
BIOGRAPHICAL SKETCH

Provide the following information for the Senior/key personnel and other significant contributors.
Follow this format for each person. **DO NOT EXCEED FIVE PAGES.**

NAME Lemos, Carolina	POSITION TITLE Associated Professor at ICBAS, University of Porto
PhD	

Carolina Lemos graduated in Biology in 2002 by the Faculty of Sciences of University of Porto. She finished her PhD in 2009, at UNIGENe, IBMC and Institute of Biomedical Sciences Abel Salazar (ICBAS). Her PhD project focused on the genetic and epidemiologic study of migraine, a complex disease. Her main research interest is in the genetic epidemiology of complex and mendelian genetic diseases, focusing nowadays in Primary Headaches and Familial Amyloidotic Polyneuropathy (FAP) research. Carolina Lemos published 60 articles in peer-reviewed journals. She is the author of 33 oral communications by invitation in national and international meetings, presenting the main results of her research group activities and author/co-author of 56 oral communications and 101 posters in national and international scientific meetings.

During the last 5 years, she published 5 papers in peer-reviewed journals as last author, 20 papers as co-author. The publications reflect the commitment with her scientific lines of research, where she devoted to translational research, from bench to analysis of clinical data. She devoted herself to show the evidence of a genetic component of migraine in the Portuguese population and the importance of the anticipation of age-at-onset in Familial Amyloid Polyneuropathy. She and her team received 19 prize (s) and / or honors in national and international meetings, recognizing the value of the research that is made in this field. She had the opportunity to co-supervise/supervise during the last 15 years: 1 Post-Doc, 11 PhD students, 8 MSc students and 24 Medicine MSc Students. She is an Assistant Professor at ICBAS, teaching Genetics and Statistics to Medical; Aquatic sciences; Biochemistry and Public Health students. She is Treasurer of the Board of Headache Portuguese Society and ANDO-Associação Nacional de Displasias Ósseas. Participates actively in the dissemination of science in schools as Science Ambassador.

Contributions to Science

39. Dias A, Santos M, Carvalho E, Felício D, Silva P, Alves I, Pinho T, Sousa A, Alves-Ferreira M, LEMOS C. Functional characterization of a novel PRRT2 variant found in a Portuguese patient with hemiplegic migraine. *Clinical Genetics* 2023, 1-7. doi:10.1111/cge.14379. (JCR® IF: 4.296, Quartile: Q1)
38. 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, et al. 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. (JCR® IF: 51·1, Quartile: Q1).
37. Marcelino, V.; Paço, M.; Dias, A.; Almeida, V.; Rocha, J.C.; Azevedo, R.; Alves-Ferreira, M.; LEMOS, C.; Pinho, T. The Role of Pain Inflexibility and Acceptance among Headache and Temporomandibular Disorders Patients. *Int. J. Environ. Res. Public Health* 2022, 19(13), 7974. 10.3390/ijerph19137974
36. 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. 10.3390/brainsci12050526
35. Dias A, Mariz T, Sousa A, LEMOS C, Alves-Ferreira M. A review of migraine genetics: gathering genomic and transcriptomic factors. *Human Genetics*, (), 1-14 10.1007/s00439-021-02389-7

34. Alves-Ferreira M, Quintas M, Neto JL, Sequeiros J, Sousa A, Pereira-Monteiro J, Alonso I, Lemos C. A genetic interaction of *Neurexin* with *GABAA-receptor, synaptotagmin* and *CASK* in migraine patients: a case-control study. *J Headache Pain* 2021 doi: 10.1186/s10194-021-01266-y
33. M. Alves-Ferreira, A. Azevedo, T. Coelho, D. Santos, J. Sequeiros, I. Alonso, A. Sousa, C. LEMOS. Beyond Val30Met transthyretin (TTR): variants associated with age-at-onset in hereditary ATTRv amyloidosis. *Amyloid* 2020 doi: 10.1080/13506129.2020.1857236
32. Quintas M, Neto JL, Sequeiros J, Sousa A, Pereira-Monteiro J, LEMOS C, Alonso I. Going Deep into Synaptic Vesicle Machinery Genes and Migraine Susceptibility - A Case-Control Association Study. *Headache*. 2020 Sep 26. doi: 10.1111/head.13957. Online ahead of print.
31. Ferreira LT, Orr B, Rajendraprasad G, Pereira AJ, LEMOS C, Lima JT, Guasch Boldú C, Ferreira JG, Barisic M, Maiato H. α -Tubulin detyrosination impairs mitotic error correction by suppressing MCAK centromeric activity. *J Cell Biol* (2020) 219 (4): e201910064. (<https://doi.org/10.1083/jcb.201910064>)
30. Dias A, Santos D, Coelho T, Alves-Ferreira M, Sequeiros J, Alonso I, Sousa A, LEMOS C. C1QA and C1QC modify age-at-onset in familial amyloid polyneuropathy patients. *Ann Clin Transl Neurol*. 2019 Mar 7;6(4):748-754.
29. Santos D, Coelho T, Alves-Ferreira M, Sequeiros J, Mendonça D, Alonso I, Sousa A, LEMOS C. Large normal alleles of ATXN2 decrease age-at-onset in TTR-FAP Val30Met patients. *Ann Neurol*. 2019 Feb;85(2):251-258.
28. Madhivanan K, Greiner ER, Alves-Ferreira M, Soriano-Castell D, Rouzbeh N, Aguirre C, Paulsson JF, Chapman J, Jiang X, Ooi FK, LEMOS C, Dillin A, Prahlad V, Kelly JW, Encalada SE. Cellular clearance of circulating transthyretin decreases cell-nonautonomous proteotoxicity in *Caenorhabditis elegans*. *Proc Natl Acad Sci U S A*. Aug 14;115(33):E7710-E7719.
27. Santos D., Santos MJ., Alves-Ferreira M., Coelho T., Sequeiros J., Alonso I., Oliveira P., Sousa A., LEMOS C., Grazina M. mtDNA copy number associated with age of onset in familial amyloid.polyneuropathy. *J Neurol Neurosurg Psychiatry*. 2018;89(3):300-304
26. Alves-Ferreira M., Coelho T., Santos D., Sequeiros J., Alonso I., Sousa A., LEMOS C. (2017). A trans-acting factor may modify age-at-onset in familial amyloid polyneuropathy ATTRV30M in Portugal. *Mol Neurobiol* (DOI 10.1007/s12035-017-0593-4).
25. Santos, D., Coelho, T., Alves-Ferreira, M., Sequeiros, J., Mendonça, D., Alonso, I., Lemos, C. and Sousa, A. (2016), Familial amyloid polyneuropathy in Portugal: New genes modulating age-at-onset. *Ann Clin Transl Neurol* 2016. doi:10.1002/acn3.380
24. Santos D., Coelho T., Alves-Ferreira M., Sequeiros J., Mendonça D., Alonso I., LEMOS C., Sousa A. Variants in *RBP4* and *AR* genes modulate age-at-onset in Familial Amyloid Polyneuropathy (FAP ATTRV30M). *EJHG* 2016 May; ;24(5):756-60
23. Beirão JM, Malheiro J, LEMOS C, Beirão I, Costa P, Torres P. Ophthalmological manifestations in hereditary transthyretin (ATTR V30M) carriers: a review of 513 cases. *Amyloid*. 2015 Jun;22 (2):117-22

22. Beirão JM, Malheiro J, LEMOS C, Matos E, Beirão I, Pinho-Costa P, Torres P. Impact of liver transplantation on the natural history of oculopathy in Portuguese patients with transthyretin (V30M) amyloidosis. *Amyloid*. 2015 Mar;22(1):31-5
21. Alves-Ferreira M, Pinho T, Sousa A, Sequeiros J, LEMOS C, Alonso I. Identification of Genetic Risk Factors for Maxillary Lateral Incisor Agenesis. *J Dent Res*. 2014 May;93(5):452-8
20. Barros J, Ferreira A, Brandão AF, LEMOS C, Correia F, Damásio J, Tuna A, Sequeiros J, Coutinho P, Alonso I, Pereira-Monteiro J. Familial hemiplegic migraine due to L263V SCN1A mutation: Discordance for epilepsy between two kindreds from Douro Valley. *Cephalgia*. 2014 Oct;34(12):1015-20
19. LEMOS C, Coelho T, Alves-Ferreira M, Martins-da-Silva A, Sequeiros J, Mendonça D, Sousa A. Overcoming artefact: anticipation in 284 Portuguese kindreds with familial amyloid polyneuropathy FAP ATTRV30M. *Neurol Neurosurg Psychiatry*. 2014 Mar;85(3):326-30
12. LEMOS C, Alonso I, Barros J, Sequeiros J, Pereira-Monteiro J, Mendonça D, Sousa A. Assessing risk factors for migraine: differences in gender transmission. *PLoS One* 2012, 7:e50626.
11. LEMOS C, Neto JL, Pereira-Monteiro J, Mendonça D, Barros J, Sequeiros J, Alonso I, Sousa A. A role for endothelin receptor type A in migraine without aura susceptibility? A study in Portuguese patients. *Eur J Neurol* 2011, 18: 649–655
10. LEMOS C, Mendonça D, Pereira-Monteiro J, Barros J, Sequeiros J, Alonso I, Sousa A. BDNF and CGRP interaction: implications in migraine susceptibility. *Cephalgia* 2010; 30(11):1375-82
9. Pinho T, Maciel P, LEMOS C, Sousa A. Familial aggregation maxillary lateral incisors agenesis. *J Dent Res* 2010;89(6):621-5.
8. LEMOS C, Pereira-Monteiro J, Mendonça D, Ramos EM, Barros J, Sequeiros J, Alonso I, Sousa A. Evidence of Syntaxin 1A Involvement in Migraine Susceptibility: A Portuguese Study. *Arch Neurol*.2010;67(4):422-427.
7. M Panque, C LEMOS, A Sousa, L Velázquez, J Sequeiros, M Fleming. The role of the disease in the psychological impact of pre-symptomatic testing for SCA2 and FAP ATTRV30M: knowledge of the disease in the family, degree of kinship and gender of the transmitting parent. *J Genet Couns* 2009;18(5):483-93
6. Ferro A, Castro MJ, LEMOS C, Santos M, Sousa A, Pereira-Monteiro J, Sequeiros J, Maciel P. The C677T polymorphism in MTHFR is not associated with migraine in Portugal. *Dis Markers*.2008;25(2):107-13.
5. MJ Castro, AH Stam, C LEMOS, B de Vries, KRJ Vanmolkot, J Barros, GM Terwindt, RR Frants, J Sequeiros, MD Ferrari, JM Pereira-Monteiro, AMJM van den Maagdenberg. First mutation in the voltage-gated NaV1.1 subunit gene SCN1A with co-occurring familial hemiplegic migraine and epilepsy. *Cephalgia*. 2009;29(3):308-13.
4. LEMOS C, Castro MJ, Barros J, Sequeiros J, Pereira-Monteiro J, Mendonça D, Sousa A. Familial Clustering of Migraine: Further Evidence From a Portuguese Study. *Headache*. 2009; 49(3): 404-411.
3. Castro MJ, Nunes B, de Vries B, LEMOS C, Vanmolkot KR, van den Heuvel JJ, Temudo T, Barros J, Sequeiros J, Frants RR, Koenderink JB, Pereira-Monteiro JM, van den Maagdenberg AM. Two novel functional mutations in the Na⁺,K⁺-ATPase alpha2-subunit ATP1A2 gene in patients with familial hemiplegic migraine and associated neurological phenotypes. *Clin Genet*. 2008;73(1):37-43.

2.Castro MJ, Stam AH, LEMOS C, Barros J, Gouveia RG, Martins IP, Koenderink JB, Vanmolkot KR, Mendes AP, Frants RR, Ferrari MD, Sequeiros J, Pereira-Monteiro JM, van den Maagdenberg AM.Recurrent ATP1A2 mutations in Portuguese families with familial hemiplegic migraine. *J Hum Genet.*2007;52(12):990-8,

1.Paneque M, LEMOS C, Escalona K, Prieto L, Reynaldo R, Velazquez M, Quevedo J, Santos N, Almaguer LE, Velazquez L, Sousa A, Fleming M, Sequeiros J. Psychological Follow-up of Presymptomatic Genetic Testing for Spinocerebellar Ataxia Type 2 (SCA2) in Cuba. *J Genet Couns.* 2007;16(4):469-79.