

Dr Júlia Baptista

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EDUCATION

Jan-05/Mar-08 **PhD in Human Genetics, University of Southampton, UK**
Thesis: "Detailed molecular studies of chromosome rearrangements in man" with Dr John Crolla, Prof Pat Jacobs and Prof Nic Cross

Sep-97/Oct-02 **Degree in Biology, University of Lisbon, Portugal**
5-year degree in biology with specialisation in Microbial Biology and Genetics.

AWARDS

May 2013/May 2016 **Young Investigators Award, Italian Ministry for Health**
Award of €267k. This project aimed to identify and characterise novel genes for limb defects using exome sequencing and functional assays in the zebrafish animal model.

Jan 2005/Mar 2008 **School of Medicine, University of Southampton**
Award of £1350 towards my part-time PhD tuition fees whilst an employee of the University of Southampton.

PROFESSIONAL EXPERIENCE

Sep-16/present:
University of Exeter Medical School, UK
Honorary Lecturer (Genomic Medicine)

I am involved in the delivery of the MSc degree in Genomic Medicine and am the Clinical Lead of the "Omics" technologies module. My role requires designing and delivering lectures and tutoring duties.

Nov-15/present:
Department of Molecular Genetics, Royal Devon and Exeter NHS Foundation Trust. UK
Clinical Scientist

I am involved in the delivery of genetic service, for patients with rare diseases. I am currently the Lead Scientist of the Exeter Exome Service.

May-13/Nov-15:

Medical Genetics Unit, Bologna University Hospital S.Orsola-Malpighi. Italy

Research Fellow

P.I. to the project: "Unravelling the genetic basis of limb defects through a combined genomic and functional approach".

- Sep-14/Nov-15:

University of Exeter Medical School (UEMS), UK

Honorary Research Fellow

I set up a collaboration with the UEMS and with Prof Charles Tyler at the University of Exeter to expand my zebrafish research skills.

- May-13/Aug-14:

Department of Biology and Biochemistry, University of Bath, UK

Visiting Research Fellow

I set up a collaboration with Prof R Kelsh in order to learn the zebrafish techniques and the developmental biology concepts needed to undertake my research.

Oct-10/May-13:

Career break for maternity reasons

Oct-09/ Sep-10:

Romeo Lab, Medical Genetics Unit, University of Bologna, Italy

Postdoctoral Research Fellow

Research project "Improving diagnoses of mental retardation in children in Central Eastern Europe and Central Asia through genetic characterisation and bioinformatics/-statistics" funded by the EU 7th Framework.

Apr-08/Sep-09

Career break for maternity reasons

Sep-03/Mar-08

Division of Human Genetics, University of Southampton- Based at the Wessex Regional Genetics Laboratory, Salisbury District Hospital, UK

Research Technician grade D

I conducted molecular cytogenetics research on individuals with chromosomal translocations on a project funded by the Wellcome Trust.

Oct-01/Jul-02

Serologic Laboratory, Medinfar Group, Portugal

Industrial Student Placement

This was a one year mandatory placement whilst on the 5th and final year of my undergraduate studies.

Thesis: "Selection of high-toxin producer strains of *Clostridium perfringens* type D"

Dr J Baptista- CV

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TEACHING AND SUPERVISION EXPERIENCE

Faculty at the "Basic and advanced course in genetic counselling in practice". European School of Genetic Medicine, Bologna.

I have participated as faculty in 7 editions of this residential course aimed at clinicians, nurses, genetic counsellors and other professionals with an interest in genetic counselling (e.g. psychologists and lawyers).

Sep-2007

Workshop "Human genome data mining: Ensembl and other web resources". Wessex Clinical Genetics Service, Southampton

I delivered a practical workshop aimed at helping clinicians use web resources (DECIPHER, Ecaruca, Ensembl, NCBI) useful for understanding molecular cytogenetic laboratory reports.

Informal teaching of Students, Technical Staff and Clinicians

I have provided on-the-job training to technical staff, students and clinicians on various aspects of genomic medicine. I have directly supervised 2 undergraduate theses during my postdoctoral period at the Bologna University Hospital. Furthermore, in 2014 and 2015, I have hosted internships for a total of 10 weeks for 2 students.

PUBLICATIONS

Stals KL, Wakeling M, **Baptista J** et al. "Diagnosis of lethal or prenatal-onset autosomal recessive disorders by parental exome sequencing. *Prenat Diagn.* 2018 Jan;38(1):33-43.

Thomas NS, Morris JK, **Baptista J** et al. "De novo apparently balanced translocations in man are predominantly paternal in origin and associated with a significant increase in paternal age." *J Med Genet.* 2010 Feb;47(2):112-5.

Murphy R, **Baptista J**, Holly J et al. "Severe intrauterine growth retardation and atypical diabetes associated with a translocation breakpoint disrupting regulation of the insulin-like growth factor 2 gene." *J Clin Endocrinol Metab.* 2008 Nov;93(11):4373-80.

Baptista J, Mercer C, Prigmore E et al.. "Breakpoint mapping and array CGH in translocations: comparison of a phenotypically normal and an abnormal cohort." *Am J Hum Genet.* 2008 Apr;82(4):927-36.

Gribble SM, Kalaitzopoulos D, Burford DC, Prigmore E, Selzer RR, Ng BL, Matthews NS, Porter KM, Curley R, Lindsay SJ, **Baptista J** et al. "Ultra-high resolution array painting facilitates breakpoint sequencing." *J Med Genet.* 2007 Jan;44(1):51-8.

Baptista J, Prigmore E, Gribble SM et al. "Molecular cytogenetics analyses of breakpoints in apparently balanced reciprocal translocations carried by phenotypically normal individuals." *Eur J Hum Genet.* 2005 Nov;13(11):1205-12.

ADDITIONAL SKILLS

- Holder of a Personal Home Office Licence for scientific procedures in guinea-pigs, mice, rats and zebrafish.
- Extensive experience with packages for fluorescence microscopy imaging (MacProbe, Genikon Workstation, ISIS suite), software for microarray imaging and analysis (Agilent CGH analytics and BlueFuse) and web resources for genomic data mining
- Fluent in European Portuguese (native language), English (IELTS score 8/9) and Italian.


17/05/2018