelli R, Kawamura B, Nothnagel M, **Pinto N*.** Comprehensive Modelling of Recombination and Statistical Methodologies for X-STR Kinship Evaluations.

3. Rodrigues P*, **Pinto N**, Prata M, Flores-Espinoza R, Burgos G, Nakanishi H, Perini J, Basta P, Børsting C, Gusmão L, Pereira V. MHappaMundi: A new microhaplotype panel for ancestry inference.
4. Costa C*, Trindade B, Minervino A, Carnevali E, Onofri M, Angeletti S, Rena V, Gutierrez Mendez R, Castillo Amezquita M, Camps Bel L, Cortes Sierra S, Cordoba S, Guinudinik A, Dorigón Lezana M, Palencia L, García Fernández O, Alvarez Merino J, Vinueza-Espinosa D, Biagini S, Ginart S, Sala A, Caputo M, Nicolotti M, Wirz L, Mosquera A, Puente M, Miozzo C, Ramella M, Prieto L, **Pinto N**. Statistical Interpretation of Cases Involving Mixtures: A Spanish and Portuguese-Speaking Working Group (GHEP-ISFG) Collaborative Exercise.

1.2. Books

1. Amorim A, **Pinto N**, "An Introduction to Forensic Genetics for Non-Geneticists", ISBN: 9781003836599, Edition: 12-2023, Editor: CRC PRESS. doi: 10.1201/9781003266716.

1.3. Book Chapters

1. Amorim A, Faustino M, **Pinto N**, Prata MJ, Gomes I (2025). Applications of Haplodiploid Markers in Forensic Genetics. In: Dash, H.R., Elkins, K.M., Al-Snan, N.R. (eds) Advances in Forensic Biology and Genetics. Springer, Singapore. https://doi.org/10.1007/978-981-96-4585-5_16
2. Amorim A, **Pinto N**, Haplodiploid Markers and Their Forensic Relevance, 2022, In: Dash H.R., Shrivastava P., Lorente J.A. (eds) Handbook of DNA Profiling. Springer, Singapore. Doi: 10.1007/978-981-16-4318-7_7
3. Pita C, Figueruelo M, Rodríguez M, Maturana-Candelas A, Hornero R, Poza J, Ruiz-Gómez S J, García M, Duraes L, Rocha R, Álvarez L, Martins S, Lopes A M, Gomes I, Arenas M, Sousa P, Gómez C, **Pinto N**, Improvements in the early diagnosis of Alzheimer's disease through the correlation between genome and brain activity, Envelhecimento como prespetiva futura, 2019, Edition Aranzadi – Thomson Reuters, ISBN: 978-84-1309-827-2
4. Oliveira M, Arenas M, **Pinto N**, Amorim A, Genética Forense No Humana, Genética Forense: Del laboratorio a los Tribunales, 2019, Ediciones Díaz de Santos, ISBN: 978-84-9052-213-4, Depósito Legal: M-37475-2018
5. **Pinto N**, X Chromosome, Forensic Genetics: Biodiversity and heredity in civil and criminal investigation, October 2016, Imperial College Press, ISBN: 9781783268344, ISBN-10: 1783268344. Doi: 10.1142/q0023
6. **Pinto N**, Amorim A, Identity-by-descent, Brenner's Encyclopedia of Genetics 2nd edition, April 2013, Elsevier, Electronic ISBN: 9780080961569, Print ISBN: 9780123749840. doi.org/10.1016/B978-0-12-374984-0.00764-6. Scopus indexed.

1.4. Conference International Proceedings (Scopus and WoS indexed, ≥ 2 pages)

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2. Antão-Sousa S, Conde-Sousa E, Gusmão L, Amorim A, **Pinto N***. How often have X- and autosomal-STRs mutations equivocal parental origin been assigned? Forensic Science International: Genetics Supplement Series, 2022, 8, pp. 99–101. doi.org/10.1016/j.fsigss.2022.09.035
3. Faustino M*, Turchi C, Kling D, Gusmão L, Amorim A, **Pinto N.** Paternal half-sisters or unrelated – How can the X-chromosomal evidence be quantified when one female presents trisomy X? Forensic Science International: Genetics Supplement Series, 2022, 8, pp. 138–139. doi.org/10.1016/j.fsigss.2022.10.011
4. Costa C*, Figueiredo C, Amorim A, ..., Ferreira PM, **Pinto N**. Statistical analysis tools of mixture DNA samples: When the same software provides different results. Forensic Science International: Genetics Supplement Series, 2022, 8, pp. 38–39. doi.org/10.1016/j.fsigss.2022.09.014
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8. Conde-Sousa E, **Pinto N**, Amorim A*. Reference DNA databases for forensic species identification: Auditing algorithms. Forensic science international Forensic Science International: Genetics Supplement Series. 2019; 7(1): e564-e566. doi.org/10.1016/j.fsigss.2019.10.091.
9. Garcia MG, Catanesi CI, Penacino GA, **Pinto N*.** Weighing substructure in Argentina considering different population clusters. Forensic science international Forensic Science International: Genetics Supplement Series. 2019; 7(1): e773-e775. doi.org/10.1016/j.fsigss.2019.10.172.
10. Maturana-Candelas A, Gómez C*, Poza J, Ruiz-Gomez S, Núñez P, Rodríguez M, Figueruelo M, Pita C, **Pinto N**, Martins S, Lopes A M, Gomes I, Hornero R. Analysis of Spontaneous EEG Activity in Alzheimer's Disease Patients by Means of Multiscale Spectral Entropy. In: Masia L., Micera S., Akay M., Pons J. (eds) Converging Clinical and Engineering Research on Neurorehabilitation III. ICNR 2018. Biosystems & Biorobotics, vol 21. Springer, Cham. https://doi.org/10.1007/978-3-030-01845-0_116. Scopus-indexed as book chapter.
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14. García MG*, Gusmão L, Catanesi CI, Penacino GA, **Pinto N.** Mutation rate of 12 X-STRs from Investigator Argus X-12 kit in argentine population. Forensic Science International: Genetics Supplement Series. 2017; 6(1): e562-e564. doi.org/10.1016/j.fsigss.2017.09.219.
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17. Pereira V*, Tomas Mas C, **Pinto N**, Amorim A, Gusmão L, Prata MJ, Morling N. Assessing the potential application of X-chromosomal haploblocks in population genetics and forensic studies. Forensic Science International: Genetics Supplement Series. 2013; 4(1): e9-e10. doi.org/10.1016/j.fsigss.2013.10.004.
18. Gomes C, Magalhães M, Amorim A, Alves C, **Pinto N**, Gusmão L*. How useful is your X in discerning pedigrees? Forensic Science International: Genetics Supplement Series. 2011; 3(1): e161-e162. doi.org/10.1016/j.fsigss.2011.08.081.
19. Magalhães M*, **Pinto N**, Gomes C, Pereira R, Amorim A, Alves C, Gusmão L. When the alleged father is a close relative of the real father: the utility of insertion/deletion polymorphisms. Forensic Science International: Genetics Supplement Series. 2011; 3(1): e9-e10. doi.org/10.1016/j.fsigss.2011.08.004.
20. **Pinto N***, Gusmão L, Silva P V, Amorim A. Estimating coancestry from genotypes using a linear regression method. Forensic Science International: Genetics Supplement Series. 2011; 3(1): e373-e374. doi.org/10.1016/j.fsigss.2011.09.048.
21. **Pinto N***, Gusmão L, Amorim A. Distinguishing kinship from genealogical likelihoods. Forensic Science International: Genetics Supplement Series. 2009;2(1):453-4. doi.org/10.1016/j.fsigss.2009.08.018.

1.5. Conference Abstracts (WoS indexed)

1. Carvalho E, Dias A, Sousa A, Lopes AM, Martins S, **Pinto N**, Lemos C, Alves-Ferreira M. RAMP1 gene promoter and female migraine susceptibility: new clues in epigenetic processes. 55th European-Society-of-Human-Genetics (ESHG) Conference, JUN 11-14, 2022, Vienna, Austria. Volume 31, Page 318-318, Supplement 1, Meeting Abstract EP21.002. WOS:001050507001110

2. Felício D, Martins S, Santos M, Lopes AM, Lemos C, **Pinto N**, Alves-Ferreira M. Candidate regulatory variants in SNARE complex genes and their involvement in migraine susceptibility. 55th European-Society-of-Human-Genetics (ESHG) Conference, JUN 11-14, 2022, Vienna, Austria. Volume 31, Page 196-196, Supplement 1, Meeting Abstract EP10.032. WOS: 001050507000495
3. Felício D, Martins S, Santos M, Lopes AM, Lemos C, **Pinto N**, Alves-Ferreira M. Integrating Functional Genomic Data to Prioritize Candidate Non-Coding Variants in Migraine Susceptibility. Medicine 102(13). 26th Annual Meeting of the Portuguese-Society-of-Human-Genetics (SPGH). NOV 17-19, 2022. Coimbra, Portugal. WOS:001005724900037
4. Carvalho E, Dias A, Guerrero AL, Gómez C, Sousa A, Lopes AM, Martins S, **Pinto N**, Lemos C, Alves-Ferreira M, Ramp1 Promoter Methylation Status in Portuguese and Spanish Women with Migraine. Medicine 102(13). 26th Annual Meeting of the Portuguese-Society-of-Human-Genetics (SPGH). NOV 17-19, 2022. Coimbra, Portugal. WOS: 001005724900048
5. Carvalho E, Dias A, Sousa A, Lopes AM, Martins S, **Pinto N**, Lemos C, Alves-Ferreira M, Role of Epigenetics in Migraine: Methylation Levels in The Cgrp's Receptors, Cephalalgia 42 (1_SUPPL), 35-35. 19th Biennial Migraine Trust International Symposium (MTIS), SEP 08-11, 2022, London, England. WOS:000851579900039
6. Carvalho E, Dias A, Sousa A, Sequeiros J, Lopes A, Martins S, **Pinto N**, Lemos C, Alves-Ferreira M. Epigenetic Analysis of the CGRP Pathway Genes Involved in Migraine. Medicine 101(30). 2022. WOS:000832873200121

1.6. Other Publications

1. Prata M J., Gusmão L, Gomes I, Pinto N, Martins S, Gomes V, Carracedo A. (2025). António Amorim (in memoriam). Forensic Science International: Genetics, https://doi.org/10.1016/j.fsigen.2025.103254