

Annual Report 2012



INSTITUT de
BIOLOGIA
EVOLUTIVA

ANNUAL REPORT 2012



INSTITUT de
BIOLOGIA
EVOLUTIVA

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- **Scientific coordinator and layout:** Carles Lalueza-Fox
- **Technical coordinator and layout:** Anna Pérez-Lezaun
- **Text and photography:** IBE Researchers
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FOREWORD

by Xavier Bellés, Director of the IBE



We all go through tough times in our careers and our research, as well as those times when we can celebrate our successes. If I had to give one piece of advice, I would say keep going through the difficult times. Do not give up.

Julia Goodfellow, Vice-Chancellor of the University of Kent

The year 2012 has been the year of the evaluation of the IBE by the External Scientific Committee (CCE). After its foundation in July 2008, our Institute has been functioning with our original criteria concerning aspects of paramount importance, such as scientific structure or governance. Thus, the evaluation by the CCE became necessary to get qualified and independent opinions covering all aspects –general or particular– of our Institute. We must thank the efficiency of the CCE members, who were hectically working during their visit in March 2012, and then, from home, produced a synthetic but clear-cut analysis of our reality, and suggested a number of constructive ideas to improve our Institute’s project. The implementation of some of these suggestions, such as having our own building as soon as we could, was beyond our possibilities, but applying many other ideas was in our hands. As a result, we have sensibly modified the scientific organization, including changes in aims, composition and names of the groups and programs. As the reader will see in the following pages, the changes have been already adopted in the general organization chart and in the description of the programs and groups. In my view, one of the most exciting suggestions of the CCE was the convenience of organizing a course on basic concepts of evolutionary biology addressed to the predoctoral students newly arrived to the IBE. This would benefit not only the formation of the youngest members of our Institute, but also our social cohesion. The idea was enthusiastically received and the course was immediately organized. Thus, the first edition will be held by the beginning of next year.

Despite being immersed in a perfect storm of financial crisis, the current life of the Institute has not been significantly affected (yet). Income from grants and projects has decreased somewhat, but publications have notably increased, in quantitative, but also qualitative terms. But this output may be an inertial consequence of recent past times, which were financially better, and inertia ends up with immobility soon or later if no more energetic inputs are added, as Galileo showed in the early 1500s.

Unfortunately, our policy-makers are affected by an acute myopia, which is leading to dramatic mistakes of perspective when deciding what to do with our science. It is hard to believe that they do not apparently understand such obvious things as that sustained scientific investment is required, for example, to produce medical breakthroughs, to obtain technological innovation and to solve pressing environmental issues. The role of science, spurring new ventures based on scientific discoveries and creating new jobs, is of paramount importance in economic recovery. Reduced job opportunities due to hiring freezes and downsizing in academia and the industry will lead to the best senior scientists to emigrate to more welcoming scientific climates. Perhaps worse, those young researchers who have just finished a long and arduous training and are not able to get funding will also migrate or leave research. Indeed, «solutions» involving draconian cuttings and freezing can lead to lose an entire generation of good researchers, and once lost, it can take more than ten years to build up that experience again. As a recent editorial of *Nature Cell Biology* told us, in times of economic crisis it is important to remember the old adage: research is a long-distance race, not a sprint. Investing in research is investing in the future, and requires a long-term commitment to the accumulation of knowledge, the testing of basic principles, and the translation of these discoveries into practical applications that impact everyday life. Safeguarding the future of scientific development will require bold decisions and long-term vision by policy-makers.

What can we do, individually and as a relatively small scientific collective? We should increase our creativity, not only to get financial support, but also to adapt our scientific focus and approaches to new situations, to resist the temptation to become more conservative in science, as this might be one of the worse consequences of the austerity context. Briefly, we should continue doing good science, despite the unfortunate context. We cannot give up, among other things, because nobody in further generations would forgive us.

INTRODUCTION TO THE IBE



SCOPE AND GENERAL GOALS

The Institute of Evolutionary Biology (IBE) was formally founded in July 2008, as a joint Institute of the Spanish National Research Council (CSIC) and the Pompeu Fabra University (UPF). The Institute functional structure was not fully operative until mid 2009 when the “Management Unit” was complete and ongoing. Initially, the IBE was created with 11 independent research groups from the Molecular Biology Department (CID, CSIC) and 6 independent research groups from the Evolutionary Biology Unit (dCEXS, UPF). Nowadays, IBE activity involves more than a hundred people and 18 independent research groups distributed in five scientific programs related to Evolutionary Biology research.

One of the great challenges of the 21st century, after the publication of the Human Genome Sequence and many other species, is the study of biodiversity, either within species (variation, polymorphism) and/or between species (divergence), as a crucial element to understand the essential mechanisms of life. In this context, evolutionary biology provides the key tools and concepts and the main IBE mission is to promote knowledge and research excellence in this field of knowledge. The basis of the IBE, and its main peculiarity, is the ability to address biodiversity studies describing functional and evolutionary genomics at all levels of observation: molecular, biochemical, physiological and morphological.

Briefly, the IBE project vision, defined as the projection of future long-term of the Institute, is to be a centre of international reference in the study of biodiversity, in the broadest sense, and its evolution, from a molecular and genomic perspective and including human diversity.

GENERAL STRUCTURE

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

BOARD OF TRUSTEES

IBE main managing structure is the “Board of Trustees” composed by two representatives of both partner Institutions (CSIC and UPF), which has the highest responsibilities in all aspects of IBE strategy and functioning, comprising the direction, composition, research lines, structure and functioning rules of IBE.

Members of Board of Trustees along 2012:

Luis Calvo

| CSIC Institutional Coordinator in Catalonia

José García Montalvo

| UPF Vicechancellor of Scientific Policy

Francesc Posas

| CEXS-UPF Department Director

José Ramón Urquijo Goitia

| CSIC Vicepresident of Institutional Relationships and Organisation

EXTERNAL SCIENTIFIC COMMITTEE (CCE)

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

The External Scientific Committee was approved by the Board of Trustees in 2011, and its composition is as follows:

Chairman



Andrés Moya |
Universitat de València,
València, Spain

Members



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



Stuart Reynolds |
University of Bath, Bath, UK



Brian Charlesworth |
University of Edinburgh,
Edinburg, UK



Luis Serrano |
Centre de Regulació
Genòmica, Barcelona, Spain



Gonzalo Giribet |
Harvard University, Cambridge,
MA, USA



Eske Willerslev |
University of Copenhagen,
Copenhagen, Denmark

The first meeting and evaluation of the CCE was held in the IBE on the 1st and 2nd March 2012 with the participation of all members, except Carlos Bustamante, who did not attend due to last minute academic compromises at Stanford University. Evaluation covered all levels of IBE activities.

EXECUTIVE BOARD

The IBE Executive Board is composed by 7 members:

IBE Director

- | Xavier Bellés

IBE Vicedirector

- | Arcadi Navarro (acting also as the Coordinator of the "*Comparative and computational genetics*" program)

Current Members

- | José Castresana (acting as the Coordinator of the "*Animal biodiversity and evolution*" program)
- | David Comas (acting as the Coordinator of the "*Population genetics*" Program)
- | Maria-Dolors Piulachs (acting as the Coordinator of the "*Functional genomics and evolution*" program)
- | Ricard Solé (acting as the Coordinator of the "*Complex systems*" program)

General Manager and Board Secretary

- | Anna Pérez-Lezaun

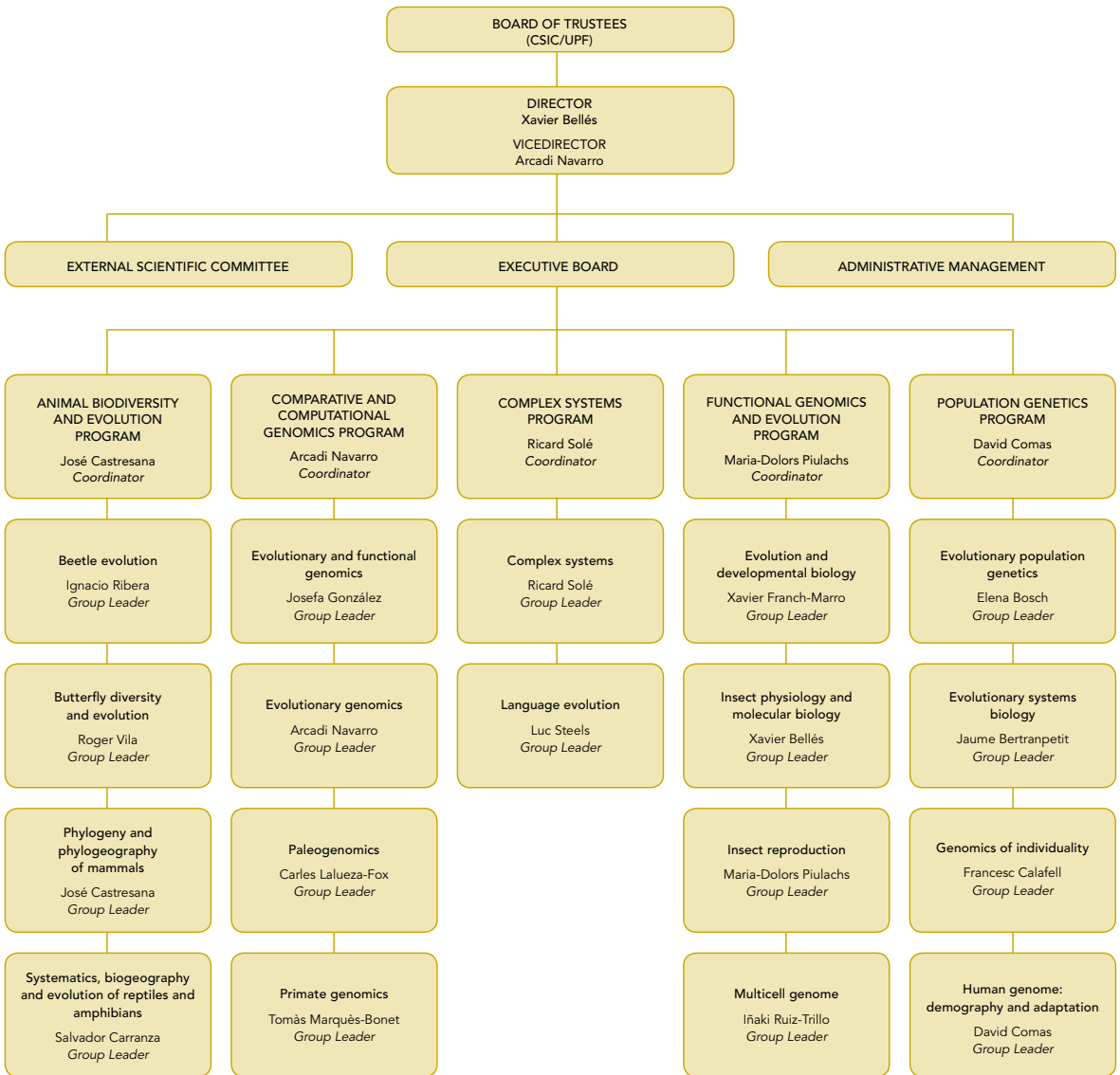
SCIENTIFIC STRUCTURE

As a result of the recommendations emerged from evaluation by the CCE, the structure of scientific groups was modified from an initial number of 23 to the present 18. The name, composition and scope of some Scientific Programs was also modified.

The new scientific structure distributes the present 18 groups into the following Programs:

- | Animal biodiversity and evolution
- | Comparative and computational genomics
- | Complex systems
- | Functional genomics and evolution
- | Population genetics

IBE ORGANISATION CHART



SERVICE UNITS

In support of the IBE scientific structure three service units have been planned: One "Management Unit" already functional, and two technical Units still to be fully completed: "Bioinformatics Unit", and "Experimental Techniques Unit".

MANAGEMENT UNIT

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



MANAGEMENT UNIT

General Manager

Anna Pérez-Lezaun | UPF

ViceManager and Accountant

Rita Arias | CSIC

Administrative Support

Emiliano González | CSIC

Blanca Álvarez | CSIC

Judit Sainz | UPF

EXPERIMENTAL TECHNIQUES

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

Members:

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

BIOINFORMATICS UNIT

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

Members:

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

PROGRAM RESEARCH ASSISTANTS

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

Members:

- | Eva Ramallo | UPF Contract • Supporting Complex systems and Population genetics program
- | Mònica Vallés | Technical Staff (UPF) • Supporting Population genetics Program and Comparative and computational genetics program
- | Maria Niño | JAETEC (CSIC) • Supporting Comparative and computational genetics program



Laura Gutiérrez

Mónica Vallés

Maria Niño

José María Heredia

Cristina Olivella

PROGRAMS RESEARCH ASSISTANTS

Experimental Techniques

Cristina Olivella | Technical Staff (CSIC)

Bioinformatics Unit

José María Heredia | CSIC Contract (PTA-MICINN)

Program Research Assistants

Eva Ramallo | UPF Contract · Supporting Complex systems and Population genetics Program

Mónica Vallés | Technical Staff UPF · Supporting Population genetics Program and Comparative and computational genetics Program

Laura Gutiérrez | JAETEC CSIC · Supporting UPF Genomics Core facility

PERSONNEL

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

Table 1. IBE personnel distribution by categories. December 2012.

	2012	2011	2010	2009
Faculty	18	18	17	16
Long-term Researchers* (ICREA, Marie Curie and Ramon y Cajal programs)	5	5	3	3
Postdoctoral Researchers	23	25	21	17
Predocctoral Researchers	50	43	42	33
Support Personnel				
Laboratory Technicians	11	8	9	4
Bioinformaticians	9	6	5	4
Administrative Staff	5	5	5	5
Others (project support)	0	9	2	2
Long-term Visitors (> 1 month)	19	11	14	8
TOTAL	140	130	118	92

*Marie Curie, Ramón y Cajal or ICREA researchers

LOCALISATION

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

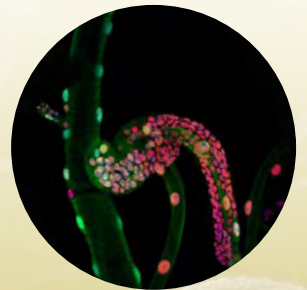
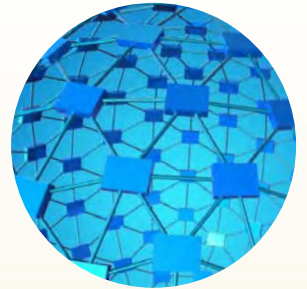
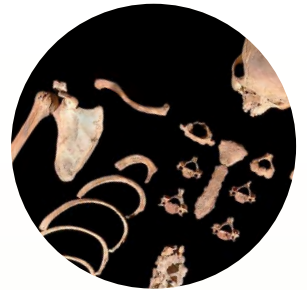
IBE at the CMIMA building:
Passeig Marítim de la Barceloneta, 37-49.
08003 Barcelona, Spain.

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





IBE RESEARCH PROGRAMS







PROGRAM

animal biodiversity and evolution

RESEARCH GROUPS

Beetle evolution

Ignacio Ribera, Group Leader

Subgroups:

Water and cave beetle evolution

| *Ignacio Ribera, PI*

Herbivore beetle evolution

| *Jesús Gómez-Zurita, PI*

Butterfly diversity and evolution

Roger Vila, Group Leader

Phylogeny and phylogeography of mammals

José Castresana, Group Leader

Systematics, biogeography and evolution of reptiles and amphibians

Salvador Carranza, Group Leader

Members of this research program carry out research on animal biodiversity from a phylogenetic perspective with the aim of gaining further insights 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, genomics, proteomics, bioinformatics, morphometry 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. The use of genomic data and large-scale phylogenetic analyses (both in terms of species considered and sequenced data) is helping to obtain more robust phylogenies and evolutionary conclusions. Phylogenetic trees are a common framework for many evolutionary studies and, therefore, this research program provides many points of contact with other programs at the IBE.

GROUP
BEETLE EVOLUTION



Valeria Rizzo



Enrico Ruzzier



David García Vázquez

David Sánchez-Fernández

André Silva Fernandes

Ignacio Ribera

Andrey Rudoy

Rocío Alonso Rodríguez



Nguyen Thi Dinh



Anna Papadopoulou

Gissela De la Cadena

Anabela Cardoso

Jesús Gómez-Zurita

Tinguaro Montelongo



Amparo Hidalgo

GROUP MEMBERS

Ignacio Ribera, Group Leader

| Research Scientist, CSIC



| Subgroup: Water and cave beetle evolution

Ignacio Ribera | Research Scientist, CSIC

David Sánchez-Fernández | Post-doc, Juan de la Cierva Program

David García Vázquez | PhD Student, FPI Scholarship, MICINN

Amparo Hidalgo Galiana | PhD Student, FPI Scholarship, MICINN

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

Andrey Rudoy | PhD Student, JAE Scholarship, CSIC

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

Enrico Ruzzier | MSc Student, Università degli Studi di Padova

Rocío Alonso Rodríguez | Laboratory Technician, Contract Associated to Project

| Subgroup: Herbivore beetle evolution

Jesús Gómez-Zurita | Tenured Scientist, CSIC

Anna Papadopoulou | Post-doc, Juan de la Cierva Program

Anabela Cardoso | Lab Manager and PhD Student, MICINN Contract

Gissela De la Cadena | PhD Student, MAEC-AECID Scholarship

Tinguaro Montelongo | PhD Student, MICINN Scholarship

Nguyen Thi Dinh | PhD Student, CSIC Scholarship (International Cooperation)

Helena Vizán | PhD Student, MICINN Scholarship

RESEARCH OUTLINE

We study different evolutionary processes using beetles. One sub-group (led by IR) is centered around the macroevolutionary consequences of range size changes and the ecomorphological and phylogenetic diversification of species-rich lineages of cave and water beetles. The other (led by JGZ) focuses on the diversity and evolution of plant-insect interactions, the origin and evolutionary particularities of unisexual (and hybrid) lineages and conservation of tropical faunas.

RESEARCH LINES

SUBGROUP: WATER AND CAVE BEETLE EVOLUTION

1. Thermal tolerance and pleistocene range expansions
2. Origin and diversification of the cave beetles of the genus *Troglocharinus*
3. Evolution of the complex male genitalia in hydraenidae
4. Riffle beetles of the amazonian region
5. Evolution of the sensory organs in the antennae of leptodirini cave beetles

SUBGROUP: HERBIVORE BEETLE EVOLUTION

1. Evolutionary consequences of unisexuality: evolution of gender-specific genes
2. Biodiversity, structure and conservation of tropical dry forests
3. Diversity and diversification of new caledonian eumolpinae
4. Systematics of chrysomelidae



Fig. 1: Many species of insects have what seems unnecessary complex male genitalia, usually though to be the result of either sexual selection or species recognition mechanisms. We investigate the evolution of this complexity in a group of aquatic beetles. In the photo, an axial slice of the penis of *Limnebius truncatellus*, with the quitinised tissue in dark blue.

Image: Andrey Rudoy.

ISI Articles

- Andújar, C., Gómez-Zurita, J., Rasplus, J.Y., and Serrano, J. 2012. Molecular systematics and evolution of the subgenus *Mesocarabus* Thomson 1875 (Coleoptera: Carabidae: *Carabus*), based on mitochondrial and nuclear DNA. *Zoologic Journal of the Linnean Society* 166: 787-804.
- Andújar, C., Serrano, J., and Gómez-Zurita, J. 2012. Winding up the molecular clock in the genus *Carabus* (Coleoptera: Carabidae) — Effects of methodological decisions on rate and node age estimation. *BMC Evolutionary Biology* 12: 40.
- Arribas, P., Velasco, J., Abellán, P., Sánchez-Fernández, D., Andújar, C., Calosi, P., Millán, A., Ribera, I., and Bilton, D.T. 2012. Dispersal ability rather than ecological tolerance drives differences in range size between lentic and lotic water beetles (Coleoptera: Hydrophilidae). *Journal of Biogeography* 39: 984-994.
- Bergsten, J., Bilton, D.T., Fujisawa, T., Elliott, M., Monaghan, M.T., Balke, M., Hendrich, L., Geijer, J., Herrmann, J., Foster, G.N., Ribera, I., Nilsson, A.N., Barraclough, T.G., and Vogler, A.P. 2012. The effect of geographical scale of sampling on DNA barcoding. *Systematic Biology* 61: 851-869.
- Fernandes, A.S., and Hamada, N. 2012. Description and notes on the bionomics of a new species of *Potamophilops* Grouvelle, 1896 (Coleoptera: Elmidae: Larainae), from the Cerrado biome in Brazil. *Journal of Natural History* 46: 717-727.
- Ge, D.Y., Gómez-Zurita, J., Chesters, D., Yang, X.K., and Vogler, A.P. 2012. Suprageneric systematics of flea beetles (Chrysomelidae: Alticinae) inferred from multilocus sequence data. *Molecular Phylogenetics and Evolution* 62: 793-805.
- Gebiola, M., Gómez-Zurita, J., Monti, M.M., Navone, P., and Bernardo, U. 2012. Integration of molecular, ecological, morphological and endosymbionts data for species delimitation within the *Pnigalio soemius* complex (Hymenoptera: Eulophidae). *Molecular Ecology* 21: 1190-1208.
- Gómez-Zurita, J., Sassi, D., Cardoso, A., and Balke, M. 2012. Evolution of *Cryptocephalus* leaf beetles related to *C. sericeus* (Coleoptera: Chrysomelidae) and the role of hybridization in generating species mtDNA paraphyly. *Zoologica Scripta* 41: 47-67.
- Hernando, C., Castro, A., Aguilera, P., and Ribera, I. 2012. A new interstitial species of the *Hydroporus ferrugineus* group from north-western Turkey, with a molecular phylogeny of the *H. memnonius* and related groups (Coleoptera: Dytiscidae: Hydroporinae). *Zootaxa* 3173: 37-53.
- Przewozny, M., and Fernandes, A.S. 2012. *Portelmis guianensis* sp.nov. from French Guiana (Coleoptera: Elmidae). *Zootaxa* 3196: 58-63.
- Sánchez-Fernández, D., Lobo, J.M., Millán, A., and Ribera, I. 2012. Habitat persistence mediates time to equilibrium in the geographical distribution of Iberian diving beetles. *Global Ecology and Biogeography* 21: 988-997.



Fig. 2: Molecular methods can be used to match adults and larvae of insects with a complete metamorphosis, such as the species of riffle beetle pictured here (*Phanoceroideus aquaticus*), from the Amazonian basin. Image: Andre Silva Fernandes.

- Sánchez-Fernández, D., Aragón, P., Bilton, D.T., and Lobo, J.M. 2012. Assessing the congruence of thermal niche estimations derived from distribution and physiological data. A test using diving beetles. *PLOS ONE* 7(10): e48163.

Books/Book Chapters

- Millán, A., Abellán, P., Sánchez-Fernández, D., Picazo, F., Velasco, J., Lobo, J.M., and Ribera, I. 2012. Efectividad de la red de Parques Nacionales peninsulares en la conservación de la biodiversidad acuática. In: Ramírez, L., and Asensio, B. (eds.) *Proyectos de investigación en Parques Nacionales: 2008-2011*. Organismo Autónomo de Parques Nacionales, Madrid, pp 151-181.
- Ribera, I., and Beutel, R.G. 2012. Coleópteros. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 312-321.
- Ribera, I., and Beutel, R.G. 2012. Hexápodos. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 280-291.

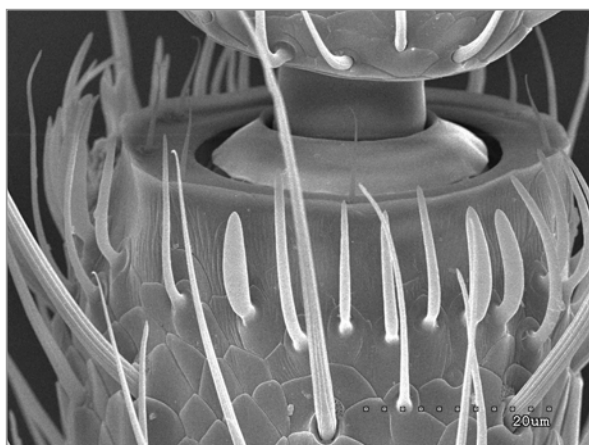


Fig. 3: The extreme modifications of the species living in the subterranean environment have always fascinated evolutionary biologists, and in particular those referring to the sensory organs. We investigate the evolution of the antennal sensory receptors in a group of Mediterranean subterranean beetles, such as the Turkish *Karadeniziella omodei*, pictured here.

Image: Enrico Ruzzier.

Other Publications

- Ribera, I., Hernando, C., and Aguilera, P. 2012. Coleópteros acuáticos del humedal de Laku (Araba). *Estudios del Museo de Ciencias Naturales de Álava* 23 (2009-2010): 215-222.
- Salgado, J.M., Luque, C.G., Labrada, L., Fresneda, J., and Ribera, I. 2012. Revisión del género *Cantabrogeus* Salgado, 2000, con la descripción de tres nuevas especies hipogeas endémicas de la Cordillera Cantábrica (Coleoptera, Leiodidae, Cholevinae, Leptodirini). *Animal Biodiversity and Conservation* 35: 27-50.

FUNDED PROJECTS

Project Title: Análisis a escala genómica de las consecuencias evolutivas del abandono del sexo: Explorando el destino de la función masculina

Financed by: Ministerio de Ciencia e Innovación

Years: 2012-2014

PI: Jesús Gómez-Zurita

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

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: Ignacio Ribera

Fig. 4: Islands are favourite places for evolutionary biologists. In our lab we study the origin of island faunas in both the Macaronesia and the Mediterranean. In the photo, Lagoa do Negro, in Terceira (Azores), home to several endemic species of water beetles.

Image: Alexandra Cieslak.



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 BBVA

Years: 2009-2012

PI: Jesús Gómez-Zurita

Project Title: Sentando las bases para la creación del Banco de Germoplasma del Bosque Seco Tropical Americano, el BGBST «El Sálamo»

Financed by: Programa de Cooperación Interuniversitaria e Investigación Científica (Ministry of Foreign Affairs and Cooperation)

Years: 2012

PI: Jesús Gómez-Zurita

Project Title: Grup de Recerca Consolidat en Sistemàtica i Evolució Zoològica – ZOOSYSEVO

Financed by: Generalitat de Catalunya

Years: 2009-2013

PI: Salvador Carranza

Project Title: Atlas y Libro Rojo de los Coleópteros acuáticos de España

Financed by: Ministerio de Medio Ambiente

Years: 2010-2012

PI: Andrés Millán (Universidad de Murcia)

GROUP

BUTTERFLY DIVERSITY AND EVOLUTION



Roger Vila

Claudia Sañudo

Vlad Eugen Dinca

Gerard Talavera

Raluca Voda

Marga Marín

GROUP MEMBERS

Roger Vila, Group Leader

| Research Scientist, CSIC



Claudia Sañudo, PhD Student | FBBVA Project Scholarship

Gerard Talavera, PhD Student | FPI Scholarship, MEC

Raluca Voda, PhD Student | FPU Scholarship, MEC

Martha Erazo | MSc Student, Master UAB

Mònica Navarro | Undergraduate Student, Practicum UB

Marga Marín, Laboratory Technician

RESEARCH OUTLINE

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



Fig. 1: A spectacular congregation of *Lysandra bellargus* males on salt-rich moist soil. *Lysandra* display high karyotype instability and constitute a great system to study chromosomal evolution.

Image: Vlad Dinca.



Fig. 2: The hesperiids in general, and the genus *Spialia* in particular, are the focus for our efforts to discover cryptic diversity and reconstruct species' biogeographical history. Image: Vlad Dinca.

RESEARCH LINES

1. Characterization of butterfly diversity with DNA barcoding

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

2. Uncovering cryptic butterfly biodiversity in Europe

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

3. Ecological factors determining butterfly biogeography

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

4. Chromosomal evolution in *Polyommatus* and *Leptidea*

Some butterfly groups have remarkably unstable chromosomes and display unusual patterns in their karyotypes. They constitute an ideal group to study



Fig. 3: Butterflies are beautiful: a simple but powerful reason to love studying them. In the picture, an *Aglais io* in Montseny Mt. Image: Roger Vila.

chromosomal evolution in action. We are focusing our studies on understanding the origin and evolutionary consequences of karyotype instability in *Polyommatus* and *Leptidea*.

PUBLICATIONS 2012

ISI Articles

- Dapporto, L., Bruschini, C., Dinca, V., Vila, R., and Dennis, R.L.H. 2012. Identifying zones of phenetic compression in West Mediterranean butterflies (Satyrinae): refugia, invasion and hybridization. *Diversity and Distributions* 18: 1066-1076.
- Hernández-Roldán, J.L., Munguira, M.L., Wagner, W., and Vila, R. 2012. Comparative analysis and taxonomic use of the morphology of immature stages and natural history traits in European species of *Pyrgus* Hübner, 1819 (Lepidoptera: HesperIIDae, Pyrginae). *Zootaxa* 3470: 1-71.
- Talavera, G., Lukhtanov, V.A., Pierce, N.E., and Vila, R. 2012. Establishing criteria for higher level taxonomic classification using molecular data: an example with blue butterflies (Lepidoptera, Lycaenidae). *Cladistics* 1-27. doi: 10.1111/j.1096-0031.2012.00421.x.

Book Chapters

- Vila, R. 2012. Lepidópteros. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de seres vivos*. Madrid. pp 342-351.

Other Publications

- Arrizabalaga, A., Stefanescu, C., Vallhonrat, F., Dantart, J., Vila, R., Jubany, J., Sesma, J.M., Viader, S., and Dinca, V. 2012. Proposta de noms comuns per a les papallones diürnes (ropalòcers) catalanes. *Butlletí de la Societat Catalana de Lepidopterologia* 103: 5-28; 1 XII.

Fig. 4: Adult butterflies are collected in warm weather, but overwintering caterpillars can be found in the worst conditions. Roger Vila and colleagues Juan L. Hernández-Roldán and Santi Viader are looking for larvae in Albarracín Mts. under a blizzard. Image: Juan Carlos Vicente Arranz.



FUNDED PROJECTS

Project Title: Faunal genetic comparisons to infer large-scale biogeographical patterns: the colonization of Western Mediterranean islands by butterflies (CGL2010-21226/BOS)

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: Roger Vila

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

Financed by: Fundación BBVA

Years: 2009-2012

PI: Roger Vila

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

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

Years: 2009-2013

PI: Alberto Tinaut

Project Title: Species Recovery Program (SRP) for 4 of the 15 threatened endemic species of butterflies in continental Europe - phase I

Financed by: MAVA Foundation Pour la Nature

Years: 2012-2015

PI: Miguel López Munguira

Fig. 5: Vlad Dinca (left) and Leonardo Dapporto (center) are key collaborators for our project on the West Mediterranean butterflies. In the picture, collecting in Marettimo Island (West of Sicily).

Image: Vlad Dinca.



GROUP

PHYLOGENY AND PHYLOGEOGRAPHY OF MAMMALS



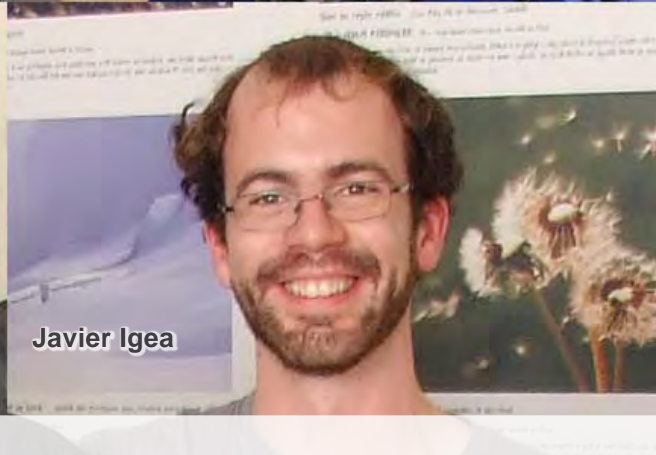
Marina Querejeta



José Castresana



Ana Rodríguez-Prieto



Javier Igea

GROUP MEMBERS

José Castresana, Group Leader

| Research Scientist, CSIC



Javier Igea, PhD Student | JAEPRE-CSIC Fellowship

Marina Querejeta, PhD Student | FPI Fellowship, MINECO

Ana Rodríguez-Prieto, PhD Student | FPI Fellowship, MINECO

RESEARCH OUTLINE

Our main goal is the application of phylogenetic and genomic analyses to study animal biodiversity and evolution. Using different types of markers, we are studying the phylogeographic patterns and the population history of several species of small mammals, some of them of great conservation importance. Another aspect of these studies applied to groups of closely related species is to try to better understand how speciation occurred in mammals. We are also interested in the study of global diversification patterns and in the factors that affect the net generation of species. Finally, since phylogenies are one of the main tools of our work, we are always trying to improve phylogenetic analyses by introducing new methodologies and programs.

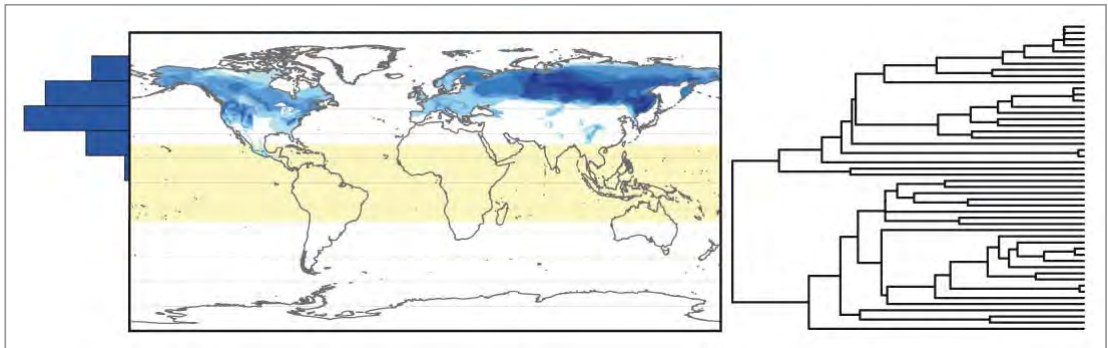


Fig. 1: We have studied latitudinal differences in diversification rates in order to test if a greater diversification rate in the Tropics could explain the latitudinal gradient of diversity. In this example, we show the distribution map of all the species of the genus *Sorex*, with color intensity reflecting species richness. The histogram on the left panel indicates the number of species in different latitudinal bands. From the dated phylogenetic tree of this genus (right panel), diversification rates are calculated. The same analysis for many mammalian genera indicated that diversification rates were not different for groups with different latitudinal distribution.

RESEARCH LINES

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

The reconstruction of species trees of closely related species requires the use of multiple genetic markers. To be able to effectively use these techniques in mammals, we are developing intronic markers from the mammalian genomes available in databases. In addition, we are sequencing these markers in different groups of small mammals (water shrews of the genus *Neomys* and Mediterranean voles of the genus *Microtus*) to study patterns of genetic diversity, gene flow and, in general, to better understand how speciation occurred in mammals. We are also interested in helping to detect cryptic lineages with these markers. For these studies we are making extensive use of noninvasive samples such as skulls obtained from owl pellets.

2. Evolution and conservation genetics of the pyrenean desman (*Galemys pyrenaicus*)

The Pyrenean desman (*Galemys pyrenaicus*) is a small semi-aquatic mammal endemic to the northern half of the Iberian Peninsula. It is a species of great evolutionary and ecological interest, but its genetic structure is yet to be investigated. Using mitochondrial and nuclear data, we are studying the phylogeography and population history of this unique species. Much of the material that we use for genetic studies comes from the droppings that desmans deposit on emerged rocks of the rivers. To get additional samples and to carry out this research, we are collaborating with scientists from many different institutions. The results we are obtaining may have crucial implications for the conservation of this endangered species.

3. Analysis of species diversification in mammals

Species-level phylogenies contain plenty of information in their patterns of branch splits. For example, it is possible to estimate diversification rates from phylogenetic trees that have an adequate sampling of species. It is also possible to study the variability of diversification rates between different groups or across the globe. The availability of a large amount of sequences in databases allows us to perform large-scale phylogenetic analyses in mammals with the purpose of studying their global diversification patterns.

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

Phylogenetic trees are essential in evolutionary biology and, therefore, it is important to understand their potentials and limitations. With this aim, we are working on different aspects of tree reconstruction and on the detection of artifacts that may affect phylogenetic analyses. We are interested in all steps of these analyses, including the generation of alignments, the reconstruction of phylogenetic trees, the comparison of these trees and the extraction of useful information from them. Furthermore, we are interested in methodologies at the interphase between phylogenetics and population genetics, and we work on devising the best conditions for estimating species trees from gene trees. Methods and software that we develop are made available online.

PUBLICATIONS 2012

ISI Articles

- Sánchez-Gracia, A., and Castresana, J. 2012. Impact of deep coalescence on the reliability of species tree inference from different types of DNA markers in mammals. *PLOS ONE* 7: e30239.
- Soria-Carrasco, V., and Castresana, J. 2012. Diversification rates and the latitudinal gradient of diversity in mammals. *Proceedings of the Royal Society B: Biological Sciences* 279: 4148-4155.

Book Chapters

- Castresana, J. 2012. Deutoróstomos. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 352-359.

FUNDED PROJECTS

Project Title: Reconstruction of species trees with genomic markers and its application to the study of mammalian speciation

Financed by: Ministerio de Ciencia e Innovación (CGL2011-22640)

Years: 2012-2014

PI: José Castresana

Project Title: Distribution and genetic diversity of the Iberian desman

(Galemys pyrenaicus) in the Spanish Network of National Parks

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

Years: 2009-2012

PI: Joaquim Gosálbez

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO

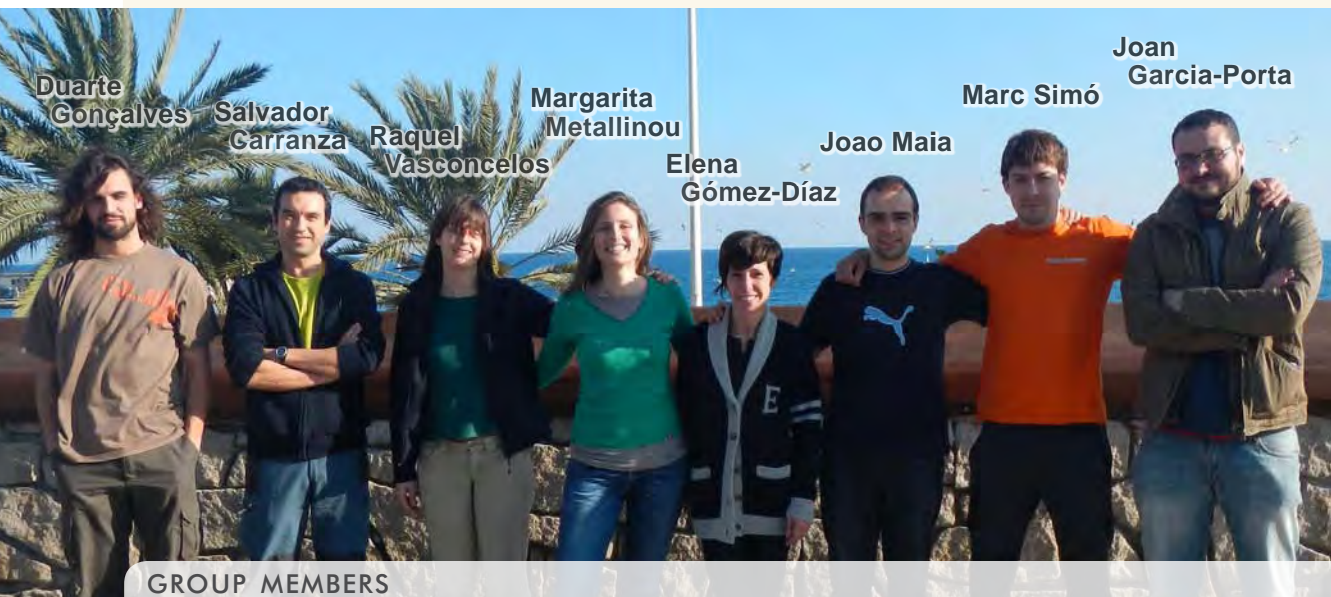
Financed by: Generalitat de Catalunya (2009 SGR 1462)

Years: 2009-2013

PI: Salvador Carranza

GROUP

SYSTEMATICS, BIOGEOGRAPHY AND EVOLUTION
OF REPTILES AND AMPHIBIANS



GROUP MEMBERS

Salvador Carranza, Group Leader

| Tenured Scientist, CSIC



- Elena Gómez-Díaz, Post-doc Researcher | Juan de la Cierva Contract
- Raquel Vasconcelos, Post-doc Researcher | FCT Scholarship, Portugal
- Mafalda Barata, PhD Student Co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal
- Joan Garcia-Porta, PhD Student | JAEPRE-CSIC Fellowship
- Duarte Gonçalves, PhD Student Co-supervised with Dr. J.C. Brito, CIBIO, Portugal | FCT Scholarship, Portugal
- Joao Maia, PhD Student Co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal
- Joana Mendes, PhD Student Co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal
- Margarita Metallinou, PhD Student | FPU Scholarship, MEC
- Philip de Pous, PhD Student Co-supervised with Delfi Sanuy, UDL | FI Scholarship
- Catarina Rato, PhD Student Co-supervised with Dr. D.J. Harris, CIBIO, Portugal | FCT Scholarship, Portugal
- Emilio Valbuena Ureña, PhD Student | Teaching Assistant, UAB, Barcelona
- Marc Simó, MSc Student | Master in Biodiversity, University of Barcelona

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 research lines focus mainly on the faunas of the Mediterranean Basin and Arabia, including some oceanic and continental islands, as for instance the Canary and the Cape Verde islands in the Atlantic Ocean and, since 2010, the unique archipelago of Socotra in the Indian Ocean.



Fig. 1: *Hemidactylus luqueorum*. A new species of *Hemidactylus* endemic to the *Jebel Akhdar*, *Oman*, described in 2012 by S. Carranza and E.N. Arnold.

Fig. 2: *Hemidactylus paucituberculatus*. A new species of *Hemidactylus* from *Salalah*, *South Oman*, described in 2012 by S. Carranza and E.N. Arnold.

RESEARCH LINES

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; and 3) 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 in 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 island 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.



Fig. 3: *Telescopus dhara* from Wadi Ayun, South Oman.

PUBLICATIONS 2012

ISI Articles

- Barata, M., Carranza, S., and Harris, D.J. 2012. Extreme genetic diversity in the lizard *Atlantolacerta andreaskyi* (Werner, 1929): a montane cryptic species complex. *BMC Evolutionary Biology* 12: 167.
- Carranza, S., and Arnold, E.N. 2012. A review of the geckos of the genus *Hemidactylus* (Squamata: Gekkonidae) from Oman based on morphology, mitochondrial and nuclear data, with descriptions of eight new species. *Zootaxa* 3378: 1-95.
- Dietrich, M., Kempf, F., Gómez-Díaz, E., Kitaysky, A.S., Hipfner, J.M., Boulinier, T., and McCoy, K.D. 2012. Inter-oceanic variation in patterns of host-associated divergence in a seabird ectoparasite. *Journal of Biogeography* 39: 545-555.
- Garcia-Porta, J., Litvinchuc, S., Crochet, P.A., Romano, A., Geniez, Ph., Lo-Valvo, M., Lymberakis, P., and Carranza, S. 2012. Molecular phylogenetics and historical biogeography of the west-palaearctic common toads (*Bufo bufo* species complex). *Molecular Phylogenetics and Evolution* 63:1 13-130.
- Gómez-Díaz, E., Jordà, M., Peinado, M.A., and Rivero, A. 2012. Epigenetics of host-pathogen interactions: the road ahead and the road behind. *PLoS Pathogens* 8 (11): e1003007.
- Gómez-Díaz, E., Morris-Pocock, J., González-Solís, J., and McCoy, K.D. 2012. Trans-oceanic host dispersal explains high seabird tick diversity on Cape Verde islands. *Biology Letters* 23: 616-619.
- Gómez-Díaz, E., Sindaco, R., Pupin, F., Fasola, M., and Carranza, S. 2012. Origin and in situ diversification in *Hemidactylus* geckos of the Socotra Archipelago. *Molecular Ecology* 21: 4074-4092.
- Llorente, G.A., Vidal-García, M., Garriga, N., Carranza, S., Pleguezuelos, J.M., and Santos, X. 2012. Lessons from a complex biogeographic scenario: morphological

characters match mitochondrial lineages within Iberian *Coronella austriaca* (Reptilia: Colubridae). *Biological Journal of the Linnean Society* 106: 210-223.

- Maia, J., Gómez-Díaz, E., and Harris, D.J. 2012. Apicomplexa primers amplify *Proteromonas* (Stramenopiles, Slopalinida, Proteromonadidae) in tissue and blood samples from lizards. *Acta parasitologica*, 57: 337-341.
- Metallinou, M., Arnold, E.N., Crochet, P.A., Geniez, Ph., Brito, J.C., Lymberakis, P., Baha el Din, Sh., Sindaco, R., Robinson, M., and Carranza, S. 2012. Conquering the Sahara and Arabian deserts: Systematics and biogeography of *Stenodactylus* geckos (Reptilia: Gekkonidae). *BMC Evolutionary Biology* 12: 258.



Fig. 4: *Stenodactylus arabicus* from the Wahiba Sands, Oman.

- Noel, V.L., Gómez-Díaz, E., Risterucci, A., and McCoy, K.D. 2012. Isolation and characterization of new polymorphic microsatellite markers for the tick *Ixodes ricinus* (Acari, Ixodidae). *Acarologia* 52 (2): 123-128.
- Ord, T.J., and Garcia-Porta, J. 2012. Is sociality required for the evolution of communicative complexity? Evidence weighed against alternative hypotheses in diverse taxonomic groups. *Philosophical Transactions of the Royal Society of London Series B*. 316: 1811-1828.
- Rato, C., Carranza, S., and Harris, D.J. 2012. Evolutionary history of the genus *Tarentola* (Gekkota: Phyllodactylidae) from the Mediterranean Basin, estimated using multilocus sequence data. *BMC Evolutionary Biology* 12:14.
- Santos, X., Rato, C., Carranza, S., Carretero, M., and Pleguezuelos, J.M. 2012. Complex phylogeography in the Southern Smooth snake (*Coronella girondica*) supported by mtDNA sequences. *Journal of Zoological Systematics and Evolutionary Research* 50: 210-219.
- Santos, X., Rato, C., Carranza, S., Carretero, M., and Pleguezuelos, J.M. 2012. Morphology matches mtDNA lineages in the Southern Smooth snake *Coronella girondica* from southeastern Iberia. *Zoology* 115: 398-404.
- Sindaco, R., Metallinou, M., Pupin, F., Fasola, M., and Carranza, S. 2012. Forgotten in the ocean: systematics, biogeography and evolution of the *Trachylepis* skinks of the Socotra Archipelago. *Zoologica Scripta* 41: 346-362.
- Stekolnikov, A.A., Carranza, S., and Gómez-Díaz, E. 2012. A new genus and species of Apoloniinae (Acari: Trombiculidae) from Oman. *Zootaxa* 3499: 74-80.
- Vasconcelos, R., Brito, J., Carvalho, S.B., Carranza, S., and Harris, D.J. 2012. Identifying priority areas for island endemics using genetic versus specific diversity - the case of terrestrial reptiles of the Cape Verde Islands. *Biological Conservation* 153: 276-286.

- Vasconcelos, R., Perera, A., Geniez, Ph., Harris, D.J., and Carranza, S. 2012. An integrative taxonomic revision of the *Tarentola* geckos (Squamata, Phyllodactylidae) of the Cape Verde Islands. *Zoological Journal of the Linnean Society* 164: 328-360.
- Vasconcelos, R., Santos, X., and Carretero, M.A. 2012. High temperatures constrain microhabitat selection and activity patterns of the insular Cape Verde wall gecko. *Journal of Arid Environments* 8: 18-25.

Book Chapters

- Carranza, S. 2012. Sauropsidos. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 424-433.

Other Publications

- de Pous, P., Beukema, W., Dingemans, D., Donaire, D., Geniez, P., and El Mouden, H. 2012. Distribution review, habitat suitability and conservation of the endangered and endemic Moroccan spadefoot toad (*Pelobates varaldii*). *Basic and Applied Herpetology* 26: 57-71.
- de Pous, P., Speybroeck, J., Bogaerts, S., Pasmans, F., and Beukema, W. 2012. A contribution to the atlas of the terrestrial herpetofauna of Sardinia. *Herpetology Notes* 5: 391-405.

FUNDED PROJECTS

Project Title: Zoological Systematics and Evolution Research Group – ZOOSYSEVO

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

Years: 2009-2013

PI: Salvador Carranza

Project Title: Using Agamid lizards to understand the origin and colonization of North African and Arabian deserts

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

Years: 2011-2012

PI: Salvador Carranza

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

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

Years: 2010-2012

PI: Salvador Carranza

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

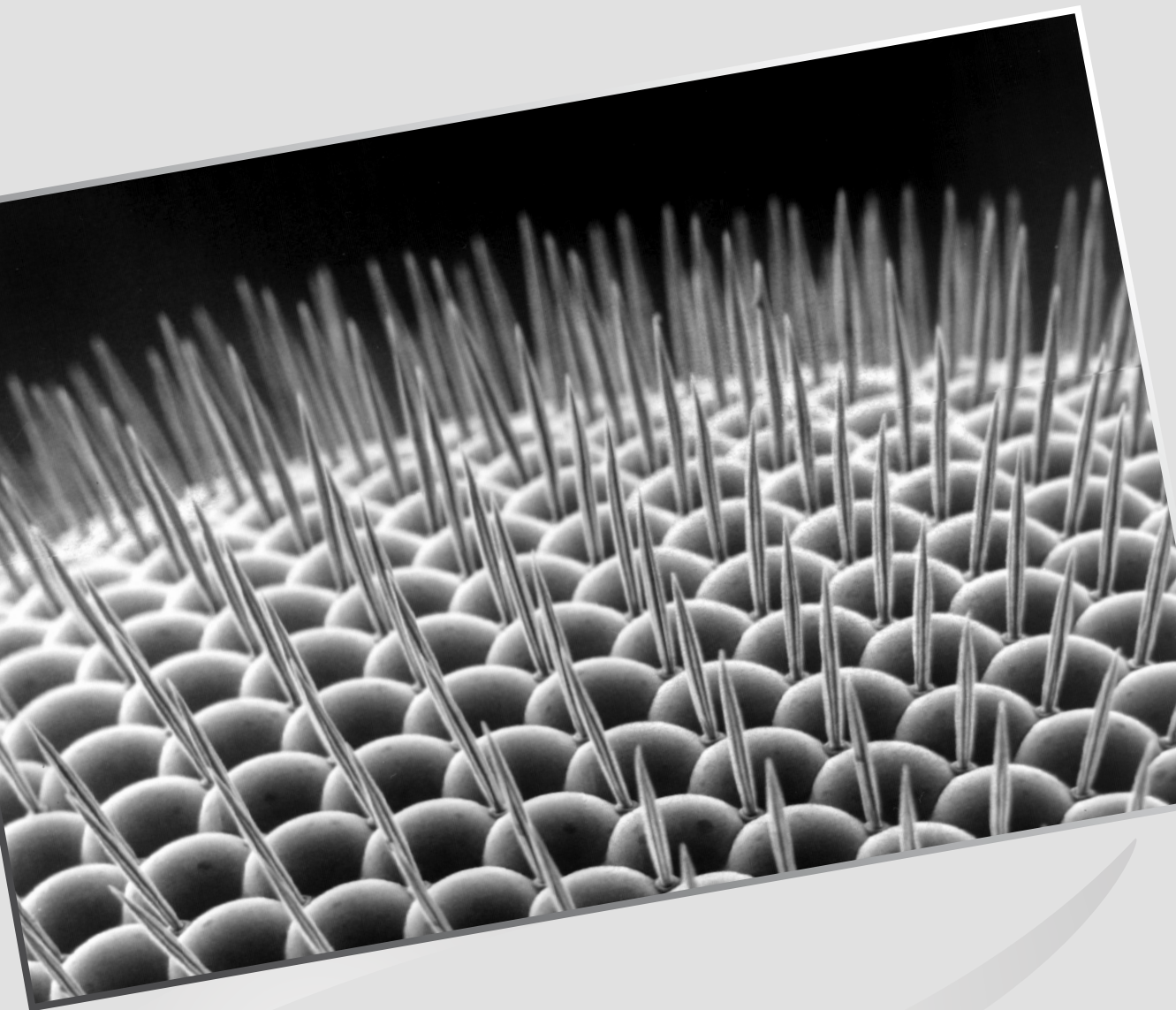
Financed by: European Commission, FP7-PEOPLE-Reintegration Grants. ERG-PARIS-276838.


Years: 2010-2013

PI: Elena Gómez-Díaz



Fig. 5: Wadi Tanuf with the old city of Tanuf in the foreground.





PROGRAM

comparative and computational genomics

RESEARCH GROUPS

Evolutionary and functional genomics

Josefa González, Group Leader

Subgroups:

Evolutionary and functional genomics

| *Josefa González, PI*

Drosophila telomeres

| *Elena Casacuberta, PI*

Evolutionary genomics

Arcadi Navarro, Group Leader

Paleogenomics

Carles Lalueza-Fox, Group Leader

Primate genomics

Tomàs Marquès-Bonet, Group Leader

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 species. Thus, studying full genomes is key to answer the basic questions posed eight decades ago by the research paradigm created by the Synthetic Theory of Evolution: how much adaptation can we detect in nature? In addition to the study of adaptation, genomics allows us to answer questions about other crucial evolutionary phenomena such as chromosomal evolution, speciation or the dynamics of transposable elements. Understanding these phenomena is fundamental in shedding light in issues as varied as hominization or the genetic architecture of complex phenotypes.

In the Comparative and Computational Genomics program, genes and 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 genome diversity and function, with a recent emphasis on phenotypic differences between individuals and species. To achieve these goals, we deploy state-of-the-art techniques at both the experimental and nuclear level.

GROUP

EVOLUTIONARY AND FUNCTIONAL GENOMICS



**Karine
Ribeiro**

**Marta
Perera**

**Josefa
González**

**Anna
Ullastres**

Lidia Mateo

Lain Guio

**Miriam
Merenciano**



**Elena
Casacuberta**

Elisenda López

Rute Souza

GROUP MEMBERS

Josefa González, Group Leader

| Ramón y Cajal Researcher



| Subgroup: Evolutionary and functional genomics

Josefa González | Ramón y Cajal Researcher

Lain Guio, PhD Student | FI Fellowship

Anna Ullastres, PhD Student | FPI Fellowship

Lidia Mateo, Undergraduate Student | JAE-Intro Fellowship,
UPF Contract

Miriam Merenciano, Undergraduate Student

Marta Perera, Undergraduate Student

Karine Ribeiro, Undergraduate Student | Science Without
Borders Fellowship

| Subgroup: Drosophila telomeres

Elena Casacuberta | Tenured Scientist, CSIC

Elisenda López Panadès, PhD Student | UPF Fellowship

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

RESEARCH OUTLINE

The Evolutionary and Functional Genomics group uses transposable elements as a tool to unravel genome function and evolution. The group contains two subgroups: "Evolutionary and functional genomics", lead by Josefa González, and "Drosophila telomeres", lead by Elena Casacuberta.

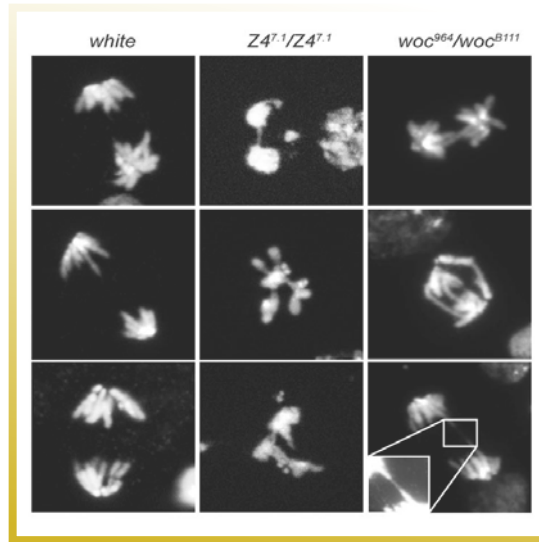


Fig. 1: Anaphase preparations of *Drosophila* mitotic chromosomes. 1st column: negative control; 2nd column: homozygous combination of the $Z4^{7.1}$ mutant allele; and 3rd column: positive control, heteroallelic combination of woc^{964}/woc^{B111} .

RESEARCH LINES

SUBGROUP: EVOLUTIONARY AND FUNCTIONAL GENOMICS

Adaptation is the key concept in Evolutionary Biology. Understanding adaptation has important scientific and social implications since adaptation underlies processes such as the ability of species to survive in changing environments, host-pathogen interactions, or resistance to pesticides and drugs (e.g. antibiotics or cancer chemotherapies). Although there are some brilliant examples in which adaptive mutations have been connected to their phenotypic and fitness effects, adaptation is to date a poorly understood process. This is largely because current approaches to the study of adaptation are often exclusively based on a priori candidate genes or on searching for signals of selection at the DNA level, giving us an incomplete and biased picture of the adaptive process.

In our lab we aimed at understanding the molecular process of adaptation and its functional consequences. Towards this end, we study recent transposable element (TE)-induced adaptations in *Drosophila melanogaster*. We combine omics strategies with detailed molecular and functional analyses of the identified adaptive mutations in order to arrive at a comprehensive picture of adaptation.

SUBGROUP: DROSOPHILA TELOMERES

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. We are also isolating and identifying protein complexes using the telomeric proteins as bait in order to understand which are the cellular partners that assist the telomeric proteins throughout their life cycle.

2. Telomere replication in the *Drosophila* germ line

We have isolated some interacting partners of the telomere retrotransposons that are essential genes for the development of the *Drosophila* germ line and the oocytes. We are currently investigating the life cycle of the telomere retrotransposons in the different germ cells of the developing ovary in wild type and mutant strains in order to understand which are the necessary interactions that the telomere proteins establish to replicate the telomeres in the germ line tissue.

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.

PUBLICATIONS 2012

ISI Articles

- Calvete, O., González, J., Betran, E., and Ruiz, A. 2012. Segmental duplication, microinversion and gene loss associated with a complex inversion breakpoint region in *Drosophila*. *Molecular Biology and Evolution* 29 (7): 1875-1889.
- Petit, N., Piñeyro, D., López, E., Casacuberta, E., and Navarro, A. 2012. HeT_A_pi1, a piRNA target sequence in the *Drosophila* telomeric retrotransposon HeT-A is extremely conserved across copies and species. *PLOS ONE* 7 (5): e37405.
- Silva-Sousa, R., López, E., Piñeyro, D., and Casacuberta, E. 2012. The Chromosomal Proteins JIL-1 and Z4/Putzig Regulate the Telomeric Chromatin in *Drosophila melanogaster*. *PLoS Genetics* 8 (12): e1003153.
- Zhu, Y., Bergland, A.O., González, J., and Petrov, D.A. 2012. Empirical validation of pooled genome population re-sequencing in *Drosophila melanogaster*. *PLOS ONE* 7 (7): e41901.

Books/Book Chapters

- González, J., and Petrov, D.A. 2012. Evolution of genome content: population dynamics of transposable elements in flies and humans. *Methods in Molecular Biology* 855: 361-383.
- Silva-Sousa, R., López-Panades, E., and Casacuberta, E. 2012. *Drosophila* Telomeres: an Example of Co-Evolution with Transposable Elements. Garrido-Ramos MA (ed.) In: *Repetitive DNA. Genome Dyn.* Basel, Karger, vol 7, pp 46-67. doi: 10.1159/000337127.

FUNDED PROJECTS

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

Financed by: Ministerio de Ciencia e Innovación

Years: 2012-2014

PI: Josefa González

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

Financed by: European Commission

Years: 2011-2014

PI: Josefa González

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

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2015

PI: Josefa González

Project Title: Estudio de los aspectos funcionales y evolutivos de los telomeros de Drosophila

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

Years: 2010-2012

PI: Elena Casacuberta

OUTREACH PROJECTS

Project Title: La ciència és part del teu món (2012ACDC 00025)

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

Year: 2012

PI: Josefa González

Project Title: La ciencia en tu mundo (FCT-12-4240)

Financed by: Fundación Española para la Ciencia y la Tecnología

Years: 2012-2013

PI: Josefa González

Project Title: Reconstruint el nostre passat evolutiu

Financed by: Fundació CatalunyaCaixa

Year: 2012

PI: Josefa González

GROUP

EVOLUTIONARY GENOMICS



Carlos Morcillo

Txema Heredia

Juan Antonio Rodríguez-Pérez

Urko Martínez

Fernando Muñiz

Jordi Rambla

Ángel Carreño

Gabriel Santpere

Diego Hartasánchez

María Niño

Arcadi Navarro

Natalia Petit

GROUP MEMBERS

Arcadi Navarro, Group Leader

| Professor, UPF Research Professor, ICREA



Rui Faria, Post-doc | FCT Fellowship

Carlos Morcillo, Post-doc | Project Contract

Gabriel Santpere, Post-doc | Project Contract

Natalia Petit, Post-doc | JAE Contract

Diego Hartasánchez, PhD Student | JAE Contract

Urko Martínez, PhD Student | UPF Scholarship

Juan Antonio Rodríguez-Pérez, PhD Student | Project Contract

Ángel Carreño, IT Technician | INB (National Bioinformatics Institute)

Txema Heredia, IT Technician | INB (National Bioinformatics Institute)

Fernando Muñiz, IT Technician | INB (National Bioinformatics Institute)

Dan Ouchi, IT Technician | INB (National Bioinformatics Institute)

María Niño, IT Technician | JAE Contract

Jordi Rambla, IT Technician | INB (National Bioinformatics Institute)

RESEARCH OUTLINE

Life as we see it in our planet today has been shaped by many different biological processes during billions of years. These processes leave a signature in our genomes in the form of differences between species, or between individuals of the same species. Interrogating these patterns of genome diversity we can infer what are the forces that affect living organisms, how and when they act, and how do they affect such various things as biodiversity, human emotions or the differential susceptibility of different people to certain diseases. All this knowledge empowers us to control our future but, above all, it is fun to obtain.

In practical terms, there are two main kinds of research we do in our group. First, we develop and analyze models that help to answer questions concerning chromosomal evolution, genome dynamics, speciation, comparative gene expression and the genetic architecture of complex human traits (including disease). The tools we use for these research lines include analytical and simulation studies, together with data mining and analysis. Second, we also carry out wet-lab research. Lately, we have focused on array CGH of primate genomes to study the evolution of copy-number variation and on Genome-Wide Association Studies of human socio-economic traits.

RESEARCH LINES

1. Chromosomal evolution and speciation

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

2. Segmental duplications and copy-number variation in primates

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

3. Detecting positive selection in the human lineage

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

4. World-wide distribution of human disease

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

5. Genoeconomics

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

ISI Articles

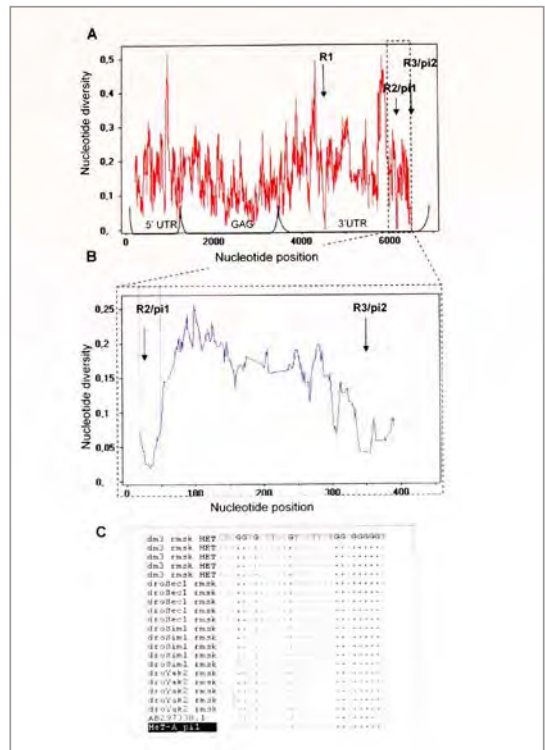
- Cantó, E., Reverter, F., Morcillo-Suárez, C., Matesanz, F., Fernández, O., Izquierdo, G., Vandenbroeck, K., Rodríguez-Antigüedad, A., Urcelay, E., Arroyo, R., Otaegui, D., Olascoaga, J., Saiz, A., Navarro, A., Sanchez, A., Domínguez, C., Caminero, A., Horga, A., Tintoré, M., Montalban, X., and Comabella, M. 2012. Chitinase 3-like 1 plasma levels are increased in patients with progressive forms of multiple sclerosis. *Multiple Sclerosis Journal* 18 (7): 983-990.
- Dickhaus, T., Straßburger, K., Schunk, D., Morcillo-Suárez, C., Illig, T., and Navarro, A. 2012. How to analyze many contingency tables simultaneously in genetic association studies. *Statistical Applications in Genetics and Molecular Biology* 11 (4): ISSN (Online) 1544-6115.
- Garcia-Mas, J., Benjak, A., Sanseverino, W., Bourgeoisa, M., Mira, G., González, V.M., Hénaff, E., Cámara, F., Cozzuto, L., Lowy, E., Alioto, T., Capella-Gutiérrez, S., Blanca, J., Cañizares, J., Ziarsolo, P., Gonzalez-Ibeas, D., Rodríguez-Moreno, L., Droege, M., Dui, L., Álvarez-Tejado, M., Lorente-Galdós, B., Melé, M., Yang, L., Weng, Y., Navarro, A., Marqués-Bonet, T., Aranda, M.A., Nuez, F., Picó, B., Gabaldón, T., Roma, G., Guigó, R., Casacuberta, J.M., Arús, P., and Puigdomènech, P. 2012. The genome of melon (*Cucumis melo* L.). Genome amplification in the absence of recent duplication in an old widely cultivated species. *Proceedings of the National Academy of Sciences USA* 109 (29): 11872-11877.
- Lorente-Galdós, B., Medina, I., Morcillo-Suárez, C., Sangrós, R., Alegre, J., Pita, G., Vellalta, G., Malats, N., Pisano, D.G., Dopazo, J., and Navarro, A. 2012. SYSNPs (Select Your SNPs): a web tool for automatic and massive selection of SNPs. *International Journal of Data Mining and Bioinformatics* 6 (3): 324-334.
- Petit, N., Piñeyro, D., López-Panadès, E., Casacuberta, E., and Navarro, A. 2012. HeT-A_pi1, a piRNA target sequence in the *Drosophila* telomeric retrotransposon HeT-A, is extremely conserved across copies and species. *PLOS ONE* 7 (5): e37405.

Fig. 1: Nucleotide diversity estimates among elements and species.

(A) Conservation analysis of the six complete HeT-A elements from *D. melanogaster*. Sliding windows of 25 ntds size and 1 ntd steps are represented. The significantly conserved regions (R) in the 3'UTR are marked with arrows.

(B) Conservation analysis of the last 500 ntds of the 3'UTR among species (*D. melanogaster*, *D. sechellia*, *D. simulans*, *D. yakuba*). Graph constructed with an alignment of homologous sequences, longer than 350 ntds, obtained from the Blast analysis with a window size of 25 ntds and step size of 1 ntds (see Supplementary Results). Number of aligned sequences: *D. melanogaster* 26, *D. sechellia* 26, *D. yakuba* 6 and *D. simulans* 12. The estimated average nucleotide diversity among all 72 sequences is 0.13571. For nucleotide diversity within each species see Supplementary Figure S1.

(C) Alignment of the piRNA target sequence among HeT-A copies from four *Drosophila* species. Nucleotide diversities: all, 0.048; *D. sechellia*, 0.028, *D. yakuba*, 0.021; *D. simulans*, 0.058; and *D. melanogaster*, 0.0437.



Books/Book Chapters

- Freitas, A.T., and Navarro, A. 2012. Bioinformatics in Personalized Medicine. Series: *Lecture Notes in Computer Science*. Subseries: *Lecture Notes in Bioinformatics*. Vol. 6620. ISBN 978-3-642-28061-0. Springer-Verlag (Berlin-Heidelberg).
- Muñoz-Fernandez, F., Carreño-Torres, A., Morcillo-Suárez, C., and Navarro, A. 2012. Application of Array-Oriented Scientific Data Formats (NetCDF) to Genotype Data, GWASpi as an Example. In: *Bioinformatics in Personalized Medicine*. Series: *Lecture Notes in Computer Science*. Subseries: *Lecture Notes in Bioinformatics*. A. Freitas, A.T., and Navarro, A. (eds.) *Lecture Notes in Computer Science 6620*: 8-20.
- Navarro, A. 2012. Genes, dinero y Darwin: una nueva aproximación al estudio de la evolución humana. In: *Visiones del ser humano - Del pasado al presente*, pp 13-21. Mateos-Cachorro, A., and Perote-Alejandre, A. (eds.) ISBN: 978-84-7867-085-7. CNIEH (Burgos, Spain).

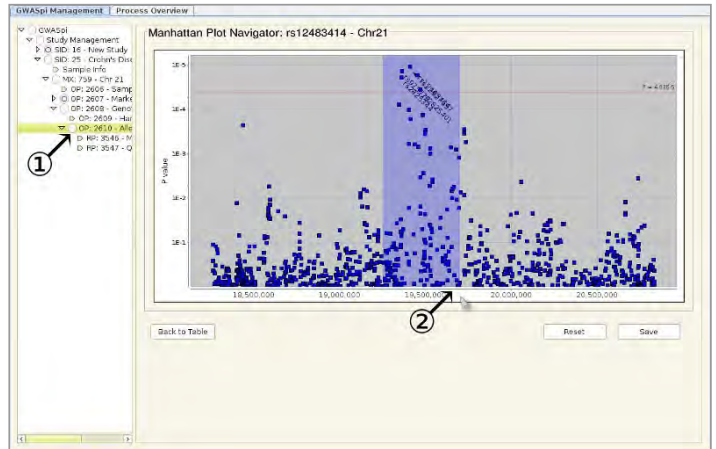


Fig. 2: Screenshot of GWASpi's Manhattan plot navigator.

FUNDED PROJECTS

Project Title: Identifying evolutionary novelties and adaptation in duplicated regions of the genomes of primates

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

Years: 2009-2012

PI: Arcadi Navarro

Project Title: Exploring the behavioral genetics of trade and cooperation

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

Years: 2008-2012

PI: Arcadi Navarro

Project Title: INB GN8

Financed by: Genoma España (Instituto Nacional de Bioinformática)

Years: 2003-2012

PI: Arcadi Navarro

Project Title: Red Española de Esclerosis Múltiple

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

Years: 2009-2012

PI: Arcadi Navarro

GROUP
PALEOGENOMICS



GROUP MEMBERS

Carles Lalueza-Fox, Group Leader

| Research Scientist, CSIC



Oscar Ramírez | JAEDOC, CSIC Contract

Iñigo Olalde, PhD Student | Basque Country Scholarship

Federico Sánchez-Quinto, PhD Student | FPI Scholarship, MICINN

Laura Matas-Lalueza | Master Student

RESEARCH OUTLINE

Our research group focuses on paleogenomics, the study of structure, function and organization of ancestral genomes. We are interested in different evolutionary problems that can be answered with ancient DNA data, involving human evolution, population dynamics and diversity, phylogenetics, phylogeography and adaptive processes. We work with different animal species, 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). We are also investigating the evolutionary dynamics of the Prehistory of Europe through the analysis of Mesolithic and Neolithic human genomes, with a special focus on Southern Europe.

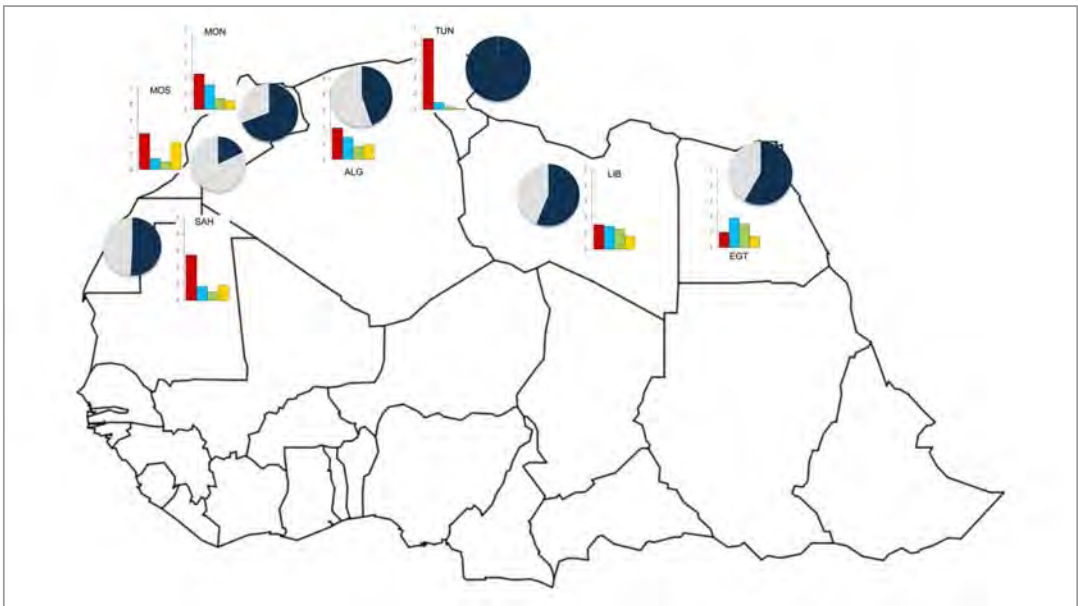


Fig. 1: Neanderthal genetic introgression in North African populations as a fraction of that found in Europeans. Relative proportion of Neanderthal ancestry for each population is presented as the dark blue section of the pies on a map of North Africa. Additionally, each population is also represented as a barplot of the different geographic genetic components: in red, North African; in blue, Sub-Saharan; in green, European; and in yellow, Middle Eastern.

RESEARCH LINES

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 13 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 and the kinship relationships within a Neandertal family group.

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

We are studying different extinct species, to answer specific questions about their phylogeny, adaptation and evolution. We are also interested in the analysis of domestication processes at the genomic level, and we are currently comparing structural variations (CNVs) between dogs and wolves.

4. Extinct modern human populations

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

5. Personal genomics

We are conducting a pioneer project for obtaining complete genomes from historical characters, including Louis XVI -king of France-, thus bringing the personal genomics to a new dimension that could have historical implications.

PUBLICATIONS 2012

ISI Articles

- Burbano, H., Green, R.E., Maricic, T., Lalueza-Fox, C., de la Rasilla, M., Rosas, A., Kelso, J., Pollard, K.S., Lachmann, M., and Paabo, S. 2012. Analysis of Human Accelerated DNA Regions Using Archaic Hominin Genomes. *PLOS ONE* 7 (3): e32877.
- Cánovas, A., Pena, R.N., Gallardo, D., Ramírez, O., Amills, M., and Quintanilla, R. 2012. Segregation of regulatory polymorphisms with effects on the gluteus medius transcriptome in a purebred pig population. *PLOS ONE* 7 (4): e35583.
- Frayer, D., Fiore, I., Lalueza-Fox, C., Radovčić, J., and Bondioli, L. 2012. Replay to Benítez-Burraco and Longa: When is enough, enough? *Journal of Anthropological Science* 90: 1-6.
- Gallardo, D., Pena, R.N., Quintanilla, R., Ramírez, O., Almuzara, D., Noguera, J.L., and Amills, M. 2012. Quantitative trait loci analysis of a Duroc commercial population highlights differences in the genetic determination of meat quality traits at two different muscles. *Animal Genetics* 43 (6): 800-804.
- González-Ruiz, M., Santos, C., Jordana, X., Simón, M., Lalueza-Fox, C., Gigli, E., Aluja, M.P., and Malgosa, A. 2012. Tracing the origin of the East-west population admixture in the Altai region (central Asia). *PLOS ONE* 7 (11): e48904.
- Hardy, K., Buckley, S., Collins, M.J., Estalrich, A., Brothwell, D., Copeland, L., García-Tabernero, A., García-Vargas, S., de la Rasilla, M., Lalueza-Fox, C., Huguet, R., Bastir, M., Santamaría, D., Madella, M., Fernández Cortés, A., and Rosas, A. 2012. Neanderthal medics? Evidence for food, cooking and medicinal plants entrapped in Neanderthal dental calculus. *Naturwissenschaften* 99 (8): 617-626.

- Lalueza-Fox, C., Gigli, E., Sánchez-Quinto, F., de la Rasilla, M., Fortea, J., and Rosas, A. 2012. Issues from Neandertal genomics: Diversity, adaptation and hybridisation revised from the El Sidrón case study. *Quaternary International* 247: 10-14.
- Lalueza-Fox, C., Rosas, A., and de la Rasilla, M. 2012. Palaeogenetic research at El Sidrón Neandertal site. *Annals of Anatomy* 194 (1): 133-137.
- López-Valenzuela, M., Ramírez, O., Rosas, A., García-Vargas, S., de la Rasilla, M., Lalueza-Fox, C., and Espinosa-Parrilla, Y. 2012. An ancestral allele of miR-1304 present in Neanderthals regulates genes involved in enamel formation and could explain dental differences with modern humans. *Molecular Biology and Evolution* 29 (7):1797-1806.
- Mannino, M., Catalano, G., Talamo, S., Mannino, G., Di Salvo, R., Schimmenti, V., Lalueza-Fox, C., Messina, A., Petruso, D., Caramelli, D., Richards, MP., Sineo, L. 2012. Origin and Diet of the Prehistoric Hunter-Gatherers on the Mediterranean Island of Favignana (Égadi Islands, Sicily). *PLOS ONE* 7 (11).
- Rosas, A., Estalrich, A., García-Taberner, A., Bastir, M., García-Vargas, S., Sánchez-Meseguer, A., Huguet, R., Lalueza-Fox, C., Peña-Melián, A., Kranioti, E.F, Santamaría, D., de la Rasilla, M., and Fortea, F. 2012. Les Néandertaliens d'El Sidrón (Asturies, Espagne). Actualisation d'un nouvel échantillon. *L'Anthropologie* 116 (1): 57-76.
- Sánchez-Quinto, F., Rodríguez-Botigué, L., Civit, S., Arenas, C., Ávila-Arcos, M.C., Bustamante, C.D., Comas, D., and Lalueza-Fox, C. 2012. North African populations carry the signature of admixture with Neandertals. *PLOS ONE* 7 (10): e47765.
- Sánchez-Quinto, F., Schroeder, H., Ramírez, O., Ávila-Arcos, M.C., Pybus, M., Olalde, I., Velazquez, A.M.V., Prada Marcos, M.E., Vidal Encinas, J.M., Bertranpetit, J., Orlando, L., Gilbert, M.T.P., and Lalueza-Fox, C. 2012. Genomic affinities of two 7,000-year-old Iberian hunter-gatherers. *Current Biology* 22: 1494-1499. PMID: 22748318.

FUNDED PROJECTS

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

Financed by: Ministerio de Ciencia e Innovación

Years: 2010-2012

PI: Carles Lalueza-Fox

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

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

Years: 2009-2012

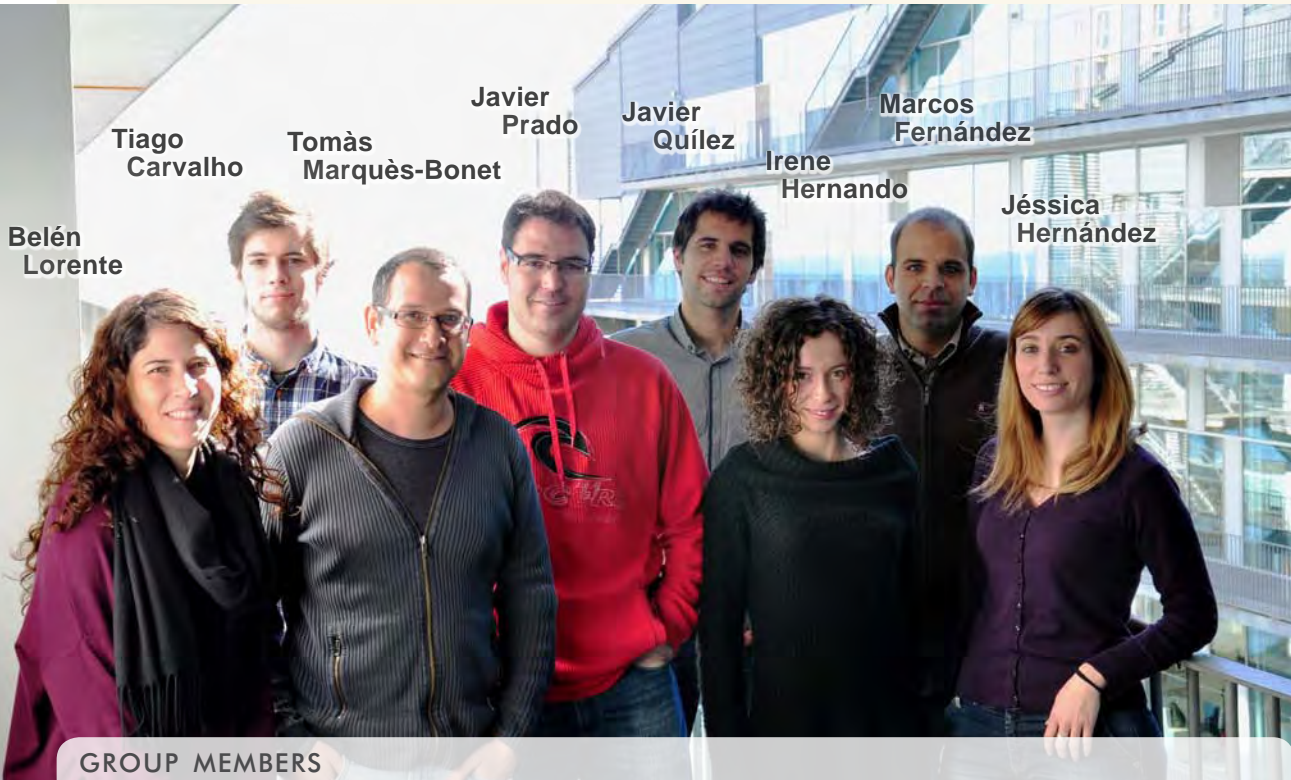
PI: Jaume Bertranpetit

Fig. 2: Image of La Braña 1 (León, Spain) of a 7,000 years-old mesolithic skeleton from which nuclear and mitochondrial DNA has been retrieved.



GROUP

PRIMATE GENOMICS



GROUP MEMBERS

Tomàs Marquès-Bonet, Group Leader

| ICREA Research Professor



Belén Lorente, Postdoc | ERC StGt Project Contract

Javier Quílez, Postdoc | ERC StGt Project Contract

Tiago Carvalho, PhD Student | ERC StGt Project Contract

Irene Hernando, PhD Student | FI Scholarship, Generalitat de Catalunya

Javier Prado, PhD Student | FI Scholarship, MEC

Marcos Fernández, Computational Support | ERC StGt Project Contract

Jéssica Hernández, Experimental Support | ERC StGt Project Contract

RESEARCH OUTLINE

Our main line of research is centered in the discovery of the extent of all kinds of genome variation within the great ape species and other mammalian genomes. The goal is to create an integrated view of genome 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 LINES

1. Genomic variation in ape genomes

Despite international efforts to characterize thousands of human genomes to understand the extent of structural variants in the human species, primates (our closest relatives) have somehow been forgotten. Yet, they are the ideal set of species to study the evolution of these features from both mechanistic and adaptive points of view. Most genome projects include only one individual as a reference, but in order to understand the impact of structural variants in the evolution of every species we need to re-sequence multiple individuals of each species. We can only understand the origins of genomic variants and phenotypical differences among species if we can model variation within species and compare it to a proper perspective with the differences among species.

2. Snowflake genome. Study of albinism in gorillas.

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

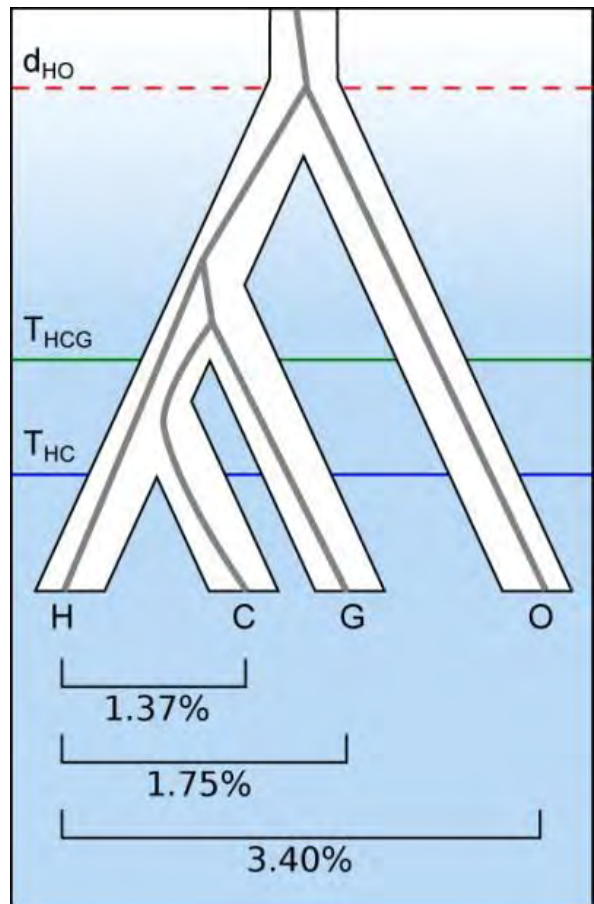
3. Epigenetics and transcriptomics of non-human primates

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

ISI Articles

- Garcia-Mas, J., Benjak, A., Sanseverino, W., Bourgeoisa, M., Mira, G., González, V.M, Hénaff, E., Cámara, F., Cozzuto, L., Lowy, E., Alioto, T., Capella-Gutiérrez, S., Blanca, J., Cañizares, J., Ziarsolo, P., Gonzalez-Ibeas, D., Rodríguez-Moreno, L., Droege, M., Dui, L., Álvarez-Tejado, M., Lorente-Galdós, B., Melé, M., Yang, L., Weng, Y., Navarro, A., Marquès-Bonet, T., Aranda, M.A, Nuez, F., Picó, B., Gabaldón, T., Roma, G., Guigó, R., Casacuberta, J.M, Arús, P., and Puigdomènech, P. 2012. The genome of melon (*Cucumis melo* L.). Genome amplification in the absence of recent duplication in an old widely cultivated species. *Proceedings of the National Academy of Sciences USA* 109 (29): 11872-11877.
- Prüfer, K., Munch, K., Hellmann, I., Akagi, K., Miller, J.R., Walenz, B., Koren, S., Sutton, G., Kodira, C., Winer, R., Knight, J.R., Mullikin, J.C., Meader, S.J., Ponting, C.P., Lunter, G., Higashino, S., Hobolth, A., Dutheil, J., Karakoç, E., Alkan, C., Sajjadian, S., Catacchio, C.R., Ventura, M., Marquès-Bonet, T., Eichler, E.E., André, C., Atencia, R., Mugisha, L., Junhold, J., Patterson, N., Siebauer, M., Good, J.M., Fischer, A., Ptak, S.E., Lachmann, M., Symer, D.E., Mailund, T., Schierup, M.H., Andrés, A.M., Kelso, J., and Pääbo, S. 2012. The bonobo genome compared with the chimpanzee and human genomes. *Nature* 486 (7404): 527-5231.

Fig. 1: Interior grey lines illustrate an example of incomplete lineage sorting at a particular genetic locus –in this case $((C, G), H), O$ rather than $((H, C), G), O$. Below are mean nucleotide divergences between human and the other great apes from the EPO alignment. (Scally et al., 2012. Nature).



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FUNDED PROJECTS

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

Financed by: European Research Council

Years: 2010-2014

PI: Tomàs Marquès-Bonet

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

Financed by: Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: Tomàs Marquès-Bonet





PROGRAM

complex systems

RESEARCH GROUPS

Complex systems

Ricard Solé, Group Leader

Language evolution

Luc Steels, Group Leader

This program involves the study of the evolution and organizing principles of both natural and artificial complexity. Using theoretical as well as experimental methods, we study the design principles of natural, technological and synthetic systems and how major transitions can occur. We also explore the possible and the actual in artificially designed systems spanning multiple scales, from engineered bacteria to humanoid robots. Among our major fields of analysis, we study the origins of innovation and universal laws of organization associated to communication, computation, cultural and technological evolution, multicellularity and collective intelligence.

GROUP
COMPLEX SYSTEMS



Sergi Valverde

Jordi
Delgado

Carlos
Rodríguez-Caso

Salvador Durán

Bernat
Corominas-Murtra

Núria Conde

Ricard Solé

Javier Macía

GROUP MEMBERS

Ricard Solé, Group Leader

| Professor, UPF • ICREA Researcher



Bernat Corominas-Murtra | Post-doc Researcher

Javier Macía | Post-doc Researcher

Carlos Rodríguez-Caso | Post-doc Researcher

Sergi Valverde | Post-doc Researcher

Adriano Bonforti | Pre-doc Researcher

Max Carbonel | Pre-doc Researcher

Núria Conde | Pre-doc Researcher

Salvador Durán | Pre-doc Researcher

Luis Seoane | Pre-doc Researcher

Ben Shirt-Ediss | Pre-doc Researcher

Eva García Ramallo | Lab Technician

RESEARCH OUTLINE

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

RESEARCH SUBLINES

1. Evolutionary innovations

We explore theoretical approaches to the origins of evolutionary innovations and major transitions in biological, artificial and technological systems. Using methods from statistical physics we explore potential scenarios for the development of innovations and the potential patterns of universality common to all these disparate classes of systems. Simulated artificial ecosystems, information technology systems, language networks and other case studies are considered.

2. Multicellularity: origins, maintenance and decay

We want to develop a general theoretical framework for the origins and development of complex multicellular systems, including early emergence through evolution (evo-devo), the elegiac of tissue organization and the role played by evolution in cancer development. We also have a theoretical/experimental research approach based on synthetic multicellularity, involving the development of synthetic, engineered cell-cell communication in order to force cells to behave as multicellular entities.

3. Emergence of complex behavior

We explore the emergence of communication, collective intelligence and language in natural and artificial systems. The main goal here is to understand the nature of the major transitions associated to the shift from single individuals to cooperative systems, as well as the emergence of a complex language as a result of interactions among words. Here we also use synthetic biology to study the potential collective behaviors arising from manipulated, single-cell bacterial communities.

4. Biological computation

We study the nature, origins and evolution of living computational systems, both natural and synthetic. Using a number of methods from complex systems theory, we want to make a map of the landscape of computational processes that can occur in nature and figure out how we can move beyond that landscape. This work has several branches, both theoretical and applied to biomedical research.

PUBLICATIONS 2012

ISI Articles

- Macía, J., Posas, F., and Solé, R. 2012. Distributed computation: the new wave of synthetic biology devices. *Trends in Biotechnology* 30: 342-349.
- Macía, J., Solé, R., and Elena, S. 2012. The causes of epistasis in genetic networks. *Evolution* 66: 586-596.
- Valverde, S., Solé, R., and Elena, S.F. 2012. Evolved Modular Epistasis in Artificial Organisms. *Artificial Life* 13: 111-115.
- Widder, S., Solé, R., and Macía, J. 2012. Evolvability of feed-forward loop architecture biases its abundance in transcription networks. *BMC Systems Biology* 6 (1): 7.

Books

- Solé, R. 2012. *Vidas sintéticas*. Tusquets Metatemas.

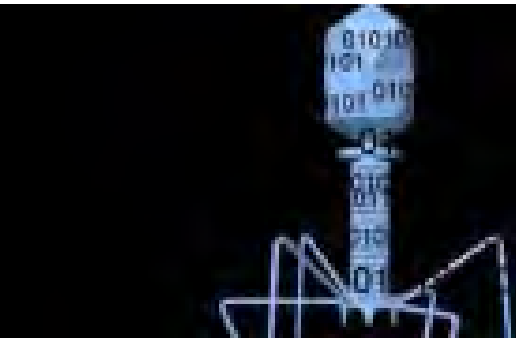


Fig.1: Complex systems

FUNDED PROJECTS

Project Title: SYNCOM

Financed by: European Research Council (ERC)

Years: 2012-2017

PIs: Ricard Solé and F. Posas

Project Title: Cellular Computation

Financed by: Fundación Marcelino Botín

Years: 2010-2016

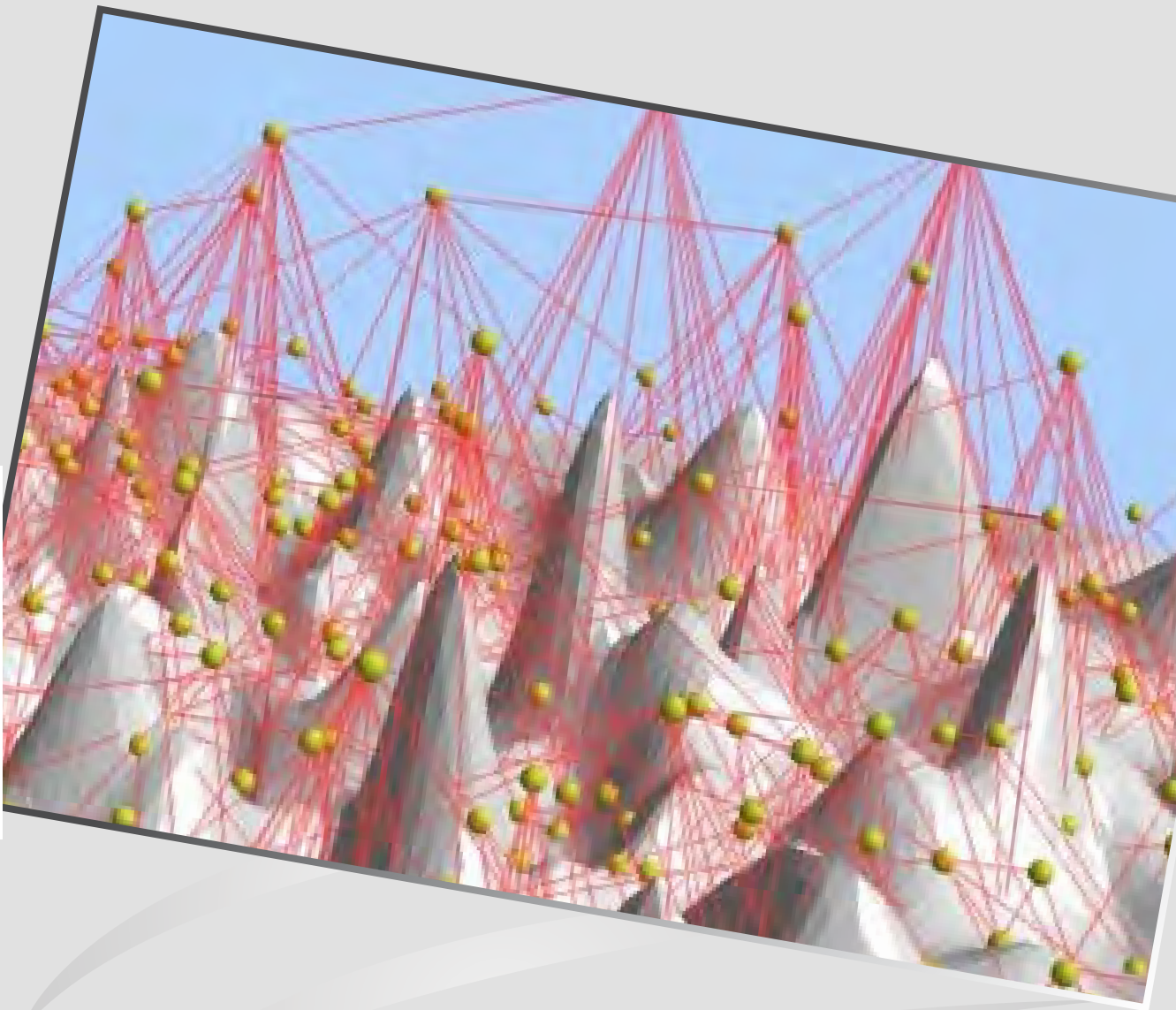
PI: Ricard Solé

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

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

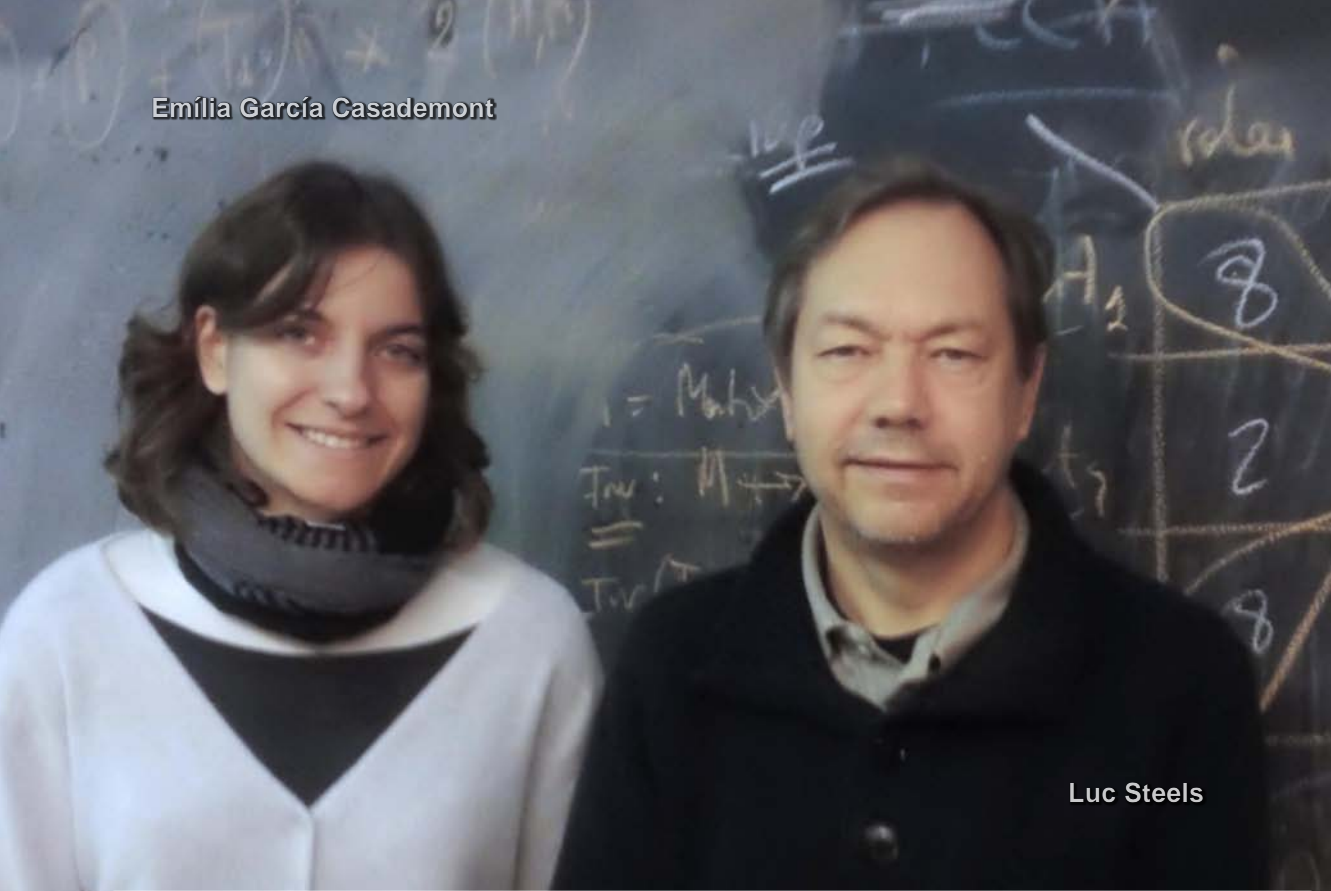
Years: 2010-2012

PI: Ricard Solé



GROUP
LANGUAGE EVOLUTION

Emília García Casademont



Luc Steels

GROUP MEMBERS

Luc Steels, Group Leader

| ICREA Researcher



Emília García Casademont | Pre-doc Researcher

RESEARCH OUTLINE

The goal of our research is to develop a theory for the origins and evolution of language. Such a theory necessarily involves three aspects: social, cultural, and biological. The social aspect should give us answers to the question why humans started to talk. The cultural aspect should answer how new language forms arise in language and keep on changing over time. The biological aspect addresses the question how the biological foundations for language may have arisen. We focus mostly on the cultural aspect, developing and testing agent-based models explaining how features of language, such as agreement systems, arise and culturally evolve.

RESEARCH SUBLINES

1. Origins and evolution of grammatical structures

Although there are a lot of data about the historical change in language, there is virtually no theory of the fundamental processes underlying this kind of evolution. We try to understand what cognitive mechanisms, interaction patterns, and collective dynamics that could explain how grammatical structures arise in human language by building agent-based models and using the hypothesis that self-organization and (linguistic) selection are the primary driving forces. We analyze the behavior of our models using the tools of complex systems science and compare the results with phenomena observed in human languages. At this point we focus in particular on the origins of agreement systems and of grammatical patterns (such as noun phrases).

2. Fluid construction grammar (FCG)

In order to do agent-based experiments in language evolution it is necessary to have a computational formalism that is capable to handle variation, flexibility and change. We are therefore working in collaboration with other research centers on the development of such a formalism. The formalism takes a construction grammar viewpoint which is more appropriate for modeling language evolution. It consists of data structures for representing linguistic knowledge and mechanisms for parsing, production, and language learning. FCG has been released in open source and is being used by a growing community: <http://www.fcg-net.org/>

3. Neural implementations of fluid construction grammar

To bridge the gap between computational models and neurobiology, we are investigating how a replicator dynamics model of the brain could potentially be used to implement the highly complex operations that Fluid Construction Grammar demands.

ISI Articles

- Steels, L. 2012. Interactions between cultural, social and biological explanations for language evolution. *Physics of Life Reviews* 9(1): 5-8.
- Van Trijp, R., and Steels, L. 2012. Multilevel alignment maintains language systematicity. *Advances in Complex Systems* 15(3/4).

Books/Book Chapters

- Beuls, K., Steels, L., and Hofer, S. 2012. The emergence of internal agreement systems. In: Steels, L. (ed.) *Experiments in Cultural Language Evolution*. John Benjamins Pub., Amsterdam, pp 233-256.
- Spranger, M., Loetzsch, M., and Steels, L. 2012. A perceptual system for language game experiments. In: Steels, L., and M. Hild (eds.) *Language Grounding in Robots*. Springer-Verlag, New York, pp 95-116.
- Spranger, M., Pauw, S., Loetzsch, M., and Steels, L. 2012. Open-ended Procedural Semantics. In: Steels, L., and M. Hild (eds.) *Language Grounding in Robots*. Springer-Verlag, New York, pp 159-178.
- Steels, L. 2012. Grounding language through evolutionary language games. In: Steels, L. and M. Hild (eds.) *Language Grounding in Robots*. Springer-Verlag, New York, pp 1-23.

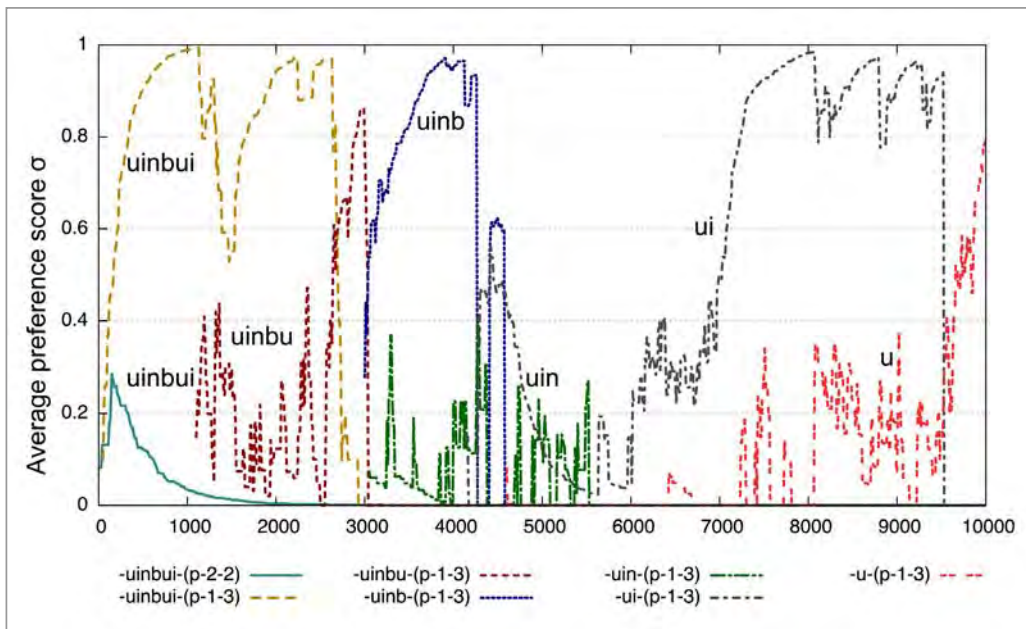


Fig. 1: Snapshot of a simulation modeling the phonological erosion of agreement markers in a population of agents.

- Steels, L. 2012. Self-organization and selection in language evolution. In: Steels, L. (ed.) *Experiments in Cultural Language Evolution*. John Benjamins, pp 1-37.
- Steels, L., De Beule, J., and Wellens, P. 2012. Fluid Construction Grammar on Real Robots. In: Steels, L. and M. Hild (eds.) *Language Grounding in Robots*. Springer-Verlag, New York, pp 201-219.
- Steels, L., and Loetzsch, M. 2012. The Grounded Naming Game. In: Steels, L. (ed) *Experiments in Cultural Language Evolution*. John Benjamins Pub., Amsterdam, pp 41-59.
- Steels, L. and M. Hild (eds.) 2012. *Language Grounding in Robots*. Springer-Verlag, New York.
- Steels, L., and Spranger, M. 2012. Emergent functional grammar for space. In: Steels, L. (ed.) *Experiments in Cultural Language Evolution*. John Benjamins Pub., Amsterdam, pp 207-232.
- Steels, L., and Spranger, M. 2012. Emergent mirror systems for body language. In: Steels, L. (ed.) *Experiments in Cultural Language Evolution*. John Benjamins Pub., Amsterdam, pp 87-109.
- Steels, L. (ed.) 2012. *Computational Issues in Fluid Construction Grammar*. LNAI 7249. Springer-Verlag, Berlin.
- Steels, L. (ed.) 2012. *Experiments in Cultural Language Evolution*. John Benjamins Pub., Amsterdam.
- Steels, L., Spranger, M., Van Trijp, R., and Hild, M. 2012. Emergent action language on real robots. In: Steels, L. and M. Hild (eds.) *Language Grounding in Robots*. Springer-Verlag, New York, pp 261-282.

Conference Publications

- Steels, L. 2012. Evolutionary Language Games as a paradigm for Integrated AI research. In: 2012 AAAI Spring Symposium, AAAI Press.

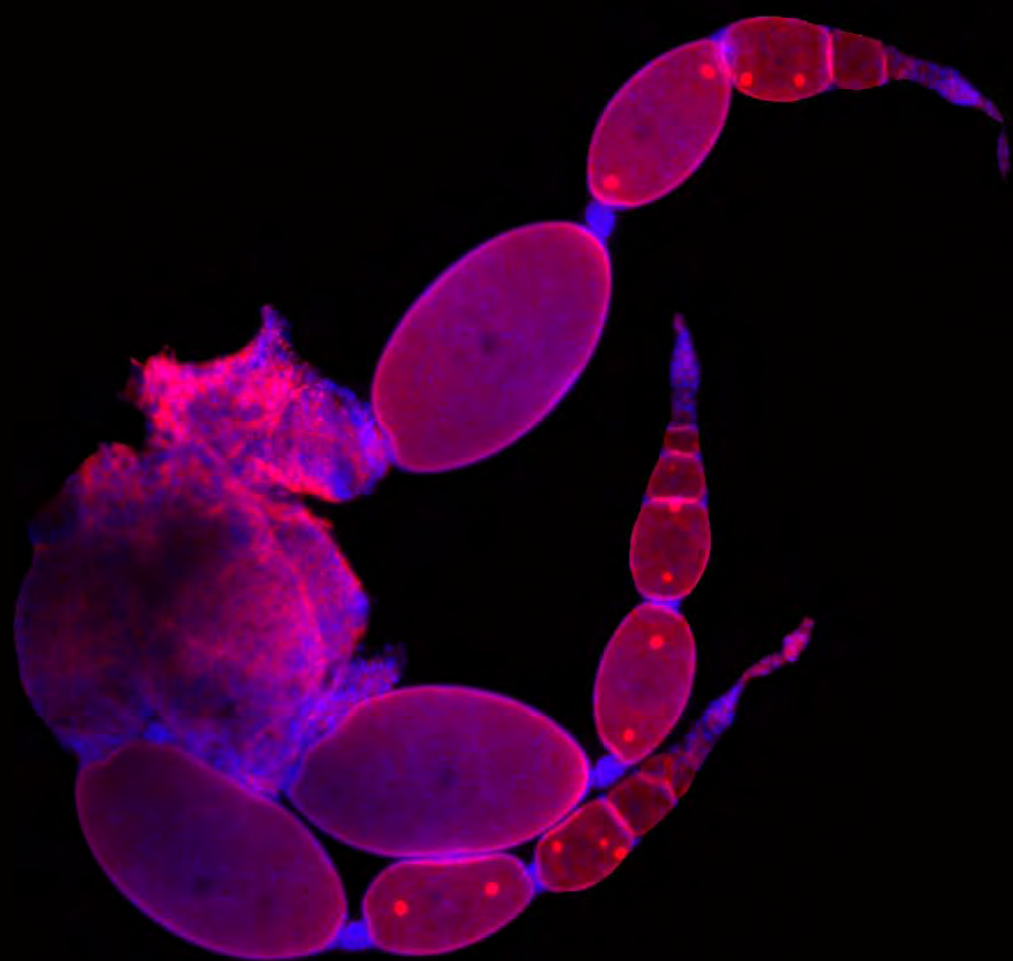
FUNDED PROJECTS

Project Title: Artificial Language Evolution for Autonomous Agents

Financed by: EU Marie Curie Integration Grant

Years: 2011-2013

PI: Luc Steels





PROGRAM

functional genomics and evolution

RESEARCH GROUPS

Evolution and developmental biology

Xavier Franch-Marro, Group Leader

Subgroups:

Morphology and signalling

| Xavier Franch-Marro, PI

Hormonal control of insect development

| David Martín, PI

Insect physiology and molecular biology

Xavier Bellés, Group Leader

Subgroups:

Evolution of insect metamorphosis

| Xavier Bellés, PI

Nutritional signals in insects

| José Luis Maestro, PI

Insect reproduction

Maria-Dolors Piulachs, Group Leader

Multicell genome

Iñaki Ruiz-Trillo, Group Leader

The synthesis of evolution, genomics and development led to the *new* field of Evolution and Development (so called EvoDevo). The aim of EvoDevo is to approach basic evolutionary questions taking into account the embryological (developmental) data but with a wider, comparative perspective. Our program goes one step forward, by combining evo-devo analyses with functional genomics approaches. The goal is to study fundamental biological questions, such as the evolution of multicellularity, development, growth, metamorphosis and oogenesis.

Most evolutionary research has been restricted to model animal systems, some of which turned out to be rather derived taxa. Our program aims at exploring new horizons by creating new data from yet neglected taxa. Thus, to address our questions, we use both model (*Drosophila melanogaster*) and non-model species (cockroaches, like *Blattella germanica*, beetles, like *Tribolium castaneum*, and unicellular eukaryotes, like *Capsaspora owczarzaki* and *Creolimax fragrantissima*). By further developing these new non-model species, we aim to generate data promising to provide new insights into these important evolutionary questions.

In the context of the IBE, this program follows a well differentiated approach since it combines both comparative data generation on a great number of taxa, and the application of a number of different methodologies, such as cell and developmental biology as well as next generation sequencing techniques.

GROUP

EVOLUTION AND DEVELOPMENTAL BIOLOGY



Josefa Cruz Rodríguez

Ferran Borràs

David Martín Casacuberta

Cristina Manjón

Neus Bota Rabassedas

Cristina Miguel Vijandi

Enric Ureña

Xavier Franch-Marro

GROUP MEMBERS

Xavier Franch-Marro, Group Leader

| Tenured Scientist, CSIC



| Subgroup: Morphology and signalling

Xavier Franch-Marro | Tenured Scientist, CSIC

Josefa Cruz Rodríguez, Post-doc | CSIC Contract

Neus Bota Rabassedas, PhD Student | FPI Scholarship, MEC

Cristina Miguel Vijandi, PhD Student | FPI Scholarship, MEC

| Subgroup: Hormonal control of insect development

David Martín Casacuberta | Tenured Scientist, CSIC

Cristina Manjón, Post-doc | CSIC Contract

Enric Ureña, PhD Student | CSIC Contract

RESEARCH OUTLINE

Evolution along Earth history has developed a great number of different organisms with a consequent incredibly variety of forms and sizes. Those morphologies are tailored during development, by modifying the expression pattern of key genes as well as by modulating hormone activation, which controls the timing of development. Thus, our main goal is to understand how changes in hormone regulation affect morphology evolution and to identify which genes and what kinds of changes in their sequences are responsible for the evolution of the mentioned morphological diversity. We address those questions comparing *Drosophila melanogaster*, *Tribolium castaneum* and *Blattella germanica* development.

RESEARCH LINES

SUBGROUP: MORPHOLOGY AND SIGNALLING

1. Tracheal system remodeling and morphogenesis

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

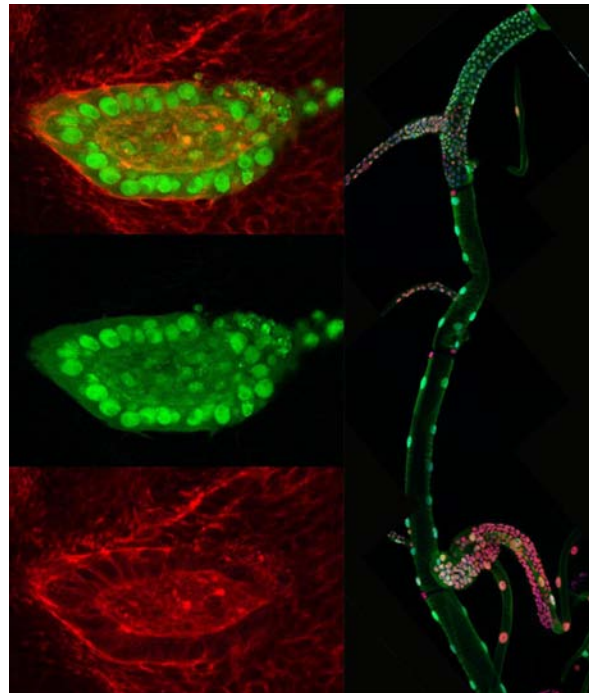


Fig. 1: Larval tracheal system of *Drosophila melanogaster*. Detail of air sac primordium. Tracheal cells are shown in green.

2. Tracheal system evolution

Tracheal system is the respiratory organ of insects. It consists of a network of tubes that transport oxygen to all the tissues. Insects present different morphology of the tracheal network depending on its habitat. For instance, we have found that *Drosophila* tracheal network presents some morphological innovations compared to the tracheal morphology of a most primitive insect such as *Tribolium*. The main goal of this project is to discover the genetic changes that have allowed the generation of those morphological adaptations along evolution.

4. Wingless signaling in size control and evolution

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

SUBGROUP: HORMONAL CONTROL OF INSECT DEVELOPMENT

1. Molecular analysis of ecdysteroid and juvenile hormone action in insect development and metamorphosis

All immature animals undergo remarkable morphological and physiological changes to become mature adults. In winged insects, metamorphic changes are either limited to a few tissues (hemimetaboly) or involve a dramatic wholesale reorganization of most tissues and organs (holometaboly). In both cases,

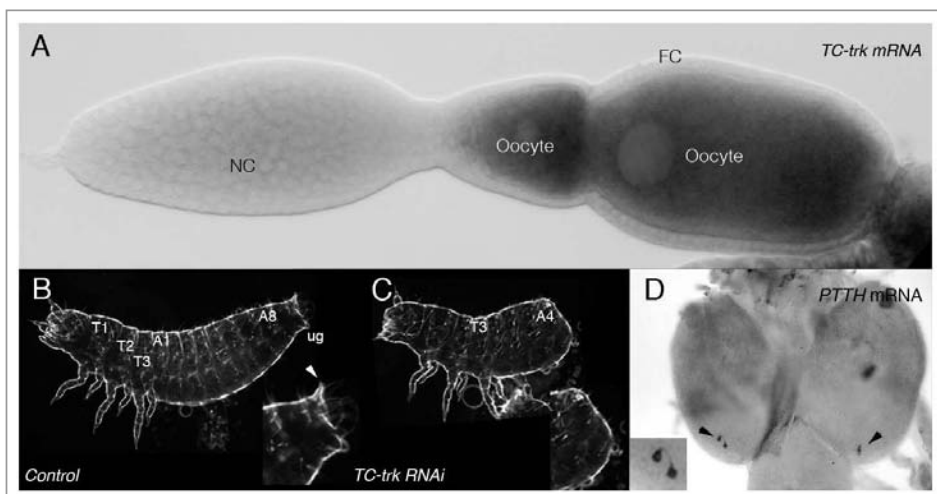


Fig. 2: *trk* and *ptth* in *Tribolium castaneum*. (A) *Tc-trk* mRNA is detected in the developing oocyte; no expression is found in nurse (NC) and follicular cells (FC); (B) *Tribolium* larva from control injection; (C) *Tc-trk* RNAi injections impair terminal patterning; (D) *Tc-ptth* mRNA is detected in two pairs of neurons in the larval brain.

however, two hormones control all the developmental changes associated to the metamorphic process. The steroid hormone 20-hydroxyecdysone (20E) triggers major developmental transitions through a genetic cascade of transcription factors that belong to the Nuclear Hormone Receptor superfamily (NRs). Furthermore, Juvenile Hormone (JH), the other hormone of paramount importance in development, prevents metamorphosis by coordinating multiple 20E-dependent developmental and physiological processes. The main goal of this project is the molecular characterization of the regulatory role of both, 20E and JH, in the metamorphosis of the hemimetabolous insect *Blattella germanica* (using RNAi in vivo and parental RNAi procedures) and the holometabolous insects *Tribolium castaneum* and *Drosophila melanogaster* (mutational analysis). These studies have already demonstrated critical roles of several transcription factors on ecdysteroid production, programmed cell death, tissue growth and morphogenesis, ovary follicle proliferation and molting behaviour in both types of insects.

2. Embryonic development in hemimetabolous short germ band insects

The main goal of this project is to characterize the major morphogenetic events in direct-developing hemimetabolous insects, which mainly occur during early-embryogenesis. For that, we are analyzing the role of each 20E-dependent NR on the morphogenetic events during the embryogenesis of the insect model *B. germanica*. This project will provide a valuable model to understand the molecular basis for how shifts in the hormonal context can lead to the creation of different body forms.

3. Control of developmentally regulated programmed cell death by ecdysteroids 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 anti-apoptotic role of JH upon this process.

4. Sumoylation in insect 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, Spain), 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 2012

ISI Articles

- Chaumot, A., Da Lage, J.L., Maestro, O., Martín, D., Iwema, T., Brunet, F., Bellés, X., Laudet, V., and Bonneton, F. 2012. Molecular adaptation and resilience of the insect's nuclear receptor USP. *BMC Evolutionary Biology* 12 (1): 199. doi: 10.1186/1471-2148-12-199.
- Grillo, M., Furriols, M., de Miguel, C., Franch-Marro, X., and Casanova, J. 2012. Conserved and divergent elements in Torso RTK activation in *Drosophila* development. *Scientific Reports* 2, 762; doi: 10.1038/srep00762.
- Mané-Padrós, D., Borràs-Castells, F., Bellés, X., and Martín, D. 2012. Nuclear receptor HR4 plays an essential role in the ecdysteroid-triggered gene cascade in the development of the hemimetabolous insect *Blattella germanica*. *Molecular and Cellular Endocrinology* 348: 322-330.

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 (BFU2009-08748)

Years: 2010-2013

PI: Xavier Franch-Marro

Project Title: Bases moleculares de la acción de los ecdisteroides y de la hormona juvenil en el desarrollo de los insectos. El papel de los receptores nucleares.

Financed by: Ministerio de Ciencia e Innovación (BFU2009-10571)

Years: 2010-2013

PI: David Martín Casacuberta

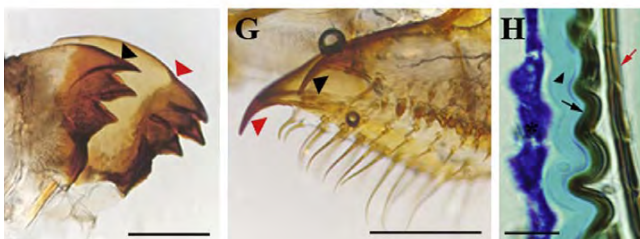
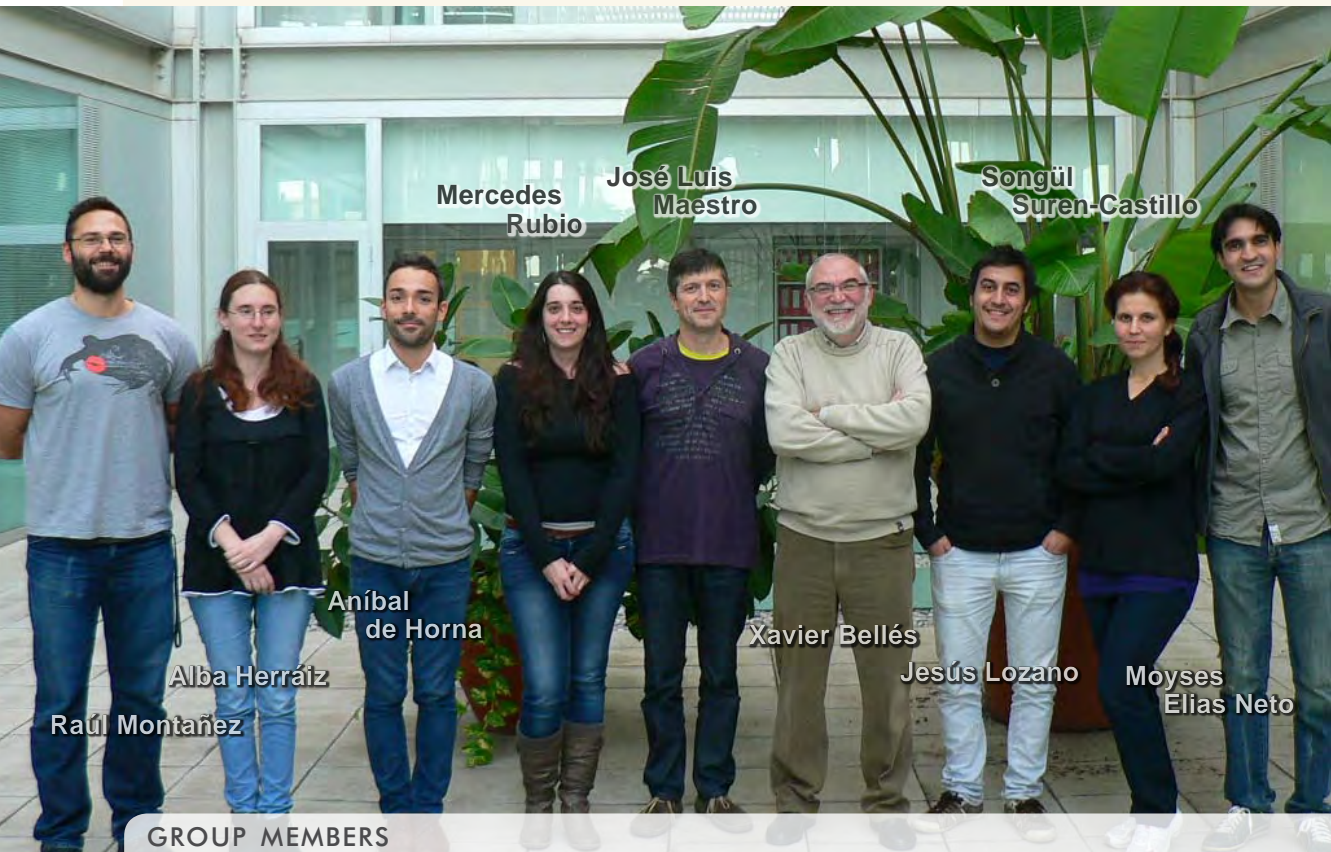


Fig. 3: Silencing of *BgHR4* by RNAi in vivo in sixth instar female nymphs of *B. germanica*. *dsBghR4-1*-treated specimen of the same age arresting development without molting. The arrested specimens show duplication of ectodermal-derived structures (nymphal structures indicated with a red arrowhead, and adult structures with black arrowheads), like double mandibles and double lacinia.

GROUP

INSECT PHYSIOLOGY AND MOLECULAR BIOLOGY



Xavier Bellés, Group Leader

| Research Professor, CSIC



| Subgroup: Evolution of insect metamorphosis

Xavier Bellés | Research Professor, CSIC

Moyses Elias Neto, Post-doc | FAPESP Grant, São Paulo, Brazil

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

Alba Herráiz, PhD Student | Scholarship CSIC (JAE Program)

Jesús Lozano, PhD Student | Scholarship MICINN

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

Aníbal de Horna, Lab Manager | Bioinformatician, Project Contract

| Subgroup: Nutritional signals in insects

José Luis Maestro | Tenured Scientist, CSIC

Songül Suren-Castillo, Post-doc | CSIC Contract (JAE Program) and Project Contract

RESEARCH OUTLINE

Historically, our goals and interests have been traditionally diverse, embracing a number of subjects around the physiology of the insect. In general, research has focused on physiological processes regulated by hormones. Therefore, we have studied the biochemical and regulatory aspects of the hormones themselves (juvenile hormone, ecdysteroids and regulatory peptides), and also the processes dependent on them, like metamorphosis and vitellogenesis (especially in relation to juvenile hormone and ecdysteroids), molting, oogenesis and chorion formation (in relation to ecdysteroids), and food intake and growth (in relation to peptides and proteins).

At present, most of our research focuses on insect metamorphosis, a research line headed by Xavier Bellés, and on the study of the physiological and developmental effects of nutritional signals, which is headed by José Luis Maestro. We are interested in the regulation of these processes from a mechanistic point of view and from an evolutionary perspective. As most information has been obtained in highly modified, holometabolous species (mainly in the fly *Drosophila melanogaster*), we concentrate on the cockroach *Blattella germanica*, a phylogenetically basal, hemimetabolous species.



Fig. 1: Last nymphal instar, precocious adult and normal adult of *Blattella germanica*, the precocious adult results from depleting *Met* expression in penultimate nymphal instar.

RESEARCH LINES

SUBGROUP: EVOLUTION OF INSECT METAMORPHOSIS

We aim to elucidate the mechanisms regulating metamorphosis in *B. germanica* and then comparing them with those operating in holometabolous species. The idea is to infer the evolutionary history underlying the transition from hemimetaboly to holometaboly. During 2012, we have been working in the following four main subjects.

1. Minimal models of metamorphosis

We concentrated in two metamorphic processes, i) the formation of the wing, which is also important to understand the origin of metamorphosis, and ii) the

formation of the male tergal gland, which is a complex morphologic structure that forms during the imaginal molt in the tergites 7 and 8 of males. We have approached these two processes by monitoring the morphological changes, but also at molecular level, by comparing transcriptomes of the respective tissues in metamorphic and non-metamorphic transitions. A number of transcripts that are specific of the last instar nymph have been identified as «metamorphosis triggers».

2. Transcription factors and hormonal signaling

Metamorphosis is mainly regulated by two hormone types: ecdysteroids and juvenile hormones. We are interested in the transcription factors involved in the signaling pathways elicited by both hormone types, considering that there are not two separate pathways, but an intricate network of interaction between JH- and ecdysone-associated factors. During 2012, we have unveiled the functions of a number of these transcription factors, like Krüppel homolog 1 and Broad complex, using RNAi techniques.

3. Small RNAs

Our hypothesis is that miRNAs play a modulatory role in the shift from juvenile to adult developmental programs. Using miRNA libraries from penultimate and last instar nymph and carrying out functional studies with anti-miR molecules and miR mimics, we have characterized a number of miRNAs that play significant roles in the transition from nymph to adult. Moreover, we have also investigated a number of aspects on the biochemical machinery involved in the RNAi process.

4. Complex networks

Metamorphosis involves complex networks of gene regulation, and the idea is to reduce this complexity to graphs capturing the main properties of these networks. During 2012, we have focused on networks of interaction mRNA-miRNA, comparing metamorphic and non-metamorphic transitions in holometabolans and hemimetabolans species.

SUBGROUP: NUTRITIONAL SIGNALS IN INSECTS

Our research is directed to understand how nutritional status is detected by the insect and how this status signals to the different processes that depend on it, with a special focus on reproduction. All Metazoans have two well conserved nutritional signaling pathways, the *Insulin Receptor* and the *target of rapamycin* (TOR) pathways. They are involved in the detection of the nutritional status and the activation of really crucial processes, such as growth, cellular proliferation, longevity, cancer or reproduction. During 2012, we have been mainly working in the following subjects.

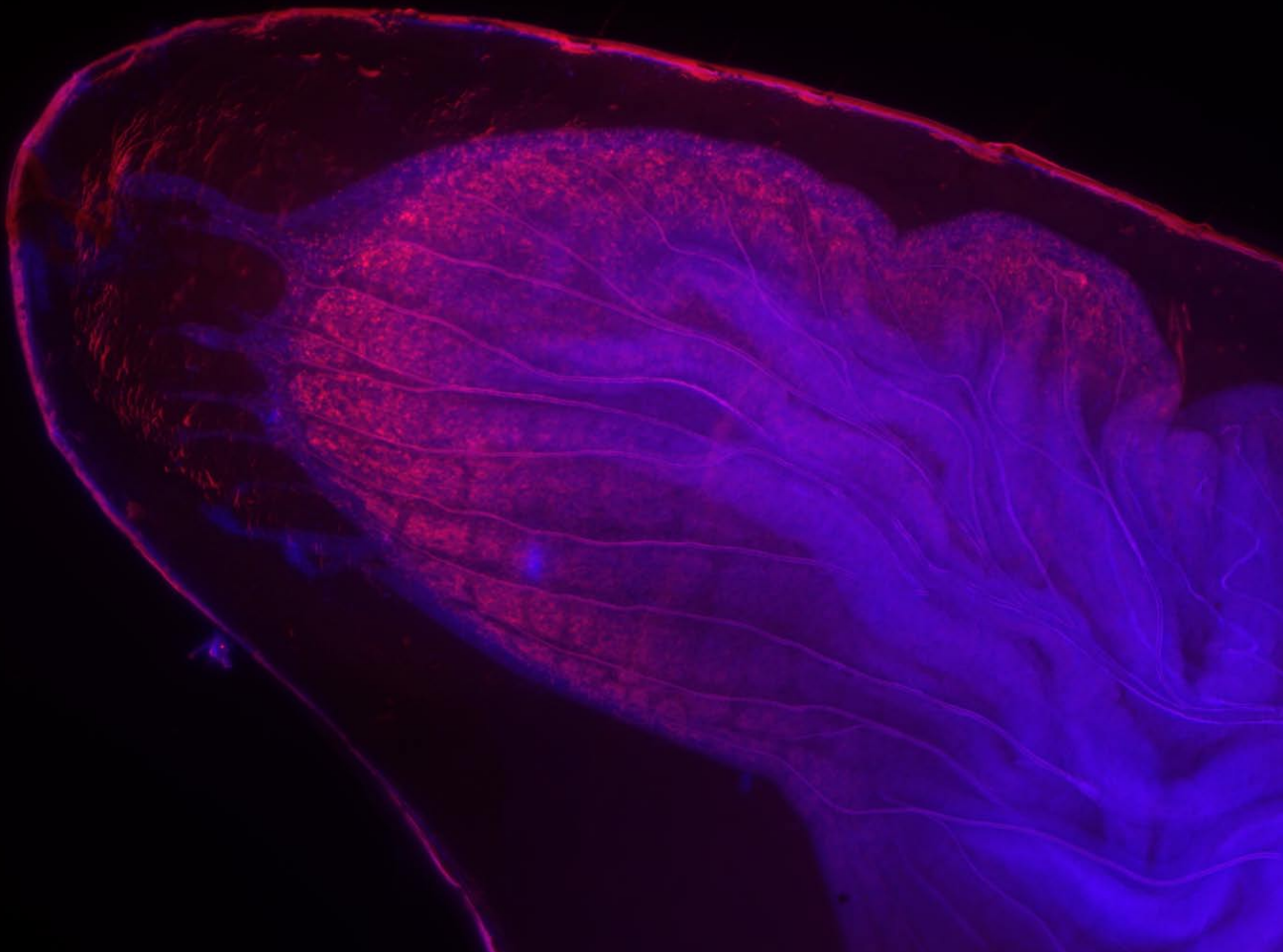
1. FoxO transcription factor

FoxO proteins are evolutionary conserved transcription factors which have been described as key players of the Insulin Receptor pathway. During 2012 we have demonstrated that FoxO is involved in regulating different processes related to the response to starvation, including among these processes the hormonal regulation of reproduction, different metabolic responses, oxidative stress resistance and longevity.

2. Insulin receptor and insulins

Our second research objective has been to study the activity of the Insulin Receptor itself and the role of the insect Insulin-like peptides (ILPs). During 2012 we have continued our studies on Insulin Receptor function, its relation with the TOR pathway and its role in regulating reproduction, growth and longevity. In addition, we have elucidated the sequence of five different ILPs in our cockroach model, and have started a series of experiments in order to analyze their implication in the juvenile hormone signaling.

Fig. 2: Developing hind wing of a specimen of *Blattella germanica* in last nymphal instar where the expression of *Broad complex* had been depleted. Notice the absence of growth all around the edge. Double labeling EdU (red spots) and DAPI (blue color).



PUBLICATIONS 2012

ISI Articles

- Boher, F., Trefault, N., Piulachs, M.D., Bellés, X., Godoy-Herrera, R., and Bozinovic, F. 2012. Biogeographic origin and thermal acclimation interact to determine survival and hsp90 expression in *Drosophila* species submitted to thermal stress. *Comparative Biochemistry and Physiology, Part A* 162: 391-396.
- Chaumot, A., Da Lage, J.L., Maestro, O., Martín, D., Iwema, T., Brunet, F., Bellés, X., Laudet, V., and Bonneton, F. 2012. Molecular adaptation and resilience of the insect's nuclear receptor USP. *BMC Evolutionary Biology* 12 (1): 199. doi: 10.1186/1471-2148-12-199.
- Huang, J.H., Bellés, X., and Lee, H.J. 2012. Functional characterization of hypertrehalosemic hormone receptor in relation to hemolymph trehalose and to oxidative stress in the cockroach *Blattella germanica*. *Frontiers in Experimental Endocrinology* 2: 114. doi: 10.3389/fendo.2011.00114.
- Lozano, J., Gómez-Orte, E., Lee, H.J., and Bellés, X. 2012. Super-induction of Dicer-2 expression by alien double-stranded RNAs. An evolutionary ancient response to viral infection? *Development, Genes and Evolution* 222: 229-235.
- Mané-Padrós, D., Borràs-Castells, F., Bellés, X., and Martín, D. 2012. Nuclear receptor HR4 plays an essential role in the ecdysteroid-triggered gene cascade in the development of the hemimetabolous insect *Blattella germanica*. *Molecular and Cellular Endocrinology* 348: 322-330.
- Rubio, M., de Horna, A., and Bellés, X. 2012. MicroRNAs in metamorphic and non-metamorphic transitions in hemimetabolous insect metamorphosis. *BMC Genomics* 13: 386. doi: 10.1186/1471-2164-13-386.
- Süren-Castillo, S., Abrisqueta, M., and Maestro, J.L. 2012. FoxO inhibits juvenile hormone biosynthesis and vitellogenin production in the German cockroach. *Insect Biochemistry and Molecular Biology* 42: 491-498.

Other Publications

- Bellés, X. 2012. *Ptinus bertranpetiti*, a new species of spider beetle from Socotra Island (Coleoptera: Ptinidae). *Acta Entomologica Musei Nationalis Pragae* 52 (supplementum 2): 219-222.
- Bellés, X. 2012. Una mirada a la biología de la conservación desde la biología evolutiva. *Lychnos* 9: 57-61.

FUNDED PROJECTS

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

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

Years: 2009-2013

PI: Xavier Bellés

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

Financed by: Ministerio de Ciencia e Innovación. Plan Nacional.

Year: 2011-2013

PI: José Luis Maestro

Project Title: Insect Control with RNAi
Financed by: CSIC and the National Taiwan University
(2010TW0019, Formosa program)
Years: 2011-2013
PI: Xavier Bellés

Project Title: Global change and physiological diversity
Financed by: International Laboratory of Global Change (LINCGlobal),
CSIC (Spain)-PUC (Chile)
Years: 2009-2012
PI: Xavier Bellés and Francisco Bozinovic

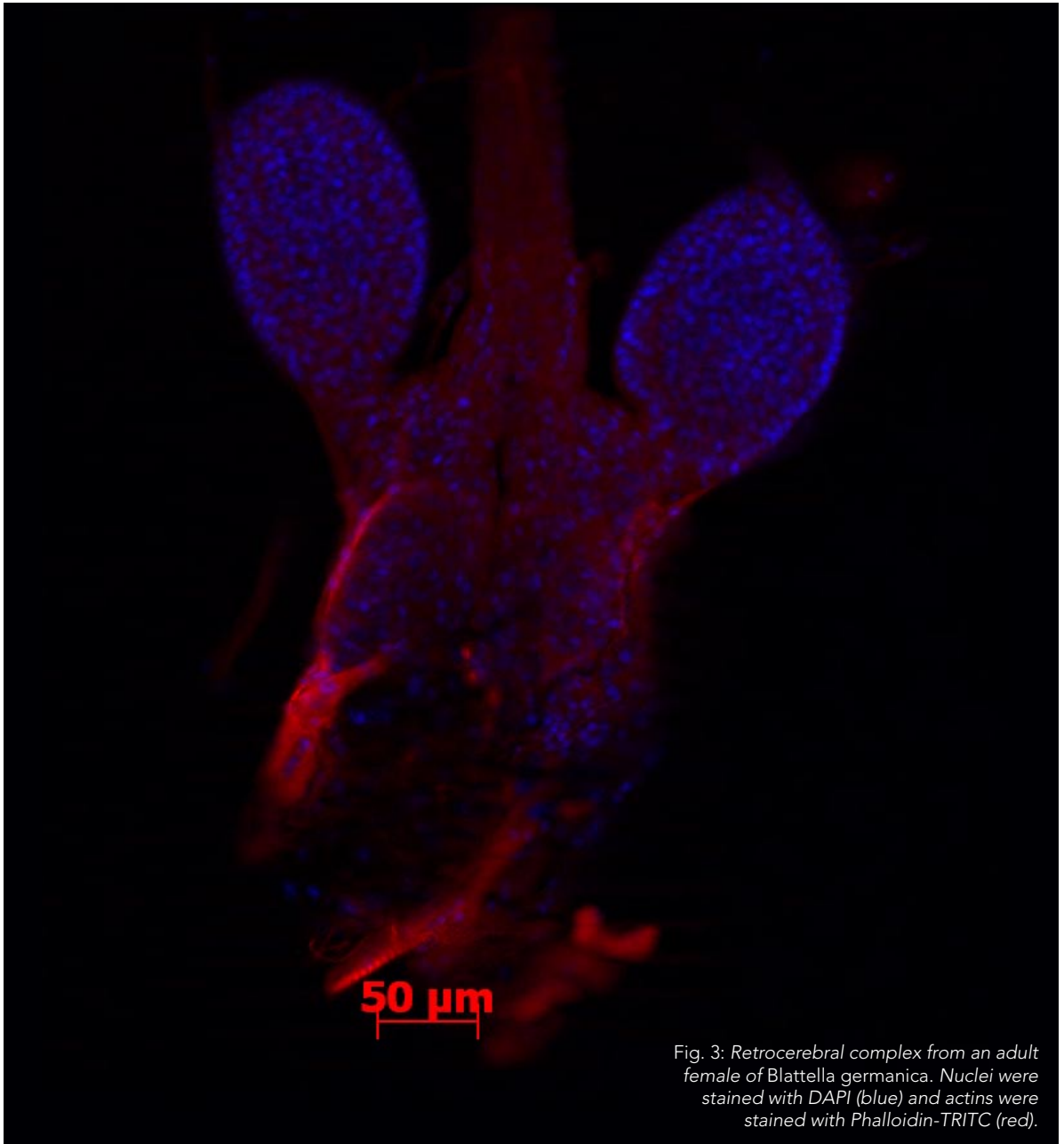


Fig. 3: Retrocerebral complex from an adult female of *Blattella germanica*. Nuclei were stained with DAPI (blue) and actins were stained with Phalloidin-TRITC (red).

GROUP

INSECT REPRODUCTION



Carlos Vásquez

Nashwa Elshaer

Maria-Dolors
Piulachs

Alba Herráiz

Paula Irlles

Aníbal
de Horna

GROUP MEMBERS

Maria-Dolors Piulachs, Group Leader

| Research Scientist, CSIC



Paula Irlles, Post-doc | CONICYT Fellowship Contract

Nashwa Elshaer, PhD Student | JAEPRE-CSIC Fellowship

Alba Herráiz, PhD Student | JAEPRE-CSIC Fellowship · LINCGlobal

Carlos Vásquez, PhD Student | CONICYT Fellowship Contract

Aníbal de Horna, Bioinformatics Support | CSIC Project Contract

RESEARCH OUTLINE

Our long term objective is to elucidate how is insect oogenesis regulated, considering the structural diversity of ovary types and their respective evolutionary history. Two main types of ovaries can be distinguished in insects: panoistic and meroistic. The panoistic type is more generalized in phylogenetically basal species, whereas the meroistic type predominates in more modified insect groups. Our general objective is to understand the transition from panoistic to meroistic type of ovaries during evolution, and to approach this end objective it is crucial to gather molecular information in both ovarian types. Most of the studies on insect oogenesis published to date have been carried out on *Drosophila melanogaster*, a species with meroistic ovaries. Thus, our project is based on a species with panoistic ovaries: the cockroach *Blattella germanica*. Our group has already obtained a great deal of information related to oogenesis and reproduction in this species, and during the last year our research was addressed to study the genes that determine oocyte polarization, and those that control follicular cell proliferation.

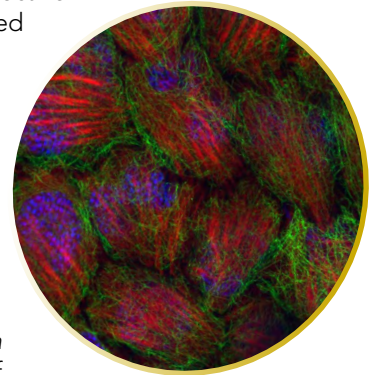


Fig. 1: Actins in the follicular cells surrounding the oocyte of *Blattella germanica*. Actins were stained with Phalloidin-TRITC (red), tubulins were revealed using a β -tubulin-anti-mouse (green) and nuclei were stained with DAPI (blue).

RESEARCH LINES

During this year our research has focused on two main subjects, using *Blattella germanica* as experimental subject:

1. Genes that in more modified species determine oocyte polarization

To develop this objective we chose a number of genes involved in this function that have been well studied in *Drosophila melanogaster*, like *capicua*, *oskar* and *EGFR*, among others. We studied their expression, localization and function in a panoistic ovary and, with this information, we established when and where the corresponding mRNAs are transcribed and in which moment the oocyte polarization occurs. The final idea is to compare these results with those obtained in *D. melanogaster* in order to test whether the function of these genes are conserved or have changed in the transition from the panoistic to the meroistic ovarian type.

2. Regulation of cell proliferation during oogenesis

Control of cell proliferation is really important to control size and form in different organs, and a number of proteins belonging to the Hippo pathway play key roles in this process. Most of these proteins are conserved through the animal evolution, from worms to humans. To understand the regulation of the Hippo pathway it is important to study not only how do cells proliferate, but also how do they avoid overproliferation, which could lead to tumorigenesis. Our objective is to study the Hippo pathway in vivo, using our cockroach model. We aim to identify their components, describe their precise function using the RNA interference (RNAi) methodologies, and determine the possible regulatory role of miRNAs in the modulation of the different components of the pathway.

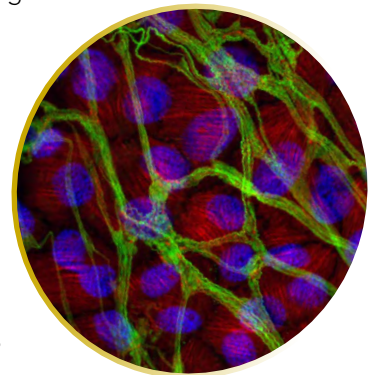


Fig. 2: Follicular epithelium from an oocyte of *Blattella germanica* with lining fragments of the tunica propria. Actins were stained with Phalloidin-TRITC (red), tubulins were revealed using a β -tubulin-anti-mouse (green) and nuclei were stained with DAPI (blue).

PUBLICATIONS 2012

ISI Articles

- Boher, F., Trefault, N., Piulachs, M.D., Bellés, X., Godoy-Herrera, R., and Bozinovic, F. 2012. Biogeographic origin and thermal acclimation interact to determine survival and hsp90 expression in *Drosophila* species submitted to thermal stress. *Comparative Biochemistry and Physiology, Part A* 162: 391-396.
- Tanaka, E.D., and Piulachs, M.D. 2012. Dicer and miRNAs in *Blattella germanica* oogenesis. *Biology of the Cell* 104: 1-10 (including the cover page).

Book Chapters

- Bellés, X., Cristino, A.S., Tanaka, E.D., Rubio, M., and Piulachs, M.D. 2012. microRNAs in insects. From molecular mechanisms to biological roles. In: *Insect Molecular Biology and Biochemistry* (Lawrence I. Gilbert, ed.) pp 30-56. Pergamon Press.

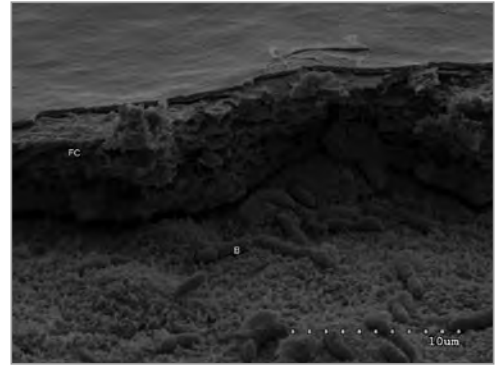


Fig. 3: Scanning electron microscopy image showing the symbiont bacteroid *Blattabacterium cuenoti* in a *Blattella germanica* oocyte. The bacteroid (B) occupies the space between follicular cells (FC) and oocyte membrane.

FUNDED PROJECTS

Project Title: Searching the origin of oocyte polarization in insects

Financed by: MINECO

Years: 2012-2014

PI: Maria-Dolors Piulachs

Project Title: Global change and physiological diversity

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

Years: 2009-2012

PIs: Xavier Bellés and Francisco Bozinovic

Project Title: Insect pest control with RNAi

Financed by: CSIC and NSC, Taiwan

Years: 2011-2012

PIs: Xavier Bellés and How-Jing Lee

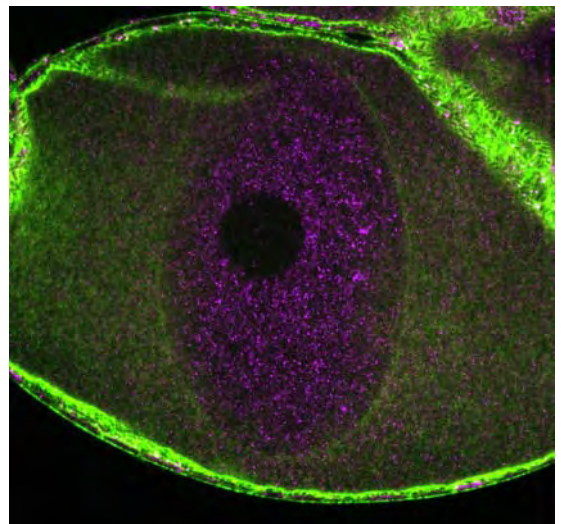


Fig. 4: Optical section of a *Blattella germanica* oocyte showing big nucleus (violet). Actins were stained with Phalloidin-TRITC (green).

GROUP

MULTICELL GENOME



GROUP MEMBERS

Iñaki Ruiz-Trillo, Group Leader

| ICREA Researcher



Javier del Campo | Post-doc Researcher

Núria Sánchez | Post-doc Researcher

Hiroshi Suga | Post-doc Researcher

Xavier Grau | PhD Student

Alex de Mendoza | PhD Student

Arnau Sebé-Pedrós | PhD Student

Guifré Torruella | PhD Student

Helena Parra | Master Student

Meritxell Antó | Research Technician

Maria José Barberà | Research Technician

RESEARCH OUTLINE

We try to understand how unicellular organisms became multicellular. Specifically, we focus on the origin of multicellular animals or metazoans. To this end, we are obtaining the genomes of animal's closest unicellular relatives in order to perform comparative and functional genomics.

RESEARCH LINES

1. Biodiversity and molecular ecology of opisthokonts

The real diversity of opisthokonts remains unknown since most protists have never been cultured. Thus, if we want to understand the real biodiversity of opisthokonts we need to analyze uncultured organisms. To this end, we are analyzing molecular data from environmental samplings to have a better idea of the real diversity of the different opisthokont lineages. If possible, we will try to isolate and culture novel protists that may eventually illuminate our understanding on the protist diversity among unicellular relatives of Metazoa and Fungi.

2. Comparative genomics to unravel the Metazoan «Genetic Starter Kit»

Our goal is to elucidate the evolutionary history of genes that are key for animal development and multicellularity. To this aim, we are part of the UNICORN (UNICellular Opisthokonts Research iNitiative) initiative: an international and multi-taxon genome project recently funded by NHGRI (National Institute for Human Genome Research), which aims to gain insights into how multicellularity first evolved in both animals and fungi. UNICORN, through the Broad Institute, will obtain the genome sequence from several of the closest unicellular relatives of both animals and fungi (see Multicellularity Project at Broad). The idea is to perform comparative genomic analyses to unravel the genome structure and gene composition of the last common unicellular ancestor that gave rise to Metazoa.

We have recently shown that the filasterean amoeboid *Capsaspora owczarzaki*, a close unicellular relative of Metazoa, expresses several genes that are required to metazoan development, such as protein tyrosine kinases, integrins and several transcription factors (i.e, T-box genes).

3. Unraveling the ancestral function of genes relevant to animal multicellularity

The aim is to unravel the ancestral function of the cell-signaling and cell-adhesion genes that are crucial for animal development. By comparative genomic analyses we have identified some homologs of genes relevant to animal multicellularity and development (e.g. integrins, receptor tyrosine kinases, T-box genes) in *Capsaspora*. We want to know what roles are those genes playing in the unicellular *Capsaspora* and how these genes were later on co-opted to the new functions in metazoans. By elucidating the «ancestral function» of those genes, we will provide significant insights into the role that cell-signaling and cell-adhesion genes played in the origin of Metazoa.

To make this happen we are currently working in developing transgenesis protocols in the filasterean *Capsaspora owczarzaki* and the ichthyosporean *Creolimax fragrantissima*.

4. Phylogenomics

If we want to approach the evolution of multicellular animals, we need a robust phylogenetic framework of the opisthokonts (i.e., the clade that comprises Metazoa, Fungi and their closest unicellular lineages). Thus, among our goals is to obtain new molecular data in order to perform phylogenetic and phylogenomic analyses to further improve the opisthokont (or the eukaryote) tree of life. For example, using single-copy protein domains rather than genes, we have recently shown that apusozoans (and not the amoebozoans) are the sister-group to the opisthokonts and that the ichthyosporeans are the sister-group to a clade formed by filastereans, choanoflagellates and metazoans.



Fig. 1: *Protist Evolution.*

PUBLICATIONS 2012

ISI Articles

- Schultheiss, K.P., Suga, H., Ruiz-Trillo, I. and Miller, W.T. 2012. Lack of Csk-mediated negative regulation in a unicellular Src kinase. *Biochemistry* 51 (41): 8267-8277.
- Sebé-Pedrós, A., Zheng, Y., Ruiz-Trillo, I., and Duojsia, P. 2012. Premetazoan origin of the Hippo signaling pathway. 2012. *Cell Reports* 1 (1): 13-20. doi: 10.1016/j.cel-rep.2011.11.004.
- Shadwick, J.D.L. and Ruiz-Trillo, I. 2012. A genomic survey shows that the haloarchaeal type Tyrosyl-Synthetase is not a Synapomorphy of Opisthokonts. *European Journal of Protistology* 48 (1): 89-93.
- Suga, H., Dacre, M., de Mendoza, A., Shalchian-Tabrizi, K., Manning, G., and Ruiz-Trillo, I. 2012. Genomic Survey of Premetazoans Shows Deep Conservation of Cytoplasmic Tyrosine Kinases and Multiple Radiations of Receptor Tyrosine Kinases. *Science Signaling* 5 (222), ra35. doi: 10.1126/scisignal.2002733.
- Torruella, G., Derelle, R., Paps, J., Lang, B.F., Roger, A.J., Shalchian-Tabrizi, K., and Ruiz-Trillo, I. 2012. Phylogenetic relationships within the Opisthokonta based on phylogenomic analyses of conserved single copy protein domains. *Molecular Biology and Evolution* 29 (2): 531-544.

Book Chapters

- Riutort, M., Paps, J., and Ruiz-Trillo, I. 2012. Bilaterales. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 188-195.
- Rodríguez-Ezpeleta, N., Moreira, D., and Ruiz-Trillo, I. 2012. Amoebozoos, Cromalveolados, Rizarios y Excavados. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 42-55.
- Ruiz-Trillo, I. Eucariotas. 2012. In: Vargas, P., and Zardoya, R. (eds.) *El árbol de la vida: sistemática y evolución de los seres vivos*. Madrid, pp 34-41.

FUNDED PROJECTS

Project Title: A comparative genomic analysis into the origin of metazoan multicellularity

Financed by: ERC (European Research Council)

Years: 2008-2013

PI: Iñaki Ruiz-Trillo


Project Title: El origen del reino animal: un análisis genómico, filogenómico y de biodiversidad de los linajes unicelulares más cercanos a los animales

Financed by: Ministerio de Ciencia e Innovación

Years: 2012-2014

PI: Iñaki Ruiz-Trillo





PROGRAM population genetics

RESEARCH GROUPS

Evolutionary population genetics

Elena Bosch, Group Leader

Evolutionary systems biology

Jaume Bertranpetit, Group Leader

Genomics of individuality

Francesc Calafell, Group Leader

Human genome: demography and adaptation

David Comas, Group Leader

Subgroups:

Human genome diversity

| *David Comas, PI*

MicroRNAs in human adaptation and disease

| *Yolanda Espinosa-Parrilla, PI*

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

GROUP

EVOLUTIONARY POPULATION GENETICS



Johannes Engelken

Nino Spataro

Elena Bosch

Elena Carnero

GROUP MEMBERS

Elena Bosh, Group Leader

| Associate Professor, UPF



Johannes Engelken, Post-doc | Volkswagenstiftung Scholarship, Germany

Elena Carnero, PhD Student | UPF Teaching Scholarship

Nino Spataro, PhD Student | UPF Scholarship

RESEARCH OUTLINE

Our research focuses on investigating different aspects of human genetic diversity. In particular, we are interested in: (i) human 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; and (ii) the architecture of the genetic predisposition to complex disease. The search for genetic signatures of selection is pursued at different levels using comparative data and 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. As for complex disease, we believe that the application of population genetic models can help in unraveling the genetic contribution to them.

RESEARCH LINES

1. 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.



Fig. 1: 454 target enrichment resequencing and 1000 Genome data comparison.

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

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

3. 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.

4. Rare variants in Parkinson's disease

Our working hypothesis is that an excess of rare variants may indicate the involvement of a gene in a complex disease such as PD. Using resequencing data and adapting classical evolutionary tests we will evaluate the possible deviations of the spectrum of allele frequencies between cases and controls in individual genes, gene pathways and in particular regulatory regions.

PUBLICATIONS 2012

ISI Articles

- Carnero-Montoro, E., Bonet, L., Engelken, J., Bielig, T., Martínez-Florensa, M., Lozano, F., and Bosch, E. 2012. Evolutionary and functional evidence for positive selection at the human *CD5* immune receptor gene. *Molecular Biology and Evolution* 29 (2): 811-823.

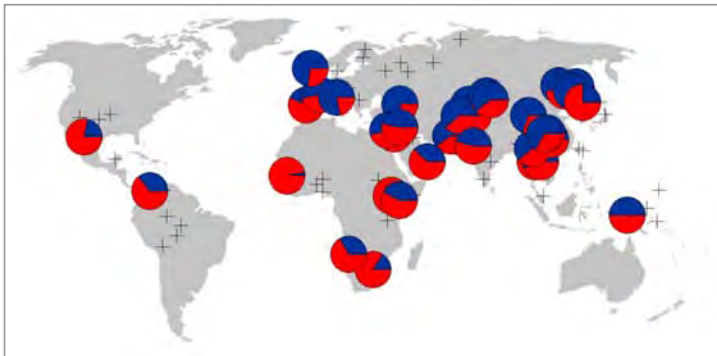


Fig. 2: Worldwide allele frequency distribution for SNP of interest.

FUNDED PROJECTS

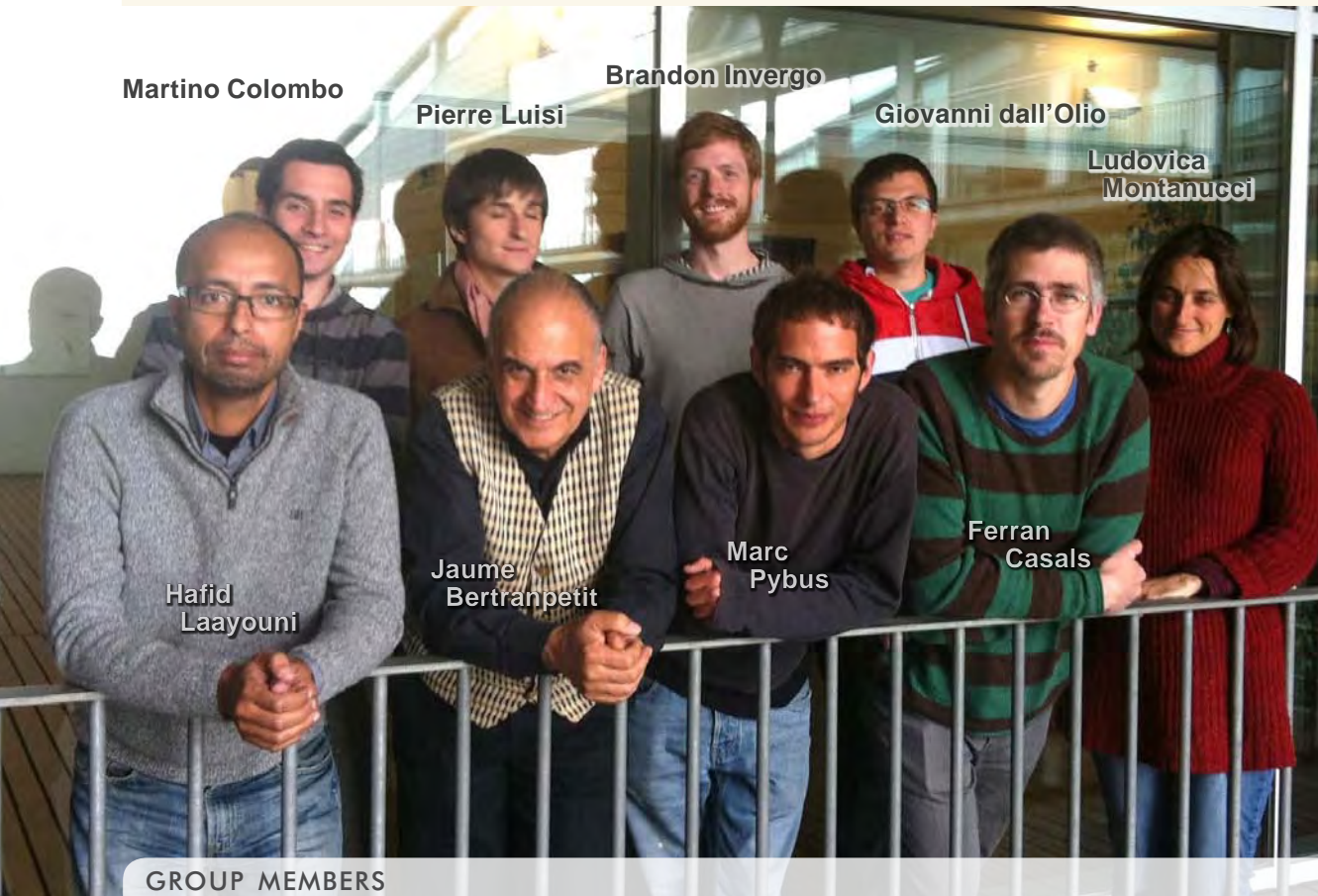
Project Title: Grup de Recerca Consolidat-SGR
Financed by: Generalitat de Catalunya (2009 SGR-1101)
Years: 2009-2013
PI: Jaume Bertranpetit

Project Title: Local Adaptation of Modern Humans to Micronutrient Deficiencies
Financed by: Volkswagenstiftung (Az: I/85 198)
Years: 2010-2012
PIs: Elena Bosch and Mark Stoneking

Project Title: Variantes genéticas raras en la enfermedad de Parkinson: aproximación evolutiva y resecuenciación de alto rendimiento
Financed by: Subdirección General de Proyectos de Investigación (SAF2011-29239)
Years: 2012-2014
PI: Elena Bosch

GROUP

EVOLUTIONARY SYSTEMS BIOLOGY



Martino Colombo

Brandon Invergo

Pierre Luisi

Giovanni dall'Olio

Ludovica
Montanucci

Hafid
Laayouni

Jaume
Bertranpetit

Marc
Pybus

Ferran
Casals

GROUP MEMBERS

Jaume Bertranpetit, Group Leader

| Professor, UPF



Ferran Casals, Senior Scientist | Beatriu de Pinós Contract

Hafid Laayouni, Senior Scientist | UPF Contract

Ludovica Montanucci, Post-doc | Juan de la Cierva Contract

Giovanni dall'Olio, PhD Student | FI scholarship, MICINN

Pierre Luisi, PhD Student | ISCIII Scholarship

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

Marc Pybus, PhD Student | FI Scholarship, MICINN

Mayukh Mondal, PhD Student | UPF Contract
(since September 2012)

Martino Colombo, Master Student (till December 2012)

RESEARCH OUTLINE

Our main research focuses on how to use molecular pathways to understand the biology of adaptation through the measures of natural selection in genes and other genomic regions. The different forms of selection (purifying, balancing and positive) are analyzed at two levels: among human populations in order to detect population-specific adaptations, and among primates in order both to recognize species-specific adaptive selection and to measure the relative strength of purifying selection.

We have also ongoing work in understanding recombination and in reconstructing population history by studying human genetic diversity. We are also collaborating with Carles Lalueza-Fox in ancient DNA studies and the functional implications of the genetic differences; with Francesc Calafell with the study of surnames and Y-chromosome diversity; with David Comas in human population studies, including the Genographic Project and the genetics of North Africa; and with Tomàs Marquès-Bonet on detecting selection in the genome of apes.

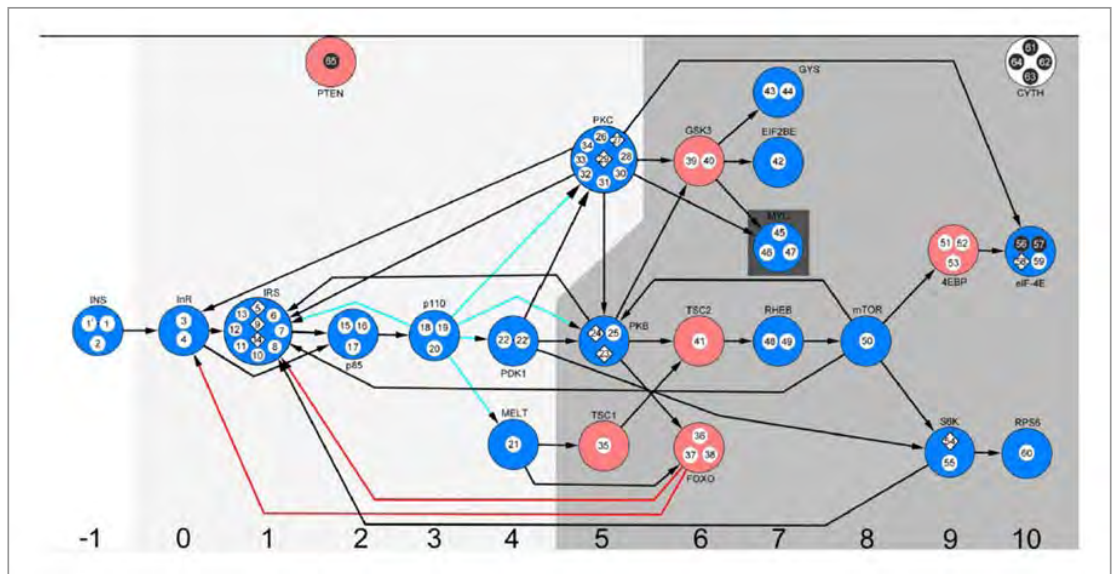


Fig. 1: Structure of the insulin/TOR signal transduction pathway. Each node represents a paralogous group. Blue nodes contain genes that activate the pathway, whereas red nodes contain pathway inhibitors. Within each paralogous group, genes are represented as circles with the numbers corresponding to genes as listed below. White circles represent genes included in our network-level analysis, whereas circles in dark gray represent genes that were excluded. The nine genes for which we observed the signature of positive selection are represented by diamonds. Black, blue, and red edges represent protein-protein, metabolic and transcriptional interactions, respectively. Gray gradations of the background represent the different cellular compartments where the proteins act in the activated pathway (from lighter to darker gray: extracellular action, plasma membrane, cytosol and nucleus).

Numbers on the lower part refers to pathway position (ie. the number of steps required for signals transduction from the insulin receptor).

Gene code: 1, IGF2; 1', INS; 2, IGF1; 3, IGFR; 4, INSR; 5, DOK1; IRS1; 7, DOK3; 8, FRS3; 9, DOK2; 10, FRS2; 11, IRS2; 12, DOK4; 13, DOK6; 14, DOK; 15, PIK3R3; 16, PIK3R1; 17, PIK3R2; 18, PIK3CD; 19, PIK3CB; 20, PIK3CA; 21, VEPH1; 22, PDPK1; 22', AC141586.2; 23, AKT3; 24, AKT1; 25, AKT1; 26, PRKCZ; 27, PRKCF; 28, PRKCD; 29, PRKO; 30, PRKCQ; 31, PRKCH; 32, PRKCB; 33, PRKCA; 34, PRKCG; 35, TSC1; 36, AC093151.2; 37, FOXO3; 38, FOXO1; 39, GSK3B; 40, GSK3A; 41, TSC2; 42, EIF2B5; 43, GYS2; 44, GYS1; 45, MYCL1; 46, MYCN; 47, MYC; 48, RHEB; 49, RHEBL1; 50, MTOR; 51, EIF4EBP3; 52, EIF4EBP1; 53, EIF4EBP2; 54, RPS6KB2; 55, RPS6KB1; 56, EIF4E2; 57, EIF4E3; 58, EIF4E; 59, EIF4E1B; 60, RPS6; 61, CYTH3; 62, CYTH1; 63, CYTH2; 64, CYTH4; 65, PTEN.

RESEARCH LINES

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 of relative importance (in cases of negative or purifying selection) or of adaptation (in cases of positive selection and in the special case of balancing selection) that will be population-specific (in the case of human diversity) or species-specific (in the case of genetic divergence). The action of selection is measured and understood not in terms of single lists of genes, but integrated in molecular physiological pathways or networks, and the aim is, in a given pathway, to understand the complex basis of adaptation and how networks have been shaped by natural selection. Indeed, to understand the complex basis of adaptation it is necessary to integrate the knowledge derived from evolutionary studies into a network framework since biological function is the result of a large number of interacting molecules organized in complex networks and arises as an emergent property from a combined effect of many different genes. The final goal is, on one hand, to understand in specific pathways how evolution has taken place, where positive selection (and balancing selection) has taken place, and where purifying selection has been shaping the genome; and on the other, to obtain possible general patterns of evolution in molecular pathways and networks.

Data is either retrieved from pre-existing databases (HapMap or 1000genomes project) or produced (ImmunoChip, WGS Illumina chip or whole genome sequences).

For the analysis of selection, in collaboration with Johannes Elgenken, we are developing a pipeline for the detection of positive selection that calculates 21 tests and, through simulation, are integrated in a machine learning algorithm (boosting) that produces a single score for specific selection footprints. The initial goal is to produce a map of positive selection in the human genome for three populations (Europe, Asia, Africa) that after will be used for other studies, mainly for the analysis of the ape genomes.

The pathways that we are analyzing are: N-glycans and of all glycosylation pathways; innate immunity; visual perception; and obesity through adiposity signals and the whole human metabolome. Special attention has been put in the quality of databases for metabolic pathways, as their quality is worse than most studies assume and manual curation is needed in all cases.

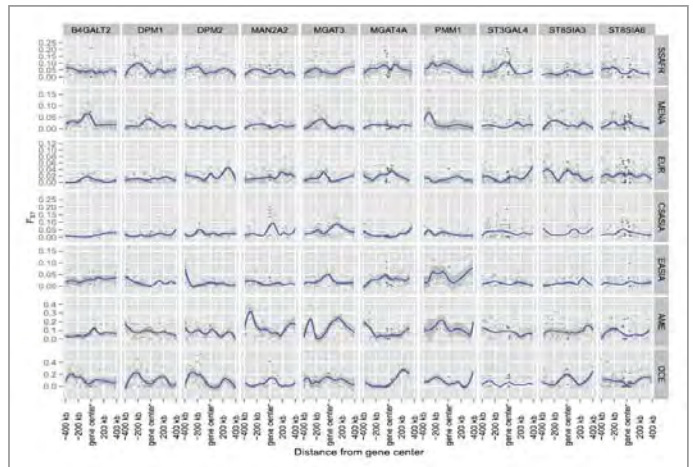


Fig. 2: Distribution of FH values per SNP on the nearby Chromosomal regions for a set of 10 genes (shown here as example) in the Asparagine N-Glycosylation pathway that show signature of population differentiation in at least one continental group.

2. Human population genetics and recombination

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

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

3. Human genetic diversity and population history

In collaboration with David Comas, 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 African, Sub-Saharan African and others).

Thanks to a new collaborative project with NIBMG, India (Prof. Partha Majumder), we have undertaken a major study of population genetics of several Indian populations.

PUBLICATIONS 2012

ISI Articles

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Recombination gives a new insight in the effective population size and the history of the Old World human populations. *Molecular Biology and Evolution* 29 (1): 25-30.

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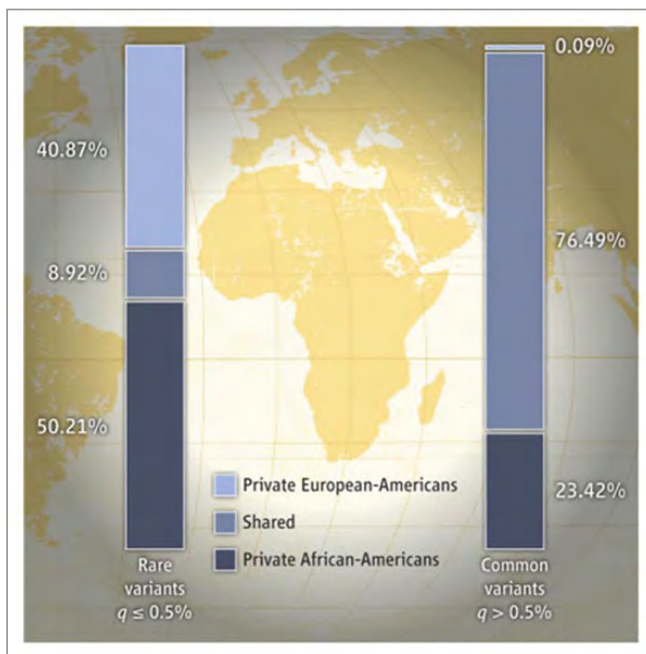


Fig. 3: A new picture of the geographic distribution of genetic variation in humans. From: Casals, F., and Bertranpetit, J. 2012. *Human Genetic Variation, Shared and Private*. Science.

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Other Publications

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FUNDED PROJECTS

Project Title: Selección natural en redes moleculares funcionales
Financed by: Ministerio de Ciencia y Tecnología (BFU2010-19443)
Years: 2011-2013
PI: Jaume Bertranpetit

Project Title: The Genographic project: Western/Central Europe region
Financed by: National Geographic and IBM
Years: 2006-2012
IPs: Jaume Bertranpetit and David Comas

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

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

GROUP

GENOMICS OF INDIVIDUALITY

Cristina
Vasconcelos

Francesc
Calafell

Neus Solé

Koldo García

Marc Garcia



GROUP MEMBERS

Francesc Calafell, Group Leader

| Associate Professor, UPF



Koldo García, Post-doc | CIBERESP and UPF Contracts

Marc Garcia, PhD Student | FU Scholarship, MEC

RESEARCH OUTLINE

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

RESEARCH LINES

1. 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.

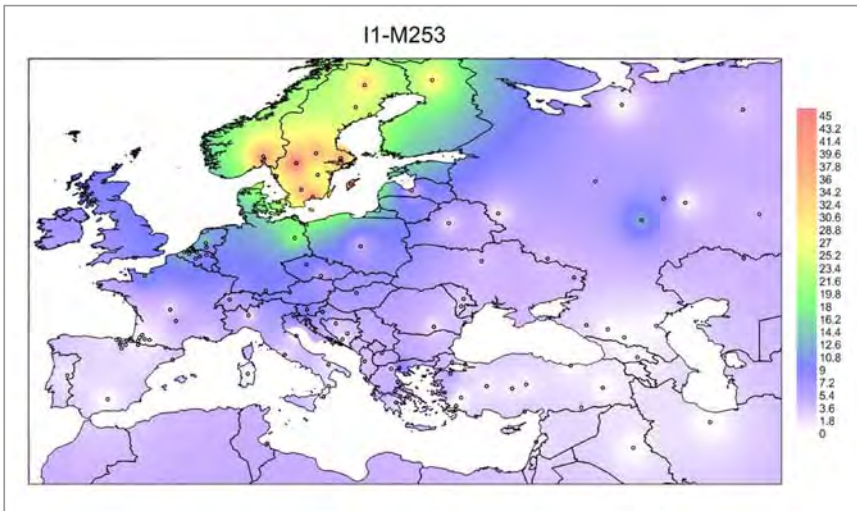


Fig. 1: Population frequencies of Y-chromosome haplogroup I1 M253. Values were taken from the literature and interpolated.

2. 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).

3. A genetic atlas of catalan surnames

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

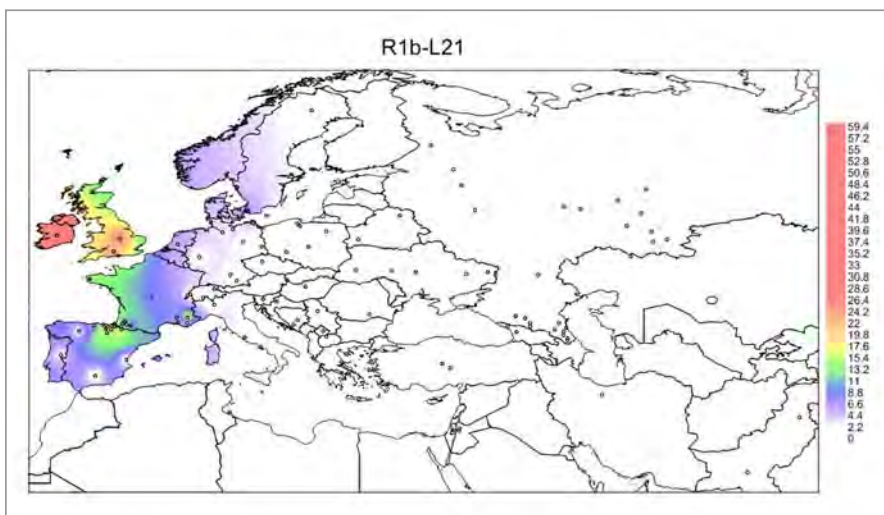


Fig. 2: Population frequencies of Y-chromosome haplogroup R1b-L21.

Catalan surnames (Alemany, Danés, Anglès, Guasch) denote geographic origin (they mean «German», «Dane», «English», and «Gascon», respectively). Such an origin can be traced as long as the Y gene pools of the region of origin and of Catalonia are different enough.

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

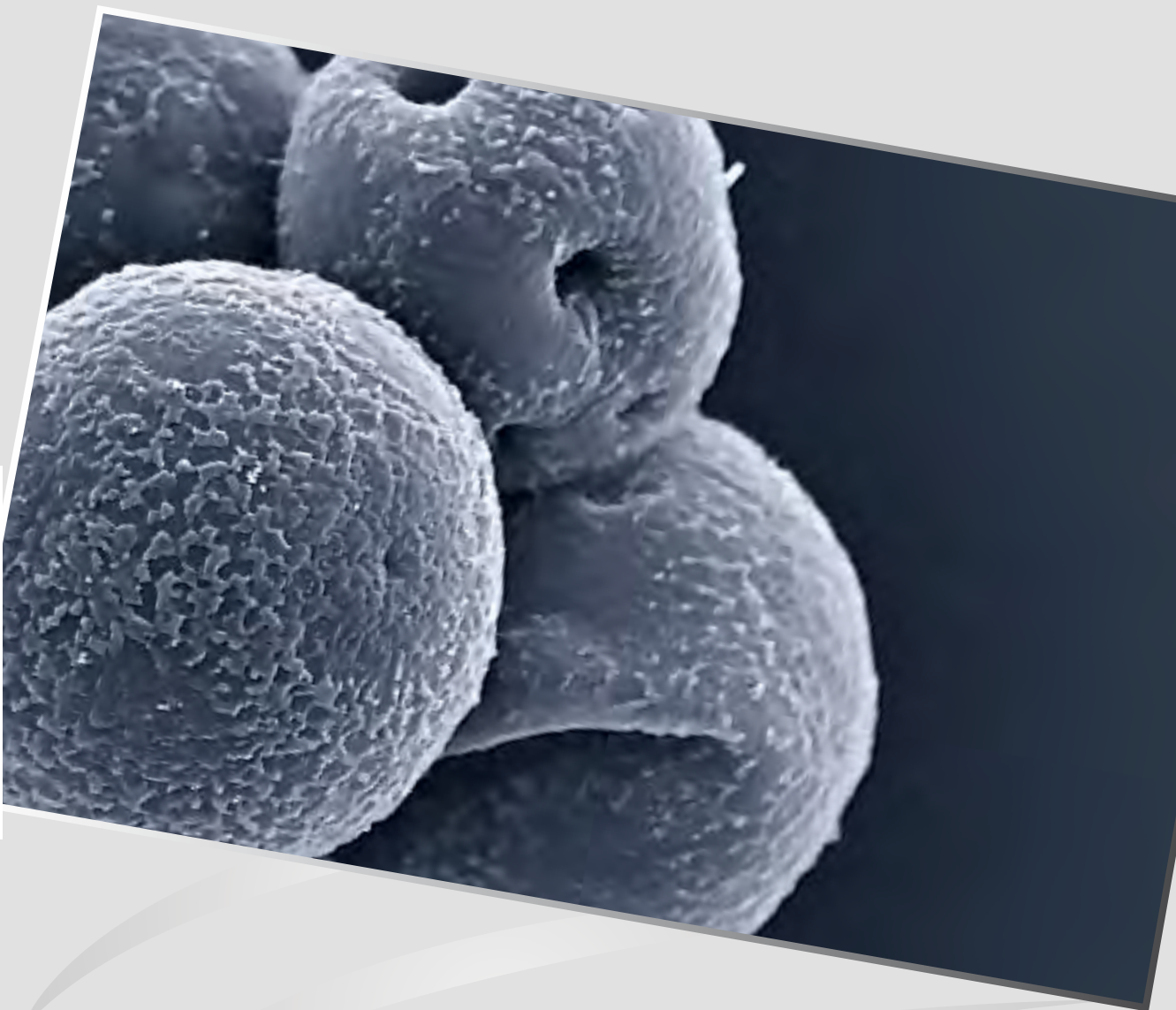
PUBLICATIONS 2012

ISI Articles

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- Garcia-Garcerà, M., Coscollà, M., Garcia-Etxebarria, K., Martín-Caballero, J., González-Candelas, F., Latorre, A., and Calafell, F. 2012. *Staphylococcus* prevails in the skin microbiota of long-term immunodeficient mice. *Environmental Microbiology* 14: 2087-2098.
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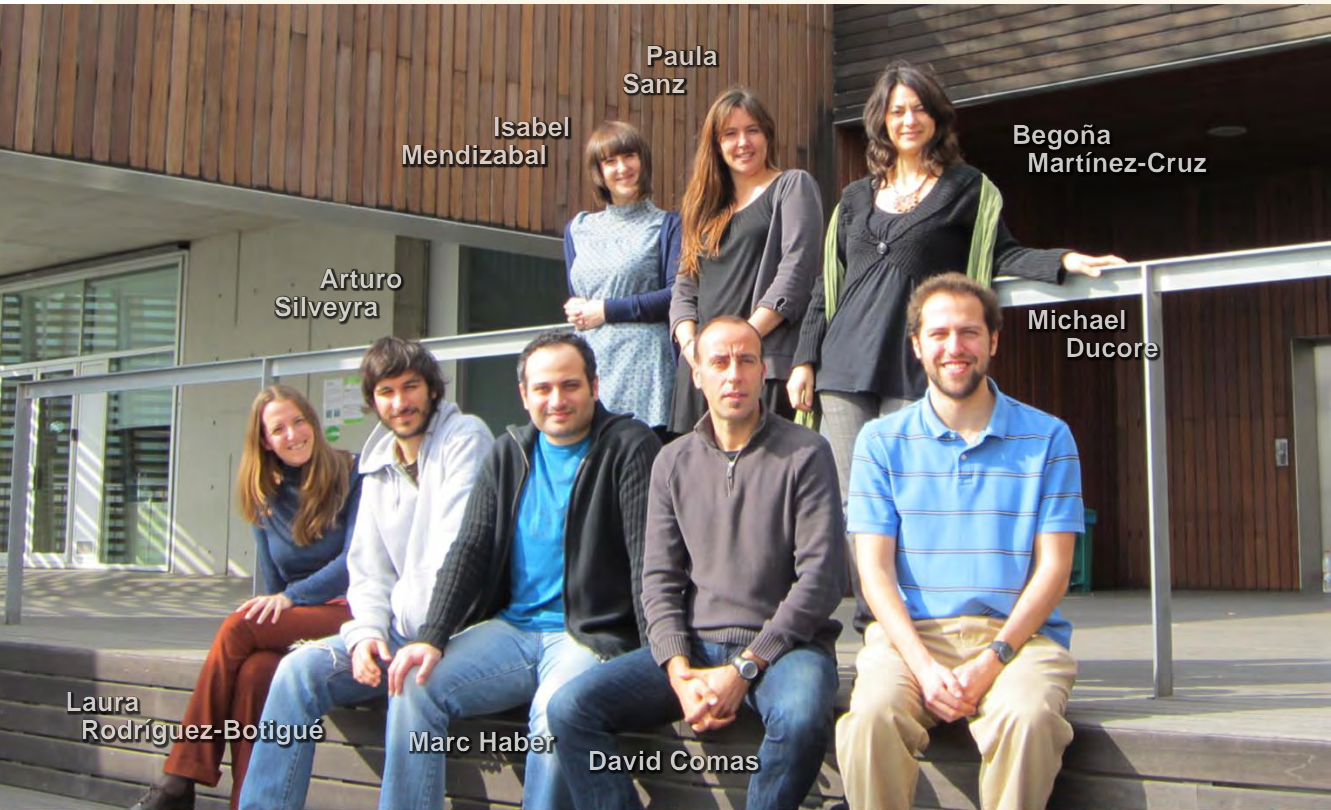
FUNDED PROJECT

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



GROUP

HUMAN GENOME: DEMOGRAPHY AND ADAPTATION



Paula Sanz

Isabel Mendizabal

Begoña Martínez-Cruz

Arturo Silveyra

Michael Ducore

Laura Rodríguez-Botigué

Marc Haber

David Comas

Alicia Gallego

Ignasi Torruella

Maria López-Valenzuela

Yolanda Espinosa-Parrilla



GROUP MEMBERS

David Comas, Group Leader

| Associate Professor, UPF



| Subgroup: Human genome diversity

David Comas, Associate Professor | UPF

Begoña Martínez-Cruz, Post-doc | The Genographic Project

Isabel Mendizabal, Post-doc | ICREA Scholarship

Laura Rodríguez-Botigué, PhD Student | Scholarship MICINN

Marc Haber, PhD Student | The Genographic Project

Arturo Silveyra, PhD Student | Scholarship CSIC (JAE Program)

Michael Ducore, Master Student | UPF

Paula Sanz, Lab Technician | UPF

| Subgroup: MicroRNAs in human adaptation and disease

Yolanda Espinosa-Parrilla, Marie Curie Researcher | UPF

Maria López-Valenzuela, PhD Student | FI-AGAUR Scholarship

Ignasi Torruella, PhD Student | FPI-MINECO Scholarship

Alicia Gallego, PhD Student | FPU-MEC Scholarship

RESEARCH OUTLINE

Our group is focused on the analysis of the human genome and related species in order to understand the processes that have modeled the extant genetic diversity of humans. We are interested in unraveling the demographic and adaptative processes that have given place to the genetic composition of human populations and their consequences in health and disease.

RESEARCH LINES

SUBGROUP: HUMAN GENOME DIVERSITY

1. Demographic history of European populations: differential migrations and genetic composition of some European minorities
2. Migrations and adaptations in North African populations
3. Genomic composition of African populations: demography and adaptation using complete genomes

SUBGROUP: MicroRNAS IN HUMAN ADAPTATION AND DISEASE

1. Involvement of MicroRNA related mechanisms in human disease susceptibility
2. Molecular evolution of MiRNAs in primates
3. Functional analysis of MicroRNAs in stress responses

PUBLICATIONS 2012

ISI Articles

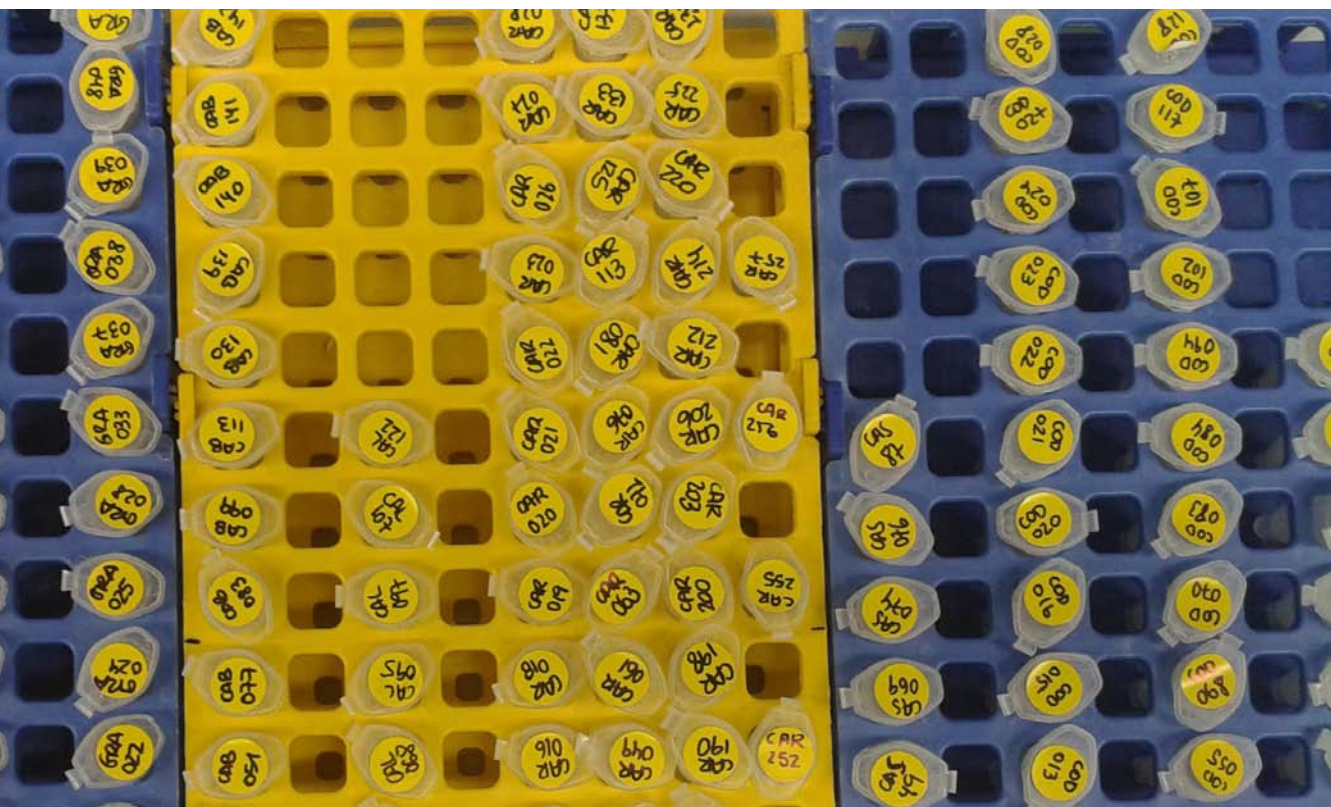
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Fig. 1: DNA samples are the starting point of the population genetics analyses.

Bustamante, C.D., and Comas, D. 2012. Genomic Ancestry of North Africans Supports Back-to-Africa Migrations. *PLoS Genetics* 8 (1): e1002397.

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- Pimenoff, V., Laval, G., Comas, D., Palo, J.U., Gut, I., Cann, H., Excoffier, L., and Sajantilla, A. 2012. Similarity in recombination rate and linkage disequilibrium at CYP2C and CYP2D cytochrome P450 gene regions among Europeans indicates signs of selection and no advantage of using tagSNPs in population isolates. *Pharmacogenetics and Genomics* 22: 846-857.
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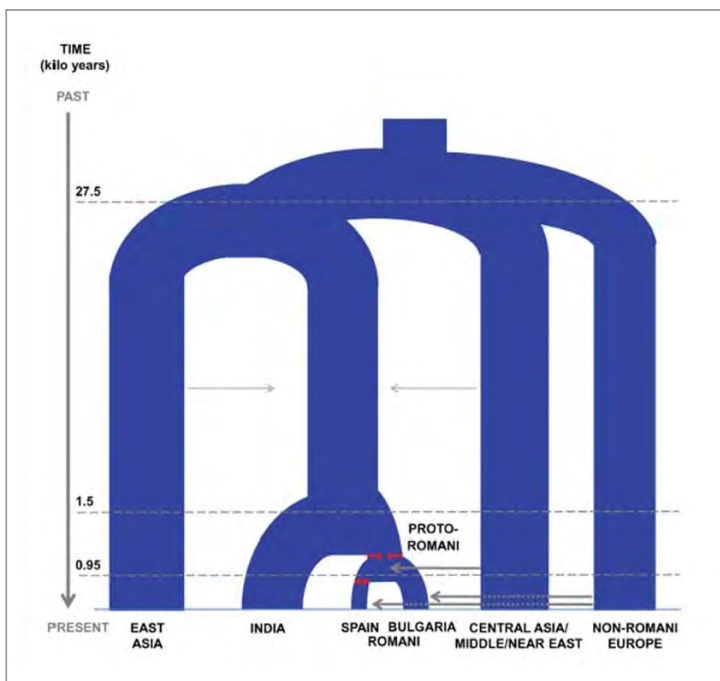


Fig. 2: Demographic inferences of the population history of Romani, aka Gypsies, based on genome-wide data.

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

Financed by: Dirección General de Investigación Científica y Técnica, Ministerio de Ciencia e Innovación

Years: 2011-2013

PI: David Comas

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

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

Years: 2011-2013

PI: David Comas

Project Title: Análisis de la diversidad genética humana en poblaciones mediterráneas (A1/040218/11)

Financed by: Programa de Cooperación Interuniversitaria e Investigación Científica (AECI): Spain-Tunisia

Years: 2011-2012

PI: David Comas

Project Title: MapbyAdmixtureChI-Mapping Genes involved in Psychiatric Disorders by Admixture Linkage Disequilibrium in Chilean populations (PIOF-GA-2008-236836)

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

Years: 2009-2012

PIs: David Comas and Yolanda Espinosa-Parrilla

Project Title: Variabilidad genética y genómica en regiones de microRNAs: hacia la identificación de novedades evolutivas y funcionales en microRNAs

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Fig. 1: Graptodytes eremitus

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Fig. 2: *Cerastes gasparetti*

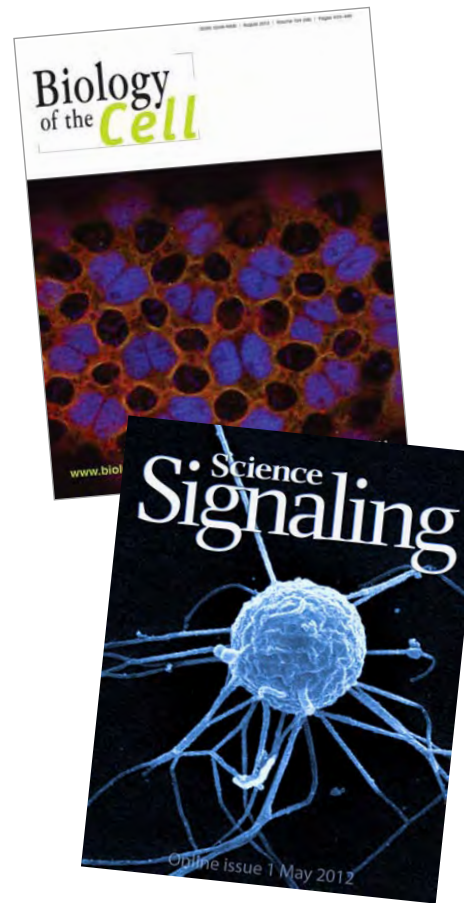
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Fig. 4: Pristurus rupestris

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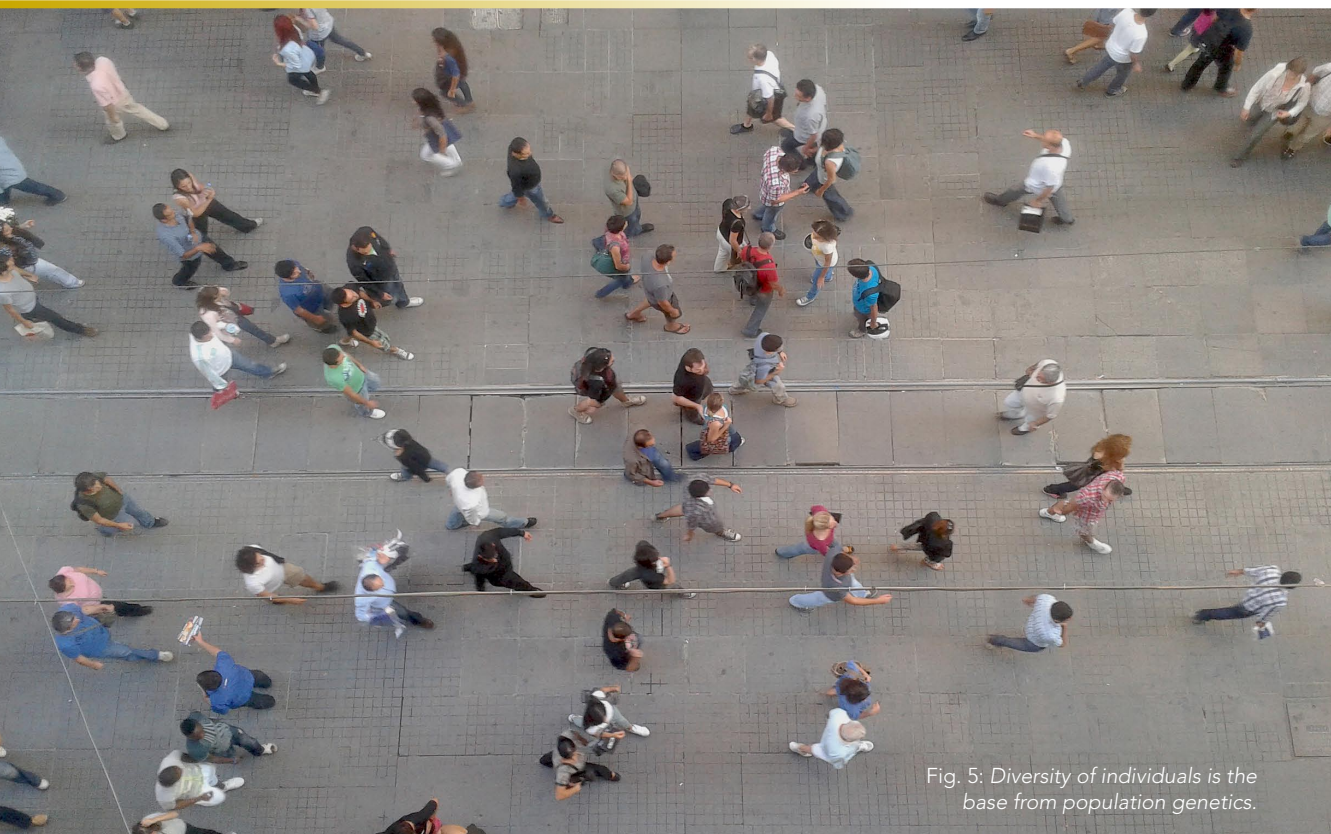


Fig. 5: Diversity of individuals is the base from population genetics.

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PREMETAZOAN ORIGIN OF THE CELL PROLIFERATION HIPPO PATHWAY

The study of metazoans' closest unicellular relatives, for example the filasterean *Capsaspora owczarzaki*, can provide crucial insights to understand a major evolutionary transition as the origin of animal multicellularity. The recent sequencing of the genome of *Capsaspora*, together with other available genomes like those of choanoflagellates, allows us to precisely reconstruct the evolutionary history of the molecular toolkit of animal multicellularity.

Cell signaling pathways are important components of this toolkit, and the Hippo pathway, discovered in recent years, is a key mechanism for controlling cell proliferation (an essential function for a multicellular entity). Like many other important genes for multicellularity, it was considered a metazoan innovation. By comparative genomics, we reconstructed the evolutionary history of this pathway and discovered that, in fact, the pathway evolved long before the origin of Metazoa, as we found all its core elements in *Capsaspora*. Moreover, we cloned some of these core elements and performed heterologous expression experiments in *Drosophila melanogaster* using an eye-specific promoter. We showed that all elements were functionally conserved, both positive and negative regulators of the pathway, in an extremely phylogenetically distant context.

These results tell us a story of a machinery that evolved in a unicellular context and was later co-opted to function in a multicellular one. We don't know what the original function of this pathway was, but by studying its current function in the unicellular *Capsaspora* we may infer it. Interestingly, the upstream regulators of the pathway are metazoan innovations, which suggests that this intracellular signal transduction cascade was activated by different (unknown) upstream clues and later plugged into new upstream receptors.

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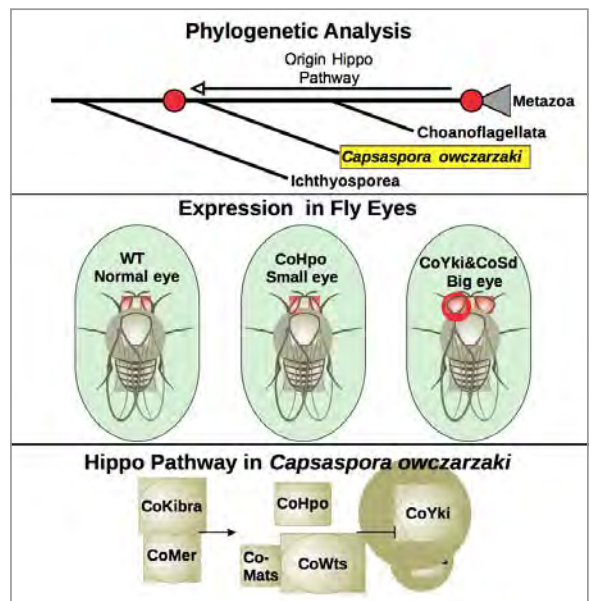


Fig. 1: Hippo signaling pathway, thought to be a key innovation of animal multicellularity, has been found in the unicellular *Capsaspora owczarzaki* (upper panel), which possesses all core components of the pathway (bottom panel) and they are functionally conserved in heterologous expression experiments in *Drosophila* (middle panel).

FIRST GENOMIC DATA FROM 7,000-YEAR-OLD IBERIAN HUNTER-GATHERERS SUPPORT A MESOLITHIC-NEOLITHIC GENETIC DISCONTINUITY

The European Mesolithic is a poorly studied period from a genetic point of view, partly due to the scarcity of the remains and the inherent methodological difficulties of the ancient human DNA research. However, the new massively parallel sequencing technologies can provide information on the nucleotide misincorporations and the fragmentation patterns that can help in determining the authenticity of the ancient sequences. In this work, we present the complete mitochondrial DNA (mtDNA) genome and shotgun genomic data retrieved from two 7,000-year-old hunter-gatherers individuals from La Braña-Arintero, a site in León (NorthWestern Spain). Both specimens show the same U5b2c1 mtDNA haplotype that is interestingly present in many other Mesolithic sites from Northern and Central Europe. This points to a remarkable genetic uniformity over a large geographic area of the pre-Neolithic populations in Europe, suggestive of a small effective population size and no phylogeographic structure. In addition, the genomic data of these two individuals, that correspond to ca. 1.34% and 0.53% of their nuclear genomes, containing ca. 50,000 and 20,000 ancestry informative SNPs, indicate that these hunter-gatherers are not related to modern Iberian populations; instead they seem to be related to current Northern European groups. Overall, our results support the existence of a clear genetic discontinuity between the Mesolithic and the Neolithic, at least in Southern Europe.

Reference Article

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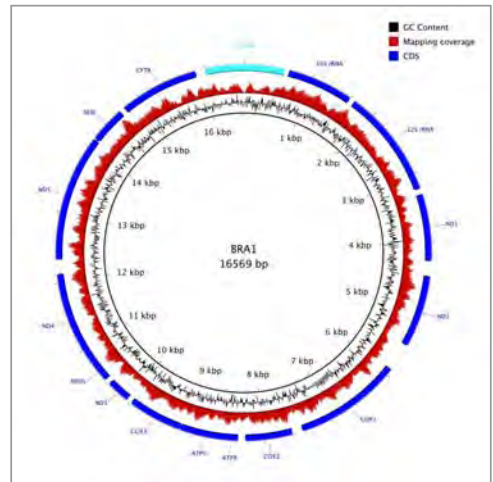


Fig. 1: The La Braña 1 complete mitochondrial genome at 28x coverage. This is the first complete mtDNA genome ever retrieved from the European Mesolithic period.

INCOMPLETE LINEAGE SORTING AMONG HUMANS AND GREAT APES

During 2012, the last two great-ape genomes were published closing our genomic perspective to human evolution. In both projects, incomplete lineage sorting (the distribution of alleles that do not correspond to the known phylogeny) is an important part of the paper. In 30% of the genome, gorilla is closer to human or chimpanzee than the latter are to each other, whereas more than 3% of the human genome is more closely related to either the bonobo or the chimpanzee genome than these are to each other. From this projects we have learnt that these events are rarer around coding genes, indicating pervasive selection throughout great ape evolution. This has functional consequences in gene expression.

On top of that, in the *Sally et al.* paper there was a deep discussion about possible variation of ancestral mutation rates that would allow hominid genomic data to be consistent with the dates from the fossil record. In the *Pruefer et al.* paper, more emphasis was done on selection scans and the effect of demography on the X vs. Autosome diversity.

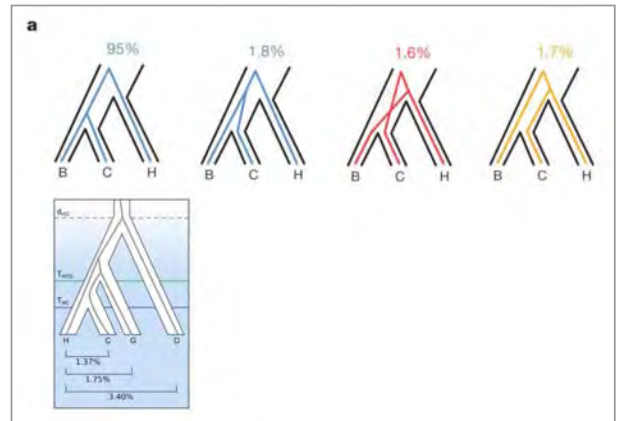


Fig. 1: Percentage of incomplete lineage sorting from genome sequence data from bonobo (above) and gorilla (below).

Reference Articles

- Prüfer, K., Munch, K., Hellmann, I., Akagi, K., Miller, J.R., Walenz, B., Koren, S., Sutton, G., Kodira, C., Winer, R., Knight, J.R., Mullikin, J.C., Meader, S.J., Ponting, C.P., Lunter, G., Higashino, S., Hobolth, A., Dutheil, J., Karakoç, E., Alkan, C., Sajjadian, S., Catacchio, C.R., Ventura, M., Marquès-Bonet, T., Eichler, E.E., André, C., Atencia, R., Mugisha, L., Junhold, J., Patterson, N., Siebauer, M., Good, J.M., Fischer, A., Ptak, S.E., Lachmann, M., Symer, D.E., Mailund, T., Schierup, M.H., Andrés, A.M., Kelso, J., and Pääbo, S. 2012. The bonobo genome compared with the chimpanzee and human genomes. *Nature* 486 (7404): 527-5231.
- Scally, A., Dutheil, J.Y., Hillier, L.W., Jordan, G.E., Goodhead, I., Herrero, J., Hobolth, A., Lappalainen, T., Mailund, T., Marquès-Bonet, T., McCarthy, S., Montgomery, S.H., Schwalie, P.C., Tang, Y.A., Ward, M.C., Xue, Y., Yngvadottir, B., Alkan, C., Andersen, L.N., Ayub, Q., Ball, E.V., Beal, K., Bradley, B.J., Chen, Y., Clee, C.M., Fitzgerald, S., Graves, T.A., Gu, Y., Heath, P., Heger, A., Karakoc, E., Kolb-Kokocinski, A., Laird, G.K., Lunter, G., Meader, S., Mort, M., Mullikin, J.C., Munch, K., O'Connor, T.D., Phillips, A.D., Prado-Martínez, J., Rogers, A.S., Sajjadian, S., Schmidt, D., Shaw, K., Simpson, J.T., Stenson, P.D., Turner, D.J., Vigilant, L., Vilella, A.J., Whitener, W., Zhu, B., Cooper, D.N., de Jong, P., Dermitzakis, E.T., Eichler, E.E., Flicek, P., Goldman, N., Mundy, N.I., Ning, Z., Odom, D.T., Ponting, C.P., Quail, M.A., Ryder, O.A., Searle, S.M., Warren, W.C., Wilson, R.K., Schierup, M.H., Rogers, J., Tyler-Smith, C., and Durbin, R. 2012. Insights into hominid evolution from the gorilla genome sequence. *Nature* 483 (7388): 169-175.

THE CHROMOSOMAL PROTEINS JIL-1 AND Z4/PUTZIG REGULATE THE TELOMERIC CHROMATIN IN *DROSOPHILA MELANOGASTER*

Drosophila telomeres constitute a remarkable exception to the general telomerase mechanism of telomere maintenance in eukaryotes. Telomere maintenance in *Drosophila* depends on the transposition of the specialized retrotransposons *HeT-A*, *TART* and *TAHRE*. Controlling the activation and silencing of these elements is crucial for a precise telomere function without compromising genomic integrity. Adding an extra layer of complexity, the telomere retrotransposons are embedded at the telomere chromatin and therefore part of this fine tuned control should be achieved through epigenetic mechanisms that regulate the telomere chromatin.

In this paper, we report the dual role of two chromosomal proteins, JIL-1 and Z4/putzig, not previously related with the telomeres, in the telomere function of *Drosophila*. The JIL-1 kinase is necessary to achieve wild type levels of *HeT-A* transcription, being the first positive regulator of telomere length described in *Drosophila*. Z4/putzig is important to maintain the presence of key factors at the telomere chromatin such as HP1a, and to guarantee telomere stability. The action of both proteins is mediated by epigenetic changes at the telomeres. We have investigated how the different chromatin marks present at the *HeT-A* promoter vary in mutant alleles of *JIL-1*, *Z4/putzig* and *Su(var)2-5* and in several allelic combinations of these three genes. With all the data extracted from quantitative RT-PCR, ChIP and Co-immunoprecipitation experiments, we propose a model for the observed different chromatin scenarios and to explain how the coordinated action of JIL-1 and Z4 results in the fine-tuned regulation of the retrotransposon telomeres. Finally, we have been able to demonstrate how the *HeT-A* Gag protein, a specific telomere protein, is responsible for the targeting of the JIL-1-Z4 complex at the telomeres. Our results are crucial to understand how the retrotransposon telomeres manage to develop an essential function in the *Drosophila* genome being of interest for the telomere community in general as well as for those groups interested in the biology of retroelements.

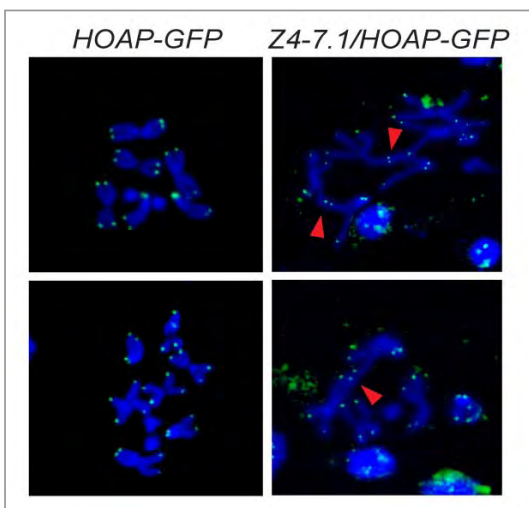


Fig. 1: 1st column: wild type chromosomes;
2nd column: Z4 mutant chromosomes where several telomere fusions can be observed.

Reference Article

- Silva-Sousa, R., López, E., Piñeyro, D., and Casacuberta, E. 2012. The Chromosomal Proteins JIL-1 and Z4/Putzig Regulate the Telomeric Chromatin in *Drosophila melanogaster*. *PLoS Genetics* 8 (12): e1003153.

WHAT EPIGENETICS CAN TELL US ABOUT HOST-PATHOGEN CO-EVOLUTION AND EPIDEMIOLOGY?

Few areas in biology attract as much current attention, and yet require as much presentation, as the field of epigenetics. The concept of epigenetics has evolved from the early definition by Conrad Waddington about the relationship between genotype and phenotype during development, towards the mechanistic understanding of the importance of DNA methylation, chromatin structure and non-coding RNAs in regulating genomic and phenotypic plasticity. This intellectual advances have been accompanied by technological breakthroughs that now make it possible to undertake sophisticated epigenomic studies across a range of organisms. The role of epigenetics in host-parasite interactions has, however, received very little attention. Yet, there is plenty to suggest that epigenetic mechanisms are implicated in the modulation of the biological interaction between hosts and pathogens.

Host-pathogen interactions are amongst the most plastic and dynamic systems in nature. To cope with the selective constraints imposed by their hosts, many pathogens have evolved an unparalleled level of phenotypic plasticity in their life history traits. Likewise, the host phenotype is drastically and rapidly altered by the presence of a pathogen, and in some cases, the parasitized phenotype is inherited across host generations. In addition,

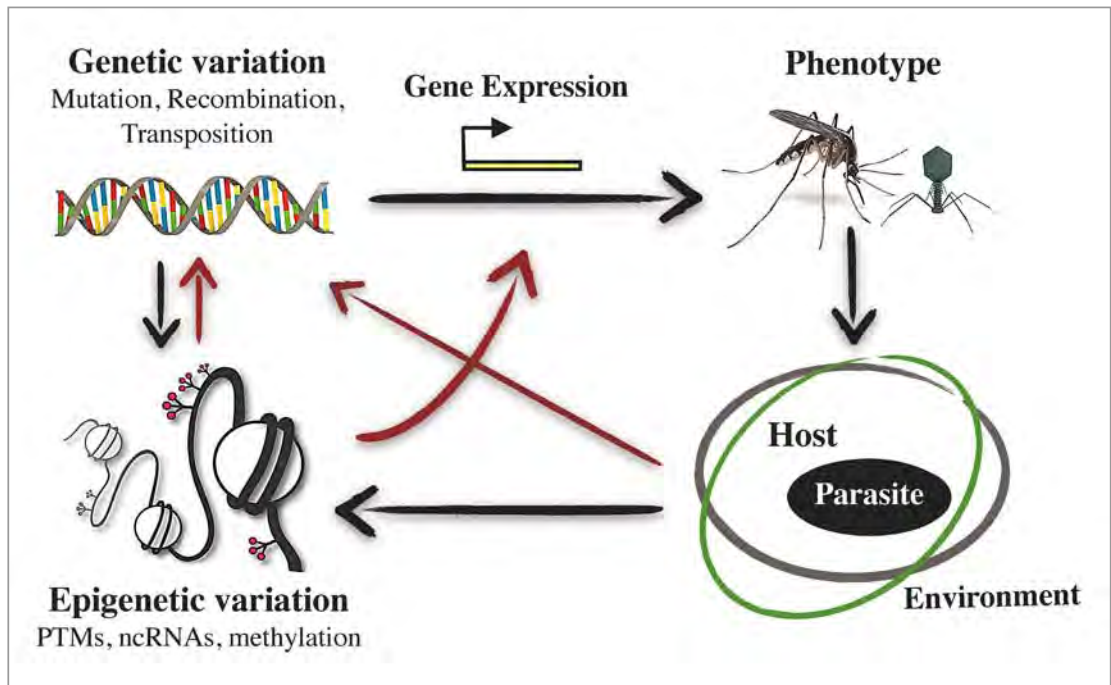


Fig. 1: Schematic representation of the interrelations between epigenetic variation, phenotypic variation and host-parasite interactions. The infection phenotype, which varies between host and parasite phenotypes and is environmentally-dependent, can induce changes at both the genomic and epigenomic levels. These changes can in turn alter gene expression patterns. Apart from these direct effects of epigenetic variation on host and parasite phenotypes, epigenetic variation can also have indirect, and transgenerational, phenotypic effects by influencing the probability of mutation, transposition and/or recombination of the DNA sequence, as well as the predisposition of a gene with a particular epigenetic mark to be selected.

co-adaptations between hosts and pathogens often occur over such short evolutionary time scales as to call into question the sole role of genetic modifications as an underlying mechanism. In this sense, epigenetic modifications may provide an accessory source of fast-acting, reversible, and readily available phenotypic variation that can be directly shaped by both host and pathogen selection pressures.

In this paper, we advocate that epigenetics not only represents a paradigm shift in our understanding of host and parasite phenotypic plasticity, but it may have crucial fundamental and applied implications. Indeed, epigenetic epidemiology has recently emerged as a promising area for future research on infectious diseases. In addition, the incorporation of epigenetic inheritance and epigenetic plasticity mechanisms to evolutionary models and empirical studies of host-pathogen interactions will provide new insights into the evolution and co-evolution of these associations. Here we review the evidence available for the role of epigenetics in shaping host-parasite co-adaptations, the methods at-hand to characterise these epigenetic modifications, as well as future directions for encouraging research on the emerging field of host-parasite epigenetics.

Reference Article

- Gómez-Díaz, E., Jordà, M., Peinado, M.A., and Rivero, A. 2012. Epigenetics of host-pathogen interactions: the road ahead and the road behind. *PLoS Pathogens* 8 (11): e1003007.

DIVERSIFICATION RATES AND THE LATITUDINAL GRADIENT OF DIVERSITY IN MAMMALS

One of the most striking patterns of species richness is the latitudinal gradient of biodiversity, which is due to the larger number of species in the tropics and its gradual decrease towards the poles. This pattern has been observed for many groups of species, including mammals. So far, the most prevalent explanation for this pattern has been a supposedly greater diversification rate in tropical regions. In order to test this hypothesis for mammals, we used a set of 232 genera taken from a mammalian supertree and, additionally, we

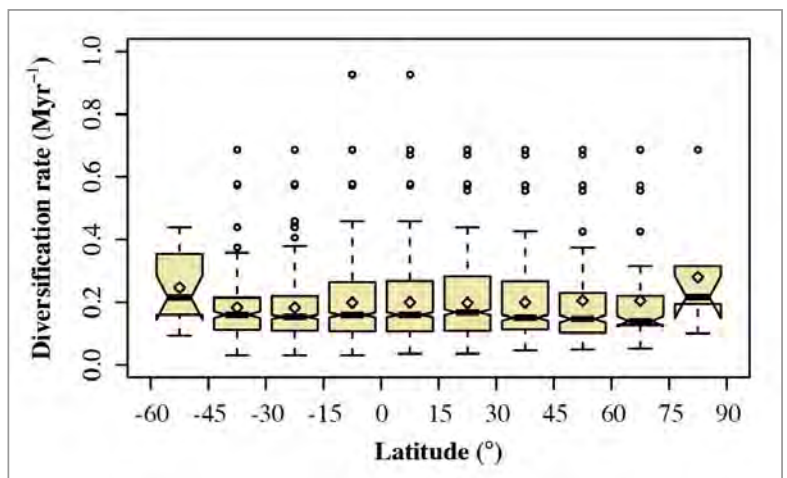


Fig. 1: Diversification rates were estimated for different mammalian genera. For each genus, we used the latitudinal band with maximum species richness and plotted the corresponding diversification rate. Box plots of analyzed genera indicate very small differences in diversification rates among latitudinal bands. Actually, there was no correlation between diversification rate and assigned latitude, also when using a phylogenetic comparative method (not shown).

reconstructed dated Bayesian phylogenetic trees of 100 genera. For each genus, we assigned the latitude considering the heterogeneity in species distribution ranges and calculated the diversification rate from the trees. For both sets of trees we found that, in fact, the net generation of species was similar at all latitudes. Therefore, other explanations for the latitudinal gradient, probably related to differences in dispersal through evolutionary time, should be further explored in the future.

Reference Article

- Soria-Carrasco, V., and Castresana, J. 2012. Diversification rates and the latitudinal gradient of diversity in mammals. *Proceedings of the Royal Society B: Biological Sciences* 279: 4148-4155.

THE EFFECT OF GEOGRAPHICAL SCALE OF SAMPLING ON DNA BARCODING

In 2003, a group of Canadian scientists proposed the use of a standard gene fragment (the 5' end of the cytochrome oxidase 1) to identify all animal species, as a kind of universal «barcode of life». The idea was simple and intuitively appealing, immediately inspiring the imagination of scientist and general public alike: the tricorder of Star Trek fame –a hand-held device able to sequence and identify any biological material in the field– would be finally made real. A flood of studies in many different groups has since then accumulated and the use of genetic barcodes is now widespread and well established. It is however necessary to assess the performance of the barcodes, to see if they can really deliver all what they are supposed to. The work by Bergsten et al. addresses one of the main potential pitfalls of the use of barcodes, which is the role of geographical scale. Basically, the use of barcodes imply finding the closest match to a query sequence among a reference pool, which ideally should include all species (and their variation) of the lineage to which the query organism belongs. However, for practical limitations the reference pool is usually drawn from a defined geographical space, which means that only part of the extant genetic variation of the lineage is included. This work uses a group of well known European diving beetles (tribe Agabini) to see how the performance of barcodes changes from local to continental scale. As expected, intraspecific variation was significantly correlated with the geographical scale of sampling, and the distance to the closest heterospecific showed a significant decrease with increasing

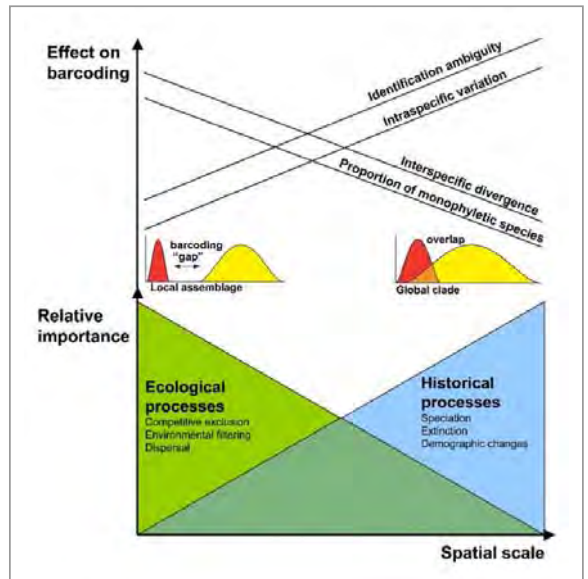


Fig. 1: Schematic representation of the relative importance of processes as spatial and temporal scale increases, and their effect on DNA barcoding parameters. The linear slopes are simplifications, as real processes could be non-continuous and chaotic across different domains of scale.

geographical scale. The average genetic distance dropped from >7% for samples within 1 km, to <3.5% for samples up to >6000 km apart. Over a third of the species were not monophyletic, and the proportion increased through locally, nationally, regionally, and continentally restricted subsets of the data. The success of identifying queries decreased with increasing spatial scale of sampling, dropping to below 50% for some methods at continental scales. The proportion of query identifications considered uncertain (more than one species <1% distance from query) escalated from zero at local, to 50% at continental scale. Finally, by resampling the most widely sampled species it was shown that even when samples are collected to maximize the geographical coverage, up to 70 individuals are required to sample 95% of intraspecific variation. The results of this study clearly show that the geographical scale of sampling has a critical impact on the global application of DNA barcoding. When used at local or regional scales and with well-known faunas they are accurate and reliable, but their use at global scales, at least with highly diverse groups, is likely to be much more problematic.

Reference Article

- Bergsten, J., Bilton, D.T., Fujisawa, T., Elliott, M., Monaghan, M.T., Balke, M., Hendrich, L., Geijer, J., Herrmann, J., Foster, G.N., Ribera, I., Nilsson, A.N., Barraclough, T.G., and Vogler, A.P. 2012. The effect of geographical scale of sampling on DNA barcoding. *Systematic Biology* 61: 851-869.

UNRAVELING THE POPULATION HISTORY OF HUMAN NORTH AFRICAN GROUPS

The population history of humans in North Africa has been determined by the ancient Paleolithic occupation of the area, migrations from the Near East, contacts with Europeans, and trans-Saharan gene flow. The relevance of each of these migrations in the modeling of extant North African groups has been analyzed by different disciplines such as archaeology, linguistics and genetics with little consensus. In order to unravel the population history of North Africans, a genome-wide analysis based on ~800,000 SNPs has provided us genetic evidence of the multilayered history of these groups. We have identified an autochthonous North African component, absent in non-North African populations, which decreases from west to east that is likely derived from a back-to-Africa migration from the Middle East and dated between 18,000 and 38,000 years ago. Ancient human occupation of the region in Paleolithic times has been recently dated back to 120,000 years ago, but our genetic data shows that there is no continuity between these ancient settlers and the extant North African groups, pointing to a recent population replacement prior to the Holocene. Besides this autochthonous component, influences from the Middle East, Europe and sub-Saharan Africa are also present in North African groups, suggesting multiple migrations in the region. We have estimated that most of the sub-Saharan gene flow detected is recent and our data agree with gene flow in historical times due to trans-Saharan slave trading. In addition, recent historical migrations, such as the Jewish Diaspora, have modeled the population history of North Africans. However, the genetic analysis of Jewish North African groups has shown that these groups cluster with other Jewish communities and show a high degree of

endogamy with little gene flow from neighboring non-Jewish North Africans, suggesting a certain degree of isolation of Jewish groups in North Africa. In summary, our genetic analyses have shown that the population history of North Africa has been a complex process of migration, admixture and isolation from diverse population sources.

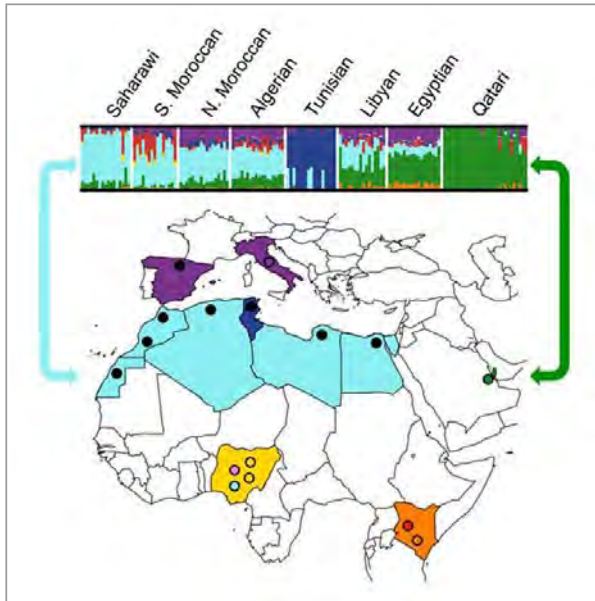


Fig. 1: Admixture analysis of North African populations showing different ancestry components based on genome-wide data.

Reference Articles

- Campbell, C.L., Palamara, P.F., Dubrovsky, M., Botigué, L.R., Fellous, M., Atzmon, G., Oddoux, C., Pearlman, A., Hao, L., Henn, B.M., Burns, E., Bustamante, C.D., Comas, D., Friedman, E., Pe’er, I., and Ostrer, H. 2012. North African Jewish and non-Jewish populations form distinctive, orthogonal clusters. *Proceedings of the National Academy of Sciences USA* 109 (34): 13865-13870.
- Henn, B.M., Botigué, L.R., Gravel, S., Wang, W., Brisbin, A., Byrnes, J.K., Fadhlou-Zid, K., Zalloua, P.A., Moreno-Estrada, A., Bertranpetit, J., Bustamante, C.D., and Comas, D. 2012. Genomic Ancestry of North Africans Supports Back-to-Africa Migrations. *PLoS Genetics* 8 (1): e1002397.



THESES, COURSES AND SEMINARS



DOCTORAL THESES PRESENTED DURING 2012

- PhD Student:* Juan Lázaro Hernández
Title: El género *Pyrgus* en Europa: sistemática, ecología y patrones biogeográficos (Lepidoptera: Hesperiidae)
Thesis Director: Roger Vila
Institution and Date: Universitat Autònoma de Barcelona, 1st April 2012
- PhD Student:* Isabel Mendizabal
Title: Demography and genetic adaptation: examples from human populations
Thesis Director: David Comas
Institution and Date: Universitat Pompeu Fabra, 27th July 2012
- PhD Student:* Carmelo Andújar
Title: Systematics and evolution of the *Carabus* (*Mesocarabus*) *lusitanicus* complex (Insecta: Coleoptera: Carabidae: Carabini) in the Iberian Peninsula
Thesis Director: José Serrano (University of Murcia) and Jesús Gómez-Zurita (IBE)
Institution and Date: Universidad de Murcia, 28th September 2012
- PhD Student:* Rute Souza
Title: The epigenetic regulation of *Drosophila* telomeres
Thesis Director: Elena Casacuberta
Institution and Date: Universitat Pompeu Fabra, 9th November 2012
- PhD Student:* Urko Martínez Marigorta
Title: Genetic architecture of Complex disease in humans: a cross-population exploration
Thesis Director: Arcadi Navarro
Institution and Date: Universitat Pompeu Fabra, 12th November 2012
- PhD Student:* Laura Rodríguez Botigué
Title: Demographic insights of human North African populations using genetic data
Thesis Director: David Comas
Institution and Date: Universitat Pompeu Fabra, 16th November 2012

PhD Student: Mercedes Rubio

Title: microRNAs and metamorphosis in the hemimetabolous insect *Blattella germanica* (L.) (Dictyoptera, Blattellidae)

Thesis Director: Xavier Bellés

Institution and Date: Universitat Pompeu Fabra, 23rd November 2012

PhD Student: Gerard Talavera

Title: Phylogenetic inference at different insect taxonomic levels

Thesis Director: Roger Vila

Institution and Date: Universitat Autònoma de Barcelona, 3rd December 2012

IBE SEMINARS 2012

Speaker	Talk	Institution	Date
Mark Cock	Evolution of complex multicellularity in the brown algae	Igal Genetics Group Station Biologique Roscoff, France	01/02/2012
Brian Charlesworth	What determines the relative variability levels for the X chromosome versus the autosomes in <i>Drosophila</i> ?	Institute of Evolutionary Biology, University of Edinburgh	01/03/2012
Antonio Rosas	Investigations on Neandertal evolution and paleobiology: the El Sidrón sample as a case study	Museo Nacional de Ciencias Naturales	13/03/2012
Karl-Wilhelm Koch	Vertebrate phototransduction- a prototypical signalling pathway	Carl von Ossietzky Universität Oldenburg, Institut für Biologie und Umweltwissenschaften, Germany	14/03/2012
Antonio Barbadilla	Mapping the footprint of natural selection throughout the genome	Facultad de Biociencias, UAB	19/03/2012
Damien Devos	Microbiology's platypus	Structural Bioinformatics EMBL	28/03/2012
Philip Donoghue	Embryology during the Cambrian explosion	Department of Earth Sciences, University of Bristol	18/04/2012
Brent Emerson	The emerging complexity of animal life in soil	Instituto de Productos Naturales y Agrobiología (IPNA), La Laguna	30/05/2012

Speaker	Talk	Institution	Date
Chiara Barbieri	Genetic variation in Khoisan of southern Africa	Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology	31/05/2012
Joanna Mountain	Web-based Participatory Genomics Research	Stanford University	22/06/2012
Steen Rasmussen	Assembling minimal life bottom up from organic and inorganic materials	Center for Fundamental Living Technology, University of Southern Denmark	10/07/2012
Martin Sikora	Inference of demographic history and natural selection in African Pygmy populations from whole-genome sequencing data	Stanford University	20/07/2012
Mattias Jakobsson	Genomic variation and evolutionary history of southern African Khoe-San populations	University of Uppsala, Sweden	27/07/2012
Harmit Malik	Genetic conflict: the usual suspects and beyond	Fred Hutchinson Cancer Research Center, Seattle, USA	05/10/2012
Ricard Solé	The evolutionary ecology of technological innovation	ICREA; Institut de Biologia Evolutiva (UPF-CSIC); Departament dCEXs UPF	17/10/2012
Helio Costa	RNAseq characterizing human transcriptome variation across seven diverse populations	Department of Genetics, Stanford University, Stanford, USA	19/10/2012
Daniel Halligan	Contribution of protein-coding and regulatory change to adaptive evolution in the mammalian genome	University of Edinburgh, UK	24/10/2012
Laura Baldo	Microbial menageries of animals in evolutionary studies: insights from insects and fishes	Zoological Institute, Basel	07/11/2012

Speaker	Talk	Institution	Date
Yikang Rong (NIH)	Telomere maintenance in <i>Drosophila</i> fast evolving genes fulfilling essential functions	National Cancer Institute, National Institutes of Health, NIH, Bethesda, USA	08/11/2012
Chantall Vaury	Struggle between transposable elements and their host genome: the story of ZAM from <i>Drosophila melanogaster</i>	Laboratoire GReD, Faculté de Médecine, UMR/CNRS6293, Clermont Université, INSERM, France	08/11/2012
Greg Gibson	Personalized genomics of wellness	Center for Integrative Genomics, Georgia Institute of Technology, Atlanta, Georgia	12/11/2012
Carlos Juan	Throwing light on the dark side: the historical biogeography of subterranean crustaceans	Universitat de les Illes Balears	28/11/2012
Laurent Duret	Evolution of robustness to splicing errors in <i>Paramecium</i> : trade-off between under-splicing and over-splicing	Université Claude Bernard - Lyon 1	12/12/2012
Mónica Medina	Coral reef responses to global climate change: a genomic perspective	University of California, Merced	17/12/2012
Manuel Irimia	Evo-Devo beyond the gene: genome structure and alternative splicing in animal evolution	Blencowe lab, University of Toronto	19/12/2012

TEACHING

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

GRADUATE STUDIES

- | Bachelor's Degree in Human Biology (Universitat Pompeu Fabra)
 - Human Evolution and Health (4 ECTS).
Coordinators: Elena Bosch and David Comas.
 - Zoology (4 ECTS). Coordinator: Ferran Casals.
 - Ecology (4 ECTS). Coordinator: Francesc Calafell.
 - Integrated Biomedicine I (4 ECTS). Coordinator: David Comas.
 - Basic Sciences 1. (7 ECTS). Coordinator: Ricard Solé.
 - Genomics (4 ECTS). Coordinator: Jaume Bertranpetit.
 - Human Biology Seminars (4 ECTS). Coordinator: Jaume Bertranpetit.
- | Bachelor's Degree in Medicine (Universitat Pompeu Fabra)
 - Human Evolution and Health (4 ECTS).
Coordinators: Elena Bosch and David Comas.
- | Bachelor's Degree in Biomedical Engineering (Universitat Pompeu Fabra)
 - Molecular Biology of the Cell II (BMCII) (4 ECTS).
Coordinator: Yolanda Espinosa Parrilla.
 - Cells and Tissues Engineering (5 ECTS). Coordinator: Ricard Solé.

MASTER STUDIES

- | Màster en Recerca Biomèdica (BIOMED) (Universitat Pompeu Fabra)
 - Subjects
 - Genomes and Systems (5 ECTS). Coordinator: Tomàs Marquès-Bonet.
- | Master in Bioinformatics for Health Sciences (BIOINFO). Joint master of the Universitat Pompeu Fabra (coordination) and Universitat de Barcelona, in cooperation with the Università di Bologna.
 - Subjects
 - Analysis of Biomedical Data (5 ECTS). Coordinator: Arcadi Navarro.
 - Biomedical Informatics (5 ECTS). Coordinator: Arcadi Navarro.

Furthermore, most IBE scientists actively participate in several international master programs and specialized courses in different universities:

- | Master on Biomedical Research (BIOMED); Universitat Pompeu Fabra (UPF)
- | Master on Human Biology; Universitat de Barcelona (UB) / Universitat Autònoma de Barcelona (UAB)
- | Master on Bioinformatics and Health Sciences (BIOINFO); Universitat Pompeu Fabra (UPF) / Universitat de Barcelona (UB) / Università di Bologna
- | Master on Public Health; Universitat Pompeu Fabra (UPF)
- | Master on Comunicación Científica, Médica y Ambiental; IDEC-UPF
- | Master on Genetic Counselling; IDEC/UPF
- | Master on Developmental Biology & Genetics; Universitat de Barcelona (UB)
- | Master on Biodiversity; Universitat de Barcelona (UB)
- | Master of Integrative Physiology; Universitat de Barcelona (UB)
- | Master on Bioinformatics (ISSCii Universidad Autonoma de Madrid)
- | The Gulbenkian Training Programme in Bioinformatics (Instituto Gulbenkian de Ciência, Oeiras, Portugal)
- | Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones; Universitat de Barcelona (UB)
- | Curs Universitari d'Estiu; Universitat Autònoma de Barcelona (UAB)
- | Specialised Course: Next Gen Sequencing for Medical & Population Genomics; Barcelona Summer School in Genomics & Bioinformatics (IBE/UPF)
- | Specialised course: Programming for Evolutionary Biology; Leipzig University
- | Specialised course: Programming for Biologists ; Cold Spring Harbor
Specialised Course: 1st Meeting in Commemoration of Alan Turing - 2012 Year of Computer Science; Universidad Menéndez y Pelayo

Last but not least, every year IBE hosts several undergraduate and master students through his/her scientific projects coming from most of Catalan Universities and some international ones.

Along 2012 IBE has hosted students from:

Universitat Pompeu Fabra (UPF), Universitat de Barcelona (UB), Universitat de Vic (uVIC), Universitat Autònoma de Barcelona (UAB), Università di Padua, Universidad Complutense de Madrid, Escola Pia de Terrassa

DISSEMINATION OF SCIENCE



TRAINING AND OUTREACH UNIT

The Training and Outreach Unit was created in May 2012 with two main objectives: to establish a post-graduate training program in Evolutionary Biology, and to inform and educate the general public about the research that is carried out at the Institut de Biologia Evolutiva (IBE). The IBE Executive Board appointed David Comas, UPF Associate Professor, and Josefa González, Ramón y Cajal Researcher, as joint coordinators of the Training and Outreach Unit.

TRAINING ACTIVITIES

During 2012, the IBE started to develop a training program for PhD and Postdoctoral students. The main goals of the program are: (i) to establish a deep knowledge in Evolutionary Biology including theoretical, analytical and experimental tools; (ii) to reinforce oral and writing abilities; (iii) to develop leadership and management qualities; and (iv) to promote the abilities to evaluate the bioethical implications of a research project. The activities of the training program will be implemented in 2013.

OUTREACH ACTIVITIES

The IBE is committed to inform and educate the general public about the research being carried out at the Institute. During 2012, the IBE has organized and participated in several outreach activities and it is also leading an outreach initiative aimed at increasing awareness of the importance and implications of Science in everyday life: *La ciència al teu món*.

La festa de la ciència (Science Festival)

16th and 17th June 2012. Organized by: Ajuntament de Barcelona.



Luc Steels participated in the Science Festival held at the Ciutadella Park in Barcelona. Dr. Steels introduced the MYON, a new humanoid robot used in experiments of language evolution in autonomous robots. The activity linked to Berlin to see the robots in action, and the language games to which robots are exposed were discussed.

Simonyi Lecture 2012

October 2012. Organized by: University of Oxford.

The 2012 Charles Simonyi Lecture was given by Luc Steels and asked if machines can be creative enough to invent their own language.

PRBB Open Day

6th October 2012, from 10am to 7pm. Organized by: PRBB.

IBE researchers participated in the PRBB open day activities by hosting visitors and giving scientific talks.



Workshops for Secondary School Teachers

Teachers and Science Program. 24th and 26th October 2012, from 5pm to 8pm.

Organized by: Fundació Catalunya Caixa and Institut de Biologia Evolutiva.



Diego Hartasánchez and Elena Gómez-Díaz organized a two day workshop entitled *Reconstruint el nostre passat evolutiu*. The goal of the workshop was to help improve the training of secondary school teachers by given them the opportunity to learn from experts. A total of 14 secondary school teachers from different schools around Catalonia attended the workshop and positively evaluated the experience.

Setmana de la ciència 2012 (2012 Science Week Activities)

20th and 21st November 2012, from 9am to 7pm.

Organized by: Institut de Ciències del Mar i Institut de Biologia Evolutiva.

Funded by: Fundación Española para la Ciencia y la Tecnología (FECYT).

The IBE participated for the first time in the science week activities by offering both schools and the general public three hands-on activities based on three different research lines that are currently being pursued at the IBE.

A total of 19 schools (502 visitors) and 96 general public visitors participated in the activities. 20 IBE members, from undergraduate students to PIs, participated in the science week activities. Both visitors and volunteers positively evaluated the experience.



Schools Visits

18th December 2012, 12.30 to 2pm. Organized by PRBB and the IBE.

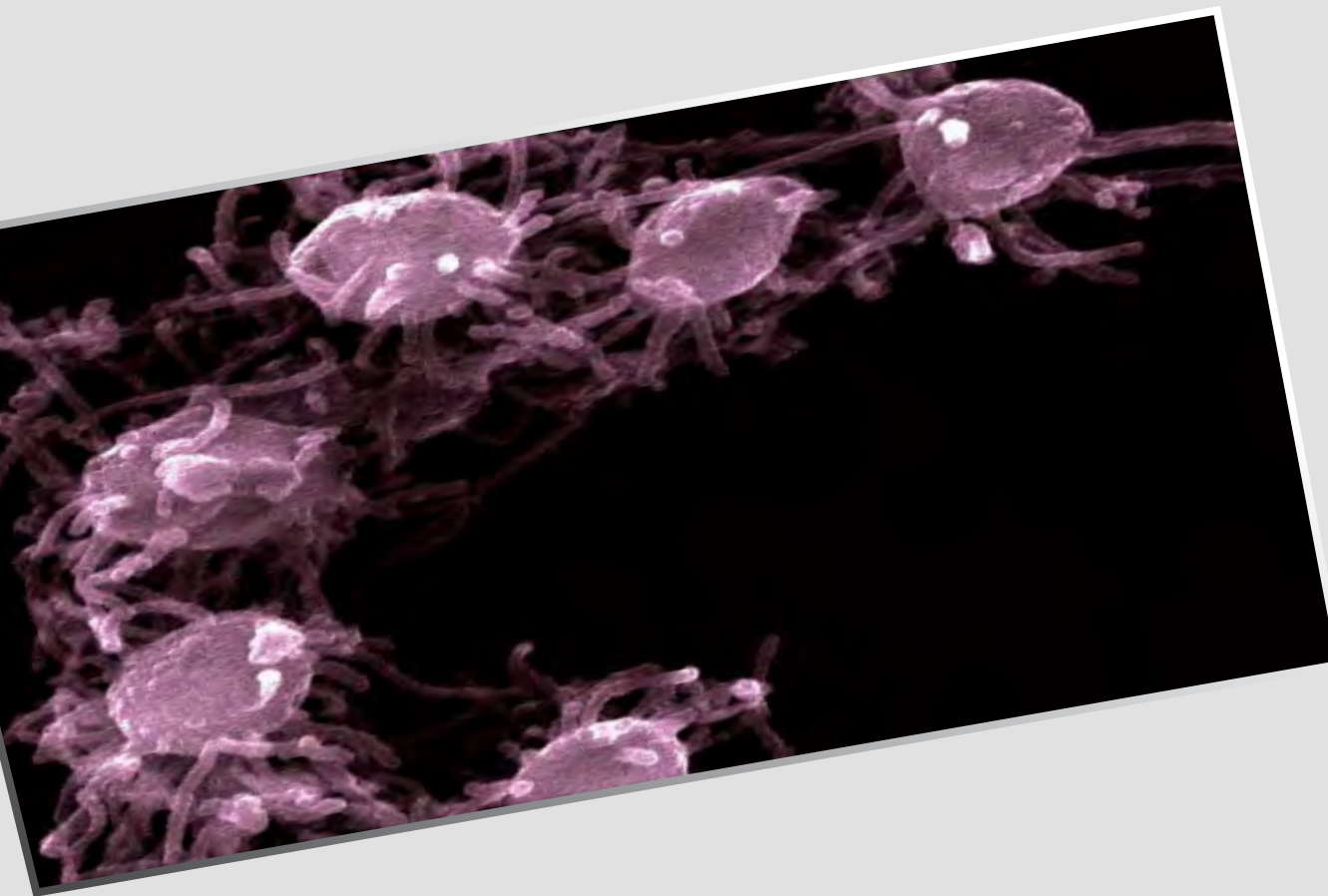
28 students and two teachers from the Oms i de Prat School in Manresa visited the Institute. The students were given the opportunity to learn about the different research lines that are currently being pursued at the IBE and to participate in two hands-on activities.

La ciència al teu món (Science is part of your world)

Funded by: *Secretaria d'Universitats i Recerca. Departament d'Economia i Coneixement. Generalitat de Catalunya* and *Fundación Española para la Ciencia y la Tecnología (FECYT)*.

Several PIs and students of the IBE are collaborating on *La ciència al teu món (LCATM)* outreach project lead by Josefa González. Besides raising awareness of the importance and the implications of Science in everyday life, *LCATM* also aims at conveying the value of a scientific way of thinking and a rational attitude to problems. *LCATM* can be found on the web www.lacienciaalteumon.cat; on twitter, [@LCATMon](https://twitter.com/LCATMon); and on Facebook, *La ciència al teu món*.





IBE RETREAT CHRONICLE



All IBE members convened at the Institut d'Estudis Catalans (Barcelona) on October 23th at the IBE retreat, organized by Salvador Carranza, Ferran Casals and Anna Pérez. The goal of this annual meeting is providing an appropriate environment for the exchanging of ideas and scientific experiences among IBE members, as well to present the IBE scientific activities. The retreat was a big success of participation, where all PhD students and master students had the opportunity of presenting their work to the whole IBE community. The new format established in this edition (giving more prominence to the poster presentation especially by the PhD students) provided an informal atmosphere that was highly appreciated by all participants, and ideal for the exchange of ideas. We can say that after this retreat every IBE member knows who is who in the Institute.

The scientific programs included two invited lectures and two lectures by young IBE researchers. The four talks were very stimulating and represent edge research in their respective fields. Cedric Notredame (from the Center for Genomic Regulation) presented the conference «ENCODE & Evolution»; Daniel Sol (from the Centre de Recerca Ecològica i Aplicacions Forestals) talked on «Unravelling the life history of successful invaders»; Ferran Casals (IBE) presented the talk «Rare is meaningful», on the excess of rare genomic variants

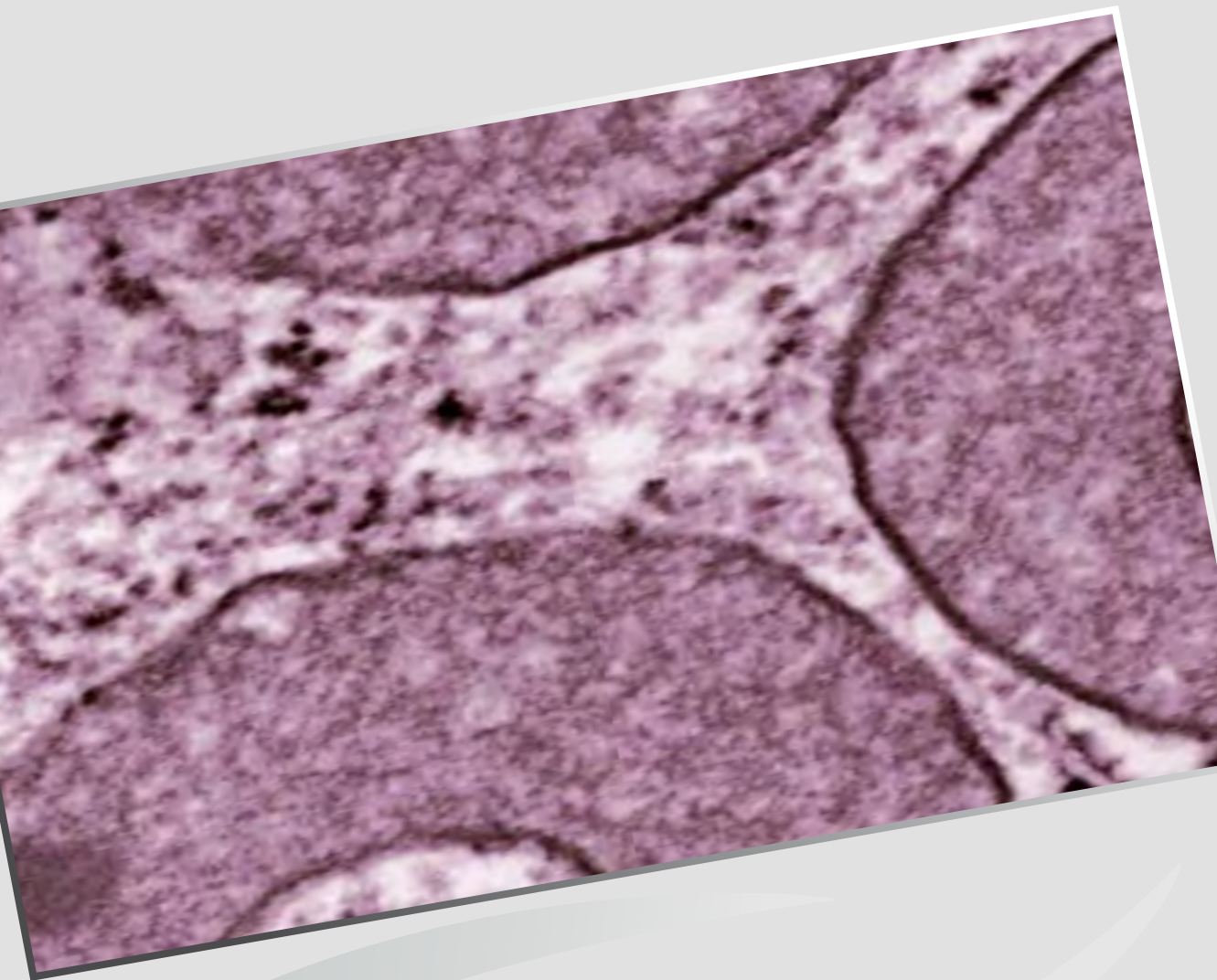
in human populations; and Iñaki Ruiz-Trillo (IBE) introduced «Origin of Multicellularity». As said above, in this retreat special attention was given to the presentation by all PhD and master students of their current work. Three posters sessions were alternated among the main lectures. The 129 attendants were divided in four groups to visit all the posters, where the students presented their work in approximately five minutes. In total, 45 posters from all the IBE research programs were presented: 8 from *Animal Biodiversity and Evolution*, 9 from *Comparative and Computational Genomics*, 12 from *Functional Genomics and Evolution*, 13 from *Population Genetics*, and 3 from *Complex Systems*.





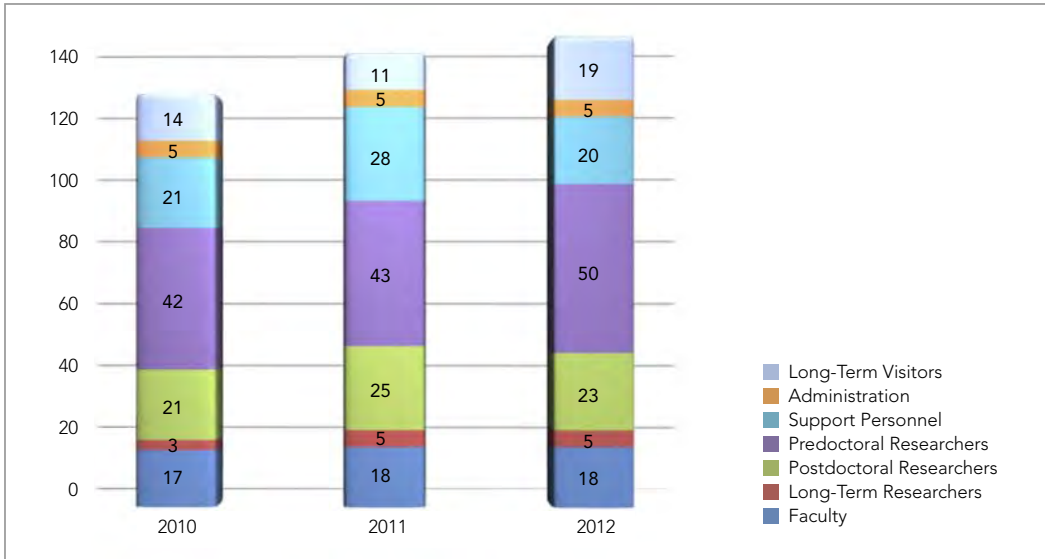
At the end of the day, David Comas and Josefa González presented the «IBE PhD Training and Outreach Program» which, starting in 2013, will be offered to all new IBE PhD students. Finally, several IBE members, mostly students, improvised a dinner near the retreat place. Although the duration of this retreat was shorter than in previous editions, it very successfully accomplished its main goal of providing a calm and informal atmosphere for the fruitful exchange of ideas and a better acquaintance among IBE members.



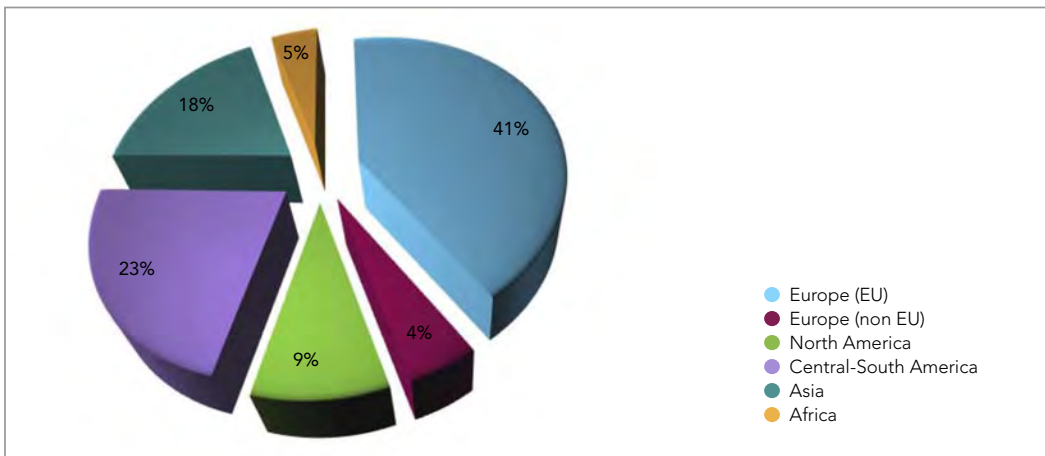


PEOPLE

Personnel distribution per categories

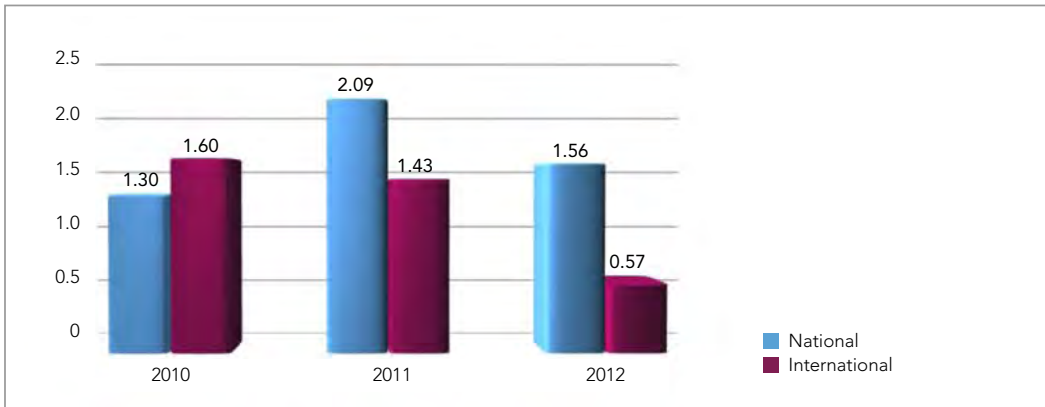


Internationalization. Foreign personnel represent 22% of the total of IBE members (visitors not included). These personnel come from 16 countries from Europe, America, Asia and Africa.

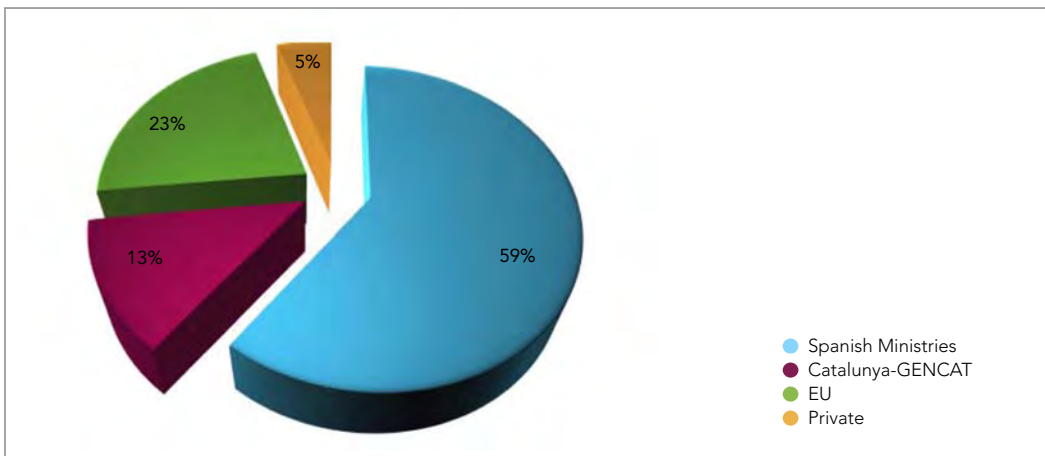


RESOURCES

Competitive funding obtained per year (in M€)

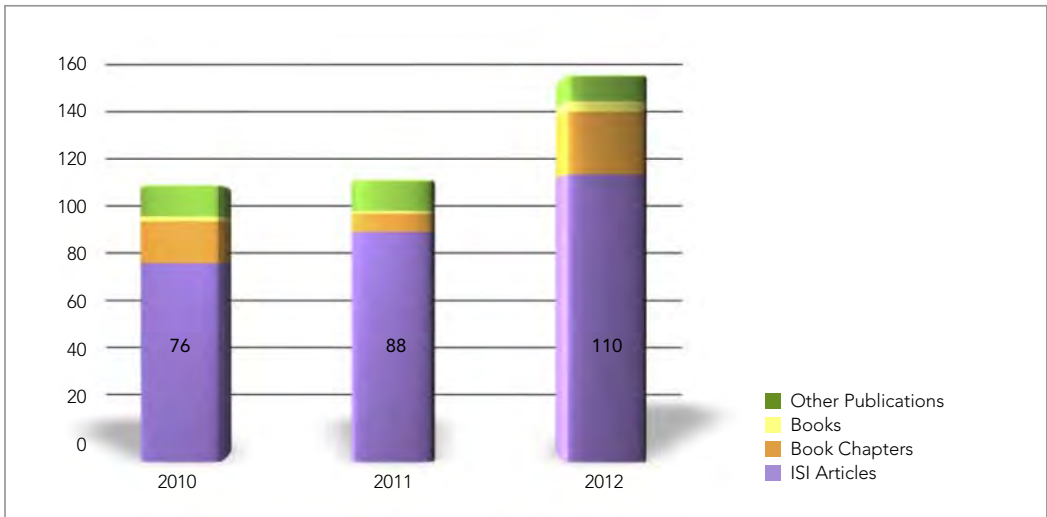


Distribution per agencies and origin of the competitive funding obtained in 2012

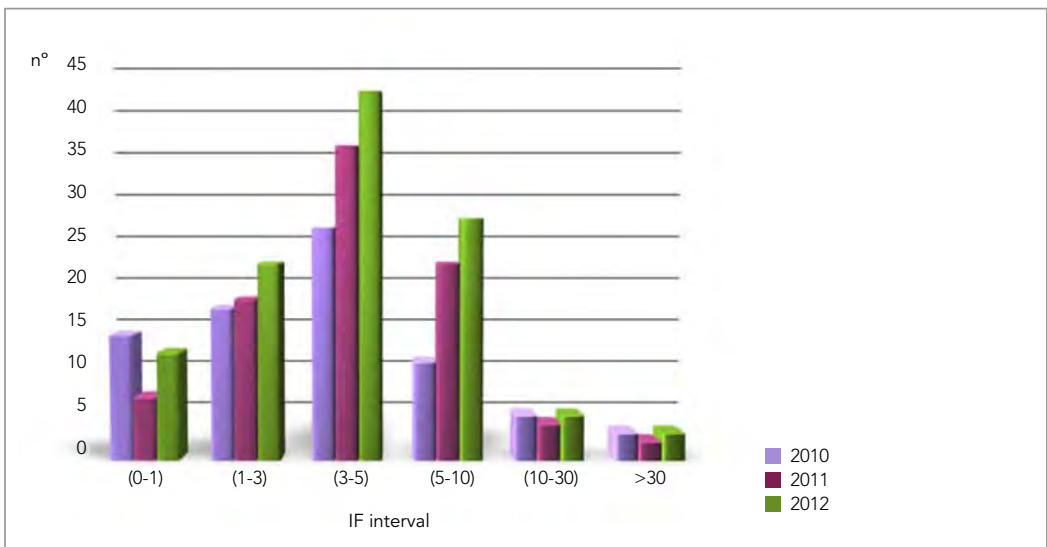


SCIENTIFIC PRODUCTION

Distribution of publications per publication type



Distribution of ISI articles per Impact Factor (IF) intervals





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