BIOGRAPHY

I received my Biology Degree at the University of Barcelona (Barcelona, Spain) in the specialization of Zoology and Ecology. After a three-year period outside of research (working in IBM), I started a PhD in the University Pompeu Fabra (Barcelona, Spain) in 2002, working on the impact of chromosomal rearrangements on the molecular evolution of primates.
During my postdoctoral period at the University of Washington (Seattle, WA) with a *Marie Curie Fellowship* from the European Union, I followed my research on non-human primate genomics and became interested in the evolution of primate genomes, especially centered on segmental duplications. I am also part of six Genome Consortia (Neanderthal (Science 2010), Bonobo (Nature 2012), Gorilla (Nature 2012), Orangutan (Nature 2011), Gibbon and Marmoset) and I have published two review papers in *Trends in Genetics* and *Annual Review of Human Genetics and Genomics*.

In 2010 I got the competitive ERC Starting Grant 2010 to establish my own group centered on the analysis genetic diversity in great apes. In 2011, I was selected as an ICREA research professor at the Universitat Pompeu Fabra (UPF).

PROJECT

European Research Council Starting Grant

Project acronym: **PRIMATESVs**

Project full title: **Identification and characterization of primate structural variation and an assessment of intra-specific patterns of selection and copy-number variation**

Overview

Studies based in both experimental and computational analyses show that ~5% of the human genome is composed of highly complex duplicated sequences. Structural variation and copy-number variant regions (CNVs) (including segmental duplications (SDs)) are usually underrepresented in genome analyses but are becoming a prominent feature in understanding the organization of genomes as well as many diseases.

The importance of studying SDs in genomics is becoming more and more evident as SDs are one of the most important features of genome evolution, having both functional and structural effects. By predisposing chromosomal architectures to be rearranged by non-allelic homologous recombination, SDs constitute genetic risk factors for many diseases (e.g. Prader-Willi, Williams-Beuren Syndromes, juvenile nephronophtisis or spinal muscular atrophy). Also, SDs are related with genes evolution because they produce partial or full coding sequence duplications that can lead to genes with new functions.

Despite international efforts to characterize thousand of human genomes to understand the extent of structural variants in the human species, primates (our closest relatives) have somehow been forgotten. Yet, they are the ideal set of species to study the evolution of these features from both mechanistic and adaptive points of view.

The object of this proposal is to discover the extent of genome structural polymorphism within the great ape species by generating next-generation sequencing datasets at high coverage from multiple individuals of diverse species and subspecies, characterizing structural variants, validating them experimentally and correlate them with gene expression. The results of these analyses will assess the rate of genome variation in primate evolution, characterize regional deletions and copy-number expansions as well as determine the patterns of selection acting upon them and whether the diversity of these segments is consistent with other forms of genetic variation among humans and great apes. In so doing, a fundamental insight will be provided into evolutionary variation of these regions among primates and into the mechanisms of disease-causing rearrangements with multiple repercussions in the understanding of evolution and human disease.