# Annual Report 2010



INSTITUT de BIOLOGIA EVOLUTIVA





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by the Director of the IBE, Xavier Bellés



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

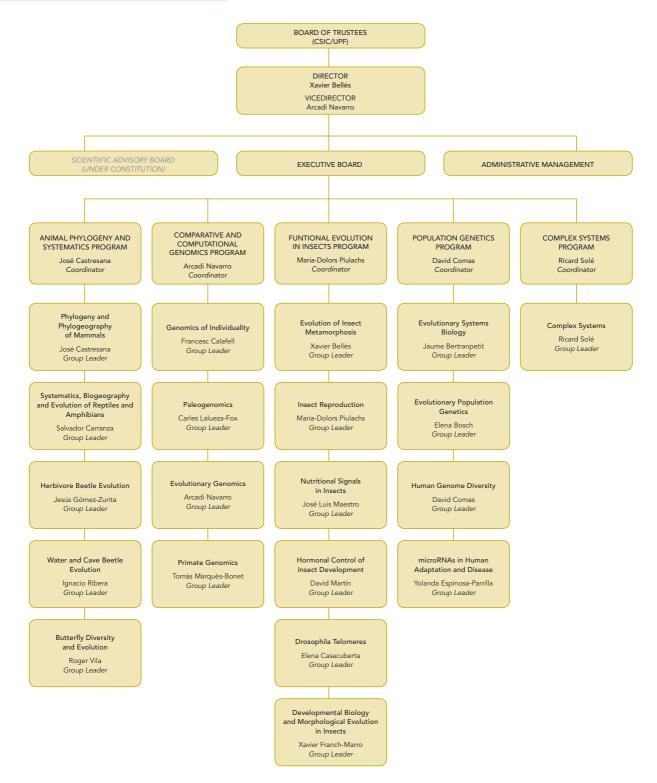
The scientific structure of the IBE is composed by twenty different groups and five Scientific Programs:

- 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".

#### IBE ORGANISATION CHART



#### 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).



Anna Pérez-Lezaun | UPF

ViceManager and Accountant Rita Arias | CSIC

#### Administrative Support

Emiliano González | CSIC Blanca Álvarez | CSIC Judit Sainz | UPF

#### 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

#### PERSONNEL

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.

Faculty	17
Long-term Researchers*	3
Postdoctoral Researchers	21
Predoctoral Researchers	44
Support Personnel	
Laboratory Technicians	9
Bioinformatics Experts	5
Management Staff	5
Others	2
Long-term Visitors (> 1 month)	14
TOTAL	118

\*Marie Curie, Ramón y Cajal or ICREA Junior researchers

#### LOCALISATION

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.



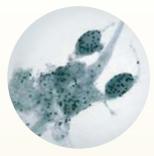


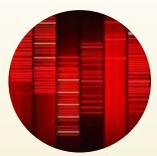


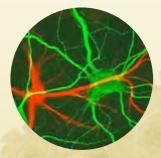


# IBE RESEARCH PROGRAMS









Lysandra coridon

# PROGRAM animal phy and systematics

#### **RESEARCH GROUPS**

Phylogeny and Phylogeography of Mammals José Castresana, Group Leader

Systematics, Biogeography and Evolution of Reptiles and Amphibians Salvador Carranza, *Group Leader* 

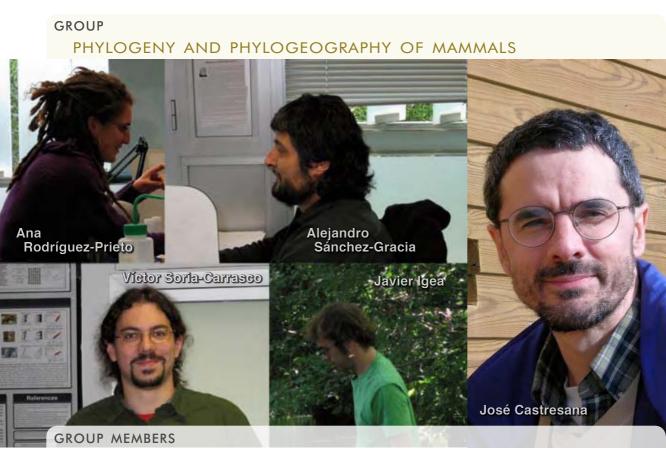
Herbivore Beetle Evolution Jesús Gómez-Zurita, Group Leader

Water and Cave Beetle Evolution Ignacio Ribera, Group Leader

#### Butterfly Diversity and Evolution

Roger Vila, Group Leader

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.



#### 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 lberian Peninsula. Finally, because 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

- Igea, J., Juste, J., and Castresana, J. 2010. Novel intron markers to study the phylogeny of closely related mammalian species. *BMC Evolutionary Biology* 10: 369.
- Sánchez-Gracia, A., Romero-Pozuelo, J., and Ferrús, A. 2010. Two frequenins in *Drosophila:* unveiling the evolutionary history of an unusual neuronal calcium sensor (NCS) duplication. *BMC Evolutionary Biology* 10: 54.

#### Other Publications

• Castresana, J. 2010. Reconstrucción filogenética, diversificación y especiación. SEBBM 165: 20-22.

#### FUNDED PROJECTS

Financed by: Years:	Distribution and genetic diversity of the Iberian desman (Galemys pyrenaicus) in the Spanish Network of National Parks Ministerio de Medio Ambiente y Medio Rural y Marino. Network of National Parks Research Funds (014/2008). 2009-2011 Joaquim Gosálbez
Financed by: Years:	Zoological Systematics and Evolution Research Group - ZOOSYSEVO Generalitat de Catalunya, AGAUR (2009 SGR 1462) 2009-2013 Salvador Carranza
Project Title:	Phylogeny and speciation of mammals studied with multiple nuclear markers
Years:	Ministerio de Ciencia e Innovación (GL2008-00434/BOS) 2009-2011 Jacé Contracado

Pl: José Castresana

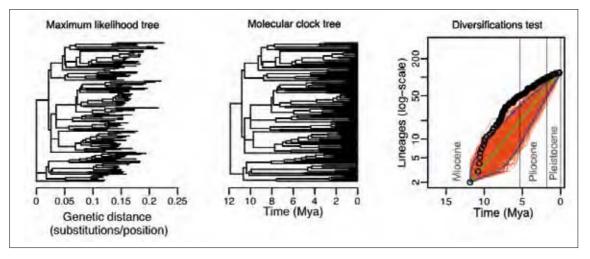


Fig. 1: Analysis of species diversification in the family Bovidae. A maximum-likelihood tree, a calibrated tree and the linage through time plots for the statistical evaluation of the lineage diversification are shown.

#### GROUP

# SYSTEMATICS, BIOGEOGRAPHY AND EVOLUTION OF REPTILES AND AMPHIBIANS



#### GROUP MEMBERS

#### 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)

#### PUBLICATIONS 2010

#### ISI Articles

- Beukema, W., de Pous, P., Donaire, D., Escoriza, D., Bogaerts, S., Toxopeus, A.G., de Bie, C.A.J.M., Roca, J., and Carranza, S. 2010. Biogeography and contemporary climatic differentiation among Moroccan Salamandra algira. Biological Journal of the Linnean Society 101: 626-641.
- Cox, S.C, Carranza, S, and Brown, R. 2010. Divergence times and colonization of the Canary Islands by *Gallotia* lizards. *Molecular Phylogenetics and Evolution* 56: 747-757.
- Mila, B., Carranza, S., Guillaume, O., and Clobert, J. 2010. Marked genetic structuring and extreme dispersal limitation in the Pyrenean brook newt *Calotriton asper* (Amphibia: Salamandridae) revealed by genome-wide AFLP but not mtDNA. *Molecular Ecology* 19: 108-120.
- Miralles, A., and Carranza, S. 2010. Systematics and biogeography of the Neotropical genus Mabuya, with special emphasis on the Amazonian skink Mabuya nigropunctata (Reptilia, Scincidae). Molecular Phylogenetics and Evolution 54: 857-869.



Fig. 1: Forest of Dracaena cinnabari, Socotra island.

- Pedraza-Lara, C., Alda, F., Carranza, S., and Doadrio, I. 2010. Mitochondrial DNA structure of the Iberian populations of the white-clawed crayfish Austropotamobius italicus italicus (Faxon, 1914). Molecular Phylogenetics and Evolution 57: 327-342.
- Rato, C., Carranza, S., Perera, A., Carretero, M.A., and Harris, D.J. 2010. Conflicting patterns of nucleotide diversity between mtDNA and nDNA in the Moorish gecko, *Tarentola mauritanica*. *Molecular Phylogenetics and Evolution* 56: 962-971.
- Vasconcelos, R., Carranza, S., and Harris, D.J. 2010. Insight into an island radiation: the *Tarentola* geckos of the Cape Verde archipelago. *Journal of Biogeography* 37: 1047-1060.
- Vasconcelos, R., Froufe, E., Brito, J.C., Carranza, S., and Harris, D.J. 2010.
   Phylogeography of the African Common Toad, *Amietophrynus regularis*, based on mitochondrial DNA sequences: inferences regarding the Cape Verde population and biogeographical patterns. *African Zoology* 45: 291-298.
- Verdú-Ricoy, J., Carranza, S., Salvador, A., Busack, S.D., and Díaz, J.A. 2010. Phylogeography of *Psammodromus algirus* revisited: systematic implications. *Amphibia-Reptilia* 31: 576-582.

#### Other Publications

- Escoriza, D., Metallinou, M., Donaire-Barroso, D., Amat, F., and Carranza, S. 2010. Biogeography of the White-Bellied Carpet Viper Echis leucogaster Roman, 1972 in Morocco, a study combining mitochondrial DNA data and ecological niche modeling. Butlletí de la Societat Catalana d'Herpetologia 18: 55-68.
- Romano, A., Amat, F., Rivera, X., Sotgiu, G., and Carranza, S. 2010. Evidence of tail autotomy in the European plethodontid *Hydromantes* (*Atylodes*) genei (Temmick and Schlegel, 1838) (Amphibia: Urodela: Plethodontidae). Acta Herpetologica 5: 199-205.



Fig. 2: Cerastes gasparetti

#### 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



Tinguaro Montelongo

Anabela Cardoso

Gissela de la Cadena

Jesús Gómez-Zurita

#### **GROUP MEMBERS**

#### Jesús Gómez-Zurita, Group Leader

| Tenured Scientist, CSIC



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 Gondwanian origin. However, this ancient origin is contentious and it was recently proposed that the island would have been colonized recently, after periods of underwater submergence in the Oligocene. The Chrysomelidae fauna in New Caledonia is very rich, with over 200 species described, mostly endemic. In this project, we aim at characterizing their species diversity using morphology and molecular markers, but also to investigate their origin, age and diversification using molecular phylogenetics, as well as their ecologies, based on field observations and the use of molecular markers (see Project 2).



Fig. 1: DNA-extracted Leaf Beetles from Nicaraguan dry forest. Insect diversity in the tropics is overwhelming, and most still remains undescribed. Molecular analyses help us to approach this problem. Image: J. Gómez-Zurita.

#### 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

- Borrero-Pérez, G.H., Gómez-Zurita, J., González-Wangüemert, M., Pérez-Ruzafa, A., and Marcos, C. 2010. Molecular systematics of the genus *Holothuria* in the Mediterranean and Northeastern Atlantic and a molecular clock for the diversification of the Holothuriidae (Echinodermata: Holothuroidea). *Molecular Phylogenetics and Evolution* 57: 899-906.
- Gómez-Zurita, J., Cardoso, A., Jurado-Rivera, J.A., Jolivet, P., Cazères, S., and Mille, C. 2010. Discovery of new species of New Caledonian *Arsipoda* Erichson, 1842 (Coleoptera, Chrysomelidae) and insights on their ecology and evolution using DNA markers. *Journal of Natural History* 44: 2557-2579.

- Lombarte, A., Palmer, M., Matallanas, J., Gómez-Zurita, J., and Morales-Nin, B. 2010. Ecomorphological trends and phylogenetic inertia of otolith sagittae in Nototheniidae. *Environmental Biology of Fishes* 89: 607-618.
- Pinzón Navarro, S., Jurado-Rivera, J.A., Gómez-Zurita, J., Lyal, C.H.C., and Vogler, A.P. 2010. DNA profiling of host-herbivore interactions in tropical forests. *Ecological Entomology* 35 (Suppl. 1): 18-32.

#### **Book Chapters**

 Gómez-Zurita, J. 2010. Tribe Timarchini Motschulsky, 1860. In: I. Löbl and A. Smetana (eds.). Catalogue of Palaearctic Coleoptera. Vol. 6. Chrysomeloidea, pp. 437-443, Apollo Books, Stenstrup, Denmark.

#### Other Publications

 Gómez-Zurita, J., and Petitpierre, E. 2010. Contribution to the knowledge of the Iberian fauna of Chrysomelidae (Coleoptera). I. New records of Criocerinae, Clytrinae and Cryptocephalinae. *Boletín de la Sociedad Entomológica Aragonesa* 47: 139-142.



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.

#### FUNDED PROJECTS

Financed by: Years:	Evolución sin sexo: Circunstancias históricas e implicaciones ecológicas del origen híbrido de insectos unisexuales Ministerio de Ciencia e Innovación (CGL2008-00007) 2009-2011 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
	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
Financed by: Years:	Zoological Systematics and Evolution Research Group – ZOOSYSEVO Generalitat de Catalunya,AGAUR (2009 SGR1462) 2009-2013 Salvador Carranza

#### GROUP WATER AND CAVE BEETLE EVOLUTION



#### Ignacio Ribera, Group Leader

Research Scientist, CSIC



Amparo Hidalgo Galiana, PhD Student | FPI Scholarship, MICINN Valeria Rizzo, Postgraduate Grant | Università La Sapienza, Roma Andrey Rudoy, PhD Student | JAE Scholarship, CSIC

#### RESEARCH OUTLINE

Beetles are the most diverse group of extant metazoa. Their 250 MY of evolutionary history and a vast ecological and morphological variation allow to use them to undertake virtually every problem in evolutionary biology, from global macroecological and macroevolutionary patterns to phylogenetics and biogeography down to population genetics and physiology. We use different groups of water and cave beetles to address some of these questions, centred in the origin and distribution of biodiversity. Our current 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 Catalonian coastal ranges and the pre-Pyrenees. It is a good example of diversification within the subterranean medium, and with the collaboration of some biospeleologists we are trying to understand its origin and evolution.



### 3. Evolution of the complex male genitalia in Hydraenidae

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

Fig. 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

#### PUBLICATIONS 2010

#### ISI Articles

- Faille, A., Bourdeau, C., and Fresneda, J. 2010. A new species of blind Trechinae from the Pyrenees of Huesca, and its position within *Aphaenops* (sensu stricto) (Coleoptera: Carabidae: Trechini). *Zootaxa* 2566: 49-56.
- Faille, A., Ribera, I., Deharveng, L., Bourdeau, C., Garnerye, L., Quéinnec, E., and Deuve, T.A. 2010.
   A molecular phylogeny shows the single origin of the Pyrenean subterranean Trechini ground beetles (Coleoptera: Carabidae). *Molecular Phylogenetics* and Evolution 54: 97-106.
- Hendrich, L., Pons, J., Ribera, I., and Balke, M. 2010. Mitochondrial cox1 sequence data reliably uncover patterns of insect diversity but may suffer from high lineage-idiosyncratic error rates. *PloS ONE* 512: e14448.
- Hidalgo-Galiana, A., Jäch, M.A., and Ribera, I. 2010. Hydrochus farsicus sp.n. from Iran and notes on other Palearctic species of the genus (Coleoptera: Hydrophiloidea: Hydrochidae). Zootaxa 2344: 61-64.
- Pons, J., Ribera, I., Bertranpetit, J., and Balke, M.
   2010. Nucleotide substitution rates for the full set of mitochondrial protein-coding genes in Coleoptera. *Molecular Phylogenetics and Evolution* 56: 796-807.
- Ribera, I., and Faille, A. 2010. A new microphthalmic stygobitic *Graptodytes* Seidlitz from Morocco, with a molecular phylogeny of the genus (Coleoptera, Dytiscidae). *Zootaxa* 2641: 1-14.
- Ribera, I., Castro, A., and Hernando, C. 2010. Ochthebius (Enicocerus) aguilerai sp.n. from central Spain, with a molecular phylogeny of the Western Palaearctic species of Enicocerus (Coleoptera, Hydraenidae). Zootaxa 2351: 1-13.
- Ribera, I., Fresneda, J., Bucur, R., Izquierdo, A., Vogler, A.P., Salgado, J.M., and Cieslak, A. 2010. Ancient origin of a Western Mediterranean radiation of subterranean beetles. BMC Evolutionary Biology 10: 29.



Fig. 2: Baronniesia delioti, a species and genus of cave beetle recently described from the Pyrenees. Image: J. Fresneda

Book Chapters, Refereed Proceedings

- Balke, M., Wewalka, G., Alarie, Y., and Ribera, I. 2010. Dytiscidae: The genus *Rhantus* Dejean (Coleoptera). In: Jäch, M.A., and Balke, M. (eds). Water beetles of New Caledonia, part 1. Zoologisch-Botanische Gesellschaft in Österreich and Wiener Coleopterologenverein, Wien, pp. 129-147.
- Beutel, R.G., Balke, M., and Ribera, I. 2010. 3.1. Aspidytidae Ribera, Beutel, Balke and Vogler, 2002. In: Leschen, R.A.B., Beutel, R.G., and Lawrence, J.F. (eds). Handbook of Zoology, Arthropoda: Insecta. Coleoptera, Beetles. Vol. 2: Morphology and Systematics (Elateroidea, Bostrichiformia, Cucujiformia partim). Walter de Gruyter, Berlin. pp. 21-28.
- Beutel, R.G., Ribera, I., and Balke, M. 2010. 3. Adephaga (Addendum). Introduction and phylogeny. In: Leschen, R.A.B., Beutel, R.G., and Lawrence, J.F. (eds). Handbook of Zoology, Arthropoda: Insecta. Coleoptera, Beetles. Vol. 2: Morphology and Systematics (Elateroidea, Bostrichiformia, Cucujiformia partim). Walter de Gruyter, Berlin. p. 21.

- Casale, A., and Ribera, I. 2010. Are Molopina of the Euro-Mediterranean region related to the Madagascan, South African and Australian Pterostichini? (Coleoptera, Carabidae). *Biogeographia (n.s.)* 29(2008): 33-44.
- Hernando, C., and Ribera, I. 2010. Limnichidae: Description of a new species from New Caledonia, and checklist of the taxa recorded from the Australian/Pacific Region (Coleoptera). In: M.A. Jäch, and Balke, M. (eds). Water beetles of New Caledonia, part 1. Zoologisch-Botanische Gesellschaft in Österreich and Wiener Coleopterologenverein, Wien, pp. 439-449.

#### 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



#### GROUP BUTTERFLY DIVERSITY AND EVOLUTION

Claudia Sañudo

GROUP MEMBERS

Vlad Dinca

#### 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

Roger Vila

Marga Marín

**Gerard Talavera** 

#### 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

- Fiz-Palacios, O., Vargas, P., Vila, R., Papadopulos, A.S.T., and Aldasoro, J.J. 2010. The uneven phylogeny and biogeography of *Erodium* (Geraniaceae): Radiations in the Mediterranean and recent recurrent intercontinental colonization. *Annals of Botany* 106: 871-884.
- Vila, R., Lukhtanov, V.A., Talavera, G., Gil-T., F., and Pierce, N.E. 2010. How common are dot-like distribution ranges? Taxonomical oversplitting in Western European *Agrodiaetus* (Lepidoptera, Lycaenidae) revealed by chromosomal and molecular markers. *Biological Journal of the Linnean Society* 101: 130154.



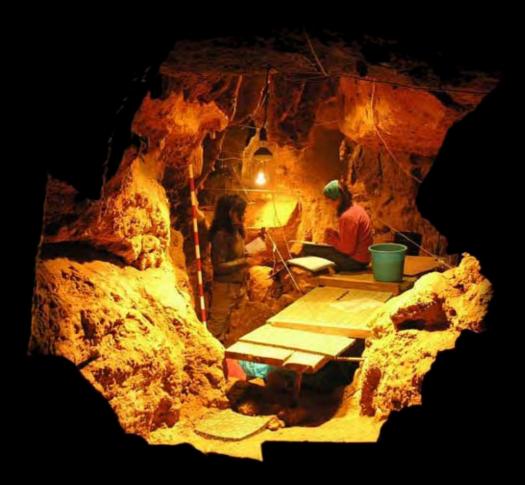
Fig. 2: Discovering cryptic biodiversity is one of our main lines of research. Careful morphological and molecular studies are needed to distinguish between very similar species, as is the case with the Aricia specimens in the picture. Image: Vlad Dinca

#### Other Publications

• Dinca, V., .Cuvelier, S., Zakharov, E.V., Hebert, P.D.N., and Vila, R. 2010. Biogeography, ecology and conservation of Erebia oeme (Hübner) in the Carpathians (Lepidoptera: Nymphalidae: Satyrinae). *Annales de la Societe Entomologique de France* 46: 486-498.

#### FUNDED PROJECTS

Financed by: Years:	Combining morphological, cytological and molecular data to study the taxonomy of Iberian Rhopalocera (Lepidoptera: Hesperioidea + Papilionoidea) Ministerio de Ciencia e Innovación (Plan Nacional I+D+I) 2007-2010 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
	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:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía.
Financed by: Years:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía. 2009-2013
Financed by: Years:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía.
Financed by: Years: PI:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía. 2009-2013 Alberto Tinaut Grup de Recerca Consolidat-SGR "Grup de Genòmica,
Financed by: Years: PI: Project Title:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía. 2009-2013 Alberto Tinaut
Financed by: Years: PI: Project Title: Funded by: Years:	de la alta montaña andaluza: Parnassius apollo. Relaciones con las poblaciones y subespecies ibéricas e implicaciones para su conservación. Proyecto de Investigación de Excelencia. Junta de Andalucía. 2009-2013 Alberto Tinaut Grup de Recerca Consolidat-SGR "Grup de Genòmica, Bioinformàtica i Evolució"



# PROGRAM comparative and computational genomics

#### **RESEARCH GROUPS**

Genomics of Individuality Francesc Calafell, Group Leader

Paleogenomics Carles Lalueza-Fox, Group Leader

Evolutionary Genomics

Arcadi Navarro, Group Leader

#### **Primate Genomics**

Tomàs Marquès-Bonet, Group Leader

In the Comparative and Computational Genomics program, genomes are compared at the intra and inter-specific levels with the general aims of understanding genome dynamics, reconstructing the evolutionary processes that generate biodiversity and linking genomic and phenotypic differences between individuals and species. To achieve these goals, we deploy both experimental and theoretical/numerical approaches, with a strong emphasis in computational techniques. Genomes contain a wealth of information, not only about how phenotypes are shaped in interaction with the environment, but also about the evolutionary history 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



GROUP MEMBERS

## Francesc Calafell, Group Leader

Associate Professor, UPF



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

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

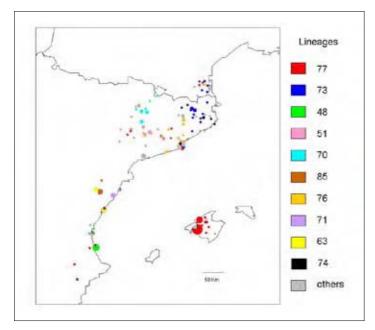
#### ISI Articles

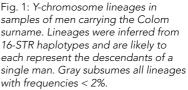
- Calafell, F., Almasy, M., Sabater-Lleal, A., Buil, A., Mordillo, C., Ramírez-Soriano, A., Sikora, M., Souto, J.C., Blangero, J., Fontcuberta, J., and Soria, J.M. 2010. Sequence variation and genetic evolution at the human *F12* locus: mapping quantitive trait nucleotides that influence FXII plasma levels. *Human Molecular Genetics* 19: 517-525.
- Laayouni, H., Calafell, F., and Bertranpetit, J. 2010. A genome-wide survey does not show the genetic distinctiveness of Basques. *Human Genetics* 127: 455-458.
- Melé, M., Javed, A., Pybus, M., Calafell, F., Parida, L., Bertranpetit, J., and The Genographic Consortium. 2010. A new method to reconstruct recombination events at a genomic scale. *PLoS Computational Biology* 6 (11): e1001010.
- Moreno-Estrada, A., Aparicio-Prat, E., Sikora, M., Engelken, J., Ramírez-Soriano, A., Calafell, F., and Bosch, E. 2010. African signatures of recent positive selection in human FOXI1. BMC Evolutionary Biology 10: 267.

#### 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
DI.	lauma Partranaatit

PI: Jaume Bertranpetit





## program 🥝 comparative and computational genomics



## GROUP MEMBERS

## 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.

## PUBLICATIONS 2010

#### ISI Articles

- de Torres, T., Ortiz, J.E., Grün, R., Eggins, S., Valladas, H., Mercier, N., Tisnérat-Laborde, N., Juliá, R., Soler, V., Martínez, E., Sánchez-Moral, S., Cañaveras, J.C., Lario, J., Badal, E., Lalueza-Fox, C., Rosas, A., Santamaría, D., de La Rasilla, M., and Fortea, J. 2010. Dating of the Hominid (Homo neanderthalensis) remains accumulation from El Sidrón cave (Piloña, Asturias, north Spain): an example of multi-methodological approach to the dating of Upper Pleistocene sites. *Archaeometry* 52 (4): 680-705.
- Frayer, D.W., Fiore, I., Lalueza-Fox, C., Radovcić, J., and Bondioli, L. 2010. Right handed Neandertals: Vindija and beyond. *Journal of Anthropological Science* 88: 113-127.
- Green, R.E., Krause, J., Briggs, A.W., Maricic, T., Stenzel, U., Kircher, M., Patterson, N., et al. 2010. A draft sequence of the Neandertal genome. *Science* 328: 710-722.
- Ramirez, O., Illera, J.C., Rando, J.C., González-Solís, J., Alcover, J.A., and Lalueza-Fox, C. 2010. Ancient DNA of the Extinct Lava Shearwater (Puffinus olsoni) from the Canary Islands Reveals Incipient Differentiation within the *P. puffinus* Complex. *PLoS ONE* 5 (12): e16072.
- Zidi, A., Amills, M., Tomás, A., Vidal, O., Ramírez, O., Carrizosa, J., Urrutia, B., Serradilla, J.M., and Clop, A. 2010. Short communication: genetic variability in the predicted microRNA target sites of caprine casein genes. *J Dairy Sci* 93: 1749-1753.

#### Other Publications

 Santamaría, D., Fortea, J., de la Rasilla, M., Martínez, L., Martínez, E., Cañaveras, J.C., Sánchez-Moral, S., Rosas, A., Estalrrich, A., García-Tabernero, A., and Lalueza-Fox, C.
 2010. The technological and typological behaviour of a Neanderthal group from El Sidrón cave (Asturias, Spain). The Technological and typological behaviour of a Neanderthal group from El Sidrón cave (Asturias, Spain). Oxford Journal of Archaeology 29 (2): 119-148.

Fig. 1: Ancient Iberian Lynx mandible



**Book Chapters** 

- Lalueza-Fox, C. 2010. ADN i arqueologia. In: Restes de vida, restes de mort; la mort en la prehistòria. Publicacions del Museu de Prehistòria de València, Diputació de València. ISBN: 978-84-7795-557-3.
- Lalueza-Fox, C. 2010. Genómica Neandertal. In: Encuentros con la Ciencia II. Del macrocosmos al microcosmos (ed. Enrique Viguera, Ana Grande y José Lozano). Publicaciones de la Universidad de Málaga. ISBN: 978-84-9747-339-2.
- Lalueza-Fox, C. 2010. El Proyecto Genoma Neandertal. In: Fósiles y moléculas. Aproximaciones a la historia evolutiva de *Homo sapiens* (ed. Antonio González-Martín). Memorias de la Real Sociedad Española de Historia Natural. Segunda época. ISBN: 978-84-936677-5-7.
- Lalueza-Fox, C. 2010. El Proyecto Genoma Neandertal: ¿hacia una definición genética de nuestra especie? In: 150 años después de Darwin: ¿evolución, futuro o crisis?. Edited by Instituto Tomás Pascual Sanz-CENIEH. ISBN: 978-84-7867-057-4.

#### 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 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

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 proteincoding 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.

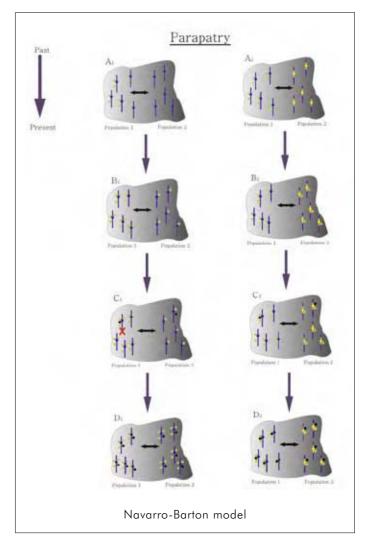
## PUBLICATIONS 2010

#### ISI Articles

- Al-Shahrour, F., Mínguez, P., Marquès-Bonet, T., Gazave, E., Navarro, A., and Dopazo, J. 2010. Selection upon genome architecture: conservation of functional clusters with changing genes. *PLoS Computational Biology* 6 (10): e1000953.
- Camiña-Tato, M., Fernández, M., Morcillo-Suárez, C., Navarro, A., Julià, E., Edo, M.C., Montalban, X., and Comabella, M. 2010. Genetic association of CASP8 polymorphisms with primary progressive multiple sclerosis. *Journal of Neuroimmunology* 222: 70-75.
- Camiña-Tato, M., Morcillo-Suárez, C., Fernández, M., Martín, R., Ortega, I., Navarro, A., Sánchez, A., Carmona, P., Julià, E., Tortola, M., Audí, L., Fossdal, R., Oksenberg, J.R., Montalban, X., and Comabella, M. 2010. Gender associated differences of perforin polymorphisms in the susceptibility to multiple sclerosis. *The Journal of Immunology* 185: 5392-5404.
- Faria, R., and Navarro, A. 2010. Chromosomal speciation: rearranging theory with pieces of evidence. *Trends in Ecology and Evolution* 25: 660-669.
- Pettifer et al. and the INB Partners (Including A. Navarro). 2010. The EMBRACE web service collection. *Nucleic Acids Research* (Advanced Online Publication May 12).

#### **Book Chapters**

 Morcillo, C., Albà, M.M., and Navarro, A. 2010. Genoma y Enfermedades Complejas. In: Eclerosis Múltiple. Ed: Pablo Villoslada. Marge Medica Books (Barcelona).



## FUNDED PROJECTS

Financed by: Years:	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 CSIC-CRUSA (2009CR0028) 2010-2011 Arcadi Navarro
Financed by: Years:	Identifying Evolutionary Novelties and Adaptation in Duplicated Regions of the Genomes of Primates Ministerio de Educación y Ciencia (BFU2009-13409-C02-02) 2009-2012 Arcadi Navarro
Financed by: Years:	Exploring the behavioral genetics of Trade and Cooperation Ministerio de Educación y Ciencia (MEC-SEJ2007-30267-E/SOCI) 2008-2011 Arcadi Navarro
Years:	IMID-Kit Ministerio de Educación y Ciencia (MEC-PSS-010000-2008-36) 2008-2010 Arcadi Navarro and Jaume Bertranpetit (Coordinator: S. Marsal)
Years:	INB GN8 Genoma España (Instituto Nacional de Bioinformática) 2003-2010 Arcadi Navarro
Financed by: Years:	Red Española de Esclerosis Múltiple Ministerio de Ciencia e Innovación. Instituto Carlos III (ISCIII-RD07/0060/2021) 2009-2011 Arcadi Navarro

## program 🥝 comparative and computational genomics

## GROUP PRIMATE GENOMICS

Tomàs Marquès-Bonet

Javier Prado

Marcos Fernández

Belén Lorente

Marta Melè rene Hernando

**GROUP MEMBERS** 

## 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

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.

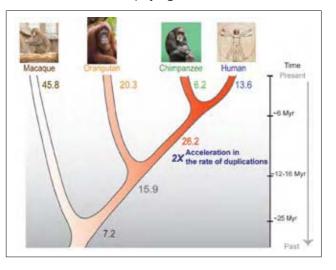
As a part of the pilot project for the ERC Starting Grant, we have sequenced Snowflake, the only known albino gorilla to perform comparative genomic approaches, both inter and intra-species, to study mutations that could explain the phenotype in this unique gorilla.

#### 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).



## PUBLICATIONS 2010

#### ISI Articles

- Al-Shahrour, F., Mínguez, P., Marquès-Bonet, T., Gazave, E., Navarro, A., and Dopazo, J.
   2010. Selection upon Genome Architecture: Conservation of Functional Neighborhoods with Changing Genes. *PLoS Computational Biology* 6: 10.
- Antonacci, F., Kidd, J.M., Marquès-Bonet, T., Teague, B., Ventura, M., Girirajan, S., Alkan, C., Campbell, C.D., Vives, L., Malig, M., Rosenfeld, J.A., Ballif, B.C., Shaffer, L.G., Graves, T.A., Wilson, R.K., Schwartz, D.C., and Eichler, E.E. 2010. A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. *Nature Genetics* 42: 745-U729.
- Fernando, C.A., Conrad, P.A., Bartels, C.F., Marquès, T., To, M., Balow, S.A., Nakamura, Y., and Warman, M.L. 2010. Temporal and Spatial Expression of CCN Genes in Zebrafish. Developmental Dynamics 239: 1755-1767.

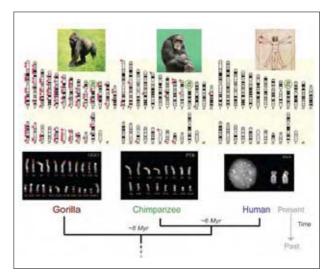


Fig. 2: A particular duplication expansion in chimpanzee and gorilla. This segment of DNA is single copy in humans. The green circle indicates the ancestral location (donor). The red arrows indicate the derivative locations in each specie based on location in the human karyotype.

- Green, R.E., Krause, J., Briggs, A.W., Maricic, T., Stenzel, U., Kircher, M., Patterson, N., Li, H., Zhai, W., Fritz, M.H., Hansen, N.F., Durand, E.Y., Malaspinas, A.S., Jensen, J.D., Marquès-Bonet, T., Alkan, C., Prufer, K., Meyer, M., Burbano, H.A., Good, J.M., Schultz, R., Aximu-Petri, A., Butthof, A., Hober, B., Hoffner, B., Siegemund, M., Weihmann, A., Nusbaum, C., Lander, E.S., Russ, C., Novod, N., Affourtit, J., Egholm, M., Verna, C., Rudan, P., Brajkovic, D., Kucan, Z., Gusic, I., Doronichev, V.B., Golovanova, L.V., Lalueza-Fox, C., de la Rasilla, M., Fortea, J., Rosas, A., Schmitz, R.W., Johnson, P.L., Eichler, E.E., Falush, D., Birney, E., Mullikin, J.C., Slatkin, M., Nielsen, R., Kelso, J., Lachmann, M., Reich, D., and Pääbo, S. 2010. A draft sequence of the Neandertal genome. *Science* 328: 710-722.
- Reich, D., Green, R.E., Kircher, M., Krause, J., Patterson, N., Durand, E.Y., Viola, B., Briggs, A.W., Stenzel, U., Johnson, P.L.F., Maricic, T., Good, J.M., Marquès-Bonet, T., Alkan, C., Fu, Q., Mallick, S., Li, H., Meyer, M., Eichler, E.E., Stoneking, M., Richards, M., Talamo, S., Shunkov, M.V., Derevianko, A.P., Hublin, J.J., Kelso, J., Slatkin, M., and Pääbo, S. 2010. Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature* 468: 1053-1060.

#### Other Publications

• Marquès-Bonet, T., and Comas, D. 2010. Com es pot mesurar la variabilitat humana? *Omnis Cellula* 25: 8-9.

#### 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
  - Pl: Tomàs Marquès-Bonet

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Ovarioles from Blattella germanica ovary. DAPI (blue), rhodamine-phalloidin (red) and tubuline (green). Image: Nashwa Elshaer and M. Dolors Piulachs

## PROGRAM functional evolution in insects

## RESEARCH GROUPS

Evolution of Insect Metamorphosis Xavier Bellés, Group Leader

Insect Reproduction Maria-Dolors Piulachs, Group Leader

## Nutritional Signals in Insects José Luis Maestro, Group Leader

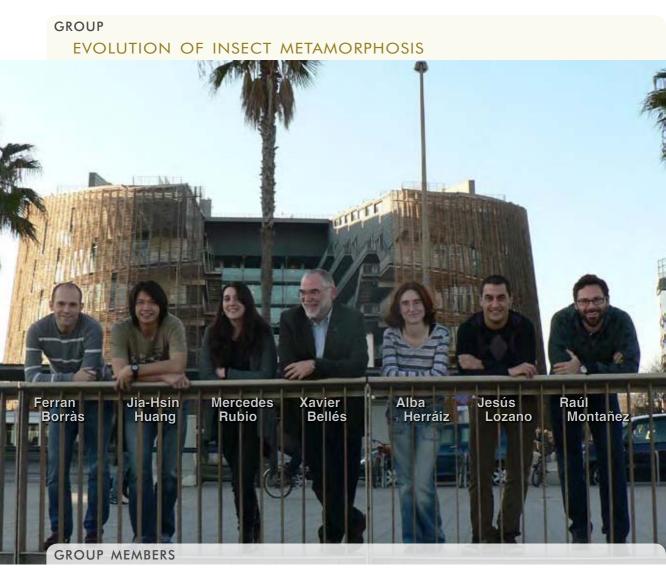
## Hormonal Control of Insect Development David Martín, Group Leader

Drosophila Telomeres Elena Casacuberta, Group Leader

## Developmental Biology and Morphological Evolution in Insects

Xavier Franch-Marro, Group Leader

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.



## 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

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

- Bellés, X. 2010. Beyond *Drosophila*. RNAi in vivo and functional genomics in insects. *Annual Review of Entomology* 55: 111-128.
- Maestro, J.L., Pascual, N., Treiblmayr, K., Lozano, J., and Bellés, X. 2010. Juvenile hormone and allatostatins in the German cockroach embryo. *Insect Biochemistry and Molecular Biology* 40: 660-665.
- Mané-Padrós, D., Cruz, J., Vilaplana, L., Nieva, C., Ureña, E., Bellés, X., and Martín, D. 2010. The hormonal pathway controlling cell death during metamorphosis in a hemimetabolous insect. *Developmental Biology* 346: 150-160.
- Montanez, R., Medina, M.A., Solé, R.V., and Rodriguez-Caso, C. 2010. When metabolism meets topology: Reconciling metabolite and reaction networks. *Bioessays* 32: 246-256.
- Piulachs, M.D., Pagone, V., and Bellés, X. 2010. Key roles of Broad-Complex transcription factors in insect embryogenesis. *Insect Biochemistry and Molecular Biology* 40: 468-475.

#### Books

- Bellés, X. 2010. Bestiari. Monografíes Mètode. Universitat de València, 96 pages.
- Bellés, X. 2010. Vivir dos vidas. Un viaje por la metamorfosis de los insectos. Casaciencias, prisma Casa de las Ciencias, La Voz de Galicia A Coruña, 112 pages.

#### Other Publications

• Bellés, X. 2010. Darwin e a expresión das emocións. Agora do Orcellón 19: 13-16.

#### FUNDED PROJECTS -----

Financed by: Years:	Clearing bacteroids with RNAi Ministerio de Ciencia e Innovación (REF: CGL2010-09266-E, Explora program) 2010-2011 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

program / functional evolution in insects

## 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.

#### **RESEARCH SUBLINES**

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.

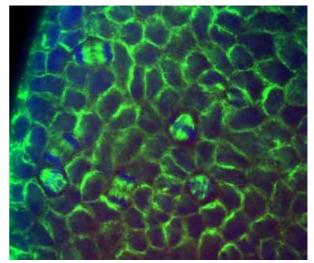


Fig. 1: Mitosis figures in the follicular epithelia from an ovary of a newly emerged adult. DAPI (blue) and tubuline (green). Image: Laura Ciudad and M. Dolors Piulachs

## **PUBLICATIONS 2010**

#### ISI Articles

• Piulachs, M.D., Pagone, V., and Bellés, X. 2010. Key roles of the Broad-Complex gene in insect embryogenesis. Insect Biochemistry and Molecular Biology 40: 468-475.

#### 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 Pls: Xavier Bellés and Francisco Bozinovic

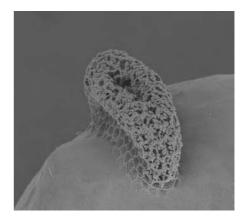


Fig. 2: SEM micrograph of the Sponge-like body from a Blattella germanica egg. Image: Paula Irles and M. Dolors Piulachs

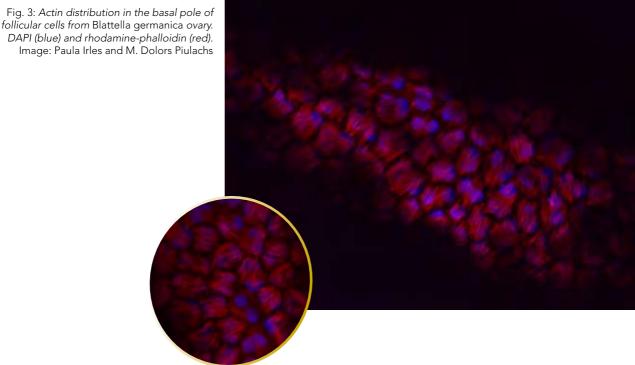
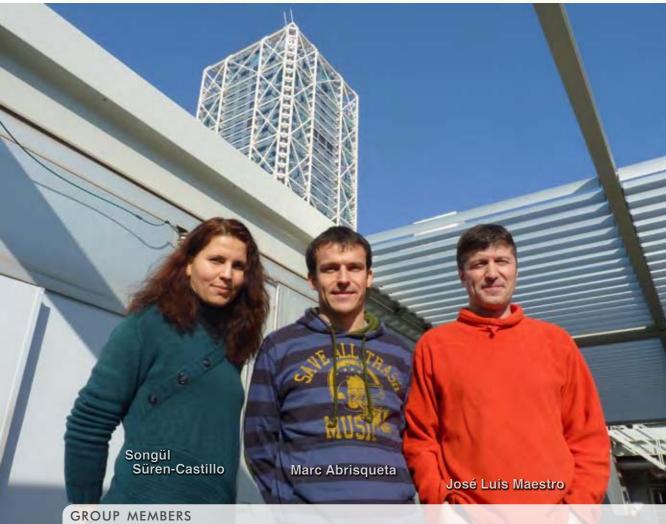


Image: Paula Irles and M. Dolors Piulachs

## GROUP NUTRITIONAL SIGNALS IN INSECTS



## José Luis Maestro, Group Leader

| Tenured Scientist, CSIC



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

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).

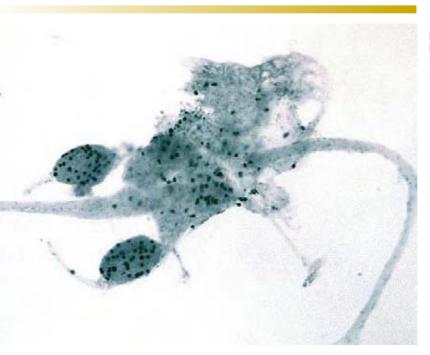


Fig. 1: Retrocerebral complex from Blattella germanica 3-day old 6th instar nymph stained for detection of BrdU labelling.

#### 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

• Maestro, J.L., Pascual, N., Treiblmayr, K., Lozano, J., and Bellés, X. 2010. Juvenile hormone and allatostatins in the German cockroach embryo. *Insect Biochemistry and Molecular Biology* 40: 660-665.

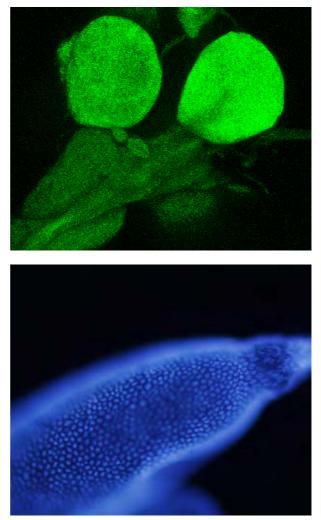


Fig. 2: Corpora allata from a 5-day old Blattella germanica adult female stained with fluorescein diacetate.

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

Cristina Manjón

Enric Ureña

## GROUP HORMONAL CONTROL OF INSECT DEVELOPMENT

Ferran Borràs

## 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

David Martín

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

- Mané-Padrós, D., Cruz, J., Vilaplana, L., Nieva, C., Ureña, E., Bellés, X., and Martín, D. 2010. The hormonal pathway controlling cell death during metamorphosis in a hemimetabolous insect. *Developmental Biology* 346: 150-160.
- Pankotai, T., Popescu, C., Martín, D., Grau, B., Zsindely, N., Bodai, L., Tora, L., Ferrús, A., and Boros, I. 2010. "Genes of the ecdysone biosynthesis pathway are regulated by the dATAC histone acetyltransferase complex in *Drosophila*". *Molecular and Cellular Biology* 30: 4254-4266.
- Talamillo, A., Martín, D., Hjerpe, R., Sánchez, J., and Barrio, R. 2010. "SUMO and ubiquitin modifications during steroid hormone synthesis and function". *Biochemical Society Transactions* 38: 54-59.
- Soin, T., De Geyter, E., Mosallanejad, H., Iga, M., Martín, D., Ozaki, S., Kitsuda, S., Harada, T., Miyagawa, H., Stefanou, D., Kotzia, G., Efrose, R., Labropoulou, V., Geelen, D., Latrou, K., Nakagawa, Y., Smagghe, G., and Swevers, L. 2010. "Assessment of species specificity of molting accelerating compounds in lepidopteran insects: comparison of activity between *Bombyx mori* and *Spodoptera littoralis* by *in vitro* reporter assay and *in vivo* toxicity assays". *Pest Management Science* 66: 526-535.

#### **Book Chapters**

• Martín, D. 2010. Functions of Nuclear Receptors in Insect Development. In: Nuclear Receptors; Current concepts and future challenges (series: Proteins and Cell Regulation). Bunce, C.M., and Campbell, M.J. (eds). Springer Publishers (UK). Vol. 8, pp. 31-61.

## FUNDED PROJECTS

 Project Title: Molecular Basis of Ecdysteroid and Juvenile Hormone Actions in Insect Development. The Role of Nuclear Hormone Receptors. (REF: BFU2009-10571)
 Financed by: Ministerio de Ciencia e Innovación Years: 2010-2012
 PI: David Martín

Fig. 2: RNAi-mediated knockdown of SUMO showing impaired wing development at the adult stage of the cockroach Blattella germanica.

## GROUP DROSOPHILA TELOMERES

Elisenda López

Elena Casacuberta

**Rute Sousa** 

David Piñeyro

## GROUP MEMBERS

## Elena Casacuberta, Group Leader

| Tenured Scientist, CSIC



David Piñeyro, Post-doc Researcher | CSIC Contract Rute Sousa, PhD Student | PhD Fellowship, Fundaçao para a Ciência e Tecnologia, Portugal Elisenda López Panadès, PhD Student | UPF Contract

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.

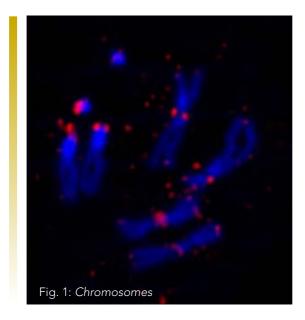
#### **RESEARCH SUBLINES**

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 telomere length and genome stability. Understanding the regulation of the telomeric transposons will shed light in both telomere length control as well as transposon regulation in *Drosophila*. We are currently focusing 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.



#### 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.

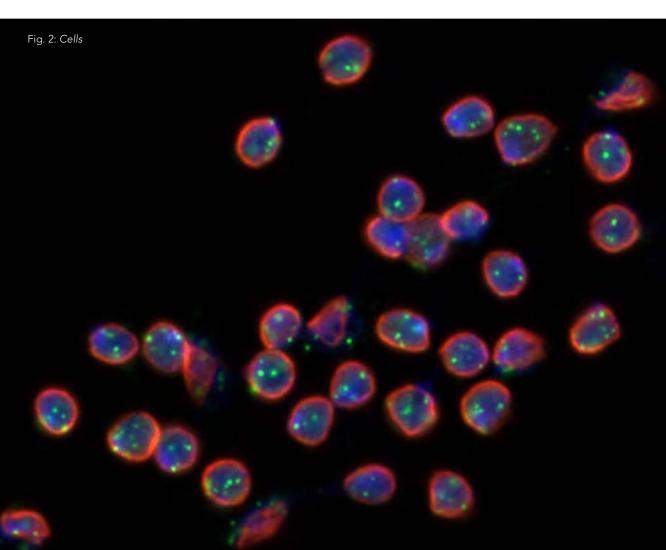
#### 4. Variability within the telomere retrotransposons

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<br/>de DrosophilaFinanced by:Ministerio de Ciencia e Innovación (BFU2009-08318)<br/>Years:Years:2010-2012

PI: Elena Casacuberta Suñe



## 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

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.

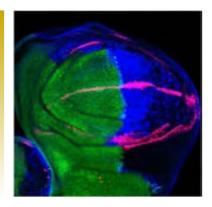


Fig. 1: Wing imaginal disc of Drosophila with Posterior compartment mutant for Wls, marked with abscense of GFP. Wg protein, shown in red, accumulates in the secreting cells of wls mutant cells.

## 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* not *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 analize 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 signalling cascades, which lead to changes in gene transcription. It has already shown that Wingless, the main Wnt *Drosophila* member, is post-transcriptional modified and then secreted by its dedicated seven-pass-transmembrane protein Wntless (Wls). However, the function of these post-transcriptional modifications is still poor understood. Thus, our goal is to understand the mechanism that controls Wg secretion by Wls and the function of its post-transcriptional modifications.

#### 4. Wingless signaling 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

 Wendler, F., Gillingham, A.K., Sinka, R., Rosa-Ferreira, C., Gordon, D.E., Franch-Marro, X., Peden, A.A., Vincent, J.P., Munro, S. 2010.
 A genome-wide RNA interference screen identifies two novel components of the metazoan secretory pathway. *Embo Journal* 29: 304-314.

## FUNDED PROJECTS

Project Title:	Formación del Gradiente del Morfógeno y de la función de
	Wingless en el control del crecimiento
Financed by:	Ministerio de Ciencia e Innovación (REF: BFU2009-08748)
Years:	2010-2012
PI:	Xavier Franch-Marro
Proiect Title:	Función de la vía de Wingless en el control del crecimiento y sus
.,	implicaciones evolutivas
Financed by:	CSIC-Proyectos Intramurales Especiales (REF: 2009201209)

- Year: 2010
  - Pl: Xavier Franch-Marro

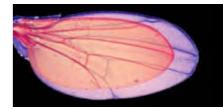


Fig. 2: Drosophila adult wings. Overexpression of Wg signalling promotes a 30% size increase of this organ.





## PROGRAM population genetics

## **RESEARCH GROUPS**

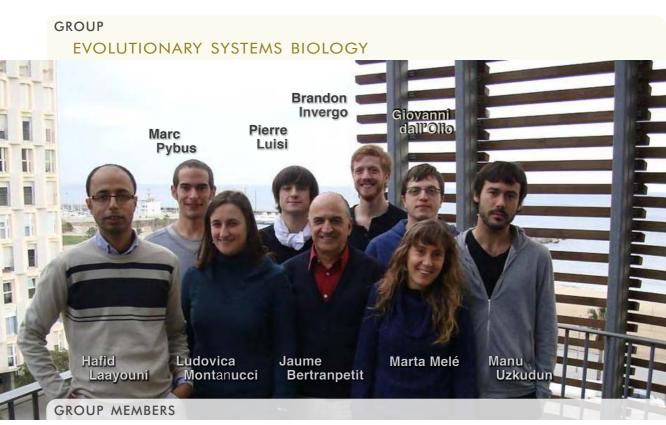
Evolutionary Systems Biology Jaume Bertranpetit, Group Leader

Evolutionary Population Genetics Elena Bosch, Group Leader

Human Genome Diversity David Comas, Group Leader

microRNAs in Human Adaptation and Disease Yolanda Espinosa-Parrilla, *Group Leader* 

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



## Jaume Bertranpetit, Group Leader

Professor, UPF



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

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

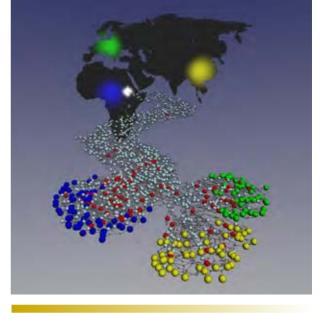
#### ISI Articles

- Acuna-Alonzo, V., Flores-Dorantes, T., Kruit, J.K., Villarreal-Molina, T., Arellano-Campos, O., Hunemeier, T., Moreno-Estrada, A., Ortiz-López, M.G., Villamil-Ramírez, H., León-Mimila, P., Villalobos-Comparan, M., Jacobo-Albavera, L., Ramírez-Jiménez, S., Sikora, M., Zhang, L.H., Pape, T.D., Granados-Silvestre, M.D., Montufar-Robles, I., Tito-Álvarez, A.M., Zurita-Salinas, C., Bustos-Arriaga, J., Cedillo-Barrón, L., Gómez-Trejo, C., Barquera-Lozano, R., Vieira, J.P., Granados, J., Romero-Hidalgo, S., Huertas-Vázquez, A., González-Martín, A., Gorostiza, A., Bonatto, S.L., Rodríguez-Cruz, M., Wang, L., Tusie-Luna, T., Aguilar-Salinas, C.A., Lisker, R., Moises, R.S., Menjivar, M., Salzano, F.M., Knowler, W.C., Bortolini, M.C., Hayden, M.R., Baier, L.J., and Canizales-Quinteros, S. 2010. *Human Molecular Genetics* 19: 2877-2885.
- Laayouni, H., Calafell, F., and Bertranpetit, J. 2010. A genome-wide survey does not show the genetic distinctiveness of Basques. *Human Genetics* 127: 455-458.
- Melé, M., Javed, A., Pybus, M., Calafell, F., Parida, L., Bertranpetit, J., and Genographic Consortium Members. 2010. A new method to reconstruct recombination events at a genomic scale. *PLoS Comput Biol* 6 (11): e1001010.
- Pons, J., Ribera, I., Bertranpetit, J., and Balke, M. 2010. Nucleotide substitution rates for the full set of mitochondrial protein-coding genes in Coleoptera. *Molecular Phylogenetics and Evolution* 56: 796-807.

As Part of the Genographic Consortium

- Haak, W., Balanovsky, O., Sánchez, J.J., Koshel, S., Zaporozhchenko, V., Adler, C.J., Der Sarkissian, C.S., Brandt, G., Schwarz, C., Nicklisch, N., Dresely, V., Fritsch, B., Balanovska, E., Villems, R., Meller, H., Alt, K.W., Cooper, A., and Consortium G. 2010. Ancient DNA from European early neolithic farmers reveals their near eastern affinities. *PLoS Biol* 8 (11): e1000536.
- Qin, Z.D., Yang, Y.J., Kang, L.L., Yan, S., Cho, K., Cai, X.Y., Lu, Y., Zheng, H.X., Zhu, D.C., Fei, D.M., Li, S.L., Jin, L., Li, H., and Consortium, G. 2010.
   A Mitochondrial Revelation of Early Human Migrations to the Tibetan Plateau Before and After the Last Glacial Maximum. *American Journal of Physical Anthropology* 143: 555-569.
- Zhadanov, S.I., Dulik, M.C., Markley, M., Jennings, G.W., Gaieski, J.B., Elias, G., Schurr, T.G., and Genographic Consortium Members. 2010. Genetic heritage and native identity of the Seaconke Wampanoag tribe of Massachusetts. *American Journal of Physical Anthropology* 142 (4): 579-589.

Fig. 1: This picture shows a simulated coalescent network generated with cosi (Shaffner et al. 2005) that represents the history of humankind with an Out of Africa event and the emergence of the different human groups, Africans, Europeans and Asians. African individuals appear in blue, Asian individuals in yellow and Europeans in green. The cyan nodes represent past recombination events, whereas the small gray nodes represent coalescent events. Red nodes are those past recombination events that were recovered by IRiS. These kind of coalescent networks were used to fine-tune and validate IRiS. Software used Pajek (http://vlado.fmf. uni-lj.si/pub/networks/pajek/). Figure by: Marc Pybus



#### **Book Chapters**

 Bertranpetit, J. Prólogo. In: Fósiles y moléculas. Aproximaciones a la historia evolutiva de Homo sapiens. Ed. Antonio González-Martín. Memoria de la Real Sociedad de Historia Natural. Segunda Época. Tomo VIII. 169-201. 2010. ISSN: 1132-0869; ISBN: 978-84-936677-5-7.

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.
- Rellegint el missatge genètic: gens, genomes, diversitat i comprensió de la vida i dels humans. Memorias de la Real Academia de Ciencias y Artes de Barcelona. Núm. 1025, Vol. LXIV, NÚM. 8. Barcelona, 2010.

## 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
Pls:	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 EVOLUTIONARY POPULATION GENETICS

Elena Bosch Mònica Vallès

Johannes Engelken

## Anna-Lena Scherr Núria Bonet

Elena Carnero

## **GROUP MEMBERS**

## Elena Bosch, Group Leader

Associate Professor, UPF



Johannes Engelken, Post-doc | Volkswagenstiftung Scholarship, Germany

Elena Carnero, PhD Student | UPF Teaching Scholarship

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

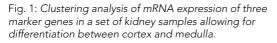
By means of a resequencing approach, we have confirmed previous SNP-based evidences of recent positive selection in East Asian populations in a lymphocyte receptor gene, which is involved in recognition of fungal cell walls. We are now currently investigating quantitative and qualitative functional differences 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 Clínic (Barcelona).

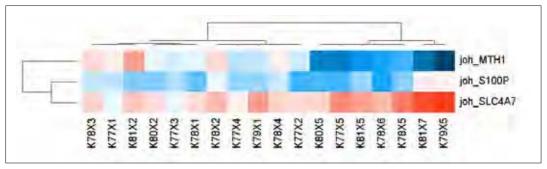
## 2. Recent human adaptation and nutrition

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

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

We are obtaining sequence data at both intraspecific and interespecific 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).





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.

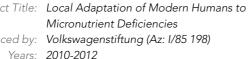
## **PUBLICATIONS 2010**

#### ISI Articles

- Engelken, J., Brinkmann, H., and Adamska, I. 2010. Taxonomic distribution and origins of the extended LHC (light-harvesting complex) antenna protein superfamily. BMC Evolutionary Biology 10:233.
- Moreno-Estrada, A., Aparicio-Prat, E., Sikora, M., Engelken, J., Ramírez-Soriano, A., Calafell, F., and Bosch, E. 2010. African signatures of recent positive selection in human FOXI1. BMC Evolutionary Biology 10:267.
- Myles, S., Lea, R.A., Ohashi, J., Chambers, G.K., Weiss, J.G., Hardouin, E., Engelken, J., Macartney-Coxson, D.P., Eccles, D.A., Naka, I., Kimura, R., Inaoka, T., Matsumura, Y., and Stoneking, M. 2010. Testing the thrifty gene hypothesis: the Gly482Ser variant in PPARGC1A is associated with BMI in Tongans. BMC Evol Biol 10:233.

## FUNDED PROJECTS

Project Title:	Detección de la selección natural en genes candidatos: aproximación a las bases de la adaptación humana
Financed by:	Subdirección General de Proyectos de
	Investigación (BFU2008-01046)
Years:	2009-2011
PI:	Elena Bosch
Project Title:	Grup de Recerca Consolidat-SGR
Financed by:	Generalitat de Catalunya (2009 SGR-1101)
Years:	2009-2013
PI:	Jaume Bertranpetit
Project Title:	Local Adaptation of Modern Humans to
,	Micronutrient Deficiencies
Financed bv:	Volkswagenstiftung (Az: 1/85 198)
V	5 5.



Pls: Elena Bosch and Mark Stoneking

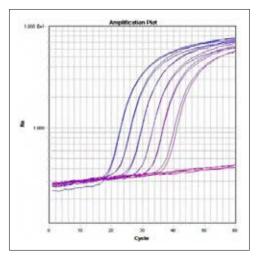


Fig. 2: qRT-PCR amplification covering cDNA concentrations diluted across 5 log ranges.

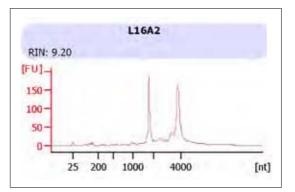


Fig. 3: RNA quality check of extracted tissue samples using Agilent 1200 Bioanalyzer.





## GROUP 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

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

- Atkinson, Q.D., Barbujani, G., Collard, M., Comas, D., and Franceschi, C. 2010. Interdisciplinary views on Molecular Anthropology in the Genomic Era. *Journal* of Anthropological Sciences 88: 231-250.
- Behar, D.M., Yunusbayev, B., Metspalu, M., Metspalu, E., Rosset, S., Parik, J., Rootsi, S., Chaubey, G., Kutuev, I., Yudkovsky, G., Khusnutdinova, E.K., Balanovsky, O., Semino, O., Pereira, L., Comas, D., Gurwitz, D., Bonne-Tamir, B., Parfitt, T., Hammer, M.F., Skorecki, K., and Villems, R. 2010. The genome-wide structure of the Jewish people. *Nature* 466 (7303): 238-242.
- Donnelly, M.P., Paschou, P., Grigorenko, E., Gurwitz, D., Mehdi, S.Q., Kajuna, S.L., Barta, C., Kungulilo, S., Karoma, N.J., Lu, R.B., Zhukova, O.V., Kim, J.J., Comas, D., Siniscalco, M., New, M., Li, P., Li, H., Manolopoulos, V.G., Speed, W.C., Rajeevan, H., Pakstis, A.J., Kidd, J.R., and Kidd, K.K. 2010. The distribution and most recent common ancestor of the 17q21 inversion in humans. *Am J Hum Genet* 86 (2): 161-171.
- Viviani, R, Sim, E.J., Lo, H., Beschoner, P., Osterfeld, N., Maier, C., Seeringer, A., Godoy, A.L., Rosa, A., Comas, D., and Kirchheiner, J. 2010. Baseline brain perfusion and the serotonin transporter promoter polymorphism. *Biol Psychiatry* 67 (4): 317-22.

As Part of the Genographic Consortium

- Comas, D. 2010. The Genographic Project: insights into Western/Central European variation. *Journal of Anthropological Science* 88: 243-244.
- Haak, W., Balanovsky, O., Sánchez, J.J., Koshel, S., Zaporozhchenko, V., Adler, C.J., Der Sarkissian, C.S., Brandt, G., Schwarz, C., Nicklisch, N., Dresely, V., Fritsch, B., Balanovska, E., Villems, R., Meller, H., Alt, K.W., Cooper, A., and Consortium G. 2010. Ancient DNA from European early neolithic farmers reveals their near eastern affinities. *PLoS Biol* 8 (11): e1000536.
- Melé, M., Javed, A., Pybus, M., Calafell, F., Parida, L., Bertranpetit, J., and Genographic Consortium Members. 2010. A new method to reconstruct recombination events at a genomic scale. *PLoS Comput Biol* 6 (11): e1001010.
- Qin, Z., Yang, Y., Kang, L., Yan, S., Cho, K., Cai, X., Lu, Y., Zheng, H., Zhu, D., Fei, D., Li, S., Jin, L., Li, H., and Genographic Consortium Members. 2010. A mitochondrial revelation of early human migrations to the Tibetan Plateau before and after the last glacial maximum. *Am J Phys Anthropol* 143 (4): 555-569.
- Zhadanov, S.I., Dulik, M.C., Markley, M., Jennings, G.W., Gaieski, J.B., Elias, G., Schurr, T.G., and Genographic Consortium Members. 2010. Genetic heritage and native identity of the Seaconke Wampanoag tribe of Massachusetts. *American Journal of Physical Anthropology* 142 (4): 579-589.

#### Other Publications

- Comas, D. 2010. Biodiversidad en poblaciones humanas. SEBBM (Sociedad Española de Bioquímica y Biología Molecular), 165: 24-28.
- Comas, D. 2010. Evolució i diversitat del genoma humà. l'Atzavara 19: 61-68.
- Marquès-Bonet, T., and Comas, D. 2010. Com es pot mesurar la variabilitat humana? *Omnis Cellula* 25: 8-9.



## FUNDED PROJECTS

Financed by: Years:	Diversidad genética en poblaciones humanas de África Central: implicaciones demográficas, lingüísticas y culturales (CGL2007-61016/BOS) Dirección General de Investigación Científica y Técnica 2007-2010 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)
Years:	Dirección General de Investigación Científica y Técnica 2010-2012 David Comas
Financed by: Years:	Grup de Recerca en Biologia Evolutiva (2009SGR1101) Direcció General de Recerca, Comissionat per a Universitats i Recerca, Generalitat de Catalunya 2009-2012 Jaume Bertranpetit
Financed by: Years:	The Genographic project: Western/Central Europe region National Geographic and IBM 2006-2010 Jaume Bertranpetit and David Comas



## GROUP MEMBERS

## Yolanda Espinosa-Parrilla, Group Leader

Marie Curie Researcher, UPF



María López, PhD Student | FI Scholarship, AGAUR

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 noncoding 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 anwer 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, microRNAdependent 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 wit 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

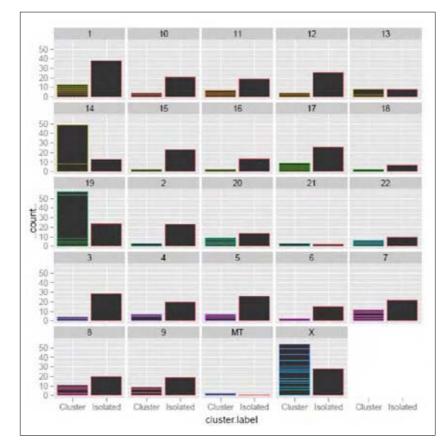
- Guidi, M., Muiños-Gimeno, M., Kagerbauer, B., Martí, E., Estivill, X., and Espinosa-Parrilla, Y. 2010. Overexpression of miR-128 specifically inhibits the truncated isoform of NTRK3 and upregulates BCL2 in SH-SY5Y neuroblastoma cells. BMC Molecular Biology 11: 95.
- Muiños-Gimeno, M., Montfort, M., Bayés, M., Estivill, X., and Espinosa-Parrilla, Y. 2010. Design and evaluation of a panel of Single Nucleotide Polymorphisms (SNPs) in microRNA genomic regions for association studies in human disease. *European Journal of Human Genetics* 18 (2): 218-226.

## FUNDED PROJECTS

Project Tittle: 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.

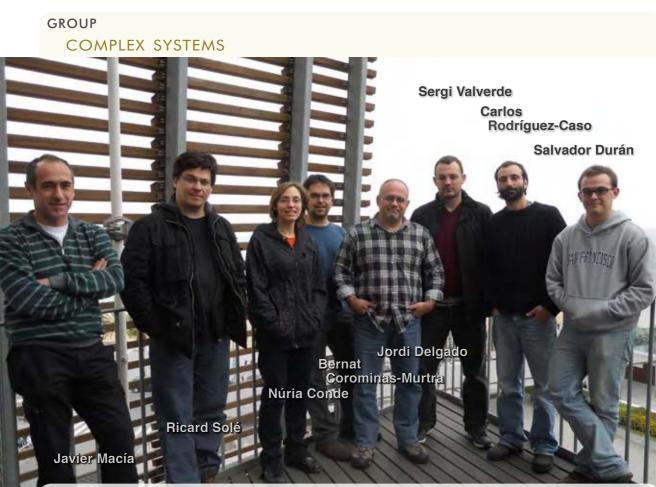


# PROGRAM complex systems

## **RESEARCH GROUPS**

Complex Systems Ricard Solé, Group Leader

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 enfasis is taken in the study of computational biology, protocell biology, synthetic systems and network biology. In particular, in the study of viruses dynamics, tissue architecture, ecological networks, cancer systems, language networks, etc.



## GROUP MEMBERS

## 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 SUBLINES

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

## **PUBLICATIONS 2010**

ISI Articles

- Corominas-Murtra, B. 2010. Percolation of arbitrary uncorrelated nested subgraphs. *European Physical Journal B* 77: 213-217.
- Corominas-Murtra, B., Rodríguez-Caso, C., Goñi, J., and Solé, R. 2010. Topological reversibility and causality in feed-forward networks. *New Journal of Physics* 12: 11.
- Corominas-Murtra, B., and Solé, R.V. 2010. Universality of Zipf's law. *Physical Review* E 82: 1.
- Elena, S.F., Solé, R.V., and Sardanyes, J. 2010. Simple genomes, complex interactions: epistasis in RNA virus. *Chaos* 20: 026106.
- Goni, J., Corominas-Murtra, B., Solé, R.V., and Rodríguez-Caso, C. 2010. Exploring the randomness of directed acyclic networks. *Phys Rev E Stat Nonlin Soft Matter Phys* 82: 066115.
- Goni, J., Martincorena, I., Corominas-Murtra, B., Arrondo, G., Ardanza-Trevijano, S., and Villoslada, P. 2010. Switcher-Random-Walks: A Cognitive-Inspired Mechanism for Network Exploration. *International Journal of Bifurcation and Chaos* 20: 913-922.
- Joppa, L.N., Montoya, J.M., Solé, R.V., Sanderson, J., and Pimm, S.L. 2010. On nestedness in ecological networks. *Evolutionary Ecology Research* 12: 35-46.
- Montanez, R., Medina, M.A., Solé, R.V., and Rodríguez-Caso, C. 2010. When metabolism meets topology: Reconciling metabolite and reaction networks. *Bioessays* 32: 246-256.
- Munteanu, A., Constante, M., Isalan, M., and Solé, R.V. 2010. Avoiding transcription factor competition at promoter level increases the chances of obtaining oscillation. *BMC Syst Biol* 4: 66.
- Solé, R.V. 2010. Genome Size, Self-Organization and DNA's Dark Matter. *Complexity* 16: 20-23.
- Solé, R.V., Corominas-Murtra, B., and Fortuny, J. 2010. Diversity, competition, extinction: the ecophysics of language change. *J R Soc Interface* 7: 1647-1664.
- Solé, R.V., Corominas-Murtra, B., Valverde, S., and Steels, L. 2010. Language Networks: Their Structure, Function, and Evolution. *Complexity* 15: 20-26.

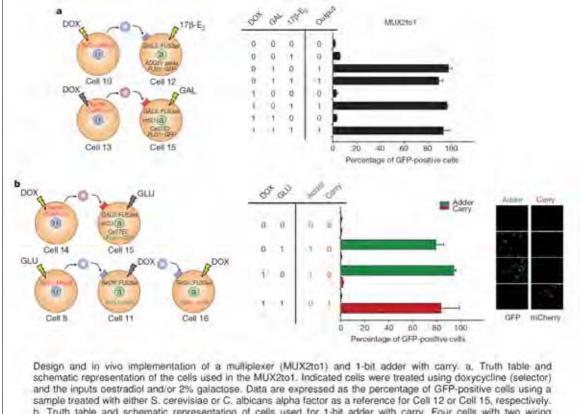
• Solé, R.V., Saldana, J., Montoya, J.M., and Erwin, D.H. 2010. Simple model of recovery dynamics after mass extinction. *J Theor Biol* 267: 193-200.

#### **Book Chapters**

- Corominas-Murtra, B., Valverde, S., and Solé, R.V. 2010. Emergence of Scale-Free Syntax Networks. In Evolution of Communication and Language in Embodied Agents, pp. 83--101. Springer Verlag Berlín, Heidelberg, Germany.
- Solé, R.V. 2010. Catastrophes and Complex Networks. In: Multiscale Cancer Modeling pp. 67-86. Ed: T. Deisboeck and G. StakamatoChapman & Hall-CRG Press. (Virginia)

Other Publications

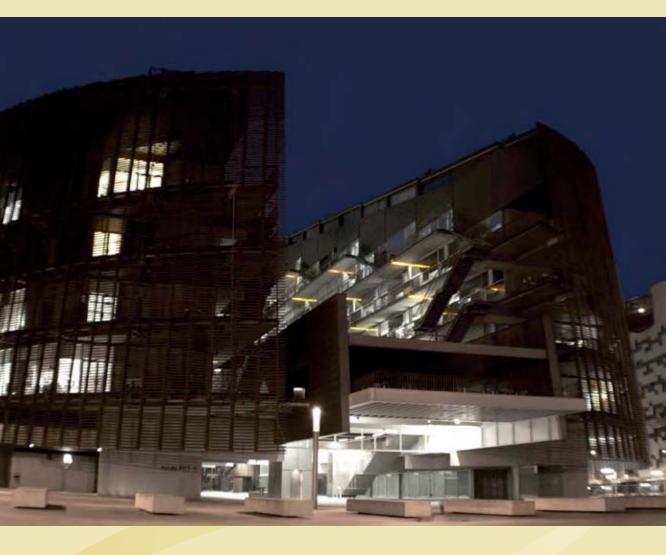
- Fortuny, J., and Corominas-Murtra, B. 2010. Some formal considerations on the generation of hierarchically structured expressions. *Catalan Journal of Linguistics* 8: 99-111.
- Horta-Bernús, R., Rosas-Casals, M., and Valverde, S. 2010. Discerning Electricity Consumption Patterns from Urban Allometric Scaling. 2010 Complexity in Engineering: Compeng 2010, Proceedings 49-51.



b, Truth table and schematic representation of cells used for 1-bit adder with carry. Four cells with two wiring systemsthat respond to glucose and doxycycline with an XOR logic were combined with an extra cell that respond to same stimuli but with an AND logic in which instead of GFP, mCherry was expressed as output. The final outcome was measured as in Fig. 3a. Green bars indicate the adder output (GFP) whereas red bars represent the carry bit (mCherry). GFP and mCherry images of cells are shown (right panels). Data represent the mean and standard deviation of three independent experiments. (Figure from Regot et al. (2010) Nature 467, 207-11)

## FUNDED PROJECTS

Financed by: Years:	Origins of innovation in tinkered networks McDonnell Foudation USA (McDonell Award 220020117) 2007-2011 Ricard Solé
Years:	SYNLET FP7-European Union 2007-2010 Ricard Solé
Financed by: Years:	COMPLEXDIS - Unravelling complex diseases with complexity theory FP7-European Union 2007-2010 Ricard Solé
Financed by: Years:	Development of virtual tumor National Institute of Health (Grant N°: CA11304-3 and 03S3 CFDA N° 93397) 2008-2010 Ricard Solé
Financed by: Years:	Computación, replicación y rotura de simetría en sistemas protocelulares Ministerio de Ciencia e Innovación (FIS2009-12365) 2010-2012 Ricard Solé
Financed by: Years:	Convenio de colaboración en materia de apoyo a la transferencia tecnológica en el campo de la biotecnología, prof. Ricard Solé. Fundación Marcelino Botín 2009-2010 Ricard Solé



## SCIENTIFIC PUBLICATIONS

#### ISI Articles

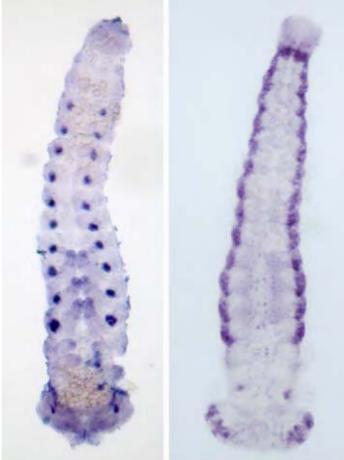
- Acuna-Alonzo, V., Flores-Dorantes, T., Kruit, J.K., Villarreal-Molina, T., Arellano-Campos, O., Hunemeier, T., Moreno-Estrada, A., Ortiz-López, M.G., Villamil-Ramírez, H., León-Mimila, P., Villalobos-Comparan, M., Jacobo-Albavera, L., Ramírez-Jiménez, S., Sikora, M., Zhang, L.H., Pape, T.D., Granados-Silvestre, M.D., Montufar-Robles, I., Tito-Álvarez, A.M., Zurita-Salinas, C., Bustos-Arriaga, J., Cedillo-Barrón, L., Gómez-Trejo, C., Barquera-Lozano, R., Vieira, J.P., Granados, J., Romero-Hidalgo, S., Huertas-Vázquez, A., González-Martín, A., Gorostiza, A., Bonatto, S.L., Rodríguez-Cruz, M., Wang, L., Tusie-Luna, T., Aguilar-Salinas, C.A., Lisker, R., Moisés, R.S., Menjivar, M., Salzano, F.M., Knowler, W.C., Bortolini, M.C., Hayden, M.R., Baier, L.J., and Canizales-Quinteros, S. 2010. A functional ABCA1 gene variant is associated with low HDL-cholesterol levels and shows evidence of positive selection in Native Americans. *Human Molecular Genetics* 19: 2877-2885.
- Al-Shahrour, F., Mínguez, P., Marquès-Bonet, T., Gazave, E., Navarro, A., and Dopazo, J. 2010. Selection upon Genome Architecture: Conservation of Functional Neighborhoods with Changing Genes. *PLoS Computational Biology* 6: 10.
- Antonacci, F., Kidd, J.M., Marquès-Bonet, T., Teague, B., Ventura, M., Girirajan, S., Alkan, C., Campbell, C.D., Vives, L., Malig, M., Rosenfeld, J.A., Ballif, B.C., Shaffer, L.G., Graves, T.A., Wilson, R.K., Schwartz, D.C., and Eichler, E.E. 2010. A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. *Nature Genetics* 42: 745-U729.
- Atkinson, Q.D., Barbujani, G., Collard, M., Comas, D., and Franceschi, C. 2010. Interdisciplinary views on Molecular Anthropology in the Genomic Era. *Journal of Anthropological Sciences* 88:231-250.
- Behar, D.M., Yunusbayev, B., Metspalu, M., Metspalu, E., Rosset, S., Parik, J., Rootsi, S., Chaubey, G., Kutuev, I., Yudkovsky, G., Khusnutdinova, E.K., Balanovsky, O., Semino, O., Pereira, L., Comas, D., Gurwitz, D., Bonne-Tamir, B., Parfitt, T., Hammer, M.F., Skorecki, K., and Villems, R. 2010. The genome-wide structure of the Jewish people. *Nature* 466 (7303): 238-242.
- Bellés, X. 2010. Beyond Drosophila. RNAi in vivo and functional genomics in insects. *Annual Review of Entomology* 55: 111–128.
- Beukema, W., de Pous, P., Donaire, D., Escoriza, D., Bogaerts, S., Toxopeus, A.G., de Bie, C.A.J.M., Roca, J., and Carranza, S. 2010. Biogeography and contemporary climatic differentiation among Moroccan Salamandra algira. Biological Journal of the Linnean Society 101: 626-641.

- Borrero-Pérez, G.H., Gómez-Zurita, J., González-Wangüemert, M., Pérez-Ruzafa, A., and Marcos, C. 2010. Molecular systematics of the genus *Holothuria* in the Mediterranean and Northeastern Atlantic and a molecular clock for the diversification of the Holothuriidae (Echinodermata: Holothuroidea). *Molecular Phylogenetics and Evolution* 57: 899-906.
- Calafell, F., Almasy, M., Sabater-Lleal, A., Buil, A., Mordillo, C., Ramírez-Soriano, A., Sikora, M., Souto, J.C., Blangero, J., Fontcuberta, J., and Soria, J.M. 2010. Sequence variation and genetic evolution at the human F12 locus: mapping quantitive trait nucleotides that influence FXII plasma levels. *Human Molecular Genetics* 19:517-525.
- Camiña-Tato, M., Fernández, M., Morcillo-Suárez, C., Navarro, A., Juli, E., Edo, M.C., Montalban, X., and Comabella, M. 2010. Genetic association of CASP8 polymorphisms with primary progressive multiple sclerosis. *Journal of Neuroimmunology* 222:70-75.
- Camiña-Tato, M., Morcillo-Suárez, C., Fernández, M., Martín, R., Ortega, I., Navarro, A., Sánchez, A., Carmona, P., Julià, E., Tortola, M., Audí, L., Fossdal, R., Oksenberg, J.R., Montalban, X., and Comabella, M. 2010. Gender associated differences of perforin polymorphisms in the susceptibility to multiple sclerosis. *The Journal of Immunology* 185: 5392 -5404.
- Comas, D. 2010. The Genographic Project: insights into Western/Central European variation. *Journal of Anthropological Science* 88: 243-244.
- Corominas-Murtra, B. 2010. Percolation of arbitrary uncorrelated nested subgraphs. *European Physical Journal* B 77: 213-217.
- Corominas-Murtra, B., Rodríguez-Caso, C., Goñi, J., and Solà, R. 2010. Topological reversibility and causality in feed-forward networks. *New Journal of Physics* 12: 11.
- Corominas-Murtra, B., and Solé, R.V. 2010. Universality of Zipf's law. *Physical Review* E 82: 1.
- Cox, S.C, Carranza, S., and Brown, R. 2010. Divergence times and colonization of the Canary Islands by *Gallotia* lizards. *Molecular Phylogenetics and Evolution* 56: 747-757.
- de Torres, T., Ortiz, J.E., Grün, R., Eggins, S., Valladas, H., Mercier, N., Tisnérat-Laborde, N., Juliá, R., Soler, V., Martínez, E., Sánchez-Moral, S., Cañaveras, J.C., Lario, J., Badal, E., Lalueza-Fox, C., Rosas, A., Santamaría, D., de La Rasilla, M., and Fortea, J. 2010. Dating of the Hominid (Homo neanderthalensis) remains accumulation from El Sidrón cave (Piloìa, Asturias, north Spain): an example of multi-methodological approach to the dating of Upper Pleistocene sites. *Archaeometry* 52 (4): 680-705.
- Donnelly, M.P., Paschou, P., Grigorenko, E., Gurwitz, D., Mehdi, S.Q., Kajuna, S.L., Barta, C., Kungulilo, S., Karoma, N.J., Lu, R.B., Zhukova, O.V., Kim, J.J., Comas, D., Siniscalco, M., New, M., Li, P., Li, H., Manolopoulos, V.G., Speed, W.C., Rajeevan, H., Pakstis, A.J., Kidd, J.R., and Kidd, K.K. 2010. The distribution and most recent common ancestor of the 17q21 inversion in humans. *Am J Hum Genet* 86 (2): 161-171.
- Elena, S.F., Solé, R.V., and Sardanyes, J. 2010. Simple genomes, complex interactions: epistasis in RNA virus. *Chaos* 20: 026106.
- Engelken, J., Brinkmann, H., and Adamska, I. 2010. Taxonomic distribution and origins of the extended LHC (light-harvesting complex) antenna protein superfamily. *BMC Evolutionary Biology* 10:233.
- Faille, A., Bourdeau, C., and Fresneda, J. 2010. A new species of blind Trechinae from the Pyrenees of Huesca, and its position within *Aphaenops* (sensu stricto) (Coleoptera: Carabidae: Trechini). *Zootaxa* 2566: 49-56.
- Faille, A., Ribera, I., Deharveng, L., Bourdeau, C., Garnerye, L., Quéinnec, E., and Deuve, T.A. 2010. A molecular phylogeny shows the single origin of the Pyrenean subterranean Trechini ground beetles (Coleoptera: Carabidae). *Molecular Phylogenetics and Evolution* 54: 97-106.

- Faria, R., and Navarro, A. 2010. Chromosomal speciation: rearranging theory with pieces of evidence. *Trends in Ecology and Evolution* 25: 660-669.
- Fernando, C.A., Conrad, P.A., Bartels, C.F., Marquès, T., To, M., Balow, S.A., Nakamura, Y., and Warman, M.L. 2010. Temporal and Spatial Expression of CCN Genes in Zebrafish. *Developmental Dynamics* 239: 1755-1767.
- Fiz-Palacios, O., Vargas, P., Vila, R., Papadopulos, A.S.T., and Aldasoro, J.J. 2010. The uneven phylogeny and biogeography of *Erodium* (Geraniaceae): Radiations in the Mediterranean and recent recurrent intercontinental colonization. *Annals of Botany* 106: 871-884.
- Frayer, D.W., Fiore, I., Lalueza-Fox, C., Radovcić, J., and Bondioli,
   L. 2010. Right handed Neandertals: Vindija and beyond. *Journal of Anthropological Science* 88:113-127.
- Gómez-Zurita, J., Cardoso, A., Jurado-Rivera, J.A., Jolivet, P., Cazères, S., and Mille, C. 2010. Discovery of new species of New Caledonian Arsipoda Erichson, 1842 (Coleoptera, Chrysomelidae) and insights on their ecology and evolution using DNA markers. Journal of Natural History 44: 2557-2579.
- Goni, J., Corominas-Murtra, B., Solé, R.V., and Rodríguez-Caso, C. 2010.
   Exploring the randomness of directed acyclic networks. *Phys Rev E Stat Nonlin Soft Matter Phys* 82: 066115.
- Goni, J., Martincorena, I., Corominas-Murtra, B., Arrondo, G., Ardanza-Trevijano, S., and Villoslada, P. 2010. Switcher-Random-Walks: A Cognitive-Inspired Mechanism for Network Exploration. International Journal of Bifurcation and Chaos 20: 913-922.
- Green, R.E., Krause, J., Briggs, A.W., Maricic, T., Stenzel, U., Kircher, M., Patterson, N., Li, H., Zhai, W., Fritz, M.H., Hansen, N.F., Durand, E.Y., Malaspinas, A.S., Jensen, J.D., Marquès-Bonet, T., Alkan, C., Prufer, K., Meyer, M., Burbano, H.A., Good, J.M., Schultz, R., Aximu-Petri, A., Butthof, A., Hober, B., Hoffner, B., Siegemund, M., Weihmann, A., Nusbaum, C., Lander, E.S., Russ, C., Novod, N., Affourtit, J., Egholm, M., Verna, C., Rudan, P., Brajkovic, D., Kucan, Z., Gusic, I., Doronichev, V.B., Golovanova, L.V., Lalueza-Fox, C., de la Rasilla, M., Fortea, J., Rosas, A., Schmitz, R.W., Johnson, P.L., Eichler, E.E., Falush, D., Birney, E., Mullikin, J.C., Slatkin, M., Nielsen, R., Kelso, J., Lachmann, M., Reich, D., and Pääbo, S. 2010. A draft sequence of the Neandertal genome. *Science* 328: 710-722.
- Guidi, M., Muiños-Gimeno, M., Kagerbauer, B., Martí, E., Estivill, X., and Espinosa-Parrilla, Y. 2010. Overexpression of miR-128 specifically inhibits the truncated isoform of NTRK3 and upregulates BCL2 in SH-SY5Y neuroblastoma cells. *BMC Molecular Biology* 11: 95.
- Haak, W., Balanovsky, O., Sánchez, J.J., Koshel, S., Zaporozhchenko, V., Adler, C.J., Der Sarkissian, C.S., Brandt, G., Schwarz, C., Nicklisch, N., Dresely, V., Fritsch, B., Balanovska, E., Villems, R., Meller, H., Alt, K.W., and Cooper, A. 2010. Ancient DNA from European early neolithic farmers reveals their near eastern affinities. Members of the Genographic Consortium. *PLoS Biol* 8 (11):e1000536.
- Hendrich, L., Pons, J., Ribera, I., and Balke, M. 2010. Mitochondrial cox1 sequence data reliably uncover patterns of insect diversity but may suffer from high lineageidiosyncratic error rates. *PloS ONE* 512: e14448.

Wadi Ayoun, Dhofar, Southern Oman

- Hidalgo-Galiana, A., Jäch, M.A., and Ribera, I. 2010. Hydrochus farsicus sp.n. from Iran and notes on other Palearctic species of the genus (Coleoptera: Hydrophiloidea: Hydrochidae). *Zootaxa* 2344: 61-64.
- Igea, J., Juste, J., and Castresana, J. 2010. Novel intron markers to study the phylogeny of closely related mammalian species. *BMC Evolutionary Biology* 10: 369.
- Joppa, L.N., Montoya, J.M., Solé, R.V., Sanderson, J., and Pimm, S.L. 2010. On nestedness in ecological networks. *Evolutionary Ecology Research* 12: 35-46.
- Laayouni, H., Calafell, F., and Bertranpetit, J. A genome-wide survey does not show the genetic distinctiveness of Basques. *Human Genetics* 127:455-458.
- Lombarte, A., Palmer, M., Matallanas, J., Gómez-Zurita, J., and Morales-Nin, B. 2010. Ecomorphological trends and phylogenetic inertia of otolith sagittae in Nototheniidae. *Environmental Biology of Fishes* 89: 607-618.
- Maestro, J.L., Pascual, N., Treiblmayr, K., Lozano, J., and Bellés, X. 2010. Juvenile hormone and allatostatins in the German cockroach embryo. *Insect Biochemistry and Molecular Biology* 40: 660-665.
- Mané-Padrós, D., Cruz, J., Vilaplana, L., Nieva, C., Ureña, E., Bellés, X., and Martín, D. 2010. The hormonal pathway controlling cell death during metamorphosis in a hemimetabolous insect. *Developmental Biology* 346: 150-160.
- Melé, M., Javed, A., Pybus, M., Calafell, F., Parida, L., Bertranpetit, J., and Genographic Consortium Members.
   2010. A new method to reconstruct recombination events at a genomic scale. *PLoS Computational Biology* 6 (11): e1001010.
- Mila, B., Carranza, S., Guillaume, O., and Clobert, J. 2010. Marked genetic structuring and extreme dispersal limitation in the Pyrenean brook newt *Calotriton asper* (Amphibia: Salamandridae) revealed by genome-wide AFLP but not mtDNA. *Molecular Ecology* 19: 108-120.
- Miralles, A., and Carranza, S. 2010. Systematics and biogeography of the Neotropical genus Mabuya, with special emphasis on the Amazonian skink Mabuya nigropunctata (Reptilia, Scincidae). Molecular Phylogenetics and Evolution 54: 857-869.
- Montanez, R., Medina, M.A., Solé, R.V., and Rodríguez-Caso, C. 2010. When metabolism meets topology: Reconciling metabolite and reaction networks. *Bioessays* 32: 246-256.
- Moreno-Estrada, A., Aparicio-Prat, E., Sikora, M., Engelken, J., Ramírez-Soriano, A., Calafell, F., and Bosch, E. 2010. African



In situ staining of two tracheal genes in embryos of Tribolium castaneum.

signatures of recent positive selection in human FOXI1. *BMC Evolutionary Biology* 10:267.

- Muiños-Gimeno, M., Montfort, M., Bayés, M., Estivill, X., and Espinosa-Parrilla, Y. 2010. Design and evaluation of a panel of Single Nucleotide Polymorphisms (SNPs) in microRNA genomic regions for association studies in human disease. *European Journal of Human Genetics* 18 (2): 218-226.
- Munteanu, A., Constante, M., Isalan, M., and Solé, R.V. 2010. Avoiding transcription factor competition at promoter level increases the chances of obtaining oscillation. *BMC Syst Biol* 4: 66.
- Myles, S., Lea, R.A., Ohashi, J., Chambers, G.K., Weiss, J.G., Hardouin, E., Engelken, J., Macartney-Coxson, D.P., Eccles, D.A., Naka, I., Kimura, R., Inaoka, T., Matsumura, Y., and Stoneking, M. 2010. Testing the thrifty gene hypothesis: the Gly482Ser variant in PPARGC1A is associated with BMI in Tongans. *BMC Evol Biol* 10:233.
- Pankotai, T., Popescu, C., Martín, D., Grau, B., Zsindely, N., Bodai, L., Tora, L., Ferrús, A., and Boros, I. 2010. "Genes of the ecdysone biosynthesis pathway are regulated by the dATAC histone acetyltransferase complex in Drosophila". *Molecular and Cellular Biology* 30: 4254-4266.
- Pedraza-Lara, C., Alda, F., Carranza, S., and Doadrio, I. 2010. Mitochondrial DNA structure of the Iberian populations of the white-clawed crayfish Austropotamobius italicus italicus (Faxon, 1914). Molecular Phylogenetics and Evolution 57: 327-342.
- Pettifer, S., Ison, J., Kalas, M., Thorne, D., McDermott, P., Jonassen, I., Liaquat, A., Fernández, J.M., Rodríguez, J.M., Pisano, D.G., Blanchet, C., Uludag, M., Rice, P., Bartaseviciute, E., Rapacki, K., Hekkelman, M., Sand, O., Stockinger, H., Clegg, A.B., Bongcam-Rudloff, E., Salzemann, J., Breton, V., Attwood, T.K., Cameron, G., Vriend, G., and INB-Partners. 2010. The EMBRACE web service collection. *Nucleic Acids Research* 38: W683-W688.
- Pinzón Navarro, S., Jurado-Rivera, J.A., Gómez-Zurita, J., Lyal, C.H.C., and Vogler, A.P. 2010. DNA profiling of host-herbivore interactions in tropical forests. *Ecological Entomology* 35 (Suppl. 1): 18-32.
- Piulachs, M.D., Pagone, V., and Bellés, X. 2010. Key roles of Broad-Complex transcription factors in insect embryogenesis. *Insect Biochemistry and Molecular Biology* 40: 468-475.
- Pons, J., Ribera, I., Bertranpetit, J., and Balke, M. 2010. Nucleotide substitution rates for the full set of mitochondrial protein-coding genes in Coleoptera. *Molecular Phylogenetics and Evolution* 56 (2): 796-807.
- Qin, Z., Yang, Y., Kang, L., Yan, S., Cho, K., Cai, X., Lu, Y., Zheng, H., Zhu, D., Fei, D., Li, S., Jin, L., Li, H., and Genographic Consortium Members. 2010. A mitochondrial revelation of early human migrations to the Tibetan Plateau before and after the last glacial maximum. *Am J Phys Anthropol* 143 (4): 555-569.
- Ramírez, O., Illera, J.C., Rando, J.C., González-Solís, J., Alcover, J.A., and Lalueza-Fox, C. 2010. Ancient DNA of the Extinct Lava Shearwater (Puffinus olsoni) from the Canary Islands Reveals Incipient Differentiation within the P. puffinus Complex. *PLoS ONE* 5 (12): e16072.
- Rato, C., Carranza, S., Perera, A., Carretero, M.A., and Harris, D.J. 2010. Conflicting patterns of nucleotide diversity between mtDNA and nDNA in the Moorish gecko, *Tarentola mauritanica. Molecular Phylogenetics and Evolution* 56: 962-971.
- Reich, D., Green, R.E., Kircher, M., Krause, J., Patterson, N., Durand, E.Y., Viola, B., Briggs, A.W., Stenzel, U., Johnson, P.L.F., Maricic, T., Good, J.M., Marquès-Bonet, T., Alkan, C., Fu, Q., Mallick, S., Li, H., Meyer, M., Eichler, E.E., Stoneking, M., Richards, M., Talamo, S., Shunkov, M.V., Derevianko, A.P., Hublin, J.J., Kelso, J., Slatkin, M., and

Pääbo, S. 2010. Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature* 468: 1053-1060.

- Ribera, I., and Faille, A. 2010. A new microphthalmic stygobitic *Graptodytes* Seidlitz from Morocco, with a molecular phylogeny of the genus (Coleoptera, Dytiscidae). *Zootaxa* 2641: 1-14.
- Ribera, I., Castro, A., and Hernando, C. 2010. *Ochthebius (Enicocerus) aguilerai* sp.n. from central Spain, with a molecular phylogeny of the Western Palaearctic species of *Enicocerus* (Coleoptera, Hydraenidae). *Zootaxa* 2351: 1-13.
- Ribera, I., Fresneda, J., Bucur, R., Izquierdo, A., Vogler, A.P., Salgado, J.M., and Cieslak, A. 2010. Ancient origin of a Western Mediterranean radiation of subterranean beetles. *BMC Evolutionary Biology* 10: 29.
- Sánchez-Gracia, A., Romero-Pozuelo, J., and Ferrús, A. 2010. Two frequenins in *Drosophila:* unveiling the evolutionary history of an unusual neuronal calcium sensor (NCS) duplication. *BMC Evolutionary Biology* 10: 54.
- Soin, T., De Geyter, E., Mosallanejad, H., Iga, M., Martín, D., Ozaki, S., Kitsuda, S., Harada, T., Miyagawa, H., Stefanou, D., Kotzia, G., Efrose, R., Labropoulou, V., Geelen, D., Latrou, K., Nakagawa, Y., Smagghe, G., and Swevers, L. 2010. "Assessment of species specificity of molting accelerating compounds in lepidopteran insects: comparison of activity between *Bombyx mori* and *Spodoptera littoralis* by in vitro reporter assay and in vivo toxicity assays". *Pest Management Science* 66: 526-535.
- Solé, R.V. 2010. Genome Size, Self-Organization and DNA's Dark Matter. *Complexity* 16: 20-23.
- Solé, R.V., Corominas-Murtra, B., and Fortuny, J. 2010. Diversity, competition, extinction: the ecophysics of language change. *J R Soc Interface* 7: 1647-1664.
- Solé, R.V., Corominas-Murtra, B., Valverde, S., and Steels, L. 2010. Language Networks: Their Structure, Function, and Evolution. *Complexity* 15: 20-26.
- Solé, R.V., Saldana, J., Montoya, J.M., and Erwin, D.H. 2010. Simple model of recovery dynamics after mass extinction. *J Theor Biol* 267: 193-200.
- Talamillo, A., Martín, D., Hjerpe, R., Sánchez, J., and Barrio, R. 2010. "SUMO and ubiquitin modifications during steroid hormone synthesis and function". *Biochemical Society Transactions* 38: 54-59.
- Vasconcelos, R., Carranza, S., and Harris, D.J. 2010. Insight into an island radiation: the *Tarentola* geckos of the Cape Verde archipelago. *Journal of Biogeography* 37: 1047-1060.
- Vasconcelos, R., Froufe, E., Brito, J.C., Carranza, S., and Harris, D.J. 2010.
   Phylogeography of the African Common Toad, *Amietophrynus regularis*, based on mitochondrial DNA sequences: inferences regarding the Cape Verde population and biogeographical patterns. *African Zoology* 45: 291-298.
- Verdú-Ricoy, J., Carranza, S., Salvador, A., Busack, S.D., and Díaz, J.A. 2010. Phylogeography of Psammodromus algirus revisited: systematic implications. *Amphibia-Reptilia* 31: 576-582.
- Vila, R., Lukhtanov, V.A., Talavera, G., Gil-T., F., and Pierce, N.E. 2010. How common are dot-like distribution ranges? Taxonomical oversplitting in Western European *Agrodiaetus* (Lepidoptera, Lycaenidae) revealed by chromosomal and molecular markers. *Biological Journal of the Linnean Society* 101: 130-154.
- Viviani, R., Sim, E.J., Lo, H., Beschoner, P., Osterfeld, N., Maier, C., Seeringer, A., Godoy, A.L., Rosa, A., Comas, D., and Kirchheiner, J. 2010 Baseline brain perfusion and the serotonin transporter promoter polymorphism. *Biol Psychiatry* 67(4): 317-22.

- Wendler, F., Gillingham, A.K., Sinka, R., Rosa-Ferreira, C., Gordon, D.E., Franch-Marro, X., Peden, A.A., Vincent, J.P., Munro, S. 2010. A genome-wide RNA interference screen identifies two novel components of the metazoan secretory pathway. *Embo Journal* 29: 304-314.
- Zhadanov, S.I., Dulik, M.C., Markley, M., Jennings, G.W., Gaieski, J.B., Elias, G., Schurr, T.G., and Genographic Consortium Members. 2010. Genetic heritage and native identity of the Seaconke Wampanoag tribe of Massachusetts. *American Journal of Physical Anthropology* 142 (4): 579-589.
- Zidi, A., Amills, M., Tomás, A., Vidal, O., Ramírez, O., Carrizosa, J., Urrutia, B., Serradilla, J.M., and Clop, A. 2010. Short communication: genetic variability in the predicted microRNA target sites of caprine casein genes. *J Dairy Sci* 93: 1749-1753.

#### Books

- Bellés, X. 2010. Bestiari. Monografies Mètode. Universitat de València, 96 pages.
- Bellés, X. 2010. Vivir dos vidas. Un viaje por la metamorfosis de los insectos. Casaciencias, prisma Casa de las Ciencias, *La Voz de Galicia* A Coruña, 112 pages.

## Book Chapters

- Balke, M., Wewalka, G., Alarie, Y., and Ribera, I. 2010. Dytiscidae: The genus Rhantus Dejean (Coleoptera). In: Jäch, M.A., and Balke, M. (eds). Water beetles of New Caledonia, part 1. Zoologisch-Botanische Gesellschaft in Österreich and Wiener Coleopterologenverein, Wien, pp. 129-147.
- Bertranpetit, J. Prólogo. In: Fósiles y moléculas. Aproximaciones a la historia evolutiva de Homo sapiens. Ed. Antonio González-Martín. Memoria de la Real Sociedad de Historia Natural. Segunda Época. Tomo VIII. 169-201. 2010. ISSN: 1132-0869; ISBN: 978-84-936677-5-7.
- Beutel, R.G., Balke, M., and Ribera, I. 2010. 3.1. Aspidytidae Ribera, Beutel, Balke and Vogler, 2002. In: Leschen, R.A.B., Beutel, R.G., and Lawrence, J.F. (eds). Handbook of Zoology, Arthropoda: Insecta. Coleoptera, Beetles. Vol. 2: Morphology and Systematics (Elateroidea, Bostrichiformia, Cucujiformia partim). Walter de Gruyter, Berlin. pp. 21-28.
- Beutel, R.G., Ribera, I., and Balke, M. 2010. 3. Adephaga (Addendum). Introduction and phylogeny. In: Leschen, R.A.B., Beutel, R.G., and Lawrence, J.F. (eds). Handbook of Zoology, Arthropoda: Insecta. Coleoptera, Beetles. Vol. 2: Morphology and Systematics (Elateroidea, Bostrichiformia, Cucujiformia partim). Walter de Gruyter, Berlin. p. 21.
- Casale, A., and Ribera, I. 2010. Are Molopina of the Euro-Mediterranean region related to the Madagascan, South African and Australian Pterostichini? (Coleoptera, Carabidae). Biogeographia (n.s.) 29 (2008): 33-44.
- Corominas-Murtra, B., Valverde, S., and Solé, R.V. 2010. Emergence of Scale-Free Syntax Networks. In: Evolution of Communication and Language in Embodied Agents, pp. 83-101. Springer Verlag Berlín, Heidelberg, Germany.
- Gómez-Zurita, J. 2010. Tribe Timarchini Motschulsky, 1860. In: I. Löbl and A. Smetana (eds.). Catalogue of Palaearctic Coleoptera. Vol. 6. Chrysomeloidea, pp. 437-443. Apollo Books, Stenstrup, Denmark.

- Hernando, C., and Ribera, I. 2010. Limnichidae: Description of a new species from New Caledonia, and checklist of the taxa recorded from the Australian/Pacific Region (Coleoptera). In: M.A. Jäch, and Balke, M. (eds). Water beetles of New Caledonia, part 1. Zoologisch-Botanische Gesellschaft in Österreich and Wiener Coleopterologenverein, Wien, pp. 439-449.
- Lalueza-Fox, C. 2010. ADN i arqueologia. In: Restes de vida, restes de mort; la mort en la prehistòria. Publicacions del Museu de Prehistòria de València, Diputació de València. ISBN: 978-84-7795-557-3.
- Lalueza-Fox, C. 2010. Genómica Neandertal. In: Encuentros con la Ciencia II. Del macrocosmos al microcosmos (ed. Enrique Viguera, Ana Grande y José Lozano). Publicaciones de la Universidad de Málaga. ISBN: 978-84-9747-339-2.
- Lalueza-Fox, C. 2010. El Proyecto Genoma Neandertal. In: Fósiles y moléculas. Aproximaciones a la historia evolutiva de *Homo sapiens* (ed. Antonio González-Martín). Memorias de la Real Sociedad Española de Historia Natural. Segunda época. ISBN: 978-84-936677-5-7.
- Lalueza-Fox, C. 2010. El Proyecto Genoma Neandertal: ¿hacia una definición genética de nuestra especie? In: 150 años después de Darwin: ¿evolución, futuro o crisis?. Edited by Instituto Tomás Pascual Sanz-CENIEH. ISBN: 978-84-7867-057-4.
- Martín, D. 2010. Functions of Nuclear Receptors in Insect Development. In: Nuclear Receptors; Current concepts and future challenges (series: Proteins and Cell Regulation) Bunce, C.M., and Campbell, M.J. (Eds). Springer Publishers (UK). Vol. 8. pp. 31-61.
- Morcillo, C., Albà, M.M., and Navarro, A. 2010. Genoma y Enfermedades Complejas. In: Eclerosis Múltiple. Ed: Pablo Villoslada. Marge Medica Books (Barcelona).
- Solé, R.V. 2010. Catastrophes and Complex Networks. In: Multiscale Cancer Modeling pp. 67-86. Ed: T. Deisboeck and G. StakamatoChapman & Hall-CRG Press. (Virginia)



#### Other Publications

- Bellés, X. 2010. Darwin e a expresión das emocións. Agora do Orcellón 19: 13-16.
- Casale, A., and Ribera, I. 2010. Are Molopina of the Euro-Mediterranean region related to the Madagascan, South African and Australian Pterostichini? (Coleoptera, Carabidae). Biogeographia (n.s.) 29 (2008): 33-44.
- Castresana, J. 2010. Reconstrucción filogenética, diversificación y especiación. SEBBM 165: 20-22.
- Comas, D. 2010. Biodiversidad en poblaciones humanas. SEBBM (Sociedad Española de Bioquímica y Biología Molecular), 165: 24-28.
- Comas, D. 2010. Evolució i diversitat del genoma humà. l'Atzavara 19: 61-68.
- 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.
- Dinca, V., .Cuvelier, S., Zakharov, E.V., Hebert, P D.N., and Vila, R. 2010. Biogeography, ecology and conservation of Erebia oeme (Hübner) in the Carpathians (Lepidoptera: Nymphalidae: Satyrinae). Annales de la Societe Entomologique de France 46: 486-498.
- Escoriza, D., Metallinou, M., Donaire-Barroso, D., Amat, F., and Carranza, S. 2010. Biogeography of the White-Bellied Carpet Viper *Echis leucogaster* Roman, 1972 in Morocco, a study combining mitochondrial DNA data and ecological niche modeling. *Butlletí de la Societat Catalana d'Herpetologia* 18: 55-68.
- Fortuny, J., and Corominas-Murtra, B. 2010. Some formal considerations on the generation of hierarchically structured expressions. *Catalan Journal of Linguistics* 8: 99-111.
- Gómez-Zurita, J., and Petitpierre, E. 2010. Contribution to the knowledge of the Iberian fauna of Chrysomelidae (Coleoptera). I. New records of Criocerinae, Clytrinae and Cryptocephalinae. *Boletín de la Sociedad Entomológica Aragonesa* 47: 139-142.
- Horta-Bernús, R., Rosas-Casals, M., and Valverde, S. 2010. Discerning Electricity Consumption Patterns from Urban Allometric Scaling. 2010 Complexity in Engineering: Compeng 2010, Proceedings 49-51.
- Marquès-Bonet, T., and Comas, D. 2010. Com es pot mesurar la variabilitat humana? Omnis Cellula 25: 8-9
- Rellegint el missatge genètic: gens, genomes, diversitat i comprensió de la vida i dels humans. Memorias de la Real Academia de Ciencias y Artes de Barcelona. Núm. 1025, Vol. LXIV, NÚM. 8. Barcelona, 2010.
- Romano, A., Amat, F., Rivera, X., Sotgiu, G., and Carranza, S. 2010. Evidence of tail autotomy in the European plethodontid Hydromantes (Atylodes) genei (Temmick and Schlegel, 1838) (Amphibia: Urodela: Plethodontidae). *Acta Herpetologica* 5: 199-205.
- Santamaría, D., Fortea, J., de la Rasilla, M., Martínez, L., Martínez, E., Cañaveras, J.C., Sánchez-Moral, S., Rosas, A., Estalrrich, A., García-Tabernero, A., and Lalueza-Fox, C.
   2010. The technological and typological behaviour of a Neanderthal group from El Sidrón cave (Asturias, Spain). The Technological and typological behaviour of a Neanderthal group from El Sidrón cave (Asturias, Spain). Oxford Journal of Archaeology 29 (2): 119-148.



# IBE HIGHLIGTED

## GENOMES FROM EXTINCT HOMININS CHALLENGE CURRENT THEORIES ON MODERN HUMAN ORIGINS

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 posible to determine that 78 genes have been recently selected in our lineage. Those genes include some related to physiological, metabolic and cognitive aspects, but nevertheles 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



Fig.1: Vindija bones Image: Max Planck Institute

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.

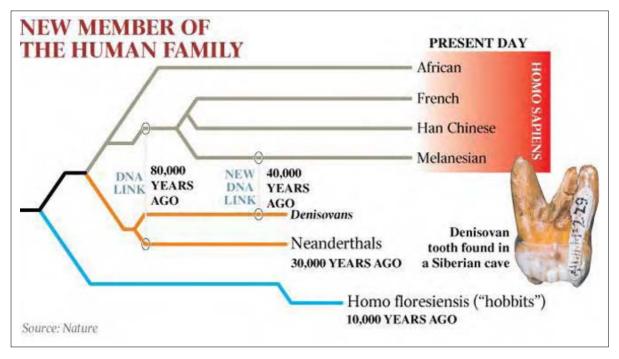


Fig. 2: Modern human origins and the interactions between Homo sapiens and extinct hominins (Neandertals and Denisovans), as suggested from new paleogenomic data.

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

- Green, R.E., Krause, J., Briggs, A.W., Maricic, T., Stenzel, U., Kircher, M., Patterson, N., Li, H., Zhai, W., Fritz, M.H.Y., Hansen, N.F., Durand, E.Y., Malaspinas, A.S., Jensen, J.D., Marquès-Bonet, T., Alkan, C., Prüfer, K., Meyer, M., Burbano, H.A., Good, J.M., Schult, R., Aximu-Petri, A., Butthof, A., Höber, B., Höffner, B., Siegemenud, M., Weihmann, A., Nusbaum, C., Lander, E.S., Russ, C., Novod, N., Affourtit, J., Egholm, M., Verna, C., Rudan, P., Brajkovic, D., Kucan, Z., Gusic, I., Doronichev, V.B., Golovanova, L.V., Lalueza-Fox, C., De la Rasilla, M., Fortea, J., Rosas, A., Schmitz, R.W., Johnson, P., Eichler, E., Falush, D., Birney, E., Mullikin, J.C., Slatkin, M., Nielsen, R., Kelso, J., Lachmann, M., Reich, D., Pääbo, S. 2010. A draft sequence of the Neandertal genome. *Science* 328: 710-722.
- Reich, D., Green, R.E., Kircher, M., Krause, J., Patterson, N., Durand, E.Y., Viola, B., Briggs, A.W., Stenzel, U., Johnson, P.L.F., Maricic, T., Good, J.M., Marquès-Bonet, T., Alkan, C., Fu, Q., Mallick, S., Li, H., Meyer, M., Eichler, E.E., Stoneking, M., Richards, M., Talamo, S., Shunkov, M.V., Derevianko, A.P., Hublin, J.J., Kelso, J., Slatkin, M., Pääbo, S. 2010. Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature* 468: 1053-1060.

# 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 carstic 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

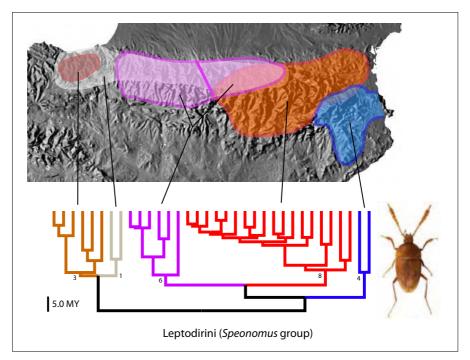


Fig. 1: Phylogeny of the Pyrenean clade of subterranean beetles of tribe Leptodirini. The main lineages are as old as the chain itself, and did colonize it from the edges to the center.

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

• Ribera, I., Fresneda, J., Bucur, R., Izquierdo, A., Vogler, A.P., Salgado, J.M., and Cieslak, A. 2010. Ancient origin of a Western Mediterranean radiation of subterranean beetles. *BMC Evolutionary Biology* 10: 29.

The Article has been Featured in

• Juan, C., and Emerson, B. 2010. Minireview. Evolution underground: shedding light on the diversification of subterranean insects. *Journal of Biology* 9: 17.

#### 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.

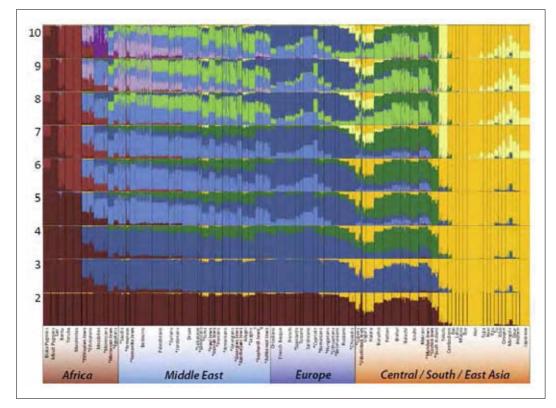


Fig. 1: Population structure plot (ADMIXTURE analysis) based on more than 225,000 SNPs in human worldwide populations including Jewish communities.

#### Reference Article

 Behar, D.M., Yunusbayev, B., Metspalu, M., Metspalu, E., Rosset, S., Parik, J., Rootsi, S., Chaubey, G., Kutuev, I., Yudkovsky, G., Khusnutdinova, E.K., Balanovsky, O., Semino, O., Pereira, L., Comas, D., Gurwitz, D., Bonne-Tamir, B., Parfitt, T., Hammer, M.F., Skorecki, K., Villems, R. 2010. The genome-wide structure of the Jewish people. *Nature* 466: 238-242.

### 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

 Antonacci, F., Kidd, J.M., Marquès-Bonet, T., Teague, B., Ventura, M., Girirajan, S., Alkan, C., Campbell, C.D., Vives, L., Malig, M., Rosenfeld, J.A., Ballif, B.C., Shaffer, L.G., Graves, T.A., Wilson, R.K., Schwartz, D.C., Eichler, E.E. 2010. "A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. *Nature Genetics* 42: 745-U729.

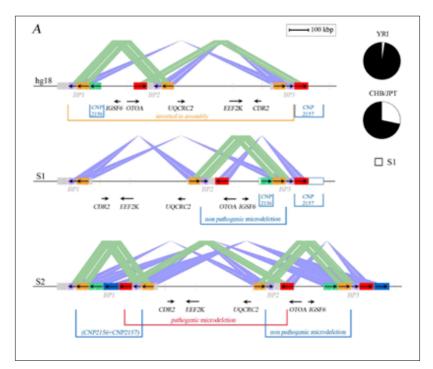


Fig. 1: Schematic representation of the 16p12 region. The top track represents the human genome (hg18). The following 2 tracks represents the 2 major haplotypes found in humans. Only the S2 haplotype predisposes to the pathogenic microdeletion. Color boxes denotes the location of segmental duplications and green and blue lines represents the orientation among block (direct and reverse, respectively).

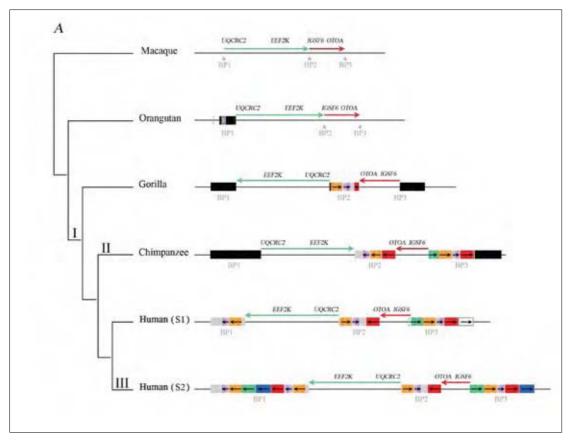
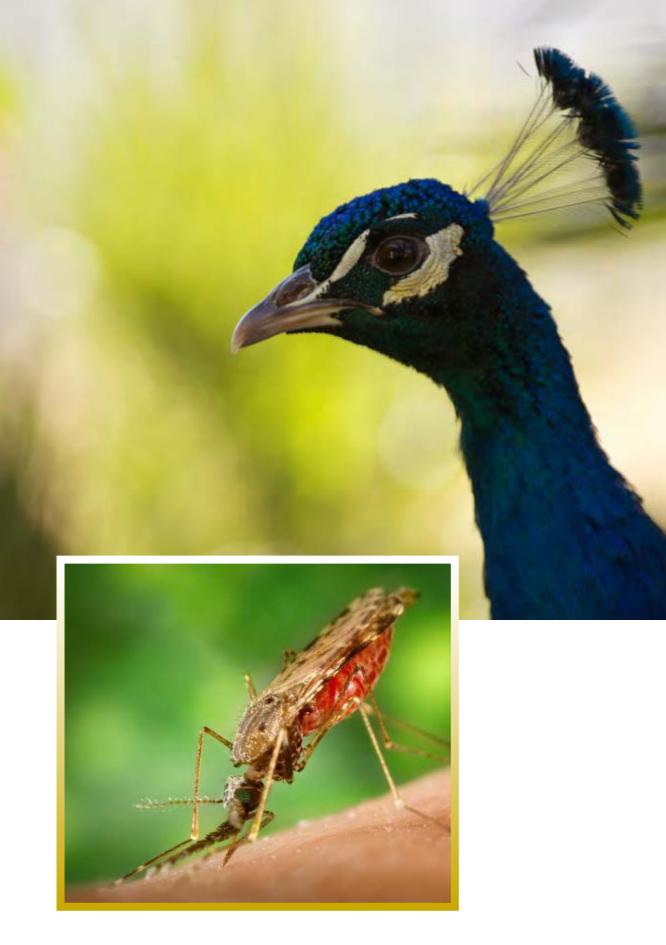


Fig. 2: (A) 16p12.1 genomic organization in macaque, orangutan, gorilla, chimpanzee and human. Green and red arrows indicate the orientation of the regions between breakpoint (BP1-BP2 and BP2-BP3). Black and colored rectangles show segmental duplications. Sequence and FISH data indicate that the inverted 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 be 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).





# DOCTORAL THESES PRESENTED DURING 2010

PhD Student: Title: Thesis Director: Institution and Date:	Paula Irles Bases moleculares de la oogénesis en la cucaracha <i>Blattella germanica</i> (L.) (Dictyoptera, Blattellidae) Maria-Dolors Piulachs Universitat de Barcelona, 15th February 2010	
=	Martin Sikora Evolutionary genetics of malaria: genetic susceptibility and natural selection	
Thesis Directors: Institution and Date:		
PhD Student: Title:	Karla Sandoval Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica.	
Title: Thesis Director:	Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica. David Comas	
Title: Thesis Director:	Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica.	
Title: Thesis Director: Institution and Date: PhD Student:	Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica. David Comas Universitat Pompeu Fabra, 19th June 2010 Gemma Berniell	
Thesis Director: Institution and Date: PhD Student: Title:	Ethnicity, linguistics, and genetic diversity in native mexicans: reconstructing the population history of Mesoamerica. David Comas Universitat Pompeu Fabra, 19th June 2010	

# DISSEMINATION OF SCIENCE: BIODIVERSITY YEAR

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

# IBE SEMINARS 2010

Speaker	Talk	Institution	Date
Luc Steels	The origins of human language. Genes, memes or cognition?	University of Brussels (VUB AI lab) and Sony Computer Science Laboratory (Paris)	04/02/2010
David Vieites	Research prospects for historical biogeography and evolutionary biology in Madagascar	Museo Nacional de Ciencias Naturales (CSIC) (Madrid)	22/02/2010
Alfred Vogler	DNA taxonomy and the study of biodiversity patterns	Department of Biology, Imperial College (London) and The Natural History Museum (London)	26/04/2010
Stuart Reynolds	Partners in crime: Evolution of a pathogenic symbiosis between Photorhabdus bacteria and heterorhabditid nematodes, and how the insect host can fight back	Department of Biology and Biochemistry, University of Bath (Bath)	31/05/2010
Alex Kacelnik	A biologist's reflections on rationality	Behavioral Ecology Research Group, University of Oxford (Oxford)	14/06/2010

Speaker	Talk	Institution	Date
Frederic Bartumeus	Anomalous diffusion and stochastic search strategies in animal movement: an evolutionary perspective	Centre d'Estudis Avancats de Blanes, CSIC (Blanes)	16/06/2010
Marcos Pérez-Losada	Phylogeny, conservation and speciation of Aegla (Anomura: Decapoda: Aeglidae) freshwater crabs from Patagonia	CIBIO, Centro de Investigação em Biodiversidade e Recursos Genéticos Universidade do Porto (Porto)	28/06/2010
Francisco Úbeda	Solving the recombination hotspot paradox	Department of Ecology and Evolutionary Biology University of Tennessee (Knoxville Tennessee)	12/07/2010
Mohamed Noor	Recombination, speciation, and nucleotide diversity in the Drosophila pseudoobscura species group	Biology Department, Duke University (Durham NC)	13/09/2010
Teresa Romero	Resolución de conflictos, consolación y empatía en primates	University of Tokyo (Tokyo)	22/09/2010
Jordi Bascompte	Species mutualistic networks: the architecture of biodiversity	Integrative Ecology Group, Estación Biológica de Doñana, CSIC (Sevilla)	18/10/2010
Hernán Burnano	Targeted sequencing of Neandertal DNA: insights into recent coding and non-coding evolution on the human lineage	Department of Evolutionary Genetics, Max Planck Institute of Evolutionary Anthropology (Leipzig)	19/10/2010
José M. Eirin López	Histones in regalia: flourishing diversity on the verge of germ chromatin evolution	Universidade da Coruña (A Coruña)	25/10/2010
David de Lorenzo	Senses and Neuromarketing: Darwin goes shopping	Facultat de Medicina, Universitat de Lleida (Lleida)	05/11/2010
Patrick Fitze	Determinants of the intensity of sexual selection and its implications for conservation	Museo Nacional de Ciencias Naturales, CSIC (Madrid)	29/11/2010

# TEACHING

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

# **GRADUATE STUDIES**

Bachelor's degree in 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

A 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.

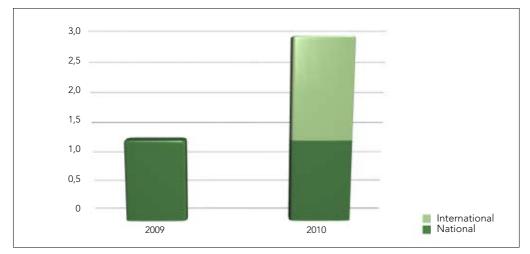
Subject	In: Master	University	Teacher
"Genetic variation dynamics in populations" In: Genomes and Systems	Master in Biomedical Research (BIOMED)	Pompeu Fabra University (UPF)	Elena Bosch
"Statistics in forensic genetics" In: Grounds in Forensic Anthropology	Master in Human Biology	(UB / UAB)	Francesc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

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

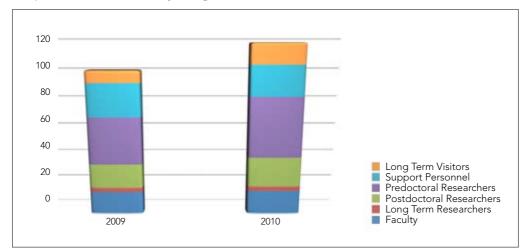
# IBE IN NUMBERS

# 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).



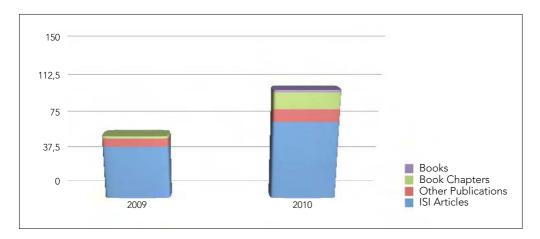
# PEOPLE



IBE personnel distributed by categories.

# PUBLICATIONS

IBE publications distributed by categories.



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

