Annual Report 2010
# Index

## Foreword

5

## Introduction to the IBE

7
- Scope and General Goals ................................................................. 7
- General Structure ........................................................................ 8
- Scientific Structure ...................................................................... 9
- Service Units ................................................................................ 9
- IBE Organisation Chart ............................................................... 10
- Personnel ...................................................................................... 13
- Localisation .................................................................................. 13

## IBE Research Programs

15
- **Animal Phylogeny and Systematics Program** ........................................ 17
  - Phylogeny and Phylogeography of Mammals
    - José Castresana, Group Leader .................................................. 18
  - Systematics, Biogeography and Evolution of Reptiles and Amphibians
    - Salvador Carranza, Group Leader .............................................. 21
  - Herbivore Beetle Evolution
    - Jesús Gómez-Zurita, Group Leader ........................................... 25
  - Water and Cave Beetle Evolution
    - Ignacio Ribera, Group Leader ................................................... 29
  - Butterfly Diversity and Evolution
    - Roger Vila, Group Leader ......................................................... 33

- **Comparative and Computational Genomics Program** .......................... 37
  - Genomics of Individuality
    - Francesc Calafell, Group Leader ............................................... 38
  - Paleogenomics
    - Carles Lalueza-Fox, Group Leader .......................................... 41
  - Evolutionary Genomics
    - Arcadi Navarro, Group Leader ................................................ 45
  - Primate Genomics
    - Tomàs Marquès-Bonet, Group Leader ..................................... 49
FUNCTIONAL EVOLUTION IN INSECTS PROGRAM ........................................... 53
Evolution of Insect Metamorphosis
  | Xavier Bellés, Group Leader ........................................... 54
Insect Reproduction
  | Maria-Dolors Piulachs, Group Leader .................................. 58
Nutritional Signals in Insects
  | José Luis Maestro, Group Leader .......................................................... 61
Hormonal Control of Insect Development
  | David Martín, Group Leader .......................................................... 64
Drosophila Telomeres
  | Elena Casacuberta, Group Leader ......................................................... 68
Developmental Biology and Morphological Evolution in Insects
  | Xavier Franch-Marro, Group Leader .......................................................... 71

POPULATION GENETICS PROGRAM ............................................................... 75
Evolutionary Systems Biology
  | Jaume Bertranpetit, Group Leader ......................................................... 76
Evolutionary Population Genetics
  | Elena Bosch, Group Leader .......................................................... 80
Human Genome Diversity
  | David Comas, Group Leader .......................................................... 83
microRNAs in Human Adaptation and Disease
  | Yolanda Espinosa-Parrilla, Group Leader ............................................. 87

COMPLEX SYSTEMS PROGRAM ................................................................. 91
Complex Systems
  | Ricard Solé, Group Leader .......................................................... 92

SCIENTIFIC PUBLICATIONS ......................................................................... 97

IBE HIGHLIGHTED PAPERS ......................................................................... 107
Genomes from Extinct Hominins Challenge Current Theories on Modern Human Origins ............................................. 107
The Colonization of Pyrenean Caves by a Hyperdiverse Tribe of Beetles ........................................................................ 109
Common Genetic Origins of Contemporary Jewish Communities ........................................................................ 110
A Large and Complex Structural Polymorphism at 16p12.1 Underlies Microdeletion Disease Risk ............................................. 112

OTHER ACTIVITIES ....................................................................................... 115
Doctoral Theses Presented During 2010 .......................................................... 115
Dissemination of Science: Biodiversity Year .......................................................... 116
IBE Seminars 2010 ....................................................................................... 116
Teaching .......................................................................................... 118

IBE IN NUMBERS .......................................................................................... 121
The capacity of blunder slightly is the real marvel of DNA. Without this special attribute, we would still be anaerobic bacteria and there would be no music.

Lewis Thomas. The Medusa and the Snail (New York, 1979)

This special attribute of DNA to “blunder slightly” has led to the wonderful biodiversity we enjoy today in our planet. Biodiversity, however, is at risk because the high rates of species extinction mainly provoked by human myopia, abuse and stupidity. Edward O. Wilson, the most renowned biodiversity advocate, has estimated that between 4,000 to 6,000 species become extinguished every year, an estimation that can be debatable but that readily suggests that we are facing a serious problem. The responsibility of preserving biodiversity is a huge one, as also is the challenge of discovering and describing the 90% of the world’s biodiversity that remains unknown to science. These circumstances lead the United Nations to declare 2010 as The International Year of Biodiversity, aiming at raising awareness of the importance of biodiversity through activities and events all over the world.

One of the most important IBE missions is to place biodiversity into a robust evolutionary frame. We especially aim to decipher the mechanisms explaining the interspecific and intraspecific differences, as well as to find out how these differences are fixed, maintained, distributed and transmitted, and how they interact with the environment. All this may explain how is biodiversity generated and maintained, which doubtlessly helps to preserve it. The IBE has been busy this year, as it has organized a great number of conference cycles, lectures and seminars; published popular science articles and books; and participated in the celebrations of other institutions addressed to commemorate the Year of Biodiversity.
2010 has been a year of additional activities and achievements. Importantly, the Board of Trustees approved the IBE Regulations of Functioning, and it is now studying the composition of the External Scientific Committee according to the proposal submitted by our Executive Board. We hope that this will be solved within the next weeks, so the External Scientific Committee will be fully operative in 2011. Included in the hardware organization of the IBE there is the issue of the new building. Although some shadows glide over the financial aspects, the planned schedule is being followed steadily, so we are confident that we can have the IBE building by 2013.

From a point of view of scientific structure, the main novelty is the incorporation of a new program to the IBE, that of Complex Systems, coordinated by Ricard Solé (ICREA Research Professor). This is great news because the high scientific standing of the program leader, and because we see living and evolving organisms as having emergent properties that can be properly studied only under the light of the complex systems concepts.

Moving to more prosaic IBE issues, the financial income coming from competitive sources has increased dramatically in 2010 (2,906,086 euros) with respect to 2009 (1,304,355 euros), these figures calculated as the sum of amounts granted in the respective year for pluriannual projects. Of note, the ERC starting grant awarded to Tomàs Marquès, “Ramón y Cajal” Researcher recently incorporated to the Comparative and Computational Genomics program, represents a substantial part of the whole amount, which made the increase so spectacular.

Concerning publications, ISI articles signed by IBE members augmented from 52 in 2009 (3.2 papers per PI) to 76 in 2010 (3.8 papers per PI). Quantity may be important but quality is essential for us. In this sense, the number of articles increased in practically all Impact Factor (IF) intervals used to classify the journals, except in that between 5 and 10 (15 articles published in journals of this IF interval in 2009, versus 12 in 2010). Conversely, we increased the number of papers published in high ranked journals, with an IF between 10 and 30 (2 in 2009, 5 in 2010), and in very high ranked journals, with an IF higher than 30 (2 in 2009, 3 in 2010).

Considering all categories, IBE personnel augmented from 100 (2009) to 119 (2010) people, but most substantial increases were in predoctoral (33 in 2009 versus 42 in 2010) and postdoctoral researchers (17 in 2009 versus 21 in 2010). In addition to the aforementioned Ricard Solé and Tomàs Marquès, we welcome also the incorporation of two PIs with long contracts: Roger Vila (ICREA Junior Researcher), that joined the Animal Phylogeny and Systematics program, and Yolanda Espinosa (“Marie Curie” Researcher), that joined the Population Genetics program. Needless to say, IBE people are their greatest asset. They are intelligence in action, but with the capacities for wonder, astonishment and fun still intact. People like Sigfried after he tasted the dragon’s blood, when he found to his surprise that he could understand the language of birds. And above all, open minded people, because, as James Dewar once said, “Minds are like parachutes. They only function when they are open”.


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, IBE was created with 11 independent research groups from the Insect Molecular Biology Department (CID, CSIC) and 6 independent research groups from the Evolutive Biology Unit (dCEXS, UPF). Nowadays, IBE activity involves a hundred people and 20 independent research groups distributed in five scientific programs related to Evolutionary Biology research. The scope and general research goals of the IBE focus on biological evolution.

Evolutionary biology aims at studying the processes and mechanisms generating biodiversity. After the Human Genome project, the description and comprehension of biodiversity is one of the most important scientific challenges for the XXI century, and coping to this challenge requires the methods and concepts of evolutionary biology. In particular, we need to understand the genetic basis of the difference among species (divergence), as well as within every species (variation, polymorphism), paying special attention at how differences are fixed and how are distributed, transmitted, maintained and how they interact with the environment. The description of genomes and the understanding of genomic differences in organisms are of paramount importance to understand the basic mechanisms of life and to place biodiversity into a robust evolutionary frame.

The general objective of the IBE is to work on these contexts, using all the available new tools, experimental and computational, to understand the basic functioning of life, to describe and put into context their diversity, to unveil the mechanisms generating biological innovations and evolutionary changes and, finally, to preserve biodiversity and to promote its use in a sustainable way. In particular, the basis of the IBE, and its main peculiarity, is the capacity to face biodiversity studies describing genomic and functional evolution at any observational scale: molecular, biochemical, physiological or morphological. The IBE aims at becoming an international reference in these subjects.
GENERAL STRUCTURE

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

BOARD OF TRUSTEES

IBE main governing body is the “Board of Trustees”, composed by two representatives of both partner Institutions (CSIC-UPF):

- **Luis Calvo**
  - CSIC Institutional Coordinator in Catalonia

- **Teresa García Milà**
  - UPF Vicechancellor of Scientific Policy

- **Francisco Montero**
  - CSIC Vicepresident of Institutional Relationships and Organisation

- **Francesc Posas**
  - CEXS-UPF Department Director

EXTERNAL SCIENTIFIC COMMITTEE (ESC)

The IBE External Scientific Committee (ESC) will be a group of scientific experts with international recognition in the Evolutionary Biology field whose main task will be 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.

After the approval of the IBE Regulations of functioning by the Board of Trustees, they are now studying the composition of the ESC proposed by our Executive Board. The ESC is expected to be fully operative by mid 2011.

The composition of the ESC proposed by IBE Executive Board (now under approval process) is as follows:

- **Chairman**
  - Andrés Moya (Universitat de València, València, Spain)

- **Members**
  - Carlos Bustamante (Stanford University, Palo Alto, CA, USA)
  - Brian Charlesworth (University of Edinburgh, Edinburgh, UK)
  - Gonzalo Giribet (Harvard University, Cambridge, MA, USA)
  - Stuart Reynolds (University of Bath, Bath, UK)
  - Luis Serrano (Centre de Regulació Genòmica, Barcelona, Spain)
  - 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
| Jaume Bertranpetit

| José Castresana (acting also as the Coordinator of the “Animal Phylogeny and Systematics” Program)

| David Comas (acting also as the Coordinator of the “Population Genetics” Program)

| Maria-Dolors Piulachs (acting also as the Coordinator of the “Functional Evolution in Insects” Program)

General Manager and Board Secretary
| Anna Pérez-Lezaun

SCIENTIFIC STRUCTURE

The scientific structure of the IBE is composed by twenty different groups and five Scientific Programs:

| Animal Phylogeny and Systematics
| Comparative and Computational Genomics

| Functional Evolution in Insects
| Population Genetics

| Complex Systems

SERVICE UNITS

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

The IBE management unit was formally constituted by mid 2009 with the incorporation of an 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 cover at a micro scale level all the basic Institute running processes (Accounting, Human resources, purchasing, logistics and safety, and support to projects).

EXPERIMENTAL TECHNIQUES

As mentioned, this unit is still under constitution process. Right now, it counts with one technician from the JAETEC (CSIC) program incorporated to IBE activity in September 2010 to coordinate the maintenance and use of specialized common technical instrumentation and installations.
BIOINFORMATICS UNIT

This unit is planned to start its activity in 2011 with the incorporation of a Bioinformatics expert at the beginning of 2011.

It is expected that, in the next future, this unit could give support to all IBE programs in the area of Bioinformatics by making available to researchers a coordinated and stable team of Bioinformatics experts to support the increasingly strong computational needs of IBE research teams, both offering highly specialized personnel support and availability to high capacity calculation and storage hardware.

RESEARCH ASSISTANTS

Apart from the mentioned formal units, the IBE also counts with four laboratory technicians that give scientific key support to different IBE programs.

Rocío Alonso, JAETEC-CSIC Contract | Animal Phylogeny and Systematics Program
Cristina Olivella, Technical Staff CSIC | Functional Evolution in Insects Program
José Martínez, JAETEC-CSIC Contract | Experimental Techniques Unit
Mónica Vallés, Technical Staff UPF | Population Genetics and Comparative and Computational Genomics Programs
At the end of 2010, the IBE had 118 members (Table 1) with a ratio men/women close to 50% and an internalization ratio of more than 26% (maximum ratio in postdoctoral researchers where internalization level increases up to 33 %).

Table 1. IBE personnel distribution by categories. December 2010.

<table>
<thead>
<tr>
<th>Category</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Faculty</td>
<td>17</td>
</tr>
<tr>
<td>Long-term Researchers*</td>
<td>3</td>
</tr>
<tr>
<td>Postdoctoral Researchers</td>
<td>21</td>
</tr>
<tr>
<td>Predoctoral Researchers</td>
<td>44</td>
</tr>
<tr>
<td>Support Personnel</td>
<td></td>
</tr>
<tr>
<td>Laboratory Technicians</td>
<td>9</td>
</tr>
<tr>
<td>Bioinformatics Experts</td>
<td>5</td>
</tr>
<tr>
<td>Management Staff</td>
<td>5</td>
</tr>
<tr>
<td>Others</td>
<td>2</td>
</tr>
<tr>
<td>Long-term Visitors (&gt; 1 month)</td>
<td>14</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td><strong>118</strong></td>
</tr>
</tbody>
</table>

*Marie Curie, Ramón y Cajal or ICREA Junior researchers

While not having an our specific building, whose construction is planned towards 2013, 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.
Members of this research program carry out research on animal biodiversity from a phylogenetic perspective aiming at gaining further insight into the tree of life. The program’s 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

Ana Rodríguez-Prieto, PhD Student | FPI Scholarship, MICINN
Javier Igea, PhD Student | JAEPRE-CSIC Fellowship
Víctor Soria-Carrasco, PhD Student | FPI Scholarship, MICINN
Alejandro Sánchez-Gracia, Post-doc | Juan de la Cierva Contract
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, because 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 multiple genetic markers. With this aim, we have analyzed the available mammalian genomes to compile new sets of intronic markers that may help in these works. We are currently sequencing these markers together with mitochondrial genes in different mammalian species, particularly small mammals of the Iberian Peninsula, to better understand speciation scenarios, patterns of genetic variability, 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 the 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 used at the interphase between phylogenetics and population genetics. We are also developing databases that allow the integration of specimen data, geographic coordinates and multiple markers for both phylogenetic and phylogeographic analyses. Methods and software that we develop are made available online.

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 2010

ISI Articles

Other Publications

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, AGAUR (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/BOS)*  
Years: 2009-2011  
PI: José Castresana

Fig. 1: Analysis of species diversification in the family Bovidae. A maximum-likelihood tree, a calibrated tree and the lineage through time plots for the statistical evaluation of the lineage diversification are shown.
GROUP

SYSTEMATICS, BIOGEOGRAPHY AND EVOLUTION OF REPTILES AND AMPHIBIANS

Salvador Carranza, Group Leader
| Tenured Scientist, CSIC

Margarita Metallinou, PhD Student | FPU Scholarship, MEC
Hernán Morales, MSc Student | Master in Evolutionary Biology, University of Groningen, The Netherlands
Joan Garcia-Porta, PhD Student | JAEPRE-CSIC Fellowship
Josep Roca, Technician | Project Contract
Elena Gómez-Díaz, Post-doc Researcher | Beatriu de Pinós Contract
Catarina Rato, PhD Student co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal
Raquel Vasconcelos, 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
RESEARCH OUTLINE

Our research focuses on the application of phylogenetic analyses of reptiles and amphibians to understand how biodiversity is generated and maintained. Moreover, 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 island, 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. The main objectives of the project 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 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)
ISI Articles


Other Publications

Funded Projects

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO
Financed by: Generalitat de Catalunya, AGAUR (2009 SGR1462)
Years: 2009-2013
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: 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 Gosálbez

Fig. 3: Pristurus rupestris, Wadi Dharbat, Oman.
GROUP

HERBIVORE BEETLE EVOLUTION

GROUP MEMBERS

Jesús Gómez-Zurita, Group Leader
| Tenured Scientist, CSIC

Anna Papadopoulou

Tinguaro Montelongo

Anabela Cardoso

Gissela de la Cadena

Jesús Gómez-Zurita

Tinguaro Montelongo, PhD Student | FPI Scholarship, MEC
Anabela Cardoso, Research Assistant | CSIC
Gissela de la Cadena, PhD Student | AECID Scholarship, MAEC
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 the study of biological processes such as hybridization, unisexuality and insect-plant associations from an evolutionary perspective. The basic approach we follow to tackle this broad spectrum of research topics is a phylogenetic framework based upon 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 molecular signatures of the hybrid origin of unisexual taxa. We are also interested in ecological aspects of this system, investigating performance and niche characteristics of sympatric bisexual and unisexual species, relevant to understand their interaction and respective evolutionary advantages.

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 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 and trophic networking 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 that isolated from other continental land masses by the end of the Cretaceous, and remained isolated since that period. It hosts a singular biota with high endemicity and in some cases purportedly very ancient, perhaps relicts of Gondwanan 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 (see Project 2).
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. hypochaeridis*-complex, which include 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 finishing now the revision of the bulk of the genus in its Southern Nearctic and Neotropical distribution.

**PUBLICATIONS 2010**

**ISI Articles**


**Book Chapters**


**Other Publications**


**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**: Systematic Revision of the poorly studied southern Nearctic and Neotropical Calligrapha Chevrolat  
  **Financed by**: Harvard University, Ernst Mayr Grant  
  **Year**: 2010  
  **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. 2: Entomological Campaign in New Caledonia. Field work is the first very important step for biodiversity surveys and evolutionary studies by our group. We get fresh samples for DNA work, learn a lot about the biology of the animals we study... and it is fun!

Image: J. Gómez-Zurita.
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 focuses are 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).

### RESEARCH SUBLINES

1. **Thermal tolerance and Pleistocene range expansions**
   
The *Agabus brunneus* complex includes species of diving beetles both with narrow and wide geographical ranges, which also diverge in their thermal tolerances. With the analyses of the whole proteome expression through 2D electrophoresis we try to establish the role of shifts in thermal tolerance in the Pleistocene range expansion 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 Catalan 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. 1:** Graptodytes eremitus, a stygobitic diving beetle described in Ribera & Faille (2010) from a single specimen found in a cave in Morocco. Image: J. Fresneda and J. Gómez-Zurita
ISI Articles


Book Chapters, Refereed Proceedings


Funded Projects

Project Title: The evolution of the size of the geographical range as a key factor in the generation of biodiversity
Funded by: Ministerio de Ciencia e Innovación (CGL2007-61665)
Years: 2007-2010
PI: Ignacio Ribera

Project Title: Effectiveness of the peninsular National Park Net in the conservation of aquatic biodiversity
Funded by: Ministerio de Ciencia e Innovación (Research program on National Parks, 023/2007).
Years: 2007-2011
PI: Andrés Millán (Universidad de Murcia)

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO
Funded by: Generalitat de Catalunya, AGAUR (2009 SGR-1462)
Years: 2009-2013
PI: Salvador Carranza

Fig. 3: Valeria Rizzo collecting specimens of Troglocharinus. Image: Joan Cabeza
GROUP

BUTTERFLY DIVERSITY AND EVOLUTION

GROUP MEMBERS

Roger Vila, Group Leader
| ICREA Researcher

Vlad Dinca, PhD Student | FI Scholarship, UAB
Claudia Sañudo, PhD Student | FBBVA Project Scholarship
Gerard Talavera, PhD Student | FPI Scholarship, MEC
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.

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. Our main goals are to test the efficiency of the method at large scale, and to develope 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 to deeply analyse each case, and to be able to assess 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.

Fig. 1: Leptidea is a very interesting genus for us: geographically isolated from nearest relatives, with unstable chromosome numbers, and with high potential to host cryptic species. Image: Vlad Dinca
PUBLICATIONS 2010

ISI Articles

Other Publications

Funded Projects

Project Title: Combining morphological, cytological and molecular data to study the taxonomy of Iberian Rhopalocera (Lepidoptera: Hesperioidea + Papilionoidea)
Financed by: Ministerio de Ciencia e Innovación (Plan Nacional I+D+i)
Years: 2007-2010
PI: Roger Vila

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: Grup de Recerca Consolidat-SGR “Grup de Genòmica, Bioinformàtica i Evolució”
Financed by: Generalitat de Catalunya (2009 SGR-0088)
Years: 2009-2013
PI: Alfredo Ruiz

Fig. 2: Discovering cryptic biodiversity is one of our main lines of research. Careful morphological and molecular studies are needed to distinguish between very similar species, as is the case with the *Aricia* specimens in the picture.
Image: Vlad Dinca
“El Sidrón” Cave
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 of the species these genomes come from. Studying full genomes is, thus, 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 key 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

Francesc Calafell, Group Leader
| Associate Professor, UPF

Núria Bonet, Laboratory Technician | CIBERESP Contract
Marc García, PhD Student | FU Scholarship, MEC
Koldo García, Post-doc | CIBERESP Contract
Marta Melé, PhD Student | FI Scholarship, MEC
Mireia Coscollà, Post-doc | CIBERESP Contract
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 three 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; and 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.

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).
PUBLICATIONS 2010

ISI Articles


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

Fig. 1: Y-chromosome lineages in samples of men carrying the Colom surname. Lineages were inferred from 16-STR haplotypes and are likely to each represent the descendants of a single man. Gray subsumes all lineages with frequencies < 2%.
Carles Lalueza-Fox, Group Leader

| Research Scientist, CSIC

Elena Gigli, PhD Student | FPI Scholarship, MICINN
Federico Sánchez, PhD Student | FPI Scholarship, MICINN
Oscar Ramírez, Visitant Professor, UPF
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* 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 Sidrón 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 Sidrón 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 Sidrón offers the unique opportunity of launching a genomic project for understanding the diversity within the Neandertal lineage.

3. Phylogenetics, phylogeography and adaptation in extinct species
   We are studying different extinct species, including *Puffinus*, lynx, *Myotragus balearicus*, and mammoths, 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.

4. Extinct modern human populations
   We are genetically analysing ancient samples from prehistoric European populations to reconstruct past human migrations and also sample from aboriginal human groups recently vanished. We are trying to developed new methodological tools for capturing targeted genomic regions combined with massively parallel sequencing technologies.
ISI Articles


Other Publications


**Fig. 1:** Ancient Iberian Lynx mandible
Book Chapters


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
GROUP

EVOLUTIONARY GENOMICS

GROUP MEMBERS

Arcadi Navarro, Group Leader
   | Professor, UPF - Research Professor, ICREA

Natalia Petit, Post-doc | JAE Contract
Gabriel Santpere, Post-doc | Project Contract
Belén Lorente, PhD Student | UPF Contract
Urko Martínez, PhD Student | UPF Scholarship
Diego Hartasánchez, PhD Student | JAE Contract
Oriol Vallès, Masters Student | Project Contract
Carlos Morcillo, Project Manager | INB (National Bioinformatics Institute)
Ángel Carreño, IT Technician | INB (National Bioinformatics Institute)
Txema Heredia, IT Technician | INB (National Bioinformatics Institute)
Fernando Muñiz, IT Technician | INB (National Bioinformatics Institute)
Rui Faria, Visiting Scientist | FCT Fellowship
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 of single-copy protein-coding regions. We focus in how natural selection may have shaped regulatory regions and the functional content of SDs.

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.
ISI Articles


Book Chapters

**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:** IMID-Kit  
**Financed by:** Ministerio de Educación y Ciencia (MEC-PSS-010000-2008-36)  
**Years:** 2008-2010  
**PIs:** Arcadi Navarro and Jaume Bertranpetit (Coordinator: S. Marsal)  

**Project Title:** INB GN8  
**Financed by:** Genoma España (Instituto Nacional de Bioinformática)  
**Years:** 2003-2010  
**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
Tomàs Marquès-Bonet, Group Leader
| Ramón y Cajal Researcher, UPF

Belén Lorente, PhD Student | INB Project Contract
Javier Prado, PhD Student | FI Scholarship, MEC
Irene Hernando, PhD Student | ERC StGt Project Contract
Marcos Fernández, Computational Support | ERC StGt Project Contract
RESEARCH OUTLINE

Our main line of research is centered in the discovery of the extent of genome structural polymorphism within the great ape species. The goal is to create an integrated view of structural variation evolution by studying changes in the composition, frequency, size and location at every major branchpoint 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. Structural 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.

3. Recurrent duplications in great-ape evolution
We now know that most human segmental duplications arose during a narrow window of evolution (7-10 million years ago) within the ancestral lineage of humans and great apes. The interspersed organization and complexity of duplication blocks are greatest among humans and African ape. However, one of the most interesting finding was the detection of some regions that showed a pattern of duplication inconsistent with the known consensus phylogenetic relationship of great-apes. These regions are highly relevant because they represent either incomplete lineage sorting at the time of speciation or recurrent events that happened independently during the course of evolution.

Fig. 1: A burst of duplications in the genome of the common ancestor of African great-Apes (around 8-12 million years ago). All numbers are in megabases (1 million of basepairs).
ISI Articles


Other Publications


**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

Fig. 2: A particular duplication expansion in chimpanzee and gorilla. This segment of DNA is single copy in humans. The green circle indicates the ancestral location (donor). The red arrows indicate the derivative locations in each species based on location in the human karyotype.
Ovarioles from Blattella germanica ovary. DAPI (blue), rhodamine-phalloidin (red) and tubuline (green).
Image: Nashwa Elshaer and M. Dolors Piulachs
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

This program aims at studying fundamental processes in animal life –such as metamorphosis, reproduction, development, cell division or telomere regulation–, 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), bees (Apis) 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 lasts 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, it affords the tools of experimental biology, which are of paramount importance to understand the adaptive mechanisms of evolution.
GROUP

EVOLUTION OF INSECT METAMORPHOSIS

GROUP MEMBERS

Xavier Bellés, Group Leader
| Research Professor, CSIC

Raúl Montañez, Post-doc | JAEDOC-CSIC Contract
Ferran Borràs, PhD Student | FPI Scholarship MICINN
Mercedes Rubio, PhD Student | JAEPRE-CSIC Fellowship
Alba Herráiz, PhD Student | JAEPRE-CSIC Fellowship
Jesús Lozano, PhD Student | FPI Scholarship MICINN
Jia-Hsin Huang, PhD Student | Scholarship National Taiwan University
RESEARCH OUTLINE

The endocrine regulation of insect metamorphosis has been a long standing theme of research and training in the Bellés group, which started in the late 1970’s. We are interested in the regulation of metamorphosis from a developmental point of view and from an evolutionary perspective. As most information has been obtained in highly modified, holometabolan species (mainly in Drosophila melanogaster), we focus on the cockroach Blattella germanica, a phylogenetically basal, hemimetabolan species. We aim to elucidate the mechanisms regulating metamorphosis in B. germanica and then comparing them with those operating in holometabolans. The idea is to infer the evolutionary history underlying the transition from hemimetaboly to holometaboly.

RESEARCH SUBLINES

1. Endocrine basis of metamorphosis
The study of the physiological effects of the hormones regulating metamorphosis (juvenile hormone and ecdysone) is one of our fields of interest. Also important is the determination of the hormonal titers and rates of biosynthesis in metamorphic and non-metamorphic transitions, in order to have accurate endocrine frames for functional studies.

2. Qualitative metamorphic changes
We investigate qualitative metamorphic changes that occur in the transition from nymph to adult. An example is the formation of the adult wing, whose study is being approached not only by monitoring the morphological changes, but also at molecular level, by comparing wing bud transcriptomes in metamorphic and non-metamorphic transitions.

Fig. 1: Role of Broad Complex in wing morphogenesis in Blattella germanica.
Image: Jia-Hsin Huang
3. Transcription factors and hormonal signaling
A great deal of data is available on transcription factors of the ecdysone pathway. Thus, we concentrate on transcription factors typically belonging to the juvenile hormone pathway (like Krüppel homolog 1 and Methoprene-tolerant), and those intersecting both pathways (like Broad Complex). We use RNAi approaches as the main tool to unveil their functions during metamorphosis.

4. Small RNAs
We investigate the regulatory role of miRNAs in metamorphosis under the hypothesis that miRNAs play a crucial role in the shift of “genetic program” from juvenile to adult. Work is not only descriptive but also functional, using anti-miR molecules to reveal the functions of the miRNAs under study. We are also interested in studying the biochemical machinery involved in the RNAi process and in miRNA generation.

5. 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 can study their topological properties in metamorphic and non-metamorphic transitions, infer regulatory mechanisms, and validate them experimentally. At present, we focus on networks of interaction mRNA-miRNA comparing holometabolan and hemimetabolan models.

PUBLICATIONS 2010

ISI Articles

Books

Other Publications
FUENDED PROJECTS

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 Ros

Project Title: Insect Control with RNAi
Financed by: CSIC and the National Taiwan University
(REF: 2010TW0019, Formosa program)
Years: 2010-2011
PI: Xavier Bellés Ros

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 Ros

Fig. 2: Precocious metamorphosis induced in Blattella germanica by depleting Krüppel homolog 1 with RNAi.
Image: Jesús Lozano
GROUP

INSECT REPRODUCTION

GROUP MEMBERS

Maria-Dolors Piulachs, Group Leader
| Research Scientist, CSIC

Erica D. Tanaka, Post-doc | JAEDOC-CSIC Contract
Paula Irles, Post-doc Researcher | CSIC Contract
Alba Herráiz, PhD Student | JAEPRE-CSIC Fellowship · LINCGlobal
Jelle Caers, Erasmus Student
Our aim is to understand how the oogenesis in insects is regulated, considering the structural diversity of ovary types and their respective evolutionary history. Until now, ovarian maturation has been studied in detail only in the dipterans *Drosophila melanogaster* and *Aedes aegypti*, which are highly modified species, with meroistic ovaries regulated by ecdysteroids. In our case, we use as a model the cockroach *Blattella germanica*, which is an hemimetabolan species with panoistic ovaries, whose vitellogenesis and oocyte growth are regulated by juvenile hormone. Our working hypothesis is that some genes will be conserved in structure and function in panoistic and meroistic ovaries, whereas other genes will be specific to one of these types or will have different functions in the two ovarian types. The approach is to study the genes that are inducible by JH in the ovary of *B. germanica* and their functions, and to establish how many of these genes and functions are conserved in other insect species with different ovary types.

Our research focuses on three main subjects:

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 and previtellogenesis, oocyte development in vitellogenesis, and chorion synthesis. Attention is also paid to the role of miRNA in regulating oogenesis.

3. **Evolution of ovarian structure and function**
   The idea is to study the function of genes expressed in the ovary of *B. germanica* and to establish how many of these genes and functions are conserved in meroistic models. Data obtained might suggest functional scenarios underlying the transition between the panoistic and the meroistic type.

*Fig. 1: Mitosis figures in the follicular epithelia from an ovary of a newly emerged adult. DAPI (blue) and tubuline (green). Image: Laura Ciudad and M. Dolors Piulachs*
PUBLICATIONS 2010

ISI Articles


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

Fig. 2: SEM micrograph of the Sponge-like body from a Blattella germanica egg. Image: Paula Irles and M. Dolors Piulachs

Fig. 3: Actin distribution in the basal pole of follicular cells from Blattella germanica ovary. DAPI (blue) and rhodamine-phalloidin (red). Image: Paula Irles and M. Dolors Piulachs
GROUP

NUTRITIONAL SIGNALS IN INSECTS

GROUP MEMBERS

José Luis Maestro, Group Leader
| Tenured Scientist, CSIC

Songül Süren-Castillo, Post-doc | JAEDOC-CSIC Contract
Marc Abrisqueta, PhD Student | FPI Scholarship, MICINN
RESEARCH OUTLINE

The main research interest of our group focuses on the study of how the organisms can detect their nutritional status and how this information will regulate some critical processes, especially reproduction, but also growth, longevity, etc. A typical case in which the flow of information from nutrition to physiological events becomes crucial is that of the anautogenous organisms. Anautogeny is a successful reproductive strategy typically used by different mosquito species, but also by other insects. Females of the anautogenous species do not initiate the reproductive processes until they have made a food intake. This intake will trigger a series of signals that will activate the physiological, metabolic or endocrine processes that will lead to reproduction. The German cockroach, *Blattella germanica*, is a typical anautogenous insect that will not start the production of reproductive hormones or yolk proteins until it has eaten. This species is a very good model of insect physiology because its reproduction is governed, as in most insects, by juvenile hormone and not by ecdysteroids as it only happens in dipterans (flies and mosquitoes). For these reasons, this cockroach is our main insect model.

RESEARCH SUBLINES

1. Nutritional signaling pathways: Insulin receptor and TOR

Two of the pathways that contribute to the nutritional signal transmission, in insects and other organisms, are the insulin receptor (InR) and the target of rapamycin (TOR) pathways. They are involved in detecting nutritional signals and activating different processes, such as growth, cell proliferation, longevity and cancer. Our group studies these pathways in *B. germanica* and their relationships to the activation of reproduction, among other processes. 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).

Fig. 1: Retrocerebral complex from *Blattella germanica* 3-day old 6th instar nymph stained for detection of BrdU labelling.
2. FOXO transcription factor

FOXO proteins are evolutionary conserved transcription factors which have been described as key players in 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 the 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 this transcription factor. We are also interested in the role of FOXO in some other processes such as starvation resistance, oxidative stress and longevity.

PUBLICATIONS 2010

ISI Articles


Fig. 2: Corpora allata from a 5-day old *Blattella germanica* adult female stained with fluorescein diacetate.

Fig. 3: Detail of DAPI staining of the basal structure of an oocyte from a 5-day old *Blattella germanica* treated with TOR dsRNA.
GROUP

HORMONAL CONTROL OF INSECT DEVELOPMENT

GROUP MEMBERS

David Martín, Group Leader
- Tenured Scientist, CSIC

Cristina Manjón, Post-doc
- Juan de la Cierva Contract, MICINN

Ferran Borràs, PhD Student
- FPI Scholarship, MICINN

Enric Ureña, PhD Student
- JAEPRE-CSIC Fellowship
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 a holometabolous insect with complete metamorphosis (Drosophila melanogaster).

RESEARCH SUBLINES

1. Molecular analysis of Ecdysteroids and Juvenile Hormone action
   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. Unfortunately, the molecular mechanism of JH action is still largely unknown. 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 (using RNAi in vivo and parental RNAi procedures) and 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 both types of insects. On the other hand, to elucidate the JH signalling pathway, we are currently analyzing in detail the function of two genes, methoprene-tolerant and ultraspiracle, the putative JH receptors.

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. 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.

Fig. 1: Adult Drosophila melanogaster Met RNAi knockdown showing defects in wing morphogenesis.
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.

**PUBLICATIONS 2010**

**ISI Articles**


**Book Chapters**

Funded Projects

Project Title: Molecular Basis of Ecdysone 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

![RNAi-mediated knockdown of SUMO showing impaired wing development at the adult stage of the cockroach Blattella germanica.](image-url)
GROUP

DROSOPHILA TELOMERES

GROUP MEMBERS

Elena Casacuberta, Group Leader
Tenured Scientist, CSIC

David Piñeyro, Post-doc Researcher
CSIC Contract

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, tumorogenesis 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 telomeric 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 onto 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.

Fig. 1: Chromosomes
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.

FUNDED PROJECTS

Project Title: Estudio de los aspectos funcionales y evolutivos de los telomeros de Drosophila
Financed by: Ministerio de Ciencia e Innovación (BFU2009-08318)
Years: 2010-2012
PI: Elena Casacuberta Suñe

Fig. 2: Cells
GROUP

DEVELOPMENTAL BIOLOGY AND MORPHOLOGICAL EVOLUTION IN INSECTS

GROUP MEMBERS

Xavier Franch-Marro, Group Leader
| Tenured Scientist, CSIC

Neus Bota Rabassedas, PhD Student | FPI Scholarship, MICINN
Cristina Miguel Vijandi, PhD Student | FPI Scholarship, MICINN
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 kinds of changes in their sequences are responsible for the evolution of the mentioned morphological diversity. Using *Drosophila melanogaster* and *Tribolium castaneum* as organisms models, the aim of our lab is to elucidate the mechanisms that allows 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 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.

RESEARCH SUBLINES

1. Tracheal System Evolution

Traqueal system is the respiratory organ of Insects. It consists of a network of tubes that transport oxigen 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 basal 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.

Fig. 1: Wing imaginal disc of Drosophila with Posterior compartment mutant for Wls, marked with absense of GFP. Wg protein, shown in red, accumulates in the secreting cells of wls mutant cells.

2. Terminal system Evolution

In *Drosophila*, terminal system determines the anterior and posterior most terminal body regions of the embryo. Basically, the terminal system consists of the interaction of a group of maternal genes that results eventually in the local activation of the receptor tyrosine kinase Torso by its processed ligand Trunk, at the egg poles. This activation of the signaling pathway leads to transcription of several target genes. Recent studies have shown that terminal-group genes are conserved in the Coleoptera *Tribolium*, where are responsible, as in *Drosophila*, of the development of the posterior segments. In contrast, in Hymenoptera *Apis mellifera* neither *Tor* nor *trunk* have
been found in its genome. This raises the question of whether terminal system is a relatively new mechanism to control the formation of terminal regions of the embryo or in contrast it is an ancient system that has been lost in Hymenoptera. To solve this question, we want to isolate Torso in hemimetabolous insects and analyze its possible function regulating terminal body regions in these ancient organisms.

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 signaling 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 poorly 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 and size control
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.

PUBLICATIONS 2010

ISI Articles

Funded Projects

<table>
<thead>
<tr>
<th>Project Title</th>
<th>Formación del Gradiente del Morfógeno y de la función de Wingless en el control del crecimiento</th>
</tr>
</thead>
<tbody>
<tr>
<td>Financed by</td>
<td>Ministerio de Ciencia e Innovación (REF: BFU2009-08748)</td>
</tr>
<tr>
<td>Years</td>
<td>2010-2012</td>
</tr>
<tr>
<td>PI</td>
<td>Xavier Franch-Marro</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Project Title</th>
<th>Función de la vía de Wingless en el control del crecimiento y sus implicaciones evolutivas</th>
</tr>
</thead>
<tbody>
<tr>
<td>Financed by</td>
<td>CSIC-Proyectos Intramurales Especiales (REF: 2009201209)</td>
</tr>
<tr>
<td>Year</td>
<td>2010</td>
</tr>
<tr>
<td>PI</td>
<td>Xavier Franch-Marro</td>
</tr>
</tbody>
</table>
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.
Jaume Bertranpetit, Group Leader

Hafid Laayouni, Senior Scientist | CIBEResp Researcher
Ludovica Montanucci, Post-doc | UPF Contract
Martin Sikora, PhD Student | FU Scholarship, MICINN
Marta Melé, PhD Student | FU Scholarship, MICINN
Giovanni dall’Olio, PhD Student | FI Scholarship, MICINN
Pierre Luisi, PhD Student | ISCIII Scholarship
Brandon Invergo, PhD Student | Project Scholarship, FI Scholarship, Generalitat de Catalunya
Marc Pybus | Master - PhD Student
Manu Uzkudun | Master Student, 1000genomes Project
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.

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. 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.

2. Malaria
Taking also a pathway approach we have been studying the genetic susceptibility to placental malaria in a case-control setting in a population in Manhiça, Mozambique. This project is in collaboration with Clara Menéndez and other members of the Barcelona Center for International Health Research (CRESIB). High-throughput genotyping data has been generated and the analysis has been centered mostly in innate immunity and glycosylation.

3. 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. Francesc Calafell is also involved in this project (see his page for more details).
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.

4. 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 a variety of population-specific studies (including Basques, North Africa, South Saharan Africa and others).

PUBLICATIONS 2010

ISI Articles

As Part of the Genographic Consortium
Book Chapters

Other Publications
• Dall’Olio, G.M., Bertranpetit, J., and Laayouni, H. 2010. The annotation and the usage of scientific databases could be improved with public issue tracker software. Database (Oxford) 2010: baq035.

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
Funded by: National Geographic and IBM
Years: 2006-2010
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
GROUP
EVIOLUTARY POPULATION GENETICS

GROUP MEMBERS

Elena Bosch, Group Leader
Associate Professor, UPF

Johannes Engelken, Post-doc
Volkswagenstiftung Scholarship, Germany

Elena Carnero, PhD Student
UPF Teaching Scholarship

Mònica Vallès
Anna-Lena Scherr
Núria Bonet
**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 are now currently investigating quantitative and qualitative functional differences between the alleles at two nonsynonymous substitutions linked to the genetic signature of selection. This project is developed in collaboration with Francisco Lozano’s group from the Hospital Clinic (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: Clustering analysis of mRNA expression of three marker genes in a set of kidney samples allowing for differentiation between cortex and medulla.](image-url)
4. Bioinformatic analysis of 1000genomes 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 1000genomes project.

PUBLICATIONS 2010

ISI Articles

FUNDED PROJECTS

Project Title: Detecção 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

Fig. 2: qRT-PCR amplification covering cDNA concentrations diluted across 5 log ranges.

Fig. 3: RNA quality check of extracted tissue samples using Agilent 1200 Bioanalyzer.
GROUP
HUMAN GENOME DIVERSITY

GROUP MEMBERS

David Comas, Group Leader
| Associate Professor, UPF

Begoña Martínez-Cruz, Post-doc | UPF Contract
David Soria, Post-doc | UPF Contract
Isabel Mendizabal, PhD Student | Basque Country Scholarship
Laura Rodríguez-Botigué, PhD Student | FI Scholarship
Paula Sanz, Technician | UPF Contract
Graciela Sotelo, Technician | UPF Contract
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 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 the 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 2010

ISI Articles

As Part of the Genographic Consortium


Other Publications

**FUNDED PROJECTS**

Project Title: *Diversidad genética en poblaciones humanas de África Central: implicaciones demográficas, lingüísticas y culturales*  
(CGL2007-61016/BOS)  
Financed by: Dirección General de Investigación Científica y Técnica  
Years: 2007-2010  
PI: David Comas

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: Dirección General de Investigación Científica y Técnica  
Years: 2010-2012  
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: *The Genographic project: Western/Central Europe region*  
Financed by: National Geographic and IBM  
Years: 2006-2010  
PIs: Jaume Bertranpetit and David Comas
GROUP

micrORNAS IN HUMAN ADAPTATION AND DISEASE

GROUP MEMBERS

Yolanda Espinosa-Parrilla, Group Leader
| Marie Curie Researcher, UPF

María López, PhD Student | FI Scholarship, AGAUR
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 adaptative 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.

Fig. 1: Optimal secondary structure for a human specific miRNA according to RNAfold.
PUBLICATIONS 2010

ISI Articles


FUNDED PROJECTS

Project Title: MapbyAdmixtureChl-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

Fig. 2: Organization of the human collection of microRNAs per chromosomes according to their aggregation in clusters.
This program, which is still in its initial phase of implementation, 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.
Ricard Solé, Group Leader
Professor, UPF, ICREA Researcher

Javier Macía | Post-doc Researcher
Sergi Valverde | Post-doc Researcher
Jordi Delgado | Post-doc Researcher
Carlos Rodríguez-Caso | Post-doc Researcher
Bernat Corominas-Murtra | Post-doc Researcher
Martí Rosas Casals | Post-doc Researcher
Stefanie Widder | Post-doc Researcher
Ben Shirt-Ediss | Pre-doc Researcher
Núria Conde | Pre-doc Researcher
Salvador Durán | Pre-doc Researcher
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. Our research spans a broad range of systems, with special attention to biological computation, protocell biology, synthetic systems and network biology.

RESEARCH OUTLINE

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

PUBLICATIONS 2010

ISI Articles


**Book Chapters**


**Other Publications**


FUNDED PROJECTS

Project Title: Origins of innovation in tinkered networks
Financed by: McDonnell Foudation USA (McDonell Award 220020117)
Years: 2007-2011
PI: Ricard Solé

Project Title: SYNLET
Financed by: FP7-European Union
Years: 2007-2010
PI: Ricard Solé

Project Title: COMPLEXDIS - Unravelling complex diseases with complexity theory
Financed by: FP7-European Union
Years: 2007-2010
PI: Ricard Solé

Project Title: Development of virtual tumor
Financed by: National Institute of Health
(Grant N°: CA11304-3 and 03S3 CFDA N° 93397)
Years: 2008-2010
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

Project Title: Convenio de colaboración en materia de apoyo a la transferencia tecnológica en el campo de la biotecnología, prof. Ricard Solé.
Financed by: Fundación Marcelino Botín
Years: 2009-2010
PI: Ricard Solé
ISI Articles


• Laayouni, H., Calafell, F., and Bertranpetit, J. A genome-wide survey does not show the genetic distinctiveness of Basques. Human Genetics 127:455-458.


Books

Book Chapters


Other Publications

Year 2010 will be remembered in the field of human evolution by two notable scientific landmarks: the Neandertal genome, published in May, and the Denisova hominin genome, published in December.

The Neandertal genome, up to a 1.5x coverage, has been generated from three bone samples belonging to three different individuals from Vindija Cave in Croatia and dating to around 40,000 years ago. It has been complemented with partial genomic data from three other Neandertals: Mezmaiskaya in Russia; Feldhofer in Germany; and El Sidrón in Spain. By comparing those coding protein positions were Neandertals carry the ancestral allele and modern humans a fixed, derived allele, it has been possible to determine that 78 genes have been recently selected in our lineage. Those genes include some related to physiological, metabolic and cognitive aspects, but nevertheless the exact biological meaning of these genetic differences will need to be functionally investigated in the future. In addition to, the analysis of the Neandertal has shown that interbreeding occurred between Neandertals and non-Africans modern humans, with the former contributing up to 2% of the latter genomes. This previously undetected process of hybridisation likely took place in the Near East, where Neandertals and modern humans co-existed, between 50,000 and 100,000 years ago.

The second extinct hominin genome has been obtained from a 30,000 to 50,000-year-old finger bone from Denisova Cave, in southern Siberia. Although no morphological analysis can be undertaken from this small bone, a tooth found in the same cave and belonging to a second individual shows morphological traits that are distinct from Neandertals and modern humans and resemble much older hominin forms, such as *Homo habilis* and *Homo erectus*. The authors have temptatively named these hominins Denisovans. Based on the genome sequence, Denisovans appear to have been a group of hominins that shared a common ancient origin with Neandertals, but subsequently had a distinct evolutionary history. Unlike Neandertals, Denisovans did not contribute genes to all present-day Eurasians.
Instead, present day Melanesian populations carry 4-6% of Densiovan genetic material in their genomes, suggesting that there was interbreeding with the ancestors of Melanesians.

Both the Neandertal and the Denisova genomes suggest a much more complex picture of genetic interactions between our ancestors and different ancient hominin groups than has been previously assumed.

Reference Articles

THE COLONIZATION OF PYRENEAN CAVES BY A HYPERDIVERSE TRIBE OF BEETLES

The year of diversity was a good opportunity to literally “unearth” one of the most diverse and neglected faunas, that of the subterranean environment. The extreme diversity of the organisms living in the underground offers multiple opportunities to address general evolutionary questions concerning the origin and distribution of diversity, and cave organisms have been used as models for evolution and biogeography since Darwin. Most current hypotheses on the origin of subterranean species assume, generally without further evidence, that they arose recently from surface dwelling, dispersive close relatives. This work deals with the origin of the Iberian fauna of one of the most diverse groups, the beetles of tribe Leptodirini, with more than 250 species. All of them have restricted distributions, in many cases reduced to a single cave or group of nearby caves in the same karstic system. The use of molecular data from nuclear and mitochondrial genes allowed to infer the phylogenetic relationships among the main groups, and the presence of several species in Sardinia, most likely as the result of the tectonic separation of the Corso-Sardinian plate ca. 30 million years ago, allowed to calibrate the resulting tree using a molecular clock approach. The phylogenetic analysis revealed strongly supported clades, each of them restricted to a major mountain system in the Iberian Peninsula. The molecular clock calibration established a rate of 2.0% divergence per MY for a combination of five mitochondrial genes (4% for the most commonly used

![Fig. 1: Phylogeny of the Pyrenean clade of subterranean beetles of tribe Leptodirini. The main lineages are as old as the chain itself, and did colonize it from the edges to the center.](image-url)
cox1 alone) and dated the nodes separating the main subterranean lineages before the Early Oligocene. The colonisation of the Pyrenean chain, by a lineage not closely related to those found elsewhere in the Iberian Peninsula, began soon after the subterranean habitat became available in the Early Oligocene, and progressed from the edges to the centre. The results of the study strongly suggest that by the Early-Mid Oligocene the main lineages of Western Mediterranean Leptodirini had developed all modifications adapted to the subterranean life and were already present in the main geographical areas in which they are found today. The origin of the currently recognised genera can be dated to the Late Oligocene-Miocene, and their diversification can thus be traced to Miocene ancestors fully adapted to subterranean life, with no evidence of extinct epigean, less modified lineages. The close correspondence of organismal evolution and geological record confirms them as a most interesting case study for historical biogeography and molecular evolution.

ISI Article

The Article has been Featured in

COMMON GENETIC ORIGINS OF CONTEMPORARY JEWISH COMMUNITIES

Contemporary Jews comprise diverse ethno-religious groups scattered in different geographical regions. It is assumed that Jews have a common origin in the Middle East and that they dispersed through Europe, Asia and Africa in historical times (Jewish Diaspora). Although several genetic studies based on molecular diseases and uniparental (mitochondrial and Y-chromosome) markers have approached this issue, there are some relevant questions related to the population history and genetic structure of the Jewish communities that remain poorly understood. It is still not clear what the genetic relationships between contemporary Jewish groups, their Diaspora neighbours and Middle Eastern populations are.

Fourteen contemporary Jewish communities comprising European, Asian and African Jews were analyzed together with several Middle Eastern and neighbouring non-Jewish populations. The genetic data consisted of more than half a million markers (Single Nucleotide Polymorphisms, SNPs) scattered through the genome, which provided the first genome-wide view of the Jewry.

The present analysis shows that, despite their dispersion and isolation during millennia, Jewish communities exhibit a common genetic origin and a genetic continuity with non-Jewish Middle Eastern populations. The only exceptions are the Jewish populations from India and Ethiopia that exhibit a genetic pool similar to their non-Jewish neighbours (Indians and Ethiopians, respectively) and distant to the rest of Jewish groups and Middle Eastern
samples. This result suggests that both these Jewish communities might have been originated by autochthonous individuals with posterior religious and cultural diffusion from other Jewish groups. The rest of Jewish communities can be genetically assigned to three general groups: firstly, Yemenite Jews; secondly, Middle Eastern (Iranian and Iraqi Jews) jointly with Caucasian Jews; and finally a third group that includes Ashkenazi (central European Jews), Sephardi (coming from the exile from the Iberian Peninsula), and north African Jews. Despite the presence of small genetic differences among these three Jewish groups, they exhibit little genetic admixture with non-Jewish neighbours with whom they have been sharing territory for millennia. This has kept the ancestral genetic pool from the Middle East in their ethno-religious communities.

Fig. 1: Population structure plot (ADMIXTURE analysis) based on more than 225,000 SNPs in human worldwide populations including Jewish communities.

Reference Article
A LARGE AND COMPLEX STRUCTURAL POLYMORPHISM AT 16P12.1 UNDERLIES MICRODELETION DISEASE RISK

It has been ten years after the first release of the human genome assembly, a milestone for our research, and four until it was declared as finished. It is striking then, that one of the most scrutinized region in chromosome 16p12, reported as a risk factor for childhood intellectual disability and developmental delay, still contained a blatant error in the assembly.

In this paper, we identified what appeared to be one of the largest inconsistencies of the human reference genome. We experimentally and computationally showed that all humans are homozygously inverted for the gene order of a 1.1 Mbp region compared to the representation of 16p12.1 in the reference genome. The new reconstructed organization showed that at least two human haplotypes are segregating in human populations and more importantly, only one of them predisposes this configuration alone to non-allelic homologous recombination and hence to potential damaging microdeletion events.

Reference Article


Fig. 1: Schematic representation of the 16p12 region. The top track represents the human genome (hg18). The following 2 tracks represents the 2 major haplotypes found in humans. Only the S2 haplotype predisposes to the pathogenic microdeletion. Color boxes denotes the location of segmental duplications and green and blue lines represents the orientation among block (direct and reverse, respectively).
Fig. 2: (A) 16p12.1 genomic organization in macaque, orangutan, gorilla, chimpanzee and human. Green and red arrows indicate the orientation of the regions between breakpoint (BP1-BP2 and BP2-BP3). Black and colored rectangles show segmental duplications. Sequence and FISH data indicate that the inverted configuration as found in orangutan and macaque is likely the ancestral state. Inversions of the regions from BP1 to BP2 and from BP2 to BP3 are likely to have occurred in the African Great Apes ancestor after formation and expansion of the segmental duplications (I). Subsequently, the region spanning from BP1 to BP2 might have flipped back to the direct orientation in the chimpanzee lineage (II). The region has become increasingly complex in human leading to the addition of another polymorphic 333 kbp at BP1 specifically in the human lineage (III).
<table>
<thead>
<tr>
<th>PhD Student</th>
<th>Paula Irles</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Title</strong></td>
<td>Bases moleculares de la oogénesis en la cucaracha <em>Blattella germanica</em> (L.) (Dictyoptera, Blattellidae)</td>
</tr>
<tr>
<td><strong>Thesis Director</strong></td>
<td>Maria-Dolors Piulachs</td>
</tr>
<tr>
<td><strong>Institution and Date</strong></td>
<td>Universitat de Barcelona, 15th February 2010</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PhD Student</th>
<th>Martin Sikora</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Title</strong></td>
<td>Evolutionary genetics of malaria: genetic susceptibility and natural selection</td>
</tr>
<tr>
<td><strong>Thesis Directors</strong></td>
<td>Jaume Bertranpetit and Ferran Casals</td>
</tr>
<tr>
<td><strong>Institution and Date</strong></td>
<td>Universitat Pompeu Fabra, 4th June 2010</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PhD Student</th>
<th>Karla Sandoval</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Title</strong></td>
<td>Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica.</td>
</tr>
<tr>
<td><strong>Thesis Director</strong></td>
<td>David Comas</td>
</tr>
<tr>
<td><strong>Institution and Date</strong></td>
<td>Universitat Pompeu Fabra, 19th June 2010</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PhD Student</th>
<th>Gemma Berniell</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Title</strong></td>
<td>Genes, people and languages in central Africa</td>
</tr>
<tr>
<td><strong>Thesis Director</strong></td>
<td>David Comas</td>
</tr>
<tr>
<td><strong>Institution and Date</strong></td>
<td>Universitat Pompeu Fabra, 19th July 2010</td>
</tr>
</tbody>
</table>
## DISSEMINATION OF SCIENCE: BIODIVERSITY YEAR

<table>
<thead>
<tr>
<th>Activity</th>
<th>Date and Place</th>
<th>Organisers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cycle of Lectures: “Biodiversidad en acción”</td>
<td>3rd November 2010, Fundación BBVA, Madrid (Spain)</td>
<td>IBE and Fundación BBVA</td>
</tr>
<tr>
<td>Workshop: “Frontiers In Biodiversity. A Phylogenetic Perspective”</td>
<td>1st to 2nd October 2010, Barcelona (Spain)</td>
<td>IBE, IRBIO, GRC, IEC</td>
</tr>
<tr>
<td>Cycle Of Lectures: “Biodiversitat. Del Museu A La Naturalesa”</td>
<td>4th to 25th October 2010, Barcelona (Spain)</td>
<td>IBE and Residencia Investigadors del CSIC a Catalunya</td>
</tr>
</tbody>
</table>

### IBE SEMINARS 2010

<table>
<thead>
<tr>
<th>Speaker</th>
<th>Talk</th>
<th>Institution</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>Luc Steels</td>
<td>The origins of human language. Genes, memes or cognition?</td>
<td>University of Brussels (VUB AI lab) and Sony Computer Science Laboratory (Paris)</td>
<td>04/02/2010</td>
</tr>
<tr>
<td>David Vieites</td>
<td>Research prospects for historical biogeography and evolutionary biology in Madagascar</td>
<td>Museo Nacional de Ciencias Naturales (CSIC) (Madrid)</td>
<td>22/02/2010</td>
</tr>
<tr>
<td>Alfred Vogler</td>
<td>DNA taxonomy and the study of biodiversity patterns</td>
<td>Department of Biology, Imperial College (London) and The Natural History Museum (London)</td>
<td>26/04/2010</td>
</tr>
<tr>
<td>Stuart Reynolds</td>
<td>Partners in crime: Evolution of a pathogenic symbiosis between Photorhabdus bacteria and heterorhabditid nematodes, and how the insect host can fight back</td>
<td>Department of Biology and Biochemistry, University of Bath (Bath)</td>
<td>31/05/2010</td>
</tr>
<tr>
<td>Alex Kacelnik</td>
<td>A biologist’s reflections on rationality</td>
<td>Behavioral Ecology Research Group, University of Oxford (Oxford)</td>
<td>14/06/2010</td>
</tr>
<tr>
<td>Speaker</td>
<td>Talk</td>
<td>Institution</td>
<td>Date</td>
</tr>
<tr>
<td>-------------------------------</td>
<td>----------------------------------------------------------------------</td>
<td>----------------------------------------------------------------------------</td>
<td>------------</td>
</tr>
<tr>
<td>Frederic Bartumeus</td>
<td>Anomalous diffusion and stochastic search strategies in animal movement: an evolutionary perspective</td>
<td>Centre d’Estudis Avancats de Blanes, CSIC (Blanes)</td>
<td>16/06/2010</td>
</tr>
<tr>
<td>Marcos Pérez-Losada</td>
<td>Phylogeny, conservation and speciation of Aegla (Anomura: Decapoda: Aeglidae) freshwater crabs from Patagonia</td>
<td>CIBIO, Centro de Investigação em Biodiversidade e Recursos Genéticos Universidade do Porto (Porto)</td>
<td>28/06/2010</td>
</tr>
<tr>
<td>Francisco Úbeda</td>
<td>Solving the recombination hotspot paradox</td>
<td>Department of Ecology and Evolutionary Biology University of Tennessee (Knoxville Tennessee)</td>
<td>12/07/2010</td>
</tr>
<tr>
<td>Mohamed Noor</td>
<td>Recombination, speciation, and nucleotide diversity in the Drosophila pseudoobscura species group</td>
<td>Biology Department, Duke University (Durham NC)</td>
<td>13/09/2010</td>
</tr>
<tr>
<td>Teresa Romero</td>
<td>Resolución de conflictos, consolación y empatía en primates</td>
<td>University of Tokyo (Tokyo)</td>
<td>22/09/2010</td>
</tr>
<tr>
<td>Jordi Bascompte</td>
<td>Species mutualistic networks: the architecture of biodiversity</td>
<td>Integrative Ecology Group, Estación Biológica de Doñana, CSIC (Sevilla)</td>
<td>18/10/2010</td>
</tr>
<tr>
<td>Hernán Burnano</td>
<td>Targeted sequencing of Neandertal DNA: insights into recent coding and non-coding evolution on the human lineage</td>
<td>Department of Evolutionary Genetics, Max Planck Institute of Evolutionary Anthropology (Leipzig)</td>
<td>19/10/2010</td>
</tr>
<tr>
<td>José M. Eirin López</td>
<td>Histones in regalia: flourishing diversity on the verge of germ chromatin evolution</td>
<td>Universidade da Coruña (A Coruña)</td>
<td>25/10/2010</td>
</tr>
<tr>
<td>David de Lorenzo</td>
<td>Senses and Neuromarketing: Darwin goes shopping</td>
<td>Facultat de Medicina, Universitat de Lleida (Lleida)</td>
<td>05/11/2010</td>
</tr>
</tbody>
</table>
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 Biology (Pompeu Fabra University)
  - “Evolution” (4.5 ECTS). Coordinator: Elena Bosch

- Bachelor’s degree in Human Biology (Pompeu Fabra University)
  - “Antropology and Evolution” (4 ECTS). Coordinators: Elena Bosch and David Comas
  - “Zoology” Coordinator: Oscar Ramírez
  - “Ecology” 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

- Bachelor’s degree in Medicine (Pompeu Fabra University)
  - “Anthropology and Evolution” (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

- Master in Biochemical Research (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 University of Bologna
  - “Analisis 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 international master programs and specialized courses of different universities giving specialized lectures, as summarized in the following table.

<table>
<thead>
<tr>
<th>Subject</th>
<th>In: Master</th>
<th>University</th>
<th>Teacher</th>
</tr>
</thead>
<tbody>
<tr>
<td>“Genetic variation dynamics in populations” In: Genomes and Systems</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>Pompeu Fabra University (UPF)</td>
<td>Elena Bosch</td>
</tr>
<tr>
<td>“Statistics in forensic genetics” In: Grounds in Forensic Anthropology</td>
<td>Master in Human Biology</td>
<td>(UB / UAB)</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Probability calculations in STRs” In: Molecular Analysis and Statistics</td>
<td>Master in Human Biology</td>
<td>(UB / UAB)</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Transmission Disequilibrium Test” In: Biomedical Informatics</td>
<td>Bioinformatics and Health Sciences (BIOINFO)</td>
<td>UPF / UB / Università di Bologna</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Pharmacogenomics” In: Biomedical Informatics</td>
<td>Bioinformatics and Health Sciences (BIOINFO)</td>
<td>UPF / UB / Università di Bologna</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Linkage Disequilibrium” In: Genomes and Systems</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Genowide Association Studies” In: Genomes and Systems</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“The genomic view into the genetic bases of diseases” In: Genomes and Systems</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Francesc Calafell</td>
</tr>
<tr>
<td>“Genome variation: the measure of the genetic variation at the DNA level” In: Genomes and Systems</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>David Comas</td>
</tr>
<tr>
<td>“Population genetics” In: Genetic Epidemiology</td>
<td>Master on Public Health (UPF)</td>
<td>UPF</td>
<td>David Comas</td>
</tr>
<tr>
<td>“Anàlisi genètica” In: Desenterrant silencis; les fosses de la guerra civil</td>
<td>Curs Universitat d’Estiu</td>
<td>UAB</td>
<td>Carles Lalueza-Fox</td>
</tr>
<tr>
<td>“Evolución Humana”</td>
<td>Comunicación Científica, Médica y Ambiental</td>
<td>IDEC-UPF</td>
<td>Carles Lalueza-Fox</td>
</tr>
<tr>
<td>Subject</td>
<td>In: Master</td>
<td>University</td>
<td>Teacher</td>
</tr>
<tr>
<td>--------------------------------------------------------------</td>
<td>------------------------------------------------</td>
<td>--------------</td>
<td>----------------------------------</td>
</tr>
<tr>
<td>“Paleogenomics”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Carles Lalueza-Fox</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Mobile Elements and Genome Evolution”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Elena Casacuberta</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Mobile elements and Genome Evolution”</td>
<td>Máster en Biología del Desarrollo y Genética</td>
<td>UB</td>
<td>Elena Casacuberta</td>
</tr>
<tr>
<td>In: “Molecular Evolution”</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Introduction to genomic variation”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Jaume Bertranpetit</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Capturing the global human variation”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Jaume Bertranpetit / Tomàs Marquès-Bonet</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“El Modelo Icrea”</td>
<td>Màster en Lideratge i Gestió de la Ciencia i la Innovació</td>
<td>IDEC-UPF</td>
<td>Jaume Bertranpetit</td>
</tr>
<tr>
<td>“Alineamiento múltiple”</td>
<td>Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones</td>
<td>UB</td>
<td>José Castresana</td>
</tr>
<tr>
<td>“Métodos de reconstrucción filogenética aplicados a la filogenómica”</td>
<td>Máster en Biología del Desarrollo y Genética</td>
<td>UB</td>
<td>José Castresana</td>
</tr>
<tr>
<td>“Genetic drift and its effects”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Hafid Laayouni</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Selection”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Arcadi Navarro</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“The non-coding genome”</td>
<td>Master in Biomedical Research (BIOMED)</td>
<td>UPF</td>
<td>Yolanda Espinosa-Parrilla</td>
</tr>
<tr>
<td>In: Genomes and Systems</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>“Origin of Bilaterian animals”</td>
<td>Master in Biodiversity</td>
<td>UB</td>
<td>Salvador Carranza</td>
</tr>
<tr>
<td>“Topology tests”</td>
<td>Master in Biodiversity</td>
<td>UB</td>
<td>Salvador Carranza</td>
</tr>
<tr>
<td>“Molecular Phylogenetics”</td>
<td>Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones</td>
<td>UB</td>
<td>Salvador Carranza</td>
</tr>
</tbody>
</table>
**PROJECTS**

Evolution of financial income (in million euros) coming from competitive sources (only new projects granted in 2010). Figures include whole amount granted (pluriannual projects).

**PEOPLE**

IBE personnel distributed by categories.
IBE publications distributed by categories.

IBE ISI articles distributed by Impact Factor (IF) intervals of the journals used for publication.