Annual Report 2013

ANNUAL REPORT 2013

INSTITUT de BIOLOGIA EVOLUTIVA

CSIC UPF

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FOREWORD

by Xavier Bellés, Director of the IBE

So, that like produces like is the oldest biological observation. What science has accomplished is to tell us that this happens because organisms contain genes in them and that the future organism is written in this somehow. And 'somehow' is what we have to explain. We have to say not somehow, but how.

Sydney Brenner, *My life in science* (2001)

In February 2013, I received a manuscript letter from Sydney Brenner announcing that he was going to attend the 8th European Zebrafish Meeting, which would take place in Barcelona the second week of July, and asking whether he could take advantage of the trip to visit the IBE. This was a great surprise and a great joy for us, and obviously I invited him to visit our Institute, what he did on 10 July 2013. This was a happy coincidence because the IBE was formally founded on 16 July 2008, and thus the visit of Prof. Brenner roughly coincided with our 5th anniversary, as if it were an unexpected and wonderful present.

The visit was first a privilege but also a great pleasure. Bright and restless, he made us think again about the challenges of present science (how to convert "somehow" into "how") and about our job of scientific researchers. We realized that most, practically all of us, gradually lose our enthusiasm along our career, as if it were a kind of ontogenetic development. As young graduate students, we are energetic, resourceful, open minded, and full of ideas, and even bright. However, time runs against (most of) these qualities, and only a few privileged minds are able to keep an age of, say, a 20-year old somewhere in a hiding place of the brain and heart. These singular people are never bound by dominant disciplinary paradigms, and continue questioning the existing information in order to produce new understanding through creative discovery.

These privileged scientists produce breakthrough science and enlarge the field of knowledge in unexplored conceptual territories during their entire careers and lives. Sydney Brenner showed us again that he is one of the best living examples of this attitude.

I said before that the IBE is just 5 years old. At this age, children have a longer attention span, talk a lot, ask many questions, want real adult things, keep art projects, test physical skills and courage with caution, reveal feeling in dramatic play, like to share things with friends, do not like to lose, share and take turns sometimes. In addition to needs from previous years, children at this age require opportunities to engage in problem-solving, become more and more interested in final products, practice teamwork, develop sense of personal competency, learn cooperation by helping and sharing, practice questioning and observing, and acquire basic life skills. Although metaphorically, the assessment shows that our program for the next years is vast and exciting. Five years old may also be an age of consolidation, and we can consider our Institute fully consolidated. There may be a few spoilsports that solemnly declare that the IBE is a sort of abstraction, and will be so until we do not have the building that we all wish. Make no mistake about this point: the building is important and we will fiercely pursuit its construction in order to complete the whole scientific project of the IBE, and it will come in due time. But what make an Institute is an organized group of people that want to be an Institute, and we have achieved that. Along the last 30 years, I have seen many research institutes all around the world, and some were sheltered in nice buildings but swam in mediocrity, whereas others were creative and full of projects, talent and enthusiasm, even though they lacked their own building, or even many important facilities. Technological resources are very important, but can become useless if they are not accompanied by ideas.

2013 has seen a chain of changes of persons in the IBE organization. The representation of the Universitat Pompeu Fabra (UPF) in the Board of Trustees has changed, Jose García Montalvo being replaced by Antoni Bosch, as a consequence of the election of Jaume Casals as new Rector of the UPF. In the same context, the IBE vice director, Arcadi Navarro, was nominated head of the Department of Experimental and Health Sciences of the UPF, and he has been replaced by David Comas in the vice direction of our Institute. This, in turn, triggered the changes of coordinator of the Programs "Population genetics" (now coordinated by Jaume Bertranpetit) and "Comparative and computational genomics" (Carles Lalueza-Fox as new coordinator). Beyond these organization changes, 2013 has been the year when Iñaki Ruiz-Trillo was awarded an ERC consolidator project, and when Tomàs Marquès-Bonet joined the network of EMBO Young Investigators. Income from grants and projects has decreased significantly, but the most remarkable change has been the source of funds, which mostly (88%) come from the EU. The number of publications has remained approximately stable, but quality has increased if measured, for example, by the higher number of papers published in journals with an Impact Factor greater than 10.

And 2014 will be better than 2013 according to the perspectives that we have now, and because, as ever, we will face it being aware of the problems but with energy and resolution. Stated concisely, and borrowing the words of Antonio Gramsci, with the pessimism of the intellect and the optimism of the will.

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). Initially the IBE was created with 11 research groups from the Molecular Biology Department (CID, CSIC) and 6 research groups from the Evolutionary Biology Unit (dCEXS, UPF). Nowadays, IBE activity involves more than a hundred people and 18 research groups distributed in five scientific programs related to Evolutionary Biology research.

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

The IBE project vision, defined as the projection of the long-term future 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 clear molecular and genomic perspective, including human diversity. Establishing the foundations of a multidisciplinary approach, not limited to the evolution from a mere biological approach, but extended to the human sciences in the broadest sense.

GENERAL STRUCTURE

In addition to the classical figures of Director, Vice director, 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

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

Members of the Board of Trustees during 2013:

Antoni Bosch
- UPF Vice chancellor for Economy and Strategic Projects

Lluis Calvo
- CSIC Institutional Coordinator in Catalonia

Francesc Posas
- UPF Vice chancellor for Scientific Policy

José Ramón Urquijo Goitia
- CSIC Vice president 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 to recruit talent and widen the scientific strength of the Institute.

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

The composition of the CCE is as follows.

Chairman

Andrés Moya |
Universitat de València,
València, Spain

Members

Brian Charlesworth |
University of Edinburgh,
Edinburg, UK

Stuart Reynolds |
University of Bath, Bath, UK

Gonzalo Giribet |
Harvard University, Cambridge,
MA, USA

Luis Serrano |
Centre de Regulació
Genòmica, Barcelona, Spain

Eske Willerslev |
University of Copenhagen,
Copenhagen, Denmark

EXECUTIVE BOARD

The IBE Executive Board is composed of 7 members:

IBE Director

- Xavier Belles

IBE Vicedirector

- David Comas

Current Members

- Jaume Bertranpetit • Coordinator of the *"Population genetics"* program
- José Castresana • Coordinator of the *"Animal biodiversity and evolution"* Program
- Carles Lalueza-Fox • Coordinator of the *"Comparative and computational genomics"* program
- Maria-Dolors Piulachs • Coordinator of the *"Functional genomics and evolution"* program
- Ricard Solé • Coordinator of *"Complex systems"* program

General Manager and Board Secretary

- Anna Pérez-Lezaun

GOOD PRACTICES AND SCIENTIFIC INTEGRITY

- Jose Luis Maestro • Tenured Scientist, CSIC
- Elena Bosch • Associate Professor, UPF

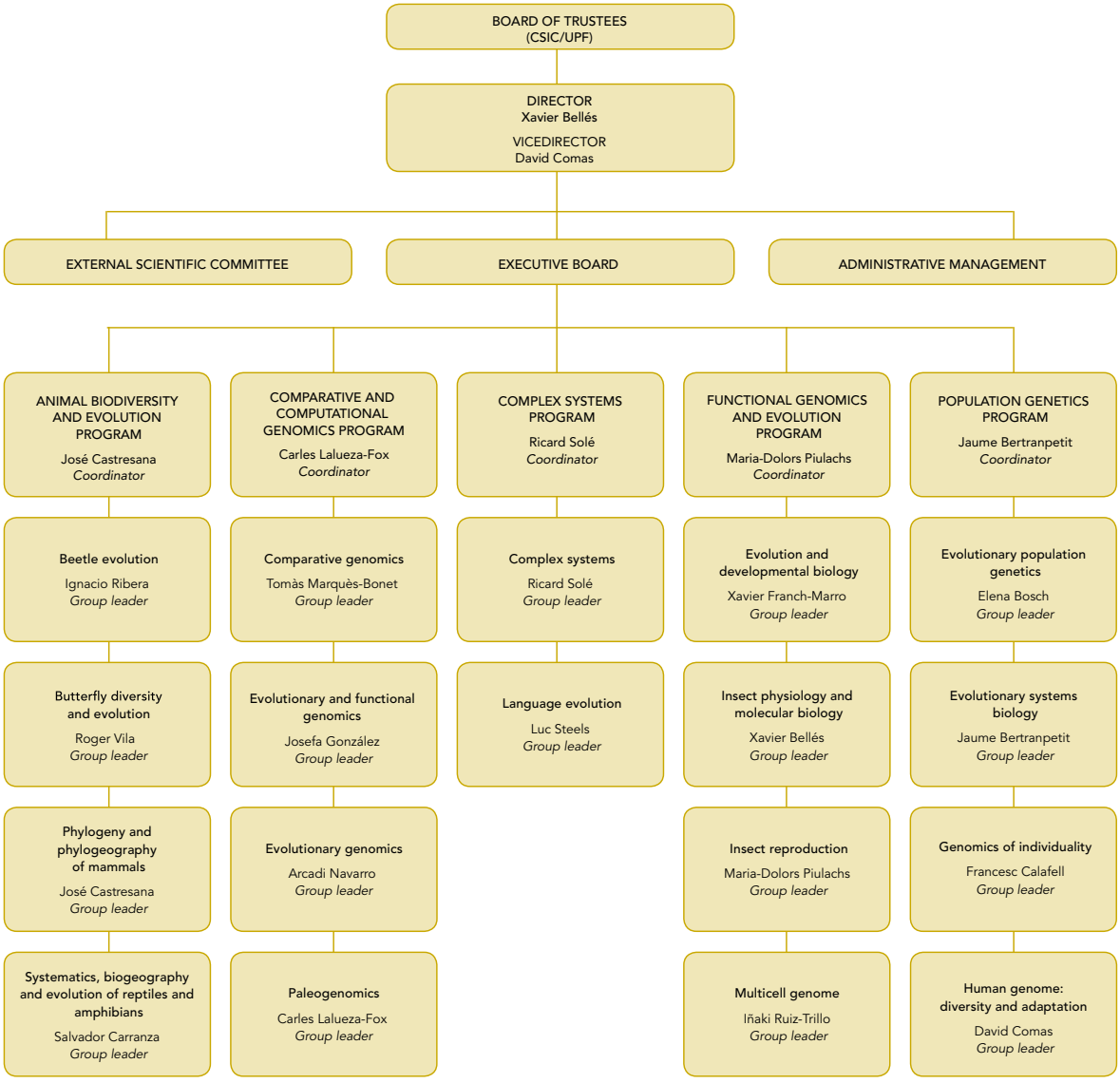
SCIENTIFIC STRUCTURE

As a result of the recommendations that emerged from the *in situ* evaluation by the CCE in March 2012, the structure of research groups was modified, from the former 23 to 18 in 2013. The name and scope of some Scientific Programs were also modified although the number of them remains five.

The new scientific structure distributes its actual eighteen 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 an "Experimental techniques Unit".

MANAGEMENT UNIT

Nowadays, the IBE management unit is composed of 5 people and covers at a micro scale level all 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 UNIT

This unit coordinates the maintenance and use of the insect colonies and of the specialized technical instrumentation and facilities mainly related to the activities of those groups belonging to the Functional genomics and evolution program. Right now, it depends on a staff technician from the CSIC. It is planned that the personnel and functions of this unit should be enlarged in the near future to give support to other programs and technological needs.

Technical support:

| Cristina Olivella | Technical Staff (CSIC)

BIOINFORMATICS UNIT

This unit started formally functioning at the beginning of 2011 with the incorporation of a shared specialized bioinformatician (MICINN-PTA) who joined a group of IT technicians from the research groups of the Comparative and Computational Genomics Program. The Bioinformatics Unit coordinates the support to all IBE Programs in all tasks requiring knowledge on computational biology, particularly in what refers to the growing computational needs of current biological research. The unit offers highly specialized support (installation of software, creation of databases, scripting, etc.) and manages the access of IBE researchers to our local computational cluster, an IBM blade center with more than 400 cores and 500Tb of storage capacity.

Technical support:

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

PROGRAM RESEARCH ASSISTANTS

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

Members:

- Mònica Vallés | Technical Staff (UPF) • Supporting the *"Population genetics"* program and the *"Comparative and computational genetics"* program
- Eva García-Ramallo | UPF Contract • Supporting the *"Complex systems"* program
- Maria Niño | JAETEC CSIC • Supporting *"Comparative and computational genetics"* program
- Laura Gutiérrez | JAETEC CSIC • Supporting UPF genomics core facility

PERSONNEL

At the end of 2013, the IBE had 120 members (visitors not included; Table 1) with a ratio of men to women around 1.66, and an internationalization level of close to 22% foreign members (in postdoctoral researchers this percentage increases up to 33 %).

Table 1. Distribution of IBE personnel by categories. December 2013.

	2013	2012	2011	2010	2009
Faculty	18	18	18	17	16
Long-term researchers * (ICREA, Marie Curie R&C…)	5	5	5	3	3
Postdoctoral researchers	21	23	25	21	17
Predoctoral researchers	48	50	43	42	33
Support personnel (Lab techn., bioinformaticians,)	23	20	23	16	10
Administrative staff	5	5	5	5	5
Long-term visitors (> 1 month)	16	19	11	14	8
TOTAL	136	140	130	118	92

*Marie Curie, Ramón y Cajal or ICREA researchers

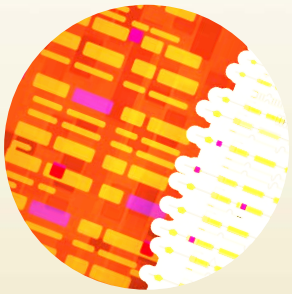
LOCALISATION

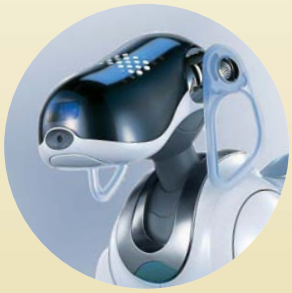
While not having a specific building,
the IBE has two different headquarters:

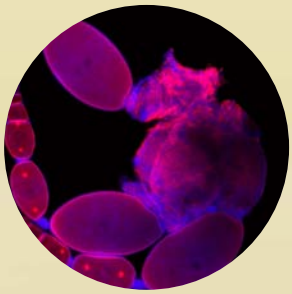
IBE at the CMIMA building:
Passeig Marítim de la Barceloneta, 37-49.
08003 Barcelona, Spain.

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

IBE RESEARCH PROGRAMS







Most of our research is centred on the Mediterranean basin, an area of extreme diversity but also subjected for millennia to strong human pressure. In the image, a view of Itea and the Gulf of Corinth from Delphi, Greece.
© Alexandra Cieslak

PROGRAM
animal biodiversity and evolution

RESEARCH GROUPS

Beetle evolution
Ignacio Ribera, Group leader

Subgroups:

Water and cave 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 insight into the tree of life. The program's specific research interests include the origin and distribution of biodiversity (whether morphological, genetic, ecological or functional), systematics of certain groups, speciation, hybridization, diversification, biogeography, evolutionary ecology, 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

GROUP MEMBERS

Ignacio Ribera, Group Leader

| Research Scientist

| Subgroup: Water and cave beetle evolution

Ignacio Ribera | Research Scientist, CSIC

Rocío Alonso Rodríguez | Laboratory technician, contract associated to project

David García Vázquez, PhD Student | MICINN Scholarship,

Andrey Rudoy, PhD Student, JAE Scholarship | CSIC

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

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

| 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 centred on the ecomorphological and phylogenetic diversification of species-rich lineages of cave and water beetles, with a focus on the origin of the Mediterranean fauna and the macroevolutionary dynamics of geographical ranges. 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 of widespread species of European lotic water beetles
3. Origin and diversification of western European cave beetles
4. Evolution of the complex male genitalia in Hydraenidae
5. Conservation of Iberian water beetles
6. Riffle beetles of the Amazonian region

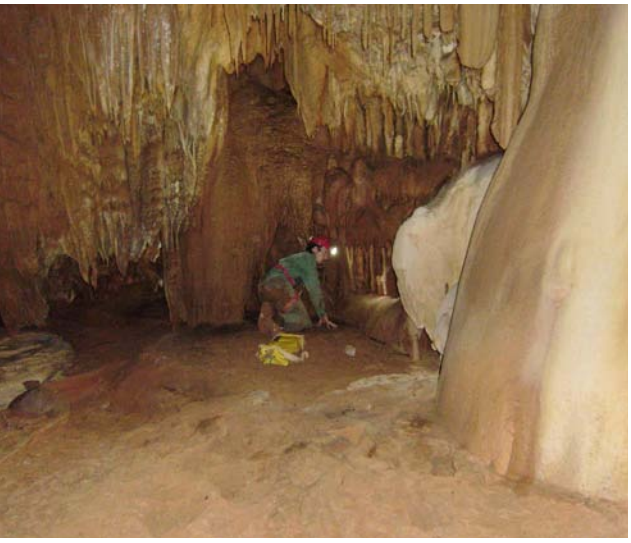


Fig. 1: *A limiting factor when studying subterranean fauna is the access to specimens, which are often rare and difficult to find. In the image, Arnaud Faille, a past member and regular collaborator of our group, searching fauna in "Sa Campana" cave, in Mallorca.*
© Charles Bourdeau

Fig. 2: *Reserva Natura (Managua). One of several private initiatives in Nicaragua to recover and protect the original exuberance of the Mesoamerican dry forest.*
© Anabela Cardoso/Gissela De la Cadena

SUBGROUP: HERBIVORE BEETLE EVOLUTION

1. Hybridization and the evolution of unisexuality
2. Evolution of male-specific genes in thelytokous species
3. Origin and diversification of New Caledonian leaf beetles
4. Systematics and evolution of American Chrysomelinae
5. Characterization of insect-plant associations in tropical dry forests
6. DNA-based species delimitation

PUBLICATIONS 2013

ISI Articles

- Abellán, P., Sánchez-Fernández, D., Picazo, F., Millán, A., Lobo, J.M., and Ribera, I. 2013. Preserving the evolutionary history of freshwater biota in Iberian National Parks. *Biological Conservation* 162: 116-126.
- Barbosa, F.F., Fernandes, A.S., and Oliveira, L.G. 2013. Three new species of *Macrelmis* Motschulsky, 1859 (Coleoptera: Elmidae: Elminae) from the Brazilian Cerrado Biome with updated key for the *Macrelmis* of Brazil. *Zootaxa* 3736: 128-142.
- Faille, A., Casale, A., Balke, M., and Ribera, I. 2013. A molecular phylogeny of Alpine subterranean Trechini (Coleoptera: Carabidae). *BMC Evolutionary Biology* 13: 248.
- Guareschi, S., Coccia, C., Sánchez-Fernández, D., Carbonell, J.A., Velasco, J., Boyero, L., Green, A.J., and Millán, A. 2013. How far could the alien boatman *Trichocorixa verticalis verticalis* spread? Worldwide estimation of its current and future potential distribution. *PLos One* 88 (3): e59757.
- Millán, A., Picazo, F., Fery, H., Moreno, J.L., and Sánchez-Fernández, D. 2013. *Stictonectes abellani* sp. n. (Coleoptera: Dytiscidae: Hydroporinae) from the Iberian Peninsula, with notes on the phylogeny, ecology and distribution of the Iberian species of the genus. *Zootaxa* 3745: 533-550.
- Montelongo, T., and Gómez-Zurita, J. 2013. Morphological and molecular characterization of a new Nearctic species of *Calligrapha* Chevrolat, 1836 (Coleoptera: Chrysomelidae, Chrysomelinae) from Central Mexico. *Proceedings of the Entomological Society of Washington* 115: 369-391.
- Papadopoulou, A., Cardoso, A., and Gómez-Zurita, J. 2013. Diversity and diversification of Eumolpinae (Coleoptera: Chrysomelidae) in New Caledonia. *Zoological Journal of the Linnean Society* 168: 473-495.
- Rizzo, V., Comas, J., Fadrique, F., Fresneda, J., and Ribera, I. 2013. Early Pliocene range expansion of a clade of subterranean Pyrenean beetles. *Journal of Biogeography* 40: 1861-1873.
- Sánchez-Fernández, D., Abellán, P., Picazo, F., Millán, A., Ribera, I., and Lobo, J.M. 2013. Do protected areas represent species' optimal climatic conditions? A test using Iberian water beetles. *Diversity and Distributions* 19: 1407-1417.

Fig. 3: *Although the focus is often on the most spectacular and colourful species, most beetles are small, brown and inconspicuous – like this new species of* Bhyrrinus *(family Limnichidae) from the island of Socotra, only a couple of millimetres long, in the process of being described by our group.* © Jiři Hájek

- Trizzino, M., Jäch, M.A., Audisio, P., Alonso, R., and Ribera, I. 2013. A molecular phylogeny of the cosmopolitan hiperdiverse genus *Hydraena* Kugelann (Coleoptera, Hydraenidae). *Systematic Entomology* 38: 192-208.

Books/Book Chapters

- Millán, A., Picazo, F., Sánchez-Fernández, D., Abellán, P., and Ribera, I. 2013. Los Coleópteros acuáticos amenzados (Coleoptera). In: Díaz Ruano, F., Tierno de Figueroa, J.M., Tinaut, J.A. (Eds). Los insectos de Sierra Nevada. 200 años de historia. Asociación Española de Entomología, pp. 442-456.

Other Publications

- Barbosa, F.F., Fernandes, A.S., and Oliveira, L.G. 2013. Taxonomic key for the genera of Elmidae (Coleoptera, Byrrhoidea) ocurring in Goiás State, Brazil, including new records and distributional notes. *Revista Brasileira de Entomologia* 57: 149-156.
- Gómez-Zurita, J. 2013. The beauty of beetles. *International Innovation*, October 2013: 86-88.

FUNDED PROJECTS

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*

Project Title: *Atlas y Libro Rojo de los Coleópteros Acuáticos de España*
Financed by: *Ministerio del Medio Ambiente*
Years: *2010-2013*
PI: *Andrés Millán (Universidad de Murcia)*

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: *Grup de Recerca Consolidat en Sistemàtica i Evolució Zoològica - ZOOSYSEVO*
Financed by: *Generalitat de Catalunya*
Years: *2009-2013*
PI: *Salvador Carranza*

Fig. 4: *Anabela at work in Neblina del Bosque (Estelí, Nicaragua). Herbivore beetles are usually collected knocking them down the plants they feed upon using beating trays as in the picture, or sweeping nets.*
© Anabela Cardoso/Gissela De la Cadena

Fig. 5: *Brood of tortoise beetle larvae collected in Nicaraguan tropical dry forest. Cassidinae constitute an example of subsocial beetles: siblings typically display a gregarious behaviour after hatching their eggs, sometimes associated to maternal care, to increase their chances of survival.*
© Anabela Cardoso/Gissela De la Cadena

GROUP

BUTTERFLY DIVERSITY AND EVOLUTION

GROUP MEMBERS

Roger Vila, Group Leader

| CSIC Researcher

Raluca Voda, PhD Student | FPU Scholarship, MEC
Gerard Talavera, Visiting Post-doc | St. Petersburg University
Vlad Dinca, Visiting Post-doc | Stockholm University
Mònica Navarro | Undergraduate Student, Practicum UB
Marga Marín | Laboratory Technician

RESEARCH OUTLINE

We study butterfly diversity patterns in time and space, and their evolutionary causes. Our final goal is to answer general questions regarding chromosomal evolution, the limits between species, and the link between phylogeography and ecology. When and following what route a group of tiny butterflies colonized the New World, how parasitism evolved from a friendly association between species, or if a given population constitutes a new species worth protecting are examples of questions we address.

RESEARCH 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 has been the first country with all butterfly species barcoded), Iberian Peninsula and Italy. We have recently started the challenging 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.

Fig. 1: *In July Raluca and Roger, together with our collaborator Leonardo Dapporto, made an expedition to Tunisia. You can read about our adventures in Raluca's blog* "En busca de papallones per la Mediterrània" *within the webpage La Ciència al teu Món.* http://lacienciaalteumon.cat/en-busca-de-papallones-per-la-mediterranea/. © Raluca Voda

2. Uncovering of cryptic butterfly biodiversity in Europe

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

3. Ecological factors determining butterfly biogeography

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

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

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

Fig. 2: *In October Gerard was invited to a course on Neotropical Biogeography held in French Guiana. He took the chance and collected many interesting butterflies. In this beach he collected the southernmost known specimens of* Vanessa cardui *in the New World, a migratory species that he is studying during his post doctorate.* © Gerard Talavera

Fig. 3: *Raluca and Vlad collected in Bulgaria and Greece in July-August. The Balkan Peninsula harbours a very diverse butterfly fauna and we are now focusing on sampling that region.* © Vlad Dinca

PUBLICATIONS 2013

ISI Articles

- Carnicer, J., Stefanescu, C., Vila, R., Dinca, V., Font, X., and Peñuelas, J. 2013. A unified framework for diversity gradients: the adaptive trait continuum. *Global Ecology and Biogeography* 22: 6-18. (Featured in the journal cover)
- Dapporto, L., Ramazzotti, M., Fattorini, S., Talavera, G., Vila, R., Dennis, R.L.H. 2013. recluster: an unbiased clustering procedure for beta-diversity turnover. *Ecography* 36: 1070-1075.
- Dinca, V., Runquist, M., Nilsson, M., and Vila, R. 2013. Dispersal, fragmentation and isolation shape the phylogeography of the European lineages of *Polyommatus (Agrodiaetus) ripartii* (Lepidoptera: Lycaenidae). *Biological Journal of the Linnean Society* 109: 817-829.
- Dinca, V., Wiklund, C., Lukhtanov, V.A., Kodandaramaiah, U., Norén, K., Dapporto, L., Wahlberg, N., Vila, R., and Friberg, M. 2013. Reproductive isolation and patterns of genetic differentiation in a cryptic butterfly species complex. *Journal of Evolutionary Biology* 26: 2095-2106. (Featured inthe journal cover)
- Giraldo, C.E., Willmott, K.R., Vila, R., Uribe, S.I. 2013. Ithomiini Butterflies (Lepidoptera: Nymphalidae) of Antioquia, Colombia. *Neotropical Entomology* doi: 10.1007/s13744-012-0102-4.
- Sañudo-Restrepo, C.P., Dinca, V., Talavera, G., Vila, R. 2013. Biogeography and systematics of *Aricia* butterflies (Lepidoptera, Lycaenidae). *Molecular Phylogenetics and Evolution* 66: 369-379.
- Talavera, G., Dinca, V., Vila, R. 2013. Factors affecting species delimitations with the GMYC model: insights from a butterfly survey. *Methods in Ecology and Evolution* 4: 1101-1110.
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- Talavera, G., Lukthanov, V.A., Rieppel, L., Pierce, N.E., Vila, R. 2013. In the shadow of phylogenetic uncertainty: the recent diversification of the *Lysandra* butterflies through chromosomal change. *Molecular Phylogenetics and Evolution* 69: 469-478.

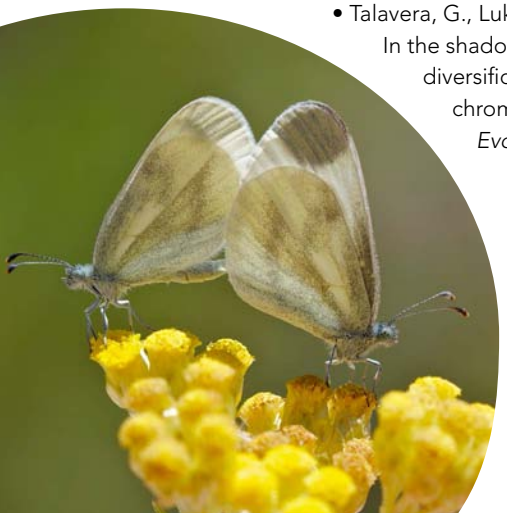


Fig. 4: Leptidea *butterflies are one of our most cherished study models. This year we have published results of female-choice experiments that confirm the existence of three cryptic species in Europe and discovered a population in the Balearic Islands after many years of searching for it. This image, which could be entitled "a pair in copula in Ibiza" was selected as the journal cover.* © Roger Vila

Fig. 5: *Vlad and Roger attended the 14th Congress of the European Society for Evolutionary Biology in Lisbon, Portugal and also collected butterflies for our collection. August was extremely hot and forest fires were a danger, like this one in Serra da Estrela.* © Roger Vila

FUNDED PROJECTS

Project Title: *Faunal genetic comparisons to infer large-scale biogeographical patterns: the colonization of Western Mediterranean islands by butterflies*
Financed by: *Spanish Ministerio de Ciencia e Innovación (CGL2010-21226/BOS)*
Years: *2011-2013*
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*

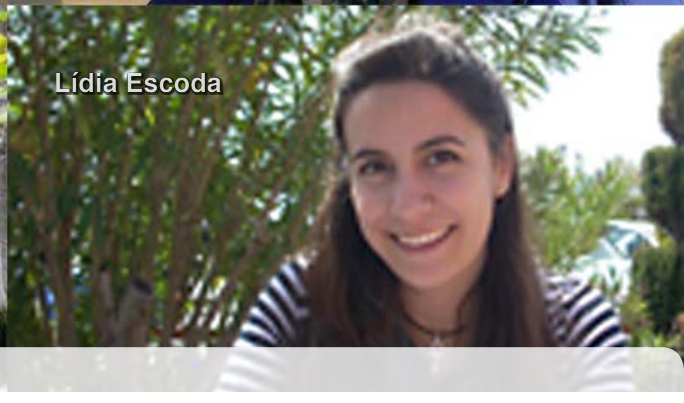
Project Title: *How climate change and extreme drought events disrupt Mediterranean food webs: an eco-evolutionary analysis*
Financed by: *Netherlands Organization for Scientific Research (NWO)*
Years: *2012-2015*
PI: *Jofre Carnicer*

Project Title: *Grup de Genòmica, Bioinformàtica i Evolució*
Financed by: *Suport als Grups de Recerca de Catalunya, Generalitat de Catalunya (SGR 2009-0088)*
Years: *2009-2013*
PI: *Alfredo Ruiz*

Fig. 6: *We are intensely studying the diversity and biogeography of West Mediterranean butterflies. In the picture,* Aglais ichnusa, *a spectacular Sardo-Corsican endemic species.* © Vlad Dinca

GROUP

PHYLOGENY AND PHYLOGEOGRAPHY OF MAMMALS



GROUP MEMBERS

José Castresana, Group Leader

| Research Scientist, CSIC

Marina Querejeta, PhD Student | FPI Fellowship, MINECO

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

Lídia Escoda Assens, Master Student | Universitat de Barcelona

RESEARCH OUTLINE

Our main goal is the application of phylogenetic and genomic analyses to study animal biodiversity and evolution. Using multiple markers as well as next-generation sequencing techniques, 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 estimate species trees and to analyze different speciation scenarios. Finally, since phylogenies are one of the main tools of our work, we are always trying to improve phylogenetic analyses by introducing new methodologies and programs.

RESEARCH SUBLINES

1. Phylogeny and speciation of mammals studied with multiple genomic markers

The reconstruction of species trees of closely related species based on multiple genomic markers can help to estimate accurate speciation times, to study patterns of genetic diversity and gene flow and, in general, to better understand how speciation occurred in mammals. 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 such as water shrews of the genus *Neomys*, shrews of the genus *Sorex*, and Mediterranean voles of the genus *Microtus*. 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. Conservation genomics 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 and is endangered in a large part of its distribution range. We are currently studying several aspects of the phylogeography and population history of this unique species using mitochondrial and nuclear data as well as next-generation sequencing techniques. 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 different institutions. The results we are obtaining may have crucial implications for the conservation of this species.

3. Methodological Aspects of Phylogenetic Reconstruction

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 such as diversification patterns. Furthermore, we are interested in methodologies at the interphase between phylogenetics, coalescence and population genetics, and in the estimation of species trees from gene trees. The software that we develop is made freely available online.

PUBLICATIONS 2013

ISI Articles

- Igea, J., Aymerich, P., Fernández-González, A., González-Esteban, J., Gómez, A., Alonso, R., Gosálbez, J., and Castresana, J. 2013. Phylogeography and postglacial expansion of the endangered semi-aquatic mammal *Galemys pyrenaicus*. *BMC Evolutionary Biology* 13, 115.

Book/Book Chapters

- Castresana, J., Igea, J., Aymerich, P., Fernández-González, A., and Gosálbez, J. 2013. Filogeografía del desmán ibérico (*Galemys pyrenaicus*) y su distribución en la Red de Parques Nacionales. In: Proyectos de investigación en parques nacionales: 2009-2012. Organismo Autónomo de Parques Nacionales (Madrid). pp 143-154.

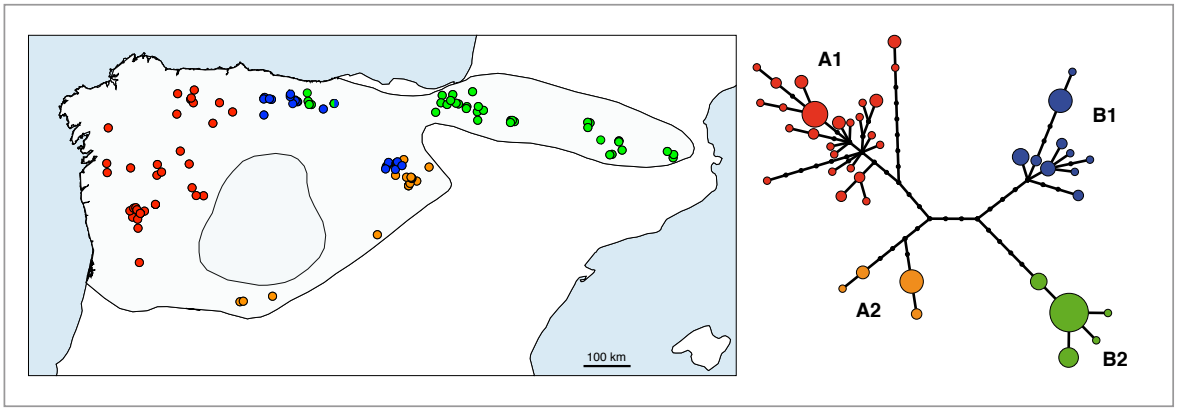


Fig. 1: *We have analyzed the phylogeography of the Pyrenean desman using mitochondrial genes and nuclear introns. The introns showed very low variability but the results with the mitochondrial data revealed a strong phylogeographic structure, with the desman populations divided into four main groups. The figure shows the northern part of the Iberian Peninsula with the historical distribution area of the Pyrenean desman in grey. Dots indicate the Pyrenean desman samples used and colors denote the four mitochondrial lineages recovered in the tree of mitochondrial haplotypes: A1, A2, B1 and B2.*

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: *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

João Campos, PhD Student co-supervised with Dr. José Carlos Brito, 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

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

João 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

Emilio Valbuena Ureña, PhD Student | Teaching Assistant UAB, Barcelona

Marc Simó, MSc Student | Master in Biodiversity, University of Barcelona

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

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 continental systems; 4) despite their small geographical size they can contain a diversity of habitats; and 5) they are often geologically dynamic with historical and contemporary volcanic and erosional activity. At the moment we are investigating both oceanic and continental reptile islands faunas from several places in the world including the Canary Islands and Cape Verde in the Atlantic Ocean and the Socotra archipelago in the Indian Ocean.

PUBLICATIONS 2013

ISI Articles

- Beukema, W., de Pous, P., Donaire-Barroso, D., Bogaerts, S., Garcia-Porta, J., Escoriza, D., Arribas, O., El Mouden, H., and Carranza, S. 2013. Review of the systematics, distribution, biogeography and natural history of Moroccan amphibians. *Zootaxa* 3661: 1-60.
- Crochet, P.A., and Metallinou, M. 2013. Correction to "Nomenclature of African species of the genus *Stenodactylus* (Squamata: Gekkonidae)" by Metallinou and Crochet (2013). *Zootaxa* 3710: 099.
- de Pous, P., Metallinou, M., Donaire-Barroso, D., Carranza, S., and Sanuy, D. 2013. Integrating mtDNA analyses and ecological niche modelling to infer the evolutionary history of *Alytes maurus* Amphibia; Alytidae) from Morocco. *Herpetological Journal* 23: 153-160.
- Garcia-Porta, J., and Ord, T.J. 2013. Key innovations and island colonization as engines of evolutionary diversification: a comparative test with the Australasian diplodactyloid geckos. *Journal of Evolutionary Biology* 26: 2662-2680.
- Lapiedra, O., Sol, D., Carranza, S., and Beaulieu, J.M. 2013. Behavioural changes and the adaptive diversification of pigeons and doves. *Proceedings of the Royal Society of London*, Series B 280: 20122893.
- Litvinchuk, S.N., Crottini, A., Federici, S., de Pous, P., Donaire, D., Andreone, F., Kalezić, M.L., Džukić, G., Veith, M., Lada, G.A., Borkin, L.J. and Rosanov, J.M. 2013. Phylogeographic patterns of genetic diversity in the common spadefoot toad, *Pelobates fuscus*, reveals evolutionary history, postglacial range expansion and secondary contact. *Organisms Diversity and Evolution* 13: 433-451.
- Metallinou, M., and Carranza, S. 2013. New species of *Stenodactylus* (Squamata: Gekkonidae) from the Sharqiyah Sands in northeastern Oman. *Zootaxa* 3745: 449-468.
- Metallinou, M., and Crochet, P.A. 2013. Nomenclature of African species of the genus *Stenodactylus* (Squamata: Gekkonidae). *Zootaxa* 3691: 365-376.
- Ramírez, O.*, Gómez-Díaz, E.*, Olalde, I., Illera, J.C., Rando, J.C., González-Solís, J., and Lalueza-Fox, C. 2013. Population connectivity buffers genetic diversity loss in a seabird. *Frontiers in Zoology* 10: 28 (*Authors contributed equally to this work)
- Rato, C., Carranza, S., and Harris, D.J. 2013. Evolutionary patterns of the mitochondrial genome in the Moorish gecko, *Tarentola mauritanica*. *Gene* 512: 166-173.

- Šmíd, J., Carranza, S., Kratochvíl, L., Gvoždík, V., Karim Nasher, A., and Moravec, J. 2013. Out of Arabia: A complex biogeographic history of multiple vicariance and dispersal events in the gecko genus *Hemidactylus* (Reptilia: Gekkonidae). *PLos One* 8: e64018.
- Šmíd, J., Moravec, J., Kratochvíl, L., Gvoždík, V., Karim Nasher, A., Busais, S.M., Wilms, T., Shobrak, M.Y., and Carranza, S. 2013. Two newly recognized species of *Hemidactylus* (Squamata, Gekkonidae) from the Arabian Peninsula and Sinai, Egypt. *ZooKeys* 355: 79-107.
- Stefan, L., Gómez-Díaz, E., and Mironov, S. 2013. Three new species of the feather mite subfamily Ingrassiinae (Acari: Xolalgidae) from petrels (Procellariiformes: Procellariidae). *Zootaxa* 3682: 105-120.
- Valbuena-Ureña, E., Amat, F., and Carranza, S. 2013. Integrative phylogeography of *Calotriton* newts (Amphibia, Salamandriade), with special remarks on the conservation of the endangered Montseny brook newt (*Calotriton arnoldi*). *PLos One* 8: e62542.
- Vasconcelos, R., Brito, J., Carranza, S., and Harris, D.J. 2013. Review of the distribution and conservation status of the terrestrial reptiles of the Cape Verde Islands. *Oryx* 47: 77-87.

Other Publications

- Amat, F., Carbonell, F., Carranza, S., and Guinart, D. 2013. Tritón del Montseny: el anfibio mas amenazado de Europa occidental. *Quercus* 328: 32-37.
- Bogaerts, S., Donaire-Barroso, D., Pasmans, F., Carranza, S., and Böhme, W. 2013. Do North African Fire Salamanders, *Salamandra algira*, occur in Tunisia?. *Herpetology Notes* 6: 301-306.

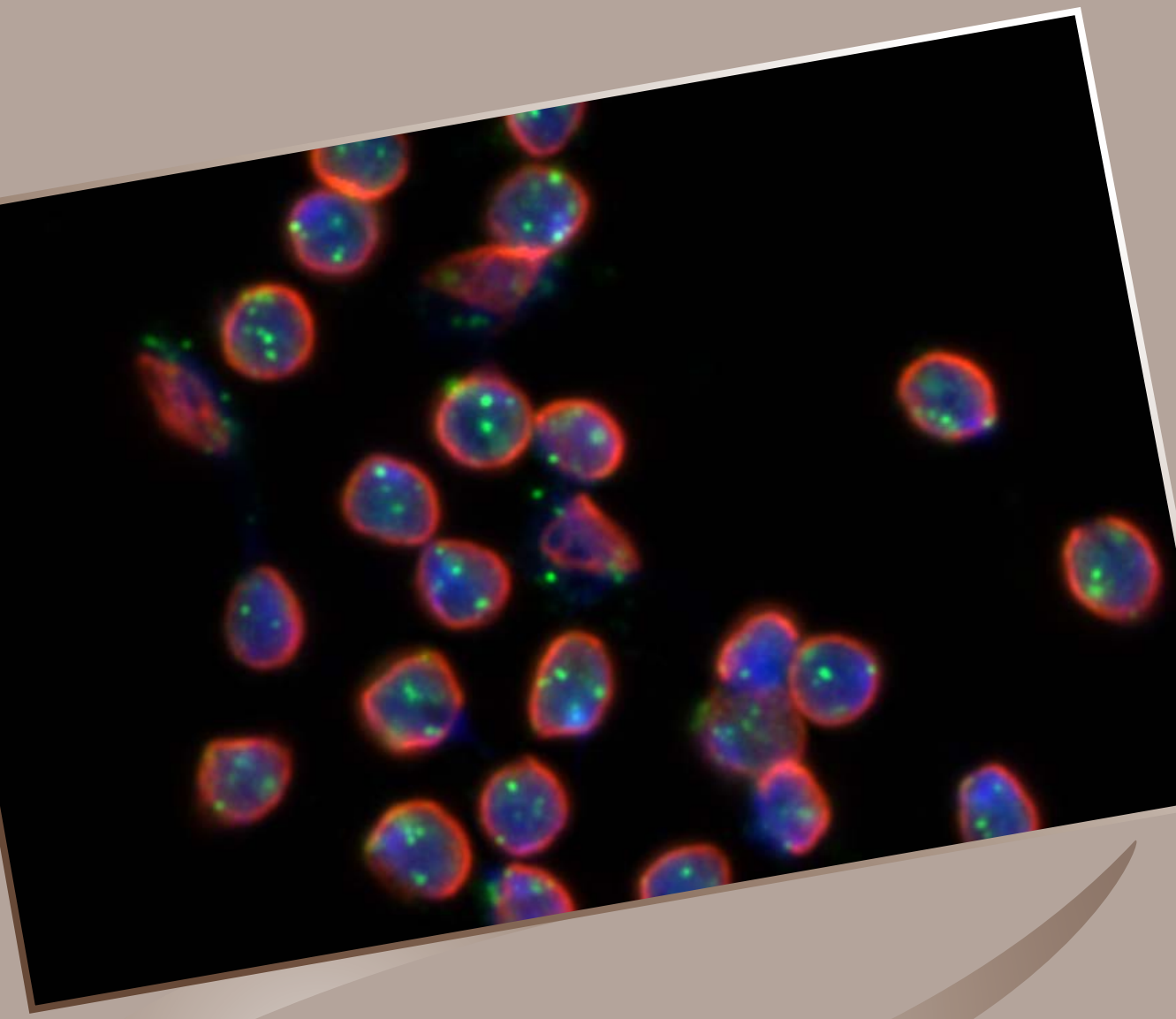
FUNDED PROJECTS

Project Title: *Zoological Systematics and Evolution Research Group - ZOOSYSEVO*
Financed by: *Generalitat de Catalunya (AGAUR) (2009SGR1462)*
Years: *2009-2013*
PI: *Salvador Carranza*

Project Title: *Parasite evolution on islands: reptiles and their parasites as a model study (PARIS)*
Financed by: *European Commission, FP7-PEOPLE-Reintegration Grants. ERG-PARIS-276838*
Years: *2010-2013*
PI: *Elena Gómez-Díaz*

Project Title: *Olvidados en el océano: los reptiles del Archipiélago de Socotra como modelo para el estudio de la Biogeografía, Evolución y conservación en islas*
Financed by: *Ministerio de Economia y Competitividad MINECO (CGL2012-36970)*
Years: *2013-2015*
PI: *Salvador Carranza*

Project Title: *Field study for the conservation of the reptiles of Oman*
Financed by: *Ministry of Environment and Climate Affairs (Sultanate of Oman). REF: 22412027*
Years: *2013*
PI: *Salvador Carranza*



PROGRAM
comparative and computational genomics

RESEARCH GROUPS

Comparative genomics
Tomàs Marquès-Bonet, Group leader

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

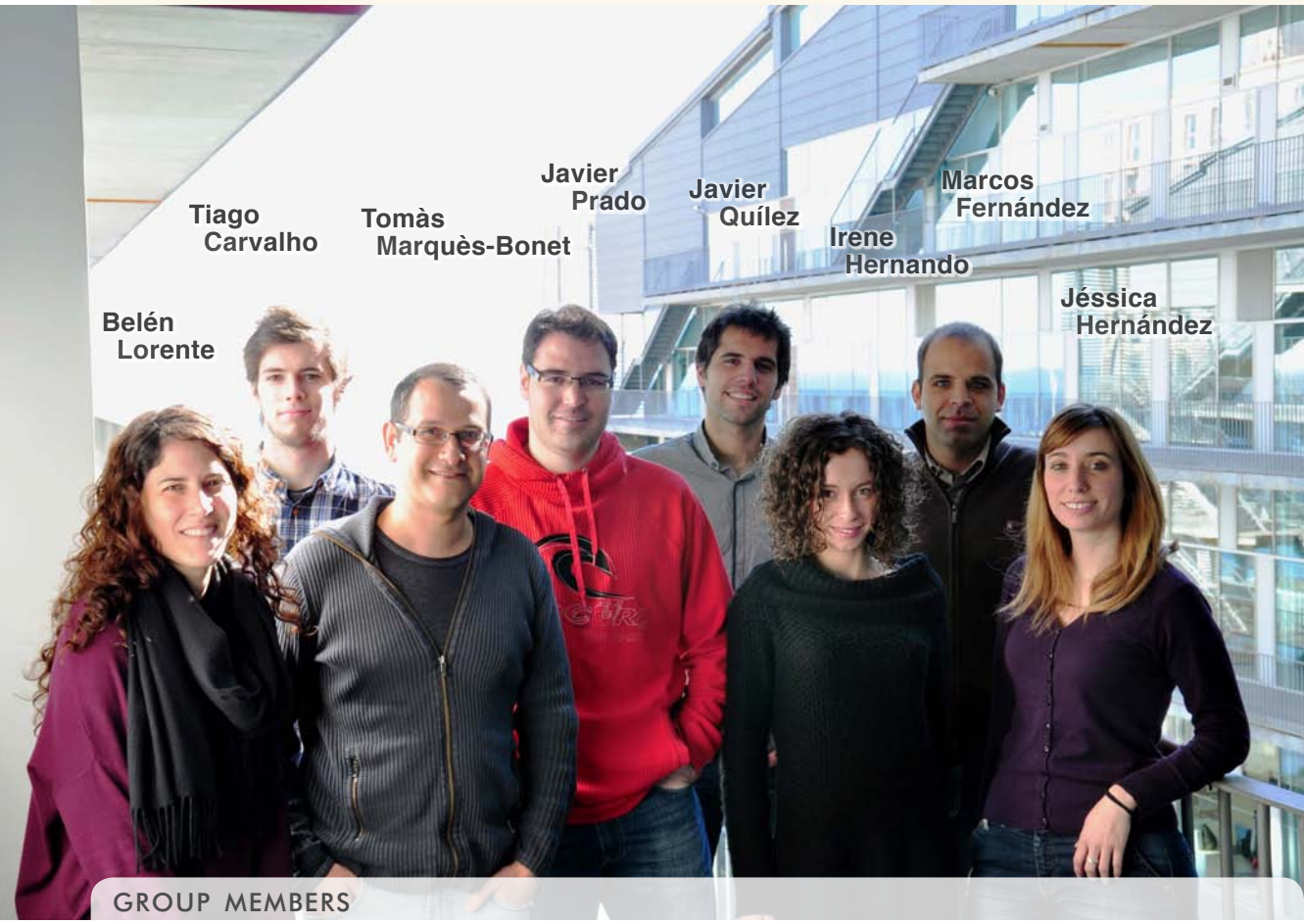
Paleogenomics
Carles Lalueza-Fox, Group leader

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 nuclerical level.

GROUP
COMPARATIVE GENOMICS



GROUP MEMBERS

Tomàs Marquès-Bonet, Group Leader

| ICREA Research Professor

Belén Lorente, Post-doc | ERC StGt Project Contract
Javier Prado, PhD Student | FI Scholarship, MEC
Irene Hernando, PhD Student | FI Scholarship, Generalitat
Tiago Carvalho, PhD Student | ERC StGt Project Contract
Raquel Garcia, PhD Student | FI Scholarship, Generalitat
Jéssica Hernadez, PhD Student | FPI Scholarship, MEC

RESEARCH OUTLINE

Our main line of research is centered in the discovery of the extent of all kinds of genome variation within different phenotypically genomes. Specifically, we study genome variation (centered on copy-number variations or CNVs), gene expression, and epigenetic differences in the human species in the context of great ape evolution and other mammalian genomes such as canids. The goal is to create an integrated view of genome evolution by studying changes in the composition, frequency, size, and location at every major branch point of recent human evolution.

RESEARCH SUBLINES

1. Genomic variation in ape genomes

Despite international efforts to characterize thousands of human genomes to understand the extent of structural variants in the human species, primates (our closest relatives) have somehow been forgotten. Yet, they are the ideal set of species to study the evolution of these features from both the 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 the variation within species and compare it with proper perspective with the differences among species.

2. 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 recent years. However, little has been done to combine a three way analysis going from genomic variants, to gene expression and epigenetics in non-human primates. In the coming years I am planning to use different tissues from the same individuals comparing human, chimps, and rhesus macaque to explore the relationship of these three layers of complexity.

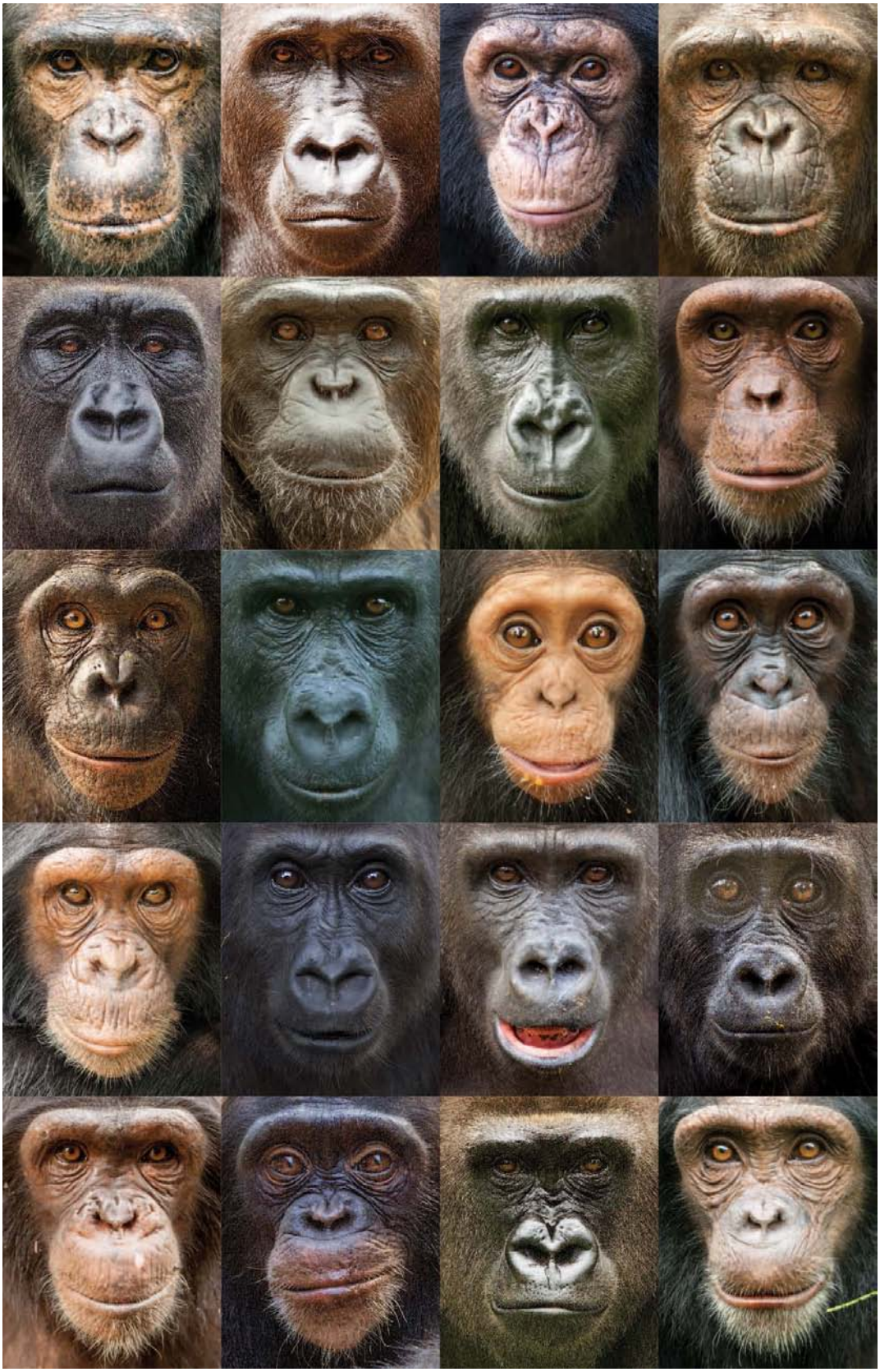
3. Canid evolution

The domestic dog has been widely recognized as an important organism for studying the relationship between selection, genome variation, and phenotypic diversity. Both dogs and wolves have been extensively surveyed using mtDNA, microsatellites, and SNPs, but structural variation, including variation in multicopy gene families, has not been fully characterized in canines.

ISI Articles

- Giannuzzi, G., Siswara, P., Malig, M., Marques-Bonet, T., NISC Comparative Sequencing Program, Mullikin, J.C., Ventura, M., and Eichler, E.E. 2013. Evolutionary dynamism of the primate LRRC37 gene family. *Genome Research* 23 (1): 46-59.
- Hernando-Herraez, I., Prado-Martínez, J., Garg, P., Fernandez-Callejo, M., Heyn, H., Hvilsom, C., Navarro, A., Esteller, M., Sharp, A.J., Marques-Bonet, T. 2013. Dynamics of DNA Methylation in Recent Human and Great Ape Evolution. *PLos Genetics* 9 (9) e1003763.
- Hormozdiari, F., Konkel, M.K., Prado-Martinez, J., Chiatante, G., Herraez, I.H., Walker, J.A., Nelson, B., Alkan, C., Sudmant, P.H., Huddleston, J., Catacchio, C.R., Ko, A., Malig, M., Baker, C., Genome Project, G.A., Marques-Bonet, T., Ventura, M., Batzer, M.A., and Eichler, E.E. 2013. Rates and patterns of great ape retrotransposition. *Proceedings of the National Academy of Sciences USA* 110: 13457-13462.
- Lorente-Galdos, B., Bleyh. J., Santpere, G., Vives, L., Ramirez, O., Hernandez, J., Anglada, R., Cooper, G.M., Navarro, A., Eichler, E.E., and Marques-Bonet, T. 2013. Accelerated exon evolution within primate segmental duplications. *Genome Biology* 14 (1): R9.
- Prado-Martinez, J., Hernando-Herraez, I., Lorente-Galdos, B., Dabad, M., Ramirez, O., Baeza-Delgado, C., Morcillo-Suarez, C., Alkan, C., Hormozdiari, F., Rainieri, E., Estelle, J., Fernandez-Callejo, M., Valles, M., Ritscher, L., Schoneberg, T., de la Calle-Mustienes, E., Casillas, S., Rubio-Acero, R., Mele, M., Engelken, J., Caceres, M., Gomez-Skarmeta, J., Gut, M., Bertranpetit, J., Gut, I., Abello, T., Eichler, E., Mingarro, I., Lalueza-Fox, C., Navarro, A., and Marques-Bonet, T. 2013. The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild. *BMC Genomics* 14 (1): 363.
- Prado-Martinez, J., Sudmant, P.H., Kidd, J.M., Li, H., Kelley, J.L., Lorente-Galdos, B., Veeramah, K.R., Woerner, A.E., O´Connor, T.D., Santpere, G., Cagan, A., Theunert, C., Casals, F., Laayouni, H., Munch, K., Hobolth, A., Halager, A.E., Malig, M., Hernandez-Rodriguez, J., Hernando-Herraez, I., Prufer, K., Pybus, M., Johnstone, L., Lachmann, M., Alkan, C., Twigg, D., Petit, N., Baker, C., Hormozdiari, F., Fernandez-Callejo, M., Dabad, M., Wilson, M.L., Stevison, L., Camprubi, C., Carvalho, T., Ruiz-Herrera, A., Vives, L., Mele, M., Abello, T., Kondova, I., Bontrop, R.E., Pusey, A., Lankester, F., Kiyang, J.A., Bergl, R.A., Lonsdorf, E., Myers, S., Ventura, M., Gagneux, P., Comas, D., Siegismund, H., Blanc, J., Agueda-Calpena, L., Gut, M., Fulton, L., Tishkoff, S.A., Mullikin, J.C., Wilson, R.K., Gut, I.G., Gonder, M.K., Ryder, O.A., Hahn, B.H., Navarro, A., Akey, J.M., Bertranpetit, J., Reich, D., Mailund, T., Schierup, M.H., Hvilsom, C., Andres, A.M., Wall, J.D., Bustamante, C.D., Hammer, M.F., Eichler, E.E., and Marques-Bonet, T. 2013. Great ape genetic diversity and population history. *Nature* 499 (7459): 471-475.

Fig. 1: *A group of great ape faces mimics the genome diversity found within these species (Prado-Martinez et al. Nature 2013).* © Javier Prado/Ian Bickerstaff



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- Sudmant, P.H., Huddleston, J., Catacchio, C.R., Malig, M., Hillier, L.W., Baker, C., Mohajeri, K., Kondova, I., Bontrop, R.E., Persengiev, S., Antonacci, F., Ventura, M., Prado-Martinez, J. Great Ape Genome Project, Marques-Bonet, T., Eichler, E.E. 2013. Evolution and diversity of copy number variation in the great ape lineage. *Genome Research* 23 (9): 1373-82.

Awards

Tomàs Marqués-Bonet. EMBO Young Investigator Awards (2013).

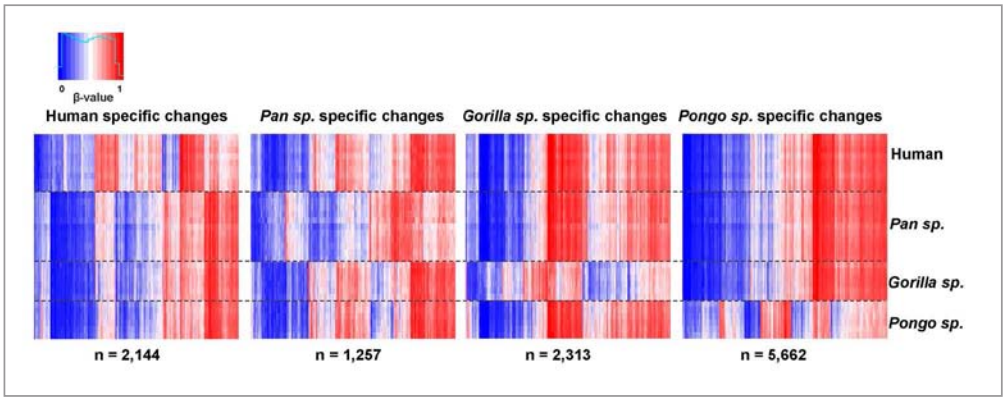


Fig.2: *Lineage specific epigenetics differences (including human) in a comparison of human and other great apes (Hernando-Herraez et al. PLos Genetics 2013).*

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: *MICINN (Spain)*
Years: *2011-2014*
PI: *Tomàs Marquès-Bonet*

GROUP
EVOLUTIONARY AND FUNCTIONAL GENOMICS

GROUP MEMBERS

Josefa González, Group Leader

| Ramón y Cajal Researcher

| Subgroup: Evolutionary and functional genomics

Lain Guio, PhD Student | FI Fellowship
Anna Ullastres, PhD Student | FPI Fellowship
Hung Le Mahn, Postdoctoral Student | VAST-CSIC Fellowship
Lidia Mateo, Master Student | CSIC Contract
Eduard Falquès, Master Student
Miriam Merenciano, Undergraduate Student
Maite G. Barrón | CSIC Contract

| Subgroup: Drosophila telomeres

Elena Casacuberta, Tenured Scientist, CSIC
Elisenda López Panadès, PhD Student | UPF Fellowship

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.

RESEARCH LINES

SUBGROUP: EVOLUTIONARY AND FUNCTIONAL GENOMICS

The key question in genomics is how genomes vary and evolve at both large and fine scales. In our lab, we are particularly interested in understanding the molecular processes underlying adaptive evolution and the functional consequences of adaptive mutations. Towards this end, we combine -omics strategies with detailed molecular and functional analyses of the candidate adaptive mutations in order to arrive at a comprehensive picture of adaptation. We study both transposable element (TE)-induced adaptations and point mutations in the model organism *Drosophila melanogaster*.

We are also interested in the population dynamics of TEs. TEs are the most active, diverse, and ancient components in a broad range of genomes. As such, a complete understanding of genome function and evolution cannot be achieved without a thorough understanding of TE impact and biology.

SUBGROUP: *DROSOPHILA* TELOMERES

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

HeT-A, TART, and TAHRE must be integrated into the cellular pathways controlling telomere length and genome stability. Understanding the regulation of the telomeric transposons will shed light on both telomere length control as well as transposon regulation in *Drosophila*. We are currently focused on 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 cellular partners 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 retrotrotransposons 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 interactions are necessary for telomere proteins to establish and 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.

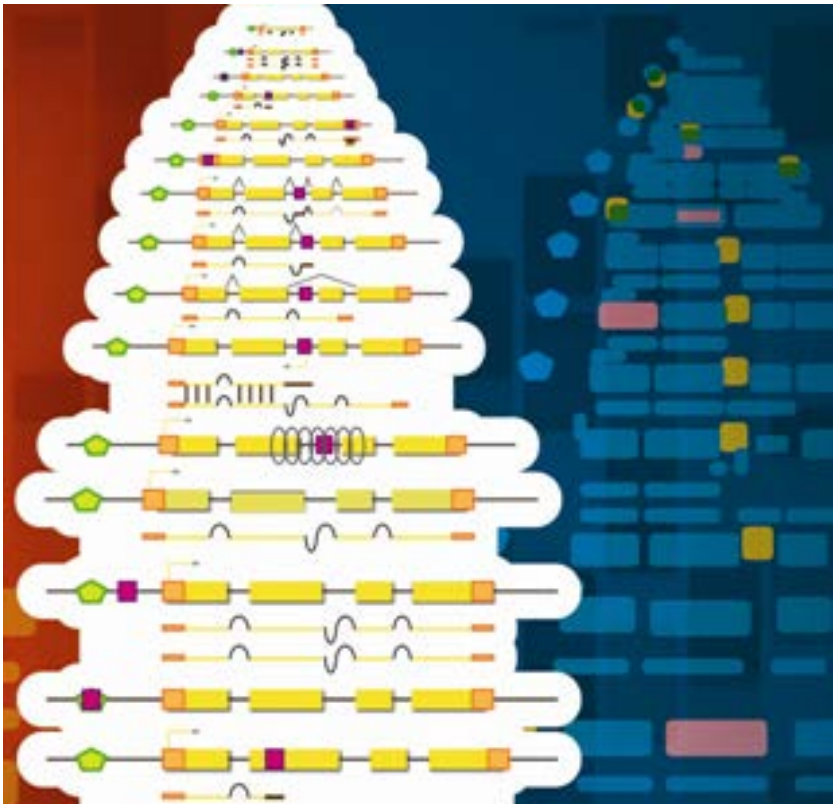


Fig. 1: *Transposable elements, first discovered in maize, generate a great variety of mutations affecting gene structure and gene expression. Casacuberta and González (2013) reviewed the most recent and compelling examples linking transposable element mutations with environmental adaptation. Pink boxes are TEs, green pentagons are regulatory regions, yellow boxes are exons and orange boxes are UTRs.*

PUBLICATIONS 2013

ISI Articles

- Casacuberta, E., and González, J. The impact of transposable elements in environmental adaptation. *Molecular Ecology* 22: 1503-1517, 2013. (Invited Review).
- Silva-Sousa, R., and Casacuberta, E. The JIL-1 kinase affects telomere expression in the different telomere domains of Drosophila. *PLos One* 8: e81543. doi: 10.1371 2013.
- Silva-Sousa, R., Varela, M.D., Casacuberta, E. 2013. The Putzig partners DREF, TRF2 and KEN are involved in the regulation of the Drosophila telomere retrotransposons, HeT-A and TART. *Mobile DNA*. 4: 18. doi: 10.1186/1759-8753-4-18.

FUNDED PROJECTS

Project Title: *El proceso molecular y las consecuencias funcionales de la adaptación (BFU2011-24397)*
Financed by: *Ministerio de Ciencia e Innovacion. Spain.*
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. Spain.*
Years: *2011-2015*
PI: *Josefa González*

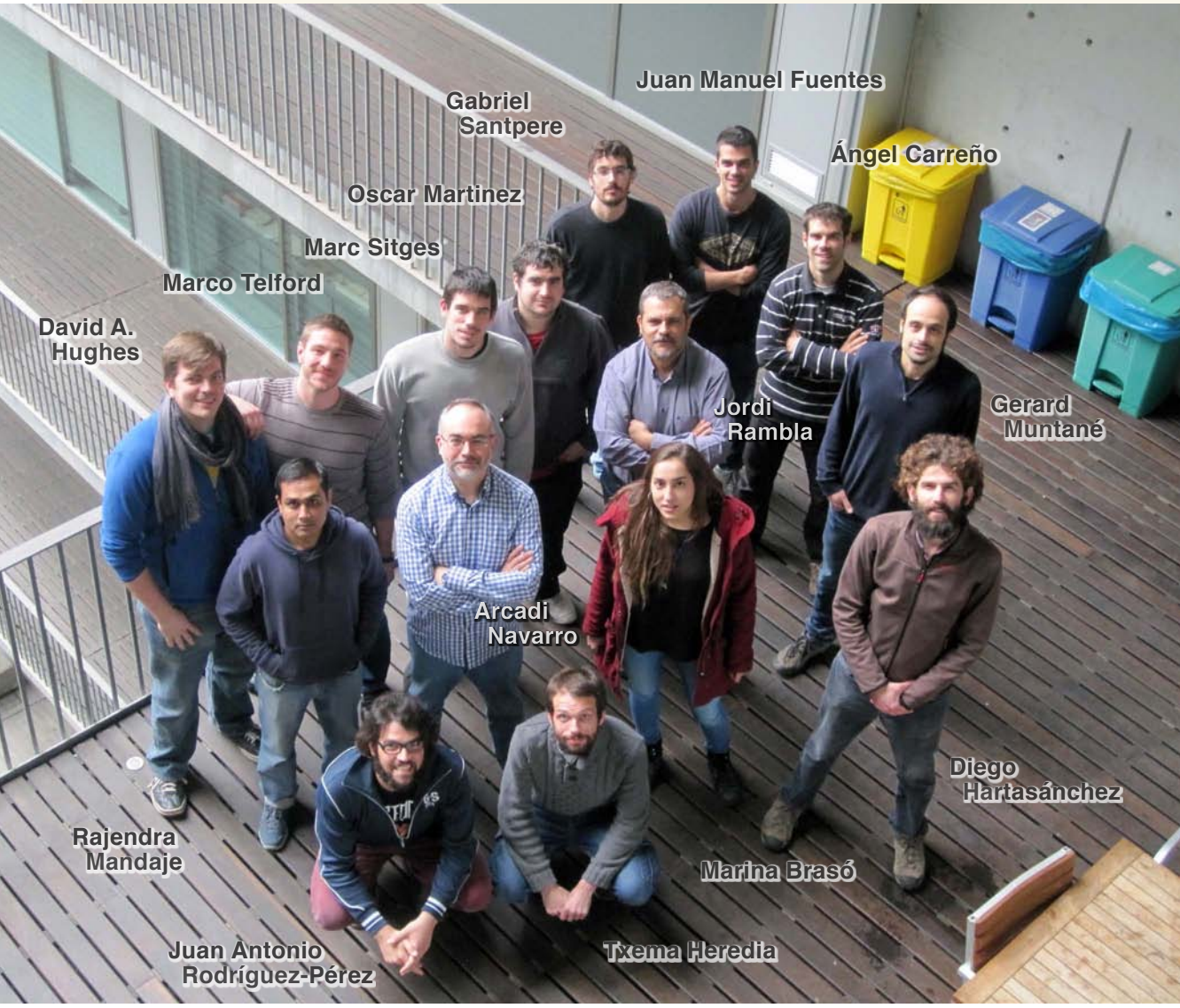
OUTREACH PROJECTS

Project Title: *La ciencia en tu mundo (FCT-12-4240)*
Financed by: *Fundación Española para la Ciencia y la Tecnologia*
Years: *2012-2013*
PI: *Josefa González*

Project Title: *R(e)volución: acércate a las fronteras del conocimiento (FCT-13-6749)*
Financed by: *Fundación Española para la Ciencia y la Tecnologia*
Years: *2013-2014*
PI: *Josefa González*

Project Title: *Com els organismes s'adapten a l'ambient*
Financed by: *Fundació Catalunya-La Pedrera*
Years: *2013*
PI: *Josefa González*

GROUP
EVOLUTIONARY GENOMICS



GROUP MEMBERS

Arcadi Navarro, Group Leader

| Professor, UPF • ICREA Research Professor

Gabriel Santpere, Post-doc | Project Contract (RETICS-ISCIII)

David A. Hughes, Post-doc | Marie Curie Fellowship Contract (COFUND-FP7 UE)

Carlos Morcillo, Post-doc | Project Contract (INB-ISCIII)

Rui Faria, Post-doc | FCT Fellowship

Urko Martínez, PhD Student | UPF Scholarship

Diego Hartasánchez, PhD Student | JAE Contract (CSIC)

Juan Antonio Rodríguez-Pérez , PhD Student | UPF PhD Grant

Marco Telford, PhD Student | Project Contract (INB-ISCIII)

Rajendra Mandaje, PhD Student | Project Contract (INB-ISCIII)

Marina Brasó , MSc Student | Project Contract (RETICS-ISCIII)

Dan Ouchi, IT Technician | Project Contract (INB-ISCIII)

María Niño, IT Technician | JAE Contract (CSIC)

Txema Heredia, IT Technician | Project Contract (INB-ISCIII)

Ángel Carreño, IT Technician | Project Contract (EGA-CRG)

Jordi Rambla, IT Technician | Project Contract (EGA-CRG)

Oscar Martinez, IT Technician | Project Contract (EGA-CRG)

Marc Sitges, IT Technician | Project Contract (EGA-CRG)

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 sequencing full viromes and on studying the levels of expression of virus genes in LCLs to study the evolution of virus-host interactions.

RESEARCH SUBLINES

1. Chromosomal evolution and speciation

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

2. Segmental duplications and copy-number variation in primates

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

3. Detecting positive selection in the human lineage

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

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.

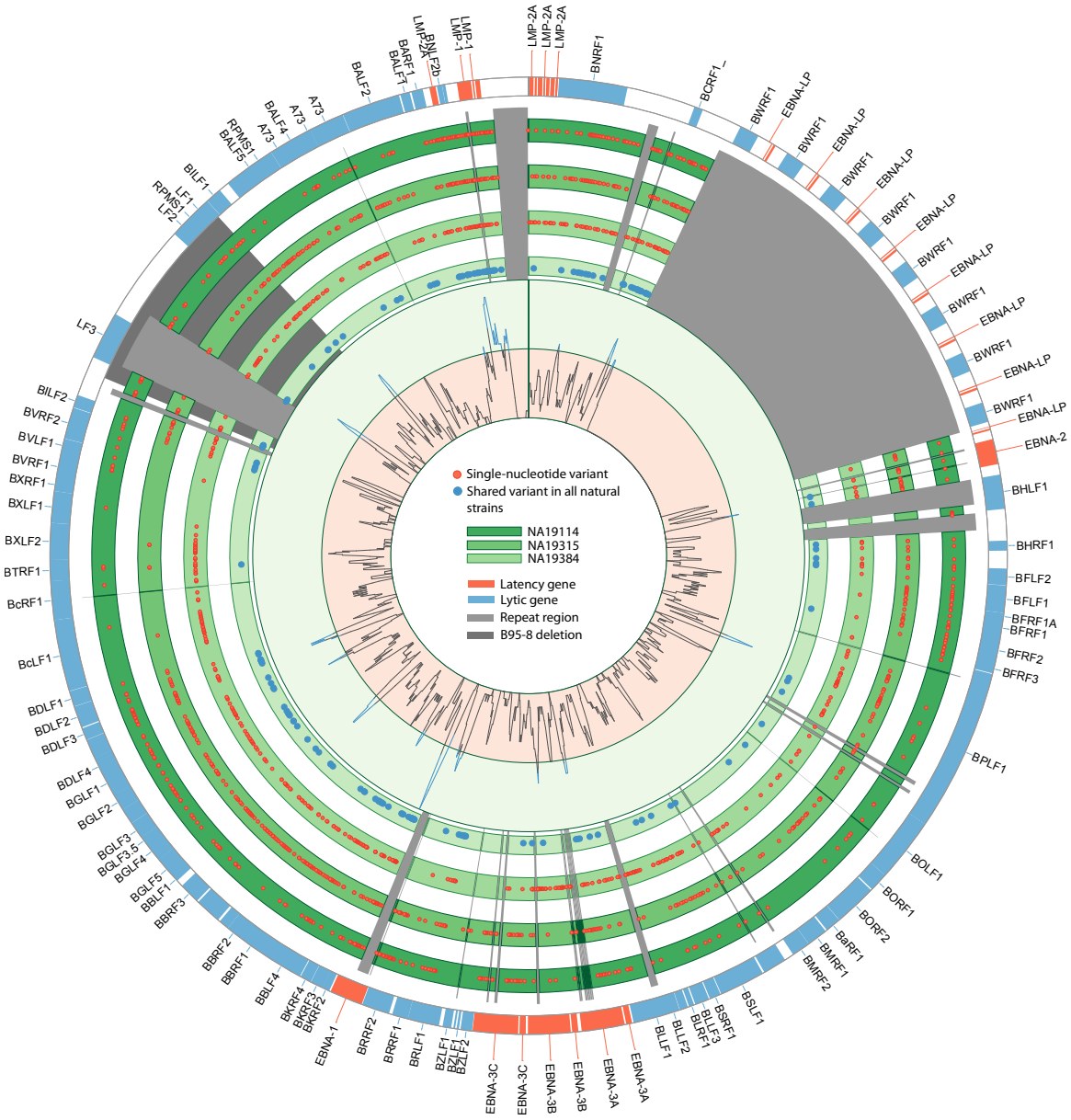


Fig. 1: *Circos plot showing natural Epstein-barr genome wide nucleotide diversity in three lymphoblastoid cell lines derived from African individuals of the 1000 Genomes Project.*

ISI Articles

- Alcina, A., Fedetz, M., Fernández, O., Saiz, A., Izquierdo, G., Lucas, M., Leyva, L., García-León, J.A., Abad-Grau, M.D., Alloza, I., Antigüedad, A., Garcia-Barcina, M.J., Vandenbroeck, K., Varadé, J., de la Hera, B., Arroyo, R., Comabella, M., Montalban, X., Petit-Marty, N., Navarro, A. , Otaegui, D., Olascoaga, J., Blanco, Y., Urcelay, E., and Matesanz, F. 2013. Identification of a functional variant in the KIF5A-CYP27B1-METTL1-FAM119B locus associated with multiple sclerosis. *Journal of Medical Genetics* 50 (1): 25-33.
- Hernando-Herraez, I., Prado-Martinez, J., Garg, P., Fernandez-Callejo, M., Heyn, H., Hvilsom, C., Navarro, A., Esteller, M., Sharp, A.J., Marques-Bonet, T. 2013. Dynamics of DNA Methylation in Recent Human and Great Ape Evolution. *PLos Genetics* 9 (9): e1003763.
- López de Maturana, E., Ye, Y., Calle, M.L., Rothman, N., Urrea, V., Kogevinas, M., Petrus, S., Chanock, S.J., Tardón, A., García-Closas, M., González-Neira, A., Vellalta, G., Carrato, A., Navarro, A., Lorente-Galdós, B., Silverman, D.T., Real, F.X., Wu, X., and Malats, N. 2013. Application of multi-SNP approaches Bayesian LASSO and AUC-RF to detect main effects of inflammatory-gene variants associated with bladder cancer risk. *PLos One* 8 (12): e83745.
- Lorente-Galdos, B., Bleyhl, J., Santpere, G., Vives, L., Ramírez, O., Hernández, J., Anglada, R., Cooper, G.M., Navarro, A., Eichler, E.E., and Marques-Bonet, T. 2013. Accelerated exon evolution within primate segmental duplications. *Genome Biology* 14 (1): R9.
- Marigorta, U.M., and Navarro, A. 2013. High Trans-ethnic Replicability of GWAS Results Implies Common Causal Variants. *PLos Genetics* 9 (6): e1003566.
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- Prado-Martinez, J., Sudmant, P.H., Kidd, J.M., Li, H., Kelley, J.L., Lorente-Galdos, B., Veeramah, K.R., Woerner, A.E., O´Connor, T.D., Santpere, G., Cagan, A., Theunert, C., Casals, F., Laayouni, H., Munch, K., Hobolth, A., Halager, A.E., Malig, M., Hernandez-Rodriguez, J., Hernando-Herraez, I., Prufer, K., Pybus, M., Johnstone, L., Lachmann, M., Alkan, C., Twigg, D., Petit, N., Baker, C., Hormozdiari, F., Fernandez-Callejo, M., Dabad, M., Wilson, M.L., Stevison, L., Camprubi, C., Carvalho, T., Ruiz-Herrera, A., Vives, L., Mele, M., Abello, T., Kondova, I., Bontrop, R.E., Pusey, A., Lankester, F., Kiyang, J.A., Bergl, R.A., Lonsdorf, E., Myers, S., Ventura, M., Gagneux, P., Comas, D., Siegismund, H., Blanc, J., Agueda-Calpena, L., Gut, M., Fulton, L., Tishkoff, S.A., Mullikin, J.C., Wilson, R.K., Gut, I.G., Gonder, M.K., Ryder, O.A., Hahn, B.H., Navarro, A., Akey, J.M., Bertranpetit, J., Reich, D., Mailund, T., Schierup, M.H., Hvilsom, C., Andres, A.M., Wall, J.D., Bustamante, C.D., Hammer, M.F., Eichler, E.E., and Marques-Bonet, T. 2013. Great ape genetic diversity and population history. *Nature* 499 (7459): 471-475.
- Serrano-Munuera, C., Corral-Juan, M., Stevanin, G., San Nicolás, G., Roig, C., Corral, J., Campos, B., De Jorge, L., Morcillo-Suarez, C., Navarro, A., Forlani, S., Durr, A., Kulisevsky, J., Brice, A., Sánchez, I., Volpini, V., and Matilla-Dueñas, A. 2013.

New subtype of spinocerebellar ataxia with altered vertical eye movements mapping to chromosome 1p32. *JAMA Neurology* 70 (6): 764-771. doi: 10.1001/jamaneurol.2013.2311.

Publications as part of the Great Ape Genome Project

- As member of the GAGC. Author(s): Hormozdiari, F., Konkel, M.K., Prado-Martinez, J. et al. Group Author(s): Great Ape Genome Project. 2013. Rates and patterns of great ape retrotransposition. *Proceedings of the National Academy of Sciences USA* 110: 13457-13462.
- As member of the GAGC. Author(s): Sudmant, P.H., Huddleston, J., Catacchio, C.R. et al. Group Author(s): Great Ape Genome Project. 2013. Evolution and diversity of copy number variation in the great ape lineage. *Genome Research* 23: 1373-1382.

FUNDED PROJECTS

Project Title: *Genómica y transcriptómica de las rutas de detoxificación en Drosophila*
Financed by: *Ministerio de Economía y Competitividad - Proyectos de Cooperación Conjuntos España Argentina (PIB2010AR-00266)*
Years: *2011-2013*
PI: *Hernán Dopazo*

Project Title: *Toward a complete view of adaptation in complete genomes. A bottom-up approach to selection acting upon multiple targets*
Financed by: *Ministerio de Economía y Competitividad - MINECO (BFU2012-38236)*
Years: *2013-2015*
PI: *Arcadi Navarro*

Project Title: *Group within the "Red Española de Esclerosis Múltiple" (Spanish Research Network in Multiple Sclerosis)*
Financed by: *Within the RETICS (Redes Españolas de Investigación Cooperativa en Salud) on Multiple Sclerosis (RD12/0032/0011)*
Years: *2013-2015*
PI: *Arcadi Navarro (Coordinator: Pablo Villoslada)*

Project Title: *INB GN8*
Financed by: *Instituto de Salud Carlos III (Instituto Nacional de Bioinformática)*
Years: *2013*
PI: *Arcadi Navarro*

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

GROUP
PALEOGENOMICS

GROUP MEMBERS

Carles Lalueza-Fox, Group Leader

| Research Scientist, CSIC

Oscar Ramírez | JAEDOC, CSIC Contract
Íñigo Olalde, PhD Student | Basque Country Scholarship
Federico Sánchez-Quinto, PhD Student | FPI Scholarship, MICINN
Daniel Gómez Sánchez, Master Student
Federica Pierini, Leonardo da Vinci grant

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, and phylogenetics and adaptive processes. We work with different animal species, and also with an extinct hominin species (the Neandertals). In our group we are interested in the genomic diversity among Neandertals, and in the individualisation of a Neandertal family group from the El Sidrón site (Asturias, Spain). We are also investigating the evolutionary dynamics of the prehistory of Europe through the analysis of Mesolithic, Neolithic, and Cooper Age human genomes.

RESEARCH LINES

1. Neandertal genomic diversity

We are analyzing different individuals from the 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 genomics project for understanding the Neandertal diversity and the kinship relationships within a Neandertal family group.

2. 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.

3. European prehistory

We are genetically analysing ancient samples from prehistoric European populations to reconstruct past human migrations and adaptive processes related to the arrival of farming in the Mesolithic-Neolithic shift. We have recently retrieved the first pre-agricultural, modern human European genome, that of La Braña-Arintero in León, Spain.

4. Historical genomics

We are conducting a pioneer project for obtaining complete genomes from relicts attributed to historical characters, such as that of Louis XVI (king of France), and from samples excavated from recent European settlements such as Quebec. Our goal is to bring the personal genomics to a new dimension that could have historical implications.

PUBLICATIONS 2013

ISI Articles

- Charlier, P., Lalueza-Fox, C., and Hervé, C. 2013. Medical recollections. The head of Henri IV: identification and ethical issues. *Revue du Praticien* 63 (2): 289-293.
- Charlier, P., Olalde, I., Solé, N., Ramírez, O., Babelon, J.P., Galland, B., Calafell, F., and Lalueza-Fox, C. 2013. Genetic comparison of the head of Henri IV and the presumptive blood from Louis XVI (both Kings of France). *Forensic Science International* 226 (1-3): 38-40.
- Dean, M.C., Rosas, A., Estalrrich, A., García-Tabernero, A., Huguet, R., Lalueza-Fox, C., Bastir, M., and de la Rasilla, M. 2013. Longstanding dental pathology in Neandertals from El Sidrón (Asturias, Spain) with a probable familial basis. *Journal of Human Evolution* 64 (6): 678-686.
- Lalueza-Fox, C. 2013. Agreements and Misunderstandings among Three Scientific Fields: Paleogenomics, Archaeology, and Human Paleontology. *Current Anthropology* 54 (S8): S214-S220.
- Lorente-Galdos, B., Bleyhl, J., Santpere, G., Vives, L., Ramírez, O., Hernandez, J., Anglada, R., Cooper, G.M., Navarro, A., Eichler, E.E., and Marques-Bonet, T. 2013. Accelerated exon evolution within primate segmental duplications. *Genome Biology* 14 (1): R9.
- Manunza, A., Zidi, A., Yeghoyan, S., Balteanu, V.A., Carsai, T.C., Scherbakov, O., Ramírez, O., Eghbalsaied, S., Castelló, A., Mercadé, A., and Amills, M. 2013. A high throughput genotyping approach reveals distinctive autosomal genetic signatures for European and Near Eastern wild boar. *PLos One* (2): e55891.
- Maricic, T., Günther, V., Georgiev, O., Gehre, S., Ćurlin, M., Schreiweis, C., Naumann, R., Burbano, H.A., Meyer, M., Lalueza-Fox, C.,de la Rasilla, M., Rosas, A., Gajović, S., Kelso, J. Enard, W., Schaffner, W., and Pääbo, S. 2013. A recent evolutionary change affects a regulatory element in the human FOXP2 gene. *Molecular Biology and Evolution* 30 (4): 844-852.
- Prado-Martinez, J., Hernando-Herraez, I., Lorente-Galdos, B., Dabad, M., Ramirez, O., Baeza-Delgado, C., Morcillo-Suarez, C., Alkan, C., Hormozdiari, F., Raineri, E., Estellé, J., Fernandez-Callejo, M., Valles, M., Ritscher, L., Schöneberg, T., de la Calle-Mustienes, E., Casillas, S., Rubio-Acero, R., Melé, M., Engelken, J., Caceres, M., Gomez-Skarmeta, J.L., Gut, M., Bertranpetit, J., Gut, I.G., Abello, T., Eichler, E.E., Mingarro, I., Lalueza-Fox, C., Navarro, A., and Marques-Bonet, T. 2013. The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild. *BMC Genomics* 14: 363.
- Ramírez, O., Gómez-Díaz, E., Olalde, I., Illera, J.C., Rando, J.C., González-Solís, J., and Lalueza-Fox, C. 2013. Population connectivity buffers genetic diversity loss in a seabird. *Frontiers in Zoology* 10 (1): 28.
- Ramírez, O., Quintanilla, R., Varona, L., Gallardo, D., Díaz, I., Pena, R.N., and Amills, M. 2013. DECR1 and ME1 genotypes are associated with lipid composition traits in Duroc pigs. *Journal of Animal Breeding and Genetics* 131: 46-52.
- Rosas, A., Estalrrich, A., García-Vargas, S., García-Tabernero, A., Huguet, R., Lalueza-Fox, C., and de la Rasilla, M. 2013. Identification of Neandertal individuals in fragmentary fossil assemblages by means of tooth associations: The case of El Sidron (Asturias, Spain). *Comptes Rendues Palevolution* 12 (5): 279-291.
- Wood, R.E., Higham, T.F.G, de Torres, T., Tisnérat-Laborde, N., Valladas, H., Ortiz, J.E., Lalueza-Fox, C., Sánchez-Moral, S., Cañaveras, J.C., Rosas, A., Santamaría, D., and de la Rasilla, M. 2013. A new date for the Neanderthals of El Sidrón Cave (Asturias, Northern Spain). *Archaeometry* 55 (1): 148-158.

Fig 1: *Collective Late Neolithic burial from El Mirador cave in Atapuerca (Burgos)*
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Books

- Lalueza-Fox, C. 2013. Palabras en el tiempo; la lucha por el genoma neandertal. Ed. Crítica; colección Drakontos. ISBN10: 8498924545, ISBN13: 978-8498924541.

FUNDED PROJECTS

Project Title: *Inferencias evolutivas a partir de la captura y secuenciación de regiones genómicas en neandertales*
Financed by: *Ministerio de Economía y Competitividad - MINECO (BFU2012-34157)*
Years: *2013-2015*
PI: *Carles Lalueza-Fox*

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

GROUP MEMBERS

Ricard Solé, Group Leader

| Professor, UPF • ICREA Research Professor

Javier Macía | Post-doct Researcher (profesor lector UPF)
Raúl Montañez | Post-doct Researcher (UPF Project Contract)
Dani Rodríguez-Amor | Post-doct Researcher (UPF Project Contract)
Carlos Rodríguez-Caso | Post-doct Researcher (UPF Project Contract)
Sergi Valverde | Post-doct Researcher (visiting professor UPF)
Adriano Bonforti | Pre-doct Researcher
Max Carbonell | Pre-doct Researcher
Salvador Durán | Pre-doct Researcher
Luis Seoane | Pre-doct Researcher
Ben Shirt-Ediss | Pre-doct 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.

1. Multicellularity: origins, maintenance and decay

We want t develop a general theoretical framework for the origins and development of complex multicellular systems, including early emergence through evolution (evo-devo), th 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 how we can move beyond that landscape. This work has several branches, both theoretical and applied to biomedical research.
http://complex.upf.edu/

ISI Articles

- Corominas Murtra, B., Fortuny, J., and Solé, R.V. 2013. Towards a mathematical theory of meaningful communication. http://arxiv.org/abs/1004.1999.
- Duarte, J., Januário, C., Rodrigues, C., and Sardanyés, J. 2013. Topological complexity and predictability in the dynamics of a tumor growth model with Shilnikov's chaos. *International Journal of Bifurcation and Chaos in Applied Sciences and Engineering* 23 (7): 1350124.
- Fort, J., and Solé, R.V. 2013. Accelerated tumor invasion under non-isotropic cell dispersal in glioblastomas. *New Journal of Physics* 15 (5), 055001.
- Murtra, B.C., Goñi, J., Solé, R.V., and Rodriguez-Caso, C. 2013. On the origins of hierarchy in complex networks. *Proceedings of the National Academy of Sciences USA* 110, 13316-13321.
- Otero-Espinar, V., Seoane, L., Nieto, J.J., and Mira, J. 2013. Analytic solution of a model of language competition with bilingualism and interlinguistic similarity. *Physica* D64: 17-26, http://arxiv.org/abs/1303.4959.
- Seoane, L., and Solé, R.V. 2013. A multiobjective optimization approach to statistical mechanics http://arxiv.org/abs/http://arxiv.org/abs/1303.5633.
- Solé, R.V., and Macía, J. 2013. Expanding the landscape of biological computation with synthetic multicellular consortia. *Natural Computing* 12, 485-497.
- Solé, R.V., and Valverde, S. 2013. Before the endless forms: embodied model of transition from single cells to aggregates and ecosystem engineering. *PLos One* 8 (4): e59664.
- Solé, R.V., and Valverde, S. 2013. Macroevolution in silico: scale, constraints and universals. *Paleontology* 56, 1327-1340.
- Solé, R.V., Valverde, S., Rosas Casals, M., Kauffman, S., Farmer, D., and Eldredgen., 2013. The evolutionary ecology of technological innovations. *Complexity* 18, 15-27.
- Weitz, J.S., Poisot, T., Meyer, J.R., Flores, C.O., Valverde, S., Sullivan, M.B., Hochberg, M.E. 2013. Phage–bacteria infection networks. *Trends in Microbiology* 2, 82-91.
- Zwart, M.P., Piljman, G.P., Sardanyés, J., Duarte, J., Januário, C., and Elena, S.F. 2013. Complex dynamics of defective interfering baculoviruses during serial passage in insect cells. *Journal of Biological Physics* 39 (2), 327-342.

Books/Book Chapters

- Solé, R.V., Duran, S. 2013. In silico transitions to multicellularity. In: Evolutionary transitions to multicellular life. A. Nedelcu, Ruiz-Trillo, I.(ed). Springer.

Other Publications

- Solé, R.V., Murtra, B.C., and Fortuny, J. 2013. Lenguaje, redes y evolución. *Investigación y ciencia* 58-67.
- Valverde, S., and Solé, R.V. 2013. Networks and the City. *Architectural Design* 83 (4), 112-119.
- Widder, S., Solé, R.V., Macia, J. 2013. Plasticity, evolvability and the abundance of feed-forward loops in transcription networks. *Networking Networks* 81-100.

FUNDED PROJECTS

Project Title: *SYNCOM*
Financed by: *European research Council (ERC)*
Years: *2012-2017*
PIs: *Ricard Solé/F. Posas*

Project Title: *Física estadística de cánceres inestables genómicamente*
Financed by: *Ministerio de Economía y Competitividad (MINECO) (FIS 2012-39288)*
Years: *2013-2015*
PI: *Ricard Solé*

Contract Title: *Cellular computation (Convenio de colaboración en materia de apoyo a la transferencia tecnológica en el campo de la biotecnología)*
Financed by: *Fundación Marcelino Botín*
Years: *2010-2016*
PI: *Ricard Solé*

Project Title: *Assessment of topological vulnerability of infrastructural systems based on extended complex network technique.*
Financed by: *Stichting Next Generation Infrastructure*
Years: *2012-2014*
PI: *Ricard Solé*

GROUP
LANGUAGE EVOLUTION

GROUP MEMBERS

Luc Steels, Group Leader

| ICREA Research Professor

Emília García Casademont | Pre-doctoral Researcher

Miquel Cornudella Gaya | Pre-doctoral 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 did humans start to talk?'. The cultural aspect should answer how new language forms arise in language and keep on changing over time. The biological aspect addresses 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 is 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 the 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 conduct 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 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.

PUBLICATIONS 2013

ISI Articles

- Beuls, K., and Steels, L. 2013. Agent-based models of strategies for the emergence and evolution of grammatical agreement. *PLos One* 8 (3).

Book Chapters

- Beuls, K., and Steels, L. 2013. Crossing complexity barriers with grammar. A case study for agreement systems. In: Mufwene, S., J-M. Hombert, F. Pellegrino and C. Coupe (eds) Complexity in Language: Developmental and Evolutionary Perspectives. Cambridge University Press, Cambridge.
- Steels, L. 2013. Breaking down barriers. In: Dor, D., C. Knight and J. Lewis 2013. The Social Origins of Language: Early Society, Communication and Polymodality. Oxford University Press, Oxford.
- Steels, L. 2013. How language emerges in situated embodied interactions. In: Lefebvre, C., B. Comrie (2013) New Perspectives on the Origins of Language. Cambridge University Press, Cambridge.

FUNDED PROJECTS

Project Title: *Artificial Language Evolution for Autonomous Agents*
Financed by: *EU Marie Curie Integration Grant*
Years: *2011-2013*
PI: *Luc Steels*

Project Title: *INSIGHT - Darwinian Neurodynamics*
Financed by: *FP7-EU*
Years: *2013-2016*
PI: *Luc Steels*

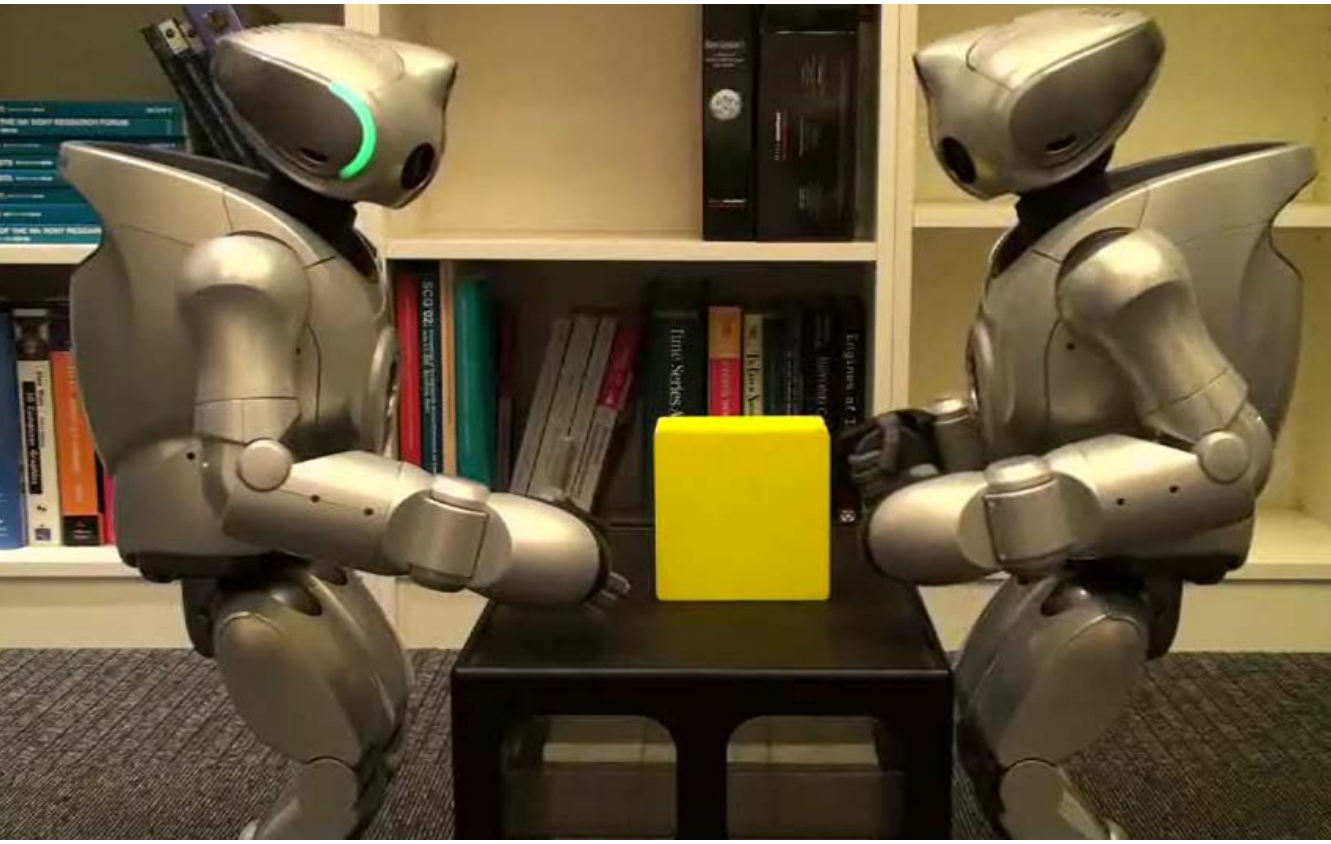


Fig. 1: *Experiments in the ontogenetic ritualization of gestures from instrumental action. Robots help each other pick up objects and a failed grasping action becomes a pointing gesture (Steels).*

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, paleontology, 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 at the application of a number of different technical methodologies, such as cell and developmental biology and comparative genomics.

GROUP

EVOLUTION AND DEVELOPMENTAL BIOLOGY

GROUP MEMBERS

Xavier Franch-Marro, Group Leader

| Tenured Scientist, CSIC

| Subgroup: Morphology and signalling

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

Neus Bota Rabassedas, PhD Student, FPI Scholarship, MEC

Cristina Miguel Vijandi, PhD Student, FPI Scholarship, MEC

Josefa Cruz Rodríguez, Post-doc, CSIC Contract

Victoria Rodríguez Marcé, Undergraduate Student, UdV

| Subgroup: Hormonal control of insect development

David Martín Casacuberta, Principal Investigator, Tenured Scientist, CSIC

Enric Ureña, PhD Student, CSIC Contract

Cristina Manjón, Post-doc, CSIC Contract

Alex Subias, Undergraduate Student, UB

RESEARCH OUTLINE

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

RESEARCH SUBLINES

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 the *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 the FGF (fibroblast growth factor) signalling pathway. At metamorphosis, the tracheal system undergoes a deep remodelling stage, giving rise to pupae and the 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 regains development potency. The genetic circuits controlling tracheal cell proliferation and dedifferentiation are only now beginning to emerge. Therefore, we aim to discover new genes and signalling pathways involved in such interesting processes.

2. Tracheal System Evolution

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

3. Wingless 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 in 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 the *Drosophila* wing disc. In parallel, we study these genes in *Tribolium castaneum* in order to gain further insights into developmental processes occuring during beetle and fly development, leading to more general conclussions for arthropod evolution.

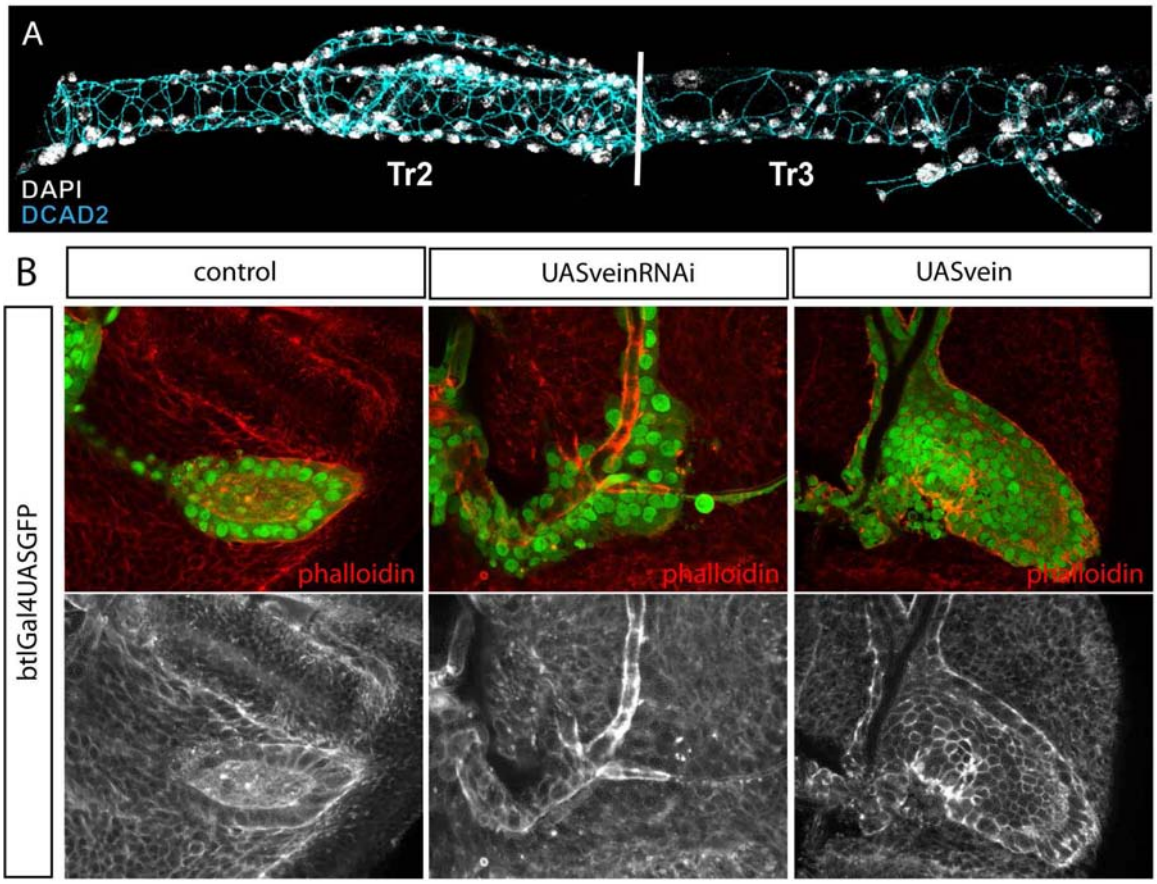


Fig. 1: *A) Dividing Tracheal cells during Drosophila larva development. Cell membrane is marked in blue and nucleus in grey.*
B) Effects of EGF ligand vein during Air sac Primordium development.

SUBGROUP: HORMONAL CONTROL OF INSECT DEVELOPMENT

1. Hormonal control of insect 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, however, two hormones control all the developmental changes associated with 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 hormones of paramount importance in development, prevents metamorphosis by coordinating multiple

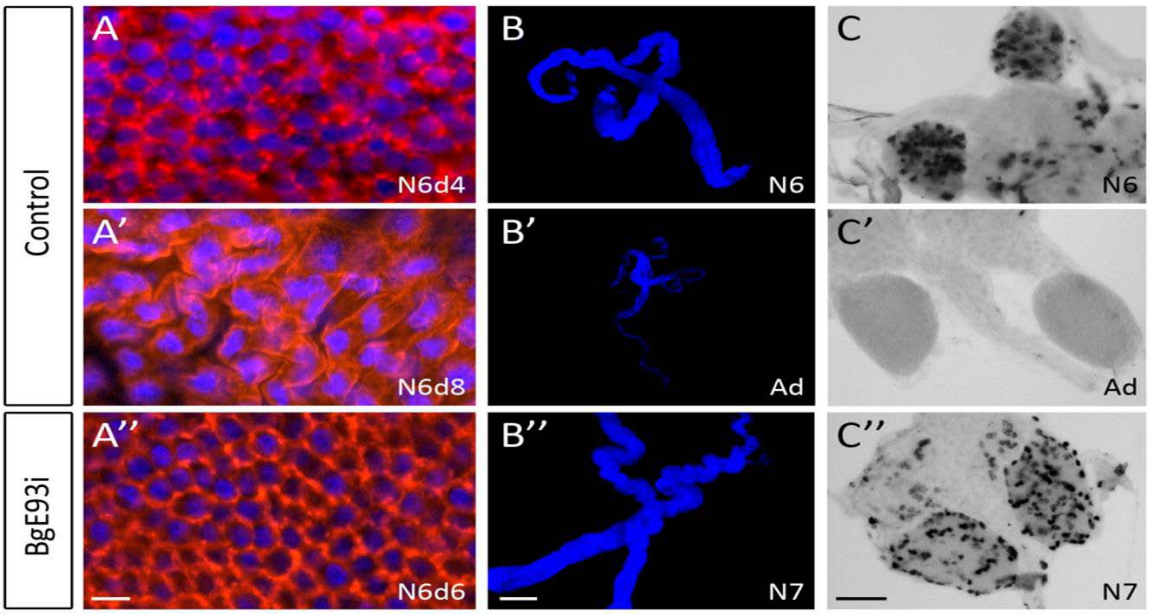


Fig. 2: *E93 is required for adult differentiation in the hemimetabolous insect* Blattella germanica. *Effect of RNAi of BgE93 on adult metamorphosis of wings (A-A"), prothoracic glands (B-B") and corpora* allata *(C-C"). Loss of E93 blocked adult metamorphosis during the N6-adult transition.*

20E-dependent developmental and physiological processes. The main goal of this project is the molecular characterization of the mechanisms underlying the regulatory roles 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* (RNAi and 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 behavior in both types of insects. In summary, this project aims to define the molecular mechanisms

underlying insect metamorphosis to understand the evolutionary conservation of the molecular program determining the developmental transition to adulthood in animals.

2. Insect body plan evolution

The body plan is highly conserved in hemimetabolous and holometabolous insects, although the molecular mechanisms underlying body patterning during embryogenesis are very different. The holometabolous *D. melanogaster* is an example of a long germ band insect. The embryonic primordium of the embryo, the germ band, spreads through the entire egg and all of the body segments are specified simultaneously. In contrast, hemimetabolous insects present short germ band development where the germ band occupies a relatively short portion of the body, and segments are added sequentially during the embryonic development from a posterior proliferation zone. The main goal of this project is to characterize how ecdysteroids and juvenile hormone control the major morphogenetic events in short germ band 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. Evolution of SUMO protein functions in insect metamorphosis

Post-translational modification with small ubiquitin-related modifier, SUMO, is a widespread mechanism for rapid and reversible changes in protein function, especially in nuclear hormone receptors. In this project, in collaboration with the laboratory of Dr. Rosa Barrio (CIC bioGUNE, Vizcaya), we are addressing the functional analysis of sumoylation on the development of the hemimetabolous model insect, *B. germanica*, and its relationship with nuclear hormone receptor function. Furthermore, we are testing whether the functions of SUMO proteins are evolutionarily conserved between hemimetabolous and holometabolous insects by performing functional analysis of the two *B. germanica* Sumo homologues using *D. melanogaster* as model system.

PUBLICATIONS 2013

ISI Articles

- Boulan, L., Martín, D., and Milán, M. 2013. *bantam* miRNA promotes systemic growth by connecting insulin signaling and ecdysone production. *Current Biology* 23: 473-478.

Other Publications

- Wendler, F., Bota-Rabassedas, N., and Franch-Marro, X. 2013. Cancer becomes wasteful: emerging roles of exosomes in cell-fate determination. *Journal of Extracellular Vesicles* 013; 2: 10.3402/jev.v2i0.22390.

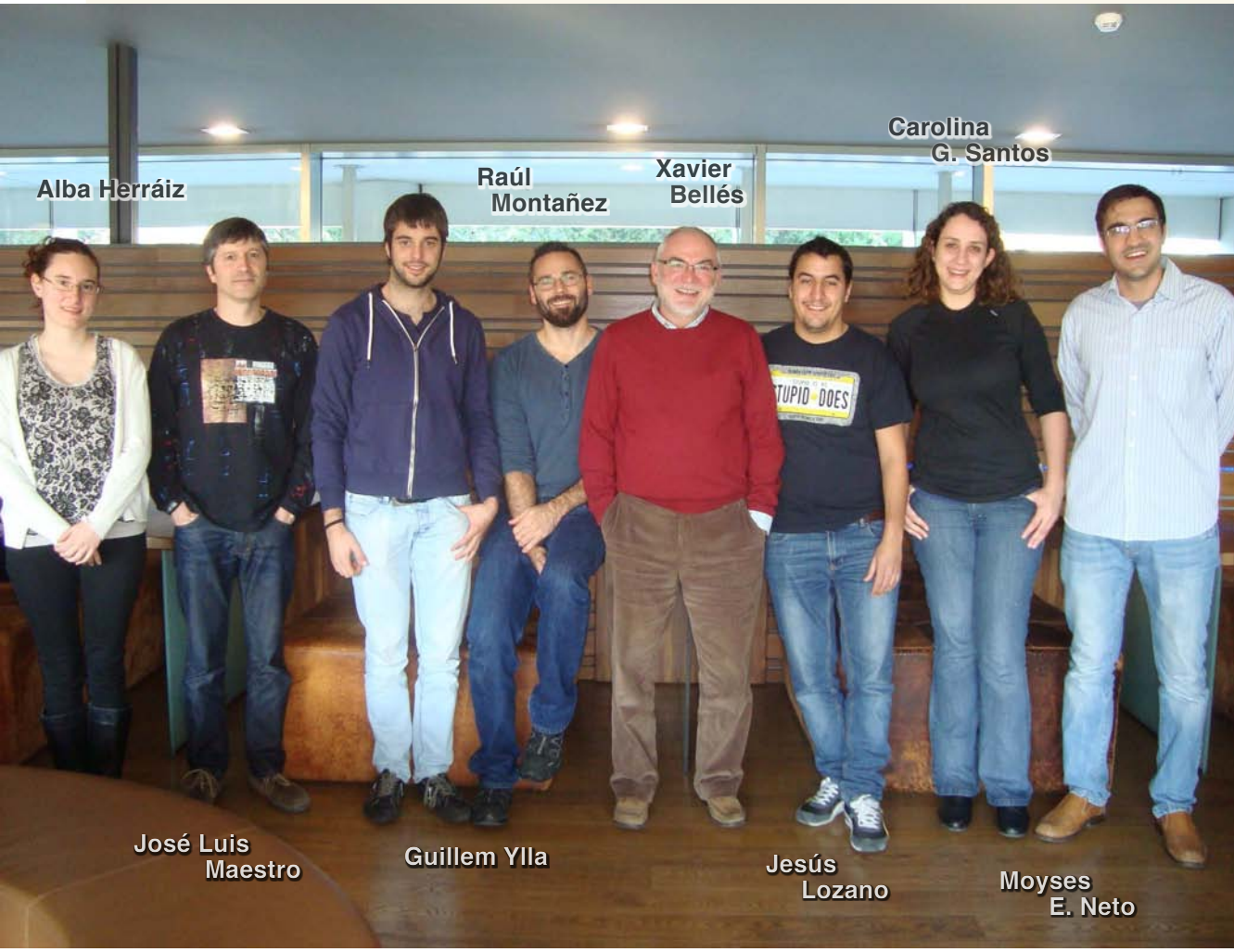
FUNDED PROJECTS

Project Title: *Formación del Gradiente del Morfógeno y de la función de Wingless en el control del crecimiento*
Financed by: *Ministerio de Ciencia e Innovación (REF: BFU2009-08748)*
Years: *2010-2013*
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 (REF: BFU2009-10571)*
Years: *2010-2013*
PI: *David Martín Casacuberta*

GROUP

INSECT PHYSIOLOGY AND MOLECULAR BIOLOGY



GROUP MEMBERS

Xavier Bellés, Group Leader

| Research Professor, CSIC

| Subgroup: Evolution of insect metamorphosis

Xavier Bellés, Group Leader, Research Professor, CSIC

Carolina Goçalves Santos, Postdoctoral Researcher, CNPq Fellowship

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

Moyses Elias Neto, Post-doc (FAPESP Grant, Sao Paulo, Brazil)

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

Jesús Lozano, PhD Student, Scholarship MICINN

Guillem Ylla Bou, Bioinformatician, Project Contract

| Subgroup: Nutritional signals in insects

José Luis Maestro, Tenured Scientist, CSIC

Songül Süren-Castillo, Post-doc, Project Contract

RESEARCH OUTLINE

Our goals and interests have been traditionally diverse, embracing a number of subjects around the physiology of the insect, but most of the work focused our research 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.

At present, we concentrated on the origin and evolution of insect metamorphosis, a research line headed by Xavier Bellés, and on the study of the physiological and developmental effects of nutritional signals, line 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, holometabolan species (mainly in the fly *Drosophila melanogaster*), we currently use the cockroach *Blattella germanica* as model, which is a phylogenetically basal, hemimetabolan species. Therefore, results obtained in this cockroach can be used as a reference baseline when establishing comparisons with other more modified insect species.

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 holometabolan species. The idea is to infer the evolutionary history underlying the transition from hemimetaboly to holometaboly. During 2013, we have been working in the following five main subjects.

1. Using the development of cockroach tergal glands as a minimal model of metamorphosis

The male tergal gland of cockroaches is a complex morphologic structure that forms during the imaginal molt in the tergites 7 and 8 (T7-8). We have followed a transcriptomic approach, and we have compared mRNA libraries of T7-8 in metamorphic and non-metamorphic transitions. A number of transcripts that are differentially expressed at the beginning of the last instar nymph, like the transcription factor E93, have been identified as "metamorphosis triggers".

2. Transcription factors and hormonal signalling

Metamorphosis is mainly regulated by two hormone types: ecdysteroids and juvenile hormones. We are interested in the transcription factors involved in the signalling 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 2013 we continued the functional characterization of the transcription factors Krüppel homolog 1 and Broad complex, the aforementioned E93, and the two components of the JH receptor: the bHLH-PAS proteins Methoprene-tolerant and Taiman.

Fig. 1: *Winglet-shaped tergal expansions formed in the pronotum of a German cockroach as a result of depleting the expression of the gene* sex comb reduced. *A control specimen showing a normal pronotum is showed to the right.* © Moyses E. Neto

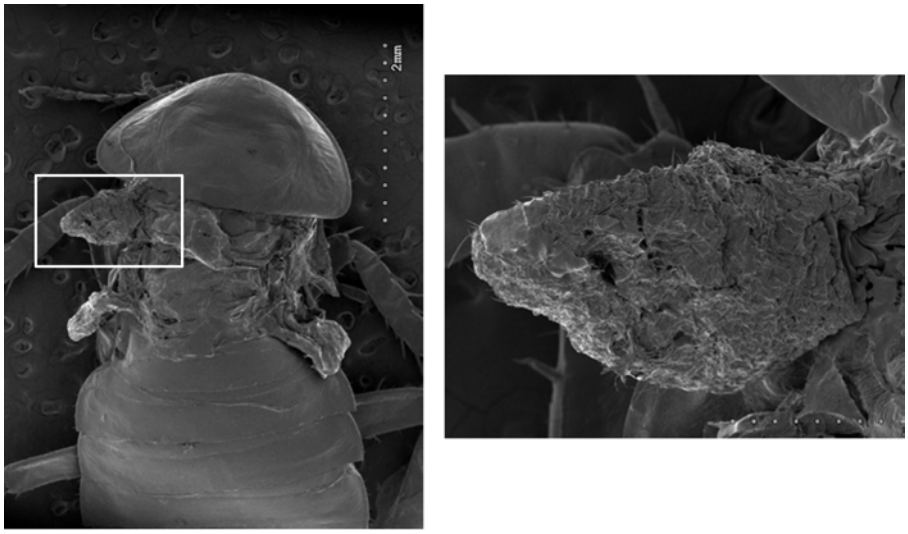


Fig. 2: *Precocious adult of German cockroach resulting from depleting the expression of* Taiman *gene in nymphal stages. The detail shows a sub-developed tegmina-like structure in the mesothorax.* © Jesús Lozano

3. Small RNAs and RNAi

Our hypothesis is that miRNAs play a modulatory role in the shift from juvenile to adult developmental programs. Comparing miRNA libraries from penultimate and last instar nymph we identified a number of miRNAs that where differentially expressed and can thus play significant roles in adult morphogenesis. Among these were miR-8, miR-100, miR-123, let-7 and miR-2a. Moreover, experiments using anti-miR molecules and miR mimics led to characterize the function of

these miRNAs in the transition from nymph to adult. Moreover, we have also investigated a number of aspects on the biochemical machinery involved in miRNA generation and in RNA interference (RNAi).

4. Determination of wing identity

A project developed in 2013 involved the identification of factors determining the identity of wing structure. Our functional studies using RNAi have revealed that *ultrabithorax* gene determines the membranous structure of the metathoracic wings, *apterous* is crucial for the formation of the coriaceous structure of the mesothoracic tegmina, whereas *sex comb reduced* represses the formation of wings in the prothorax.

5. Complex networks

Metamorphosis involves complex networks of gene regulation, and the idea is to reduce this complexity to graphs capturing the main properties of these networks. During 2013, we have focused on networks of interaction mRNA-miRNA, comparing metamorphic and non-metamorphic transitions, and interaction networks protein-protein and DNA-protein in the process of wing formation.

SUBGROUP: NUTRITIONAL SIGNALS IN INSECTS

The goal of our research line is the study of the mechanisms by which nutritional signalling pathways are able to activate physiological and endocrine processes that lead to reproduction. At present, we mainly study the insulin receptor pathway and how it works. During 2013, we worked in the following subjects.

1. Function of FoxO transcription factor on adult female metabolism

FoxO protein is the main transcriptional effector of the insulin receptor pathway. During 2013 we demonstrated that FoxO activates different catabolic pathways in response to starvation. Thus, in addition to activate some key enzymes of the classic catabolic pathways, FoxO is able to transcriptionally induce the expression of hypertrehalosemic hormone and, in this way, to act through a new and essentially different mode of action.

2. Insulin receptor and insulin-like peptides (ILPs)

During 2013 we continued different projects on the role of insulin receptor in the regulation of reproduction, starvation resistance and longevity. In addition, and using the sequence of five ILPs peptides obtained from a brain cDNA library, we studied the expression profile of the five ILPs in the different tissues and how the expression is differentially regulated in response to the nutritional status.

ISI Articles

- Garbutt, J.S., Belles, X., Richards, E.H., and Reynolds, S.E. 2013. Persistence of double-stranded RNA in insect hemolymph as a potential determiner of RNA interference success: Evidence from *Manduca sexta* and *Blattella germanica*. *Journal of Insect Physiology* 59: 171-178.
- Huang, J-H, Lozano, J, and Belles, X. 2013. *Broad-complex* functions in postembryonic development of the cockroach *Blattella germanica* shed new light on the evolution of insect metamorphosis. *Biochimica et Biophysica Acta, General Subjects* 1830: 2178-2187.

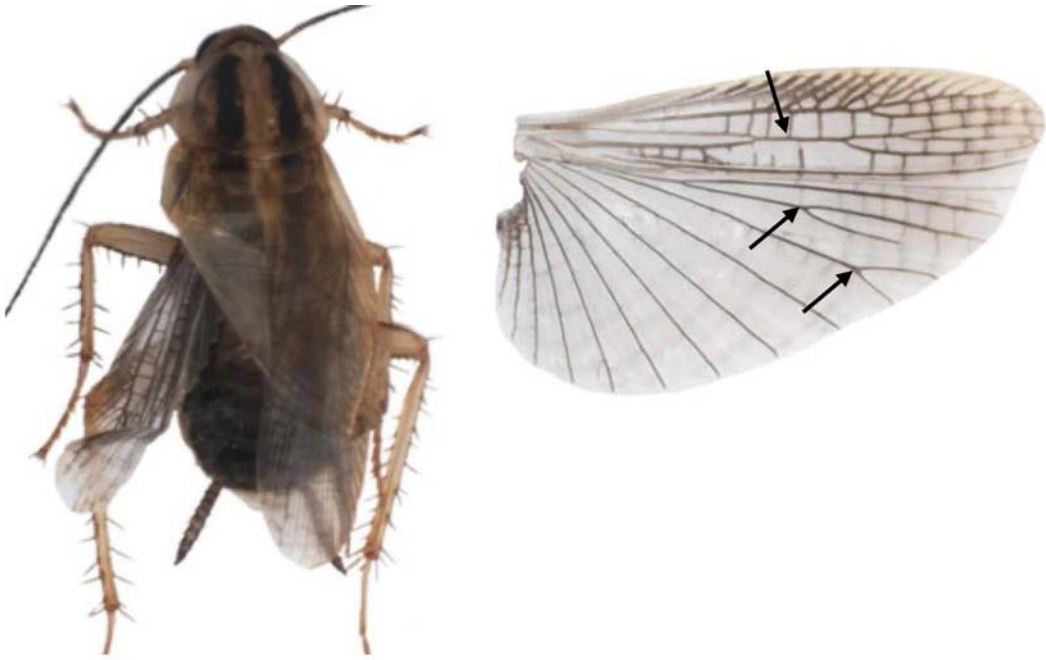


Fig. 3: *Adult female of German cockroach resulting from depleting the levels of the miRNA let-7 (note the malformed wings and tegmina), and detail of the membranous hindwings with defects in the vein patterning.* © Mercedes Rubio

- Rubio, M., and Belles, X. 2013. Subtle roles of microRNAs let-7, miR-100 and miR-125 on wing morphogenesis in hemimetabolan metamorphosis. *Journal of Insect Physiology* 59: 1089-1094.
- Rubio, M., Montañez, R., Perez, L., Milan, M., and Belles, X. 2013. Regulation of atrophin by both strands of the mir-8 precursor. *Insect Biochemistry and Molecular Biology* 43: 1009-1014.

Books/Book Chapters

- Belles, X. 2013. *La metamorfosis de los insectos.* Editorial Catarata. Colección ¿Qué sabemos de? Madrid. 102 pp.

Other Publications

- Belles, X. 2013. Eclipsat per Darwin. Cultura/s (Suplement cultural de La Vanguardia). 04/12/2013.
- Belles, X. 2013. Los libros de Alfred Russel Wallace en España. Boletín de la Sociedad Entomológica Aragonesa 53: 3-6.
- Belles, X. 2013. Naturalista que s'avançà a Darwin. Mètode 79: 19-25.

FUNDED PROJECTS

Project Title: *Global change and physiological diversity.*
Finaced by: *International Laboratory of Global Change (LINCGlobal), CSIC y Pontificia Universidad Católica de Chile.*
Years: *2008-2014*
PIs: *Xavier Bellés (España) and Francisco Bozinovic (Chile)*

Project Title: *Silencing the silencers. Mechanistic bases of metamorphosis regulation in insects.*
Finaced by: *MEC (REF: CGL2008-03517/BOS)*
Years: *2009-2013 (Consolider project)*
PI: *Xavier Bellés Ros*

Project: *Regulation of cockroach tergal gland morphogenesis as a minimal model of insect metamorphosis.*
Finaced by: *MINECO. CGL2012-36251.*
Years: *2013-2015*
PI: *Xavier Bellés Ros*

Project: *Nutritional signals and reproduction in insects. Role of the transcription factor FoxO.*
Finaced by: *MINECO (BFU2010-15906)*
Years: *2011-2013.*
PI: *José Luis Maestro Garriga*

GROUP
INSECT REPRODUCTION

GROUP MEMBERS

Maria-Dolors Piulachs, Group Leader

| Research Scientist, CSIC

Paula Irles, Post-doc | CONICYT Fellowship Contract
Alba Herráiz, PhD Student | JAEPRE-CSIC Fellowship • LINCGlobal
Nashwa Elshaer, PhD Student | JAEPRE-CSIC Fellowship
Carlos Vásquez, PhD Student | CONICYT Fellowship Contract
Guillem Ylla, Bioinformatics Support | CSIC Project Contract

RESEARCH OUTLINE

In our group, we aim at understanding the mechanisms that regulate insect oogenesis. The peculiarity of our research in this field is to use a poorly modified insect like the cockroach *Blattella germanica*, as a model. We are working on the identification of genes responsible for the maturation of the oocyte and the establishment of anterior-posterior and dorsal-ventral axes, using ovary transcriptomes. We have identified genes that are of key importance in the regulation of these processes, and through RNA interference (RNAi) methodologies we are unveiling the function of some of them in the oocyte development of *B. germanica*, an insect with a panoistic ovary type, the most primitive type between insects. The next step in our research will be to study how is regulated the function of these genes and which are the relationships between them that allow the proper development of the oocyte. Comparison of the results obtained with those already described in other, more modified insect species, suggests that some functions are conserved in evolution, although the regulation of these functions could have changed.

RESEARCH SUBLINES

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

1. Studying the genes that in more modified species determine oocyte polarization

The establishment of a symmetry axis is crucial for development in many organisms. In animals this occurs in the early steps of embryogenesis by an asymmetric distribution of mRNAs from maternal origin that localize in particular regions of the oocyte, determining its polarization. To study oocyte polarization in our primitive model, we have chosen genes that have been well studied in

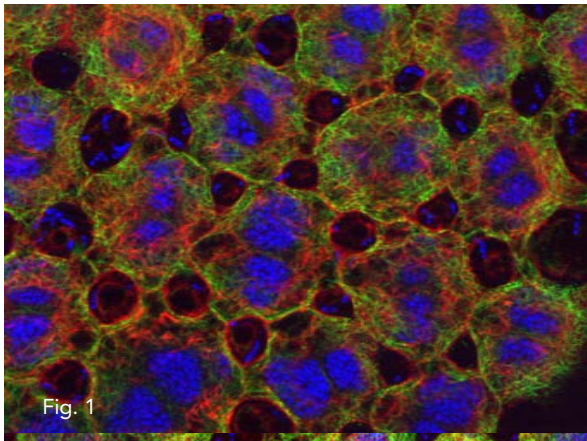


Fig. 1

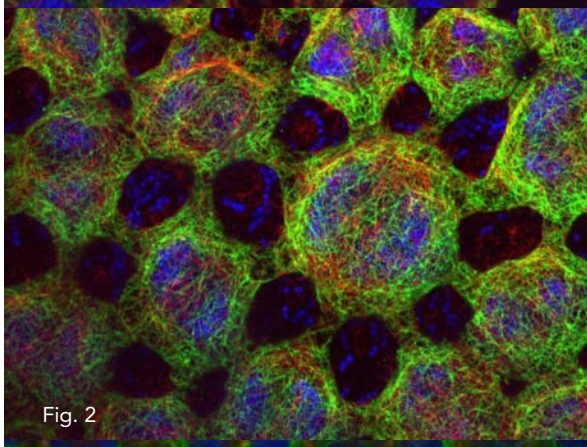


Fig. 2

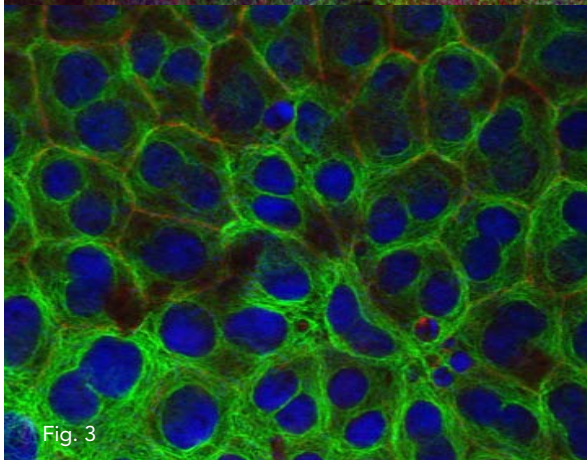


Fig. 3

Fig. 1: *Follicular cells from an ovarian follicle of* Blattella germanica *at vitellogenesis. Microtubules of ß-tubulin are labeled in green, actin fibers in red (phalloidin-TRICT) and the nuclei in blue (DAPI).* © M.D. Piulachs

Fig. 2: *Follicular cells from an ovarian follicle of* Blattella germanica *at vitellogenesis. Microtubules of ß-tubulin are labeled in green, actin fibers in red (phalloidin-TRICT) and the nuclei in blue (DAPI).* © M.D. Piulachs

Fig. 3: *Follicular cells from an ovarian follicle of* Blattella germanica *at the beginning of vitellogenesis. Microtubules of ß-tubulin are labeled in green and the nuclei appeared in blue (DAPI).* © M.D. Piulachs

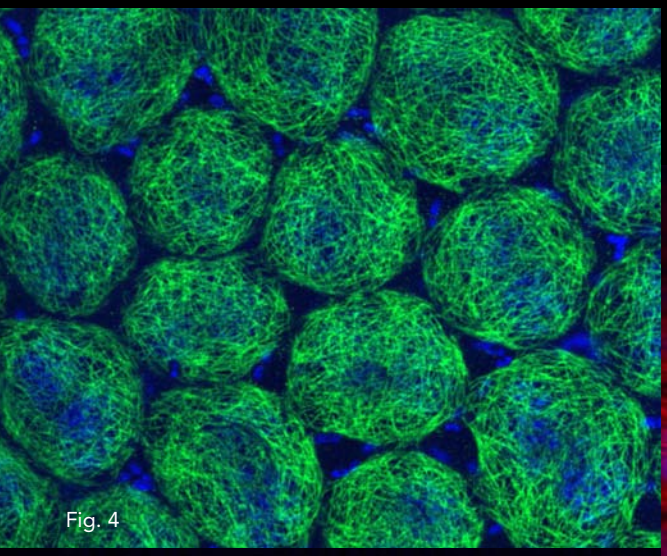


Fig. 4

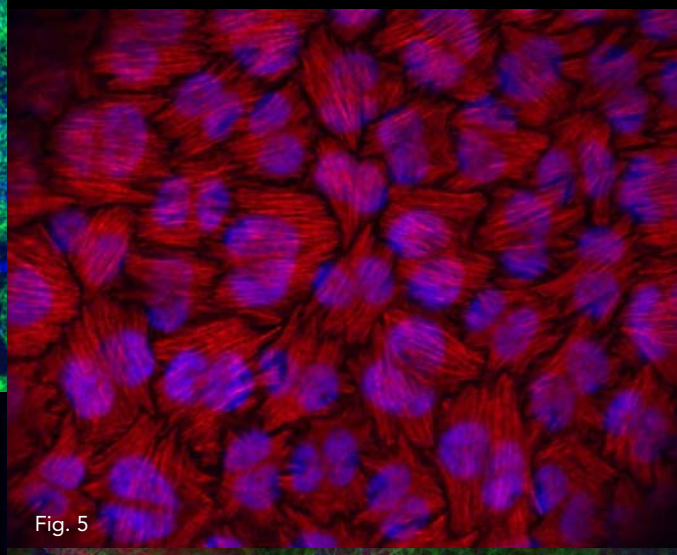


Fig. 5

Fig. 6

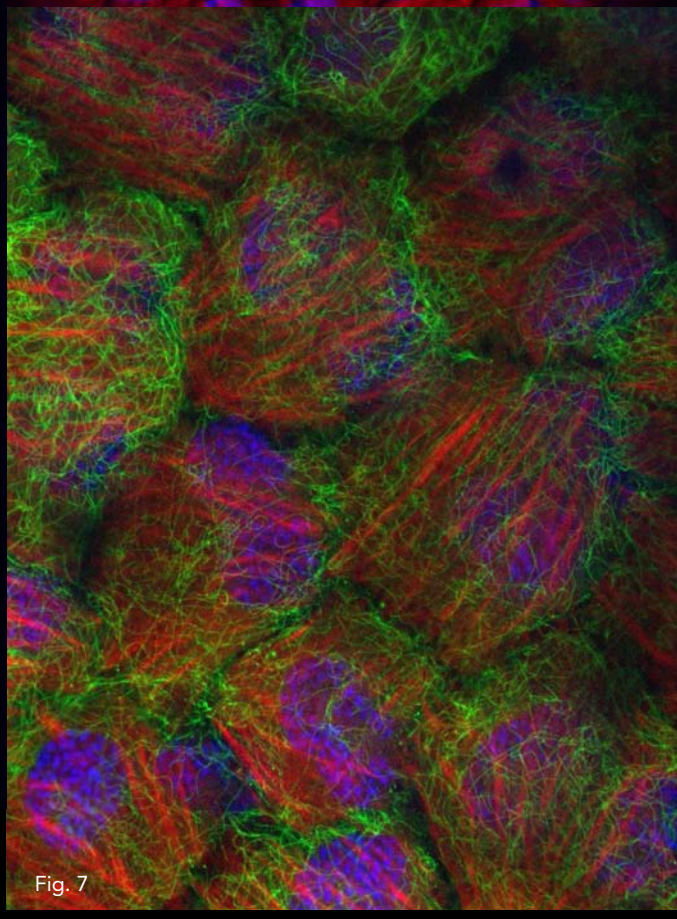


Fig. 7

Drosophila melanogaster like *capicua*, *oskar* and *EGFR* among others, and we are describing their function in a panoistic ovary. Our aim is to understand whether the function of these genes is conserved or has changed in the transition from the panoistic to the meroistic ovarian type.

2. Studying the regulation of cell proliferation during oogenesis

The rate between cell proliferation and cell death is a critical parameter determining tissue growth, and the Hippo pathway is crucial to understand the dynamics of the growth process. Understanding the regulation of the Hippo pathway is important to study not only how do cells proliferate, but also how do they avoid overproliferation, which could lead to tumorigenesis. Our objective has been to study the Hippo pathway in vivo, using our cockroach model. We identified their components, described their precise function using the RNAi methodologies, and we are also determining the possible regulatory role of miRNAs in the modulation of the different components of the pathway.

PUBLICATIONS 2013

ISI Articles

- Irles, P., Silva-Torres, F.A., and Piulachs, M.D. 2013. RNAi reveals the key role of Nervana 1 in cockroach oogenesis and embryo development. *Insect Biochemistry and Molecular Biology* 43: 178-188.

FUNDED PROJECTS

Project Title: *Searching the origin of oocyte polarization in insects*
Financed by: *MINECO*
Years: *2012-2014*
PI: *Maria-Dolors Piulachs*

Project Title: *Insect pest control with RNAi*
Financed by: *CSIC and NSC, Taiwan*
Years: *2011-2013*
PIs: *Xavier Bellés and How-Jing Lee*

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

Fig. 4: *Microtubules of ß-tubulins (green) surrounding the follicular cells of* Blattella germanica. *In blue (DAPI) is stained the cell nuclei.* © M.D. Piulachs

Fig. 5: *Follicular cells from an ovarian follicle of* Blattella germanica *at the end of the gonadotrophic cycle. Actin fibers (red) are stained with phalloidin-TRICT and nuclei with DAPI (blue).* © M.D. Piulachs

Fig. 6: Blattella germanica *panoistic ovariole. Actin fibers (red) are stained with phalloidin-TRICT and nuclei with DAPI (blue).* © M.D. Piulachs

Fig. 7: *Follicular cells from an ovarian follicle of* Blattella germanica *at the end of the gonadotrophic cycle. Microtubules of ß-tubulin are labeled in green, actin fibers in red (phalloidin-TRICT) and the nuclei in blue (DAPI).* © M.D. Piulachs

GROUP

MULTICELLGENOME

GROUP MEMBERS

Iñaki Ruiz-Trillo, Group Leader

| ICREA Research Professor

Hiroshi Suga, Postdoctoral Researcher
Núria Sánchez, Postdoctoral Researcher
Javier del Campo, Postdoctoral Researcher
Matija Harcet, Marie Curie Postdoctoral Researcher
Arnau Sebé-Pedrós, PhD Student
Alex de Mendoza, PhD Student
Guifré Torruella, PhD Student
Xavier Grau-Bové, PhD Student
Helena Parra, PhD Student
David López-Escardó, Master Student
Meritxell Antó, Research Technician
Maria José Barberà, Research Technician
Lourdes Riquelme, Master Student

RESEARCH OUTLINE

We want to understand how unicellular organisms became multicellular. Specifically, we focus on the origin of multicellular animals or metazoans. To this end, we compare the genomes of animals with the genomes of their closest unicellular relatives.

RESEARCH LINES

1. Biodiversity and Molecular Ecology of Opisthokonts

The real diversity of opisthokonts remains unknown. To address this we analysed environmental data and identified several novel opisthokont clades. To increase our understanding, we are currently analysing molecular data from the Biomarks project to have a better idea of the real diversity of the different opisthokont lineages.

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, is obtaining the genome sequence from several of the closest unicellular relatives of both animals and fungi (see the Multicellularity Project at Broad). By performing comparative genomic analyses we will unravel the genome structure and gene composition of the last common unicellular ancestor that gave rise to Metazoa.

For example, we have recently analysed the genome sequence of the filasterean amoeboid *Capsaspora owczarzaki*, a close unicellular relative of Metazoa. We identified in the *Capsaspora* genome several genes that are required for metazoan development, such as protein tyrosine kinases, integrins, and several transcription factors. This implies that the unicellular ancestor of animals was much more complex than previously thought.

Currently we are obtaining the genome sequence of several ichthyosporean taxa.

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

We want to know what roles the genes involved in multicellularity are playing in the unicellular *Capsaspora*, and how these genes were later on co-opted to the new functions in metazoans. Thus, 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 on 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 closes 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. We are currently working on having the most taxon-rich phylogenomic analysis of the opisthokonts.

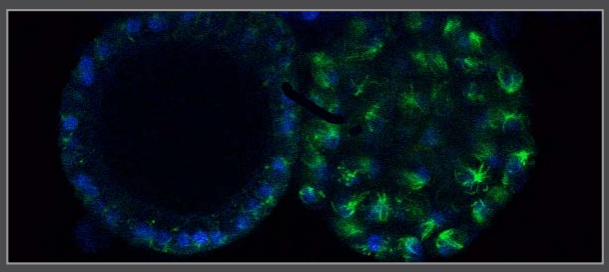


Fig. 1: *Immuno staining of* Creolimax fragrantissima. *Nucleous (DAPI- blue) and tubiline (green).* © H. Parra

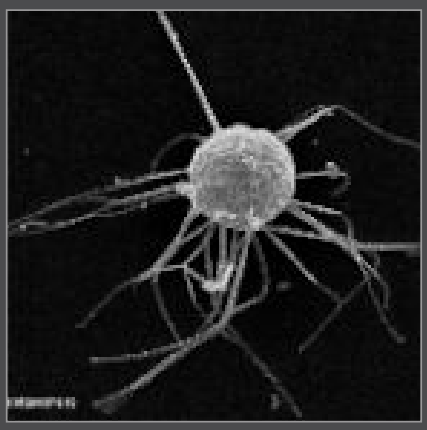


Fig. 2: Ministeria vibrans. *SEM picture.* © A. Sebé-Padrós

PUBLICATIONS 2013

ISI Articles

- del Campo, J., and Ruiz-Trillo, I. Environmental survey meta-analysis reveals hidden diversity among unicellular opisthokonts. *Molecular Biology and Evolution* 30 (4): 802-805.
- de Mendoza, A., Sebé-Pedrós, A., Šestak, M.S., Matejčić, M., Torruella, G., Domazet-Lošo, T., Ruiz-Trillo, I. 2013. Transcription factor evolution in eukaryotes and the assembly of the regulatory toolkit in multicellular lineages. *Proceedings National Academic of Sciences USA* 110 (50) E4858-E4866.
- Grau-Bové, X., Sebé-Pedrós, A., and Ruiz-Trillo, I. A genomic survey of HECT ubiquitin ligases in eukaryotes reveals independent expansions of the HECT system in several lineages. *Genome Biology and Evolution* 5 (5): 833-47. doi: 10.1093/gbe/evt052.
- Martín-Durán, J.M., de Mendoza, A., Sebé-Pedrós, A., Ruiz-Trillo, I., and Hejnol, A. A broad genomic survey reveals multiple origins and frequent losses in the evolution of respiratory hemerythrins and hemocyanins. *Genome Biology and Evolution* 5 (7): 1435-42.
- Paps, J., Medina-Chacón, L.A., Marshall, W., Suga, H., and Ruiz-Trillo, I. Molecular phylogeny of Unikonts: new insights into the position of apusomonads and ancyromonads and the internal relationships of opisthokonts. *Protist* 164 (1): 2-12.

- Sebé-Pedrós, A., Ariza-Cosano, A., Weirauch, M.T., Leininger, S., Yang, A., Torruella, G., Adamski, M., Adamska, M., Hughes, T.R., Gómez-Skarmeta, J.L., Ruiz-Trillo, I. 2013. Early evolution of the T-box transcription factor family. *Proceedings National Academic of Sciences USA* 110 (40): 16050-5.
- Sebé-Pedrós, A., Burkhardt, P., Sánchez-Pons, N., Fairclough, S.R., Lang, B.F., King, N., and Ruiz-Trillo, I. Insights into the origin of metazoan filopodia and microvilli. *Molecular Biology and Evolution* 30 (9): 2013-23.
- Sebé-Pedrós, A., Irimia, M., del Campo, J., Parra-Acero, H., Russ, C., Nusbaum, C., Blencowe, B.J., Ruiz-Trillo, I., Tautz, D. 2013. Regulated aggregative multicellularity in a close unicellular relative of Metazoa. *eLife* 2013;2: e01287.
- Suga, H., and Ruiz-Trillo, I. 2013. Development of ichthyosporeans sheds light on the origin of metazoan multicellularity. *Developmental Biology* 377, 284-292.
- Suga, H., Chen, Z., de Mendoza, A., Sebé-Pedrós, A., Brown, M.W., Kramer, E., Carr, M., Kerner, P., Vervoort, M., Sánchez-Pons, N., Torruella, G., Derelle, R., Manning, G., Lang, B.F., Russ, C., Haas, B.J., Roger, A.J., Nusbaum, C., Ruiz-Trillo, I. The Capsaspora genome reveals a complex unicellular prehistory of animals. *Nature Communications* 4: 2325 doi: 10.1038/ncomms3325 (2013).

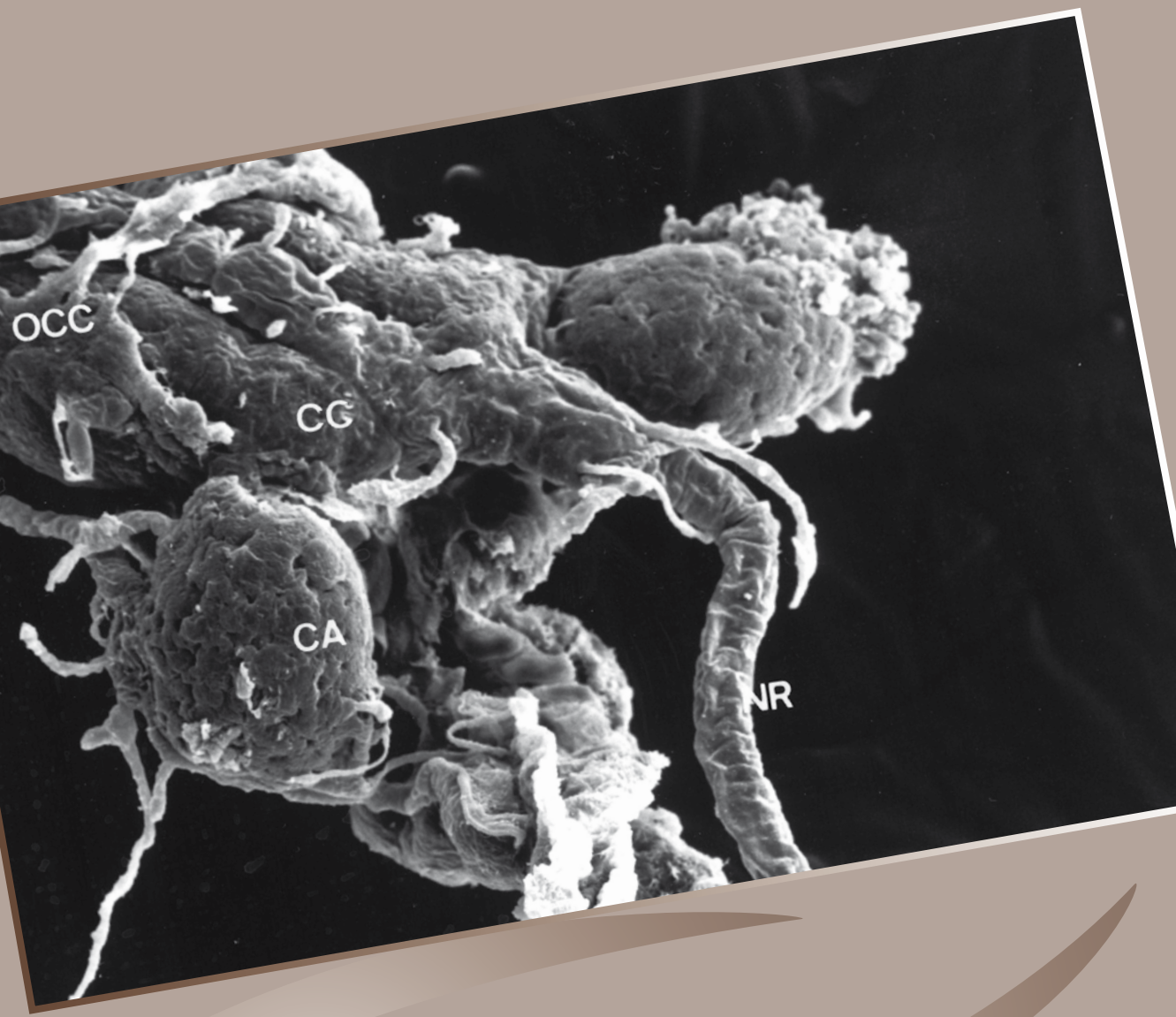
Other Publications

- de Mendoza, A., Sebé-Pedrós, A., and Ruiz-Trillo, I. El origen de la multicelularidad. *Investigación y Ciencia*, Febrero 2013 pp. 32-39.

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*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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: diversity 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 theprocesses 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

GROUP MEMBERS

Elena Bosch, Group Leader

| Associate Professor, UPF

Johannes Engelken, Post-doc | Visiting Researcher, 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 SUBLINES

1. Recent human adaptation and immunity

We are experimentally testing a number of functional variants for candidate genes related to immunity which do show signatures of recent adaptation in human populations, possibly as an adaptive response to pathogen interaction. As an example, we have identified a non-synonymous polymorphism in the human ZIP4 transporter with a strong geographical population differentiation. Further, we speculate that the reduced zinc uptake we detected for the derived variant may have been advantageous in Sub-Saharan Africa, possibly by reducing access of a geographically restricted pathogen to this micronutrient.

2. Recent human adaptation and nutrition

Micronutrients play an important role in human health and their physiological and cellular concentrations are kept in homeostasis by a number of membrane transport proteins and metal-binding proteins. Our goal is describe the interplay between genetic variation, mRNA and protein expression, together with trace elements content in different human tissue samples in order to gain insight into possible adaptive responses to nutrient availability and diet changes occurred in our past.

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

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

4. Rare variants in Parkinson's disease (PD)

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.

ISI Articles

- Farfán, M., Spataro, N., Sanglas, A., Albarral, V., Lorén, J.G., Bosch, E., and Fusté, M.C. 2013. Draft Genome Sequence of the *Aeromonas diversa* Type Strain. *Genome Announcements* 1 (3).
- Prado-Martinez, J., Hernando-Herraez, I., Lorente-Galdos, B., Dabad, M., Ramirez, O., Baeza-Delgado, C., Morcillo-Suarez, C., Alkan, C., Hormozdiari, F., Rainieri, E., Estelle, J., Fernandez-Callejo, M., Valles, M., Ritscher, L., Schoneberg, T., de la Calle-Mustienes, E., Casillas, S., Rubio-Acero, R., Mele, M., Engelken, J., Caceres, M., Gomez-Skarmeta, J., Gut, M., Bertranpetit, J., Gut, I., Abello, T., Eichler, E.E., Mingarro, I., Lalueza-Fox, C., Navarro, A., and Marques-Bonet, T. 2013. The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild. *BMC Genomics* 14 (1): 363..
- Sturm, S., Engelken, J., Gruber, A., Vugrinec, S., G Kroth, P., Adamska, I., and Lavaud, J. 2013. A novel type of light-harvesting antenna protein of red algal origin in algae with secondary plastids. *BMC Evolutionary Biology* 13 (1): 159.

Other Publications

- Spataro, N., Farfán, M., Albarral, V., Sanglas, A., Lorén, J.G., Fusté, M.C., Bosch, E. 2013. Draft genome sequence of *Aeromonas molluscorum* strain 848TT, isolated from bivalve molluscs. *Genome Announcements* 1 (3): e00382-13

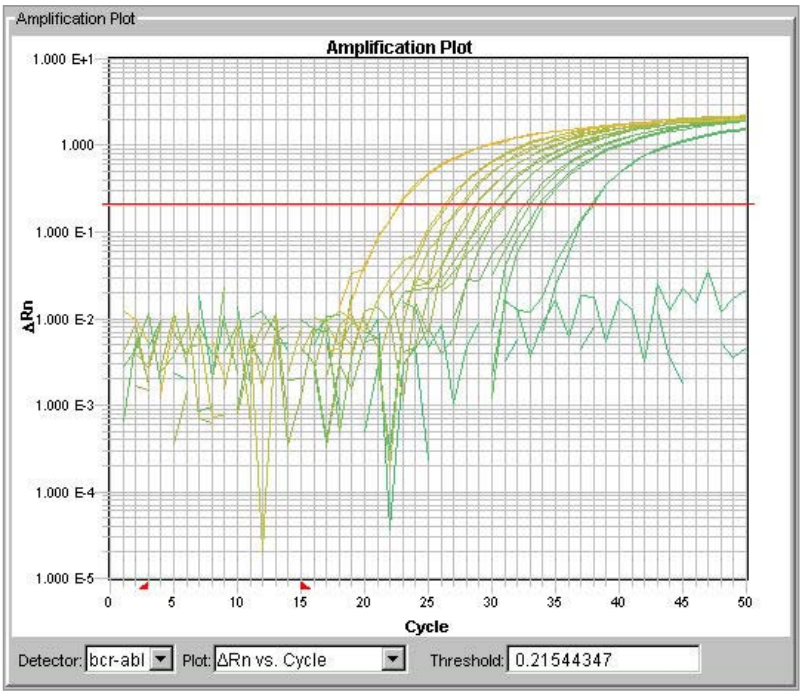

Fig. 1: *"Plot Real Time": Standard amplification plot for absolute gene expression quantification.*

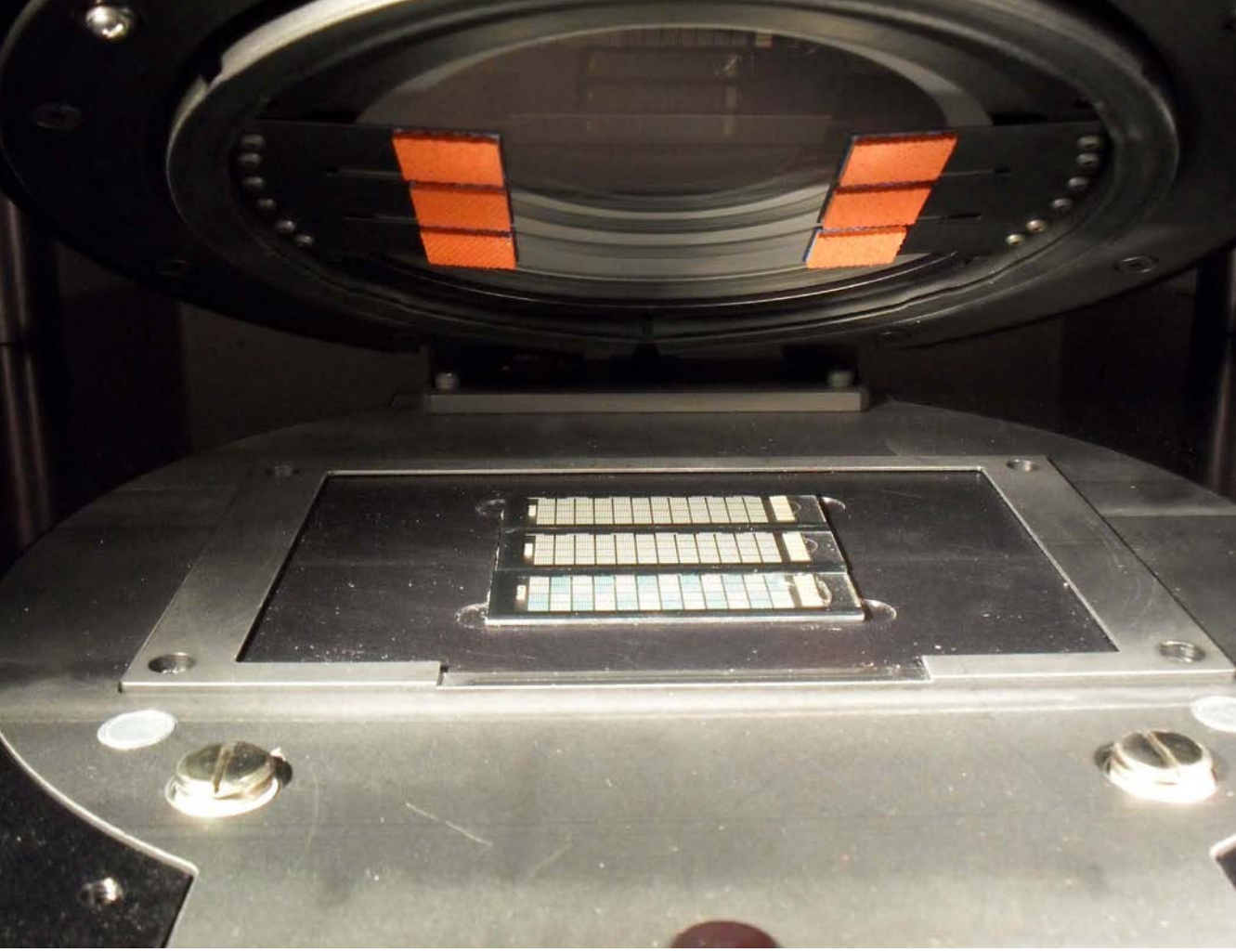

Fig. 2: *"Open Array": Open array system for gene expression quantification.*

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: *Variantes genéticas raras en la enfermedad de Parkinson: Aproximación evolutiva y re-secuenciación de alto rendimiento*
Financed by: *Subdirección General de Proyectos de Investigación (SAF2011-29239)*
Years: *2012-2014*
PI: *Elena Bosch*

GROUP
EVOLUTIONARY SYSTEMS BIOLOGY

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 | FI Scholarship, Generalitat de Catalunya

Begoña Dobón Berenguer | Master Student from UPF

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

RESEARCH SUBLINES

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

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

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

For the analysis of selection, in collaboration with Johannes Engelken, we have developed a pipeline for the detection of positive selection that calculates 21 tests which, 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, and Africa) that after will be used for other studies, in particular for the analysis of the ape genomes.

The pathways that we have analyzed are: N-glycosylation pathway; innate immunity; visual perception; obesity through adiposity signals; and the whole human metabolome. Special attention has been put into the quality of the

databases for the metabolic pathways, as their quality is worse than most studies assume and manual curation is needed in all cases.

2. Human genetic diversity and population history

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, including whole genome sequences for 70 individuals. In collaboration with Mihai Netea (Nijmegen Medical Center, The Netherlands) we have studied adaptation in Gypsies and Romanians, in a project that now includes whole genome sequences.

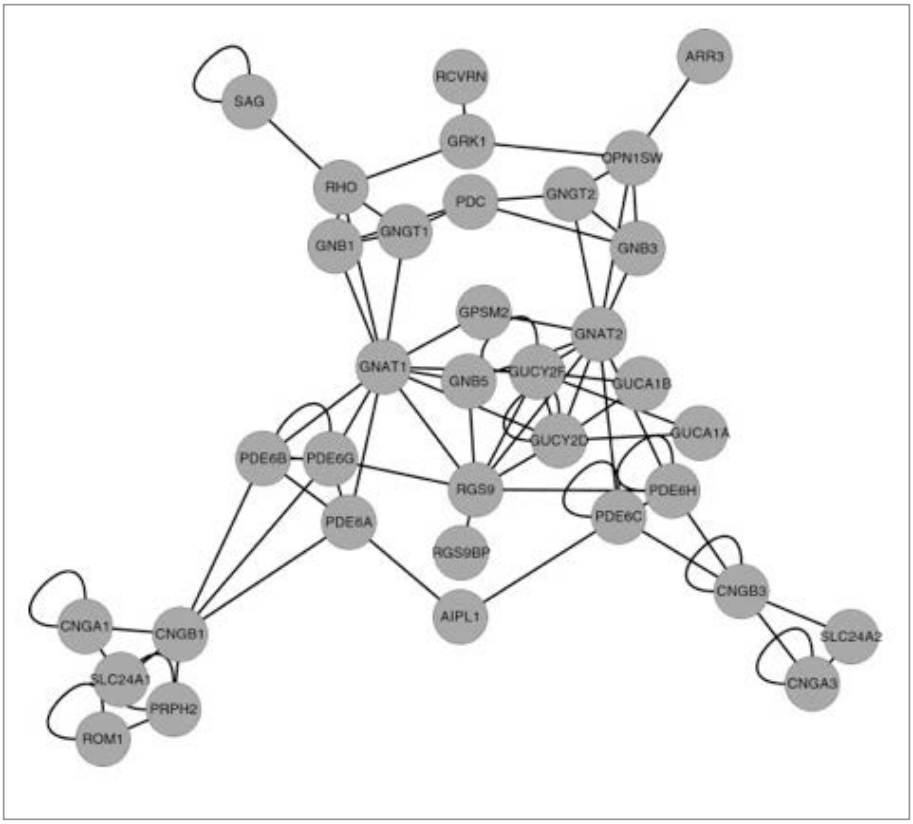

Fig. 1: *Phototransduction network. A network representation of the phototransduction signaling cascade. Nodes represent proteins and edges represent physical interactions between them.*

PUBLICATIONS 2013

ISI Articles

- Botigue, L.R., Henn, B.M., Gravel, S.Maples, B.K., Gignoux, C.R. Corona, E., Atzmon, G., Burns, E., Ostrer, H., Flores, C., Bertranpetit, J., Comas, D., and Bustamante, C.D. 2013. Gene flow from North Africa contributes to differential human genetic diversity in southern Europe. *Proceedings of the National Academy of Sciences USA* 110 (29): 11791-11796.
- Cagliani, R., Guerini, F.R., Rubio-Acero, R., Baglio, F., Forni, D., Agliardi, C., Griffanti, L., Fumagalli, M., Pozzoli, U., Riva, S., Calabrese, E., Sikora, M., Casals, F., Comi, G.P., Bresolin, N., Cáceres, M., Clerici, M., and Sironi, M. 2013. Long-Standing Balancing Selection in the THBS4 Gene: Influence on Sex-Specific Brain Expression and Gray Matter Volumes in Alzheimer Disease. *Human Mutation* 34: 743-753.
- Haber, M., Gauguier, D., Youhanna, S., Patterson, N., Moorjani, P., Botigué, L.R., Platt, D.E., Matisoo-Smith, E., Soria-Hernanz, D.F., Wells, R.S., Bertranpetit, J., Tyler-Smith, C., Comas, D., and Zalloua, P.A. 2013. Genome-Wide Diversity in the Levant Reveals Recent Structuring by Culture. *PLos Genetics* 9 (2): e1003316.
- Invergo, B.M., Montanucci, L., Koch, K.W., Bertranpetit, J., and Dell Orco, D. 2013. Exploring the rate-limiting steps in visual phototransduction recovery by bottom-up kinetic modeling. *Cell Communication and Signaling* 11 (1): 36.
- Invergo, B.M., Montanucci, L., Laayouni, H., and Bertranpetit, J. 2013. A system-level, molecular evolutionary analysis of mammalian phototransduction. *BMC Evolutionary Biology* 13 (1): 52.
- Prado-Martinez, J., Hernando-Herraez, I., Lorente-Galdos, B., Dabad, M., Ramirez, O., Baeza-Delgado, C., Morcillo-Suarez, C., Alkan, C., Hormozdiari, F., Rainieri, E., Estelle, J., Fernandez-Callejo, M., Valles, M., Ritscher, L., Schoneberg, T.,de la Calle-Mustienes, E., Casillas, S., Rubio-Acero, R., Mele, M., Engelken, J., Caceres, M., Gomez-Skarmeta, J., Gut, M., Bertranpetit, J., Gut, I., Abello, T., Eichler, E., Mingarro, I., Lalueza-Fox, C., Navarro, A., Marques-Bonet, T. 2013. The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild. *BMC Genomics* 14 (1): 363.

Publications as part of The Genographic Consortium

- Badro, D.A., Douaihy, B., Haber, M., Youhanna, S.C., Salloum, A., Ghassibe-Sabbagh, M., Johnsrud, B., Khazen, G., Matisoo-Smith, E., Soria-Hernanz, D.F., Wells, R.S., Tyler-Smith, C., Platt, D.E., and Zalloua, P.A., The Genographic, Consortium. 2013. Y-Chromosome and mtDNA Genetics Reveal Significant Contrasts in Affinities of Modern Middle Eastern Populations with European and African Populations. *PLos One* 8 (1): e54616.
- Brotherton, P., Haak, W., Templeton, J., Brandt, G., Soubrier, J., Jane Adler, C., Richards, S.M., Sarkissian, C.D., Ganslmeier, R., Friederich, S., Dresely, V., van Oven, M., Kenyon, R., Van der Hoek, M.B., Korlach, J., Luong, K., Ho, S.Y., Quintana-Murci, L., Behar, D.M., Meller, H., Alt, K.W, Cooper, A.; Genographic Consortium, Adhikarla, S., Ganesh Prasad, A.K., Pitchappan, R., Varatharajan Santhakumari, A., Balanovska, E., Balanovsky, O., Bertranpetit, J., Comas, D., Martínez-Cruz, B., Melé, M., Clarke, A.C., Matisoo-Smith, E.A., Dulik, M.C., Gaieski, J.B., Owings, A.C., Schurr, T.G., Vilar, M.G., Hobbs, A., Soodyall, H., Javed, A., Parida, L., Platt, D.E., Royyuru, A.K., Jin, L., Li, S., Kaplan, M.E., Merchant, N.C., John Mitchell, R., Der Sarkissian, C., Balanovsky, O., Brandt, G., Khartanovich, V., Buzhilova, A., Koshel, S., Zaporozhchenko, V., Gronenborn, D., Moiseyev, V., Kolpakov, E., Shumkin, V., Alt, K.W., Balanovska, E., Cooper, A., Haak, W.; Genographic Consortium. 2013. Ancient DNA reveals prehistoric

gene-flow from siberia in the complex human population history of North East Europe. *PLos Genetics* 9 (2): e1003296.
- Elhaik, E., Greenspan, E., Staats, S., Krahn, T., Tyler-Smith, C., Xue, Y., Tofanelli, S., Francalacci, P., Cucca, F., Pagani, L., Jin, L., Li, H., Schurr, T.G., Greenspan, B., Spencer Wells, R.; Genographic Consortium. 2013. The GenoChip: a new tool for genetic anthropology. *Genome Biology and Evolution* 5 (5): 1021-31.
- Haber, M., Gauguier, D., Youhanna, S., Patterson, N., Moorjani, P., Botigué, L.R., Platt, D.E., Matisoo-Smith, E., Soria-Hernanz, D.F., Wells, R.S., Bertranpetit, J., Tyler-Smith, C., Comas, D., and Zalloua, P.A. 2013. Genome-wide diversity in the Levant reveals recent structuring by culture. *PLos Genetics* 9 (2): e1003316.
- Renfrew, C., Lacerda, D.R., Santos, F.R., Soria Hernanz, D.F., Spencer Wells, R., Swamikrishnan, P., Tyler-Smith, C., Paulo Vieira, P., and Ziegle, J.S. 2013. Neolithic mitochondrial haplogroup H genomes and the genetic origins of Europeans. *Nature Communications* 4: 1764.

FUNDED PROJECTS

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

Project Title: *Genómica medica de la immunodeficiencia común variable (IDCV)*
Financed by: *Ministerio de Ciencia y Tecnología (BFU2010-19443)*
Years: *2013-2015*
PI: *Ferran Casals*

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: *MICINN acciones integradas con India*
Years: *2012-2014*
PI: *Jaume Bertranpetit*

GROUP

GENOMICS OF INDIVIDUALITY

GROUP MEMBERS

Francesc Calafell, Group Leader

| Associate Professor, UPF

Neus Solé Morata, Support Personnel | UPF Contract

RESEARCH OUTLINE

The general topics that interest us revolve around the genomics of individuality: what is there in our genomes that make 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 investigating Y-chromosome genetic diversity within samples of men carrying the same surname 2) we are characterizing the skin microbiome in healthy and psoriatic skin to reveal whether psoriasis has a microbial trigger; and 3) we are working on a case-control association study to detect any host genetic determinant of a poor progression in 2009 A (H1N1) influenza.

RESEARCH SUBLINES

1. A genetic atlas of Catalan surnames

Given their transmission, surnames behave as alleles at a locus in the Y chromosome, and they also carry linguistic, social, and historic information. We have selected a list of 50 Catalan surnames and have gathered ~50 men for each of those surnames, for a total sample of 2,550. We have typed 17

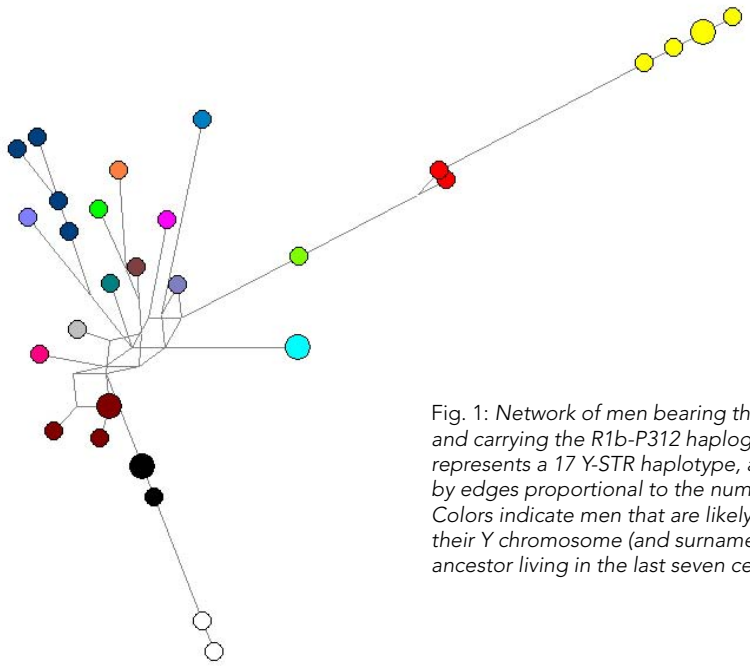

Fig. 1: *Network of men bearing the surname Soler and carrying the R1b-P312 haplogroup. Each circle represents a 17 Y-STR haplotype, and are connected by edges proportional to the number of mutations. Colors indicate men that are likely to have inherited their Y chromosome (and surname) from a common ancestor living in the last seven centuries.*

Y-chromosome STRs and 68 SNPs 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 polyphiletism, 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 is former first names of Germanic origin (Albert, Robert, Grau, etc). We will compare some of those to patronymic surnames of Latin origin (Oriol, Pons, etc). 3) Is that also the case for ethnonym surnames? Some 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. 4) What is the probability of identifying the surname of the anonymous donor of a biological sample, in a forensic situation? What would be the false positive rate?

This project is in collaboration with David Comas and Jaume Bertranpetit (IBE), and is mostly undertaken by Neus Solé.

2. The skin microbial biota in health and disease

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

3. Genetic susceptibility factors in poor influenza progression

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

PUBLICATIONS 2013

ISI Articles

- Charlier, P., Olalde, I., Solé-Morata, N., Ramírez, O., Babelon, J.P., Galland, B., Calafell, F., and Lalueza-Fox, C. 2013. Genetic comparison of the head of Henri IV and the presumptive blood from Louis XVI (both Kings of France). *Forensic Science International* 226: 38-40.
- Garcia-Garcerà, M., Garcia-Etxebarria, K., Coscollà, M., Latorre, A., and Calafell, F. 2013. A new method for extracting skin microbes allows metagenomic analysis of whole-deep skin. *PLos One* 8 (9): e74914.

Other Publications

- Calafell, F. 2013. Els camins paral.lels de gens i cognoms. *Paratge*, 25: 59-68.
- Calafell, F. 2013. Els noms dels catalans del segle XXI. *Paratge*, 26: 159-176.

FUNDED PROJECTS

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

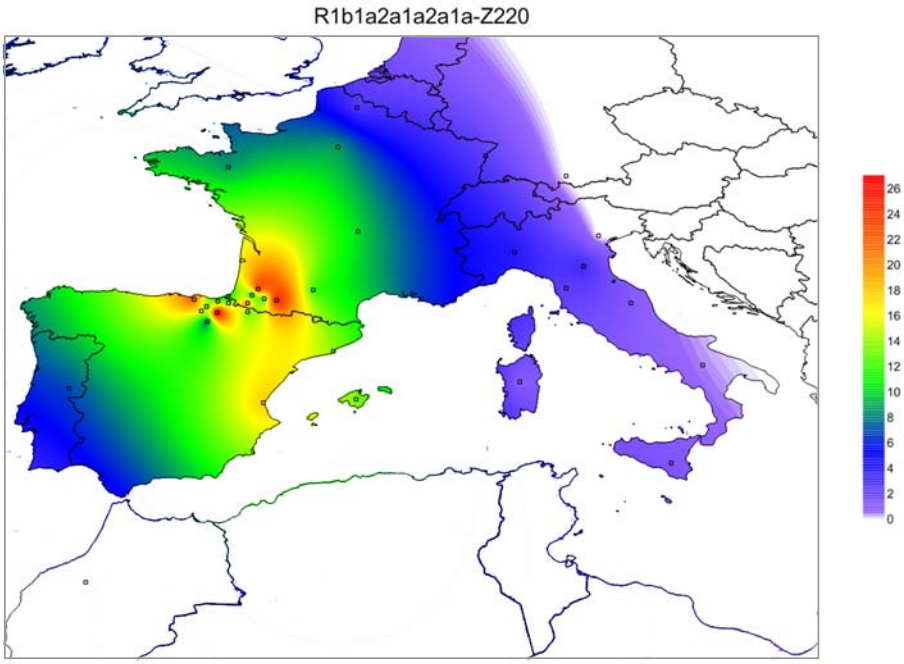

Fig. 2: *Estimated population frequencies (in percent) of haplogrup R1b-Z220 in Western Europe.*

GROUP

HUMAN GENOME: DIVERSITY AND ADAPTATION

GROUP MEMBERS

David Comas, Group Leader

| Associate Professor, UPF

| Subgroup: Human genome diversity

David Comas, Associate Professor, UPF
Isabel Mendizabal, Post-doc, ICREA Scholarship
Marc Haber, PhD Student, The Genographic Project
Lara Rubio Arauna, PhD Student, UPF Scholarship
Javier Mendoza, Visiting Predoctoral Researcher
Gerard Serra, Master Student, UPF
Àlex Mas, Master Student, UPF

| Subgroup: microRNAs in human adaptation and disease

Yolanda Espinosa-Parrilla, Visitor Professor, UPF
Ingrid Balcells, Post-doc, ERC StGt Project Contract
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 closest 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 taking into consideration both the protein-coding and non-protein coding portions of the genome.

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 microRNAs in primates

PUBLICATIONS 2013

ISI Articles

- Boattini, A., Martinez-Cruz, B., Sarno, S., Harmant, C., Useli, A., Sanz, P., Yang-Yao, D., Manry, J., Ciani, G., Luiselli, D., Quintana-Murci, L., Comas, D., Pettener, D. and the Genographic Consortium. 2013. Uniparental markers in Italy reveal a sex-biased genetic structure and different historical strata. *PLos One* 8 (5) e65441.
- Botigué, L.R., Henn, B.M., Gravel, S., Maples, B.K., Gignoux, C.R., Corona, E., Atzmon, G., Burns, E., Ostrer, H., Flores, C., Bertranpetit, J., Comas, D., and Bustamante, C.D. 2013. Gene flow from North Africa contributes to differential human genetic diversity in Southern Europe. *Proceedings of the National Academy of Sciences USA* 110: 11791-11796.
- Fadhlaoui-Zid, K., Haber, M., Martinez-Cruz, B., Zalloua, P., Elgaaied, A.B., and Comas, D. 2013. Genome-wide and paternal diversity reveal a recent origin of human populations in North Africa. *PLos One* 8 (11): e80293.
- Haber, M., Gauguier, D., Youhanna, S., Patterson, N., Moorjani, P., Botigué, L.R., Platt, D.E., Matisoo-Smith, E., Soria-Hernanz, D.F., Wells, S., Bertranpetit, J., Tyler-Smith, C., Comas, D., and Zalloua, P.A. 2013. Genome-wide diversity in the Levant reveals recent structuring by culture. *PLos Genetics* 9 (2): e1003316.
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Fig. 1: *Routine transfection experiment of mammalian cultured cells with reporter vectors expressing different microRNAs under study and a green fluorescent protein.* © E. García-Ramallo

• Prado-Martinez, J., Sudmant, P.H., Kidd, J.M., Li, H., Kelley, J.L., Lorente-Galdos, B., Veeramah, K.R., Woerner, A.E., O´Connor, T.D., Santpere, G., Cagan, A., Theunert, C., Casals, F., Laayouni, H., Munch, K., Hobolth, A., Halager, A.E., Malig, M., Hernandez-Rodriguez, J., Hernando-Herraez, I., Prufer, K., Pybus, M., Johnstone, L., Lachmann, M., Alkan, C., Twigg, D., Petit, N., Baker, C., Hormozdiari, F., Fernandez-Callejo, M., Dabad, M., Wilson, M.L., Stevison, L., Camprubi, C., Carvalho, T., Ruiz-Herrera, A., Vives, L., Mele, M., Abello, T., Kondova, I., Bontrop, R.E., Pusey, A., Lankester, F., Kiyang, J.A., Bergl, R.A., Lonsdorf, E., Myers, S., Ventura, M., Gagneux, P., Comas, D., Siegismund, H., Blanc, J., Agueda-Calpena, L., Gut, M., Fulton, L., Tishkoff, S.A., Mullikin, J.C., Wilson, R.K., Gut, I.G., Gonder, M.K., Ryder, O.A., Hahn, B.H., Navarro, A., Akey, J.M., Bertranpetit, J., Reich, D., Mailund, T., Schierup, M.H., Hvilsom, C., Andres, A.M., Wall, J.D., Bustamante, C.D., Hammer, M.F., Eichler, E.E., and Marques-Bonet, T. 2013. Great ape genetic diversity and population history. *Nature* 499 (7459): 471-475.

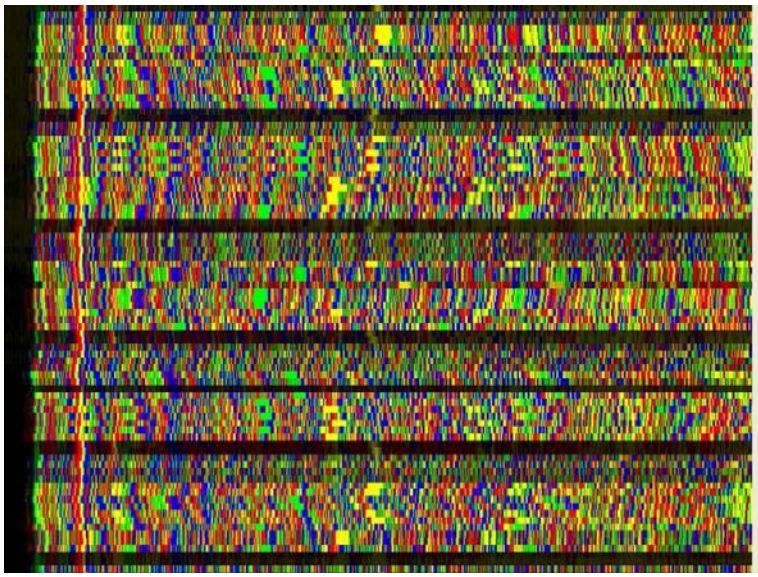

Fig. 2: *Raw data for Sanger sequencing in a* 96-capillary *ABI 3730xl DNA Analyzer.* © R. Anglada

• Prieto, L., Alves, C., Zimmermann, B., Tagliabracci, A., Prieto, V., Montesino, M., Whittle, M.R., Anjos, M.J., Cardoso, S., Heinrichs, B., Hernandez, A., López-Parra, A.M., Sala, A., Saragoni, V.G., Burgos, G., Marino, M., Paredes, M., Mora-Torres, C.A., Angulo, R., Chemale, G., Vullo, C., Sánchez-Simón, M., Comas, D., Puente, J., López-Cubría, C.M., Modesti, N., Aler, M., Merigioli, S., Betancor, E., Pedrosa, S., Plaza, G., Masciovecchio, M.V., Schneider, P.M., and Parson, W. 2013. GHEP-ISFG proficiency test 2011: Paper challenge on evaluation of mitochondrial DNA results. *Forensic Science International: Genetics* 7 (1): 10-15.
• Rebala, K., Martinez-Cruz, B., Tonjes, A., Kovacs, P., Stumvoll, M., Lindner, I., Buttner, A., Wichmann, H.E., Sivakova, D., Sotak, M., Quintana-Murci, L., Szczerkowska, Z., and Comas, D. 2013. Contemporary paternal genetic landscape of Polish and German populations: from early medieval Slavic expansion to post-World War II resettlements. *European Journal of Human Genetics* 21 (4): 415-422.

Other Publications

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FUNDED PROJECTS

Project Title: *Diversidad genómica en poblaciones humanas del norte de África y en poblaciones vecinas: inferencias sobre la estructura poblacional y migraciones (CGL2010-14944/BOS)*
Financed by: *Dirección General de Investigación Científica y Técnica, MCINN*
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: *MCINN. Programa "CSIC para el Desarrollo"*
Years: *2011-2013*
PI: *David Comas*

Project Title: *Variabilidad genética y genómica en regiones de microRNAs: hacía la identificación de novedades evolutivas y funcionales en microRNAs*
Financed by: *Ministerio de Educación y Ciencia (BFU2010-18477)*
Years: *2011-2013*
PI: *Yolanda Espinosa-Parrilla*

Fig. 3: *Individuals are the basis for population genetics analyses.* © D. Comas

SCIENTIFIC PUBLICATIONS

ISI Articles

- Abellán, P., Sánchez-Fernández, D., Picazo, F., Millán, A., Lobo, J.M., and Ribera, I. 2013. Preserving the evolutionary history of freshwater biota in Iberian National Parks. *Biological Conservation* 162: 116-126.
- Alcina, A., Fedetz, M., Fernández, O., Saiz, A., Izquierdo, G., Lucas, M., Leyva, L., García-León, J.A., Abad-Grau, M.D., Alloza, I., Antigüedad, A., Garcia-Barcina, M.J., Vandenbroeck, K., Varadé, J., de la Hera, B., Arroyo, R., Comabella, M., Montalban, X., Petit-Marty, N., Navarro, A. , Otaegui, D., Olascoaga, J., Blanco, Y., Urcelay, E., and Matesanz, F. 2013. Identification of a functional variant in the KIF5A-CYP27B1-METTL1-FAM119B locus associated with multiple sclerosis. *Journal of Medical Genetics* 50 (1): 25-33.
- Barbosa, F.F., Fernandes, A.S., and Oliveira, L.G. 2013. Three new species of *Macrelmis* Motschulsky, 1859 (Coleoptera: Elmidae: Elminae) from the Brazilian Cerrado Biome with updated key for the *Macrelmis* of Brazil. *Zootaxa* 3736: 128-142.
- Barbosa, F.F., Fernandes, A.S., Oliveira, L.G. 2013. Taxonomic key for the genera of Elmidae (Coleoptera, Byrrhoidea) occurring in Goiás State, Brazil, including new records and distributional notes. *Revista Brasileira de Entomologia* 57 (2): 149-156.
- Beukema, W., de Pous, P., Donaire-Barroso, D., Bogaerts, S., Garcia-Porta, J., Escoriza, D., Arribas, O., El Mouden, H., and Carranza, S. 2013. Review of the systematics, distribution, biogeography and natural history of Moroccan amphibians. *Zootaxa* 3661: 1-60.
- Beuls, K., and Steels, L. 2013. Agent-based models of strategies for the emergence and evolution of grammatical agreement. *PLos One* 8 (3).
- Boattini, A., Martinez-Cruz, B., Sarno, S., Harmant, C., Useli, A., Sanz, P., Yang-Yao, D., Manry, J., Ciani, G., Luiselli, D., Quintana-Murci, L., Comas, D., Pettener, D. and the Genographic Consortium. 2013. Uniparental markers in Italy reveal a sex-biased genetic structure and different historical strata. *PLos One* 8 (5) e65441.
- Botigué, L.R., Henn, B.M., Gravel, S., Maples, B.K., Gignoux, C.R., Corona, E., Atzmon, G., Burns, E., Ostrer, H., Flores, C., Bertranpetit, J., Comas, D., and Bustamante, C.D. 2013. Gene flow from North Africa contributes to differential human genetic diversity in Southern Europe. *Proceedings of the National Academy of Sciences USA* 110: 11791-11796.
- Boulan, L., Martín, D., Milán, M. 2013. bantam miRNA Promotes Systemic Growth by Connecting Insulin Signaling and Ecdysone Production. *Current Biology* 23 (6): 473-478.
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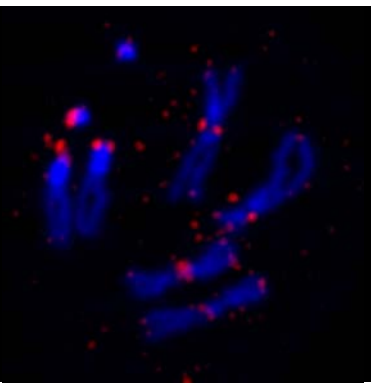

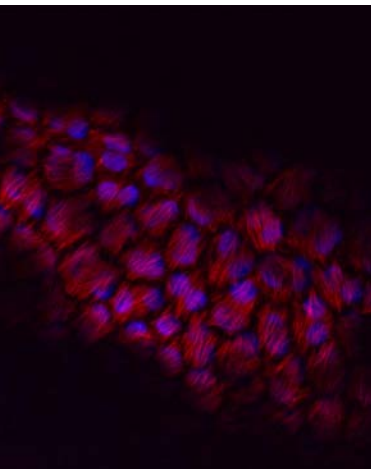

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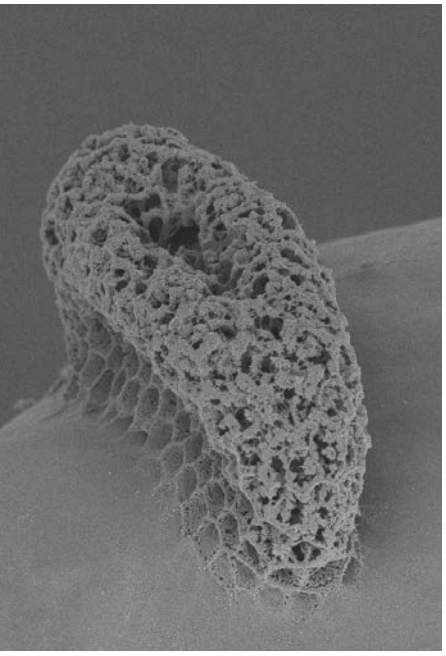

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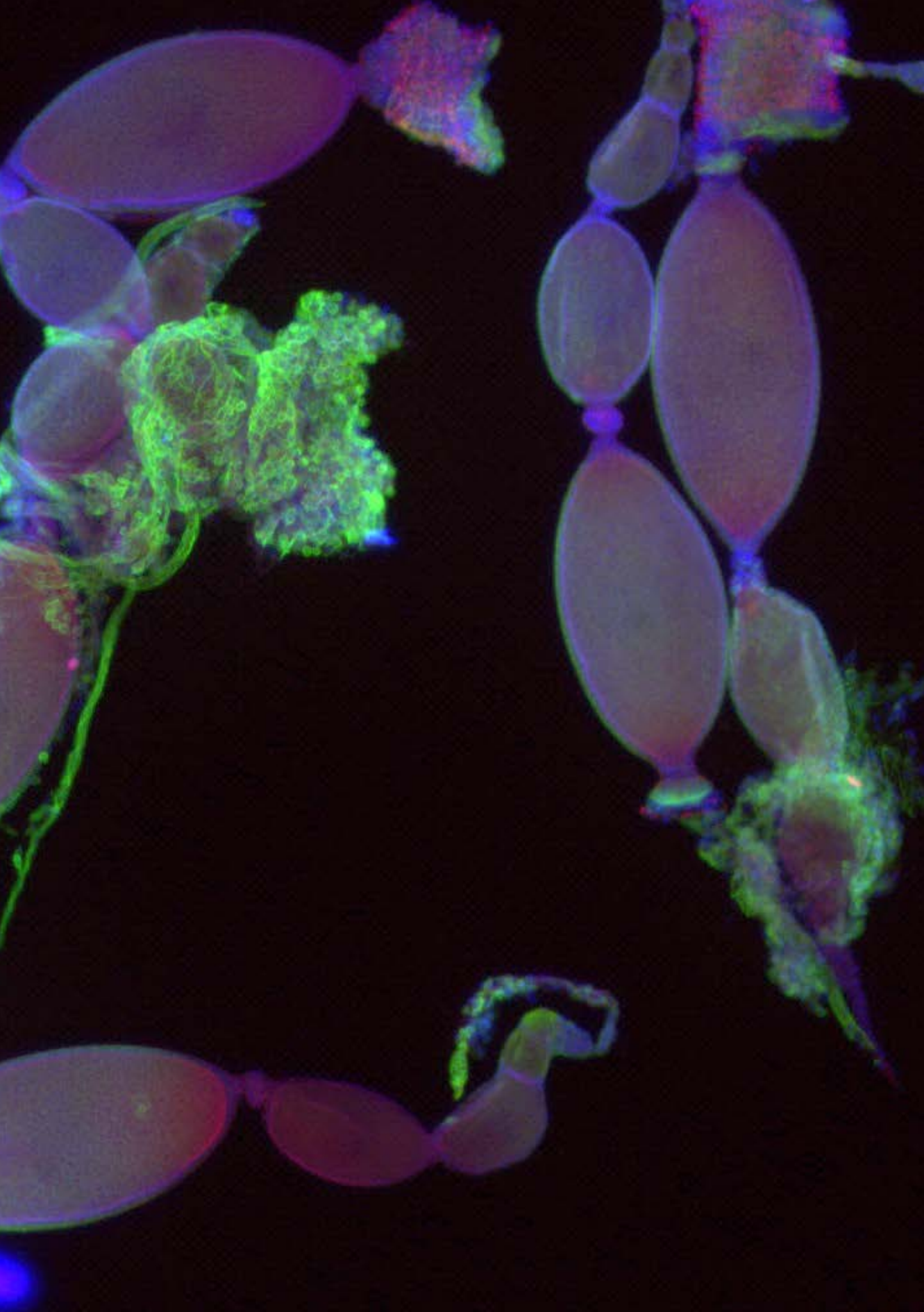

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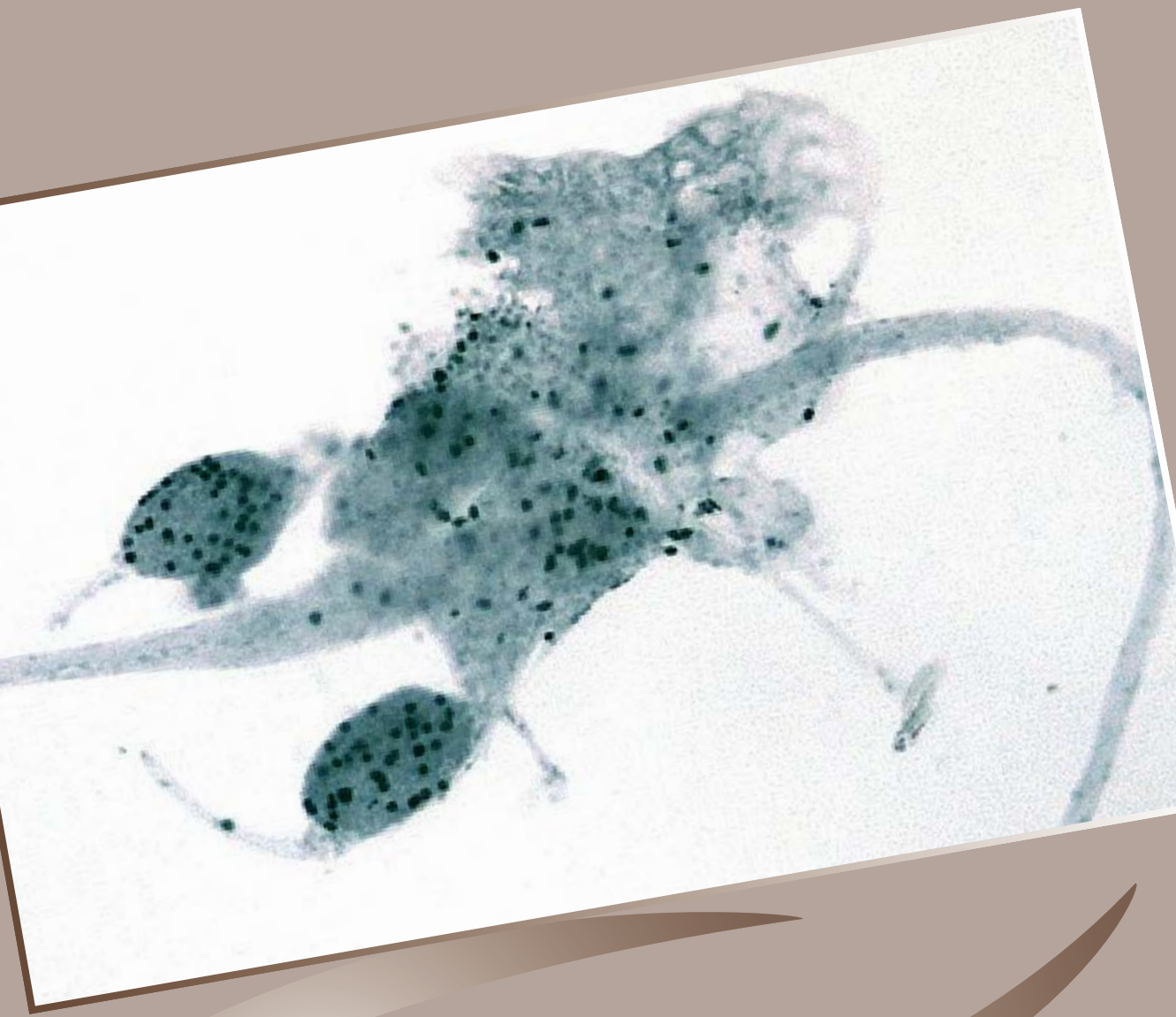

IBE HIGHLIGHTED PAPERS

GENETIC DIVERSITY AND POPULATION HISTORY IN HUMANS AND GREAT APES AND SELECTION FOR DUPLICATED SEQUENCES

Recent advances in genome sequencing technologies have allowed researchers to learn an enormous amount about human genomes and genetic diversity by sequencing individuals of our own species. In contrast however, far less attention has been focused on our great ape relatives. This year, and for the first time, we have sequenced the genomes of a large number of great apes from across Africa and South-East Asia. The study provides one of the most detailed and comprehensive analyses of genetic diversity of wild-born great apes to date—species which are now all considered endangered. We found that human genomes show relatively little variation between each other in comparison to most great apes, and that all of these species showed evidence of severe bottlenecks in their ancient history, possibly explaining the reduced genetic diversity.

One of the individuals analyzed is the world-famous gorilla "Snowflake" that was known for being the only albino gorilla. In a subsequent study, we found the mutation responsible for this unique phenotype and most surprisingly, the proportion of runs of homozygosity genome-wide revealed that this particular gorilla was a descendant of close relatives.

In the context of exploring new aspects of primate genomics, we also developed a new method to assess patterns of fast exon evolution, even in highly complex duplicated sequences without the need to rely on genome reference assemblies. We applied this method to study recent primate evolution and we showed for the first time that rapid exon evolution seemed to be more common than anticipated, possibly determining the evolutionary fate of newly originated sequences.

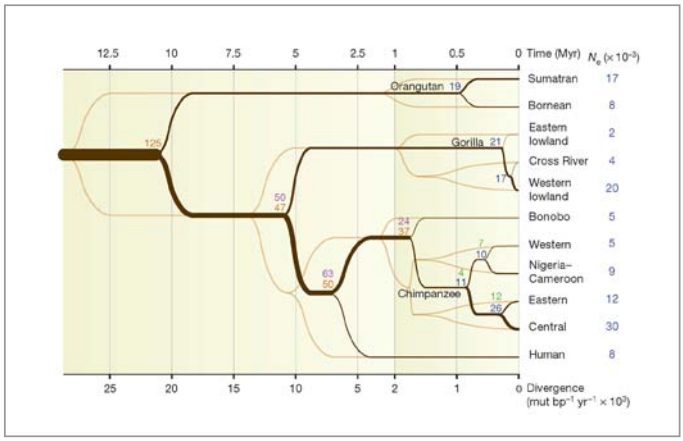

Fig. 1: *Inferred population history of the great apes. Population splits and effective population sizes (Ne) during great ape evolution. Split times (dark brown) and divergence times (light brown) are plotted as a function of divergence (d) on the bottom and time on top. The terminal Ne corresponds to the effective population size after the last split event.*

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DEMOGRAPHIC, LINGUISTIC AND CULTURAL CROSSROADS IN THE MEDITERRANEAN REVEALED BY GENETIC ANALYSES

The population history of humans in the Mediterranean has been characterized by multiple migrations and contacts among peoples from three different continents: Europe, Africa, and Asia. The Mediterranean Sea has acted as a bridge and as a barrier to demographic movements, and at times this has been correlated to cultural affinities. The availability of genomic data and the development of analytical tools to infer demographic events have allowed us to understand the past migration of human movements at the crossroads of the Mediterranean.

Focusing on the European continent, the highest genetic diversity of southern Europeans and the clinal gradient of genetic diversity in the continent have been suggested to be the result of three demographic processes: 1) the refugia in southern Europe during the last glacial maximum and the subsequent colonization of the continent from southern latitudes; 2) the gene flow coming from the Near East that has been associated with the dispersion of agriculture; and 3) the presence of migrations coming from the African continent into southern Europe. We tested this last scenario by the analysis of genome-wide data in several Mediterranean populations. Our data confirm that part of the highest diversity shown in southern European populations is in fact the result of gene flow from North Africa that has affected differentially extant European populations, with the highest genetic diversity observed in the Iberian Peninsula. Our data indicate that this gene flow is independent of the gene

flow from the Near East that has been detected in Europe. Our analyses suggest that the North African gene flow has been recent, and it could be linked to the Muslim arrival in Iberia through the Gibraltar Straits in historical times.

On the opposite eastern edge of the Mediterranean, in the Levant, our genetic data shows that recent cultural processes have prevented gene flow among close populations. The genome-wide data obtained from more than one thousand individuals shows a clear genetic structure driven by religious affiliations that have facilitated the admixture between culturally similar peoples and preventing gene flow from culturally different groups. Consequently, two major genetic components are found in the eastern part of the Mediterranean: a component related to Europe and Central Asia, and a second component with closer affinities to the Middle East and eastern Africa. The differentiation of

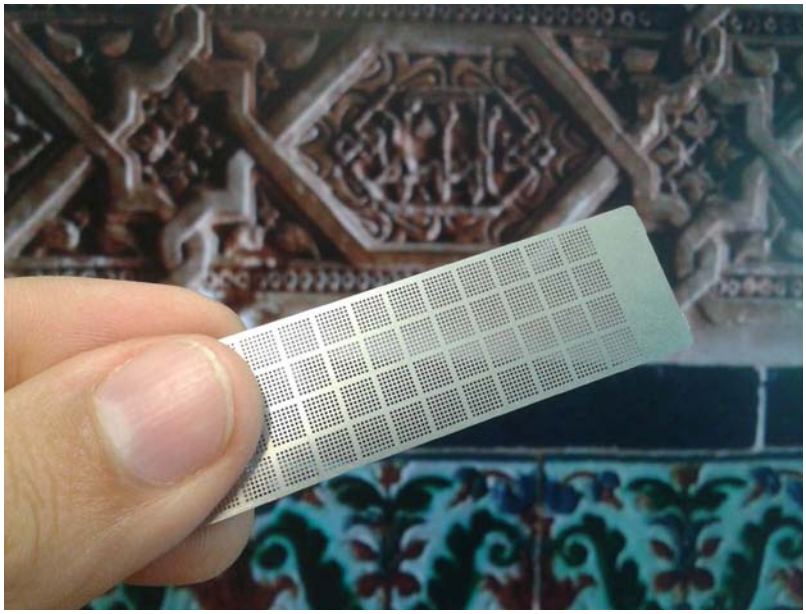

Fig. 1: *Human North African gene flow to Southern Europe has been evaluated with SNP chip arrays.*
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these genetic components is associated with the last glacial period and the start of farming in the region.

In summary, our genetic analyses have shown that the population history of the Mediterranean has been an amalgam of different and opposite processes of migration, admixture, and isolation from diverse population sources.

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THE GENOME OF *CAPSASPORA OWCZARZAKI* REVEALS THAT THE UNICELLULAR ANCESTOR OF METAZOA WAS GENETICALLY COMPLEX

The origin of multicellular animals or metazoans from their closest unicellular relatives is one of the most important evolutionary transitions in life history. However, despite its importance, this process remains poorly understood. To provide insights into this question, the genome of a close unicellular relative of Metazoa, that of *Capsaspora owczarzaki*, was sequenced (Suga et al. 2013). Analyses of this genome showed that the unicellular ancestor of Metazoa was much more complex than previously thought, since it already had a good repertoire of genes and pathways involved in cell adhesion and cell signaling.

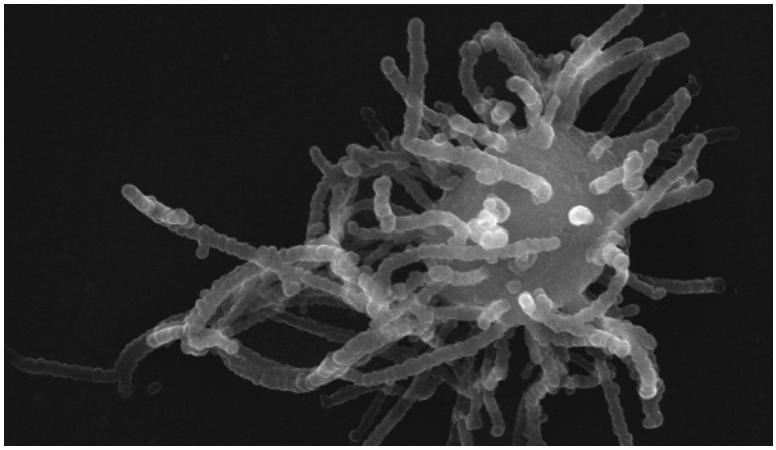

Fig. 1: Capsaspora owczarzaki, *a close unicellular relative of animals.*

Moreover, the life cycle of *C. owczarzaki* was deciphered and it includes an aggregative cell stage, the first one reported so far among close unicellular relatives of Metazoa (Sebé-Pedrós et al.). Indeed, *C. owczarzaki* has three different cell stages: filopodiated, cystic, and aggregative stages. RNAseq data obtained from three different replicates of each cell stage show that transitions between these three different cell stages are highly regulated at the level of gene expression and alternative splicing. Interestingly, genes involved in integrin adhesion are all significantly up-regulated in the aggregative stage, which suggests that integrins were anciently involved in the formation of aggregates before being co-opted into their function in metazoans.

The evolution of T-box genes, previously believed to be metazoan-specific, was analyzed (Sebé-Pedrós et al.). T-box genes are present in both filastereans and ichthyosporean, two of the three closest unicellular relatives of Metazoa. Interestingly, *C. owczarzaki* has a homolog of the T-box gene Brachyury, which

in animals is involved in gastrulation. Notably, by using heterologous expression in Xenopus, it was shown that Capsaspora-Brachyury mimics the endogenous Xenopus-Brachyury, showing that Capsaspora-Brachyury is highly conserved at a functional level. Further comparative analysis of the DNA binding motifs and the molecular behaviour of metazoans versus Capsaspora Brachyurys suggested that sub-functionalization of the T-box genes came at the onset of Metazoa, and probably, by the action of different co-factors.

Finally, the evolution of the genes involved in transcriptional regulation was analyzed at the level of all eukaryotes (de Mendoza et al. 2013). The analysis showed that both animals and plants have the most complex repertoire of genes involved in transcriptional regulation, both at the level of genes and protein domains. In contrast, other multicellular lineages, such as fungi or algae, have less transcription factors. This may be due to the fact that both animals and plants share a complex embryonic development, which requires a very strict control and, therefore, more transcription factors.

Reference Article

- de Mendoza, A., Sebé-Pedrós, A., Šestak, M.S., Matejčić, M., Torruella, G., Domazet-Lošo, T., Ruiz-Trillo, I. 2013. Transcription factor evolution in eukaryotes and the assembly of the regulatory toolkit in multicellular lineages. *Proceedings National Academic of Sciences USA* 110 (50) E4858-E4866. Ahead of print.
- Sebé-Pedrós, A., Ariza-Cosano, A., Weirauch, M.T., Leininger, S., Yang, A., Torruella, G., Adamski, M., Adamska, M., Hughes, T.R., Gómez-Skarmeta, J., LuisRuiz-Trillo, I. 2013. Early evolution of the T-box transcription factor family. *Proceedings of the National Academy of Sciences USA*. 110 (40):16050-16055. Ahead of print; doi: 10.1073/pnas.1311818110
- Sebé-Pedrós, A., Irimia, M., del Campo, J., Parra-Acero, H., Russ, C., Nusbaum, C., Blencowe, B.J., Ruiz-Trillo, I., Tautz, D. 2013. Regulated aggregative multicellularity in a close unicellular relative of metazoa. *eLife*. 2.
- Suga, H., Chen, Z., de Mendoza, A., Sebé-Pedrós, A., Brown, M.W., Kramer, E., Carr, M., Kerner, P., Vervoort, M., Sánchez-Pons, N., Torruella, G., Derelle, R., Manning, G., Lang, B.F., Russ, C., Haas, B.J., Roger, A.J., Nusbaum, C., Ruiz-Trillo, I. 2013. The Capsaspora genome reveals a complex unicellular prehistory of animals. *Nature Communications* 4.

TOGETHER IN HEALTH AND SICKNESS: EUROPEANS AND EAST ASIANS SHARE RISK ALLELES FOR THE MOST PREVALENT COMPLEX DISEASES

Describing and identifying the genetic variants that increase risk for complex diseases remains a central focus of human genetics and is fundamental for the emergent field of personalized medicine. Over the last seven years, Genome-Wide Association Studies (GWAS) have revolutionized the field, and hundreds of disease loci have been discovered thanks to this approach. However, with only a handful of exceptions, the causal variants that generate the associations unveiled by GWAS have not been identified, and their frequency and degree of sharing across populations remains unknown.

This study is the first comprehensive comparison of GWAS results published so far, and was carefully designed to try to understand the nature of causal variants. By examining the results of GWAS for 28 complex diseases that have been performed with individuals of European, East Asian, and African ancestries, Marigorta and Navarro reached some unexpected conclusions.

They started with the first systematic study of patterns of replication of GWAS results. Their first finding was that, in sharp contrast with past results from candidate gene association studies, the findings of GWAS tended to be positively replicated by independent studies. After showing that the results of GWAS are solid, the authors proceeded to cross-continental analysis, examining all the available results from GWAS performed with either European or East Asian individuals. In that second part of their analysis, they reported that the genes linked to disease are almost always the same in the two ethnic groups under study. In a third analysis, they show that the genetic markers that are associated with each disease within each gene tend to be the same across continents. Moreover: the magnitude of their effects is surprisingly similar, with odd ratios across continents having a significant correlation of r>0.8. Finally, they demonstrate that GWAS performed with larger sample sizes have detected variants with weaker effects, rather than variants with lower frequencies.

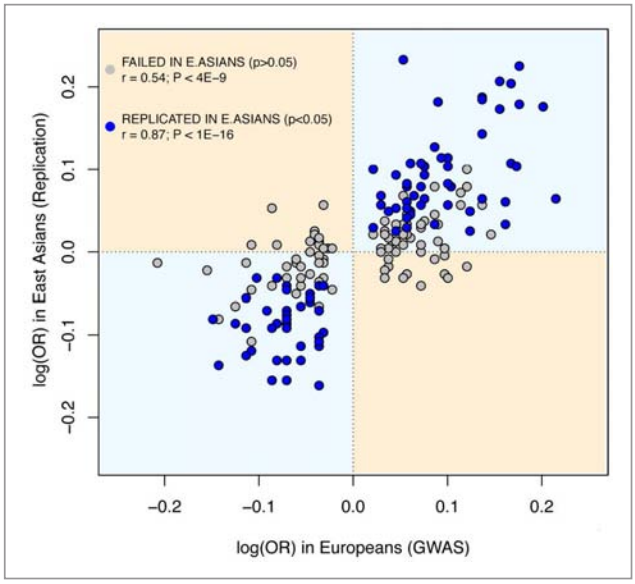

Fig. 1: *"Correlation between the effects of SNPs discovered in Europeans and the same SNPs in East Asians. The correlation exists even for failed replications, but is stronger for SNPs that are positively replicated with East Asians".* © J. Gómez-Zurita.

All these results suggest that the majority of SNP-disease associations afforded by GWAS are caused by common causal variants that are shared across Eurasians. In that sense, we all would be "Together in health and sickness". But that is not all; the findings by Marigorta and Navarro allow for several inferences. First, they contribute to the debate on the possible synthetic origin of GWAS associations, since trans-continental replicability confirms that most, and possibly all of the associations detected by GWAS cannot be caused by population-specific, rare variants (because rare variants are not usually shared across continents). Second, they clarify the contribution of common variants to extant GWAS results, since practically all GWAS have delivered precisely what they were designed to detect: associations with common variants. Finally, they show that a substantial proportion of the causal variants that are shared across European and East Asian populations probably lie in the regions close to marker SNPs, which may allow leveraging on the increasingly varied ancestries of GWAS to track them down.

This is one of several papers that shows that taking an evolutionary approach to the study of human disease helps to shed light not only on the origin of disease, but also on the potential design of strategies for disease prevention and treatment. For instance, thanks to this work we now know that human evolution allows for a majority of shared drug targets across the World, plus we are starting to have a better idea of what genes and what diseases may be exceptions to that rule.

Reference Article

- Marigorta, U., Navarro, A. 2013. High trans-ethnic replicability of GWAS results implies common causal variants. *PLos Genetics* 9 (6) e1003566

BANTAM miRNA PROMOTES SYSTEMIC GROWTH BY CONNECTIN INSULING SIGNALING AND ECDYSTEOID PRODUCTION

Understanding how animal growth is regulated is one of the main goals of biological research. During the development of higher organisms, cell and tissue growth is controlled by systemic signaling molecules, mostly hormones, which continuously integrate developmental, environmental, and physiological cues. These systemic signals, in turn, act in concert with tissue-autonomous regulatory systems to finely coordinate the final size of an organism. In the fruit fly *Drosophila melanogaster*, the main hormone controlling growth is the steroid ecdysone which acts at two different levels; whereas, peaks of this hormone act as developmental timers by promoting every developmental transition, basal ecdysone levels inhibit tissue growth during the intermolt period. Besides ecdysone, the second important signaling pathway involved in the control of growth is the insulin/IGF pathway. When nutrients are available during the larval growth stage in *Drosophila*, insulin-like peptides are released to the systemic circulation pathway promoting cell and tissue growth. The insulin-dependent cell-growth promoting effect is counteracted by the inhibitory effect of ecdysone. Remarkably, a regulatory feedback balance between insulin and ecdysone operates to control the final body size of the fruitfly as the insulin/IGF pathway itself also activates ecdysone biosynthesis in the prothoracic gland.

We have unraveled a very interesting role of a new component of the described feedback loop between ecdysone and insulin, the microRNA (miRNA) *bantam*. It is well known that *bantam*, the first miRNA-encoding gene

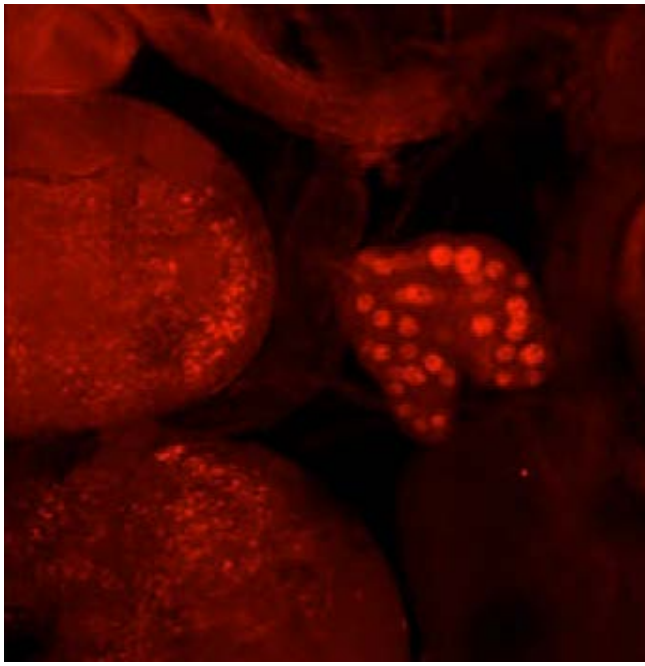

Fig. 1: *The image shows the* Drosophila melanogaster *ring gland. Part of this gland, the prothoracic gland, is responsible for the synthesis of the steroid hormone ecdysone, which controls animal growth and development. The nuclei of the prothoracic gland are marked (red).* © D. Martín

identified in *Drosophila*, controls cell and tissue growth. Ectopic expression of *bantam* induces cell overgrowth, while loss of function mutation leads to flies with severely reduced size. In this work, we show that, in addition to its well-characterized role as a cell-autonomous growth inducer, *bantam* also promotes systemic control of tissue and body growth by repressing ecdysone production during larval development. Furthermore, we show that the regulation of ecdysone synthesis by insulin relies on the repression of bantam activity in the prothoracic gland of the fly. In summary, this work has unveiled bantam as a key factor in the cross-talk between ecdysone and insulin in the control of final body size.

Reference Article

- Boulan, L., Martín, D., Milán, M. 2013. *bantam* miRNA Promotes Systemic Growth by Connecting Insulin Signaling and Ecdysone Production. *Current Biology* 23 (6): 473-478.

ON THE ORIGINS OF HIERARCHY IN COMPLEX NETWORKS

Hierarchy seems to pervade complexity in both living and artificial systems. The examples span from social interactions, to cell function, development, ecosystems, brain organization, and macroevolution. Fifty years ago, Herbert Simon defined complex systems as nested hierarchical networks of components organized as interconnected modules, but hierarchy is still a polysemous work and despite its relevance, no general theory that captures all features of hierarchy and its origins has yet been proposed. As a consequence of that and in particular from an evolutionary perspective, some questions remain unanswered: is hierarchy a widespread feature of living and non-living organization? What types of hierarchies do exist? Are hierarchies the result of selection pressures or, conversely, do they arise as a by-product of structural constraints?

To explore this issue, we departed from the intuitive idea of hierarchy as a perfect feedforward tree. That view coincides with the idea of a structure of control or relation with a pyramidal organization for which the ambiguity in who orders whom is not allowed. Figure 1 shows a collection of possible hierarchies according to our work. By means of the definition of three descriptors: Orderability, Feedforwardness, and Treenesss that basically accounted for (1) how cyclic is a network, (2) where those cycles are located in the structure and (3) how pyramidal is the structure, we obtained a formalism to quantitatively dissect the deviations of every system represented by a directed network from the ideal structure of hierarchy. As shown in figure 2a the construction of the hierarchical morphospace showed the space of possible organizations from perfectly hierarchical structures represented by an ideal tree to a full cyclic network. After a careful analysis of the morphospace by the examination of different random null models shown in Figure 2 b-c, Figure 2d-e shows the characterization of an asset of 125 networks encompassing eleven natural and artificial systems. The study revealed four major groups of hierarchical systems. Two of them matched the expected from random networks with similar connectivity, suggesting that non-adaptive factors were at work in the conformation of their organization. By contrast, ecological and gene regulatory networks defined the other two, indicating that their topological order may be by the result of functional constraints. These results were consistent with an exploration of the morphospace using *in silico* evolved networks, indicating

that some regions or the morphospace were non trivially accessible. This work implies a formal approximation for the study of hierarchy in complex systems where evolutionary constraints can be evaluated.

Future work in the development of generative models for the study of the emergence of hierarchy from an evolutionary perspective will be of strong interest in the study of dynamics in the exploration of the limits of what is possible for natural, technological, and social hierarchical organizations.

Reference Article

- Corominas-Murtra, B., Goñi, J., Sole, R.V., Rodriguez-Caso, C. 2013. On the origins of hierarchy in complex networks. *Proceedings of the National Academy of Sciences USA* 110 (33): 13316-13321.

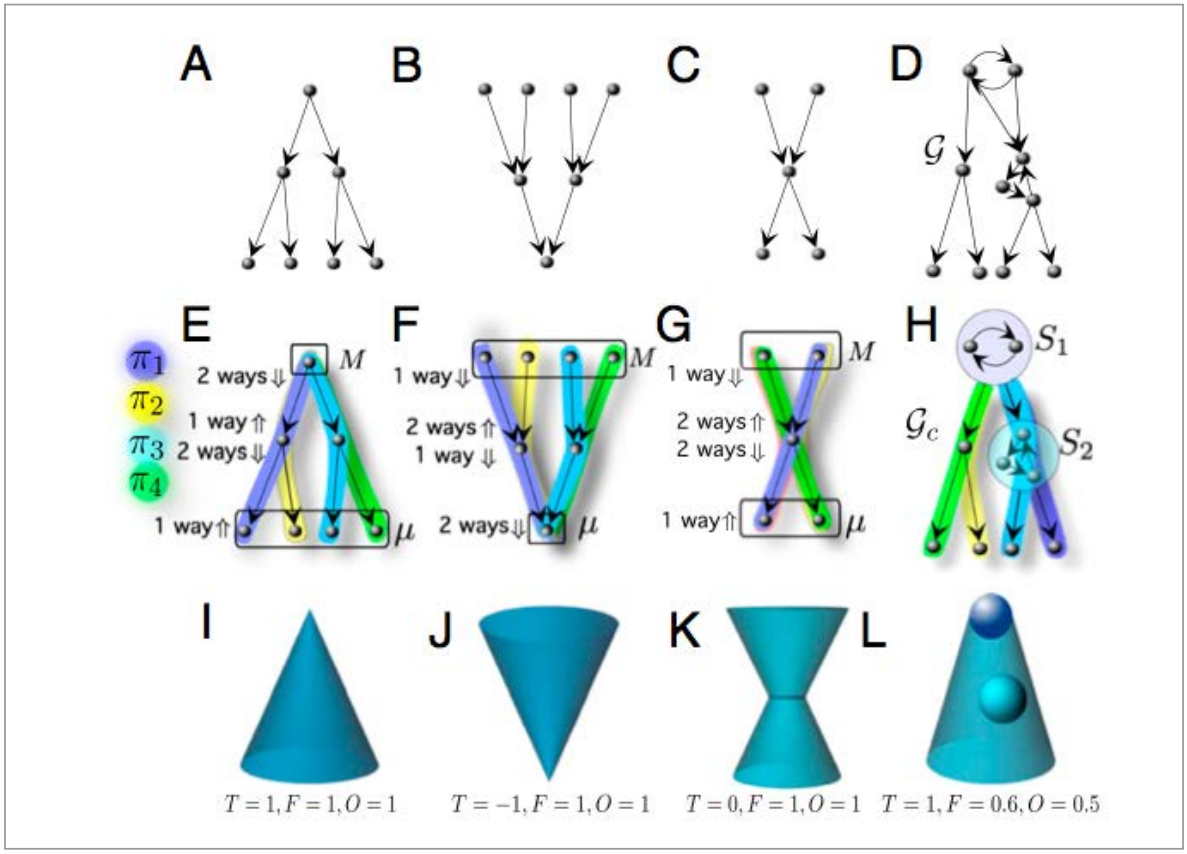

Fig. 1: *Different network organizations following the idea of hierarchy.* (A) *The ideal hierarchy seen as a tree-like feedforward graph.* (B) *An inverted tree matching the antihierarchica l. (C and* D) *A nonhierarchical fan-like feedforward graph* (C) *and a graph G displaying cycles* (D). *In* E *and* F, *all of them present tour pathways ($\pi_1, \pi_2, \pi_3, \pi_4$) from maximals* M *(top nodes) to minimals μ (bottom nodes). In* E, *downstream diversity of paths is $H_f(G)$ =log 4 without uncertainty when reversing them; i.e., $H_b(G)$ =0.* F *depicts the opposite behavior: $H_f(G)$ = 0 and $H_b(G)$ = log 4.* (G) *A nonhierarchical feedforward structure with the same forward and backward uncertainties, where $H_f(G)$ = H(G)= log 2. The node-weighted condensed graph, G_c, was computed by SCC detection labeled by every S_i.* (H) *For S_1, and S_2, the node weights are $\propto_{1=2}$ and $\propto_{2=3}$, respectively.* (I-L) *With a representative icon involving cones and balls, charts show representative* TFO *values for* (A-D) *graphs.*

THESES, COURSES AND SEMINARS

DOCTORAL THESES PRESENTED DURING 2013

PhD Student: Javier Igea de Castro
Title: Desarrollo de nuevos marcadores genómicos y su aplicación a la filogenia y variabilidad genética de mamíferos
Thesis Director: José Castresana Villamor
Institution & Date: Universitat de Barcelona, 18 th January 2013

PhD Student: Mafalda Cotrim Roberto Barata
Title: High altitude phylogeography of selected Moroccan herpetofauna
Thesis Director: Salvador Carranza
Institution & Date: Universidade do Porto,12th March 2013

PhD Student: Arnau Sebé Pedrós
Title: The origin of metazoan multicellularity, a genomic and functional approach
Thesis Director: Iñaki Ruiz-Trillo/Jaume Baguñà
Institution & Date: Universitat de Barcelona, 07th June 2013

PhD Student: Marc García Garcerá
Title: Methodological preparation and characterization of the microbial ecology of the skin
Thesis Director: Francesc Calafell
Institution & Date: Universitat Pompeu Fabra, 21 th June 2013

PhD Student: Marc Abrisqueta Carol
Title: El receptor d'insulina com a element clau en la senyalització nutricional a la panerola Blattella germanica (L.) (Dictyoptera, Blatellidae)
Thesis Director: José Luis Maestro
Institution & Date: Universitat de Barcelona, 27th September

PhD Student: Ana Rodríguez Prieto
Title: Filogenía, morfometría y especiación de dos topillos ibéricos: Microtus duodecimcostatus y *Microtus lusitanicus*
Thesis Director: José Castresana
Institution & Date: Universitat de Barcelona, 4th October 2013

PhD Student: Elena Carnero
Title: Genomic and Functional approaches to genetic Adaptation
Thesis Director: Elena Bosch
Institution & Date: Universitat Pompeu Fabra, 25th October 2013

PhD Student: Giovanni Marco Dall'Olio
Title: Applications of network theory to human population genetics: from pathways to genotype networks
Thesis Director: Jaume Bertranpetit
Institution & Date: Universitat Pompeu Fabra, November 2013

PhD Student: Brandon Invergo
Title: A system-level, molecular-evolutionary analysis of mammalian phototransduction
Thesis Director: Jaume Bertranpetit/Ludovica Montanucci
Institution & Date: Universitat Pompeu Fabra, 22nd November 2013

PhD Student: Marc Haber
Title: Study of human genetic diversity. Inferences on population origin and history
Thesis Director: David Comas
Institution & Date: Universitat Pompeu Fabra, 5th December 2013

PhD Student: Enric Ureña
Title: Regulació de la metamorfosi en insects hemimetàbols I holometàbols. Caracterització functional del gen E93 i del process de sumoilació
Thesis Director: David Martín
Institution & Date: Universitat de Barcelona, 5th December 2013

Speaker	Talk	Institution	Date
Aurora Ruiz-Herrera	"Genomic shuffling and recombination: insights into mammalian evolution"	Departament de Biologia Celular, Fisiologia e Inmunologia, Universitat Autònoma de Barcelona, Spain.	5/02/2013
Joao Zilhao	"Neandertals and early modern humans: an overview of recent developments"	Department of Archaeology and Anthropology University of Bristol, UK.	12/03/2013
José Manuel Cano Arias	"Thermal and time-constraint adaptations in amphibians: an integrative approach from genes to populations"	Research Unit of Biodiversity (UO-CSIC-PA), Oviedo, Spain.	26/03/2013
Christina Hvilsom	"Extensive X-linked adaptive evolution in central chimpanzees and genetic charaterization of captive population"	Copenhagen Zoo, DK 2000 Frederiksberg, Denmark.	02/04/2013
Alfred Cortes	Adaptation of malaria parasites to changes in their environment: playing dice to survive	CRESIB, Hospital Clinic, Barcelona, Spain.	10/04/2013
Toby Pennington	Using phylogenies and molecular clocks to understand the evolution of tropical plant diversity	Tropical Diversity Section of the Royal Botanic Garden of Edinburgh, UK.	30/04/2013
Joshua Weitz	Death or dinner? Phage-bacteria infections and consequences at the ecosystem scale	School of Biology and School of Physics Georgia Institute of Technology Atlanta, GA, US.	07/05/2013
Francisco M. De La Vega	Scalable variant identification across multigenerational pedigrees and population samples from high-throughput sequencing data	Dept. of Genetics, Stanford School of Medicine, Stanford, CA, US.	17/05/2013

Speaker	Talk	Institution	Date
David Reich	"Genetic evidence for interbreeding between archaic and modern humans"	Department of genetics of the Harvard medical School, US.	27/05/2013 (*)
Andrés Moreno	Human population genomics in the Americas: new approaches to address long-standing evolutionary riddles	Dept. of Genetics, Stanford School of Medicine, Stanford, CA, US.	26/06/2013
Xesús Abalo	Transducin paralogs in the retina and the pineal complex: different specialisations after the teleost tetraploidisation	Uppsala biomedicinska centrum, Uppsala University, Sweeden.	09/07/2013
Antonio Lazcano	"When the world was made of RNA"	Evolutionary Biology Lab, Facultad de Ciencias, Universidad Nacional Autónoma de México, México.	30/09/2013 (*)
Josephine Daub	"Detection of polygenic selection at different evolutionary time scales"	Population Genetics CMPG lab, Universität Bern, Switzerland.	29/10/2013
George Perry	"Conservation and extinction genomics of Madagascar's lemurs: Anthropogenic effects"	Department of Biology, Eberly College of Science, Pennsylvania State University. University Park, US.	11/11/2013
Lucia Carbone	"Insight on the accelerated chromosome evolution of small apes from the gibbon genome project"	Oregon Health & Sciences University, US.	17/12/2013

(*) This seminar has been part of the "PRBB-CRG Sessions" (sponsored by PRBB and CRG)


eXplorist
Z10
MAGELLAN
BOURAIL
FARINO
MOINDOU
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BOULOUPARI
PAITA
DUMBÉA
NOUMÉA
LE MONT-DORE

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 (English) (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

- Master in Biomedical Research (BIOMED) (Universitat Pompeu Fabra)
  - Genomes and Systems (5 ECTS).
    Coordinator: Tomàs Marquès-Bonet.
  - Introduction to Biomedicine (5 ECTS).
    Coordinator: David Comas.
- Master in Bioinformatics for Health Sciences (BIOINFO). Joint master of the Universitat Pompeu Fabra (coordination) and Universitat de Barcelona, in cooperation with the Università di Bologna.
  - Analisis of Biomedical Data (5 ECTS).
    Coordinator: Arcadi Navarro.
  - Biomedical Informatics (5 ECTS).
    Coordinator: Arcadi Navarro.
  - Introduction to Biomedicine (5 ECTS).
    Coordinator: David Comas.

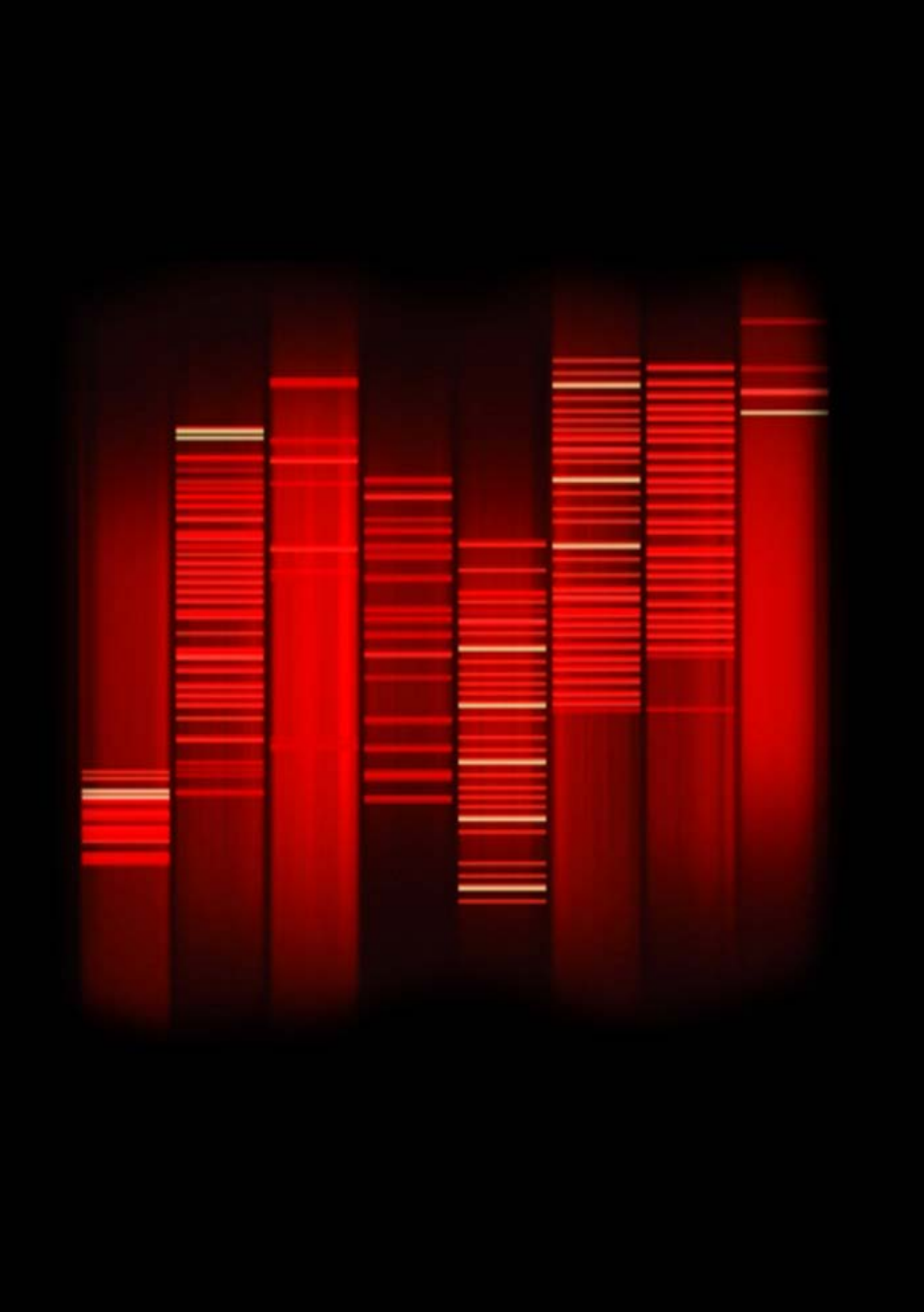
Furthermore, most IBE scientists actively participate in several international master programs and specialized courses in different universities:

- Master: Human Biology; Universitat de Barcelona (UB) / Universitat Autònoma de Barcelona (UAB).
  Teacher: Francesc Calafell
- Master: Genetic Counselling; IDEC / UPF.
  Teacher: Francesc Calafell
- Master: Biodiversity; Universitat de Barcelona (UB).
  Teachers: J. González, S. Carranza, I. Ruiz-Trillo, J. Goméz-Zurita
- Postgraduate Course: Filogenias y Genealogías de DNA: Reconstrucción y Aplicaciones; Universitat de Barcelona (UB).
  Teachers: J. Castresana, S. Carranza, I. Ruiz-Trillo
- Master: Anàlisi de Dades Òmiques / Omics Data Analysis; UVic.
  Teacher: Francesc Calafell

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

Along 2013 IBE has hosted a total of 35 students. In particular:

- 8 High school seniors and juniors from "Programa Joves i Ciència" (5)- financed by *Fundació La Caixa-la Pedrera*-, Col.legi Pare Manyanet (1), Escola Pía de Terrassa (1), Institut La Guineueta (1)
- 20 undergraduate students (practicums) from: Universitat Pompeu Fabra (10), Universitat de Barcelona (7), Universitat Autònoma de Barcelona (2), Universitat de Vic (1)
- 5 master students from: Universitat Pompeu Fabra (2), IDEC (2), Universitat de Barcelona (1)



DISSEMINATION OF SCIENCE

TRAINING AND OUTREACH UNIT (TAO)

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

IBE PhD training course, 21st-29th January 2013

Following a suggestion of the IBE External Scientific Committee, and with the aim of providing training to new PhD students in Evolutionary Biology, the Training and Outreach unit designed a course to be implemented regularly. The goal was to provide an overview of the basic concepts in the diverse Evolutionary Biology fields to new PhD students (who necessarily have different backgrounds) when enrolling the IBE.

On the first edition of this "PhD training course" main lectures and seminars were carried out by IBE PIs. Postdocs volunteered to lead hands-on sessions and paper discussions.

The topics included:

- A brief history of Evolutionary Biology
- Microevolution and the basis of population genetics
- Macroevolution and diversification processes
- Coalescence, dating trees, and molecular clocks
- Basic knowledge of techniques and models used in Evolutionary Biology: next generation sequencing, measures of gene expression, model organisms
- Comparative genomics, genome evolution, and variation
- Phylogenetics: variation in time and space
- Genome regulation, expression, and correlation between genotypes and phenotypes
- Genetic algorithms, artificial and synthetic systems, and complex networks
- Hands-on sessions on genome browsers, population genetics software, phylogenetic resources

The course was compulsory for 1st and 2nd year PhD students and included a final evaluation test. The audience of the sessions also included senior PhD students, postdocs, and PIs.

Furthermore, in the context of PRBB centres, IBE members have access to the Intervals Programme, an interdisciplinary education programme for professionals working in the Barcelona Biomedical Research Park (PRBB). The activities of the Intervals programme currently focus on:

A: Leadership, Management, and Career Development
B: Communication
C: Biomedicine, Society & Good Scientific Practice

The most popular intervals courses in 2013 were "Leading for success in science" (PIs and postdocs edition), and "Write it clearly: fundamentals of good scientific writing".

OUTREACH ACTIVITIES

The Institute of Evolutionary Biology is committed to informing and educating the general public about the research being carried out at the Institute. During 2013, IBE organized and participated in several outreach activities.

La Ciència Al Teu Món (Science is part of your world)
February 2013. Organized by: IBE.
Several PIs and students at the IBE are collaborating on *La Ciència Al Teu Món (LCATM)* outreach project lead by Josefa González (*Ramón y Cajal* Researcher at IBE). 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 towards problems.

In February 2013, a short-video, photography, and illustration contest aimed at secondary school students was launched. Students were asked to portray their dreams and interests showing how science could help to achieve them. The winners were awarded with Science Trips, tablets, 3D cameras, and outdoor experiences, thanks to the collaboration of the *Institut de Ciències del Mar* (CSIC) and several private companies.

LCATM is funded by the *Secretaria d'Universitats i Recerca, Departament d'Economia i Coneixement, Generalitat de Catalunya*, and the *Fundación*

Española para la Ciencia Y la Tecnologia (FECYT), and can be found on the web www.lacienciaalteumon.cat, on twitter, *@ LCATMon*, and on Facebook, www.facebook.com/LCATMon.

Saló de l'Ensenyament (teaching fair)
13th to 17th March 2013.
Organized by: Universitat Pompeu Fabra.
IBE participated in the UPF stand at the *Saló de l'Ensenyament* with the activity *"Deixa el ramat i menja't el món"* (Leave the herd and conquer the world) that encouraged students to share with us their professional ambitions, and offered LCATM outreach platform as a virtual place to interact with scientists.

Programa Professors i Ciència (Science and Professors programme)
30th April and 3rd, 8th and 10th May 2013.
Organized by: Fundació Catalunya-La Pedrera.
IBE participated in the Professores i Ciència program by teaching a course on the genetic basis of environmental adaptation. We also created an interactive material that communicates in a simple and effective way the concept of adaptation by natural selection. This material is available at the platform's website (www.lacienciaalteumon.cat).

La Festa de la Ciència i la Tecnologia (Barcelona Science Fair)
15th and 16th June 2013.
Organized by: Ajuntament de Barcelona.
LCATM designed an activity for this event in which by actively participating in a game young people could understand the importance of biodiversity.

Mostra d'iniciatives per la Biodiversitat (Biodiversity activities fair)

7th June 2013. Organized by: La Fabrica del Sol, Ajuntament de Barcelona.

IBE participated in this fair with the PechaKucha presentation: How organisms adapt to their environment.

Campus Gutenberg

16th and 17th September 2013. Organized by: Observatori de Comunicació Científica-Universitat Pompeu Fabra.

The LCATM team participated in this two-day Campus held at the Universitat Pompeu Fabra by sharing the different educational tools offered by the platform including web, video, video game, social networks, and interactive materials.

PRBB Open Day

October 5th, 2013. Organized by: Parc de Recerca Biomèdica de Barcelona (PRBB)

Beside presenting the LCATM platform to the PRBB visitors, IBE also organized an activity to promote the conferences *R(e)volució: conquereix amb nosaltres les noves fronteres del coneixement* (R(e)volution: conquer with us the new frontiers of knowledge). Additionally, several IBE researchers participated as volunteers and also gave scientific talks aimed at the general public.

(Re)volució: Conquereix amb nosaltres les noves fronteres del coneixement (R(e)volution: Conquer with us the new frontiers of knowledge")

October 5th to November 12th 2013. Organized by: IBE.

Researchers at the Institute of Evolutionary Biology discussed diverse topics such as predicting the behavior of biological systems, butterfly diversity and the information they give us about climate change, the origin of multi-cellularity, the role of genes in our decisions, and the past of the human species through the genome. (Re)volució conferences, an original idea of Ricard Solé, was a unique opportunity for the public at large because, in addition to gaining knowledge on frontier research in biology, they could interact with the scientists that are leading it.

Setmana de la Ciència 2013 (Science Week Activities)

19th and 20th November 2013.

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

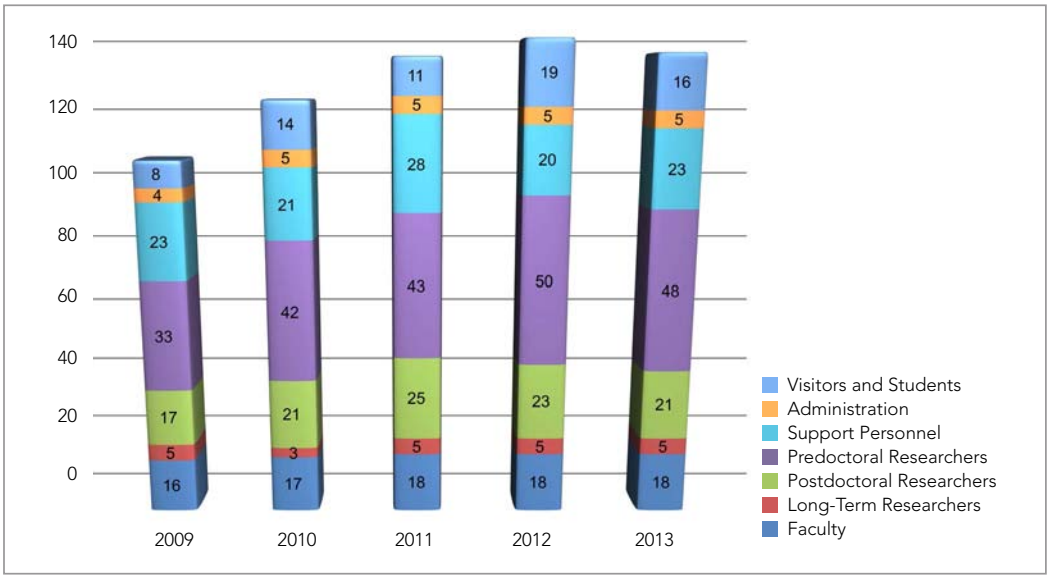
IBE participated in the science week activities by offering both schools and the general public two hands-on activities based on two different research projects that are currently being pursued at IBE.

Folding tissues: crafting a wing. This hand-on activity aimed at showing how a particular body organ is built from an already present structure, using the handbook written in the genome. For this purpose, we used as a model *Drosophila melanogaster* and the process of building an entire adult fly and all its structures, as for example the wings, from the imaginal discs during the pupa (pre-adult) stage.

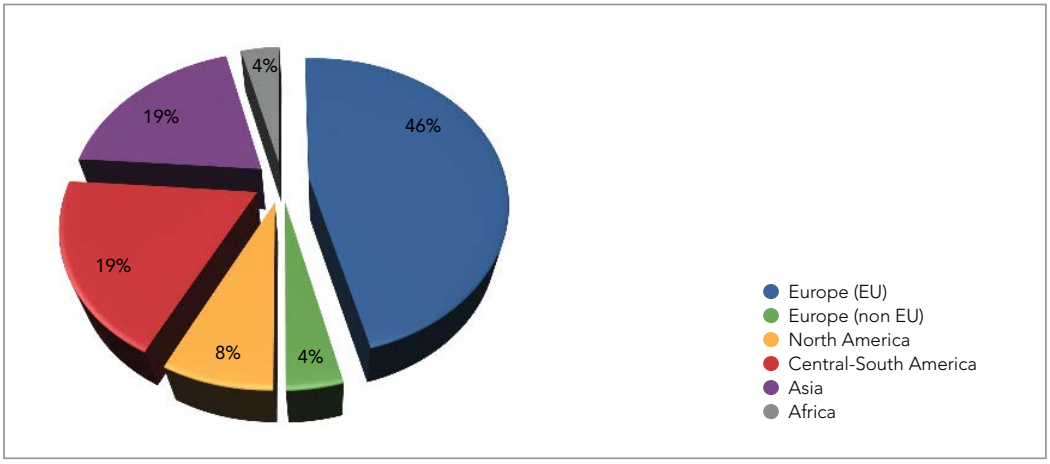
How organisms adapt to their environment. An interactive game allowed students and the general public to learn through playing the importance of biodiversity for the survival of the species.

PEOPLE

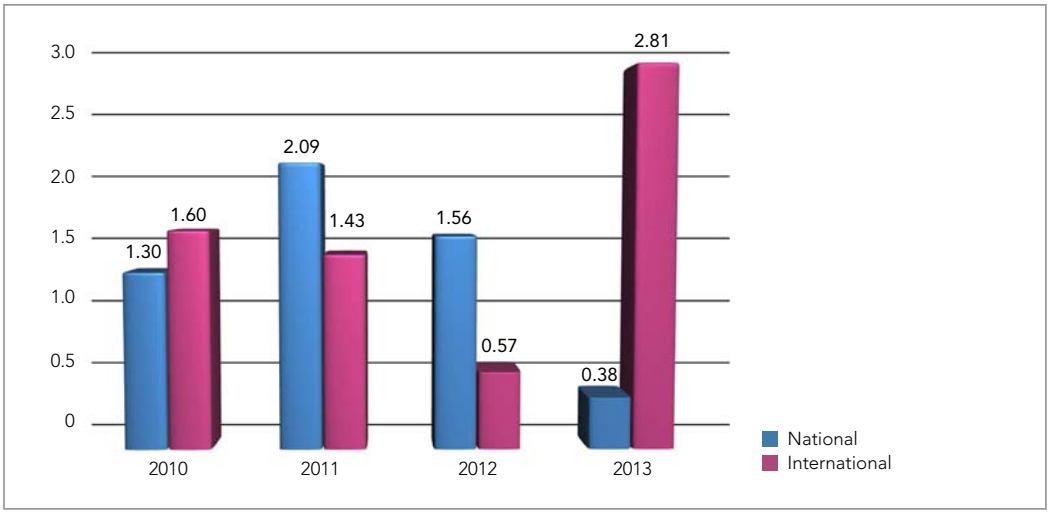
Evolution of personnel distribution per categories



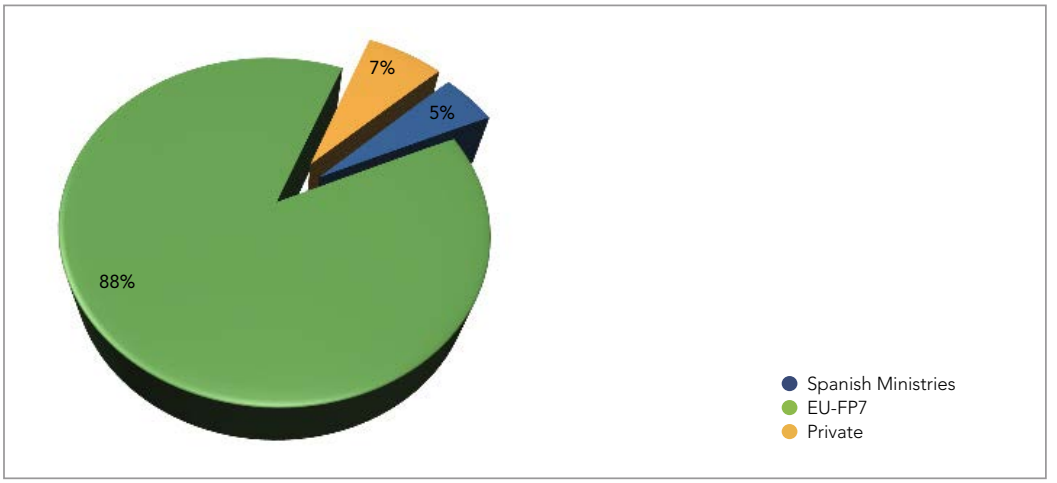
Internationalisation. Foreign personnel represents 22 % of the total of IBE members (visitors not included). A part from Spain, we have researchers from 17 different countries from Europe, America, Asia and Africa.



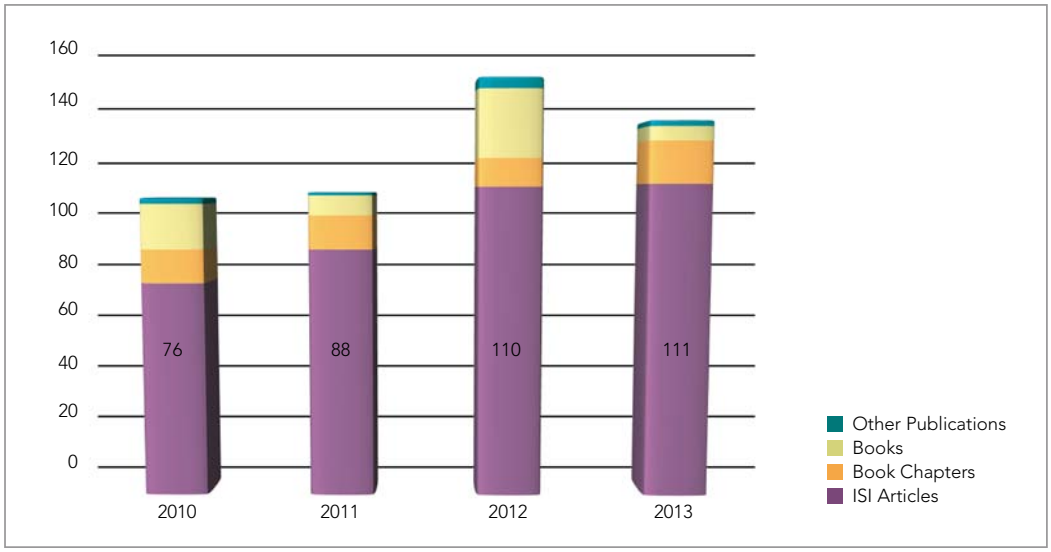
Competitive new funds raised (in M€)



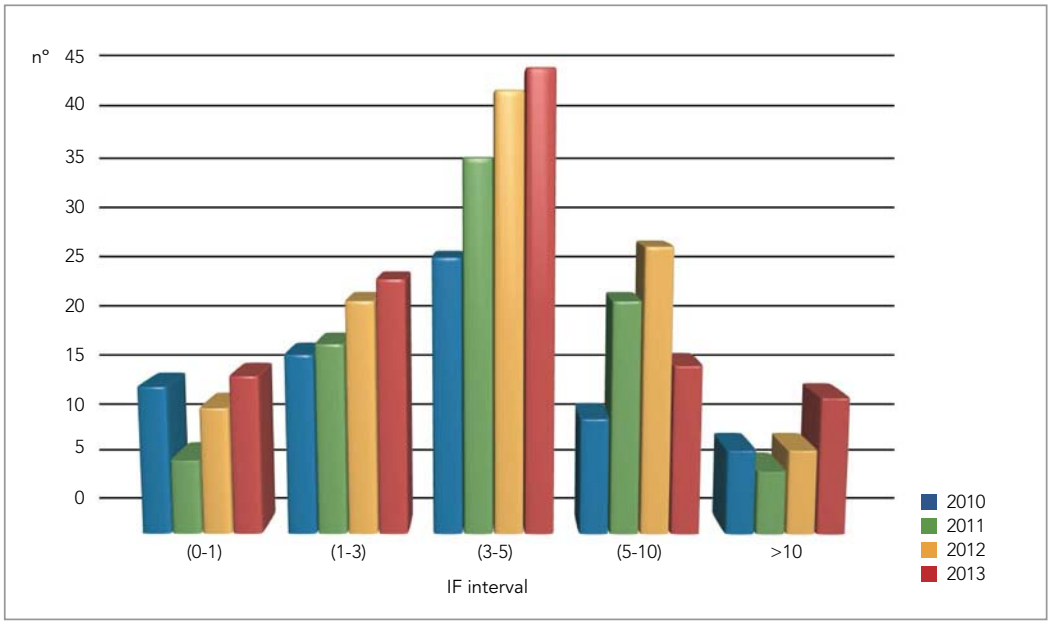
Origin per agencies of 2013 competitive funds raised



Evolution of publications distribution per kind of publication



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

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