

Annual Report 2011

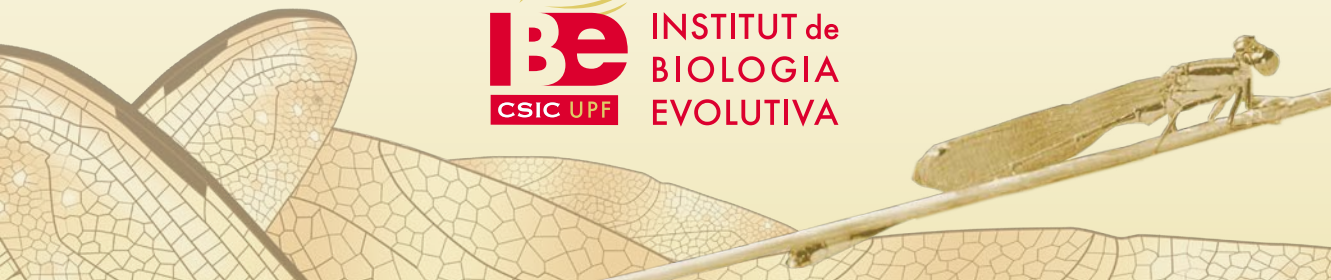


**INSTITUT de
BIOLOGIA
EVOLUTIVA**

ANNUAL REPORT 2011



INSTITUT de
BIOLOGIA
EVOLUTIVA



- **Produced by:** Institut de Biologia Evolutiva (CSIC-UPF)
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- www.ibe.upf-csic.es



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FOREWORD



by Xavier Bellés, Director of the IBE



The important thing in science is not so much to obtain new facts as to discover new ways of thinking about them.

William L. Bragg, *Nobel Guest Lecture* (1965)

Un poeta és aquell que sap reconèixer que la seva mirada de les coses no coincideix amb la dels altres. La mirada, simplement.

Miquel Bauçà, *El canvi* (1998)

Another year of the still young history of the IBE has just elapsed. Immersed in the financial crisis, 2011 has been a year of general incertitude and punctual disappointments, although a number of important achievements has been reached. Among them, the increase in the number of PIs, with the incorporation of Josefa González (in February), researcher of the Ramón y Cajal program heading the Evolutionary and Functional Genomics Lab; Luc Steels (in September), ICREA researcher and leading the Language Evolution Lab; and Iñaki Ruiz-Trillo (in December), ICREA researcher who heads the Multicellgenome Lab. These new appointments are very good news because the fields that they incorporate to the IBE research scope are exciting and complementary to those previously established. The description of the group of Josefa Gonzalez, who arrived early enough in 2011, is already included in the present report. Other very good news is the consolidation of Tomàs Marquès –a former Ramón y Cajal researcher–, as an ICREA researcher adscribed to the IBE. Moreover, some of our PIs received awards from different institutions. To mention only a few, Ricard Solé has been awarded with an ERC advanced grant, and Francesc Calafell received an ICREA Academia Award.

Concerning the inputs, we have been remarkably successful in gathering financial support from competitive calls, with 19 new granted projects that represent an income of about 3.5 million euros, half from Spanish agencies and

the other half from other sources, especially the European Union. With respect to outputs, our productivity in terms of number of ISI papers has increased from 79 (2010) to 92 (2011). However, the averaged impact factor of the journals where we published our papers decreased, and these are bad news because our goal must be to increase the quality of the papers published, rather than to increase the number of papers per year (although this should be also a goal if compatible with the increase of quality). This is an aspect of paramount importance that should deserve special attention in 2012 and beyond.

Other 2011 achievements related to the general functioning of the IBE have been the establishment of the rules of functioning approved by the Board of Trustees; the official appointment of the IBE Director, nominated by the UPF Rector and the CSIC President; and the formal nomination of the IBE Executive Board by the director. Another strategic achievement has been the official incorporation of the IBE to the Parc de Recerca Biomèdica de Barcelona (PRBB). This is great news, not only because this gives us an easier accessibility to the PRBB facilities, but also because this represents taking part of a successful mark of scientific excellence.

What would be expected for 2012? We live in a Damoclian era where all the swords are hanging over every head. Thus, the time coming will be especially complicated though plenty of exciting challenges. The first challenge is the financial consolidation of the IBE building, perhaps the more pressing need of our IBE project. Additionally, we should diversify our funding sources, we should continue our efforts to strengthen the quality of our research, we should increase our international visibility and scientific leadership, and, importantly, we should increase the IBE cohesion, fostering internal collaborations and synergies. One of the first important activities of the IBE in 2012 is the evaluation *in situ* to be carried out on the 1st and 2nd of March by the seven members of the External Scientific Committee (CEE). We count on the advice of the CEE and we are confident that it will be of great value in order to take the right decisions in the future, which is approaching fast. In science, as well as in arts, a creative way to face new and difficult challenges is to discover new ways of looking and thinking about them. As scientists (and artists in some way), this vision should be familiar enough to us, and we should be able to put every creative effort to make the challenges soluble and realistic. Let's get down working, then.

INTRODUCTION TO THE IBE



SCOPE AND GENERAL GOALS

The Institute of Evolutionary Biology (IBE) was formally founded in July 2008 as a joint institute of the Spanish National Research Council (CSIC) and the Pompeu Fabra University (UPF). The Institute functional structure was not fully operative until mid 2009, when the Management Unit was complete and ongoing. Initially, the IBE was created with 11 independent research groups from the Molecular Biology Department (CID, CSIC) and 6 independent research groups from the Evolutionary Biology Unit (CEXS, UPF). Nowadays, the IBE involves more than a hundred people and 21 independent research groups distributed in five Scientific Programs. The scope and general research goals of the IBE focus on biological evolution.

Indeed, one of the great challenges of the 21st century, after the publication of the Human Genome Sequence and the genomes of many other species, is the description and understanding of biodiversity, either within species (variation, polymorphism) and/or between species (divergence), as an essential element to understand the essential mechanisms of life. Evolutionary biology provides the key tools and concepts to make sense of these data. Thus, the main mission of the IBE is to increase knowledge and promote research of excellence in evolutionary biology. The main strength of the IBE, and its main peculiarity, is the ability to address biodiversity studies describing functional and evolutionary genomics at all levels of observation: molecular, biochemical, physiological, and morphological.

The IBE vision, defined as the projection in the long-term future of the institute, is to be a centre of international reference in the study of biodiversity and its evolution both from molecular and genomic perspectives. Our primary value is to break the segmentation traditionally imposed by the areas of knowledge in the Spanish Research System. Establishing the foundations of a multidisciplinary approach, not limited to view Evolution from a mere biological standpoint, but extended to the human sciences in the broadest sense.

GENERAL STRUCTURE

In addition to the classical figures of Director and Vicedirector, and the Executive board, the IBE counts with the important managing structures of the Board of Trustees and the External Scientific Committee.

BOARD OF TRUSTEES

IBE main managing structure is the Board of Trustees, composed by two representatives of both partner institutions (CSIC and UPF). It is competent in the direction, composition, research lines, structure and functioning rules of the IBE.

Members of Board of Trustees along 2011:

Luis Calvo

| CSIC Institutional Coordinator in Catalonia

Teresa García Milà

| UPF Vicechancellor of Scientific Policy

Francisco Montero

| CSIC Vicepresident of Institutional Relationships and Organization

Francesc Posas

| CEXS-UPF Department Director

EXTERNAL SCIENTIFIC COMMITTEE (ESC)

The IBE External Scientific Committee (ESC) is a group of scientific experts with international recognition in the Evolutionary Biology field whose main task is to help the IBE in the definition of new research lines and strategies and in the best ways to recruit talent and widen the scientific strength of the Institute.

The composition of the External Scientific Committee was approved by the Board of Trustees along 2011. The first meeting and in situ evaluation of IBE activity by this commission will take place on the 1st and 2nd March 2012.

The composition of the ESC is as follows:

Chairman

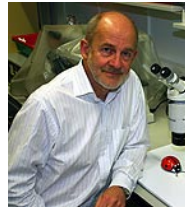


Andrés Moya |
Universitat de València,
València, Spain

Members



Carlos Bustamante |
Stanford University, Palo Alto,
CA, USA



Stuart Reynolds |
University of Bath, Bath, UK



Brian Charlesworth |
University of Edinburgh,
Edinburg, UK



Luis Serrano |
Centre de Regulació
Genòmica, Barcelona, Spain



Gonzalo Giribet |
Harvard University, Cambridge,
MA, USA



Eske Willerslev |
University of Copenhagen,
Copenhagen, Denmark

EXECUTIVE BOARD

The IBE Executive Board is composed by 7 members:

IBE Director

- | Xavier Bellés

IBE Vicedirector

- | Arcadi Navarro (acting also as the Coordinator of the "Comparative and Computational Genetics" Program)

Current Members

- | José Castresana (acting as the Coordinator of the "Animal Phylogeny and Systematics" Program)
- | David Comas (acting as the Coordinator of the "Population Genetics" Program)
- | Maria-Dolors Piulachs (acting as the Coordinator of the "Functional Evolution in Insects" Program)
- | Ricard Solé (acting as the Coordinator of the "Complex Systems" Program)

General Manager and Board Secretary

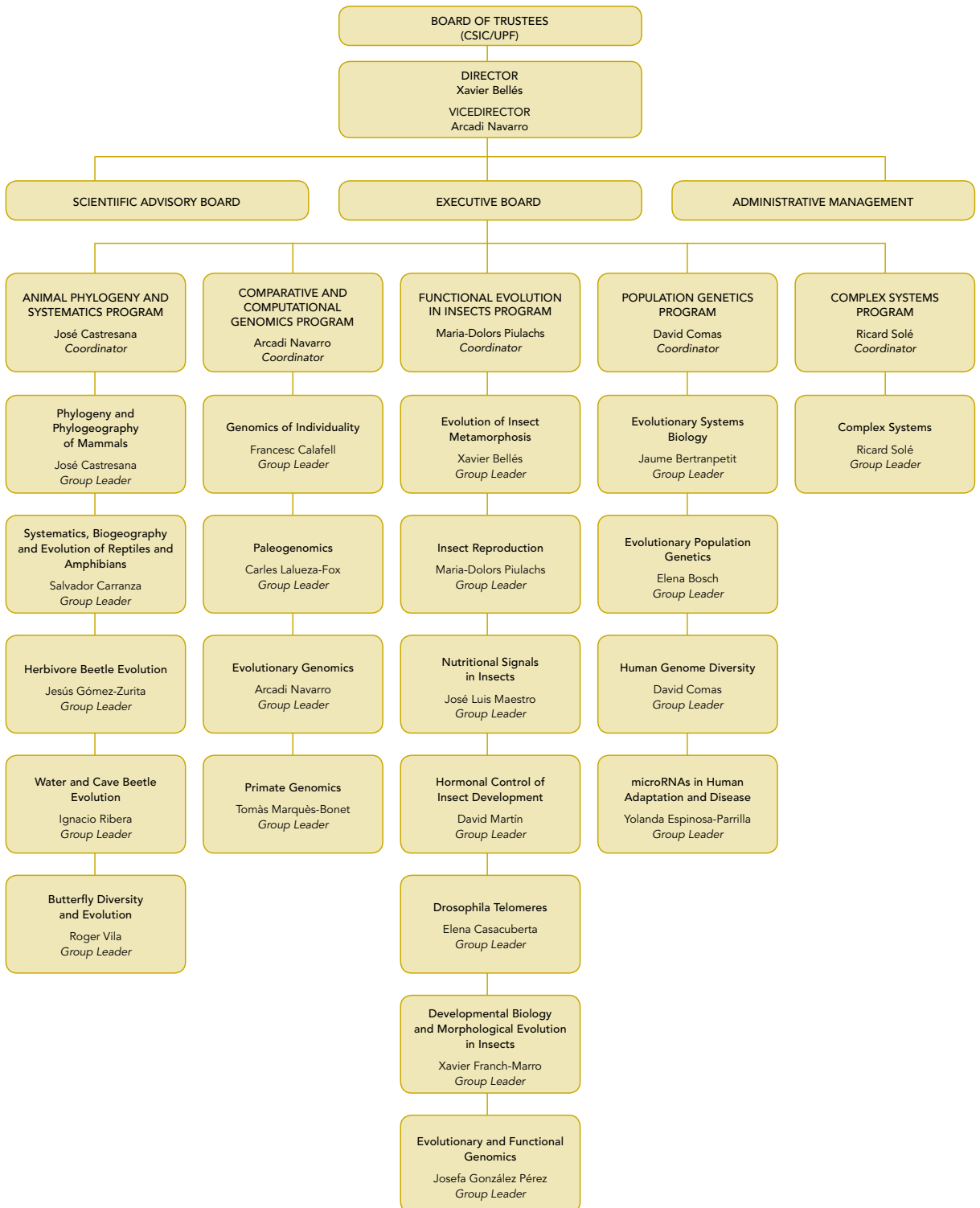
- | Anna Pérez-Lezaun

SCIENTIFIC STRUCTURE

The scientific structure of the IBE is composed by 21 different groups and five scientific programs:

- | Animal Phylogeny and Systematics
- | Comparative and Computational Genomics
- | Functional Evolution in Insects
- | Population Genetics
- | Complex Systems

IBE ORGANISATION CHART



SERVICE UNITS

In support of the IBE scientific structure, three service units have been planned: a Management Unit already functional, and two technical units under constitution, Bioinformatics Unit and Experimental Techniques Unit.

MANAGEMENT UNIT

The IBE central management unit was formally constituted by mid-2009 with the incorporation of the IBE General Manager from the Pompeu Fabra University and a ViceManager from the CSIC. Nowadays, the IBE management unit is composed by 5 people and covers at a micro scale level all the basic Institute running processes (accounting, human resources, purchasing, logistics and safety, and support to projects).



MANAGEMENT UNIT

General Manager

Anna Pérez-Lezaun | UPF

ViceManager and Accountant

Rita Arias | CSIC

Administrative Support

Emiliano González | CSIC

Blanca Álvarez | CSIC

Judit Sainz | UPF

EXPERIMENTAL TECHNIQUES

This unit coordinates the maintenance and use of the insect colonies and of the specialized technical instrumentation and facilities mainly related to the activity of those groups belonging to the Functional Evolution in Insects Program. Right now, it counts on a staff technician from the CSIC and a two-year contract technician from the JAETEC (CSIC) program appointed in the last trimester of 2010. It is planned that personnel and functions of this unit should be enlarged in the next future to give support to other programs and technological needs.

Members:

- | Cristina Olivella | Technical Staff (CSIC)
- | José Martínez | JAETEC (CSIC)

BIOINFORMATICS UNIT

This unit started formally functioning at the beginning of 2011 with the incorporation of a shared specialized bioinformatician (MICINN-PTA) who joined a group of IT technicians from the research groups of the Comparative and Computational Genomics Program. The Bioinformatics Unit coordinates the support to all IBE programs in all tasks requiring knowledge on computational biology, particularly in what refers to the growing computational needs of current biological research. The Unit offers highly specialized support (installation of software, creation of databases, scripting...) and manages the access of IBE researchers to our local computational cluster, an IBM blade center with more than 100 cores and 36 TB of storage capacity. During 2011, the Unit started preparing a major upgrading of the IBE computational capacity, which will reach ~400 cores and ~300 TB during 2012.

Members:

- | José María Heredia | CSIC Contract (PTA-MICINN)

PROGRAM RESEARCH ASSISTANTS

Apart from the mentioned formal units, the IBE also counts with three long-term laboratory technicians that give scientific key support to different IBE programs:

Members:

- | Rocío Alonso | JAETEC (CSIC) · Animal Phylogeny and Systematics Program
- | Mònica Vallés | Technical Staff (UPF) · Population Genetics and Comparative and Computational Genetics Programs
- | Eva Ramallo | UPF Contract · Complex Systems and Population Genetics Programs



PROGRAMS RESEARCH ASSISTANTS

Experimental Techniques

Cristina Olivella | Technical Staff (CSIC)

José Martínez | JAETEC (CSIC)

Bioinformatics Unit

José María Heredia | CSIC Contract (PTA-MICINN)

Program Research Assistants

Rocío Alonso, JAETEC (CSIC) | Animal Phylogeny and Systematics Program

Mónica Vallés, Technical Staff UPF | Population Genetics and Comparative and Computational Genomics Programs

Eva Ramallo, UPF Contract | Complex Systems and Population Genetics Programs

PERSONNEL

At the end of 2011, the IBE had 130 members (Table 1) with a ratio of men to women around 1.45 and an internationalization level of more than 20% foreign members (in postdoctoral researchers this percentage increases up to 33 %).

Table 1. IBE personnel distribution by categories. December 2011.

	2011	2010	2009
Faculty	18	17	16
Long-term Researchers*	5	3	3
Postdoctoral Researchers	25	21	17
Predocctoral Researchers	43	42	33
Support Personnel			
Laboratory Technicians	8	9	4
Bioinformaticians	6	5	4
Administrative Staff	5	5	5
Others (project support)	9	2	2
Long-term Visitors (> 1 month)	6	14	8
Students (undergrad or master)	5	No data available	No data available
TOTAL	130	118	92

*Marie Curie, Ramón y Cajal or ICREA researchers

LOCALISATION

While not having a specific building, the IBE has two different headquarters:

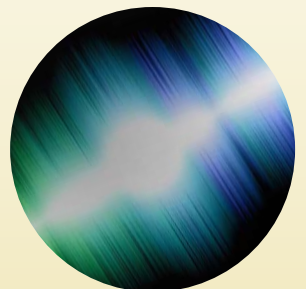
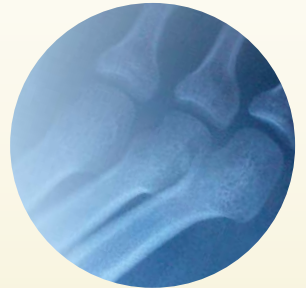
IBE at the CMIMA building:
Passeig Marítim de la Barceloneta, 37-49.
08003 Barcelona, Spain

IBE at the PRBB building:
C/ Dr. Aiguader, 88.
08003 Barcelona, Spain.





IBE RESEARCH PROGRAMS







PROGRAM

animal phylogeny and systematics

RESEARCH GROUPS

Phylogeny and Phylogeography of Mammals

José Castresana, *Group Leader*

Systematics, Biogeography and Evolution of Reptiles and Amphibians

Salvador Carranza, *Group Leader*

Herbivore Beetle Evolution

Jesús Gómez-Zurita, *Group Leader*

Water and Cave Beetle Evolution

Ignacio Ribera, *Group Leader*

Butterfly Diversity and Evolution

Roger Vila, *Group Leader*

The members of this program carry out research on animal biodiversity from a phylogenetic perspective with the aim of gaining further insight into the tree of life. The program specific research interests include the origin and distribution of biodiversity (whether morphological, genetic, ecological or functional), systematics of certain groups, speciation, hybridization, diversification, biogeography, evolutionary ecology and phylogenetic methodology. Program members work on the systematics and phylogenetic relationships among certain groups of organisms, but also on the evolutionary processes that gave rise to current biodiversity patterns. The main groups studied are mammals, reptiles, amphibians, butterflies and beetles, thus including a broad variety of animal taxa. A wide range of techniques is covered, from field work and morphological analysis, to genetic studies, genomic data mining and software development. We are increasingly using large-scale phylogenetic analyses (both in terms of species considered and sequenced data) in order to obtain more robust phylogenies and evolutionary conclusions. Phylogenetic trees are a common framework for many evolutionary studies and therefore this research program provides many points of contact with other programs at the IBE.

GROUP

PHYLOGENY AND PHYLOGEOGRAPHY OF MAMMALS



GROUP MEMBERS

José Castresana, Group Leader

| Research Scientist, CSIC



Alejandro Sánchez-Gracia, Post-doc | Juan de la Cierva Contract

Javier Igea, PhD Student | JAEPRE-CSIC Fellowship

Ana Rodríguez-Prieto, PhD Student | FPI Scholarship, MICINN

Víctor Soria-Carrasco, PhD Student | FPI Scholarship, MICINN

RESEARCH OUTLINE

Our main goal is the application of phylogenetic analyses to study animal biodiversity and its evolution, with an emphasis on mammals. We are particularly interested in the study of global diversification patterns and in the analysis of the factors that affect the net generation of species. We are also interested in the interphase between phylogenetics and population genetics, which may shed light on the analysis of speciation. In particular, we are conducting different studies to unravel speciation patterns in small mammals of the Iberian Peninsula. Finally, since phylogenies are one of the main tools of our work, we are always trying to improve phylogenetic analyses by introducing new methodologies and programs.

RESEARCH SUBLINES

1. Phylogeny, genetic diversity and speciation of mammals studied with multiple markers

Resolution of the species tree of closely related species requires the use of multiple genetic markers. With the aim of developing these techniques, we have analyzed the available mammalian genomes to compile new sets of intronic markers. We are currently sequencing these markers in different species, particularly small mammals of the Iberian Peninsula (*Galemys pyrenaicus*, *Neomys anomalus* and *Microtus sp.*), to better understand patterns of genetic diversity, speciation scenarios, and gene flow within species. We are also interested in detecting cryptic lineages with the help of both mitochondrial and the novel intronic markers.

2. Methodological aspects of phylogenetic reconstruction: gene trees and species trees

Phylogenetic trees are essential in evolutionary biology and therefore understanding their potentials and limitations is important to make better use of them. With this aim, we are working on different aspects of tree reconstruction and on the detection of artifacts that may affect phylogenetic analyses. We are interested in all steps of these analyses, including the generation of alignments, the reconstruction of phylogenetic trees, and the comparison of these trees. More recently, we have become interested in methodologies at the interphase between phylogenetics and population genetics, and in the estimation of species trees from gene trees.

3. Analysis of species diversification in mammals

Species-level phylogenies contain important information in their patterns of branch splits. For example, current statistical techniques make possible to estimate rates of diversification from phylogenetic trees that have an adequate sampling of species. It is also possible to study the variability of diversification rates along the evolution of a group. The availability of a large amount of sequences in databases allows us to perform large-scale phylogenetic analyses with the aim of studying global diversification patterns in mammals.

PUBLICATIONS 2011

ISI Articles

- Soria-Carrasco, V., and Castresana, J. 2011. Patterns of mammalian diversification in recent evolutionary times: global tendencies and methodological issues. *Journal of Evolutionary Biology* 24: 2611-2623.

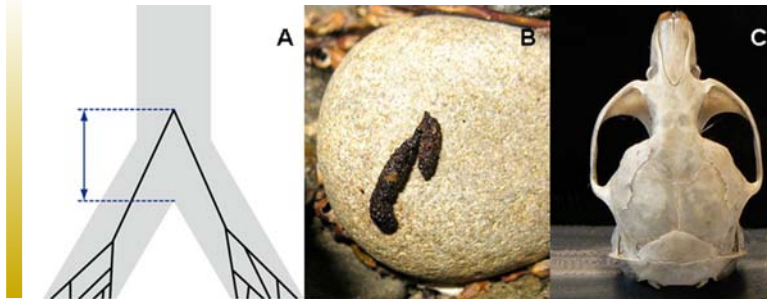


Fig. 1: We have been working on methodological aspects of speciation time estimation using species trees and gene trees (the discrepancy between both estimates is shown with an arrow; A). We are now testing these ideas in different mammalian species groups; for these studies, we are making extensive use of noninvasive samples such as excrements of *Neomys* sp. (B) and skulls of *Microtus* sp. obtained from owl pellets (C).

FUNDED PROJECTS

Project Title: Distribution and genetic diversity of the Iberian desman (*Galemys pyrenaicus*) in the Spanish Network of National Parks

Financed by: Ministerio de Medio Ambiente y Medio Rural y Marino. Network of National Parks Research Funds (014/2008).

Years: 2009-2011

PI: Joaquim Gosálbez

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO

Financed by: Generalitat de Catalunya (2009 SGR 1462)

Years: 2009-2013

PI: Salvador Carranza

Project Title: Phylogeny and speciation of mammals studied with multiple nuclear markers

Financed by: Ministerio de Ciencia e Innovación (GL2008-00434)

Years: 2009-2011

PI: José Castresana

GROUP

SYSTEMATICS, BIOGEOGRAPHY AND EVOLUTION OF REPTILES AND AMPHIBIANS



GROUP MEMBERS

Salvador Carranza, Group Leader

| Tenured Scientist, CSIC



Elena Gómez-Díaz, Post-doc Researcher | Beatriu de Pinós Contract

Josep Roca, Technician | Project Contract

Joan García-Porta, PhD Student | JAEPRE-CSIC Fellowship

Margarita Metallinou, PhD Student | FPU Scholarship, MEC

Raquel Vasconcelos, PhD Student co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal

Catarina Rato, PhD Student co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal

Mafalda Barata, PhD Student co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal

Emilio Valbuena Ureña, MSc Student | Master UAB

Hernán Morales, MSc Student | Master in Evolutionary Biology, University of Groningen, The Netherlands

RESEARCH OUTLINE

Our research focuses on the application of phylogenetic analyses of reptiles and amphibians to understand how biodiversity is generated and maintained. We are also interested in inferring the biogeographical and evolutionary patterns of the different groups studied, to revise their taxonomy and to use all this information to address conservation issues. Although our investigations include the study of many different reptile and amphibian groups, our main research sublines focus mainly on the faunas of the Mediterranean Basin and Arabia, including some oceanic and continental islands, as for instance the Canary and the Cape Verde islands in the Atlantic Ocean and the archipelago of Socotra in the Indian Ocean.

RESEARCH SUBLINES

1. Historical biogeography and evolution of the reptiles and amphibians around the westernmost Mediterranean

Our main objectives are: 1) infer the geographical history and evolution of the reptiles and amphibians around the westernmost Mediterranean basin; 2) characterize and compare the molecular evolutionary rates of reptiles and amphibians; 3) identify using phylogenies the possible existence of Pliocene fossil islands in the area of study; and 4) test the current taxonomy of the groups concerned.

2. Uses of phylogenies to study evolutionary, ecological and biogeographical processes: the North African and Arabian arid reptile faunas

In this project, we are using molecular phylogenies from multiple reptile taxa to address a whole range of evolutionary, ecological and biogeographical questions. Our main objectives are: 1) to understand how deserts gain and maintain their endemic faunas; 2) to infer the age of the Sahara and Arabian deserts; 3) to compare the diversification rates of several desert lineages; and 4) to test and improve the current taxonomy of the groups concerned.

3. Island biogeography and evolution

The main goal of this research line is to take advantage of the excellent experimental conditions of the island systems to try to understand how biodiversity is generated and maintained. Island systems offer great opportunities to study evolution, and are especially attractive environments for several reasons: 1) they present discrete geographical entities within defined oceanic boundaries; 2) gene flow between individual islands is reduced by oceanic barriers; 3) their often small geographical size has made the cataloguing of flora and fauna easier than continental systems; 4) despite their small geographical size they can contain a diversity of habitats; and 5) they are often geologically dynamic with historical and contemporary volcanic and erosional activity. At the moment we are



Fig. 1: *Asaccus platyrhynchus*, N. Oman

investigating both oceanic and continental reptile islands faunas from several places in the world including the Canary Islands and Cape Verde in the Atlantic Ocean and the Socotra archipelago in the Indian Ocean.

4. Distribution and genetic diversity of the Iberian desman (*Galemys pyrenaicus*) in the Spanish Network of National Parks (In collaboration with José Castresana)



Fig. 2: Camp site in Masirah Island, Oman

PUBLICATIONS 2011

ISI Articles

- Bauer, A.M., Parham, J.M., Brown, R.M., Stewart, B.L., Grismer, L., Papenfuss, T.J., Böhme, W., Savage, J.M., Carranza, S., Grismer, J.L., Wagner, P., Schmitz, A., Ananjeva, N.B., and Inger, R.F. 2011. Availability of new Bayesian-delimited gecko names and the importance of character-based species descriptions. *Proceedings of the Royal Society B-Biological Sciences* 278: 490-492.
- de Pous, P., Mora, E., Metallinou, M., Escoriza, D., Comas, M., Donaire, D., Pleguezuelos J.M., and Carranza, S. 2011. Elusive but widespread? The potential distribution and genetic variation of *Hyalosaurus koellikeri* (Günther, 1873) in the Maghreb. *Amphibia-Reptilia* 32: 385-397.
- Gómez-Díaz, E., Boulinier, T., Sertour, N., Cornet, M., Ferquel, E., and McCoy, D.K. 2011. Genetic structure of marine *Borrelia garinii* and population admixture with the terrestrial cycle of Lyme borreliosis. *Environmental Microbiology* 13: 2453–2467.
- Lobato, E., Pearce-Duvel, J., Staszewski, V., Gómez-Díaz, E., González-Solís, J., Kitaysky, A., McCoy, K.D., and Boulinier, T. 2011. Seabirds and the circulation of Lyme borreliosis bacteria in the North Pacific. *Vector-Borne and Zoonotic Diseases* 11: 1521-1527.
- Martínez-Silvestre, A., Amat, F., Bargalló, F., and Carranza, S. 2011. Incidence of pigmented skin tumors in a population of wild Montseny Brook Newt (*Calotriton arnoldi*). *Journal of Wildlife Diseases* 47: 410-414.
- Miralles, A., Vasconcelos, R., Perera, A., Harris, D.J., and Carranza, S. 2011. An integrative taxonomic revision of the Cape Verdean skinks (Squamata, Scincidae). *Zoologica Scripta* 40: 16-44.
- Rato, C., Carranza, S., and Harris, D.J. 2011. When selection deceives phylogeographic interpretation: The case of the Mediterranean house gecko, *Hemidactylus turcicus* (Linnaeus, 1758). *Molecular Phylogenetics and Evolution* 58: 365-373.
- Roscales, J.L., Gómez-Díaz, E., Neves, V., and González-Solís, J. 2011. Trophic versus geographic structure in stable isotope signatures of pelagic seabirds breeding in the northeast Atlantic. *Marine Ecology Progress Series* 434: 1-13.

Books

- Rivera, X., Escoriza, D., Maluquer-Margalef, J., Arribas, O., and Carranza, S. 2011. Amfibis i rèptils de Catalunya, País Valencià i Balears. Lynx, Barcelona, Spain, 276 pages.



Fig. 3: *Psammophis schokari*, Masirah Island, Oman

Other Publications

- Barata, M., Perera, A., Harris, D.J., Van Der Meijden, A., Carranza, S., Ceacero, F., García-Muñoz, E., Gonçalves, D., Henriques, S., Jorge, F., Marschall, J.C., Pedrajas, L., and Sousa, P. 2011. New observations of amphibians and reptiles in Morocco, with a special emphasis on the eastern region. *Herpetological Bulletin* 116: 4-13.

FUNDED PROJECTS

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO

Financed by: Generalitat de Catalunya (AGAUR) (2009SGR1462)

Years: 2009-2013

PI: Salvador Carranza

Project Title: Using Agamid lizards to understand the origin and colonization of North Africa and Arabia

Financed by: Proyectos conjuntos en el marco de los convenios de cooperación del DCIC: CSIC - Russian Foundation for Basic Research (RFBR) (2010RU0055)

Years: 2011-2012

PI: Salvador Carranza

Project Title: Living on the edge origin and diversification of the reptile communities of the deserts of North Africa and Arabia

Financed by: Ministerio de Ciencia e Innovación (CGL2009-11663)

Years: 2010-2012

PI: Salvador Carranza

Project Title: Parasite evolution on islands: reptiles and their parasites as a model study (PARIS)

Financed by: European Commission, FP7-PEOPLE-Reintegration Grants. (ERG-PARIS-276838)

Years: 2010-2013

PI: Elena Gómez-Díaz

Project Title: Distribution and genetic diversity of the Iberian desman (*Galemys pyrenaicus*) in the Spanish Network of National Parks

Financed by: Ministerio de Medio Ambiente y Medio Rural y Marino (014/2008)

Years: 2009-2011

PI: Joaquim Gosàlbez

Fig. 4: Wahiba Sands, Oman.



GROUP

HERBIVORE BEETLE EVOLUTION



Gissela la Cadena

Anna Papadopoulou

Anabela Cardoso

Jesús Gómez-Zurita

Tinguaro Montelongo

GROUP MEMBERS

Jesús Gómez-Zurita, Group Leader

| Tenured Scientist, CSIC



Anabela Cardoso, Research Assistant | CSIC

Gissela la Cadena, PhD Student | AECID Scholarship, MAEC

Tinguaro Montelongo, PhD Student | FPI Scholarship, MICINN

Anna Papadopoulou, Post-doc Researcher | Juan de la Cierva Programme, MICINN

RESEARCH OUTLINE

We are interested in the systematics of beetles, the analysis of geographic and ecologic speciation and of the spatial structure of genetic diversity within a temporal framework (phylogeography), as well as in the study of biological processes such as hybridization, unisexuality and insect-plant associations from an evolutionary perspective. The basic approach that we follow to tackle this broad spectrum of research topics is a phylogenetic framework based on information provided by DNA sequences. The focus of our research is a megadiverse group of herbivorous beetles, the Chrysomelidae, popularly known as «leaf beetles». With well over 35,000 known species, from the Arctic tundra to remote Pacific islands, the Chrysomelidae cover a wide range of life histories, offering suitable models to investigate any of the general evolutionary topics outlined above.

RESEARCH SUBLINES

1. Evolution of reproductive strategies: the role and importance of hybridization

The leaf beetle genus *Calligrapha* has both bisexual and unisexual species, whereby the latter seem to be the result of historical interspecific hybridization events. We study these events with a historical, phylogeographic perspective, but also from a genomic point of view, investigating the molecular signatures of the hybrid origin of these unisexual taxa.

2. Biodiversity and conservation of the tropical dry forest in Nicaragua

Tropical dry forests are one of the most threatened ecosystems on the planet. In this project we implement molecular tools to investigate both the inventory and the interactions sustained by this habitat in Nicaragua. Diagnostic DNA sequences are used for rapid biodiversity assessment of flowering plants and two groups of megadiverse phytophagous beetles, the Chrysomelidae and the Curculionidae, but also the host plants of the beetles by PCR-amplifying DNA



Fig. 1: *Biodiversity, biodiversity, biodiversity!* The overwhelming diversity of leaf beetles is a constant source of ideas and pleasure researching on its structure, evolution and interactions.

Fig. 2-3: Tropical seasonally dry forest is at the heart of several of our research and conservation initiatives. Team members collecting herbivorous beetles in the island of Juan Venado (Nicaragua).

remains of their diet to compare them with the local genetic database for the flora. This strategy will hopefully speed up considerably the much-needed tasks of inventorying diversity and ecological strategies compared to traditional approaches to the same problems.

3. Evolution in Pacific Islands: diversity and diversification of New Caledonian Chrysomelidae

New Caledonia is a continental fragment of Gondwana isolated from other continental land masses from the end of the Cretaceous. It hosts a singular biota with high endemism and in some cases purportedly very ancient, perhaps relicts of Gondwanian origin. However, this ancient origin is contentious and it was recently proposed that the island would have been colonized recently, after periods of underwater submergence in the Oligocene. The Chrysomelidae fauna in New Caledonia is very rich, with over 200 species described, mostly endemic. In this project, we aim at characterizing their species diversity using morphology and molecular markers, but also to investigate their origin, age and diversification using molecular phylogenetics, as well as their ecologies, based on field observations and the use of molecular markers

4. Climate and speciation: European species complexes of *Cryptocephalus*
Cryptocephalus (Chrysomelidae) includes two closely related species complexes in the Western Palaearctic, the *C. sericeus*-complex and the *C. hypocharididis*-complex, with several species in mountainous areas, mainly in the Pyrenees and the Alps, and offer important taxonomic problems, including clinal variation of characters and possible hybrids. These are suggestive of recent, incomplete speciation and secondary contacts due to range changes, possibly related to climate oscillations during the Quaternary. We investigate these species complexes using a combination of phylogeographic approaches with nuclear and mitochondrial markers to delimit evolutionary units and recognize putative hybrids, as well as molecular phylogenetic analyses to time their diversification.

5. Systematic revision of Central and South American *Calligrapha*

The genus *Calligrapha*, with over 100 species in the American continent, is focus of our research on the evolution of reproductive strategies and host associations. Unfortunately, the taxonomy of the group has only been revised recently for North American species, including some 30 taxa. In order to clarify the systematics of the group as a guide for our molecular phylogenetic work, we are working towards the revision of the bulk of the genus in its Southern Nearctic and Neotropical distribution.



PUBLICATIONS 2011

ISI Articles

- Ge, D.Y., Chesters, D., Gómez-Zurita, J., Zhang, L.J., Yang, X.K., and Vogler, A.P. 2011. Anti-predator defense drives parallel morphological evolution in leaf beetles. *Proceedings of the Royal Society of London. B.* 278: 2133-2141.
- Gómez-Zurita, J. 2011. A new species of Criocerinae (Chrysomelidae) from New Caledonia: *Oulema (Oulema) taophiloides* sp. nov. *Zootaxa* 2870: 63-68.
- Gómez-Zurita, J. 2011. Revision of New Caledonian Eumolpinae described by K.M. Heller (Coleoptera: Chrysomelidae). *Zootaxa* 3060: 31-46.
- Gómez-Zurita, J. 2011. *Rhyarida foensis* (Jolivet, Verma and Mille, 2007), comb. n. (Coleoptera: Chrysomelidae) and implications for the colonization of New Caledonia. *ZooKeys* 157: 33-44.

FUNDED PROJECTS

Project Title: Evolución sin sexo: Circunstancias históricas e implicaciones ecológicas del origen híbrido de insectos unisexuales.

Financed by: Ministerio de Ciencia e Innovación (CGL2008-00007)

Years: 2009-2011

PI: Jesús Gómez-Zurita

Project Title: Sincronización rápida de inventario e interacciones en estudios de biodiversidad: herramientas moleculares al servicio del conocimiento y conservación del bosque seco tropical en Nicaragua.

Financed by: Fundación Banco Bilbao Vizcaya

Years: 2009-2012

PI: Jesús Gómez-Zurita

Project Title: Sentando las bases para la creación del banco de germoplasma del bosque seco tropical americano, el BGBST «El Sálamo»

Financed by: Programa de Cooperación Interuniversitaria e Investigación Científica (PCI-AECID)

Years: 2011-2012

PI: Jesús Gómez-Zurita

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO

Financed by: Generalitat de Catalunya, AGAUR (2009 SGR1462)

Years: 2009-2013

PI: Salvador Carranza

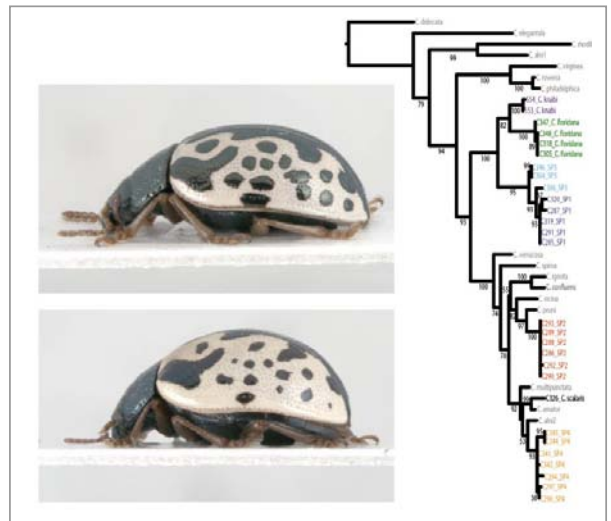
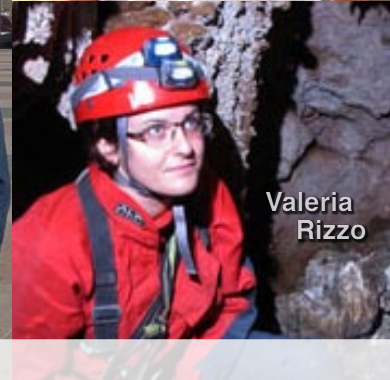


Fig. 4: Interspecific hybridization in *Calligrapha* is a frequent evolutionary process. We try to understand its causes and effects using a multifaceted approach from genetic, phylogenetic and phenotypic angles.

GROUP

WATER AND CAVE BEETLE EVOLUTION



Ignacio Ribera, Group Leader

| Research Scientist, CSIC



Amparo Hidalgo Galiana, PhD Student | FPI Scholarship, MICINN

Valeria Rizzo, Postgraduate Grant-PhD Student | Università La Sapienza, Roma

Andrey Rudoy, PhD Student | JAE Scholarship, CSIC

David García Vázquez, PhD Student | FPI Scholarship, MICINN

André Silva Fernandes, PhD Student | CAPES Grant, Government of Brazil

Rocío Alonso Rodríguez, Laboratory Technician | CSIC Contract

RESEARCH OUTLINE

Beetles are the most diverse group of extant metazoa. Their 250 MY of evolutionary history and a vast ecological and morphological variation allow to use them to undertake virtually every problem in evolutionary biology, from global macroecological and macroevolutionary patterns to phylogenetics and biogeography down to population genetics and physiology. We use different groups of water and cave beetles to address some of these questions, centred in the origin and distribution of biodiversity. Our current focus is the study of the causes and consequences of range expansions in aquatic Coleoptera, and the evolution of adaptations to the subterranean life in Leptodirini beetles (in collaboration with Alexandra Cieslak and Javier Fresneda).



Fig. 1: Head of *Hydroporus bythinicus*, a new species of diving beetle from Turkey described from an upwelling spring, with some morphological characters suggesting a subterranean lifestyle (such as the reduced eyes).

RESEARCH SUBLINES

1. Thermal tolerance and Pleistocene range expansions

There are several groups of diving beetles which include species with narrow and wide geographical ranges that are known to diverge in their thermal tolerances, such as those of the *Agabus brunneus* group and the genus *Deronectes*. With the use of phylogeographies and analyses of the whole proteome expression through 2D electrophoresis we try to establish the role of shifts in thermal tolerance in the Pleistocene range expansions of some of these species.

2. Origin and diversification of the cave beetle *Troglocharinus*

Troglocharinus is a genus of cave beetles with a wide distribution in the Catalonian coastal ranges and the pre-Pyrenees. It is a good example of diversification within the subterranean medium, and with the collaboration of some biospeleologists we are trying to understand its origin and evolution.

3. Evolution of the complex male genitalia in Hydraenidae

The extraordinary complexity of the male genitalia of some arthropods has always intrigued evolutionary biologists. Some genera of Hydraenidae combine an extreme uniformity in external morphology with a magnificent repertoire of aedeagal extravaganza. Using a comparative phylogenetic and morphometric approach we aim to discern which are the selective forces shaping the evolution of male genitalia in this group of beetles.

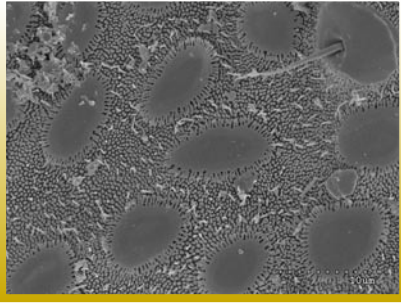


Fig. 2: The function of many of the microstructures seen in insects is totally unknown, as is the case of these flat platforms observed in the prosternum of a new genus and species of riffle beetle (Elmidae) from the Amazon basin.

4. Riffle beetles of the Amazonian region

The Elmidae (riffle beetles) are a family of Coleoptera dominant in the aquatic ecosystems of the Neotropical region, but with a largely unknown evolutionary history. With a combination of morphological and molecular data we want to reconstruct the phylogeny of the main lineages of Amazonian Elmidae, as well as to describe their immature stages by matching adults and larvae with molecular data (in collaboration with Neus Hamada, Instituto Nacional de Pesquisas da Amazônia [INPA], Brasil).

PUBLICATIONS 2011

ISI Articles

- Abellán, P., Benetti, C.J., Angus, R.B., and Ribera, I. 2011. A review of range shifts in Quaternary European aquatic Coleoptera. *Global Ecology and Biogeography* 20: 87-100.
- Abellán, P., and Ribera, I. 2011. Geographic location and phylogeny are the main determinants of the size of the geographical range in aquatic beetles. *BMC Evolutionary Biology* 11: 344.
- Andújar, C., Hernando, C., and Ribera, I. 2011. A new endogean, anophthalmous species of *Parazuphium* Jeannel from Northern Morocco (Coleoptera: Carabidae), with new molecular data for the tribe Zuphiini. *Zookeys* 103: 49-62.
- Faille, A., Casale, A., and Ribera, I. 2011. Phylogenetic relationships of Western Mediterranean subterranean Trechini groundbeetles (Coleoptera: Carabidae). *Zoologica Scripta* 40: 282-295.
- Fernandes, A.S., Passos, M.I.S., and Hamada, N. 2011. *Stegoelmis* Hinton, 1939 (Coleoptera: Elmidae: Elminae) in Brazil: two new species and a key to the Brazilian species. *Zootaxa* 2921: 56-64.
- Fresneda, J., Grevennikov, V., and Ribera, I. 2011. The phylogenetic and geographic limits of Leptodirini (Insecta: Coleoptera: Leiodidae: Cholevinae), with a description of *Sciaphyes shestakovi* sp. n. from the Russian Far East. *Arthropod Systematics & Phylogeny* 69(2): 99-123.
- Hidalgo-Galiana, A., and Ribera, I. 2011. Late Miocene diversification of the genus *Hydrochus* (Coleoptera, Hydrochidae) in the west Mediterranean area. *Molecular Phylogenetics and Evolution* 59: 377-385.

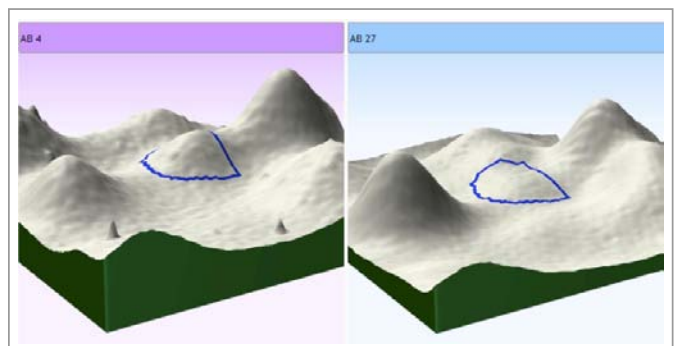


Fig. 3: The analysis of protein expression with bidimensional electrophoresis allows the comparison between different thermal treatments in closely related species. In the image, differences in the expression of a protein between specimens of the same population of *Agabus brunneus* (a diving beetle) after being exposed to low (4°C) or high (27°C) temperatures.

- Junger, B., and Faille, A. 2011. Remarkable discovery in a cave of south west Morocco: *Siagona taggadertensis* n.sp. (Carabidae: Siagoninae). *Annales de la Soci t  Entomologique de France* (n.s.), 47: 162-167.
- Ribera, I., Castro, A., D az-Pazos, J.A., Garrido, J., Izquierdo, A., J ch, M.A., and Valladares, L.F. 2011. The geography of speciation in narrow range endemics of the "Haenydra" lineage (Coleoptera, Hydraenidae, *Hydraena*). *Journal of Biogeography* 38: 502-516.
- Trizzino, M., Audisio, P.A., Antonini, G., Mancini, E., and Ribera, I. 2011. Molecular phylogeny and diversification of the "Haenydra" lineage (Hydraenidae, genus *Hydraena*), a north-Mediterranean endemic-rich group of rheophilic Coleoptera. *Molecular Phylogenetics and Evolution* 61: 772-783.
- Trizzino, M., J ch, M.A., Audisio, P., and Ribera, I. 2011. Molecular and morphological analyses confirm two new species of the *Hydraena emarginata*-saga clade (Coleoptera, Hydraenidae) from Spain and France. *Zootaxa* 2760: 29-38.

FUNDED PROJECTS

Project Title: Effectiveness of the peninsular National Park Net in the conservation of aquatic biodiversity

Financed by: Ministerio de Ciencia e Innovaci n (research program on National Parks, 023/2007).

Years: 2007-2011

PI: Andr s Mill n (Universidad de Murcia)

Project Title: Zoological Systematics and Evolution Research Group

Financed by: Generalitat de Catalunya (2009 SGR-1462)

Years: 2009-2013

PI: Salvador Carranza

Project Title: Evolution of the thermal tolerance in Pleistocene range expansions of aquatic Coleoptera from Mediterranean refugia.

Financed by: Ministerio de Ciencia e Innovaci n (CGL2010-15755).

Years: 2011-2013

PI: Ignacio Ribera

Project Title: Atlas y Libro Rojo de los Cole pteros acu ticos de Espa a [Atlas and Red Book of the Spanish Aquatic Coleoptera].

Financed by: Ministerio de Medio Ambiente

Years: 2010-2012

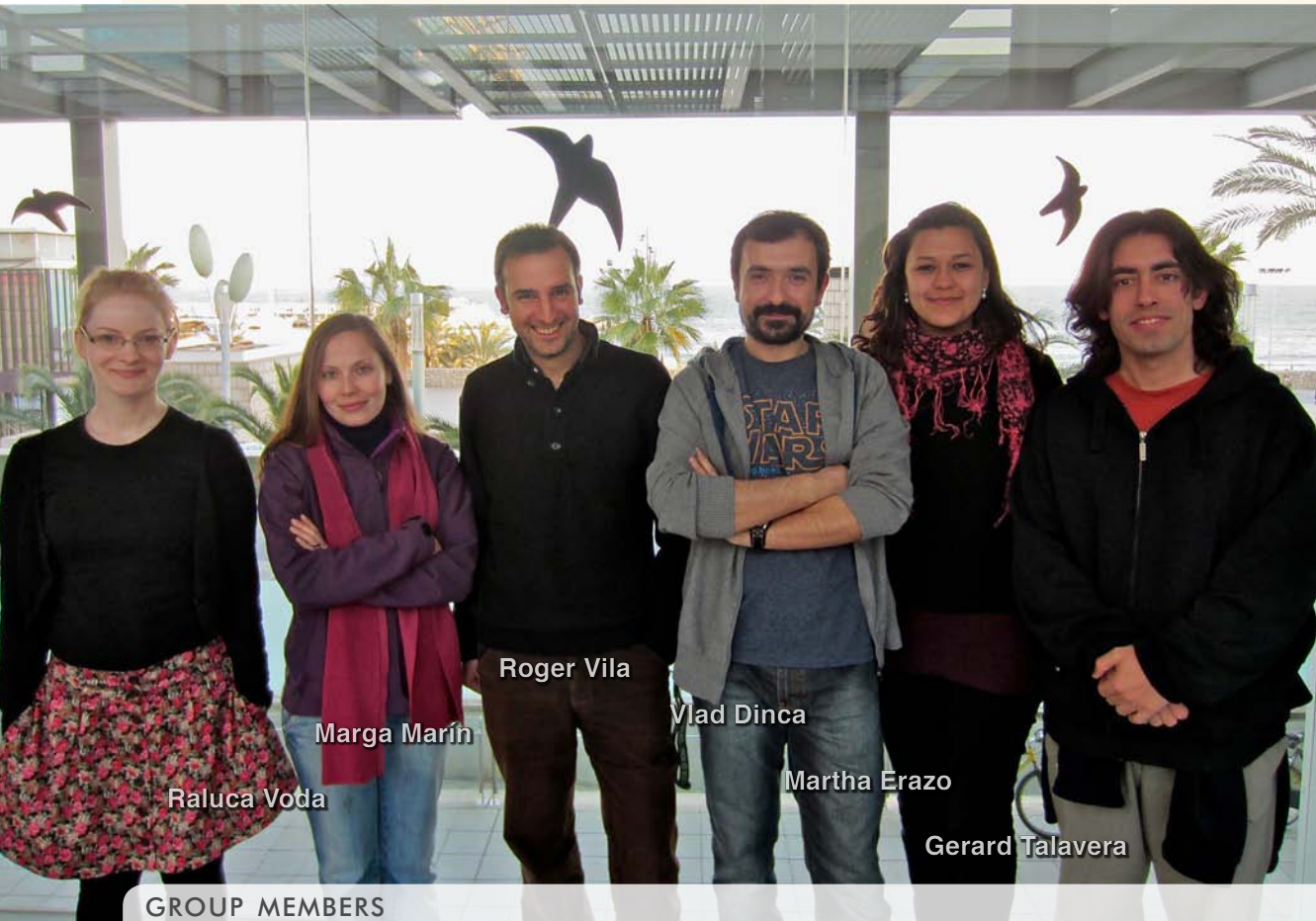
PI: Andr s Mill n (Universidad de Murcia)



Fig. 4: Cachoeira da Ferradura, in the State of Amap , Brazil, one of the places visited in search of riffle beetles (Elmidae) from the Amazon basin.

GROUP

BUTTERFLY DIVERSITY AND EVOLUTION



GROUP MEMBERS

Roger Vila, Group Leader

| ICREA Researcher



Vlad Dinca, PhD Student • Post-doc | FI Scholarship, UAB
Gerard Talavera, PhD Student | FPI Scholarship, MEC
Claudia Sañudo, PhD Student | FBBVA Project Scholarship
Raluca Voda, PhD Student | FPU Scholarship, MEC
Martha Erazo | Master Student
Marga Marín, Laboratory Technician

RESEARCH OUTLINE

We study butterfly biodiversity and evolution by integrating molecular, cytogenetic and morphological data. Our final goal is to answer general questions regarding chromosomal evolution, limits between species, and the link between phylogeography and paleoecology. When and following what route a group of tiny butterflies colonized the New World, how parasitism evolved from a friendly association between species, or if a given population constitutes a new species worth protecting are examples of questions we address.



Fig. 1: In 2011 we started a new project focusing on the diversity of butterflies in the West Mediterranean islands. They contain interesting and beautiful species, like this *Zerynthia cassandra* from Sicily (April 2011).

Image: Roger Vila.



Fig. 2: Part of the work during field expeditions consists in determining and processing the specimens collected through the day. These are *Polyommata* butterflies collected in Zambia being determined (December 2011).

Image: Roger Vila.

RESEARCH SUBLINES

1. Characterization of butterfly diversity with DNA barcoding

We are leading the implementation of DNA barcoding studies in butterflies, including the DNA barcoding of Romania (which is now the first country with all butterfly species barcoded), Iberian Peninsula and Colombia. In 2011 we started the ambitious project of obtaining a library of DNA barcodes for all the species of butterflies in the West Mediterranean. Our main goals are to test the efficiency of the method at large scale, and to develop tools based on barcoding technology to characterize diversity and phylogeography.

2. Uncovering of cryptic butterfly biodiversity in Europe

Potential cryptic species are highlighted as a result of DNA barcoding studies. We are using a wide array of techniques (nuclear and mitochondrial markers, geometric and linear morphometry, analysis of karyotype and ecological niche modelling) to deeply analyse each case, and to shed light on the origin and status of highly diverged taxa.

3. Ecological factors determining butterfly biogeography

We aim at unravelling the historical biogeography of some groups of butterflies. To do so, we combine phylogenetic methods with ecological niche modelling and paleoecological reconstruction. We are mostly interested in understanding

what ecological factors lie behind current and past distributions.

4. Chromosomal evolution in *Polyommatus* and *Leptidea*
Some butterfly groups have apparently unstable chromosomes and display unusual patterns in their karyotypes. They constitute an ideal group to study chromosomal evolution in action. We are focusing our studies on understanding the origin and evolutionary consequences of karyotype instability in *Polyommatus* and *Leptidea*.

PUBLICATIONS 2011

ISI Articles

- Dapporto, L., Schmitt, T., Vila, R., Scalercio, S., Biermann, H., Dinca, V., Gayubo, S.F., González, J.A., Lo Cascio, P., and Dennis, R.L.H. 2011. Phylogenetic island disequilibrium: evidence for ongoing long-term population dynamics in two Mediterranean butterflies. *Journal of Biogeography* 38(5): 854-867.
- Dinca, V., Dapporto, L., and Vila, R. 2011. A combined genetic-morphometric analysis unravels the complex biogeographical history of *Polyommatus icarus* and *Polyommatus celina* Common Blue butterflies. *Molecular Ecology* 20: 3921-3935.
- Dinca, V., Evgeny, E.V., Hebert, P.D.N., and Vila, R. 2011. Complete DNA barcode reference library for a country's butterfly fauna reveals high performance for temperate Europe. *Proceedings of the Royal Society Series B*. 278: 347-355.
- Dinca, V., Lukhtanov, V.A., Talavera, G., and Vila, R. 2011. Unexpected layers of cryptic diversity in wood white *Leptidea* butterflies. *Nature Communications* 2: 324.
- Hernández-Roldán, J.L., Múrria, C., Romo, H., Talavera, G., Zakharov, E., Hebert, P.D.N., and Vila, R. 2011. Tracing the origin of disjunct distributions: a case of biogeographical convergence in *Pyrgus* butterflies. *Journal of Biogeography* 38: 2006-2020.
- Lukhtanov, V.A., Dinca, V., Talavera, G., and Vila, R. 2011. Unprecedented within-species chromosome number cline in the Wood White butterfly *Leptidea sinapis* and its significance for karyotype evolution and speciation. *BMC Evolutionary Biology* 11: 109.
- Talavera, G., and Vila, R. 2011. What is the phylogenetic signal limit from mitogenomes? The reconciliation between mitochondrial and nuclear data in the Insecta class phylogeny. *BMC Evolutionary Biology* 11: 315.
- Ugelvig, L.V., Vila, R., Pierce, N.E., and Nash, D.R. 2011. A phylogenetic revision of the *Glaucoopsyche* section (Lepidoptera: Lycaenidae), with special focus on the *Phengaris-Maculinea* clade. *Molecular Phylogenetics and Evolution* 61: 237-243.
- Vila, R., Bell, C.D., Macniven, R., Goldman-Huertas, B., Ree, R.H., Marshall, C.R., Bálint, Zs., Johnson, K., Benyamini, D., and Pierce, N.E. 2011. Phylogeny and paleoecology of *Polyommatus* blue butterflies show Beringia was a climate-regulated gateway to the New World. *Proceedings of the Royal Society Series B*. 278: 2737-2744.



Fig. 3: Raluca Voda and Vlad Dinca while butterfly hunting in the Moroccan Rif Mountains. Other than nets, hats and water are key parts of the equipment (July 2011).



Fig. 4: Gerard Talavera returning to the campsite in the Altai Mountains after a day collecting species for our DNA and Tissues Butterfly Collection (May 2011).

Images: Roger Vila.

Other Publications

- Dinca, V., and Vila, R. 2011. Biodiversitat críptica i espècies falses: papallones que enganyen els científics. *Omnis Cellula* 27: 22-27.
- Dinca, V., and Vila, R. 2011. La blaveta comuna africana *Polyommatus celina*, una nova espècie a Europa. *Cynthia* 10: 16-17.
- Dinca, V., Cuvelier, S., and Mølgaard, M.S. 2011. Distribution and conservation status of *Pseudophilotes bavius* (Eversmann, 1832) (Lepidoptera: Lycaenidae) in Dobrogea (south-eastern Romania). *Phegea* 39(2): 59-67.
- Székely, L., Dinca, V., and Juhász, I. 2011. Macrolepidoptera from the steppes of Dobrogea (south-eastern Romania). *Phegea* 39(3): 85-106.

FUNDED PROJECTS

Project Title: Biodiversidad y ecología de las mariposas diurnas (Lepidoptera: Hesperioidea + Papilionoidea) de Colombia: aplicación de la técnica del código de barras genético.

Financed by: Fundación BBVA

Years: 2009-2012

PI: Roger Vila

Project Title: Estructura genética, filogenia molecular y filogeografía de un lepidóptero de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación.

Financed by: Proyecto de Investigación de Excelencia. Junta de Andalucía.

Years: 2009-2013

PI: Alberto Tinaut

Project Title: Faunal genetic comparisons to infer large-scale biogeographical patterns: the colonization of Western Mediterranean islands by butterflies.

Financed by: Ministerio de Ciencia e Innovación (CGL2010-21226/BOS)


Years: 2011-2013

PI: Roger Vila

Fig. 5: A massive concentration of *Aporia crataegi* males on salt-rich moist soil in Eastern Kazakhstan (May 2011).
Image: Roger Vila.







PROGRAM

comparative and computational genomics

RESEARCH GROUPS

Genomics of Individuality

Francesc Calafell, *Group Leader*

Paleogenomics

Carles Lalueza-Fox, *Group Leader*

Evolutionary Genomics

Arcadi Navarro, *Group Leader*

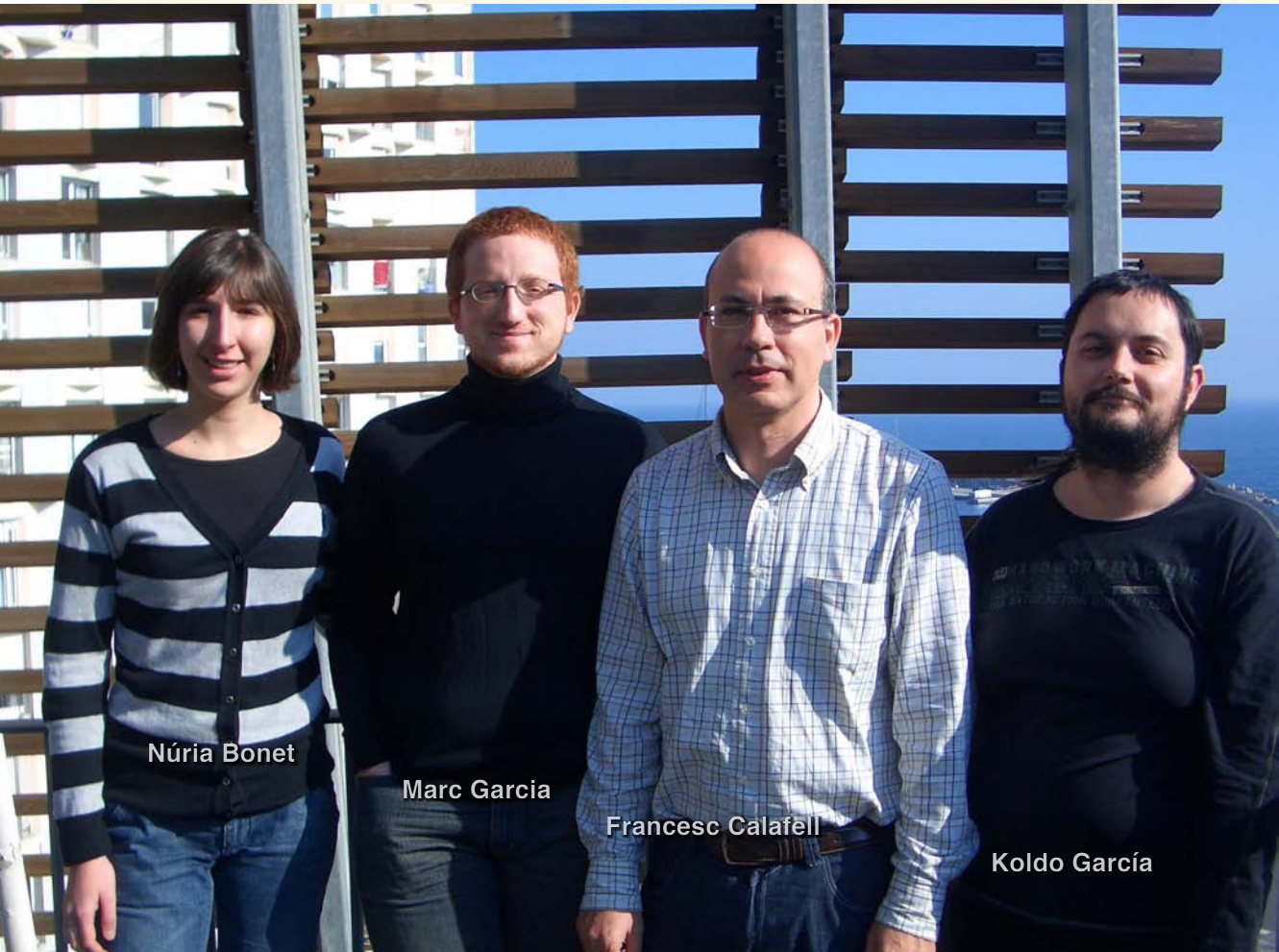
Primate Genomics

Tomàs Marquès-Bonet, *Group Leader*

In the Comparative and Computational Genomics program, genomes are compared at the intra and inter-specific levels with the general aims of understanding genome dynamics, reconstructing the evolutionary processes that generate biodiversity and linking genomic and phenotypic differences between individuals and species. To achieve these goals, we deploy both experimental and theoretical/numerical approaches, with a strong emphasis in computational techniques. Genomes contain a wealth of information, not only about how phenotypes are shaped in interaction with the environment, but also about the evolutionary history species. Thus, studying full genomes is key to answer the basic questions posed eight decades ago by the research paradigm created by the Synthetic Theory of Evolution: How much adaptation can we detect in nature? In addition to the study of adaptation, genomics allows us to answer questions about other crucial evolutionary phenomena such as speciation or the dynamics of horizontal transfer of information within the genome itself. Understanding these phenomena is fundamental to link genotypes and phenotypes, with all the implications of such knowledge in shedding light in issues such as hominization or the genetic architecture of complex phenotypes.

GROUP

GENOMICS OF INDIVIDUALITY



Núria Bonet

Marc Garcia

Francesc Calafell

Koldo García

GROUP MEMBERS

Francesc Calafell, Group Leader

| Associate Professor, UPF



Koldo García, Post-doc | CIBERESP and UPF Contracts

Marc Garcia, PhD Student | FU Scholarship, MEC

RESEARCH OUTLINE

The general topics that interest us revolve around the genomics of individuality: what is there in our genomes that makes us the way we are? What does it tell about our ancestry? How does it affect our susceptibility to diseases? How can this be applied in practical settings, i.e., in forensic genetics? This is implemented in practice in four main projects: 1) we are trying to detect past recombination events in current sequences and use them as phylogenetic markers; 2) we are characterizing the skin microbiome in healthy and psoriatic skin to reveal whether psoriasis has a microbial trigger; 3) we are working in a case-control association study to detect any host genetic determinant of a poor progression in 2009 A(H1N1) influenza, and 4) we have initiated a project to investigate Y-chromosome genetic diversity within samples of men carrying the same surname.

RESEARCH SUBLINES

1. Human populations genetics and recombination

Recombination is a problem often cited to shun autosomal genetic diversity when reconstructing population history, which is mostly carried out with the non-recombining portion of the Y chromosome and mitochondrial DNA. We are trying to turn this argument around and using recombination events as phylogeographic markers. We are working with Laxmi Parida and Asif Javed to develop software that will detect chromosomes that are descendants from recombination events. This information can be recoded into recotypes and analyzed in the same way as haplotypes are. We expect to add a new tool to analyze autosomal diversity in population genetics, but it could also be applied to detect natural selection and to understand recombination itself. This is basically the work of Marta Melé, and is carried out in collaboration with Jaume Bertranpetit.

2. The skin microbial biota in health and disease

How the human body works cannot be understood without its relationship with its associated bacterial and viral flora. Human genomic diversity can be extended to encompass the genomic diversity of the microbes living with us. We are studying the skin microbial flora to try to comprehend the skin as a complex ecosystem. We are characterizing bacterial and retroviral diversity, as well as performing bacterial metagenomics in samples from healthy individuals. We seek to establish a baseline and detect how it is affected in individuals with skin conditions such as psoriasis. Mireia Coscollà, Koldo García, and Marc Garcia work or have worked in this project, in collaboration with Marta Ferran at Hospital del Mar.

3. Genetic susceptibility factors in poor influenza progression

Little is known about the possible genetic susceptibility factors for infectious diseases beyond some classical examples in malaria. Taking the opportunity created by the 2009 A(H1N1) influenza pandemic, we are collecting confirmed influenza cases that required hospitalization and had no obvious risk factors, and comparing them with milder cases. By genotyping a whole genome array, we hope to discover genetic susceptibility factors for poor progression in influenza. The case and control collection is part of a much wider project led by Ángela Domínguez (UB), and we are collaborating with Fernando González-Candelas (UV).

4. A genetic atlas of Catalan surnames

Given their transmission, surnames behave as alleles at a locus in the Y chromosome, but they also carry linguistic, social, and historic information. We have selected a list of 50 Catalan surnames and intend to gather a sample of 50 men for each of those surnames. We will type STRs and UEPs in those samples, and we want to answer these questions: 1) How are surname frequency and genetic diversity related? The frequency of a surname may be the result of polyphyletism, namely, the fact that it may have been founded multiple times (think of *Smith* or *Jones*, John's son); in that case, surname frequency and its internal genetic diversity should be positively correlated. Alternatively, certain surnames may have become more common by natural selection: surnames may be markers of social status, which, quite often, determined survival and fertility. 2) Were the carriers of German patronymic surnames of a different genetic origin from the rest of the population? In Catalonia, as in France, a frequent source of surnames are former first names of Germanic origin (Albert, Robert, Grau...). We will compare some of those to patronymic surnames of Latin origin (Oriol, Pons...). 3) Is that also the case for ethnonym surnames? Some Catalan surnames (Alemany, Danés, Anglès, Guasch) denote geographic origin (they mean German, Dane, English, Gascon, respectively). Such an origin can be traced as long as the Y gene pools of the region of origin and of Catalonia are different enough.

This project is a collaboration with David Comas and Jaume Bertranpetit (IBE). A genetic study of the Colom and Colombo surnames has been undertaken with Francesc Albardaner (Centre d'Estudis Colombins), José Antonio Lorente (Universidad de Granada), and Cristina Martínez-Labarga (Università degli Studi di Roma "Tor Vergata").

PUBLICATIONS 2011

ISI Articles

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- Bertinetto, F.E., Calafell, F., Roggero, S., Chidichimo, R., Garino, E., Marcuccio, C., Coppo, R., Scolari, F., Frascá, G.M., Savoldi, G., Schena, and Amoroso, A. on behalf of the European IgA nephropathy Consortium. 2011. Search for genetic association between IgA nephropathy and candidate genes selected by function or by gene mapping at loci IGAN2 and IGAN3. *Nephrology Dialysis Transplantation* 0: 1-10. Ahead of print; doi: 10.1093/ndt/gfr633.
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- Javed, A., Pybus, M., Melé, M., Utro, F., Bertranpetit, J., Calafell, F., and Parida, L. 2011. IRIS*: Construction of ARG networks at genomic scales. *Bioinformatics* 27(17): 2448-2450.
- Laayouni, H., Montanucci, L., Sikora, M., Melé, M., Dall'Olio, G.M., Lorente-Galdós, B., McGee, K.M., Graffelman, J., Awadalla, P., Bosch, E., Comas, D., Navarro, A., Calafell, F., Casals, F., and Bertranpetit, J. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS ONE* 6(3): e17913.
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- Sikora, M., Laayouni, H., Calafell, F., Comas, D., and Bertranpetit, J. 2011. A genomic analysis identifies a novel component in the genetic structure of sub-Saharan African populations. *European Journal of Human Genetics* 19(1): 84-88.

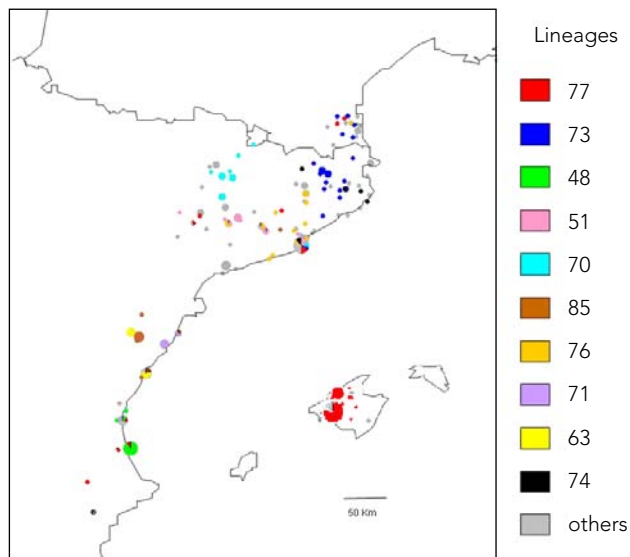


Fig. 1: Y-chromosome genetic lineages in men bearing the Colom surname. Lineages indicate descent from a common recent ancestor, probably a founder of the surname. Each lineage is indicated with an arbitrary number. Circle size is proportional to the number of men sampled in that locality.

FUNDED PROJECTS

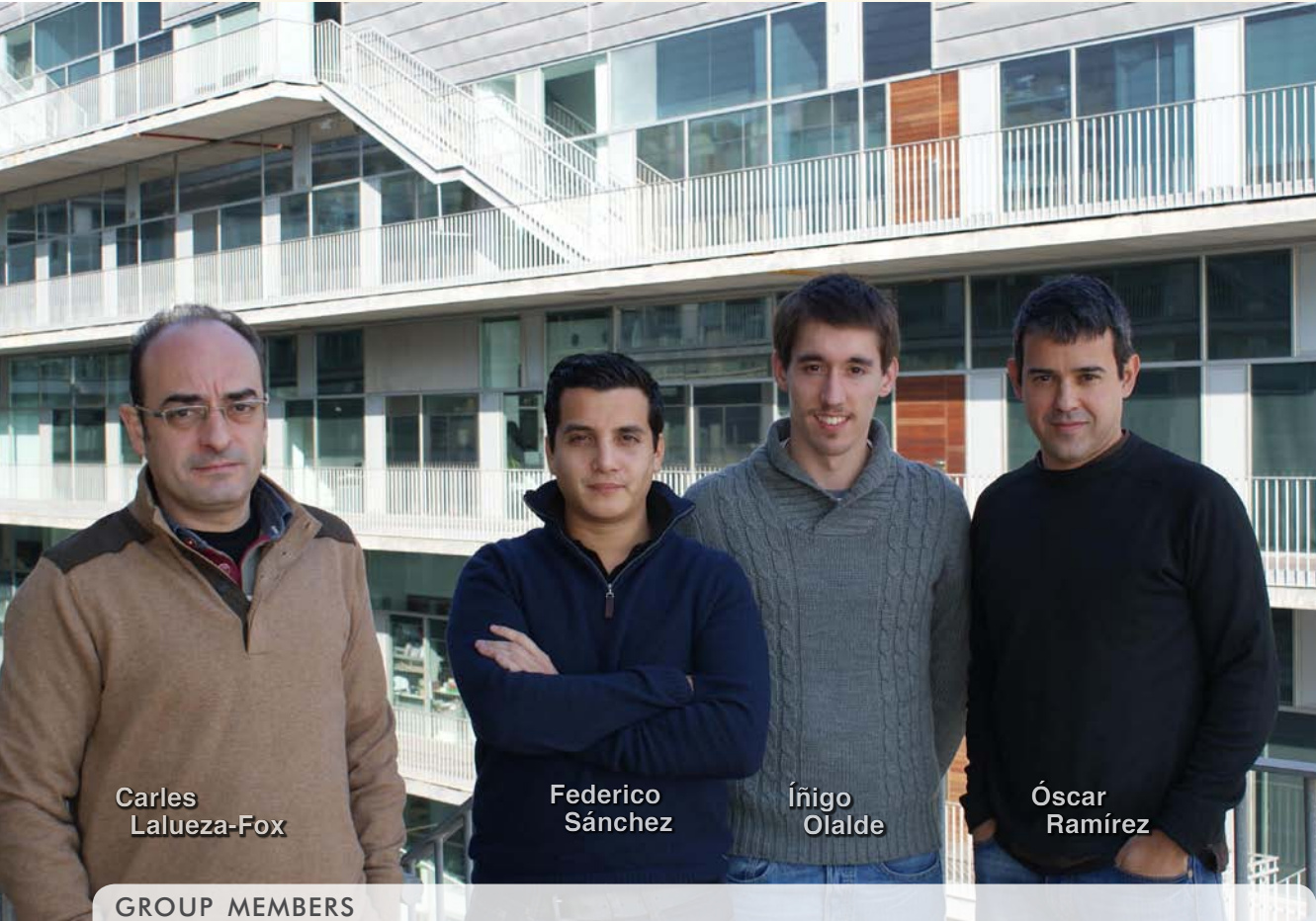
Project Title: Dinámica de la Recombinación en el genoma humano
Financed by: Ministerio de Ciencia e Innovación (REF: BFU2007-63657)
Years: 2008-2011
PI: Francesc Calafell

Project Title: Metagenomas procariotas de la piel humana sana y con psoriasis
Financed by: Ministerio de Ciencia e Innovación (SAF2010-16240)
Year: 2011
PI: Francesc Calafell

Project Title: Grup de Recerca Consolidat-SGR
Financed by: Generalitat de Catalunya (2009 SGR-1101)
Years: 2009-2013
PI: Jaume Bertranpetit

GROUP

PALEOGENOMICS



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Óscar Ramírez, Visitant Professor, UPF

Elena Gigli, PhD Student | FPI Scholarship, MICINN

Federico Sánchez, PhD Student | FPI Scholarship, MICINN

Íñigo Olalde, Master Student, La Caixa

RESEARCH OUTLINE

Our research group focuses on paleogenomics, the study of structure, function and organization of ancestral genomes. We are interested in different evolutionary problems that can be answered with ancient DNA data, involving human evolution, population dynamics and diversity, phylogenetics, phylogeography and adaptive processes. We work with different species and populations, including mammoths, cave bears, *Myotragus balearicus*, wolves and lynxs, but also with an extinct hominin species, the Neandertals. In our group we are basically interested in the genomic diversity among Neandertals, in the individualisation of a Neandertal family group from El Sidron site (Asturias, Spain) and in investigating the functional implications of genetic variants exclusively found in Neandertals.

RESEARCH SUBLINES

1. Adaptive traits and evolutionary history of Neandertals

We are currently retrieving genomic regions that are incompletely covered by the current genome draft. We are also conducting *in vitro* and *in vivo* functional studies related to genes that are known to be different between Neandertals and modern humans for trying to interpret the phenotypical consequences of these genomic differences.

2. Neandertal genomic diversity

We are analyzing different individuals from El Sidron site in Asturias, Spain. This is a family group of at least 12 Neandertal individuals that became accidentally accumulated in a single, synchronic event within a subterranean karstic system. El Sidron offers the unique opportunity of launching a genomic project for understanding the diversity and the kinship relationships within a Neandertal family group.



Fig. 1: A Neandertal bone from El Sidron site (Asturias), sampled for DNA analysis.

3. Phylogenetics, phylogeography and adaptation in extinct and living species

We are studying different extinct species, to answer specific questions about their phylogeny, adaptation and evolution. We are particularly interested in studying some insular endemics and the genomic basis of some common adaptive patterns observed among them such as body size reduction. We are also interested in the analysis of domestication processes at the genomic level, and we are currently comparing structural variations (CNVs) between dogs and wolves.

4. Extinct modern human populations

We are genetically analysing ancient samples from prehistoric European populations to reconstruct past human migrations. We are trying to develop new methodological tools for capturing targeted genomic regions and complete mitochondrial genomes combined with massively parallel sequencing technologies. We are specially interested in the Mesolithic-Neolithic shift.



Fig. 2: The Mesolithic skeletal remains found at La Braña-Arintero site in León. Image: Courtesy of J. Vidal.

PUBLICATIONS 2011

ISI Articles

- Ebenesersdóttir, S.S., Sigurðsson, A., Sánchez-Quinto, F., Lalueza-Fox, C., Stefánsson, K., and Helgason, A. 2011. *American Journal of Physical Anthropology* 144(1): 92-99.
- García-Garcerà, M., Gigli, E., Sánchez-Quinto, F., Ramírez, O., Calafell, F., Civil, S., and Lalueza-Fox, C. 2011. Fragmentation of contaminant and endogenous DNA in ancient samples determined by shotgun sequencing; prospects for human palaeogenomics. *PLoS ONE* 6(8): e24161.
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- Lalueza-Fox, C., Rosas, A., Estalrich, A., Gigli, E., Campos, P.F., García-Tabernero, A., García-Vargas, S., Sánchez-Quinto, F., Ramírez, O., Civit, S., Bastir, M., Huguet, R., Santamaría, D., Gilbert, M.T.P., Willerslev, E., and de la Rasilla, M. 2011. Genetic evidence for patrilocal mating behavior among Neandertal groups. *Proceedings of the National Academy of Sciences USA* 108(1): 250-253.
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- Rodríguez, R., Ramírez, O., Valdiosera, C.E., García, N., Alda, F., Madurell-Malapeira, J., Marmi, J., Doadrio, I., Willerslev, E., Götherström, A., Arsuaga, J.L., Thomas, M.G., Lalueza-Fox, C., and Dalén, L. 2011. 50,000 years of genetic uniformity in the critically endangered Iberian lynx. *Molecular Ecology* 20(18): 3785-3795.
- Sastre, N., Vilà, C., Salinas, M., Bologov, V.V., Urios, V., Sánchez, A., Francino, O., and Ramírez, O. 2011. Signatures of demographic bottlenecks in European wolf populations. *Conservation Genetics* 12: 701-712.

Book Chapters

- Lalueza-Fox, C. 2011. Desvelando el más íntimo código: los estudios paleogenéticos. En: La cueva de El Sidrón (Borines, Piloña, Asturias). De la Rasilla, M., Rosas, A., Cañaveras, J.C., and Lalueza, C. Eds.

FUNDED PROJECTS

Project Title: Neandertal genome diversity analyzed by ultrasequencing techniques (REF: BFU2009-06974)

Financed by: Ministerio de Ciencia e Innovación

Years: 2010-2012

PI: Carles Lalueza-Fox

Project Title: Grup de Recerca en Biologia Evolutiva (2009SGR1101)

Financed by: Direcció General de Recerca, Comissionat per a Universitats i Recerca, Generalitat de Catalunya

Years: 2009-2012

PI: Jaume Bertranpetit

Fig. 3: Pyrographically decorated gourd that presumably contains a handkerchief dipped in the blood of King Louis XVI of France after his beheading.



GROUP
EVOLUTIONARY GENOMICS



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| Professor, UPF • Research Professor, ICREA



- Natalia Petit, Post-doc | JAE Contract
- Gabriel Santpere, Post-doc | Project Contract
- Rui Faria, Post-doc | FCT Fellowship
- Belén Lorente, PhD Student | UPF Contract
- Urko Martínez, PhD Student | UPF Scholarship
- Diego Hartasánchez, PhD Student | JAE Contract
- Oriol Vallès, Master Student | Project Contract
- Carlos Morcillo, Project Manager | INB (National Bioinformatics Institute)
- Ángel Carreño, IT Technician | INB (National Bioinformatics Institute)
- Txema Heredia, IT Technician | PTA-MICINN
- Fernando Muñiz, IT Technician | INB (National Bioinformatics Institute)
- Jordi Rambla, IT Technician | INB (National Bioinformatics Institute)

RESEARCH OUTLINE

Life as we see it in our planet today has been shaped by many different biological processes during billions of years. These processes leave a signature in our genomes in the form of differences between species, or between individuals of the same species. Interrogating these patterns of genome diversity we can infer what are the forces that affect living organisms, how and when they act, and how do they affect such various things as biodiversity, human emotions or the differential susceptibility of different people to certain diseases. All this knowledge empowers us to control our future but, above all, it is fun to obtain. In practical terms, there are two main kinds of research we do in our group. First, we develop and analyze models that help to answer questions concerning chromosomal evolution, genome dynamics, speciation, comparative gene expression and the genetic architecture of complex human traits (including disease). The tools we use for these research lines include analytical and simulation studies, together with data mining and analysis. Second, we also carry out wet-lab research. Lately, we have focused on array CGH of primate genomes to study the evolution of copy-number variation and Genome-Wide Association Studies of human socio-economic traits.

RESEARCH SUBLINES

1. Chromosomal evolution and speciation

We study how large chromosomal rearrangements affect many aspects of genome structure and evolution, including how they may drive the generation of new species.

2. Segmental duplications and copy-number variation in primates

The genomes of humans and other primates show enrichment in Segmental Duplications (SDs) with high sequence identity. SDs are fundamental for the creation of novel genes and may have been key in the evolution of our lineage. We try to understand the dynamics of the molecular content of SDs.

3. Detecting positive selection in the human lineage

We try to detect the signature of adaptive changes out the usual paradigm of new mutations that arise in single-copy protein-coding regions. Recently we have been focusing on three questions: how natural selection may have shaped regulatory regions and the functional content of SDs, how natural selection has acted upon introns; and how prevalent epistatic selection (or selection upon multiple targets) has been.

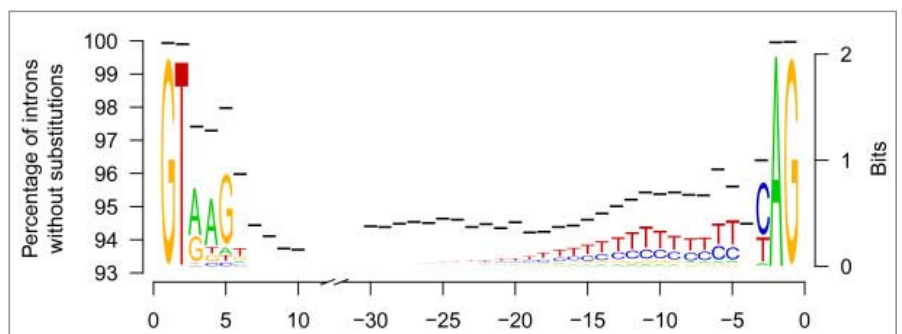


Fig. 1: Interspecific comparison of Copy Number Variation Regions (CNVR). For each species, the picture represents the proportion of its CNVRs shared with none, one, two or three other species.

4. World-wide distribution of human disease

We study world-wide patterns of disease susceptibility distribution to ascertain how these may have been influenced by recent human evolution.

5. Genoeconomics

Complex human traits that are exclusive of our lineage are the basis of our societies and have huge socio-economic impact. We deploy the latest tools of genomics for the dissection of human economic traits.

PUBLICATIONS 2011

ISI Articles

- Gazave, E., Darré, F., Morcillo-Suárez, C., Petit-Marty, N., Carreño, A., Marigorta, U.M., Ryder, O.A., Blancher, A., Rocchi, M., Bosch, E., Baker, C., Marquès-Bonet, T., Eichler, E.E., and Navarro, A. 2011. Copy number variation analysis in the great apes reveals species-specific patterns of structural variation. *Genome Research* 21(10): 1626-1639.
- Laayouni, H., Montanucci, L., Sikora, M., Melé, M., Dall'Olio, G.M., Lorente-Galdós, B., McGee, K.M., Graffelman, J., Awadalla, P., Bosch, E., Comas, D., Navarro, A., Calafell, F., Casals, F., and Bertranpetit, J. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS ONE* 6(3): e17913.
- Locke, D., LaDeana, W., Warren, W., Worley, K., Nazareth, L., Muzny, D., Yang, S., Wang, Z., Chinwalla, A., Minx, P., Mitreva, M., Cook, L., Delehaunty, K., Fronick, C., Schmidt, H., Fulton, L., Fulton, R., Nelson, J., Magrini, V., Pohl, C., Graves, T., Markovic, C., Cree, A., Dinh, H., Hume, J., Kovar, C., Fowler, G., Lunter, G., Meader, S., Heger, A., Ponting, C., Marquès-Bonet, T., Alkan, C., Chen, L., Cheng, Z., Kidd, J., Eichler, E.E., White, S., Searle, S., Vilella, A., Chen, Y., Flicek, P., Ma, J., Raney, B., Suh, B., Burhans, R., Herrero, J., Haussler, D., Faria, R., Fernando, O., Darré, F., Farré, D., Gazave, E., Oliva, M., Navarro, A., Roberto, R., Capozzi, O., Archidiacono, N., Della Valle, G., Purgato, S., Rocchi, M., Konkel, M., Walker, J., Ullmer, B., Batzer, M., Smit, A., Hubley, R., Casola, C., Schrider, D., Hahn, M., Quesada, V., Puente, X., Ordóñez, G., López-Otín, C., Vinar, T., Brejova, B., Ratan, A., Harris, R., Miller, W., Kosiol, C., Lawson, H., Taliwal, V., Martins, A., Siepel, A., Roychoudhury, A., Ma, X., Degenhardt, J., Bustamante, C., Gutenkunst, R., Mailund, T., Dutheil, J., Hobolth, A., Schierup, M., Ryder, O., Yoshinaga, Y., de Jong, P., Weinstock, G., Rogers, J., Mardis, E., Gibbs, R., and Wilson, R. 2011. Comparative and demographic analysis of orangutan genomes. *Nature* 469: 529-533.
- Malhotra, S., Morcillo-Suárez, C., Brassat, D., Goertsches, R., Lechner-Scott, J., Urcelay, E., Fernández, O., Drulovic, J., García-Merino, A., Martinelli Boneschi, F., Chan, A., Vandenbroeck, K., Navarro, A., Bustamante, M.F., Rio, J., Oksenberg, J., Montalban, X., and Comabella, M. 2011. IL28B polymorphisms are not associated with the response to interferon-beta in multiple sclerosis. *Journal of Neuroimmunology* 239: 101-104.
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- Ventura, M., Catacchio, C., Alkan, C., Marquès-Bonet, T., Sajjadian, S., Graves, T., Hormozdiari, F., Navarro, A., Malig, M., Baker, C., Lee, C., Turner, E., Chen, L., Kidd, J.,

Archidiacono, N., Shendure, J., Wilson, R.K., and Eichler, E.E. 2011. Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Research* 21(10): 1640-1649.

FUNDED PROJECTS

Project Title: Asociación entre los polimorfismos de los genes SLC6A4, DRD2 y COMT y la regulación emocional y el control cognoscitivo en la depresión infantil y juvenil

Financed by: CSIC-CRUSA (2009CR0028)

Years: 2010-2011

PI: Arcadi Navarro

Project Title: Identifying Evolutionary Novelties and Adaptation in Duplicated Regions of the Genomes of Primates

Financed by: Ministerio de Educación y Ciencia (BFU2009-13409-C02-02)

Years: 2009-2012

PI: Arcadi Navarro

Project Title: Exploring the behavioral genetics of Trade and Cooperation

Financed by: Ministerio de Educación y Ciencia (MEC-SEJ2007-30267-E/SOCI)

Years: 2008-2011

PI: Arcadi Navarro

Project Title: INB GN8

Financed by: Genoma España (Instituto Nacional de Bioinformática)

Years: 2003-2011

PI: Arcadi Navarro

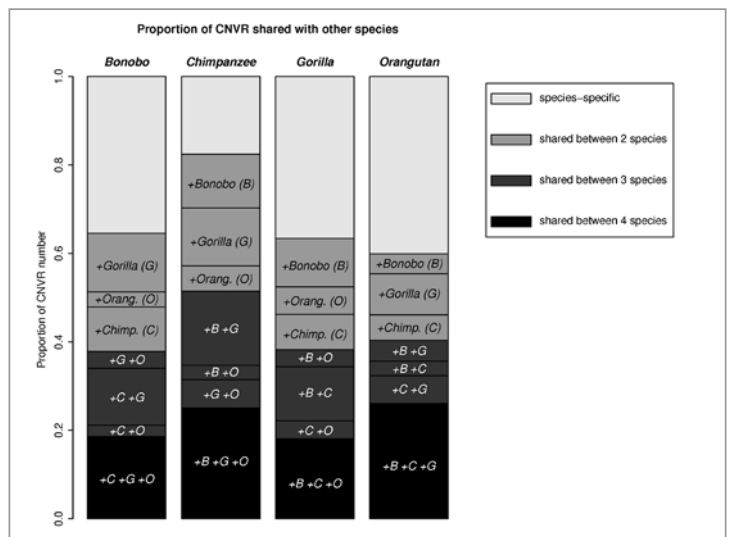
Project Title: Red Española de Esclerosis Múltiple

Financed by: Ministerio de Ciencia e Innovación. Instituto Carlos III (ISCIII-RD07/0060/2021)

Years: 2009-2011

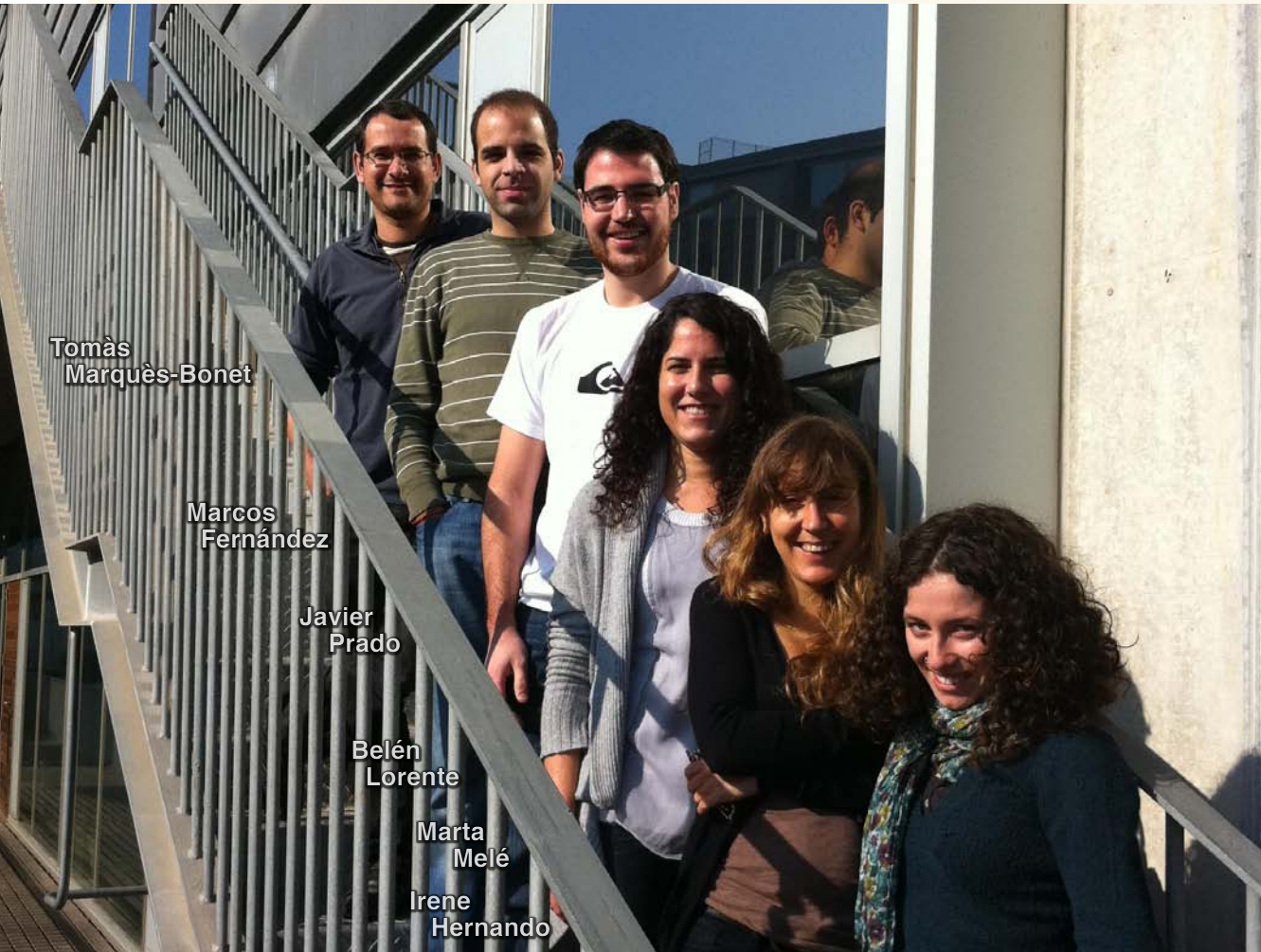
PI: Arcadi Navarro

Fig. 2: Conserved motifs in the extremes of human introns and sequence conservation in human, chimpanzee and macaque. The total height of each stack of letters corresponds to the amount of information at that position measured in bits (y-axis on the right). Within each stack letters are sorted so that the most frequent appear on top, and their height within the stack is proportional to their relative frequency. Black dashes mark the percentage (y-axis on the left) of introns with the same nucleotide in the three species (regardless of what the actual base, A, C, T or G is) in the first ten and last 30 nucleotides of introns.



GROUP

PRIMATE GENOMICS



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Marc Dabad, Computational Support | ERC StGt Project Contract

RESEARCH OUTLINE

Our main line of research is centered in the discovery of the extent of all kinds of genome variation within the great ape species. The goal is to create an integrated view of genome evolution by studying changes in the composition, frequency, size and location at every major branch point of human divergence from other primates. 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.

RESEARCH SUBLINES

1. Genomic variation in ape genomes

Despite international efforts to characterize thousands 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. Most genome projects include only one individual as a reference, but in order to understand the impact of structural variants in the evolution of every species we need to re-sequence multiple individuals of each species. We can only understand the origins of genomic variants and phenotypical differences among species if we can model variation within species and compare it to a proper perspective with the differences among species.

2. Snowflake genome. Study of albinism in gorillas.

As a part of the pilot project for the ERC Starting Grant, we have sequenced Snowflake, the only known albino gorilla to perform comparative genomic approaches, both inter and intra-species, to study mutations that could explain the phenotype in this unique gorilla. We have found the mutation causing this unique phenotype and we have used genomic tools to unravel unusual inbreeding behavior in the family of Snowflake.

3. Epigenetics and transcriptomics of non-human primates

The recognition of post-genomic modifications with high biological impact has been a focus of research in model and non-model organisms in the last years. However, little has been done combining a three way analysis going from genomic variants, to gene expression and epigenetics in non-human primates. In the next years we are planning to use different tissues from the same individual comparing human, chimps and rhesus macaque to explore the relationship of these three layers of complexity.

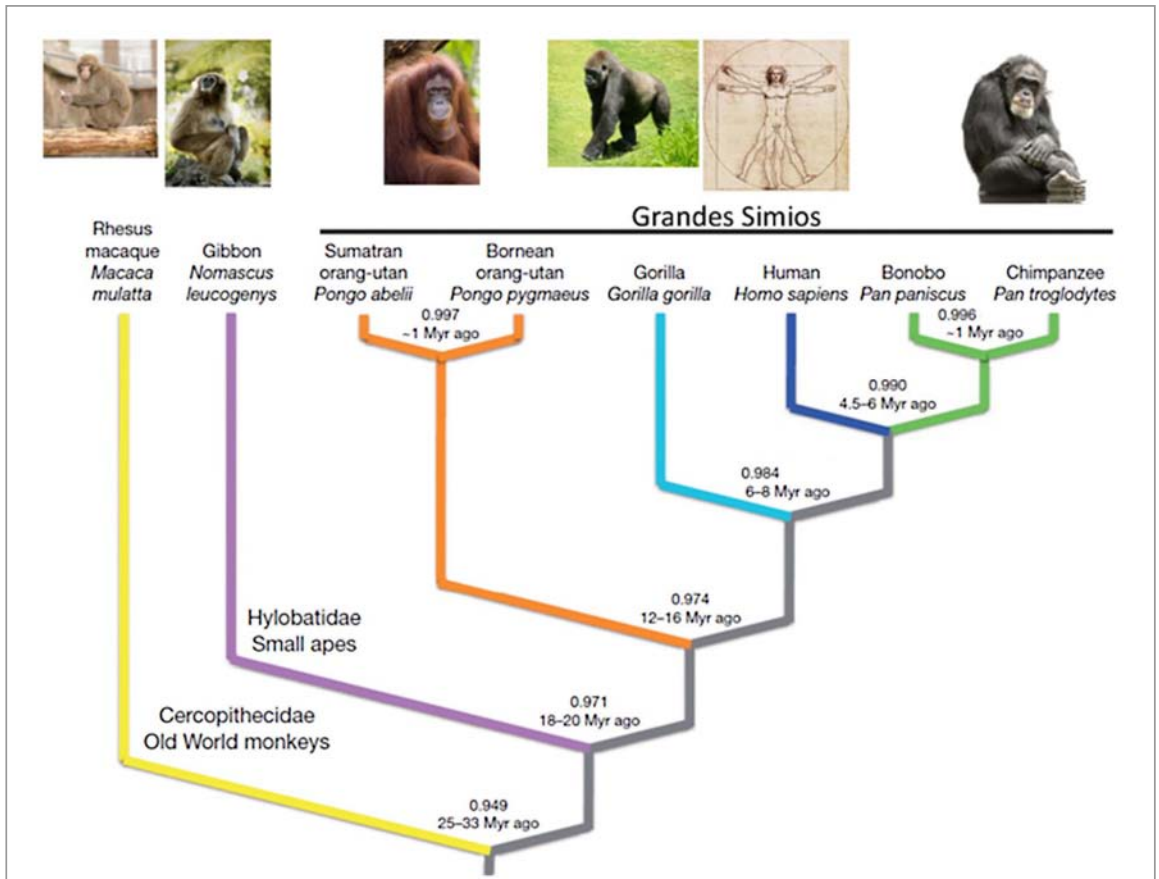


Fig. 1: Divergence time and genomic variability between the main species of Great Apes. From Locke et al. *Nature* 2011.

PUBLICATIONS 2011

ISI Articles

- Gazave, E., Darré, F., Morcillo-Suárez, C., Petit-Marty, N., Carreño, A., Marigorta, U.M., Ryder, O.A., Blancher, A., Rocchi, M., Bosch, E., Baker, C., Marquès-Bonet, T., Eichler, E.E., and Navarro, A. 2011. Copy number variation analysis in the great apes reveals species-specific patterns of structural variation. *Genome Research* 21(10): 1626-1639.
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- Locke, D., LaDeana, W., Warren, W., Worley, K., Nazareth, L., Muzny, D., Yang, S., Wang, Z., Chinwalla, A., Minx, P., Mitreva, M., Cook, L., Delehaunty, K., Fronick, C., Schmidt, H., Fulton, L., Fulton, R., Nelson, J., Magrini, V., Pohl, C., Graves, T., Markovic, C., Cree, A., Dinh, H., Hume, J., Kovar, C., Fowler, G., Lunter, G., Meader, S., Heger, A., Ponting, C., Marquès-Bonet, T., Alkan, C., Chen, L., Cheng, Z., Kidd, J., Eichler, E.E., White, S., Searle, S., Vilella, A., Chen, Y., Flicek, P., Ma, J., Raney, B., Suh, B., Burhans, R.,

Herrero, J., Haussler, D., Faria, R., Fernando, O., Darré, F., Farré, D., Gazave, E., Oliva, M., Navarro, A., Roberto, R., Capozzi, O., Archidiacono, N., Della Valle, G., Purgato, S., Rocchi, M., Konkel, M., Walker, J., Ullmer, B., Batzer, M., Smit, A., Hubley, R., Casola, C., Schrider, D., Hahn, M., Quesada, V., Puente, X., Ordóñez, G., López-Otín, C., Vinar, T., Brejova, B., Ratan, A., Harris, R., Miller, W., Kosiol, C., Lawson, H., Taliwal, V., Martins, A., Siepel, A., Roychoudhury, A., Ma, X., Degenhardt, J., Bustamante, C., Gutenkunst, R., Mailund, T., Dutheil, J., Hobolth, A., Schierup, M., Ryder, O., Yoshinaga, Y., de Jong, P., Weinstock, G., Rogers, J., Mardis, E., Gibbs, R., and Wilson, R. 2011. Comparative and demographic analysis of orangutan genomes. *Nature* 469: 529-533.

- Ventura, M., Catacchio, C., Alkan, C., Marquès-Bonet, T., Sajjadian, S., Graves, T., Hormozdiari, F., Navarro, A., Malig, M., Baker, C., Lee, C., Turner, E., Chen, L., Kidd, J., Archidiacono, N., Shendure, J., Wilson, R.K., and Eichler, E.E. 2011. Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Research* 21(10): 1640-1649.

FUNDED PROJECTS

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

Financed by: European Research Council

Years: 2010-2014

PI: Tomàs Marquès-Bonet

Project Title: Characterization of inversions and changes of gene expression in the great-ape evolution (BFU2011-28549)

Financed by: Ministerio de Ciencia e Innovación (Spain)

Years: 2011-2013

PI: Tomàs Marquès-Bonet



PROGRAM

functional evolution in insects

RESEARCH GROUPS

Evolution of Insect Metamorphosis

Xavier Bellés, *Group Leader*

Insect Reproduction

Maria-Dolors Piulachs, *Group Leader*

Nutritional Signals in Insects

José Luis Maestro, *Group Leader*

Hormonal Control of Insect Development

David Martín, *Group Leader*

Drosophila Telomeres

Elena Casacuberta, *Group Leader*

Developmental Biology and Morphological Evolution in Insects

Xavier Franch-Marro, *Group Leader*

Evolutionary and Functional Genomics

Josefa González, *Group Leader*

This program aims at studying fundamental processes in animal life –such as metamorphosis, reproduction, development, cell division or telomere regulation or adaptation to new environments– from a comparative perspective and in an evolutionary frame. Studies are focused on invertebrates, especially in insects, and they are largely based on functional genomics approaches. Until now, the models used have been insects, in particular cockroaches (*Blattella*), beetles (*Tribolium*) and flies (*Drosophila*), and works have been directed to study processes like molting, oogenesis, growth or telomere replication. In most cases, the direct or indirect regulation of these processes, either by nutritional signals, hormones, transcription factors, or by microRNAs, has been also an important subject of study. The methodologies used covered practically all scales, from morphological to molecular. During last years the use of RNA interference (RNAi) techniques allowed the study of gene functions in non-model species, which very often are not easily transformable from a genetic point of view. In these cases, RNAi offers a unique way to face functional genomics on these species. Briefly, the Functional Evolution of Insects Program combines gene sequence analysis and experimental approaches to unveil gene functions, with the aim of understanding the evolution of biological processes in insects. In the context of the general project of the IBE, the functional evolution program provides the tools of experimental biology, which are of paramount importance to understand the adaptive mechanisms of evolution.



GROUP

EVOLUTION OF INSECT METAMORPHOSIS



Raúl
Montañez

Mercedes
Rubio

Jesús
Lozano

Xavier
Bellés

Alba
Herráiz

Aníbal
de Horna

GROUP MEMBERS

Xavier Bellés, Group Leader

| Research Professor, CSIC



Raúl Montañez, Post-doc | CSIC Contract (JAE Program)

Mercedes Rubio, PhD Student | Scholarship CSIC (JAE Program)

Alba Herráiz, PhD Student | JAEPRE-CSIC Contract - LINCGlobal

Jesús Lozano, PhD Student | Scholarship MICINN

Aníbal de Horna, Bioinformatics Support | CSIC Project Contract

RESEARCH OUTLINE

Our interests focus on insect metamorphosis, and we approach its study from a mechanistic point of view and from an evolutionary perspective. As most information has been obtained in highly modified, holometabolan species (mainly in the fly *Drosophila melanogaster*, but also in the beetle *Tribolium castaneum*), we concentrate on the cockroach *Blattella germanica*, a phylogenetically basal, hemimetabolan species that can be a reasonable surrogate of the ancestral gradual metamorphosis. We aim to elucidate the mechanisms regulating metamorphosis in *B. germanica* and then comparing them with those operating in holometabolan species. The idea is to infer the evolutionary history underlying the transition from hemimetaboly to holometaboly.

RESEARCH SUBLINES

1. Minimal models of metamorphosis

To delimit the problem we concentrate in two metamorphic processes: i) the formation of the wing, which is important even to understand the origin of metamorphosis, and ii) the formation of the male tergal gland, which is a complex morphologic structure that abruptly forms after the adult molt in the tergites 7 and 8 of males. The study of these two processes is being approached not only by monitoring the morphological changes, but also at molecular level, by comparing transcriptomes of the respective tissues in metamorphic and non-metamorphic transitions.

2. Transcription factors and hormonal signaling

Metamorphosis is mainly regulated by two hormone types: ecdysteroids and juvenile hormones. We are interested in the transcription factors involved in the signaling pathways elicited by both hormone types, considering that there are not two separate pathways, but an intricate network of interaction between JH- and ecdysone-associated factors. We use RNAi techniques as the main tool to unveil the functions of the transcription factors under study.

3. Small RNAs

We investigate the regulatory role of miRNAs in metamorphosis under the hypothesis that miRNAs play a crucial role in the shift from juvenile to adult developmental programs. Work is not only descriptive but also functional, using anti-miR molecules and miR mimics to reveal the functions of the miRNAs under study. Collaterally, we are also interested in investigating the biochemical machinery involved in the RNAi process and in miRNA biogenesis.

4. Complex networks

Metamorphosis involves complex networks of gene regulation, and the idea is to reduce this complexity to graphs capturing the main properties of these networks. Then, we investigate their topological properties in metamorphic and non-metamorphic transitions. Then, we can infer regulatory mechanisms and validate them experimentally. At present, we focus on networks of interaction mRNA-miRNA comparing metamorphic and non-metamorphic transitions in holometabolan and hemimetabolan models.

PUBLICATIONS 2011

ISI Articles

- Clynen, E., Ciudad, L., Bellés, X., and Piulachs, M.D. 2011. Conservation of fruitless role as master regulator of male courtship behaviour from cockroaches to flies. *Development, Genes and Evolution* 221: 43-48.
- Cristino, A.S., Tanaka, E.D., Rubio, M., Piulachs, M.D., and Bellés, X. 2011. Deep sequencing of organ- and stage-specific microRNAs in the evolutionarily basal insect *Blattella germanica* (L.). *PLoS ONE* 6(4): e19350. doi: 10.1371 / journal.pone.0019350.
- Folguera, G., Bastías, D.A., Caers, J., Rojas, J.M., Piulachs, M.D., Belles, X., and Bozinovic, F. 2011. An experimental test of the role of environmental temperature variability on ectotherm molecular, physiological and life-history traits: implications for global warming. *Comparative Biochemistry and Physiology Part A* 159: 242-246.
- Herraiz, A., Chauvigné, F., Cerdà, J., Bellés, X., and Piulachs, M.D. 2011. Identification and functional characterization of an ovarian aquaporin from the cockroach *Blattella germanica* (L.) (Dictyoptera, Blattellidae). *Journal of Experimental Biology* 214: 3630-3638.
- Lozano, J., and Bellés, X. 2011. Conservation of the repressive function of Krüppel homolog 1 on insect metamorphosis in hemimetabolous and holometabolous species. *Scientific Reports* 1: 163. doi: 10.1038/srep00163.
- Maestro, J.L., Tobe, S.S., and Bellés, X. 2011. Leucomyosuppressin modulates cardiac rhythm in the cockroach *Blattella germanica*. *Journal of Insect Physiology* 57: 1677-1681.



Fig. 1: Inhibition of metamorphosis in *Blattella germanica*. This intermediate monster between a nymph and an adult was obtained by treating the last instar nymph with juvenile hormone.

Image: Albert Masó.

Book Chapters

- Bellés, X. 2011. Origin and Evolution of Insect Metamorphosis. In: *Encyclopedia of Life Sciences (ELS)*. John Wiley & Sons, Ltd: Chichester. <http://www.els.net> [doi: 10.1002/9780470015902.a0022854]
- Bellés, X., Alexandre, S., Tanaka, E., Rubio, M., and Piulachs, M.D. Insect MicroRNAs: From Molecular Mechanisms to Biological Roles. In: *Insect Molecular Biology and Biochemistry*. Lawrence I. Gilbert Ed., pp. 30-56. Elsevier. ISBN: 978-0-12-384747-8.

Other Publications

- Bellés, X. 2011. Systematics of the genus *Oviedinus* nov. (Coleoptera: Ptinidae), including a fossil new species from Dominican amber, biogeographical remarks and an account on fossil ptnids. *Elytron* 24: 77-88.
- Bellés, X. 2011. When inordinate tissue growth is beneficial: Improving silk production by increasing silk gland size. *Cell Research* 21: 862-863.

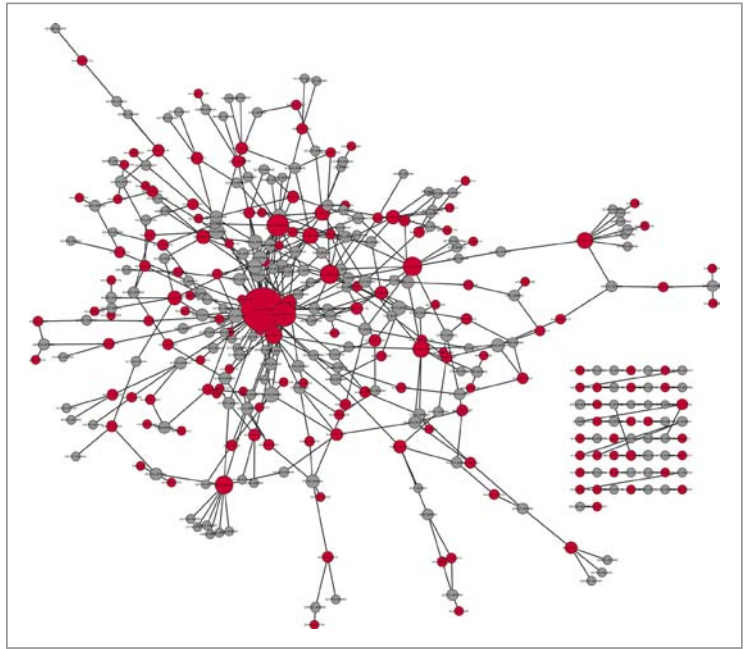


Fig. 2: Network of microRNAs and their potentially regulated targets in pupae of the silkworm, *Bombyx mori*.

Image: Raúl Montañez.

FUNDED PROJECTS

Project Title: Silencing the silencers. Mechanistic bases of metamorphosis regulation in insects.

Financed by: Ministerio de Ciencia e Innovación (REF: CGL2008-03517/BOS, Consolider Modality)

Years: 2009-2013

PI: Xavier Bellés

Project Title: Clearing bacteroids with RNAi

Financed by: Ministerio de Ciencia e Innovación (REF: CGL2010-09266-E, Explora Program)

Years: 2010-2011

PI: Xavier Bellés

Project Title: Insect Control with RNAi

Financed by: CSIC and the National Taiwan University (REF: 2010TW0019, Formosa Program)

Years: 2010-2011

PI: Xavier Bellés

Project Title: Global change and physiological diversity

Financed by: International Laboratory of Global Change (LINCGlobal), CSIC (Spain)-PUC (Chile)

Years: 2009-2012

PIs: Xavier Bellés and Francisco Bozinovic

GROUP
INSECT REPRODUCTION



Carlos Vásquez
Nashwa Elshaer
Maria-Dolors Piulachs
Alba Herráiz
Aníbal de Horna
Erica D. Tanaka
Paula Irlles

GROUP MEMBERS

Maria-Dolors Piulachs, Group Leader

| Research Scientist, CSIC



Erica D. Tanaka, Post-doc | JAEDOC-CSIC Contract
Paula Irlles, Post-doc | Becas Chile Contract
Alba Herráiz, PhD Student | JAEPRE-CSIC Contract - LINCGlobal
Nashwa Elshaer, PhD Student | JAEPRE-CSIC Fellowship
Carlos Vásquez, PhD Student | Becas Chile Contract
Aníbal de Horna, Bioinformatics Support | CSIC Project Contract

RESEARCH OUTLINE

Our aim is to elucidate how the oogenesis in insects is regulated, considering the structural diversity of ovary types and their respective evolutionary history. In insects, we can distinguish two types of ovaries: panoistic and meroistic. The panoistic type is more frequent in basal insects, whereas the meroistic one predominates in more modified groups, which suggest that the evolutionary transition was from panoistic to meroistic. Our purpose is to understand such transition and the working approach is to consider that some genes are conserved in structure and function in panoistic and meroistic ovaries, whereas other genes are specific of one of these types or have different functions in the two ovarian types. We choose a panoistic model, the cockroach *Blattella germanica*, with the idea to study the function of genes expressed in the ovary of this basal species and to establish how many of these genes and functions are conserved in meroistic models.

RESEARCH SUBLINES

1. Vitellogenesis

It is the most important process in oocyte maturation and with a pivotal role in insect reproduction. We like to understand the mechanism of vitellogenin synthesis induced by JH and to elucidate the mechanisms regulating the expression of the involved receptors.

2. Regulation of oogenesis in panoistic ovaries

We study ovarian maturation, focusing our research in oocyte capacitation during last nymphal instar, and in oocyte maturation in previtellogenesis, vitellogenesis, and chorion synthesis. Attention is also paid to the role of miRNA in regulating the expression of genes considered essential to complete oogenesis.

3. Evolution of ovarian structure and function

We focus our work in polarity of the oocyte and proliferation of follicular cells, by studying the tyrosine kinase pathway and those genes that signal the posterior fate in meroistic ovaries, by determining the polarization of microtubule cytoskeleton. The study of follicular cell proliferation will be centered on the genes of the Hippo pathway, studying their function and regulation in activation of follicular cell populations.



Fig. 1: Ovarioles from a *Blattella germanica* 6-day-old last instar nymph. Actins were stained with Phalloidin-TRITC (red) and nuclei were stained with DAPI (blue).

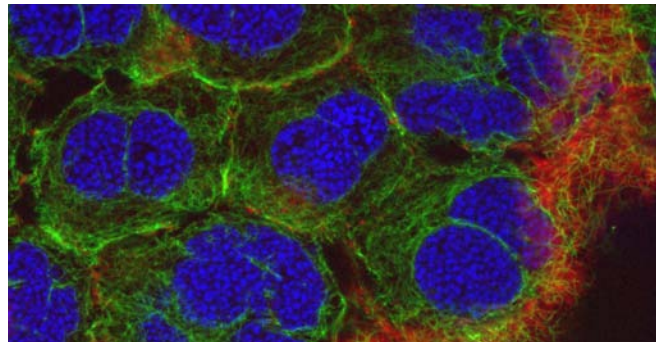


Fig. 2: Follicular cells in late chorion ovarian follicles of *Blattella germanica*. Tubulins were revealed using a β -tubulin-anti-mouse (green), actins were stained with Phalloidin-TRITC (red) and nuclei were stained with DAPI (blue).

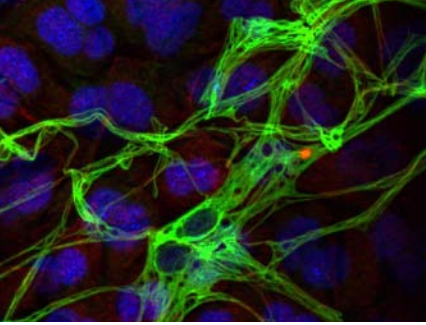


Fig. 3: Tunica propria in late chorion ovarian follicle of adult *Blattella germanica*, in a second plane appeared the follicular epithelia. Tubulins were revealed using a β -tubulin-anti-mouse (green), actins were stained with Phalloidin-TRITC (red) and nuclei were stained with DAPI (blue).

PUBLICATIONS 2011

ISI Articles

- Bortolin, F., Piulachs, M.D., Congiu, L., and Fusco, G. 2011. Cloning and expression pattern of the ecdysone receptor and retinoid X receptor from the centipede *Lithobius peregrinus* (Chilopoda, Lithobiomorpha). *General and Comparative Endocrinology* 174: 60-69.
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- Cristino, A.S., Tanaka, E.D., Rubio, M., Piulachs, M.D., and Bellés, X. 2011. Deep sequencing of organ- and stage-specific microRNAs in the evolutionarily basal insect *Blattella germanica* (L.). *PLoS ONE* 6(4): e19350. doi: 10.1371 / journal.pone.0019350.
- Folguera, G., Bastías, D.A., Caers, J., Rojas, J.M., Piulachs, M.D., Bellés, X., and Bozinovic, F. 2011. An experimental test of the role of environmental temperature variability on ectotherm molecular, physiological and life-history traits: Implications for global warming. *Comparative Biochemistry and Physiology Part A* 159: 242-246.
- Herraiz, A., Chauvigné, F., Cerdà, J., Bellés, X., and Piulachs, M.D. 2011. Identification and functional characterization of an ovarian aquaporin from the cockroach *Blattella germanica* (L.) (Dictyoptera, Blattellidae). *Journal of Experimental Biology* 214: 3630-3638.
- Irls, P., and Piulachs, M.D. 2011. Citrus, a key insect eggshell protein. *Insect Biochemistry and Molecular Biology* 41: 101-108.

FUNDED PROJECTS

Project Title: *Endocrine keys in the structural evolution of insects ovaries*

Financed by: *Ministerio de Ciencia e Innovación (BFU2008-00484)*

Years: 2009-2011

PI: *Maria-Dolors Piulachs*

Project Title: *Global change and physiological diversity*

Financed by: *International Laboratory of Global Change (LINCGlobal), CSIC (Spain)-PUC (Chile)*

Years: 2009-2012

PIs: *Xavier Bellés and Francisco Bozinovic*

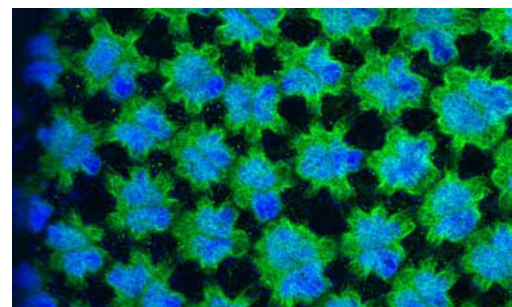
Project Title: *Eliminación de bacteroides con RNAi e implicaciones en el estudio de la simbiosis*

Financed by: *Ministerio de Ciencia e Innovación*

Year: 2011

PI: *Xavier Bellés*

Fig. 4: Basal pole of follicular cells in mid chorion ovarian follicles of adult *Blattella germanica*. Tubulins were revealed using a β -tubulin-anti-mouse (green) and nuclei were stained with DAPI (blue).



GROUP

NUTRITIONAL SIGNALS IN INSECTS



GROUP MEMBERS

José Luis Maestro, Group Leader

| Tenured Scientist, CSIC



Songül Süren-Castillo, Post-doc | JAEDOC-CSIC Fellowship

Marc Abrisqueta, PhD Student

RESEARCH OUTLINE

Our main research interest focuses on the study of nutritional signals in insects and how they regulate different processes, especially reproduction. Nutritional signaling, and in particular the *Insulin Receptor* and the *target of rapamycin* (TOR) pathways, are involved in the detection of the nutritional status and the activation of really crucial processes, such as growth, cellular proliferation, longevity, cancer or reproduction. The cockroach *Blattella germanica* is an excellent model for studying nutritional signals and their relationship with reproduction in insects for different reasons. First of all, because it is an anautogenous species, which means that we have an easy model of activation and deactivation of nutritional signaling pathways just by feeding or starving the animals. Second, cockroach vitellogenesis is activated by the juvenile hormone, as it occurs in most of the insects, and conversely as it happens in the usual models, flies and mosquitoes, which vitellogenesis is activated by ecdysteroids. This makes *B. germanica* a superb model for studying insect reproductive physiology in a general framework, and for comparing the two types of hormone-activated vitellogenesis. In addition, the fact that RNAi works exceptionally well in *B. germanica* permit us to perform valuable functional studies.

RESEARCH SUBLINES

1. Nutritional signaling pathways: Insulin receptor and TOR

The insulin receptor and the TOR pathways are involved in detecting nutritional signals and activating different processes, such as growth, cell proliferation, longevity and cancer, both in insects and in other organisms. Our group studies these pathways in *B. germanica* and their relationships to reproduction and, in particular, to the activation of juvenile hormone and vitellogenin production. We are conducting studies using different experimental models, including fed and fasted individuals and specimens in which we manipulate the expression of different proteins, such as InR, TOR or the kinase of the ribosomal protein S6 (S6K). We also perform different hormone *in vivo* and *in vitro* treatments together with quantification of hormone biosynthesis.

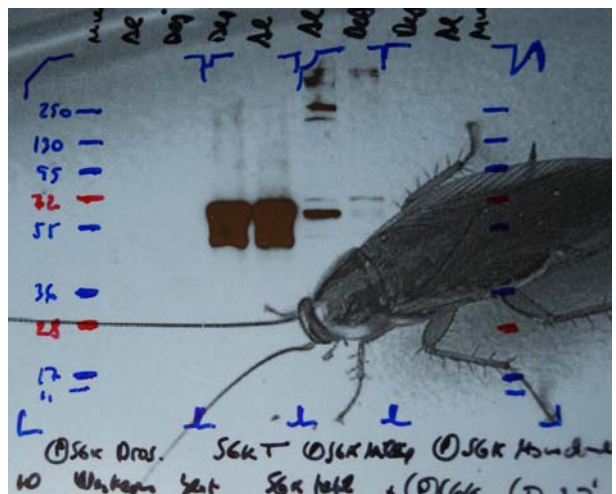


Fig. 1: S6K and phosphorylated S6K Western-blot analysis of fat body extracts from fed and starved *B. germanica* adult females.

2. FOXO transcription factor

FOXO proteins are evolutionary conserved transcription factors which have been described as key players of the InR pathway. This pathway would regulate FOXO activity through its differential phosphorylation. *B. germanica* FOXO, which has been recently cloned by our group, maintains exactly FoxO canonical phosphorylation sites, so we assume that it will be regulated in the same way. We are interested in analyzing the role of FOXO in different processes. Our main question is whether the nutritional signals activate reproduction through FOXO and which are the genes regulated by the action of FOXO. We are also interested in the role of FOXO in some other processes as starvation resistance, oxidative stress and longevity.

PUBLICATIONS 2011

ISI Articles

- Maestro, J.L., Tobe, S.S., and Bellés, X. 2011. Leucomyosuppressin modulates cardiac rhythm in the cockroach *Blattella germanica*. *Journal of Insect Physiology* 57: 1677-1681.

FUNDED PROJECTS

Project Title: Nutritional signals and reproduction in insects. Role of the transcription factor FoxO (BFU2010-15906)

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: José Luis Maestro



Fig. 2: Forewings and hindwings from *dsInR*-treated *B. germanica* females.

GROUP

HORMONAL CONTROL OF INSECT DEVELOPMENT



Enric Ureña

Ferran Borràs

David Martín

Cristina Manjón

GROUP MEMBERS

David Martín, Group Leader

| Tenured Scientist, CSIC



Cristina Manjón, Post-doc | Project Contract

Enric Ureña, PhD Student | JAEPRE-CSIC Fellowship

Ferran Borràs, PhD Student | FPI Scholarship, MEC

RESEARCH OUTLINE

The main goal of our work is to elucidate the molecular basis of the endocrine control of insect metamorphosis, particularly analyzing the roles of the two main insect hormones, Ecdysteroids and Juvenile Hormones. For that, our group is carrying out comparative studies of the role of these two hormones in the regulation of embryogenesis, post-embryonic growth, molting and metamorphosis in a direct-developing hemimetabolous insect with incomplete metamorphosis (*Blattella germanica*) and in two holometabolous insects with complete metamorphosis (*Tribolium castaneum* and *Drosophila melanogaster*).

RESEARCH SUBLINES

1. Molecular analysis of Ecdysteroids and Juvenile Hormone action during insect metamorphosis

In insects, the steroid hormone 20-hydroxyecdysone (20E) triggers major developmental transitions through a genetic cascade of transcription factors that belong to the Nuclear Hormone Receptor superfamily (NRs). Furthermore, Juvenile Hormone (JH), the other hormone of paramount importance in development, prevents metamorphosis by coordinating multiple 20E-dependent developmental and physiological processes. The main goal of this project is the characterization of the regulatory role of the NRs belonging to the 20E-triggered genetic cascade in *B. germanica* and *T. castaneum* (using RNAi in vivo and parental RNAi procedures) and in *D. melanogaster* (mutational analysis). These studies have already demonstrated critical roles of these transcription factors on ecdysteroid production, programmed cell death, tissue growth and morphogenesis, ovary follicle proliferation and molting behaviour in these three types of insects. On the other hand, to elucidate the JH signalling pathway, we are currently analyzing in detail the function of *methoprene-tolerant*, the putative JH receptor.



Fig. 1: RNAi-mediated knockdown of SUMO in *Blattella germanica* nymphs are unable to molt into adults. Wild type adult cockroach (left) and *sumo* knockdown nymph (right).

2. Embryonic development in short germ band insects

The main goal of this project is to characterize the major morphogenetic events during the early-embryogenesis of the hemimetabolous insect model *B. germanica*, analyzing the role of each 20E-dependent NR on these morphogenetic events. These embryonic morphogenetic changes constitute the truly metamorphosis in hemimetabolous short-germ band insects. This project will provide a valuable model to understand the molecular basis for how shifts in the hormonal context can lead to the creation of different body forms.

3. Control of developmentally regulated programmed cell death by steroids and juvenile hormone

In holometabolous insects, complete metamorphosis is based on the destruction of larval tissues by programmed cell death (PCD) to accommodate the growth of new adult structures. However, given that metamorphosis arose from a hemimetabolous ancestor, it would be interesting to study whether the mechanisms that coordinate stage-specific PCD were already present in more primitive hemimetabolous insects or they are a novelty of holometabolous species. Using reverse genetic studies, we are carrying out a detailed functional analysis of the 20E-mediated death of the prothoracic gland of *B. germanica*, which undergoes PCD just after the imaginal molt. Furthermore, we are also characterizing in detail the antiapoptotic role of JH.

4. Sumoylation and development

Post-translational modification with small ubiquitin-related modifier, SUMO, is a widespread mechanism for rapid and reversible changes in protein function, especially in nuclear hormone receptors. In this project, in collaboration with the laboratory of Dr. Rosa Barrio (CIC bioGUNE, Vizcaya), we are addressing the functional analysis of sumoylation on the development of the hemimetabolous model insect, *B. germanica*, and its relationship with nuclear hormone receptor function.

FUNDED PROJECTS

Project Title: *Molecular Basis of Ecdysteroid and Juvenile Hormone Actions in Insect Development. The Role of Nuclear Hormone Receptors.*
(REF: BFU2009-10571)

Financed by: *Ministerio de Ciencia e Innovación*

Years: *2010-2012*

PI: *David Martín*



Fig. 2: Adult *Drosophila melanogaster* *Seven up* RNAi knockdown showing defects in wing spreading. Wild type (left) and *Seven up* knockdown (right).

Image: Cristina Manjón.

GROUP

DROSOPHILA TELOMERES



Elisenda López

Elena Casacuberta

Rute Sousa

David Piñeyro

GROUP MEMBERS

Elena Casacuberta, Group Leader

| Tenured Scientist, CSIC



Rute Sousa, PhD Student | PhD Fellowship, Fundação para a Ciência e Tecnologia, Portugal

Elisenda López Panadès, PhD Student | UPF Contract

RESEARCH OUTLINE

Our group focuses on the study of how transposable elements interact with the eukaryote genome and how these interactions can actively contribute to evolution. To study this we use the telomeres of *Drosophila* as a model.

Although the telomeres of *Drosophila* are functionally equivalent to telomerase telomeres, they are maintained by an alternative mechanism. Instead of telomerase, *Drosophila* uses three non-LTR retrotransposons –HeT-A, TART and TAHRE– to elongate the end of the chromosomes when need it. These very special retrotransposons maintain their personality as transposons but at the same time are committed to maintain the telomeres in *Drosophila*. Telomere maintenance is crucial for processes as important as aging, tumorigenesis and genome stability.

In our group, we are studying different aspects of telomere elongation and stability in *Drosophila* and the consequences for telomere and retrotransposon evolution.

RESEARCH SUBLINES

1. Host and Retrotransposon requirements for telomere elongation and stability in *Drosophila*

HeT-A, TART and TAHRE must be integrated in the cellular pathways controlling telomere length and genome stability. Understanding the regulation of the telomeric transposons will shed light in both telomere length control as well as transposon regulation in *Drosophila*. We are currently focusing on the regulation of the telomeric chromatin and the consequences for telomere stability.

2. Interacting partners of the Telomeric proteins

In order to understand which are the cellular partners that assist the telomeric proteins throughout their life cycle and exclusively target the telomeres, we are currently isolating and identifying protein complexes using the telomeric proteins as bait.

3. Evolution of the telomere retrotransposons

The sequences of HeT-A and TART, although linked to an essential cellular role are allowed to change at a fast rate. Studying the level of variability of the telomere retrotransposons is important to better understand their dynamics and the forces that are driving their evolution. We are currently studying the possible functionality of a highly conserved piRNA target sequence found inside one of the telomeric retrotransposons.

4. Variability within the telomere retrotransposons

The different copies of each telomeric retrotransposon can be divided in different subfamilies. We are investigating if this variability is due to different necessities in telomere elongation or as a response to specific mutations.

PUBLICATIONS 2011

ISI Articles

- Piñeyro, D., López, E., Lucena Pérez, M., and Casacuberta, E. 2011. Transcriptional variability of the telomeric retrotransposon HeT-A in mutant and wild-type *Drosophila melanogaster* stocks. *BMC Genomics* 12: 573.

FUNDED PROJECTS

Project Title: Estudio de los aspectos funcionales y evolutivos de los telómeros de *Drosophila*

Financed by: Ministerio de Ciencia e Innovación (BFU2009-08318)

Years: 2010-2012

PI: Elena Casacuberta Suñer

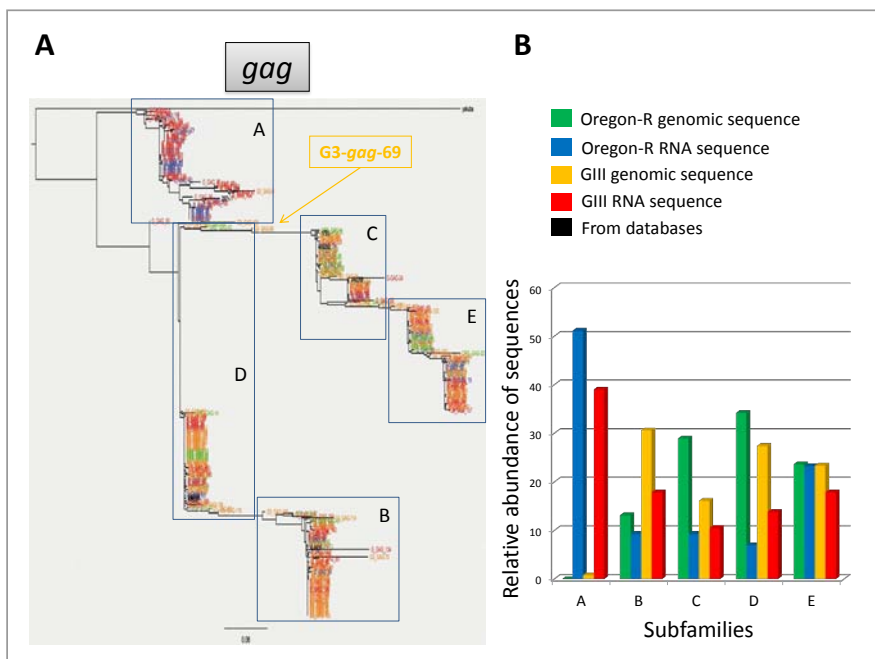


Fig. 1: Phylogenetic analysis of the sequences amplified for the gag fragment.

All amplified sequences from genomic DNA of Oregon-R and GIII (names in green and orange, respectively) and from total RNA from 3rd instar larvae of Oregon-R and GIII (names in blue and red, respectively) were used. Also included are the previously annotated HeT-A sequences containing the analyzed fragment (shown in black).

A. Phylogenetic tree constructed using all obtained sequences. The different subfamilies are indicated with boxes with the corresponding letter. Additionally, the tree includes a *Drosophila yakuba* HeT-A sequence (AF043258, named yakuba in the tree), in order to illustrate the degree of variability of HeT-A inside species and between species (D.yakuba and D.melanogaster are considered to be ~10 million years of genetic distance; source: <http://flybase.org>). The position of sequence G3-gag-69 is indicated.

B. Bar chart representing the relative abundance of each subfamily.

GROUP

DEVELOPMENTAL BIOLOGY AND MORPHOLOGICAL
EVOLUTION IN INSECTS



Cristina
Miguel Vijandi

Xavier
Franch-Marro

Neus Bota
Rabassedas

GROUP MEMBERS

Xavier Franch-Marro, Group Leader

| Tenured Scientist, CSIC



Neus Bota Rabassedas, PhD Student | FPI Scholarship, MEC

Cristina Miguel Vijandi, PhD Student | FPI Scholarship, MEC

RESEARCH OUTLINE

Evolution along Earth history has developed a great number of different organisms with a consequent incredibly variety of forms and sizes. One of the big questions in Biology is to understand which genes and what kind of changes in their sequences are responsible for the evolution of the mentioned morphological diversity. Using *Drosophila melanogaster* and *Tribolium castaneum* as organism models, the aim of our lab is to elucidate the mechanisms that allow the formation of new morphologies and sizes. We use two different organ models to address these questions: the tracheal system and the imaginal wing disc. In both systems, Wnt signalling plays a crucial role during its respective development controlling morphogenesis and size. For instance, we have found that small changes in Wnt signalling activation result in a different final size of the *Drosophila* wing. This could mean that small changes in the domain of activation of a signalling pathway in a tissue could give rise to different organ size along evolution. This effect could be achieved by either acquisition of new regulatory regions of target genes or by changes in the regulation of the activity of the pathway. Therefore, identification of new target genes of the pathway and the comparison of their expression pattern in *Drosophila* and *Tribolium*, in trachea and wing disc, will give us information of how this signalling pathway has evolved to control pattern as well as organ size along evolution.

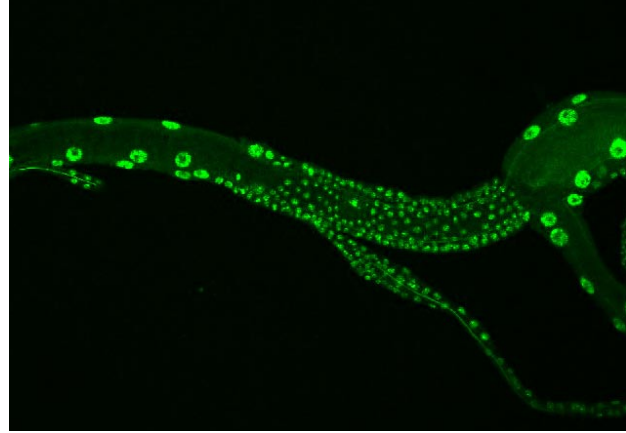


Fig. 1: Larval tracheal system. Tracheal cells of the second thoracic segment proliferate during 3rd instar larva to form the pupa and adult trachea.

RESEARCH SUBLINES

1. Tracheal System Remodeling and Morphogenesis

The tracheal system is the *Drosophila* respiratory organ and consists of epithelial tubes, the morphogenesis of which is controlled by distinct sets of signalling pathways and transcription factors. During embryogenesis, the tracheal system develops from segmentally repeated groups of ~ 80 cells that express *Trachealess* transcription factor and invaginate, forming sacs attached to the epidermis by a stalk of spiracular branch (SB) cells. Branches bud from the sacs and cells diversify primarily under control of FGF (fibroblast growth factor) signalling pathway. At metamorphosis, tracheal system undergoes a deep remodelling stage, giving rise to pupae and adult tracheal system. This remodelling involves proliferation of both a classical imaginal cell population, as in Spiracular Branch and a population of differentiated functional larval tracheal cells, as in Tr2 that re-enters the cell cycle and regain development potency. The genetic circuits controlling tracheal cell proliferation and *dedifferentiation* are only now beginning to emerge. Therefore, we aim to discover new genes and signalling pathways involved in such interesting processes.

2. Tracheal System Evolution

The tracheal system is the respiratory organ of insects. It consists of a network of tubes that transport oxygen to all the tissues. Insects present different morphology of the tracheal network depending on its habitat. For instance, we

have found that *Drosophila* tracheal network presents some morphological innovations compared to the tracheal morphology of a most primitive insect such as *Tribolium*. The main goal of this project is to discover the genetic changes that have allowed the generation of those morphological adaptations along evolution.

3. Wingless secretion

Wnt ligands comprise a large family of secreted proteins that control a variety of developmental and adult processes in all metazoan organisms. By binding to various receptors present on receiving cells, Wnt proteins initiate intracellular signalling cascades, which lead to changes in gene transcription. It has already shown that Wingless, the main Wnt *Drosophila* member, is post-transcriptional modified and then secreted by its dedicated seven-pass-transmembrane protein Wntless (Wls). However, the function of these post-transcriptional modifications is still poor understood. Thus, our goal is to understand the mechanism that controls Wg secretion by Wls and the function of its post-transcriptional modifications.

4. Wingless signaling in size control and evolution

How organ size and shape are regulated is a remaining outstanding question in developmental biology. Recently, we have shown that Wg signaling has an important role controlling growth in *Drosophila* wing imaginal discs. New experimental approaches have allowed us to find that a mild increase of Wg signaling over and above the endogenous level causes wing overgrowth by promoting cell proliferation. However, how this Wg signaling activation controls cell proliferation at a transcriptional level is still elusive. Therefore, the aim of our project is, using a microarray approach, to identify and characterize new target genes of the signaling pathway that would explain mechanistically the way Wg controls cell proliferation in *Drosophila* wing disc. In parallel, we study these genes in *Tribolium castaneum* in order to gain further insights into developmental processes occurring during beetle and fly development leading to more general conclusions for arthropods evolution.

PUBLICATIONS 2011

ISI Articles

- Campbell, K., Whissell, G., Franch-Marro, X., Batlle, E., and Casanova, J. 2011. Specific GATA Factors Act as Conserved Inducers of an Endodermal-EMT. *Developmental Cell* 21, 1051-1061.

FUNDED PROJECTS

Project Title: Formación del Gradiente del Morfógeno y de la función de Wingless en el control del crecimiento

Financed by: Ministerio de Ciencia e Innovación (REF: BFU2009-08748)

Years: 2010-2012

PI: Xavier Franch-Marro

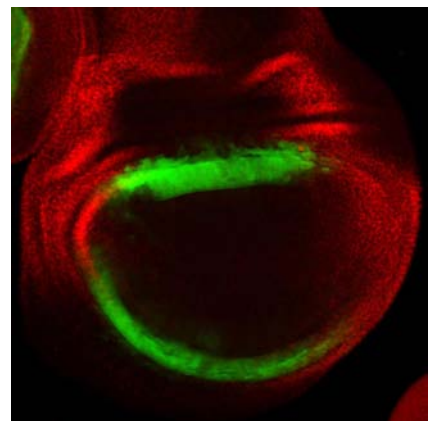


Fig. 2: Wing imaginal disc of *Drosophila* showing the expression of two genes that contribute to the patterning of the adult wing.

GROUP

EVOLUTIONARY AND FUNCTIONAL GENOMICS



Lidia
Mateo

Josefa
González

GROUP MEMBERS

Josefa González, Group Leader

| Ramon y Cajal Researcher, CSIC



Lidia Mateo, Undergraduated Student | UPF Contract

RESEARCH OUTLINE

Adaptation is the key concept in Evolutionary Biology. Understanding adaptation has important scientific and social implications, since adaptation underlies processes such as the ability of species to survive in changing environments and resistance to antibiotics and cancer chemotherapies. However, adaptation is to date a very poorly understood process, largely because the current approaches to the study of adaptation are often exclusively based on a priori candidate genes or on searching for signals of selection at the DNA level giving us an incomplete and biased picture of the adaptive process.

The long-term goal of our research is to understand the molecular process and functional consequences of adaptation. Towards this end, we focus on the study of recent transposable element (TE)-induced adaptations in *Drosophila melanogaster*, which we have already shown to be common and readily detectable. We apply a multidisciplinary approach that combines genomics, population genetics, computational biology, molecular biology and experimental evolution to identify and analyze Adaptive TE insertions.

FUNDED PROJECTS

Project Title: El proceso molecular y las consecuencias funcionales de la adaptación (BFU2011-24397)

Financed by: Ministerio de Ciencia e Innovación

Years: 2012-2014

PI: Josefa González

Project Title: The molecular process and functional consequences of adaptation (PCIG09-GA-2011-293860)

Financed by: European Commission

Years: 2011-2014

PI: Josefa González

Project Title: The process of adaptation and its functional consequences (RYC-2010-07306)

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2015

PI: Josefa González

Project Title: La ciència és part de la teva vida (2011ACDC 00060)

Financed by: Secretaria d'Universitats i Recerca. Departament d'Economia i Coneixement. Generalitat de Catalunya.

Year: 2011

PI: Josefa González

Fig. 1: Details of our fruitfly collection in a vineyard located in Southern Italy (October 2011).







PROGRAM

population genetics

RESEARCH GROUPS

Evolutionary Systems Biology

Jaume Bertranpetit, *Group Leader*

Evolutionary Population Genetics

Elena Bosch, *Group Leader*

Human Genome Diversity

David Comas, *Group Leader*

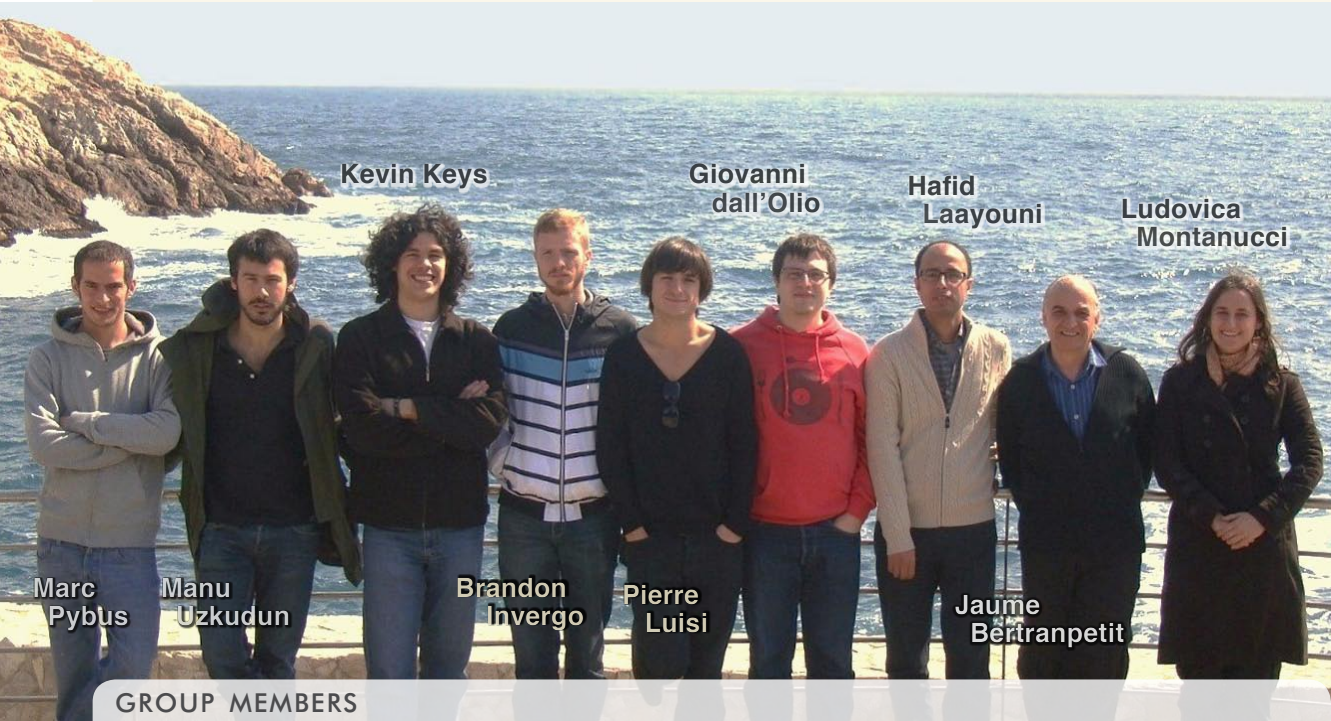
microRNAs in Human Adaptation and Disease

Yolanda Espinosa-Parrilla, *Group Leader*

In the population genetics line, intraspecific diversity patterns within populations and comparative data are explored with the general aim of reconstructing the processes that have created such a diversity. Genetic diversity is the result of the intricate interaction of different processes: some are embedded in the genome, such as mutation and recombination; others are absolutely independent from the genome and affect its entirety, such as demographic events; and finally, other processes result from the exposure of the genetic diversity to the environment, such as natural selection. Within this line, we are interested in all three types of processes mainly in humans. Namely, we investigate how recombination can be affected by genetic differences between populations; the demographic histories of particular populations or population groups; and the extent of the adaptation of humans to their pathogen exposure or to nutrient availability in their diets. In addition, the functional consequences of these processes in the human non-coding genome are also evaluated. Finally, the integration of the different levels of functional variation on genes related to particular human traits is used to understand human adaptation as a system-networking phenomenon.

GROUP

EVOLUTIONARY SYSTEMS BIOLOGY



Marc Pybus

Manu Uzkudun

Kevin Keys

Brandon Invergo

Pierre Luisi

Giovanni dall'Olio

Hafid Laayouni

Jaume Bertranpetit

Ludovica Montanucci

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| Professor, UPF



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Ludovica Montanucci, Post-doc | UPF Contract

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Pierre Luisi, PhD Student | ISCIII Scholarship

Brandon Invergo, PhD Student | FI Scholarship, Generalitat de Catalunya

Marc Pybus, PhD Student | FI Scholarship, MICINN

Manu Uzkudun | Master Student (till September)

Kevin Keys | Master Student (till July)

RESEARCH OUTLINE

Our main research focuses on how to use molecular pathways to understand the biology of adaptation through the measures of natural selection in genes and other genomic regions. The different forms of selection (purifying, balancing and positive) are analyzed at two levels: among human populations in order to detect population-specific adaptations, and among primates in order both to recognize species-specific adaptive selection and to measure the relative strength of purifying selection. We have also ongoing work in understanding recombination and in reconstructing population history by studying human genetic diversity. We are also collaborating with Carles Lalueza-Fox in ancient DNA studies and the functional implications of the genetic differences; with Francesc Calafell in the study of surnames and Y-chromosome diversity; with David Comas in human population studies, including the Genographic Project and the genetics of North Africa; and with Tomàs Marquès-Bonet on detecting selection in the genome of apes.

RESEARCH SUBLINES

1. Footprints of adaptation in humans and purifying selection in higher primates

The action of natural selection is at the base of different amounts of genetic dispensability or relative importance (in cases of negative or purifying selection) or of adaptation (in cases of positive selection and in the special case of balancing selection) that will be population-specific (in the case of human diversity) or species-specific (in the case of genetic divergence). The action of selection is measured and understood not in terms of single lists of genes, but integrated in molecular physiological pathways or networks, and the aim is, in a given pathway, to understand the complex basis of adaptation and how networks have been shaped by natural selection. Indeed to understand the complex basis of adaptation it is necessary to integrate the knowledge derived from evolutionary studies into a network framework since biological function is the result of a large number of interacting molecules organized in complex networks and arises as an emergent property from a combined effect of many different genes. The final goal is, on one hand, to understand in specific pathways how evolution has taken place, where positive selection (and balancing selection) has taken place, and where purifying selection has been shaping the genome, and on the other, to obtain possible general patterns of evolution in molecular pathways and networks. Data is retrieved mostly from pre-existing databases but, in cases of doubt, low quality or low density, we produce our own (sequences and SNPs). Inter-specific data include the sequences of several primates and intra-specific databases include: HapMap, in versions 2 and 3; SNP analysis of the HGDP panel with the 650k array of Illumina; re-sequencing projects (like Seattle-SNP), and recently the 1000 Genomes Project. The pathways that we are analyzing are: N-glycans; integration of all glycosylation pathways; innate immunity; skin pigmentation; visual perception; and obesity through adiposity signals.

Special attention has been put in the quality of databases for metabolic pathways, as their quality is worse than most studies assume and manual curation is needed in all cases.

2. Human population genetics and recombination

Recombination is a main force shaping genome diversity. In collaboration with Laxmi Parida (Computational Biology Center, IBM T J Watson Research, Yorktown, USA), we have developed an algorithm, implemented in the IRiS program, to detect past recombination events in extant sequences, with specificity of parental and recombinant sequences. The algorithm detects recombination events from tree incompatibilities found along the sequence. We have validated and calibrated the algorithm for the human genome given human demographic history and the human recombination model by means of coalescent simulations implementing a standard model of human demography.

We are also interested in the evolution of recombination and differences in rates among human populations and have demonstrated that there is stratification in the recombination rates among human populations strongly related to genetic distances.

3. Human genetic diversity and population history

In collaboration with David Comas (see his page for more details), we are participating in the Genographic Project promoted by National Geographic and IBM as responsible for Central and Western Europe and participating in variety of population-specific studies (including Basques, North African, Sub-Saharan African and others).

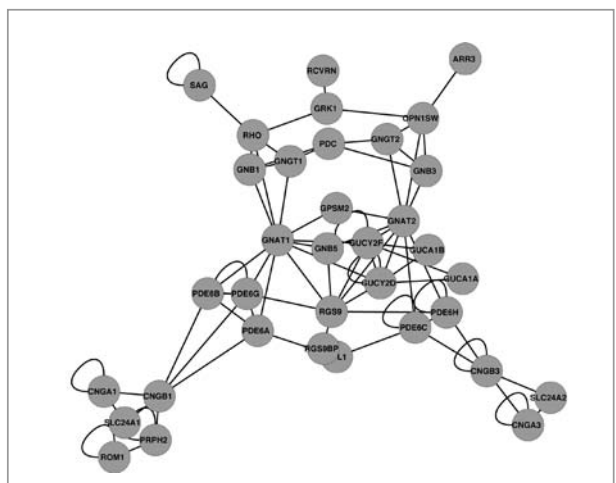
PUBLICATIONS 2011

ISI Articles

- Casals, F., Sikora, M., Laayouni, H., Montanucci, L., Muntasell, A., Lazarus, R., Calafell, F., Awadalla, P., Netea, M.G., and Bertranpetit, J. 2011. Genetic adaptation of the antibacterial human innate immunity network. *BMC Evolutionary Biology* 11(1): 202.
- Dall'Olio, G.M., Jassal, B., Montanucci, L., Gagneux, P., Bertranpetit, J., and Laayouni, H. 2011. The annotation of the Asparagine N-linked Glycosylation pathway in the Reactome Database. *Glycobiology* 21(11): 1395-1400.
- Dall'Olio, G.M., Marino, J., Schubert, M., Keys, K.L., Stefan, M.I., Gillespie, C.S., Poulain, P., Shameer, K., Sugar, R., Invergo, B.M., Jensen, L.J., Bertranpetit, J., and Laayouni, H. 2011. Ten Simple Rules for Getting Help from Online Scientific Communities. *PLoS Computational Biology* 7(9): e1002202.

Fig. 1: A network representation of the phototransduction signaling cascade. Nodes represent proteins and edges represent physical interactions between them.

From: A system-level, molecular evolutionary analysis of mammalian phototransduction. Brandon M. Invergo, Ludovica Montanucci, Hafid Laayouni and Jaume Bertranpetit (in preparation).



- Javed, A., Pybus, M., Melé, M., Utro, F., Bertranpetit, J., Calafell, F., and Parida, L. 2011. IRIS*: Construction of ARG networks at genomic scales. *Bioinformatics* 27(17): 2448-2450.
- Laayouni, H., Montanucci, L., Sikora, M., Melé, M., Dall'Olio, G.M., Lorente-Galdós, B., McGee, K.M., Graffelman, J., Awadalla, P., Bosch, E., Comas, D., Navarro, A., Calafell, F., Casals, F., and Bertranpetit, J. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS ONE* 6(3): e17913.
- Marigorta, U.M., Lao, O., Casals, F., Calafell, F., Morcillo-Suárez, C., Faria, R., Bosch, E., Serra, F., Bertranpetit, J., Dopazo, H., and Navarro, A. 2011. Recent human evolution has shaped geographical differences in susceptibility to disease. *BMC Genomics* 12: 55.
- Montanucci, L., Laayouni, H., Dall'Olio, G.M., and Bertranpetit, J. 2011. Molecular evolution and network-level analysis of the N-Glycosylation metabolic pathway across primates. *Molecular Biology and Evolution* 28(1): 813-823.
- Oostra, V., de Jong, M. A., Invergo, B. M., Kesbeke, F., Wende, F., Brakefield, P. M., Zwaan, B.J. 2011. Translating environmental gradients into discontinuous reaction norms via hormone signalling in a polyphenic butterfly. *Proceedings of the Biological Sciences* 278: 789-797.
- Parnell, L.D., Lindenbaum, P., Shameer, K., Dall'Olio, G.M., Swan, D.C., et al. 2011. BioStar: An Online Question & Answer Resource for the Bioinformatics Community. *PLoS Computational Biology* 7(10): e1002216. doi: 10.1371/journal.pcbi.1002216.
- Sikora, M., Laayouni, H., Calafell, F., Comas, D., and Bertranpetit, J. 2011. A genomic analysis identifies a novel component in the genetic structure of sub-Saharan African populations. *European Journal of Human Genetics* 19(1): 84-88.
- Sikora, M., Laayouni, H., Menéndez, C., Mayor, A., Bardaji, A., Sigauque, B., Netea, M.G., Casals, F., and Bertranpetit, J. 2011. A targeted association study of immunity genes and networks suggests novel associations with placental malaria infection. *PLoS ONE* 6(9): e24996.

As part of The Genographic Consortium

- Balanovsky, O., Dibirova, K., Dybo, A., Mudrak, O., Frolova, S., Pocheshkhova, E., Haber, M., Platt, D., Schurr, T., Haak, W., Kuznetsova, M., Radzhabov, M., Balaganskaya, O., Romanov, A., Zakharova, T., Soria-Hernanz, D.F., Zalloua, P., Koshel, S., Ruhlen, M., Renfrew, C., Wells, R.S., Tyler-Smith, C., Balanovska, E., and The Genographic Consortium. 2011. Parallel evolution of genes and languages in the Caucasus region. *Molecular Biology and Evolution* 28(10): 2905-2920.
- Cai, X., Qin, Z., Wen, B., Xu, S., Wang, Y., Lu, Y., Wei, L., Wang, C., Li, S., Huang, X., Jin, L., Li, H., and The Genographic Consortium. 2011. Human migration through bottlenecks from Southeast Asia into East Asia during Last Glacial Maximum revealed by Y chromosomes. *PLoS ONE* 6(8): e24282.
- Gaieski, J.B., Owings, A.C., Vilar, M.G., Dulik, M.C., Gaieski, D.F., Gittelman, R.M., Lindo, J., Gau, L., Schurr, T.G., and The Genographic Consortium. 2011. Genetic ancestry and indigenous heritage in a Native American descendant community in Bermuda. *American Journal of Physical Anthropology* 146(3): 392-405.
- Haber, M., Platt, D.E., Badro, D.A., Xue, Y., El-Sibai, M., Bonab, M.A., Youhanna, S.C., Saade, S., Soria-Hernanz, D.F., Royyuru, A., Wells, R.S., Tyler-Smith, C., Zalloua, P.A., and The Genographic Consortium. 2011. Influences of history, geography, and religion on genetic structure: the Maronites in Lebanon. *European Journal of Human Genetics* 19(3): 334-340.

- Yan, S., Wang, C.C., Li, H., Li, S.L., Jin, L., and The Genographic Consortium. 2011. An updated tree of Y-chromosome Haplogroup O and revised phylogenetic positions of mutations P164 and PK4. *European Journal of Human Genetics* 19(9): 1013-1015.
- Yang, K., Zheng, H., Qin, Z., Lu, Y., Farina, S.E., Li, S., Jin, L., Li, D., Li, H., and The Genographic Consortium. 2011. Positive selection on mitochondrial M7 lineages among the Gelong people in Hainan. *Journal of Human Genetics* 56(3): 253-256.

FUNDED PROJECTS

Project Title: Selección natural en redes moleculares funcionales
Financed by: Ministerio de Ciencia y Tecnología (BFU2010-19443)
Years: 2010-2012
PI: Jaume Bertranpetit

Project Title: The Genographic project: Western/Central Europe region
Financed by: National Geographic and IBM
Years: 2006-2011
PIs: Jaume Bertranpetit and David Comas

Project Title: Grup de Recerca Consolidat-SGR
Financed by: Generalitat de Catalunya (2009 SGR-1101)
Years: 2009-2013
PI: Jaume Bertranpetit

Project Title: Population genetic and functional analyses of maintenance of DNA sequence variability in response to infectious agents (human innate immune system and other responses)
Financed by: Ministerio de Ciencia e Innovación
Years: 2012-2014
PI: Jaume Bertranpetit

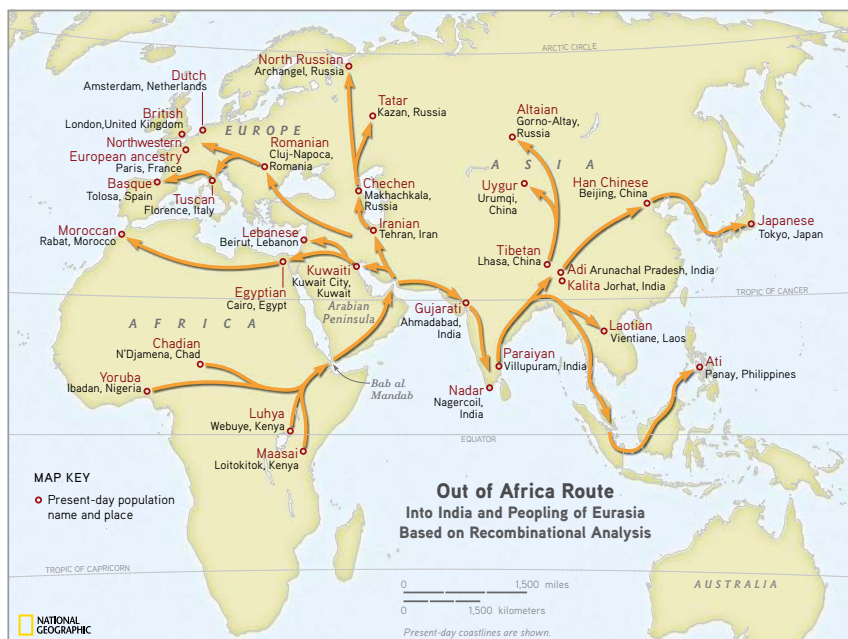


Fig. 2: Map of the Old World with the samples studied in the Recoproject, a part of the Genographic project. The study has proposed a new route of expansion of modern humans through South Arabia and not through Egypt as previously assumed. All the analysis has been based in reconstructing recombination in the populations past.

From: Melé M, Javed A. et al. *The Genographic Consortium*. 2011. Recombination gives a new insight in the effective population size and the history of the Old World human populations. *Molecular Biology and Evolution* doi: 10.1093/molbev/msr213 First published online: September 1, 2011.

GROUP

EVOLUTIONARY POPULATION GENETICS



Elena Carnero

Elena Bosch

Johannes Engelken

GROUP MEMBERS

Elena Bosch, Group Leader

| Associate Professor, UPF



Johannes Engelken, Post-doc | Volkswagenstiftung Scholarship, Germany

Elena Carnero, PhD Student | UPF Teaching Scholarship

RESEARCH OUTLINE

Our research focuses on investigating human genetic adaptation, that is, in identifying traits that have undergone positive selection during human evolution in order to understand the adaptive events that have shaped our genomes. The search of genetic signatures of selection is pursued at different levels either using comparative data and/or exploring intraspecific diversity patterns mainly within human populations but also in chimpanzees. In those cases where the imprint of selection is confirmed, we aim to determine the molecular bases of the functional adaptation. Possible adaptative variants (either coding or regulatory) are first identified through different *in silico* approaches and then experimentally tested by means of functional assays. We are currently investigating molecular phenotypes such as mRNA expression, protein localization and stability, enzyme activity and intracellular signaling.

RESEARCH SUBLINES

1. Recent human adaptation and the immunitary system

By means of a resequencing approach, we have confirmed previous SNP-based evidences of recent positive selection in East Asian populations in a lymphocyte receptor gene, which is involved in recognition of fungal cell walls. We have investigated quantitative and qualitative functional differences between the alleles at two nonsynonymous substitutions linked to the genetic signature of selection. We are also exploring their phenotypical relevant consequences in particular immunological diseases. This project is developed in collaboration with Francisco Lozano's group from the Hospital Clínic (Barcelona).

2. Recent human adaptation and nutrition

A number of mRNA expression QTLs and other additional functional variants are being experimentally tested for a set of different candidate genes related to nutrition which do show signatures of recent adaptation in human populations, possibly as an adaptive response to nutrient availability and diet changes in the past.

3. Role of selection in coding and non-coding regions of the genome

We are obtaining sequence data at both intraspecific and interspecific levels in order to investigate the role of natural selection in all coding and regulatory elements of particular gene pathways. This project is done in collaboration with Arcadi Navarro (Evolutionary Genomics Lab) and with Hernán Dopazo (Centro de Investigación Príncipe Felipe, Valencia).

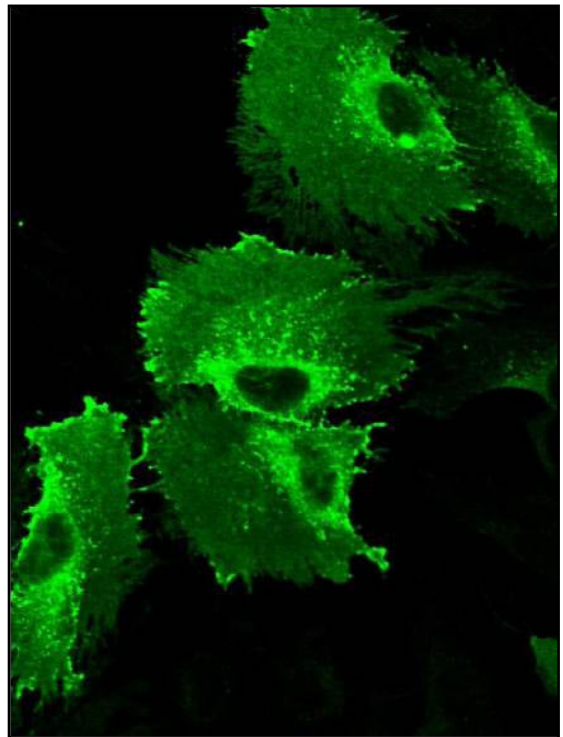


Fig. 1: Immunostained HeLa transfected cells.

4. Bioinformatic analysis of 1000 Genomes data

In collaboration with Jaume Bertranpetit's group (Evolutionary Systems Biology Lab), we have embarked on a joint project to build a flexible framework in order to analyze signatures of natural selection with an emphasis on genome-wide data from the 1000 Genomes Project.

PUBLICATIONS 2011

ISI Articles

- Gazave, E., Darré, F., Morcillo-Suárez, C., Petit-Marty, N., Carreño, A., Marigorta, U.M., Ryder, O.A., Blancher, A., Rocchi, M., Bosch, E., Baker, C., Marquès-Bonet, T., Eichler, E.E., and Navarro, A. 2011. Copy number variation analysis in the great apes reveals species-specific patterns of structural variation. *Genome Research* 21(10): 1626-1639.
- Laayouni, H., Montanucci, L., Sikora, M., Melé, M., Dall'Olio, G.M., Lorente-Galdós, B., McGee, K.M., Graffelman, J., Awadalla, P., Bosch, E., Comas, D., Navarro, A., Calafell, F., Casals, F., and Bertranpetit, J. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS ONE* 6(3): e17913.
- Marigorta, U.M., Lao, O., Casals, F., Calafell, F., Morcillo-Suárez, C., Faria, R., Bosch, E., Serra, F., Bertranpetit, J., Dopazo, H., and Navarro, A. 2011. Recent human evolution has shaped geographical differences in susceptibility to disease. *BMC Genomics* 12: 55.
- Myles, S., Lea, R.A., Ohashi, J., Chambers, G.K., Weiss, J.G., Hardouin, E., Engelken, E., Macartney-Coxson, D.P., Eccles, D.A., Naka, I., Kimura, R., Inaoka, T., Matsumura, Y., and Stoneking, M. 2011. Testing the thrifty gene hypothesis: the Gly482Ser variant in PPARGC1A is associated with BMI in Tongans. *BMC Medical Genetics* 12: 10.

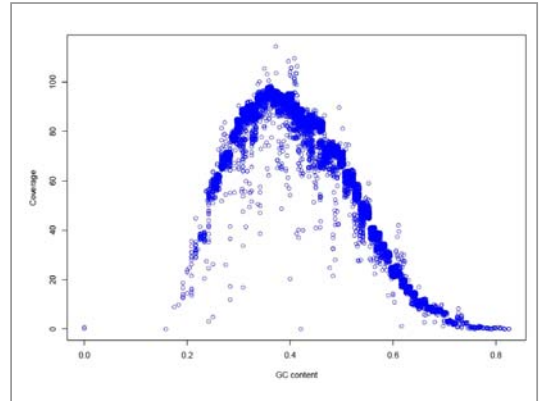


Fig. 2: Coverage versus CG content after custom enrichment and sequencing.

FUNDED PROJECTS

Project Title: Detección de la selección natural en genes candidatos: aproximación a las bases de la adaptación humana.

Financed by: Subdirección General de Proyectos de Investigación (BFU2008-01046)

Years: 2009-2011

PI: Elena Bosch

Project Title: Grup de Recerca Consolidat-SGR

Financed by: Generalitat de Catalunya (2009 SGR-1101)

Years: 2009-2013

PI: Jaume Bertranpetit

Project Title: Local Adaptation of Modern Humans to Micronutrient Deficiencies

Financed by: Volkswagenstiftung (Az: I/85 198)

Years: 2010-2012

PIs: Elena Bosch and Mark Stoneking

GROUP

HUMAN GENOME DIVERSITY



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Begoña Martínez-Cruz

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Laura Rodríguez-Botigué, PhD Student | FI Scholarship

Marc Haber, PhD Student | International Scholarship

Paula Sanz, Technician | UPF Contract

Michael Ducore, Master Student

Arturo Sylveira, Master Student from UB

RESEARCH OUTLINE

Our group's research is focused on the understanding of the current genomic diversity in human populations in order to establish the mechanisms, causes and consequences of this genetic variation. Concerning population processes, the research of our group has focused on the analysis of uniparental markers (Y-chromosome and mitochondrial DNA), as well as genome-wide autosomal markers to answer several questions about the origins of human populations. Within this framework, we have addressed questions about the population history of Central African groups, the peopling of Europe, or the diversity within North African populations. Concerning genomic processes, our research has focused on the analysis of the diversity of genes involved in complex traits and complex diseases in order to describe the standard diversity in healthy individuals and to unravel its implications in disease and phenotypes.

RESEARCH SUBLINES

1. The Genographic Project

This is a project launched by National Geographic and IBM which aims to unravel the migration history of the human species through genetic markers. We are involved in the analysis of uniparental genomes in Western and Central Europe.

2. Genetic history of North African human populations

We try to establish the evolutionary processes (demography and adaptation) that have modelled the genetic diversity of human populations in these geographical areas.

3. Genetic variants associated to phenotypes in human populations

Our goal is to describe those genetic variants related to distinct phenotypes, such as height and skin color, taking certain human populations as a model.

PUBLICATIONS 2011

ISI Articles

- Batini, C., Ferri, G., Destro-Bisol, G., Brisighelli, F., Luiselli, D., Sánchez-Diz, P., Rocha, J., Jorde, L.B., Brehm, A., Montano, V., Elwali, N.E., Spedini, G., d'Amato, M.E., Myres, N., Ebbesen, P., Comas, D., and Capelli, C. 2011. Early Y chromosome lineages in Africa: the origin and dispersal of *Homo sapiens*. *Molecular Biology and Evolution* 28: 2603-2613.
- Batini, C., Lopes, J., Behar, D.M., Calafell, F., Jorde, L.B., van der Veen, L., Quintana-Murci, L., Spedini, G., Destro-Bisol, G., and Comas, D. 2011. Insights into the demographic history of African Pygmies from complete mitochondrial genomes. *Molecular Biology and Evolution* 28: 1099-1110.
- Fadhlouzi-Zid, K., Martínez-Cruz, B., Khodjet-el-hkil, H., Mendizabal, I., Benammar-Elgaaied, A., and Comas, D. 2011. Genetic structure of Tunisian ethnic groups revealed by paternal lineages. *American Journal of Physical Anthropology* 146: 271-280.
- Fadhlouzi-Zid, K., Rodríguez-Botigué, L., Naoui, N., Benammar-Elgaaied, A., Calafell, F., and Comas, D. 2011. Mitochondrial DNA structure in North Africa reveals a genetic discontinuity in the Nile Valley. *American Journal of Physical Anthropology* 145: 107-117.

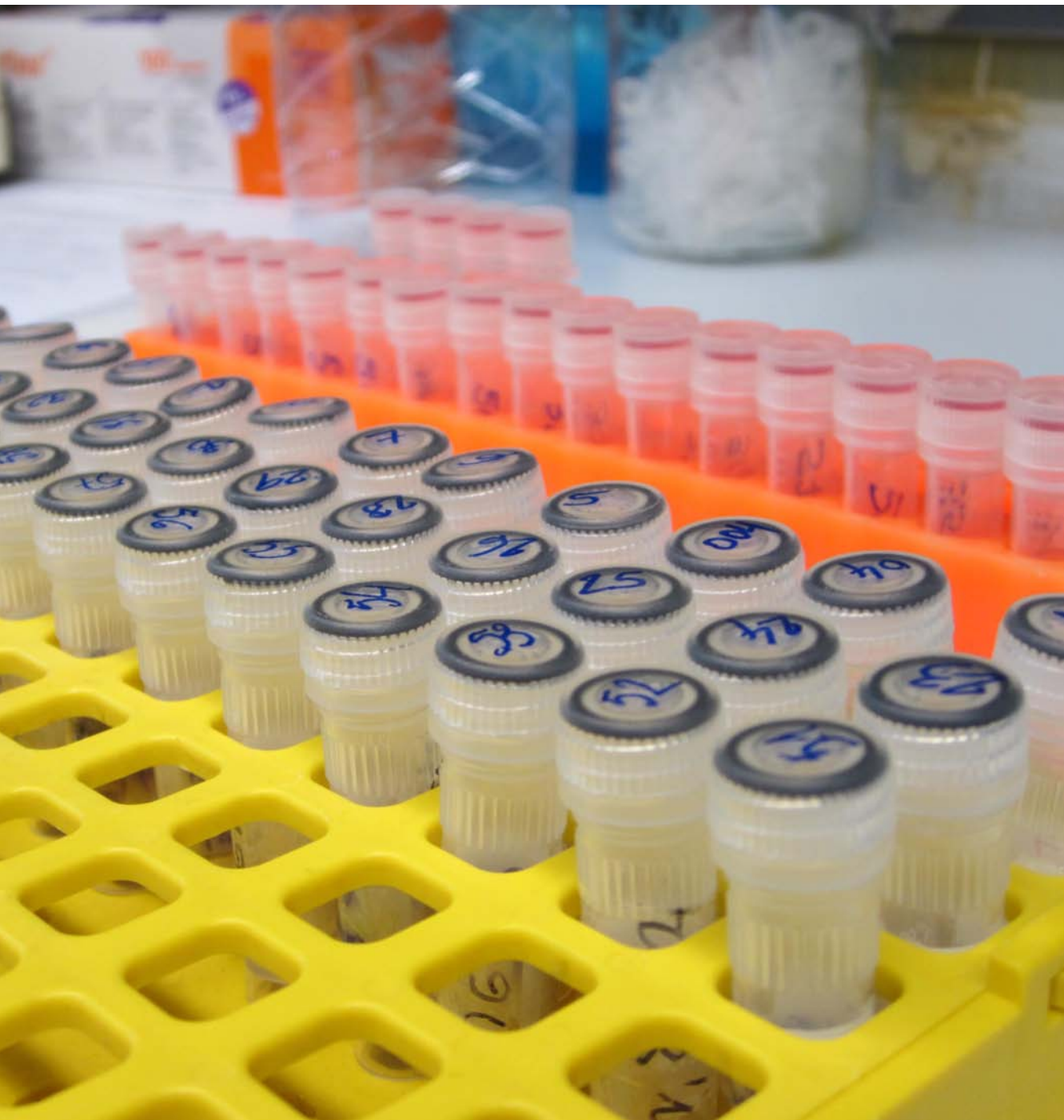


Fig. 1: Human DNA population samples.

- Henn, B.M., Gignoux, C.R., Jobin, M., Granka, J.M., Macpherson, J.M., Kidd, J.M., Rodríguez-Botigué, L., Ramachandran, S., Hon, L., Brisbin, A., Lin, A.A., Underhill, P., Comas, D., Kidd, K.K., Parham, P., Norman, P.J., Bustamante, C.D., Mountain, J.L., and Feldman, M.W. 2011. African Hunter-Gatherer Populations Maintain the Highest Levels of Human Genomic Diversity. *Proceedings of the National Academy of Sciences USA* 108: 5154-5162.
- Laayouni, H., Montanucci, L., Sikora, M., Melé, M., Dall'Olio, G.M., Lorente-Galdós, B., McGee, K.M., Graffelman, J., Awadalla, P., Bosch, E., Comas, D., Navarro, A., Calafell, F., Casals, F., and Bertranpetit, J. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS ONE* 6(3): e17913.
- Martínez-Cruz, B., Ziegler, J., Sanz, P., Sotelo, G., Anglada, R., Plaza, S., Comas, D., and Genographic Consortium. 2011. Multiplex screening of the human Y chromosome using TaqMan probes. *Investigative Genetics* 2(1): 13.
- Mendizabal, I., Valente, C., Gusmão, A., Alves, C., Gomes, V., Goios, A., Parson, W., Calafell, F., Álvarez, L., Amorim, A., Gusmão, L., Comas, D., and Prata, M.J. 2011. Reconstructing the Indian origin and dispersal of the European Roma: a maternal genetic perspective. *PLoS ONE* 6(1): e15988.
- Montano, M., Ferri, G., Marcari, V., Batini, C., Anayale, O., Destro-Bisol, G., and Comas, D. 2011. The Bantu expansion revisited: a new analysis of Y chromosome variation in Central Western Africa. *Molecular Ecology* 20: 2693-2708.
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- Sikora, M., Laayouni, H., Calafell, F., Comas, D., and Bertranpetit, J. 2011. A genomic analysis identifies a novel component in the genetic structure of sub-Saharan African populations. *European Journal of Human Genetics* 19(1): 84-88.

As part of The Genographic Consortium

- Balanovsky, O., Dibirova, K., Dybo, A., Mudrak, O., Frolova, S., Pocheshkhova, E., Haber, M., Platt, D., Schurr, T., Haak, W., Kuznetsova, M., Radzhabov, M., Balaganskaya, O., Romanov, A., Zakharova, T., Soria-Hernanz, D.F., Zalloua, P., Koshel, S., Ruhlen, M., Renfrew, C., Wells, R.S., Tyler-Smith, C., Balanovska, E., and The Genographic Consortium. 2011. Parallel evolution of genes and languages in the Caucasus region. *Molecular Biology and Evolution* 28(10): 2905-2920.
- Cai, X., Qin, Z., Wen, B., Xu, S., Wang, Y., Lu, Y., Wei, L., Wang, C., Li, S., Huang, X., Jin, L., Li, H., and The Genographic Consortium. 2011. Human migration through bottlenecks from Southeast Asia into East Asia during Last Glacial Maximum revealed by Y chromosomes. *PLoS ONE* 6(8): e24282.
- Gaieski, J.B., Owings, A.C., Vilar, M.G., Dulik, M.C., Gaieski, D.F., Gittelman, R.M., Lindo, J., Gau, L., Schurr, T.G., and The Genographic Consortium. 2011. Genetic ancestry and indigenous heritage in a Native American descendant community in Bermuda. *American Journal of Physical Anthropology* 146(3): 392-405.
- Yan, S., Wang, C.C., Li, H., Li, S.L., Jin, L., and The Genographic Consortium. 2011. An updated tree of Y-chromosome Haplogroup O and revised phylogenetic positions of mutations P164 and PK4. *European Journal of Human Genetics* 19(9): 1013-1015.
- Yang, K., Zheng, H., Qin, Z., Lu, Y., Farina, S.E., Li, S., Jin, L., Li, D., Li, H., and The Genographic Consortium. 2011. Positive selection on mitochondrial M7 lineages among the Gelong people in Hainan. *Journal of Human Genetics* 56(3): 253-256.

FUNDED PROJECTS

Project Title: Diversidad genómica en poblaciones humanas del norte de África y en poblaciones vecinas: inferencias sobre la estructura poblacional y migraciones (CGL2010-14944/BOS)

Financed by: Direcció General de Investigació Científica y Tècnica

Years: 2011-2013

PI: David Comas

Project Title: Grup de Recerca en Biologia Evolutiva (2009SGR1101)

Financed by: Direcció General de Recerca, Comissionat per a Universitats i Recerca, Generalitat de Catalunya

Years: 2009-2012

PI: Jaume Bertranpetit

Project Title: Genomic diversity of human North African populations and their neighbors: inferring population structure and migrations (I-COOP0018)

Financed by: Ministerio de Ciencia e Innovación. Programa "CSIC para el Desarrollo".

Years: 2011-2013

PI: David Comas

Project Title: The Genographic project: Western/Central Europe region

Financed by: National Geographic and IBM

Years: 2006-2012

PIs: Jaume Bertranpetit and David Comas

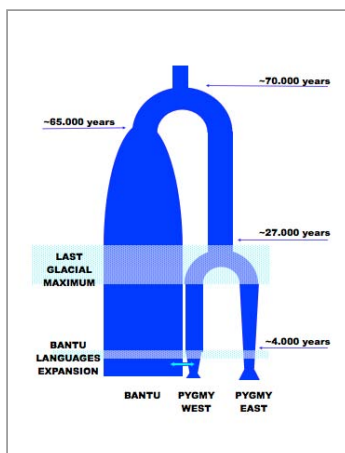


Fig. 2: Demographic inferences from mtDNA complete sequences in African Pygmy populations.

GROUP

micrornas IN HUMAN ADAPTATION AND DISEASE



Ignasi
Torruella

Yolanda
Espinosa-Parrilla

María López

GROUP MEMBERS

Yolanda Espinosa-Parrilla, Group Leader

| Marie Curie Researcher, UPF



María López, PhD Student | FI Scholarship, DGR

Ignasi Torruella Loran | FPI Scholarship, MICINN

RESEARCH OUTLINE

Answering to the question of how genomic diversity in humans has shaped phenotypes that, in the end, are linked to environment adaptation and disease is the general aim of our research. In particular, we are interested in small non-coding RNAs, among them microRNAs, as masters of gene regulation. Each microRNA can control hundreds of target genes building complex molecular pathways and influencing almost every biological process. We aim to understand how microRNAs are integrated into broader networks that, ultimately, are related to human adaptation and development of disease. Our final goal is to contribute to the deciphering of the «why and how» for the existence of such a complex regulatory network that is shaping life in changing environmental conditions. Our interdisciplinary approach combines genetic analysis of DNA sequences with functional studies, linking genomes to life.

RESEARCH SUBLINES

1. Analysis of the genetic and genomic variability in human microRNA-containing regions

The main goal of this research line is to ascertain the relationship between natural selection, structure and biological function in human miRNA diversity trying to answer to questions such as: Which are the general patterns of evolution of microRNA regions? What are the adaptive and functional roles of the evolutionary novelties generated by microRNA diversity?

2. Study of the involvement of microRNA related mechanisms in human disease susceptibility

Given the potential role of microRNAs in homeostatic control, microRNA-dependent regulatory circuits are likely to be important in disease, much of our research is thus aimed to the understanding of how microRNAs are involved in human disease by means of association studies, *in silico* target site and pathway analysis, as well as functional approaches intended to elucidate the participation of microRNAs in specific disease-related pathways.

3. Identification of genetic susceptibility factors in attention-deficit hyperactivity and substance abuse disorders in Chilean populations

This project is focused on the identification of genetic factors involved in attention-deficit hyperactivity disorder (ADHD) and substance abuse disorders in native (Aymara from Arica, North Chile) and recently admixed populations from Chile and the risk conducts associated with this disorder in the adulthood. This research is performed in collaboration with the Universidad de Chile and the Instituto de Alta Investigación de la Universidad de Tarapacá, in Chile.

PUBLICATIONS 2011

ISI Articles

- Miñones-Moyano, E., Porta, S., Escaramís, G., Rabionet, R., Iraola, S., Kagerbauer, B., Espinosa-Parrilla, Y., Ferrer, I., Estivill, X., and Martí, E. 2011. MicroRNA profiling of Parkinson's disease brains identifies early downregulation of miR-34b/c which modulate mitochondrial function. *Human Molecular Genetics* 20(15): 3067-3078.
- Muiños-Gimeno, M.*, Espinosa-Parrilla, Y.*, Guidi, M., Kagerbauer, B., Sipilä, T., Maron, E., Pettai, K., Kananen, L., Navinés, R., Martín-Santos, R., Gratacòs, M., Metspalu, A., Hovatta, I., and Estivill, X. 2011. Human microRNAs miR-22, miR-138-2, miR-148a, and miR-488 are associated with panic disorder and regulate several anxiety candidate genes and related pathways. *Biological Psychiatry* 69(6): 526-33 (*shared first authorship).

Book Chapters

- Espinosa-Parrilla Y, Muiños-Gimeno, M. 2011. MicroRNA mediated regulation and the susceptibility to anxiety disorders. In: *Anxiety Disorders*, 281-306. InTech. ISBN: 978-953-307-592-1.

FUNDED PROJECTS

Project Title: MapbyAdmixtureChI-Mapping Genes involved in Psychiatric Disorders by Admixture Linkage Disequilibrium in Chilean populations (236836)

Financed by: Marie Curie Actions-International Outgoing Fellowships (IOF)

Years: 2009-2012

PI: David Comas

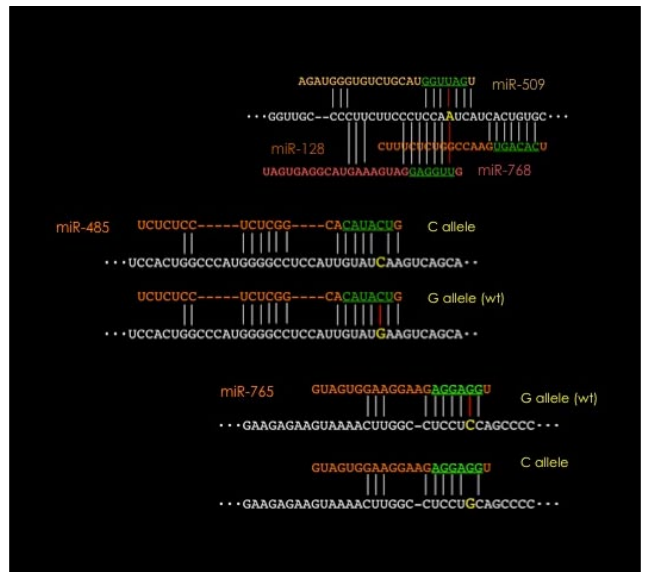
Project Title: Genetic and genomic variability in human microRNA containing regions: towards the identification of functional and evolutionary novelties in microRNAs (BFU2010-18477)

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: Yolanda Espinosa

Fig. 1: RNA sequence alignments for different allelic variants identified in functional miRNA target sites of NTRK3 showing the predicted binding to the corresponding miRNAs.







PROGRAM

complex systems

RESEARCH GROUPS

Complex Systems

Ricard Solé, *Group Leader*

This program, which is still in its initial phase of integration to the IBE, will include the study of both natural and artificial Complex Systems evolution with the aim of finding general organisation rules. Research in this program may expand to a broad range of different systems. Special emphasis is taken in the study of computational biology, protocell biology, synthetic systems and network biology. In particular, in the study of viruses dynamics, tissue architecture, ecological networks, cancer systems, language networks, etc.

GROUP
COMPLEX SYSTEMS



Sergi Valverde
Carlos Rodríguez-Caso
Salvador Durán

Jordi Delgado
Bernat Corominas-Murtra
Núria Conde

Ricard Solé

Javier Macía

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Ricard Solé, Group Leader

| Professor, UPF • ICREA Researcher



- Javier Macía | Post-doc Researcher
- Sergi Valverde | Post-doc Researcher
- Carlos Rodríguez-Caso | Post-doc Researcher
- Bernat Corominas-Murtra | Post-doc Researcher
- Ben Shirt-Ediss | Pre-doc Researcher
- Núria Conde | Pre-doc Researcher
- Salvador Durán | Pre-doc Researcher
- Max Carbonell | Pre-doc Researcher
- Luis Seoane | Pre-doc Researcher

RESEARCH OUTLINE

The Complex Systems Lab is an interdisciplinary research team exploring the evolution of complex systems, both natural and artificial, searching for their common laws of organization. We closely work in collaboration with the Santa Fe Institute (USA) and the European Centre of Living Technology (Italy). Our research spans a broad range of systems, with special attention to biological computation, protocell biology, synthetic systems, and network biology.

RESEARCH SUBLINES

1. Evolution of technology
2. In silico evodevo
3. Synthetic multicellularity
4. Evolution of viruses
5. Language, brain and cognitive networks
6. Order and disorder in cancer
7. Biological computation
8. Evolutionary synthetic biology

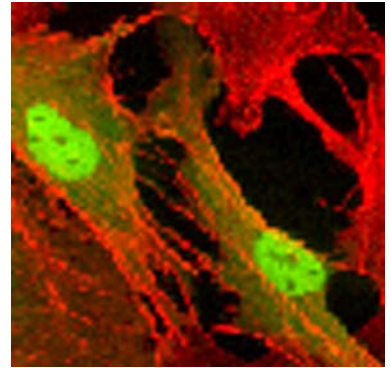


Fig. 1: Cancer cells

PUBLICATIONS 2011

ISI Articles

- Corominas-Murtra, B., Fortuny, J., et al. 2011. Emergence of Zipf's law in the evolution of communication. *Physical Review E* 83: 32767. doi: 10.1103/PhysRevE.83.036115.
- Corominas-Murtra, B., Rodríguez-Caso, C., Goñi, J., and Solé, R. 2011. Measuring the hierarchy of feedforward networks. *Chaos* 21, 016108. doi: 10.1063/1.3562548.
- Flores, C.O., Meyer, J.R., Valverde, S., Farr, L. and Weitz, J.S. 2011. Statistical structure of host-phage interactions. *Proceedings of the National Academy of Sciences* 108(28): E288-297. doi: 10.1073/pnas.1101595108.
- Regot, S., Macia, J., Conde, N., Furukawa, K., Kjellen, J., Peeters, T., Hohmann, S., de Nadal, E., Posas, F., and Solé, R. 2011. Distributed biological computation with multicellular engineered networks. *Nature* 469: 207-211.

FUNDED PROJECTS

Project Title: Origins of innovation in tinkered networks

Financed by: McDonnell Foudation USA (McDonnell Award 220020117)

Years: 2007-2011

PI: Ricard Solé

Project Title: Cellular computation

Financed by: Fundación Marcelino Botín

Years: 2010-2016

PI: Ricard Solé

Project Title: Computación, replicación y rotura de simetría en sistemas protocelulares

Financed by: Ministerio de Ciencia e Innovación (FIS2009-12365)

Years: 2010-2012

PI: Ricard Solé





ISI Articles

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- Abellán, P., and Ribera, I. 2011. Geographic location and phylogeny are the main determinants of the size of the geographical range in aquatic beetles. *BMC Evolutionary Biology* 11: 344.
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- Batini, C., Lopes, J., Behar, D.M., Calafell, F., Jorde, L.B., van der Veen, L., Quintana-Murci, L., Spedini, G., Destro-Bisol, G., and Comas, D. 2011. Insights into the demographic history of African Pygmies from complete mitochondrial genomes. *Molecular Biology and Evolution* 28: 1099-1110.
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- Campbell, K., Whissell, G., Franch-Marro, X., Battle, E., and Casanova, J. 2011. Specific GATA Factors Act as Conserved Inducers of an Endodermal-EMT. *Developmental Cell* 21, 1051-1061.

- Casals, F., Sikora, M., Laayouni, H., Montanucci, L., Muntasell, A., Lazarus, R., Calafell, F., Awadalla, P., Netea, M.G., and Bertranpetit, J. 2011. Genetic adaptation of the antibacterial human innate immunity network. *BMC Evolutionary Biology* 11(1): 202.
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- de Pous, P., Mora, E., Metallinou, M., Escoriza, D., Comas, M., Donaire, D., Pleguezuelos J.M., and Carranza, S. 2011. Elusive but widespread? The potential distribution and genetic variation of *Hyalosaurus koellikeri* (Günther, 1873) in the Maghreb. *Amphibia-Reptilia* 32: 385-397.
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- Ge, D.Y., Chesters, D., Gómez-Zurita, J., Zhang, L.J., Yang, X.K., and Vogler, A.P. 2011. Anti-predator defense drives parallel morphological evolution in leaf beetles. *Proceedings of the Royal Society of London. B* 278: 2133-2141.
- Gómez-Díaz, E., Boulinier, T., Sertour, N., Cornet, M., Ferquel, E., and McCoy, D.K. 2011. Genetic structure of marine *Borrelia garinii* and population admixture with the terrestrial cycle of Lyme borreliosis. *Environmental Microbiology* 13: 2453-2467.
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THE DISCOVERY OF A NEW SPECIES WITHIN A MODEL SYSTEM CHALLENGES OUR KNOWLEDGE ON CRYPTIC DIVERSITY

Describing the diversity of species inhabiting our planet is becoming increasingly difficult because many are becoming endangered and, after hundreds of years of scientific exploration, only the less conspicuous species remain. «Less conspicuous» does not mean few: they seem to be more than the ones already described! «Less conspicuous» does not mean that they only exist in remote exotic locations: they can be right in front of our eyes and we may not recognize them as unique. Indeed, studies using molecular techniques frequently entail discovering that what was believed to be a single species actually comprises several mutually isolated populations that cannot interbreed or exchange genes. Uncovering cryptic biodiversity is essential for understanding evolutionary processes and patterns of ecosystem functioning, as well as for nature conservation.

To know the real extent of biodiversity on Earth we need not only to finish the task of studying poorly explored taxa and regions, but also to figure out what we are missing in the already studied groups. How to quantify our own ignorance of the hidden diversity? In this study we have discovered a cryptic species within a long-studied model for speciation, a result that challenges our current knowledge and shows that deeper unexpected layers of diversity exist.

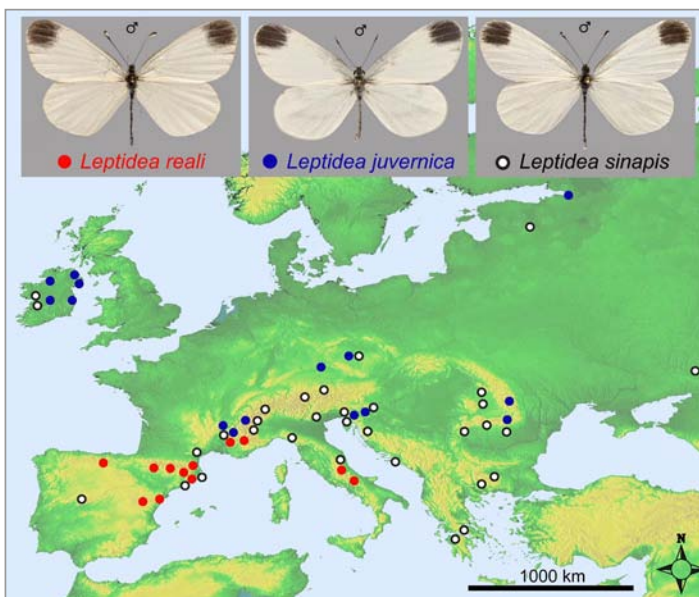


Fig. 1: The triplet of cryptic *Leptidea* species and their distribution as determined by molecular, karyological and morphometric analyses. *Leptidea reali*, described twenty years ago, was one of the first cases of cryptic species in butterflies. *Leptidea juvernica* is the cryptic species discovered in this study.

As European butterflies are arguably the best-studied group of invertebrates in the world, twenty years ago the discovery of a cryptic species within the common wood white butterfly *Leptidea sinapis* was a significant event, and these butterflies have quickly become a model to study speciation. Here we show that what has been formerly called the cryptic species pair *L. sinapis* – *L. reali* actually comprises a triplet of species. The new species can be discriminated on the basis of either DNA or karyological data, but it is morphologically indistinguishable. All previous evidence published on the *Leptidea* system needs to be reassessed under the light of the proposed classification, even more so when these taxa are widely distributed across the Palearctic region. Implications are multiple and range from aspects related to conservation to speciation and ecological differentiation.

In conclusion, we show that assessing cryptic diversity is a challenging task even in well-studied groups of organisms. Our finding exemplifies that cryptic biodiversity may consist of finely nested layers and highlights the importance of using an array of techniques when dealing with closely related species. This often involves the reconsideration of a large amount of previous data and/or hypotheses, and can have profound repercussions for various fields of research related to the target organisms.

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MATRILINEAL KINSHIP AND DIVERSITY WITHIN A NEANDERTAL FAMILY GROUP

The demography –the study of survival, fertility and population dynamics– is crucial for understanding human evolution. Two circumstances concur at El Sidrón cave in Asturias (Spain) that make this site unique for studying the demographic structure of a Neandertal group: the synchrony of the accidental assemblage of twelve Neandertal individuals and the exceptional taphonomic conditions favouring DNA preservation of all of them.

The El Sidrón Neandertal assemblage includes six adults (three males and three females) and six subadult individuals (three adolescents, two juveniles and one infant). Sampling the dentition, which was used as a morphological marker for individual attribution, it was possible to retrieve mitochondrial DNA from all of them, thus making El Sidrón the best represented site in terms of interindividual genetic preservation.

Seven out of ten individuals in the El Sidrón group carry the same mitochondrial DNA haplotype, suggesting that Neandertal groups were kinship-structured. The three adult females have different mitochondrial haplotypes, while the three adult males have identical haplotypes. This suggests the existence of female exogamy (also known as patrilocality) among Neandertal groups. This reproductive behaviour is expected to generate a wide dispersal of female-specific genetic markers such as the mtDNA, which is in agreement with the lack phylogeographic structure observed among Neandertals' mtDNA. About 70% of modern hunter-gatherer groups, as well as many traditional societies, also practise this kind of mating behaviour. Considering the age and the sharing of specific mitochondrial DNA lineages,

it was possible to estimate an interbirth interval of about 3 years. In modern hunter-gatherer groups, a similar value of interbirth interval of 3.4 years has been observed.

From these data, some demographic characteristics, such as small group sizes, patrilocal mating behaviour and long interbirth intervals, can be inferred for Neandertal groups. Some of these traits could have played a role in evolutionary processes such as interbreeding with modern humans or their final extinction about 30,000 years ago.

Reference Article

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Fig. 1: "El Sidrón" individual dentitions.

COMPARATIVE AND DEMOGRAPHIC ANALYSIS OF ORANGUTAN GENOMES

The complete genomic sequence of the «man of the forest» (according to the Malay and Indonesian words «Orang» and «Hutan»), was published on January this year (2011). Under the perspective of human evolution, the orangutan genome has proceeded much more slowly than other great apes. This is evidenced by fewer rearrangements, less segmental duplications, a lower rate of gene family turnover and surprisingly quiescent Alu repeats, which have played a major role in restructuring other primate genomes.

Besides of the female Sumatran (*Pongo abelii*) individual used to reconstruct the reference genome (Susie), five Sumatran and five Bornean (*Pongo pygmaeus*) orangutan genomes were also sequenced and analyzed at a less coverage. The analysis showed increased genetic diversity across Sumatran and Bornean orangutans compared to other great apes. This is one of the first papers to contribute to the new expanding field of population genomics.

Reference Article

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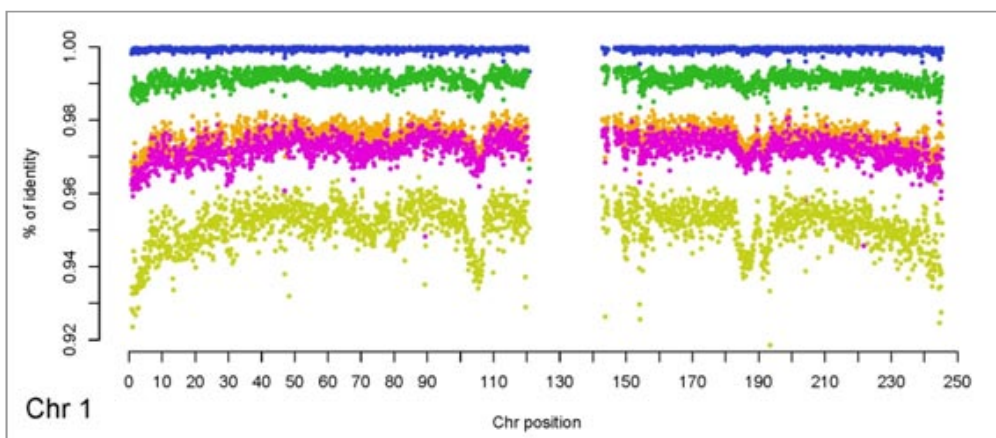


Fig. 1: Percentage of Identity of whole genome sequence data from all apes and macaque compared to the chromosome 1 of the human reference genome. In blue, human data; in green, chimpanzee; in orange, orangutan; in purple, gibbon, and in light green, macaque.

DISTRIBUTED BIOLOGICAL COMPUTATION WITH MULTICELLULAR ENGINEERED NETWORKS

Applied biotechnological designs necessarily belong to complex circuits, and one important challenge is how to create multi-purpose engineered components for a wide range of functional tasks. As standardization of basic components of molecular toolkits proceeds, new designs become possible in a reliable way. Engineering the appropriate cell circuitry is a major bottleneck in pushing synthetic circuits towards decision-making biological devices. In this context, modularization under a multicellular framework is a natural form of using whole cell types as basic modules performing simple computations. In this paper we explore both theoretically and experimentally the use of distributed computing. Distributed computation offers a shortcut towards achieving complex computations under the non-standard assumption of a multiple-output cell library with new capabilities. The circuits based on distributed computing can easily be reprogrammed by replacing few cell types by others or modifying the specific logic function that a given cell type implements by adding external effectors such as inhibitors. Reprogramming cellular circuits through external molecules is appealing because allows interaction with potential circuits located in non-accessible places, such as in several biomedical applications. Moreover, the multi-cellular approach allows for reutilization of components without additional engineering. These remarkable traits might be an important component of the future wave of synthetic designs, making possible to construct a wide range of diverse circuits.

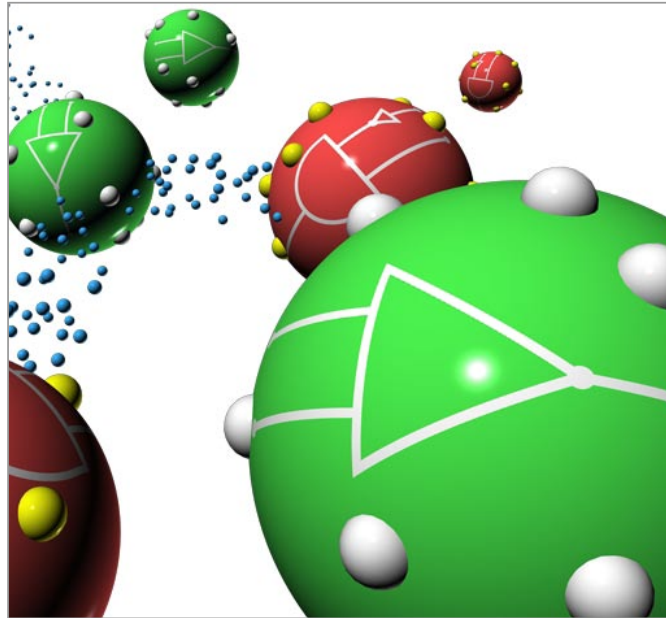


Fig. 1: Distributed computation based on the co-existence of different cellular types implementing a simple boolean function. Cells are connected with small diffusible molecules secreted performing the final computation.

Reference Article

- Regot, S., Macia, J., Conde, N., Furukawa, K., Kjellen, J., Peeters, T., Hohmann, S., de Nadal, E., Posas, F., and Solé, R. 2011. Distributed biological computation with multicellular engineered networks. *Nature* 469: 207-211.



OTHER ACTIVITIES

DOCTORAL THESES PRESENTED DURING 2011

PhD Student: Marta Melé

Title: Incorporating Recombination into the study of recent Human Evolutionary History

Thesis Directors: Jaume Bertranpetit and Francesc Calafell

Institution and Date: Universitat Pompeu Fabra, 29th March 2011

PhD Student: Vlad Eugen Dinca

Title: Diversity, Biogeography and chromosomal evolution in European Butterflies

Thesis Director: Roger Vila

Institution and Date: Universitat Autònoma de Barcelona, 1st April 2011

PhD Student: Elena Gigli

Title: Evolutionary Genetics of Homo Neanderthalensis: adaptative traits and methodological problems

Thesis Director: Carles Lalueza-Fox

Institution and Date: Universitat Pompeu Fabra, 20th June 2011

PhD Student: Bernat Coromines

Title: A unified approach to the emergence of complex communication

Thesis Director: Ricard Solé

Institution and Date: Universitat Pompeu Fabra, 12th July 2011

PhD Student: Ferran Borràs

Title: Caracterització dels factors de transcripció E74 i Seven Up en el desenvolupament de l'insecte hemimetàbol *Blattella germanica* (L.) (Dictyoptera, Blattellidae)

Thesis Directors: Xavier Bellés and David Martín

Institution and Date: Universitat de Barcelona, 26th October 2011

PhD Student: Belén Lorente Galdós

Title: The action of natural selection in recently duplicated genes

Thesis Directors: Tomàs Marquès-Bonet and Arcadi Navarro

Institution and Date: Universitat Pompeu Fabra, 11th November 2011

PhD Student: Carlos Morcillo Suárez

Title: Analysis of genetic Polymorphisms for statistical genomics: tools and applications

Thesis Director: Arcadi Navarro

Institution and Date: Universitat Pompeu Fabra, 19th December 2011

PhD Student: Víctor Soria

Title: Análisis globales de diversificación en mamíferos a partir de filogenias moleculares

Thesis Director: José Castresana Villamor

Institution and Date: Universitat de Barcelona, 19th December 2011

IBE SEMINARS 2011

Speaker	Talk	Institution	Date
Rolf Beutel	Recent progress in the phylogeny of holometabola	Institute of Systematic Zoology and Evolutionary Biology with Phyletic Museum (Jena, Germany)	31/01/2011
Elodie Gazave	The challenge of modeling the recent human population genetics and its implication for human health	Department of Biological Statistics and Computational Biology, Cornell University (Ithaca, NY)	07/02/2011
Ana Rivero	Pleiotropic effects of insecticide resistance on disease transmission	MIVEGEC, CNRS (Montpellier, France)	14/02/2011
Carles Vila	Genetic tools for the conservation of Iberian wolves: assessment of attacks and hybridization	Estación Biológica de Doñana, CSIC (Doñana, Spain)	28/02/2011
Jordi Casanova	From genes to shape: notions from the study of the <i>Drosophila</i> tracheal system	Institute for Research in Biomedicine [IRB], CSIC (Barcelona, Spain)	28/03/2011
Marco Milán	Growth control in <i>Drosophila</i>	Institut de Recerca Biomèdica [IRB] (Barcelona, Spain)	11/04/2011
Pablos Pavlidis	Detecting positive selection in natural populations	Scientific Computing Group [Exelixis Lab & HPC Infrastructure], Heidelberg Institute for Theoretical Studies [HITS gGmbH] (Heidelberg, Germany)	31/05/2011
Jordi Camí	Código de buenas prácticas científicas	Parc de Recerca Biomèdica de Barcelona [PRBB] (Barcelona, Spain)	15/06/2011

Speaker	Talk	Institution	Date
Rosa Garriga	Conservació integral a l'Àfrica: el santuari de ximpanzés de Tacugama a Sierra Leone	Tacugama Chimpanzee Sanctuary (Sierra Leone)	19/07/2011
Antonio Marco	Functional shifts during microRNA evolution	Faculty of Life Sciences, University of Manchester (UK)	27/07/2011
How-Jing Lee	Circadian regulation on oxidative stress of the German cockroach	Department of Entomology, National Taiwan University (Taiwan)	13/10/2011
Javier Herrero	50 vertebrate genomes in Ensembl	EMBL-EBI (Cambridge, UK)	25/10/2011
Daniel Villatoro	Beyond the carrot and stick in self-organized virtual societies	Instituto de Inteligencia Artificial [IIA], CSIC (Spain)	02/11/2011
Richard Ebstein	A molecular genetics approach towards understanding human altruism	Psychology Department of the Faculty of Arts and Sciences, National University of Singapore (Singapore)	18/11/2011
Marc Vía	Admixture in human populations from Latin America: genetic, demographic, and social implications	Departament de Biologia Animal, Universitat de Barcelona (Barcelona, Spain)	30/11/2011

TEACHING

IBE Scientists belonging to the Pompeu Fabra University are also academic staff at this University (CEXS Department: Evolutionary Biology Program) and are in charge of the coordination and main teaching of several academic subjects in undergraduate degrees and master programs, as follows.

GRADUATE STUDIES

- | Bachelor's degree in Human Biology (Pompeu Fabra University)
 - Human Evolution and Health (4 ECTS).
Coordinators: Elena Bosch and David Comas.
 - Zoology (4 ECTS). Coordinator: Óscar Ramírez.
 - Ecology (4 ECTS). Coordinator: Francesc Calafell.
 - Integrated Biomedicine I (4 ECTS). Coordinator: David Comas.
 - Integrated Biomedicine II (4 ECTS). Coordinator: David Comas.
 - Basic Sciences 1 (7 ECTS). Coordinator: Ricard Solé.
 - Genomics (4 ECTS). Coordinator: Jaume Bertranpetit.

- | Bachelor's degree in Medicine (Pompeu Fabra University)
 - Human Evolution and Health (4 ECTS).
Coordinators: Elena Bosch and David Comas.

- | Bachelor's degree in Humanities. Complementary degrees and courses Program.
 - Biological Understanding of Humans (4 ECTS).
Coordinator: David Comas.

MASTER STUDIES

- | Màster en Recerca Biomèdica (BIOMED) (Pompeu Fabra University)
 - Genomes and Systems (5 ECTS). Coordinator: Tomàs Marquès-Bonet.
 - Introduction to Biomedicine (5 ECTS). Coordinator: David Comas.

- | Master in Bioinformatics for Health Sciences (BIOINFO). Joint master of the Pompeu Fabra University (coordination) and University of Barcelona, in cooperation with the Bologna University.
 - Analysis of Biomedical Data (5 ECTS). Coordinator: Arcadi Navarro.
 - Biomedical Informatics (5 ECTS). Coordinator: Arcadi Navarro.
 - Introduction to Biomedicine (5 ECTS). Coordinator: David Comas.

Furthermore, most IBE scientists actively collaborate in several Undergraduate and Master programs and also in specialized courses of different Spanish universities. Either by offering students practical training through research stays or by giving specialized lectures, as summarized in the following Table.

Subject	In: Master	University	Teacher
Genetic variation dynamics in populations In: Genomes and Systems	Master in Biomedical Research (BIOMED)	Pompeu Fabra University (UPF)	Elena Bosch
Statistics in forensic genetics In: Grounds in Forensic Anthropology	Master in Human Biology	UB / UAB	Francesc Calafell
Probability calculations in STRs In: Molecular analysis and statistics	Master in Human Biology	UB / UAB	Francesc Calafell
Transmission Disequilibrium Test In: Biomedical Informatics	Bioinformatics and Health Sciences (BIOINFO)	UPF / UB / Bologna University	Francesc Calafell
Pharmacogenomics In: Biomedical Informatics	Bioinformatics and Health Sciences (BIOINFO)	UPF / UB / Bologna University	Francesc Calafell
Linkage Disequilibrium In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Francesc Calafell
Genowide Association Studies In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Francesc Calafell
The genomic view into the genetic bases of diseases In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Francesc Calafell
Genome variation: the measure of the genetic variation at the DNA level In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	David Comas
Population genetics In: Genetic Epidemiology	Master on Public Health (UPF)	UPF	David Comas
Anàlisi genètica In: Desenterrant silencis; les fosses de la guerra civil	Curs Universitari d'Estiu	UAB	Carles Lalueza
Evolución Humana	Comunicación Científica, Médica y Ambiental	IDEC-UPF	Carles Lalueza

Subject	In: Master	University	Teacher
Paleogenomics In: Genomes and Systems	Màster de Recerca Biomèdica (BIOMED)	UPF	Carles Lalueza
Mobile Elements and Genome Evolution In: Genomes and Systems	Màster de Recerca Biomèdica (BIOMED)	UPF	Josefa González
Mobile Elements and Genome Evolution In: Molecular Evolution	Biologia del Desenvolupament i Genètica	UB	Elena Casacuberta
Introduction to genomic variation In: Genomes and Systems	Màster de Recerca Biomèdica (BIOMED)	UPF	Jaume Bertranpetit
Capturing the global human variation In: Genomes and Systems	Màster de Recerca Biomèdica (BIOMED)	UPF	Jaume Bertranpetit/ Tomàs Marquès-Bonet
El Modelo ICREA	Màster en Lideratge i Gestió de la Ciència i la Innovació	IDEC-UPF	Jaume Bertranpetit
Alineamiento múltiple	Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones	UB	José Castresana
Genetic drift and its effects In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Hafid Laayouni
Selection In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Arcadi Navarro
The non-coding genome In: Genomes and Systems	Master in Biomedical Research (BIOMED)	UPF	Yolanda Espinosa
Origin of Bilaterian animals	Master in Biodiversity	UB	Salvador Carranza
Topology tests	Master in Biodiversity	UB	Salvador Carranza
Molecular Phylogenetics	Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones	UB	Salvador Carranza

Subject	In: Master	University	Teacher
Expresión génica y su regulación: Potencial diagnóstico y pronóstico de los MicroRNAs en enfermedad	Màster en Assessorament Genètic	Institut d'educació contínua de la Universitat Pompeu Fabra	Yolanda Espinosa
Analysis of the genetic variability in microRNA-mediated regulation and the susceptibility to psychiatric disorders In: Seminarios de Investigación del módulo Intensificación en genética 2	Máster interuniversitario en Biología Molecular, Celular y Genética	UV/UJI	Yolanda Espinosa



RETREAT CHRONICLE

On the 21st and 22nd of March 2011, the members of IBE convened at Sant Feliu de Guíxols for their Annual Retreat. This meeting provides a unique opportunity for all IBE members to network, share their scientific experiences, and acquire a detailed overview of the scientific activities at IBE.

The scientific program included four main lectures meant to introduce the main research projects of several IBE groups. Josefa González (Evolutionary and Functional Genomics Lab) talked on «Adaptation: molecular mechanisms and functional consequences»; Javier Macía (Complex Systems Lab) presented «Distributed computation with multicellular engineered networks»; Tomàs Marquès (Primate Genomics Lab) introduced «Three lessons I learnt from 2010»; and finally Roger Vila (Butterfly Diversity and Evolution Lab) presented «Straddling a Wellsian time-machine with Nabokov».

Alternated among the main lectures some inter- and intra-program network activities took place. The inter-program activities were coordinated by Tomàs Marquès-Bonet and Javier Macía, and involved several predoctoral researchers presentations on the latest technical developments and their possible applications to evolutionary biology.

The intra-program activities were coordinated by Josefa González and Yolanda Espinosa, and consisted in a selection of short presentations by a couple of postdoctoral fellows from each program, describing different lines and research approaches.

By the end of the first intense day, Francesc Calafell presented the results of a phylogenetic study of mitochondrial DNA lineages of IBE members started in December of previous year with the recollection of saliva samples from IBE donors. The results of this study are presented in the next pages of this report.

The isolated, calm atmosphere at the Eden Roc Hotel propitiated a lively exchange of ideas, both in the formal scientific program and in the social activities that, although not formally coordinated, were a real success of attendance and fun during the night of the 21st.

The Annual Retreat succeeded in creating a venue in which communication flowed freely and the IBE fully functioned as a cohesive unit.

METAEVOLUTION: POPULATION GENETIC ANALYSIS OF A SAMPLE OF EVOLUTIONARY BIOLOGISTS

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INTRODUCTION

Mitochondrial DNA (mtDNA) has some particular properties that have turned it into the main workhorse in population genetics and phylogenetics, in spite of the efforts by Melé et al. (2010). It does not recombine (any evidence to the contrary has been properly ignored), and it is transmitted exclusively through the maternal line (ditto). It is jam-packed with genes, except for the so-called control region where, actually, mutation runs out of control. And within the control region, two segments, hypervariable segments (HVS) I and II, are even more variable. The accumulation of genetic diversity in that region provides an optimal bang for the buck: by sequencing just 360 bp in HVSI, many different sequences can be found in a population sample. Given the tree-like accumulation of mutations and the vast literature accrued on the subject, a phylogeography has been constructed for human mtDNA. A phylogenetic tree can be constructed with the > 8,000 mtDNA molecules that have been entirely sequenced, and groups of sequences (the famous *haplogroups*) can be defined from the main branches of the tree (or from the smallest twigs, if that suits your pet population). Haplogroups tend to have restricted geographical ranges; for instance, haplogroups L0, L1, L2, L3, L4, L5, and L6 comprise >99% of African mtDNAs and are rare elsewhere (except, of course, in Afro-Americans, Afro-Caribbeans, and UK Afro-Caribbeans). Same can be said of H, I, J, K, T, U, W, and X in Europe and whereabouts (North Africa and the Middle East), or of A, B, C, D and X in Native Americans. Haplogroup nomenclature follows a hierarchical convention based on the tree structure: J1 is nested within J, J1a within J1, and so on; a star (H*, for instance), denotes a paragroup: sequences within a haplogroup that cannot be allocated to any of its subhaplogroups. And, given that mtDNA does not recombine, haplogroups can be often recognized in mtDNA just by the mutations they carry in the control region. Thus, sequencing HVSI, information can be gathered on the maternal lineage of the subject and its geographic and historic origins. Collectively, a population sample can be compared to others to explore its demographic history, in terms of population size change and migration patterns. In this paper, we have embarked in a bit of navel-gazing by sequencing mtDNA HVRI in >80 members of the Institute of Evolutionary Biology (IBE), comparing their sequences to European databases, and having a hearty laugh at the possible implications of the results.

MATERIALS AND METHODS

Samples

Buccal swabs were obtained from 86 members of the IBE staff gathered for the 2010 Christmas party. Informed consent was obtained, but, given the Christmas spirit (if not yet the Christmas spirits), consent is dubious at best. Thirty-nine individuals originated from Catalonia, one from Majorca, one from Valencia, four from the Basque Country, and 18 from elsewhere in Spain. Additionally, 12 individuals were from other European countries (four from Italy, two from Portugal, and six from six other countries), two from North Africa, one from East Asia, five from North America (including Mexico), and three from South America. This ragtag collection of a sample would never be acceptable in a human population genetics sample, although some forensic genetics papers do report resident or cosmopolitan samples. In our case, the Cosmopolitan sample is limited to subjects MMM and MBL. To the best of our dimwit knowledge, all individuals in the sample are not biologically related to each other (no, the mentor-pupil relation doesn't count).

Genetic analysis

DNA was extracted with the addictive phenol-chlorophorm method. A 360-bp fragment (from nucleotide positions 16024 to 16383) was PCR-amplified and Sanger-sequenced as in Bosch et al. (2006). Actually, the amplified fragment was longer, and we deigned to look beyond 16383 when we knew something should be there.

Statistical analysis

Haplotypes were classified into haplogroups by eyeballing them; we mean, by recognizing the HVSI motifs most recently compiled by van Oven and Kayser in the Phylotree database (www.phylotree.org) (van Oven and Kayser, 2009). Sequences were classified into the deepest phylogenetic level recognizable from HVSI variation, although we also used the HVSI sequences available for a few individuals (compare HL and RAR in Table 1, for instance). Matches were sought in a database we compiled containing sequences from 100 published papers on 22,807 individuals from Europe, W. Asia and N. Africa. This database is available from the authors for a negotiable number of coauthorships. Basic descriptive statistics were generated with Arlequin 3.5 (Excoffier and Lischer, 2010), as well as Analysis of Molecular Variance (AMOVA) statistics (Excoffier et al., 1992). A median-joining network (Bandelt et al., 1999) was produced with Network 4.6.1.0 (www.fluxus-engineering.com), by padding sequences with the coding-sequence variation that can be inferred from their haplogroups (as collected in www.phylotree.org); otherwise, the network would have been an even uglier mess. Just because more people are familiar with trees, we drew a neighbor-joining tree (Saitou and Nei, 1987) with Mega 5.1 (www.megasoftware.net), again padding the sequence with the inferred control-region variation. The IBE sample was compared with other populations by means of an *F*_{st} distance that was plotted with multidimensional scaling using Statistica 10.

RESULTS AND DISCUSSION

In a sample of 86 IBE members, 58 different sequences were found, with 61 segregating sites. Haplotype diversity was 0.9614 (which means that we could identify an IBE member from their mtDNA sequence with a probability of 96.14%), and, on average, pairs of IBE members were different at 4.097 sites. These are perfectly normal, boring, plain-vanilla figures for a European population. Sequences and their haplogroup allocations can be found in Table 1. Thirteen (http://en.wikipedia.org/wiki/Olivia_Wilde) individuals carried sequences that seem to be described for the first time; they had no matches in our database, and we could not find them in public databases for other continents. However, all of those sequences were different in just one or two nucleotide positions from known sequences, so no, those aren't mistakes and, again, this is one of the wonders of mtDNA: over 2,000 Iberians sequenced, and we still find new sequences in a stretch of 360 bp. The median-joining network and neighbor-joining tree are in Figures 1 and 2, respectively. In both, sequence NE stands out by a mile, since it belongs to haplogroup L2, found close to the root and prevalent in Subsaharan Africa, and, at lower frequencies, in N Africa and the Middle East. For both figures, close-ups excluding NE are provided. The main use of both figures is finding who your mitochondrial buddies are; for instance, JI and VS share both an office and a mtDNA sequence (and something else?).

Haplogroup frequencies are in Table 2, compared with those in Iberia and Europe. Given the cosmopolitan nature of the IBE sample, the L, A and B haplogroups were expected (A and B are found in E Asia and the Americas). OV, who carries an A sequence, is of Catalan descent, but he seems to have something of an epicanthic fold. On the contrary, the HV0 haplogroup seems to be under-represented (2.33% vs. 7.18% in Iberia). This haplogroup is more frequent among Basques, and given their representation among IBE members, this is slightly unexpected. Sixteen (18.6%) of the IBE members carry the boring CRS sequence, that is, the same sequence that was initially described for the human mtDNA and that is found in 16.6% of all Iberians or 14.0% of Europeans.

We subdivided the individuals by IBE scientific program, including the administrative staff as a sixth program. AMOVA showed that the fraction of genetic variation explained by differences among programs is -1.15% ($p=0.871$; yes, given its weird sample size correction, you can have negative fractions in AMOVA). Thus, we can reject the hypothesis that some programs have freak genetic makeups to match their research interests. Although not significant, the

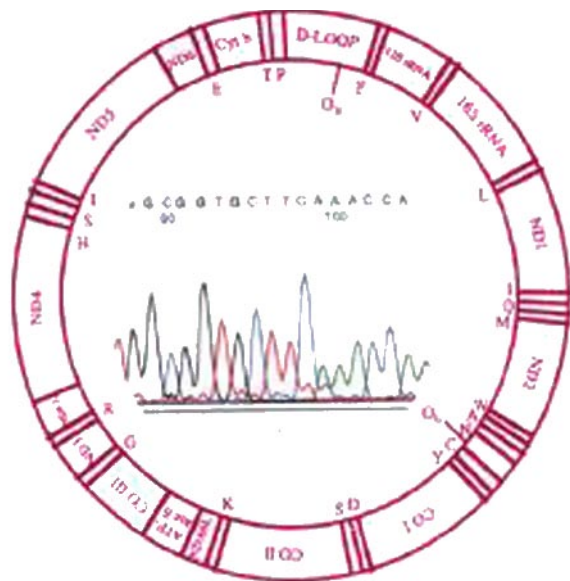


Fig. 1: Scheme of mitochondrial DNA genetic structure.

genetic difference between buildings (CMIMA vs. PRBB) was larger (0.52%, $p=0.200$), which, if confirmed with a larger sample, would mean that the buildings act as reproductive barriers. No interbuilding matings have been observed, while intrabuilding matings are common. Finally, if IBE members were grouped by status (PIs / postdocs / graduate students / technicians / administrative personnel), the fraction of genetic variation explained was -0.80% ($p=0.768$). Thus, it can be interpreted that PIs are not bluebloods.

Finally, we compared the IBE sample to an assortment of European populations with an MDS plot of the F_{st} genetic distances (Figure 3). The IBE sample appeared in a central position, as expected given its mixed origin, but was closest to Catalonia, Valencia and Andalusia, but specially to (gasp!) Sicily. We can hypothesize, then, that some mafia-like behavioral patterns by IBE members may have a genetic base.

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- Bosch, E., Calafell, F., Gonzalez-Neira, A., Flaiz, C., Mateu, E., Scheil, H.G., Huckenbeck, W., Efremovska, L., Mikerezi, I., Xirontiris, N., Grasa, C., Schmidt, H., and Comas, D. 2006. Paternal and maternal lineages in the Balkans show a homogeneous landscape over linguistic barriers, except for the isolated Aromuns. *Annals of Human Genetics* 70: 459-87.
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Table 1. Haplotypes found in their samples and their inferred haplogroups and matches in the database described in the text. 16000 should be added to all figures to recover the original position.

Haplogroup	Haplotype	Matches	Samples
L2a1*	189, 192, 223, 243, 278, 284, 294, 298, 309, 390	None	NE
L3b	124, 223, 278, 362	1.8% of Sub-saharan Africans	ANO
A4a1	223, 249, 290, 319, 362	3 Hungarians	OV
I*	129, 223, 391	0.5% of Europeans	NP
I1	129, 223, 264, 270, 311, 319, 362	3 Georgians, 1 Andalusian, 1 Xueta, 1 Greek, a few Ashkenazi Jews... and one 3,000-year-old Neolithic skeleton from Granollers!	FC
B4a*	129, 189, 217, 261, 356	1 in Hong Kong, 1 in Chinese-American	J-HH
B4*	189, 217, 270A, 316	None	FS
B4*	189, 217, 234, 266, 319	None	GC
J1b*	069, 126, 145, 222, 261, 291	None	LM
J1b1a*	069, 145, 172, 222, 261, 368	None	RA
J1c	069, 126	2.6% of Europeans	GS, MV
J1c	069, 126, 145	1 Iraqi, 2 Canaries, 2 US Caucasian, 1 Northern Greek	MM
J1c	069, 126, 278, 366	5 Spanish, 2 Açores, 1 Spanish Gypsy, 1 French, 2 Swedish	EL
T2*	126, 294, 296	0.7% of Europeans	LRB
T2*	126, 293, 294, 296	None	EG
T2*	126, 294, 296, 311	1 Aragón, 1 Andalusian, 1 Turk, 1 Greek, 1 French	MDP
T2b*	126, 294, 304	0.4% of Europeans	TH
T2b*	126, 294, 296, 304	1.2% of Europeans	CLF

Haplogroup	Haplotype	Matches	Samples
T2b*	126, 292, 294, 296, 304	4 Italians, 1 US Caucasian, 1 German, 1 Slovakian	CRC
U*	189	24 Europeans and 1 Moroccan	HL
U2e	051, 129C, 362	1 Poitou	AN
U3	343 (390)	0.8% Europeans, 7 in Spain, 52 Gypsies	EGD
U3	168, 343	1 Turkey, 1 Armenia, 5 Greece, 1 Bulgaria, 1 Egypt, 3 Spain, 1 Poland, 1 Estonia	SS
U3	172, 343, 362	1 Central Spain	CM
U4	356	0.8% of Europeans	AC, BM?
U4	186, 189, 356	2 Catalans, 2 Spanish, 1 Macedonian Aromun	RV
U4	094, 134, 356	None	NC
U5a*	256, 270, 362	1 US Caucasian, 1 Pole, 1 Hungarian, 1 Swedish, 1 Icelandic, 1 Chuvash	BI
U5a*	192, 256, 270, 297, 304, 320	None	EB
U5b*	189, 270	0.6% of Europeans	JB
U5b*	189, 270, 274	None	UMM
U5b*	167, 192, 270	None	JGZ
U5b*	192, 270, 319	11 Basques, 2 Béarn, 4 Spanish, 2 French, 1 Algerian	AH
K	224, 311	2.5% of Europeans	EC, VR, RM
K	093, 224, 311	0.7% of Europeans	BMC
K	188, 224, 311	1 Canaries, 1 Greek	CMS
K	093, 188, 224, 311	None	MLV
U6	093, 172, 189, 219, 278	1 US Caucasian	MMM
HV2	214	1 in Brittany	MT

Haplogroup	Haplotype	Matches	Samples
HV0	298	2.2% of Europeans	APL, MP
H*	CRS	13.2% of Europeans	RAB, XB, DC, GDO, SD, XF, MGG, EGz, AHD, CO, OR, MR, JS, RS, GT, EU
H*	129	0.9% Europeans, 1.8% Iberians, 8.3% Basques	YEP, TMB, DM, MBL, IM
H*	189	1.4% Europeans, 2.4% Iberia, 25% Liébana	RAR, IH
H*	311	1.9% Europeans, 0.9% Iberians	JI, VS
H*	293	0.2% Europeans, 0.5% Iberians	ECM
H*	304	1.8% Europeans, 1.2% Iberians	MA
H*	362	1.1% Europeans, 0.9% Iberians	ARP
H*	278	0.2% Europeans, 0.4% Iberians, 8% Eivissa	CDM
H*	248	31 Europeans, 10 Iberians	FM
H*	245	1 Turk, 1 Madeira	NB
H*	304, 362	1 Turk, 3 Sicilians, 1 French, 1 Swiss, 1 Bulgarian, 1 Greek, 1 Russian	JP
H*	104, 362	2 Catalans	JLM
H*	244, 278	1 Greek	JR
H*	129, 189, 212	1 Central Spain	SC
H*	189, 273	None	PL
H1a*	162	0.7% in Europeans 1 Galicia, 1 València, 1 Canaries, 1 Açores	AR, RVS
H1a3	051, 162	22 Europeans, 2 Münster, 3 Mecklenburg, 1 Andalusia, 1 C. Portugal, 1 Canaries	JE, JL
H1b	075, 172, 189, 309T, 356	None	DH
H7c	093, 265	7 Macedonian Aromun, 1 Romanian Aromun, 1 Albanian, 5 Druze, 4 Italian, 1 Hungarian, 2 Croatian, 1 Cypriot	VD

Table 2. Haplogroup percent frequencies in the IBE sample. Frequencies for Iberia and Europe are given for reference.

HG	IBE (N=86)	Iberia (N=2,117)	Europe (N=18,263)
L2a1*	1.16	0.52	0.14
L3b	1.16	0.19	0.05
A	1.16	0	0.12
I	2.33	1.28	2.07
B	3.49	0.05	0.09
J	6.98	7.46	7.97
T	6.98	8.93	9.11
U*	1.16	1.56	1.41
U2	1.16	0.85	1.09
U3	3.49	0.85	1.07
U4	4.67	1.65	2.77
U5	6.98	6.28	10.62
K	6.98	6.57	5.82
U6	1.16	1.37	0.64
HV2	1.16	0.05	0.03
HV0	2.33	7.18	6.62
H	48.84	46.15	41.17

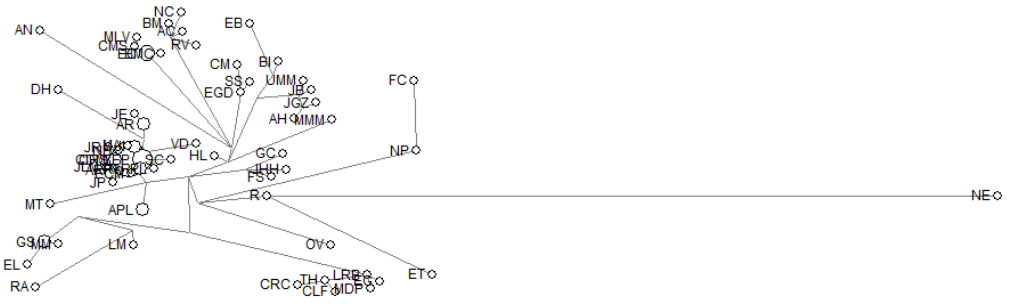


Fig. 2a: Median-joining network of IBE mtDNA sequences.

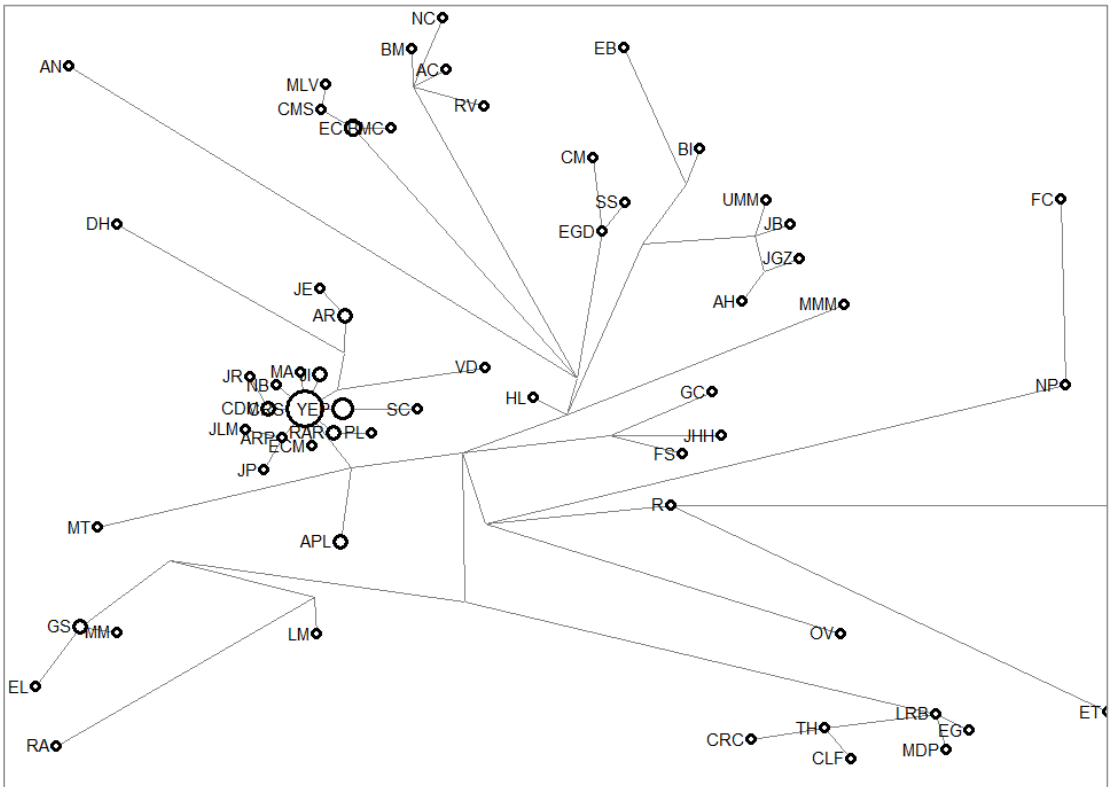


Fig. 2b: Close-up of Figure 2a.

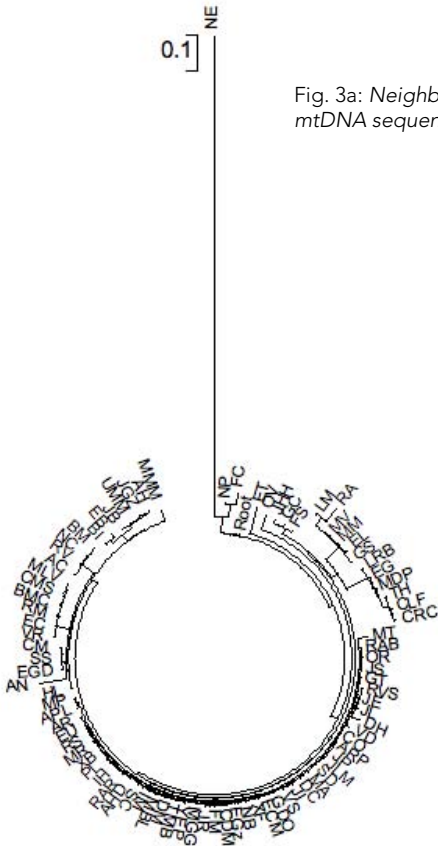


Fig. 3a: Neighbor-joining tree of the IBE mtDNA sequences

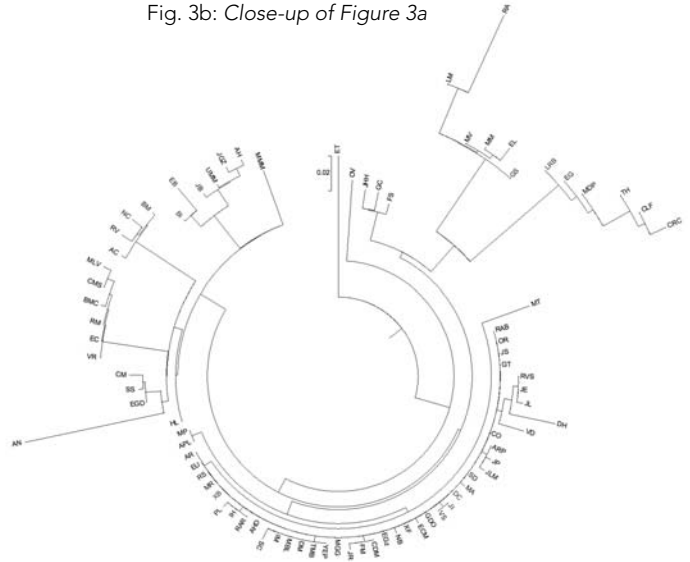


Fig. 3b: Close-up of Figure 3a

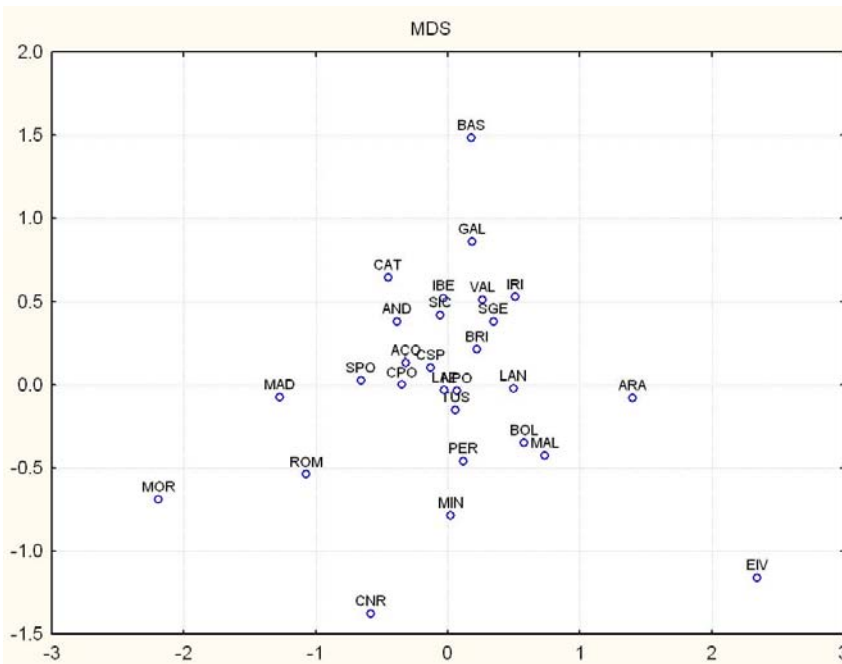
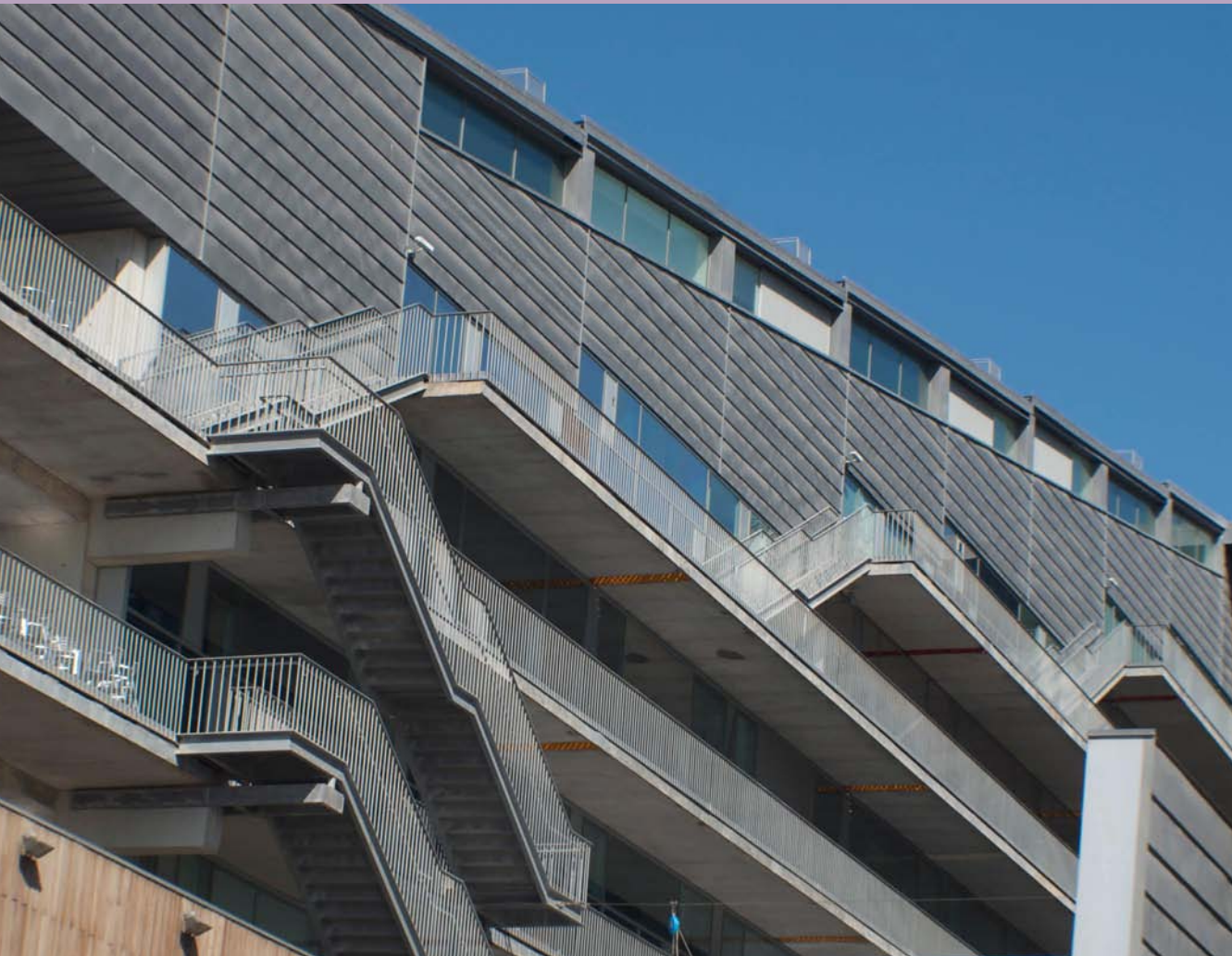


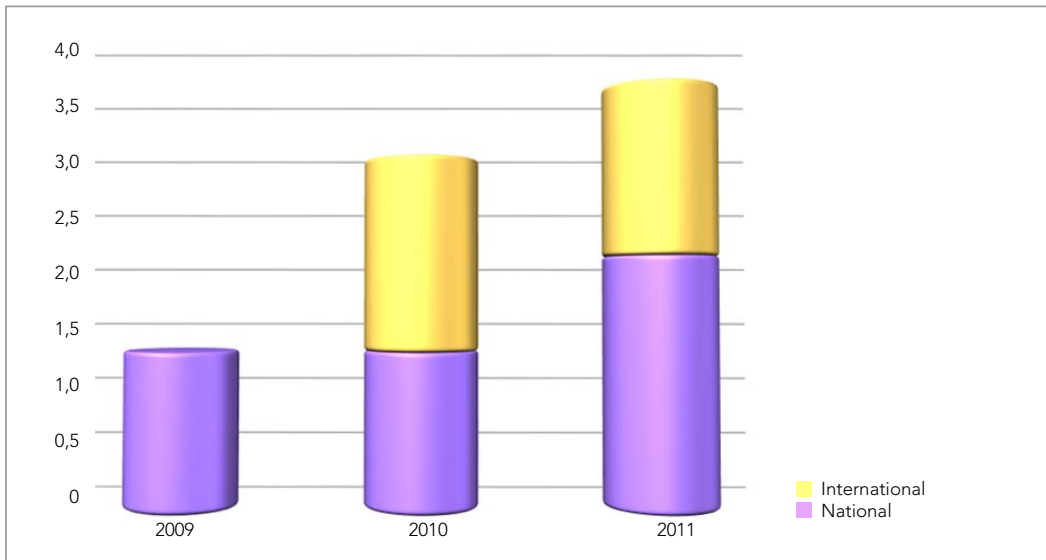
Fig. 4: MDS representation of genetic distances between the IBE and other European samples.

IBE, IBE; BAS, Basques; GAL, Galicians; CAT, Catalans; VAL, Valencians; IRI, Irish; AND, Andalusians; SIC, Sicilians; SGE, S. Germans; BRI, British; AÇO, Açoreans; CSP, Central Spanish; SPO, S. Portuguese; CPO, C. Portuguese; LAZ, Lazio; NPO, N. Portuguese; LAN, Languedocian; ARA, Aragonese; MAD, Madeiran; TUS, Tuscan; BOL, Bolognese; PER, Perigordian; MAL, Majorcan; ROM, Roman; MOR, Moroccan; MIN, Minorcan; EIV, Eivissan; CNR, Canarian.



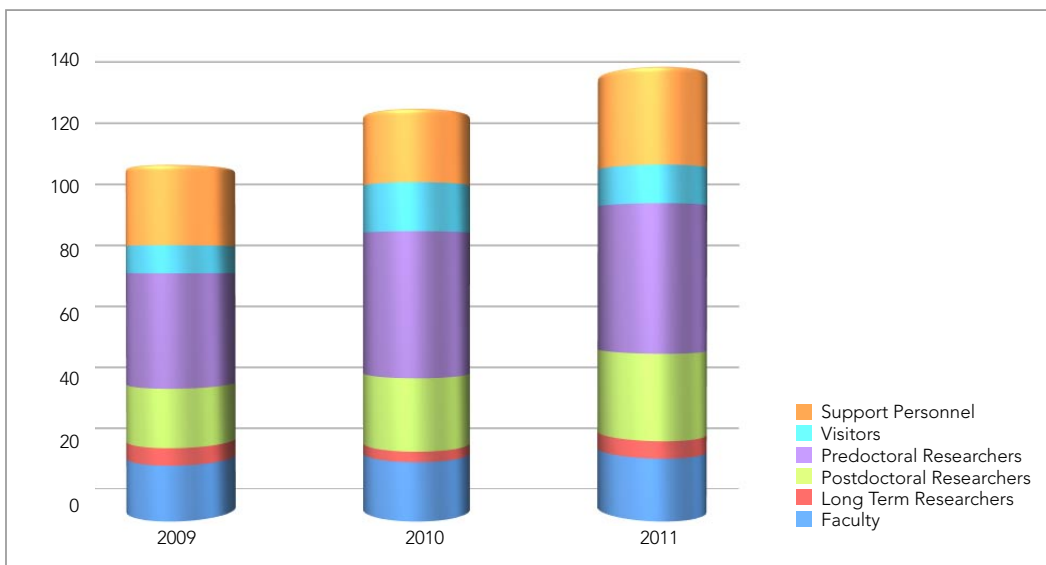
PROJECTS

Evolution of annual competitive income. Includes whole amount granted (pluriannual projects)



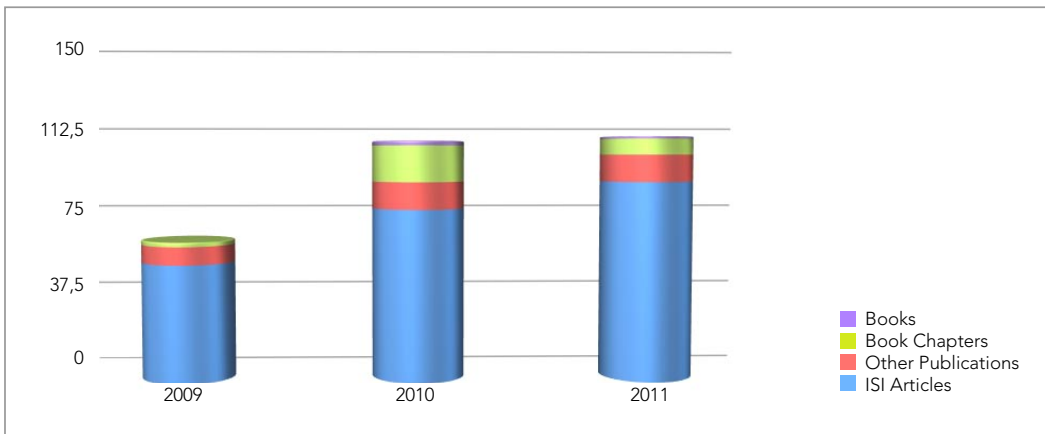
PEOPLE

Evolution of personnel distribution per categories

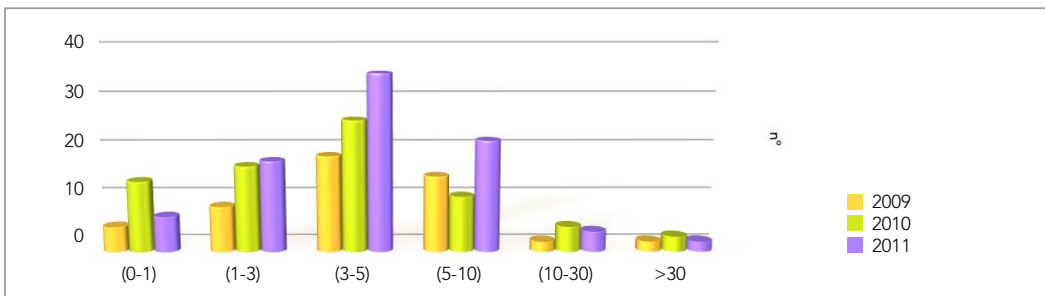


PUBLICATIONS

Evolution of publications distribution per kind of publication

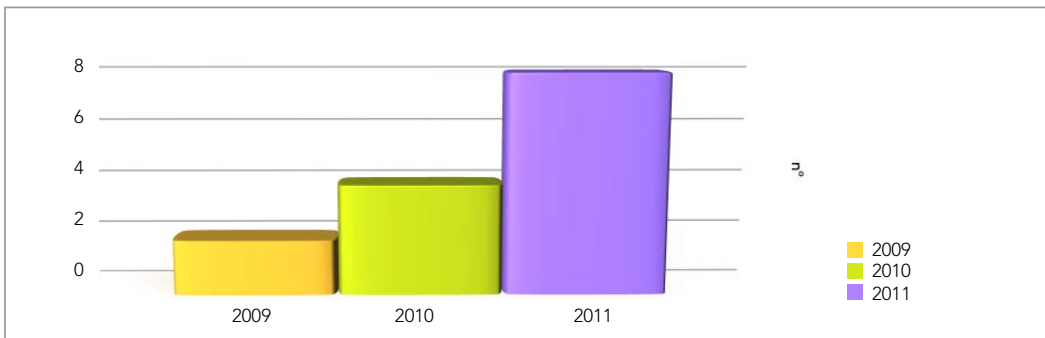


Evolution of distribution of ISI Articles per Impact Factor (IF) Intervals



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