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OVERVIEW

I am a mother of 4 sons; in parallel, I have pursued full-time University scientific and teaching careers.

I am a Full Professor of Molecular Biology at the University of Lisboa (Dept. Chemistry & Biochemistry) and Director of BioISI- Biosystems & Integrative Sciences Institute and Director of FCT-funded PhD programme BioSys.

Research in my lab focusses on the molecular and cellular mechanisms of the genetic disease Cystic Fibrosis (CF) and on translating this knowledge into the benefit of patients. To understand CF mechanisms globally we use transcriptomics, proteomics and functional genomics (functional siRNA screens). Our results translate into the clinic for better CF diagnosis, prognosis and personalized therapies.

I have authored 162 peer-reviewed papers and 4 book chapters. I edited 2 books and 2 special journal issues. My publications have been cited 5,080 (Scopus) and 7,448 (Google Scholar) times. H-index: 39 (Scopus); 48 (Google Scholar). Scopus ID: [7006683774](#) | ORCID ID: [0000-0002-0828-8630](#). I am a Highly Cited Researcher since 2020 (Mendeley, University of Stanford).

I am a member of EMBO-European Molecular Biology Organization and of the Portuguese Academy of Sciences. I received multiple awards, among which the Pfizer Award for Basic Biomedical Research and the Annual Award of European Cystic Fibrosis Society.

I have directly supervised/co-supervised 20 successfully completed PhD students (+8 ongoing); 16 post-doctoral fellows (+6 ongoing); and more than 25 junior students (BSc + MSc).

I participated in 41 grants (+6 ongoing), 33 of which as Principal Investigator, amounting to ~6M€. Eight of these were large multi-centre EU grants, of which I coordinated one (TargetScreen2) and Co-coordinated of another (EuroCareCF).

Other activities include: Coordinator of the ECFS (European Cystic Fibrosis Society) Basic Science Working Group of, Vice-Coordinator of (proposed) ECFS Organoid Working Group the and editor of Scientific Reports (Nature group). Formerly, I was member of ECFS Board, Scientific Advisory Board (SAB) CF Trust (UK), Mukoviszidose e.V (German CF Foundation) and Associate Editor of Journal Cystic Fibrosis (Elsevier).

I have registered 3 patents and I regularly develop business consulting activities with industry (BioMarin; Vertex; Facilitate; Gilead; LEK; Reuters; Novartis, Proteostasis, TranslateBio, etc).

I gave 226 invited international talks (+63 national), including the opening plenary of North-American CF Conference (2007) and 4 opening plenaries at ECFS Conferences (2004, 2008, 2013, 2016), 1 Gordon Conference and 6 EMBO Courses/workshops. I organized for 10 years the ECFS-Basic Science Conferences and 8 international training workshops for young researchers in the CF field. In total, I organized 26 international conferences, being President of 2013 ECFS-European CF Society Conference.

ACADEMIC/SCIENTIFIC BACKGROUND

- Jul 2006 "Habilitation" ("Agregação") to the title of "**Professor Agregado**", Area of **Biochemistry**, Univ Lisboa, Portugal. Unanimously Approved. Course proposal: "*Human Molecular Biology from a Systems Biology Perspective*".
- 1993 PhD in **Biochemistry/Molecular Genetics**, Univ Lisboa, Portugal & Gulbenkian Institute of Science (Oeiras, Portugal). Supervisor: Prof Claudina Rodrigues-Pousada. Final mark: *summa cum laude*. Thesis: "*Stress Proteins: Induction and Regulation in Tetrahymena pyriformis*".
- 1986 MSc (equiv) in **Biochemistry**, Univ Lisboa, Portugal & Gulbenkian Institute of Science (Oeiras, Portugal). Supervisor: Prof Claudina Rodrigues-Pousada. Final mark: *summa cum laude*. Thesis: "*The Response of the Protozoan Tetrahymena pyriformis to a Stress Agent: Sodium Meta-Arsenite*".
- 1982 BSc in **Chemistry/Biochemistry**, Univ Lisboa Portugal. Work carried out as undergraduate research student at the Faculty of Medical Sciences, New University of Lisboa, Portugal. Final mark: 17/20. Thesis: "*Lipid Content of Liver and Blood Plasma of Hepatomized Rats*".

POSITIONS/TRAINING

- Jul 2013 **Full Professor of Biochemistry/ Molecular Biology**. Dept of Chemistry & Biochemistry, Faculty of Sciences, University of Lisboa.
- 2015- **Director of BioISI- Biosystems & Integrative Sciences Institute** (evaluated in 2015: 24/25)
- 2014- **Member of the Portuguese Academy of Sciences** (Section of Sciences).
- Jan/Jun 16- **Visiting fellow at EMBL-European Molecular Biology Laboratory** (Heidelberg, Germany) at the Pepperkok group.
- 2006/2013 **Assistant Professor with "Habilitation"** (Biochemistry/ Molecular Biology), Dept of Chemistry & Biochemistry, Faculty of Sciences, University of Lisboa.
- 1993/2006 **Assistant Professor**, Faculty Sciences, Univ Lisboa, Portugal (tenure: 1998).
Invited Researcher at the Centre Human Genetics, National Institute of Health
- 1986/1993 **Teaching Assistant**, Dept. Chemistry & Biochemistry, Faculty of Sciences, University of Lisboa.
PhD student at IGC - Gulbenkian Institute of Science, Oeiras, Portugal.
- 1983/1986 **Trainee Teaching Assistant**, Dept. Chem & Biochemistry, Faculty of Sciences, University of Lisboa.
Graduate student at IGC - Gulbenkian Institute of Science, Oeiras, Portugal.

OTHER POSITIONS

- Jan/Jun 2016 **Visiting fellow at EMBL-European Molecular Biology Laboratory** (Heidelberg, Germany) at the Pepperkok group.
- Nov 2012/15 **Visiting Researcher at Faculty of Medical Sciences of UniCamp**-University of Campinas (SP, Brazil) - the, CAPES programme "Science without Borders".
- 2012/2015 **Director of the Research Centre BioFiG**-Centre for Biodiversity, Functional and Integrative Genomics.
- 2010/2011 **Vice-President** of Department of Chemistry and Biochemistry, Faculty of Sciences, University of Lisboa.
- Aug 08/Jul 10 **Visiting fellow at EMBL-European Molecular Biology Laboratory** (Heidelberg, Germany) at the Pepperkok group to coordinate EU project TargetScreen2.

- 2007** **Founding member of Research Centre BioFiG-** Centre for Biodiversity, Functional and Integrative Genomics, University of Lisboa (Portugal) and head of Membrane Protein Disorders Unit.
- 2006/2011** **Coordinator of the Cystic Fibrosis Research Unit**, Centre of Human Genetics of the National Institute of Health Ricardo Jorge, Lisboa (Portugal)

PRIZES AND HONOURS

Prizes

- 2019** John Riordan & Paul Quinton Award for CF Research, given by Bob Emmelkamp Association of parents and friends of people with Cystic Fibrosis (USA).
- 2019** Thomé Villar / Boehringer Ingelheim 2019 Award, from Portuguese Pneumology Society.
- 2016** University of Lisboa Award for the 2nd most productive scientist in the scientific area of Biology, Bioengineering, Biochemistry & Biotechnology.
- 2013** Pfizer Award for Basic Biomedical Research.
- 2010** Annual Award of European Cystic Fibrosis Society (jointly with DN Sheppard, Bristol, UK).

Honours

- 2017** Editor of Scientific Reports (Nature Group)
- 2014** Elected EMBO member
- 2014** Elected member of the Portuguese Academy of Sciences
- 2012/19** Member of the Board ECFS - European Society of Cystic Fibrosis
- 2011** Editor (with K Kunzelmann) of "*Cystic Fibrosis Protocols and Diagnosis*", Humana Press. Series: Methods in Molecular Biology Series; 2 Vols: 741 & 742
- 2011** Member of the Science Award Committee of Fundação Pulido Valente
- 2010-2011** Member of the Research Advisory Board of Mukoviszidose Institut-the Cystic Fibrosis Patients Association (Germany)
- 2008-2017** Associate Editor of Journal of Cystic Fibrosis (Elsevier)
- 2007-2010** Member of the Award Committee of EMBO/FEBS Women in Science
- 2006-2009** Member of Research Advisory Board of the Cystic Fibrosis Trust (UK)
- 2004** Chief-editor of a special supplement of *Journal of Cystic Fibrosis*.

RESEARCH INTERESTS

My lab studies human disease mechanisms associated with membrane proteins, namely those related to the genetic disease Cystic Fibrosis (CF). Thus, the major focus is on the molecular and cellular mechanisms of biogenesis, traffic and degradation of normal and mutant protein CFTR (CF transmembrane Conductance Regulator), which when mutated causes CF. Other studies include the epithelial Na⁺ channel ENaC and anoctamins to identify novel genes and small-molecules that regulate these membrane proteins. Most of our research aims to understand the molecular mechanisms that prevent mutant CFTR with F508del (the most frequent mutation found in CF patients) from reaching the cell surface. By understanding these mechanisms that retain F508del-CFTR at the endoplasmic reticulum (ER), we aim to design therapeutic strategies to restore its normal activity as a Cl⁻ channel at the cell surface, with substantial benefit for CF patients. We thus carry out drug development programmes to identify novel CFTR modulators.

To understand CF pathophysiology mechanisms in a global way, we use systems approaches based on transcriptomics, proteomics and functional genomics (functional siRNA screens). In collaboration with nanoelectronics centres, we developed novel chips for CF diagnosis.

Our research is also focussed on the characterization of other CFTR gene mutations, namely those: 1) affecting processing of mRNA (splicing and nonsense-mediated decay); 2) with intracellular trafficking defects.

We confirm all basic cellular mechanisms in **native tissues** (collected from CF patients) with the final goal of translating knowledge and results from the basic science into the clinical practice, for better CF diagnosis, prognosis and personalized therapies.

PUBLICATIONS

Articles in International Peer-Reviewed Journals [*corresponding author; impact factor publication year]

1. Simões, FB, Kmit A, Amaral MD* (2021) Cross-Talk of Inflammatory Mediators and Airway Epithelium Reveals CFTR as a Major Target. *ERJ Open Res.* Accepted for publication. DOI: [10.1183/23120541.00247-2021](https://doi.org/10.1183/23120541.00247-2021)
2. Pereira C, Mazein A, Farinha CM, Gray MA, Kunzelman K, Ostaszewski M, Balaur I, Amaral MD, Falcao AO (2021) CyFi-MAP - an interactive Pathway- based Resource for Cystic Fibrosis. *Sci Rep* **11**: 22223. [PMID: [34782688](https://pubmed.ncbi.nlm.nih.gov/34782688/)] IF **4.4**. DOI: <https://www.nature.com/articles/s41598-021-01618-3>
3. Amaral MD* (2021) How to Determine the Mechanism of Action of CFTR Modulator Compounds: A Gateway to Theranostics. *Eur J Med Chem* **210**: 112989. [PMID: [33190956](https://pubmed.ncbi.nlm.nih.gov/33190956/)] IF **6.2**. DOI: [10.1016/j.ejmech.2020.112989](https://doi.org/10.1016/j.ejmech.2020.112989).
4. Silva IAL, Railean V, Duarte A, Amaral MD* (2021) Personalized Medicine Based on Nasal Epithelial Cells: Comparative Studies with Rectal Biopsies and Intestinal Organoids. *J Pers Med* **11**: 421. PMID: [34065744](https://pubmed.ncbi.nlm.nih.gov/34065744/). IF **4.45**. DOI: [10.3390/jpm11050421](https://doi.org/10.3390/jpm11050421). *Journal cover*
5. Pinto M, Silva IAL, Figueira M; Amaral MD, Lopes-Pacheco M (2021) Pharmacological Modulation of Ion Channels for the Treatment of Cystic Fibrosis. *J Exp Pharmacol* **13**: 693-723. [PMID: [34326672](https://pubmed.ncbi.nlm.nih.gov/34326672/)] IF **2.24**. DOI: [10.2147/JEP.S255377](https://doi.org/10.2147/JEP.S255377).
6. Roda J*, Teixeira T, Silva IAL, Reis-Silva T, Ferreira R, Amaral MD, Oliveira G (2021) Paediatric Population with Cystic Fibrosis in the Centre of Portugal: candidates for new therapies. *J Ped.* Epub 9 July. [PMID: [34252371](https://pubmed.ncbi.nlm.nih.gov/34252371/)] IF **2.03**. DOI: [10.1016/j.jped.2021.05.010](https://doi.org/10.1016/j.jped.2021.05.010)
7. Bene Z, Fejes Z, Szánthó TG, Fenyvesi F, Váradi J, Clarke LA, Panyi G, Macek M Jr, Amaral MD, Balogh I, Nagy B Jr* (2021) Enhanced expression of human epididymis protein 4 (HE4) reflecting pro-inflammatory status is regulated by CFTR in cystic fibrosis bronchial epithelial cells. *Front Pharmacol* **12**: 592184. IF **4.2**. DOI: [10.3389/fphar.2021.592184](https://doi.org/10.3389/fphar.2021.592184).
8. Silva IAL, Duarte A, Marson FAL, Centeio R, Doušová T, Kunzelmann K, Amaral MD (2020) Assessment of Distinct Electrophysiological Parameters in Rectal Biopsies for the Choice of the Best Diagnosis/Prognosis Biomarkers for Cystic Fibrosis. *Front Physiol* **11**: 604580. [PMID: [33424627](https://pubmed.ncbi.nlm.nih.gov/33424627/)] IF **4.134**. DOI: [10.3389/fphys.2020.604580](https://doi.org/10.3389/fphys.2020.604580).
9. Hagemijer MC, Annelotte M. Vonk AM, Awatade NT, Silva IAL, Tischer C, Hilsenstein V, Beekman JM, Amaral MD, Botelho HM* (2020) An open-source high-content analysis workflow for CFTR function measurements using the forskolin-induced swelling assay. *Bioinformatics* **36**: 5686-5694 [PMID: [33367496](https://pubmed.ncbi.nlm.nih.gov/33367496/)] IF **5.61**. DOI: [10.1093/bioinformatics/btaa1073](https://doi.org/10.1093/bioinformatics/btaa1073).
10. Quaresma MC, Pankonien I, Clarke LA, Sousa LS, Silva IAL, Railean V, Doušová T, Fuxe J, Amaral MD* (2020) Mutant CFTR Drives TWIST1 Mediated Epithelial-Mesenchymal Transition. *Cell Death & Dis* **11**: 920. [PMID: [33106471](https://pubmed.ncbi.nlm.nih.gov/33106471/)] IF **6.30**. DOI: [10.1038/s41419-020-03119-z](https://doi.org/10.1038/s41419-020-03119-z).
11. Sousa L, Pankonien I, Simões FB, Chanson M, Amaral MD* (2020) Impact of KLF4 on cell proliferation and epithelial differentiation in the context of Cystic Fibrosis. *Int J Mol Sci* **21**: 6717. [PMID: [32937756](https://pubmed.ncbi.nlm.nih.gov/32937756/)] IF **4.56**. DOI: [10.3390/ijms21186717](https://doi.org/10.3390/ijms21186717).
12. Donegà S, Malgorzata R, Pianigiani G, Igreja S, Amaral MD, Pagani F* (2020) Rescue of common exon-skipping mutations in Cystic Fibrosis with modified U1 snRNAs. *Hum Mutat* **41**: 2143-2154. [PMID: [32935393](https://pubmed.ncbi.nlm.nih.gov/32935393/)]. IF **4.37**. DOI: [10.1002/humu.24116](https://doi.org/10.1002/humu.24116).

13. Silva-Filho LVR^{F*}, Maróstica PJC, Athanazio RA, Reis FJC, Damaceno N, Paes AT, Hira AY, Schlesinger D, Kok F, Amaral MD and The Brazilian Cystic Fibrosis Patient Registry Contributors Team (2020) Extensive CFTR Sequencing Through NGS in Brazilian Individuals with Cystic Fibrosis: Unravelling Regional Discrepancies in the Country. *J Cyst Fibros* **S1569-1993**: 30822-5. [PMID: [32819855](#)] IF **4.29**. DOI: [10.1016/j.jcf.2020.08.007](#).
14. Silva IAL, Doušová T, Ramalho S, Centeio R, Clarke LA, Railean V, Botelho HM, Holubová A, Valášková I, Yeh J-T, Hwang T-C, Farinha CM, Kunzelmann K, Amaral MD* (2020) Organoids as a Personalized Medicine Tool for Ultra-Rare Mutations in Cystic Fibrosis: the Case of S955P and 1717-2A>G. *BBA - Mol Basis Dis* **1866**: 165905. [PMID: [32730979](#)]. IF **4.33**. DOI: [10.1016/j.bbadis.2020.165905](#).
15. Lopes-Pacheco M, Silva IAL, Turner MJ, Carlile GW, Sondo E, Thomas DY, Pedemonte N, Hanrahan JW, Amaral MD* (2020) Characterization of the Mechanism of Action of RDR01752, a Novel Corrector of F508del-CFTR. *Biochem Pharmacol* **180**: 114133. [PMID: [32628927](#)] IF **4.83**. DOI: [10.1016/j.bcp.2020.114133](#).
16. Sousa L, Pankonien I, Clarke LA, Silva IAL, Kunzelmann K, Amaral MD* (2020) KLF4 Acts as a wt-CFTR Suppressor Through an AKT-Mediated Pathway. *Cells* **9**: 1607. [[32630830](#)] IF **4.37**. DOI: [10.3390/cells9071607](#).
17. Amaral MD* (2020) Letter to the editor. *J Cyst Fibros* **19**: 503. [PMID: [32600655](#)] IF **4.29**. DOI: [10.1016/j.jcf.2020.02.011](#).
18. Uliyakina I, Botelho HM, Da Paula AC, Afonso S, Lobo MJ, Felício V, Farinha CM, Amaral MD* (2020) Full Rescue of F508del-CFTR Processing and Function by CFTR Modulators Can Be Achieved by Removal of Two Unique Regulatory Regions. *Int J Mol Sci* **21**: 4524. [PMID: [32630527](#)] IF **4.56**. DOI: [10.3390/ijms21124524](#).
19. Santos JD, Pinto FR, Ferreira JF, Amaral MD, Zaccolo M, Farinha CM* (2020) Cytoskeleton regulators CAPZA2 and INF2 associate with CFTR to control its plasma membrane levels under EPAC1 activation. *Biochem J* **477**: 2561-2580. [PMID: [32573649](#)]. IF **4.33**. DOI: [10.1042/BCJ20200287](#).
20. Almeida C* & Amaral MD* (2020) A Central Role of the Endoplasmic Reticulum in the Cell Emerges from its Functional Contact Sites with Multiple Organelles. *Cell Mol Life Sci* **77**: 4729-4745. PMID: [32313974](#). IF **7.01**. DOI: [10.1007/s00018-020-03523-w](#).
21. De Boeck K, Lee T, Amaral MD, Drevinek P, Elborn JS, Fajac I, Kerem E, Davies JC* (2020) Cystic Fibrosis Drug Trial Design in the Era of CFTR Modulators Associated with Substantial Clinical Benefit: Stakeholders' Consensus View. *J Cyst Fibros* **S1569-1993**: 30162-4. [PMID: [32527602](#)]. IF **4.29**. DOI: [10.1016/j.jcf.2020.05.012](#).
22. Bene Z, Zsolt Fejesa Z, Macek M Jr, Amaral MD, Balogha I, Nagy B Jr* (2020) Laboratory Biomarkers for Lung Disease Severity and Progression in Cystic Fibrosis. *Clin Chim Acta* **508**: 277-286. [PMID: [32428503](#)]. IF **2.74**. DOI: [10.1016/j.cca.2020.05.015](#).
23. Pinto MC, Schreiber R, Lérias J, Ousingawat J, Amaral MD, Kunzelmann K* (2020) Regulation of TMEM16A by CK2 and its Role in Cellular Proliferation. *Cells* **9**: 1138. [PMID: [32380794](#)]. IF **4.37**. DOI: [10.3390/cells9051138](#).
24. Amaral MD*, Quaresma MC, Pankonien I (2020) What Role Does CFTR Play in Development, Differentiation, Regeneration and Cancer? *Int J Mol Sci* **21**: 3133. [PMID: [32365523](#)]. IF **4.56**. DOI: [10.3390/ijms21093133](#).
25. Vonk AM, van Mourik P, Ramalho AS, Silva IAL, Statia M, Kruisselbrink E, Suen SWF, Dekkers JF, Vleggaar FP, Houwen RHJ, Mullenders J, Boj SF, Vries R, Amaral MD, de Boeck K, van der Ent CK, Beekman JM* (2020) Protocol for application, standardization and validation of the forskolin induced swelling assay in Cystic Fibrosis human colon organoids. *STAR Protoc* **1**: 100019. [PMID: [33111074](#)] DOI: [10.1016/j.xpro.2020.100019](#).
26. Amaral MD, Hutt DM, Tomati V, Botelho HM, Pedemonte N* (2020) CFTR Processing, Trafficking and Interactions. *J Cyst Fibros* **9 S1**: S33-S36. [PMID: [31680043](#)]. IF **4.29**. DOI: [10.1016/j.jcf.2019.10.017](#).

27. Amaral MD*, Beekman JM (2020) Activating Alternative Chloride Channels to Treat CF: Friends or Foes? Report on the Meeting of the Basic Science Working Group in Dubrovnik, Croatia. *J Cyst Fibros* **19**: 11-15. [PMID: [31676346](#)]. IF **4.29**. DOI: [10.1016/j.jcf.2019.10.005](#).
28. Simões FB, Quaresma MC, Clarke LA, Silva IAL, Pankonien I, Railean V, Kmit A, Amaral MD* (2019) TMEM16A Chloride Channel Does Not Drive Mucus Production. *Life Sci Alliance*. **2** (6) pii: e201900462. [PMID: [31732694](#)]. DOI: [10.26508/lsa.201900462](#).
29. Amaral MD, De Boeck K* [On behalf of the ECFS Strategic Planning Task Force on 'Speeding up access to new drugs for CF'] (2019) Theranostics by testing CFTR modulators in patient-derived materials: The current status and a proposal for subjects with rare CFTR mutations. *J Cyst Fibros*. **18**: 685-69. [PMID: [31326274](#)] IF **4.29**. DOI: [10.1016/j.jcf.2019.06.010](#).
30. Matos AM, Pinto F, Barros P, Amaral MD, Pepperkok R, Matos M* (2019) Inhibition of calpain 1 restores plasma membrane stability to pharmacologically rescued Phe508del-CFTR variant. *J Biol Chem*. **294**: 13396-13410. [PMID: [31324722](#)] IF **4.106**. DOI: [10.1074/jbc.RA119.008738](#).
31. Davies JC*, Drevinek P, Elborn JS, Kerem E, Lee T; European CF Society (ECFS) Strategic Planning Task Force on 'Speeding up access to new 4 drugs for CF', Amaral MD, de Boeck K, Davies JC, Drevinek P, Elborn JS, Kerem E, Lee T (2019) Speeding up access to new drugs for CF: Considerations for clinical trial design and delivery. *J Cyst Fibros* **18**: 677-684. [PMID: [31303382](#)] IF **4.29**. DOI: [10.1016/j.jcf.2019.06.011](#).
32. Santos JD, Canato S, Carvalho AS, Botelho HM, Aloria K, Amaral MD, Matthiesen R, Falcao AO, Farinha CM* (2019) Folding status is determinant over traffic-competence in defining CFTR interactors in the endoplasmic reticulum. *Cells* **8**: E353 [PMID: [31014000](#)] IF **5.66**. DOI: [10.3390/cells8040353](#).
33. Kmit A*, Marson FAL, Pereira SV, Vinagre AM, Leite GS, Servidoni MF, Ribeiro JD, Ribeiro AF, Bertuzzo CS, Amaral MD (2019) Extent of rescue of F508del-CFTR function by VX-809 and VX-770 in human nasal epithelial cells correlates with SNP rs7512462 in SLC26A9 gene in F508del/F508del Cystic Fibrosis patients. *Biochim Biophys Acta Mol Basis Dis* **1865**: 1323-31. [PMID: [30716472](#)] IF **5.108**. DOI: [10.1016/j.bbadis.2019.01.029](#).
34. Clarke LA*, Awatade NT, Felicio VM, Silva IAL, Calucho M, Pereira L, Azevedo P, Cavaco J, Barreto C, Bertuzzo CM, Gartner S, Beekman J, Amaral MD (2019) The effect of premature termination codon mutations on CFTR mRNA abundance in human nasal epithelium and intestinal organoids: a basis for read-through therapies in Cystic Fibrosis. *Hum Mutat* **40**: 326-334 [PMID: [30488522](#)] IF **5.213**. DOI: [10.1002/humu.23692](#).
35. Patel W, Moore PJ, Sassano MF, Lopes-Pacheco M, Aleksandrov AA, Amaral MD, Tarran R, Gray MA* (2019) Increases in cytosolic Ca²⁺ induce dynamin- and calcineurin-dependent internalisation of CFTR. *Cell Mol Life Sci* **76**: 977-994. [PMID: [30547226](#)] IF **6.721**. DOI: [10.1007/s00018-018-2989-3](#)
36. Nagy B Jr*, Bene Z, Fejes Z, Heltshe SL, Reid D, Ronan NJ, McCarthy Y, Smith D, Nagy A, Joseloff E, Balla G, Kappelmayer J, Macek M Jr, Bell SC, Plant BJ, Amaral MD, Balogh I (2019) Human epididymis protein 4 (HE4) levels inversely correlate with lung function improvement (delta FEV1) in cystic fibrosis patients receiving ivacaftor treatment. *J Cyst Fibros* **18**: 271-277. [PMID: [30268371](#)] IF **4.727**. DOI: [10.1016/j.jcf.2018.08.013](#).
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Book chapters

1. Amaral MD (2011) In: Tratado de Fibrosis Quística. Salcedo A, Gartner S, Novo MDG, Girón RM, editores. Editorial Just in Time SL.
2. Kunzelmann K & Amaral MD (2008) "Novas Abordagens Terapêuticas Destinadas a corrigir o Defeito Básico na Fibrose Cística". In: *Fibrose Cística: Enfoque Multidisciplinar*. Neto NL, Coordenador. Secretaria de Estado de Santa Catarina, Florianópolis, SC, Brasil.
3. Freitas PP, Ferreira HA, Graham DL, Clarke LA, Amaral MD, Martins V, Fonseca L, Cabral JS (2004) Magnetoresistive DNA chips. In: *Magnetoelectronics*. Johnson M (Editor). Elsevier-Academic Press, Amsterdam, Netherlands, pp. 331-373.
4. Farinha CM, Amaral MD (2002) Processing and intracellular trafficking of wild-type and mutant CFTR. In: *Proc 25th European Cystic Fibrosis Conference Genova, Italy* (June 20-23). Romano L, Manno G, Galletta LJV, Eds, Monduzzi Editores, Bologna, Italy, pp.1-6. ISBN: 88-323-2622-1.

Other Publications

1. Amaral MD (2010) Cystic Fibrosis – Translating Basic Science Knowledge into Therapies. *Eur Resp Disease* **6**, 66-9.
2. Amaral MD, Clarke LA, Roxo-Rosa M, Sousa L (2006) Genomics and proteomics approaches to study the genetic disease cystic fibrosis. *Revstat* **27**, 47-54.
3. Amaral MD (2004) Editorial. *J Cyst Fibros* **3(S2)**, 3.
4. Edelman A, Amaral MD (2004) General introduction to section C: biochemistry and biophysics of CFTR. *J Cyst Fibros* **3(S2)**, 67.
5. Freitas PP, Freitas H, Graham D, Clarke L, Amaral M, Martins V, Fonseca L, Cabral JS (2003) Magnetoresistive Biochips. *Europhysics News* **34**, 224-226.

In addition, of ~300 abstracts in peer-reviewed international conferences, about half were published in international ISI-indexed journals.

PATENTS

1. Amaral MD, Almaça J, Faria D, Kunzelmann K, Schreiber R, Conrad C, Pepperkok R (2013) Drug Targets for Cystic Fibrosis And Other Conditions. Filed by University of Lisboa (Portugal) to the International Bureau of the World Intellectual Property Organization. PCT/IB2013/058851
2. Matos P, Amaral MD, Moniz S, Moraes B, Mendes AI, Jordan P (2011) Rac1 signalling stimulation rescues F508del-CFTR plasma membrane expression and function in human airway cells: a novel therapeutic approach for cystic fibrosis. Patent US 14/070,533.
3. Amaral MD, Dahimène S, Mendes F, Luz S (2011) Two novel human epithelial cell lines to be used in assays for traffic studies/ screens of CFTR protein (wild-type and with the F508del mutation). Pat Pending PT105697.

GRANTS

Ongoing

- 2020/21** CFF Cystic Fibrosis Foundation, USA (Ref. AMARAL19G0) "*PTSense: – Novel Compounds as Potential Drugs for CFTR PTC Mutations*". Budget: 151K\$; 1 yr. PI: MD Amaral.

- 2020/21** CFF Cystic Fibrosis Foundation, USA (Ref. FARINH19I0) "*DysMut2 – Characterization of Dysfunctional Mechanisms in Class II Mutations*". Budget: 108K\$; 1 yr. PI: CM Farinha; co-PI: MD Amaral.
- 2019** Vertex Pharmaceuticals (Donation grant). *Identification of Portuguese patients with Cystic Fibrosis by Complete CFTR Gene Mutation Genotyping and Rectal Biopsy Analyses*. Budget: 52K€; 1 yr. PI: MD Amaral
- 2019/21** Gilead Sciences (Research Scholars Program in Cystic Fibrosis) *Identification of novel F508del-CFTR traffic correctors among FDA-approved drugs*. Budget: 130K\$; 2 yrs. PI: M Lopes-Pacheco (Mentored by MD Amaral).
- 2018/21** FCT (PTDC/MED-QUI/28800/2017) "*iDrugCF - Identification of New Drugs for Cystic Fibrosis*". Budget: 240K€; 3 yrs. PI: MD Amaral.
- 2018/22** CF Trust Strategic Research Centre Award (Ref. SRC 013) "*Personalised Therapies for all: Restoring airway function in CF using Alternative Chloride Channels*". Total budget: 750K€. FCUL Budget: 244K€; 4 yrs. PI: M Gray, Newcastle (UK). PI for the FCUL group: MD Amaral.
- 2018/22** European Union (H2020-SC1-2017-755021). *HIT-CF – Personalised Treatment For Cystic Fibrosis Patients With Ultra-rare CFTR Mutations (and beyond)*. Total budget: 6.7M€ / FCID: 257K€; 5 yrs. Coordinator: Kors van der Ent, University Medical Centre Utrecht, Utrecht (Netherlands). Coordination FCUL Group: MD Amaral.

Previous

- 2016** FCT/POCTI (PTDC/BIM-MEC/2131/2014) "*DIFFTARGET-Novel Factors of CFTR Traffic Related to Epithelial Cell Differentiation: Potential Therapeutic Targets for Cystic Fibrosis*". Budget: 200K€; 3 yrs. PI: MD Amaral.
- 2016/19** FCT/POCTI (PTDC/QEQ-SUP/4283/2014) "*FARMTRANSANION-Anion transmembrane transport promoted by drug-like molecules: building a library of anion carriers inspired in Ataluren (PTC124)*". Budget: 200K€; 3 yrs. PI: V Félix.
- 2016/19** FCT/POCTI (PTDC/EEI-ESS/4923/2014) "*MIMED - Mining the Molecular Metric Space for Drug Design*". Budget: 127K€; 3 yrs. PI: A Falcão
- 2016/18** CFF Cystic Fibrosis Foundation, USA (Ref. AMARAL15XX1) "*RNA LIFE – Novel RNA Regulators as Potential Drug Targets for Cystic Fibrosis*". Budget: 324K\$; 2 yrs. PI: MD Amaral.
- 2015/19** ERARE15-pp-010/JTC 2015 "*INSTINCT - Induced Pluripotent Stem Cells for Identification of Novel Drug Combinations Targeting Cystic Fibrosis Lung and Liver Disease*". Total budget: 1.24 M€; 124K€ (FFCUL); 3 yrs. Principal Investigator (U Martin, Univ. Hannover, Germany). FCUL PI: MD Amaral.
- 2016/18** CFF Cystic Fibrosis Foundation, USA (Ref. AMARAL16I0) "*Characterization of Orphan CFTR mutations*". Budget: 108K\$; 2 yrs. PI: MD Amaral.
- 2016** Gilead GÉNESE Programme (Ref PGG/008/2015) "*Predicting Clinical Drug Efficacy of CFTR Protein Modulators Using Intestinal Organoids and Nasal Cells from Patients with Cystic Fibrosis*". 30K€; 1 yr. Principal Investigator: MD Amaral.
- 2016** Vertex Pharmaceuticals (Donation grant). *Complete CFTR gene mutation analysis in Portuguese patients with Cystic Fibrosis*. Budget: 20K€; 1 yr. PI: MD Amaral
- 2015/17** CFF Cystic Fibrosis Foundation, USA (Ref. AMARAL15XX0) "*CFTR mRNA Stability Studies for PTC Mutations*". Budget: 222K\$; 2 yrs. PI: MD Amaral.
- 2014/19** CF Trust Strategic Research Centre Award (Ref. SRC 003) "*INOVCF- Innovative non-CFTR Approaches for Cystic Fibrosis Therapies*". Total budget: 750K€. FCUL Budget: 178.4K€; 4 yrs. PI: M Gray, Newcastle (UK). PI for the FCUL group: MD Amaral.
- 2012/15** FCT/POCTI (PTDC/SAU-GMG/122299/2010) "*Characterization of ER-Quality Control for the F508del-CFTR Protein: Potential Therapeutic Targets for Cystic Fibrosis*". 170K€; 3 yrs. Principal Investigator: MD Amaral.

- 2010/14** COST (EU) BM1003. "*Microbial Cell Surface Determinants of Virulence as Targets for New Therapeutics in CF*". Coordinator: A Molinaro, Università di Napoli, Italy. 350K€; 4 yrs.
- 2014** Gilead GÉNESE Programme (Ref MED-2013-250) "*Diagnosis, Prognosis and Personalized Treatment of Cystic Fibrosis*". 20K€, 1 yr. PI: MD Amaral.
- 2013** Gilead GÉNESE Programme (Ref MED-2012-022) "*Incidence of CFTR mutations in Individuals with Chronic Respiratory Diseases and their Effect in Disease Expression*". 20K€, 1 yr. Principal Investigators: AS Ramalho/ MD Amaral.
- 2012/15** Science Without Borders Programme - 'Ciência Sem Fronteiras' (Ref CAPES- 053/2012) "*Diagnosis, Prognosis and Treatment of Cystic Fibrosis*". Special Visiting Researcher at UniCamp, University of Campinas (SP, Brazil) 20K + 1 post-doc fellowship + 1 PhD student PI: MD Amaral / Co-PI: Carmen Bertuzzo (UniCamp, Campinas, SP Brazil).
- 2011/13** CFF-Cystic Fibrosis Foundation, USA (Ref: 7207534) "*Identification of Novel Targets Rescuing of F508del-CFTR Traffic: Mechanism of Action*". 228US K\$, 2 yrs. Principal Investigator: MD Amaral.
- 2009/12** FCT (PIC/IC/83103/2007) "*Diagnosis, Prognosis and Treatment of Cystic Fibrosis*". 170K€; 3 yrs. PI: MD Amaral.
- 2007/11** European Union (FP6-2005-LH-7-037365). *TargetScreen2 - Novel post-genomics cell-based screens for drug targeting in membrane protein disorders*. Budget: 3.7 M€; 4 yrs. Scientific Coordinator: MD Amaral.
- 2006/10** European Union (FP6-2004-LSH-018932). *EuroCareCF – European Coordination Action for Research in Cystic Fibrosis*. 89.7K€; 3 yrs. Coordinator: David Sheppard, University of Bristol, Bristol (UK). Vice-Coordinator and PI for Coordination of Basic Research: MD Amaral.
- 2005/08** European Union (FP6-2004-IST-NMP-2-016833). *SNiP2CHIP – Development of a complete integrated SNP analysis system*. 195.4K€; 3 yrs. Coordinator: Paul Galvin, Tyndall National Institute, Cork (Ireland). PI for the FCUL group: MD Amaral.
- 2005/08** European Union (FP6-2003-LSH-512044). *NEUPROCF – Development of New Technologies for Low Abundance Proteomics: Application to Cystic Fibrosis*. 15K€; 3 yrs. Coordinator: Aleksander Edelman, Faculté de Médecine Necker – Enfants Malades, Paris (France). PI for the FCUL group: MD Amaral.
- 2005/08** BBSRC - Biotechnology and Biological Sciences Research Council, UK (BB/C517517/1). *Use of Human-Murine CFTR Chimeras to Investigate the Coupling of Permeation and Gating in the CFTR Chloride Channel*. 10K€; 3 yrs. Principal Investigator: David Sheppard, University of Bristol, Bristol (UK). PI for the FCUL group: MD Amaral.
- 2005/08** FCT /POCTI (SAU/MMO/58425/2004). *CFTR Interactome*. 95K€; 3 yrs. Principal Investigator: MD Amaral.
- 2005/08** FCT /POCTI (BIA-BCM/56609/2004). *Caenorhabditis elegans as a model to study folding of CFTR protein*. 95K€; 3 yrs. Principal Investigator: MD Amaral.
- 2005/07** FCT. *Aquisition of a small physiology unit*. Project approved by the *National Programme for Scientific Re-equipament*. 132K€. Principal Investigator: MD Amaral.
- 2003/06** FCT /POCTI (MGI/47382/2002) *Folding, Processing and Function of Normal and Mutant Cystic Fibrosis Transmembranar Conductance Regulator: Structural Implications*. 87K€; 3 yrs. Principal Investigator: MD Amaral.
- 2002/05** European Union (FP5-QLRT-2000-0182). *CF-Chip – Novel Genechip Technology for Early Detection of Cystic Fibrosis*. Budget: 147.6K€; 3 yrs. Coordinator: Paul Galvin, National Microelectronics Research Centre (NMRC), "Lee Maltings", University College, Cork (Ireland). PI for the FCUL group: MD Amaral.
- 2000/04** European Union (FP5-QLK-1999-00241, Concerted Action). *CF Network – Thematic Network around Cystic Fibrosis and Related Diseases*. 236.4K€; 4 yrs. Coordinator: Jean-Jacques Cassiman, University of Leuven (Belgium). PI for FCUL: MD Amaral.

- 2001/04** FCT/POCTI (MGI/35737/1999). *Biogenesis & Function of CFTR Protein with Different Mutations: Molecular Basis for Clinical and Therapeutic of Cystic Fibrosis?* 85K€; 3 yrs. PI: MD Amaral; Co-PI: D Penque (INSA).
- 2002/05** POCTI (MGI/40878/2001). *In search of New Molecular Targets for the Development of Novel Therapeutic Strategies for Cystic Fibrosis*. 60K€; 2 yrs. PI: D Penque (INSA); Co-PI: MD Amaral.
- 1997/00** FCT/ PraxisXXI (PSAU/P/SAU/55/96). *Cystic Fibrosis - Traffic and Cellular Function of CFTR*. 55K€; 3 yrs. PI: MD Amaral.
- 1995/97** JNICT (PBIC/C/BIA/2060/95). *Expression Studies of CFTR Gene*. 37.4K€; 3 yrs. PI: D Penque (INSA). Team member.
- 1995/97** JNICT/ French Embassy (049 C0). *Immortalization of Epithelial Cells in Portuguese Cystic Fibrosis Patients: Models to Study CFTR Gene Expression*. 1K€; 1yr. PI: MD Amaral.
- 1993/95** JNICT (PBIC/C/SAU/1587/92). *Molecular Biology of Cystic Fibrosis in the Portuguese Population: Epidemiology, Anthropogenetics and Physiopathology*. 75K€; 3 yrs. PI: J. Lavinha (INSA). Team member.

VARIOUS

Supervision: I have directly supervised **18 successfully completed PhD students** (+6 ongoing); **16 post-doctoral fellows** (+7 ongoing); and more than **25 junior students** (BSc + MSc). I also mentor(ed) **4 FCT researchers** and **1 Assistant Professor**.

Grant Reviewer: ANR – Agence Nationale de la Recherche, France; BSF – USA-Israel Binational Science Foundation; CCFF – Canadian CF Foundation; CFF – Cystic Fibrosis Foundation USA; Cystic Fibrosis Trust (UK); DFG – Deutsche Forschungsgemeinschaft (Germany); FWO (Belgium); Italian Cystic Fibrosis Foundation; NSF – National Science Foundation (USA); SFI – Science Foundation Ireland; Canadian CF Foundation; VLM – Vaincre la Mucoviscidose (French CF Foundation); Wellcome Trust (UK).

Reviewer: Science Transl Med; Nature Struct & Mol Biol; J Molecular Biology; BBA; Journal of Medical Genetics; Gene Therapy; Molecular and Cell Biology; Journal of Molecular Biology; Proteomics; Human Molecular Genetics, Thorax; Alberts 3rded - Essentials of Molecular and Cellular Biology (textbook, Garland).

Conferences organized: I organized **26 international conferences** and a current member of the Scientific Advisory Committee of the ABC – *Special FEBS Meetings on ABC Proteins*. I was President of the 36th Annual Conference of the European Cystic Fibrosis Society, 12-15 June 2013, Lisboa, Portugal; Chairman (2004-2009) of the ECFS Basic Science Conferences- *New Frontiers in Basic Science of Cystic Fibrosis*; Scientific Committee member of various European Cystic Fibrosis Society Conferences (Vice-Chairman in 2007); Co-Organizer of 2nd International Congress on Stress Responses in Biology and Medicine (2004); Chairman (2000-2003) of the European CF Network Consensus Meetings Towards Validation of CFTR Gene Expression & Functional Assays.

Invited talks: I gave **125 invited international talks** (+35 national), including one opening plenary of North-American Cystic Fibrosis Conference (2007) and 3 opening plenaries at European CF Conferences (2004, 2008, 2013).