

5<sup>th</sup> Congress  
ITALIAN SOCIETY FOR  
EVOLUTIONARY BIOLOGY  
Trento, 28-31 August 2013

**PROGRAM,  
BOOK OF ABSTRACTS,  
AUTHOR INDEX.**

**EDITED BY LINO OMETTO AND OMAR ROTA-STABELLI**



V congress of the Italian Society for Evolutionary Biology. Trento, 28-31 August 2013

# EVOLUTIONARY GENOMICS AND BIOINFORMATICS

The symposium will feature contributions dealing with any aspect evolutionary genetics and genomics in any living organism, including viruses. Descriptions of adaptive events at the genetic level, new methodological advances in the field, analysis of complete genomes or gene sets from an evolutionary perspective are invited. Bioinformatics approaches to analyze large-scale genetic data for selection signature inference will also be welcome. The aim of the symposium is providing insight into the manifold pathways to genetic adaptation and into the methodological advances to study the evolutionary history of genes and genomes.

**Chairs: Manuela Sironi (IRCCS E. MEDEA) and Dan Sargent (Fondazione Edmund Mach)**

## Reconstructing human genetic diversity in space and time

**Francois Balloux<sup>1</sup>**

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The worldwide distribution of human genetic variation is characterized by smooth clines of isolation by distance and a decrease of genetic diversity with increasing geographic distance from Sub-Saharan Africa along likely colonization routes. While these striking patterns strongly suggest a recent out-of-Africa expansion for Anatomically Modern Humans (AMHs), they shed little light on the more subtle aspects of the past demography of our species. Thus, we developed a spatially explicit model of the expansion of AMHs and using climate reconstructions over the past 120k years to quantify the possible effects of climate on human demography. The combinations of demographic parameters compatible with the current genetic makeup of worldwide populations indicate a clear effect of climate on past population densities. Our estimates of this effect, based on population genetics, capture well the observed relationship between current climate and population density in modern hunter-gatherers worldwide, providing supporting evidence for the realism of our approach. Furthermore, while we did not use any archaeological and anthropological data to inform the model, the arrival times in different continents predicted by our model are also broadly consistent with the fossil and archaeological record. Our framework arguably provides the most accurate spatiotemporal reconstruction of human demographic history but still offers considerable scope for improvement. In the last part of the presentation, I will present unpublished results including a reconstruction of the colonisation of the Americas based on whole modern and ancient genomes. Finally I will discuss the potential and limitations of explicit spatiotemporal models for inferring past demography.

## Evolution and genome divergence determine virulence traits in *Candida* spp.

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*Candida albicans* and *Candida parapsilosis* are known as the most causative agents of human fungal infections, but in healthy conditions they colonize the human gastrointestinal tract as commensals. While the pathogenicity of *C. albicans* is associated to the phenotype switch -with conversion from white to opaque colony and from cell to hyphae- affecting a variety of virulence-related genetic factors, for *C. parapsilosis* the genetic makeup associated to the virulence is still unknown. In order to investigate the intra and inter-species variability involved in the phenotypical changes we analyze the whole genome sequences of 7 candida strains (3 of *C. albicans* and 4 of *C. parapsilosis*) isolated from fecal samples of healthy and Crohn's disease patients, a severe inflammatory bowel disease. The results show two different population

structures: while *C. albicans* present an extreme variability and divergence between strains, *C. parapsilosi* seems to be more clonal, with very low number of polymorphisms. Notably, in *C. albicans* intriguing genomic plasticity have been observed. Indeed, over the high polymorphism, strain-specific gene losses, acquisition, and several miss-sense genes were found. Moreover, in *C. albicans* isolates, the most polymorphic genes codify proteins related to the cell wall and hyphal formation, external encapsulation structure and cell periphery, suggesting a continuous adaptation to adverse environments or stress conditions. Our data are confirmed by phenotypical characterization that show changes in virulence related traits (invasive growth, hyphal formation and switch from smooth to myceliated colonies) for the majority of *C. albicans* strains. Also in two *C. parapsilosis* strains similar pathogenic traits were found. Interestingly, the cytokine milieu released by human PBMCs and DCs showed that specific inflammatory responses are produced by strains with the most virulent phenotype. Overall these results provide significant insights regarding the link between host adaptation, pathogenesis and evolution.

## “Bad” species? Restriction sites associated DNA sheds light on the evolution of the *Erebia tyndarus* species complex

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The *Erebia tyndarus* species complex is a cluster of closely related alpine butterflies, representing an intriguing riddle for taxonomists, evolutionary biologists and biogeographers and a still underexploited model for understanding the role of neutral and adaptive processes in speciation. Members of the ‘tyndarus’ group have been characterized so far by morphological and ecological characters, cross-breeding experiments, karyology, and molecular data from allozymes and mitochondrial DNA (mtDNA). However, the delimitation of species, their relationships, as well as the origin of their peculiar geographic distribution, are still unclear. So much so that these taxa were cited as an example of ‘bad species’, for which the establishment of a phylogenetically ‘correct’ taxonomy could represent a desperate and almost meaningless endeavour (Descimon & Mallet, 2009). Up to five different species belonging to the ‘tyndarus’ group have been described in the Alps: *E. tyndarus*, *E. calcaria*, *E. nivalis*, *E. cassioides* and *E. carmenta* (with the latter often considered a subspecies of *E. cassioides*) all of them belonging to a unique “Alpine” clade (Albre et al., 2008). Figure 1 illustrates the distribution range of these taxa. Three species (*E. tyndarus*, *E. nivalis*, *E. calcaria*) are endemic to the Alps, while *E. cassioides* stretches its patchy distribution from Eastern Alps into the Balkans and *E. carmenta* from Western Alps into Apennines and Pyrenees. This distribution pattern includes instances of allopatry, parapatry and quasi-sympatry; the latter case represented by *E. nivalis* narrowly overlapping along an altitudinal gradient with *E. cassioides* or (in a single site) *E. tyndarus*. In this ongoing study, we are employing restriction sites associated DNA (RAD) along with more traditional mtDNA sequencing in order to clarify the systematic and genetic relationships among taxa of the *E. tyndarus* “Alpine” clade, as a basis for further evolutionary studies. Our preliminary analyses show that our RAD data are able to provide, for the first time, a clear molecular support for the recognition of four well-defined genetic units, corresponding to the species *E. tyndarus*, *E. nivalis*, *E. calcaria* and *E. cassioides+carmenta* (Figure 1c). Interestingly, these lineages do not show any synapomorphies at 1200 bp of mtDNA (Figure 1b). In striking contrast, the distribution of mtDNA haplotypes highlights a strong geographical structure within *E. cassioides+carmenta*, where RAD shows a rather continuous genetic gradient from Pyrenees to Alps and Apennines, highlighting the historical role of nuclear gene-flow and recombination in maintaining the genetic cohesion of a single evolutionary unit. Finally, RAD data are providing provisional evidence for gene flow between *E. tyndarus* and *E. nivalis* (whose range overlap is very limited), while hybridization between *E. cassioides* and *E. nivalis* (occurring in quasi-syntopy at several sites) seems to lack altogether, which suggests an intriguing correlation between reproductive isolation and ability to coexist by niche-splitting.

## A framework for the evolutionary study of uncharacterised microbial communities

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Complex microbial environments such as those analysed in metagenomic studies present a challenge in how these communities change and adapt to external stimuli. One such environment is present in the rumen of herbivores, which use