

## *Curriculum vitae*

**Full name:** Luísa Maria Sousa Mesquita Pereira  
**Date of birth:** 4th June 1974  
**Nationality:** Portuguese  
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### **Academic degrees:**

1995: B. A. Biology, Faculty of Sciences, University of Porto  
1997: M. Sc. Human Applied Genetics, Faculty of Sciences, University of Porto  
2002: D. Phil. Biology (Human population genetics), Faculty of Sciences, University of Porto

### **Present position:**

Jul 2004-present: Researcher and Group Leader, Ipatimup (Institute of Molecular Pathology and Immunology of the University of Porto)  
Feb 2006-present: Affiliated Professor, Faculty of Medicine, University of Porto

### **Past appointments:**

2002-2004: Post-doc, Ipatimup and Department of Statistics, University of Oxford (grant from FCT - The Portuguese Foundation for Science and Technology - SFRH/BPD/7121/2001)

### **Specialisation Domains:**

- Human Population Genetics
- Uniparental genetic markers (mitochondrial DNA and Y-chromosome)
- Genome-wide SNPs and association studies

### **Current Investigation**

- Study of uniparental genetic markers in human populations and investigation of the phenomena of migration, expansion, selection and geographic structure.
- Disentangling between mtDNA neutral and pathological variants in the context of complex diseases.
- Screening of genome-wide diversity at a population level and evaluation of ancestral susceptibility and resistance to complex diseases, namely cancer and dengue fever.

### **Research projects**

- 1- 01/2012-12/2016: FP7-HEALTH-2011-single-stage (DENFREE) – “Dengue research Framework for Resisting Epidemics in Europe”. Project Number: 282378 (Co-Applicant)
- 2- 11/2011-10/2015: 7<sup>th</sup> Framework Programme (EUROTAST) – “A Marie Curie Initial Training Network on the History, Archaeology and New Genetics of the Transatlantic Slave Trade”. (Co-Applicant and Member of the Steering Committee)
- 3- 06/2013-09/2015: FCT sponsored project (PTDC/IVC-ANT/4917/2012) - "Genetic history and population structure of Southeast Asia". (Team Member).
- 4- 03/2011-02/2014: FCT sponsored project (PTDC/CS-ANT/113832/2009) – “Genetic and chronological characterisation of the European settlement by modern humans in the Upper Palaeolithic”. (Principal investigator)

- 5- 06/2007-05/2010: FCT sponsored project (PTDC/ANT/66275/2006) - "Refining the geographical origin and dispersal routes of early modern humans and early farmers of the Greater Mediterranean with high-resolution genetic techniques". (Principal investigator)
- 6- 06/2007-05/2010: FCT sponsored project (PTDC/AFR/71422/2006) - "Longevity in humans of African origins - a genetic approach". (Principal investigator)
- 7- 04-2006/03-2007: Project funded by The British Academy - "Y-chromosome phylogeography in Southeast Asia and the Pacific" (Co-Investigator-1)
- 8- 05/2003-04/2006: FCT sponsored project (POCTI/ANT/45139/2002) – "Reassessment of European mitochondrial DNA diversity: present and past distributions of female lineages" (Principal investigator)

**Publications:** Co-author of **89** peer-reviewed papers in international journals; **2815** citations; H index=**29**. Co-author of **26** full papers on congress proceedings. Co-author of **1** book and **4** book chapters. **25** selected and **30** invited oral communications. **75** poster communications.

**12 Selected papers of interest to this project:**

- 1- Cavadas B, Soares P, Camacho R, Brandão A, Costa M, Fernandes V, Pereira J, Rito T, Samuels DC, **Pereira L** (2015) Fine time-scaling of purifying selection on human non-synonymous mtDNA mutations based on worldwide population tree and mother-child pairs. *Hum. Mutat.* **36**:1100-1111.
- 2- **Pereira L**, Soares P, Triska P, Rito T, Waerden Avd, Li B, Radivojac P, Samuels DC (2014) Global human frequencies of predicted nuclear pathogenic variants and the role played by protein hydrophobicity in pathogenicity potential. *Sci. Rep.* **4**:7155.
- 3- Soares P, Abrantes D, Rito T, Thomson N, Radivojac P, Li B, Macaulay V, Samuels DC, **Pereira L** (2013) Evaluating purifying selection in the mitochondrial DNA of various mammalian species. *PLoS One* **8**: e58993.
- 4- **Pereira L**, Soares P, Máximo V, Samuels DC (2012) Somatic mitochondrial DNA mutations in cancer escape purifying selection and high pathogenicity mutations lead to the oncogenic phenotype: pathogenicity analysis of reported somatic mtDNA mutations in tumors. *BMC Cancer.* **12**: 53.
- 5- Fernandes V, Alshamali F, Alves M, Costa MD, Pereira JB, Silva NM, Cherni L, Harich N, Cerny V, Soares P, Richards MB, **Pereira L** (2012) The Arabian cradle: Mitochondrial relicts of the first steps along the southern route out of Africa. *Am. J. Hum. Genet.* **90**: 347-355.
- 6- **Pereira L**, Soares P, Radivojac P, Li B, Samuels DC (2011) Comparing phylogeny and the predicted pathogenicity of protein variations reveals equal purifying selection across the global human mtDNA diversity. *Am. J. Hum. Genet.* **88**: 433-439.
- 7- Fernandes F, **Pereira L**, Freitas AT (2009) CSA: An efficient algorithm to improve circular DNA multiple alignment. *BMC Bioinformatics* **10**: 230.
- 8- **Pereira L**, Freitas F, Fernandes V, Pereira JB, Costa MD, Costa S, Máximo V, Macaulay V, Rocha R, Samuels DC (2009) The diversity present in 5,140 human mitochondrial genomes. *Am. J. Hum. Genet.* **84**: 628-640.
- 9- Costa MD, Fernandes V, Freitas F, Cherni L, El Gaaied ABA, **Pereira L** (2009) Data from complete mtDNA sequencing of Tunisian centenarians: testing haplogroup association and the "golden mean" to longevity. *Mech Ageing Dev.* **130**: 222-226.
- 10- **Pereira L**, Gonçalves J, Franco-Duarte R, Silva J, Rocha T, Arnold C, Richards M, Macaulay V (2007) No evidence for an mtDNA role in sperm motility: data from complete sequencing of asthenozoospermic males. *Mol Biol Evol.* **24**: 868-874.
- 11- Goios A, Meirinhos J, Rocha R, Lopes R, Amorim A, **Pereira L** (2006) RepeatAround: a software tool for finding and visualizing repeats in circular genomes and its application to a human mtDNA database. *Mitochondrion* **6**: 218-224.
- 12- **Pereira L**, Richards M, Goios A, Alonso A, Albarrán C, Garcia O, Behar DM, Gölge M, Hatina J, Al-Gazali L, Bradley DG, Macaulay V, Amorim A (2005) High-resolution mtDNA evidence for the late-glacial resettlement of Europe from an Iberian refugium. *Genome Res.* **15**:19-24.