Evoluzione
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SIBE

DIPARTIMENTO
SCIENZE BIOLOGICHE,
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BIGEA

VI congress of the Italian Society for Evolutionary Biology - **Bologna August 31 - September 3, 2015**

Bologna, August 31st – September 3rd 2015

Evoluzione 2015

6° Congress of the Italian Society of
Evolutionary Biology
SIBE-ISEB

Conference program

Monday, August 31st

14:00-14:20	*Conference opening and welcome*
Symposium 1	**Evolution of Genomes and Genomic Conflicts** *Chair: Marco Passamonti*
14:20 – 15:00 *Invited speaker*	**William F. Martin.** Endosymbiotic origin and differential loss of eukaryotic genes.
15:00 – 15:20	**Michele Castelli.** Insights on the evolution of the bacterial endosymbiont *Holospora caryophila* from the genome sequence.
15:20 – 15:40	**Rupinder Kaur.** Investigating the genotypic and phenotypic basis of continental specific *Drosophila-Wolbachia* endosymbiotic system.
15:40 – 16:00	**Fabrizio Ghiselli.** The Doubly Uniparental Inheritance: a model system for studying evolutionary and functional genomics of mitochondria.
16:00 – 16:20	**Liliana Milani.** Mitochondrial activity in gametes and transmission of viable mtDNA.
16:20 – 16:40	**Saverio Vicario.** Evidence for purifying selection of mtDNA mutations during human oogenesis.
16:40 – 17:00	**Federico Plazzi.** 100 of these genomes: Bivalve mitogenomics hits triple digits.
	Coffee break
17:20 – 17:40	**Andrea Luchetti.** Recombine and survive: evolutionary history of the V highly conserved domain in the mammalian genome after the V-SINE superfamily extinction.
17:40 – 18:00	**Livia Bonandin.** Reproductive biology versus transposable elements load: the role of host reproductive strategy in the study of R2 dynamics in *Bacillus* stick insects (Phasmida, Bacillidae).
18:00 – 18:20	**Chiara Pontremoli.** Adaptive evolution underlies the species-specific binding of *P. falciparum* RH5 to human basigin.
18:20 – 18:40	**Emiliano Trucchi.** Epigenetic divergence and parallel adaptation in *Heliosperma pusillum* (Caryophylaceae).
18:40 – 19:00	**Francesca Dumas.** Interstitial telomeric sequences distribution in eight Neotropical primates (Platyrrhinae) by (TTAGGG)n probe mapping: interstitial location and their possible role in evolution

Tuesday, September 1st

Symposium 2	**Evolutionary Research in Museums.** *Chair: Franco Andreone*
08:40 – 09:20 *Invited speaker*	**Angelica Crottini, Franco Andreone.** When traditional museological tools meet new cutting-edge applications: our experience with the study of species diversification patterns, taxonomy and conservation of Madagascar herpetofauna.
09:20 – 09:40	**Leonardo Latella.** The tale of specimen: what and when natural history collections can tell us about evolution.
09:40 – 10:00	**Elena Canadelli.** The "Extinction" project: valorisation of museum natural history collections as a tool to link research, outreach and conservation.
10:00 – 10:20	**Giovanni Forcina.** Unravelling the biogeography of secretive taxa by museum collections: the untold story of the black francolin (*Francolinus francolinus*, Phasianidae) in the Mediterranean.
	Coffee break
10:40 – 11:00	**Agostino Leone.** Restoring the history of sawfish in the Mediterranean from museum exhibits.
11:00 – 11:20	**Francesco Massari.** Unlocking the evolutionary history of the mighty bluefin tuna using novel paleogenetic techniques and ancient tuna remains.
Symposium 3	**Phylogenetics, Biogeography and Phylogeography** *Chairs: Gabriele Gentile, Saverio Vicario*
11:20 – 12:00 *Invited speaker*	**Alexandre Antonelli.** The evolution of neotropical mega-diversity.
12:00 – 12:20	**Laura Parducci.** Inferring past tree distribution in Nordic environments from ancient DNA.
12:20 – 12:40	**Daniela Lucente.** The role of local refugia and paleoclimatic history in the phylogeography of a cold-tolerant species: the European cave salamander *Hydromantes strinatii*
12:40 – 13:00	**Fausto Tinti.** Molecular markers, natural history and evolution in sharks and rays.
	Lunch
14:40 – 15:00	**Giulia Fassio.** Unexpected biodiversity of epizoic gastropod genus *Marseniopsis* (Velutinidae) within a phylogenetic framework for the family.
15:00 – 15:20	**Mukesh Thakur.** Problem bear translocation: is it effective management practice to mitigate bear-human conflicts in Dachigam landscape, Jammu and Kashmir, India?
15:20 – 15:40	**Matteo Vecchi.** A backboned phylogeny for invertebrates: the case and problems of eutardigrades (Tardigrada)
15:40 – 16:00	**Massimo Bernardi.** Species between conservation and evolution.

Symposium 4 **Evolution of phenotypes and Organismal Biology**

Chairs: Omar Rota Stabelli, Lino Ometto

16:20 – 17:00 *Invited speaker*	**Benjamin Proud'Homme.** Regulatory evolution and the diversification of pigmentation patterns in *Drosophila*.
17:00 – 17:20	**Silvia Paolucci.** Maternal effect on offspring size in seed harvester ants.
17:20 – 17:40	**Federica Costantini.** Genetic connectivity along depth gradient and implications for phenotypic plasticity in a gorgonian species.
17:40 – 18:00	**Ilaria Giovannini.** The phenotypic plasticity in life history traits of an Antarctic water bear as a strategy to cope with extreme environmental conditions.
18:00 – 18:20	**Sukanya Ramasamy.** The role of olfaction in the making of a pest - a genomic analysis.

19:30 – 22:00	*The Evolution of the Aperitif. Zoology Museum, via Selmi 3.*

Wednesday, September 2nd

Symposium 5 **Population Genetics and Genomics**

Chairs: Giorgio Bertorelle, Paolo Ajmone Marsan

09:00 – 09:40 *Invited speaker*	**Konrad Lohse.** Inferring population histories from genomes.
09:40 – 10:00	**Carlo Pecoraro.** The application of 2b-RAD approach for investigating the genomic population structure of yellowfin tuna (*Thunnus albacares*) at the global scale.
10:00 – 10:20	**Pierpaolo Maisano Delser.** Population genomics of *C. melanopterus* using target gene capture data: demographic inferences and conservation perspectives.
10:20 – 10:40	**Luca Calderoni.** Regressive evolution in Somalian cavefish *Phreatichthys andruzzii*: loss of selective constraint on opsin genes.
	Coffee break
11:00 – 11:20	**Livia Di Giambattista**. Population genetics, ecology, and evolution of the troglophilic isopod *Androniscus dentiger* from the Frasassi cave system.
11:20 – 11:40	**Stefano Mona.** On the role played by the carrying capacity and the ancestral population size during a range expansion.
11:40 – 12:00	**Mukesh Thakur.** Hearing hangul calls for survival: conservation genetics and demographic history of the last surviving population of red deer in India.
12:00 – 12:20	**Sara Knight.** Quantifying separation and similarity in a *Saccharomyces cerevisiae* metapopulation.
12:20 – 12:40	**Giorgio Bertorelle**. The evolution of the small and isolated population of Apennine brown bears (*Ursus arctos marsicanus*): a whole-genomes perspective.

Lunch

Symposium 6 **Evolutionary Applications**
Chair: Giorgio Binelli

14:20 – 15:00 *Invited speaker* **Berthold Heinze.** Genetic analysis and real world problems in ecology and forestry.

15:00 – 15:20 **Vincent Piou.** Importance of the phoretic phase in the reproduction of *Varroa destructor*: ectoparasite of the honey bee (*Apis mellifera*).

15:20 – 15:40 **Omar Rota Stabelli.** Evolution and genomics quickly provide with useful clues for insect pest management: the case of *Drosophila suzukii*.

15:40 – 16:00 **Isabella Vanetti.** Taxonomy and genetic diversity of anodontine mussels in Lake Maggiore.

16:00 – 16:20 **Caterina Maria Antognazza.** Genetics survey of European barbel (*Barbus barbus*) populations in England and Wales: implication for future restocking management.

16:20 – 16:40 **Emanuele Fasola.** Frogs' microbiome from metal polluted and metal non-polluted sites.

16:40 – 17:00 **Anna Sandionigi.** Next-gen monitoring strategies. A warning system for EXPO2015.

17:00 – 17:20 **Giacinto Libertini.** Evolutionary classification of diseases and similar conditions.

SIBE Lecture

17:40 – 18:20 **Pietro Omodeo.** Every evolutionary step is an addition of information to an already existing system.

18:20 – 19:40 *SIBE Members Meeting*

21:00 – 24:00 *Social Dinner*

Thursday, September 3rd

Symposium 7 **New Frontiers in Evolutionary Anthropology***
Chair: Silvia Ghirotto
** Co-hosted with the Italian Anthropological Association*

09:30 – 10:30 *Invited speaker* **Eske Willerslev**. How we settled the world and developed our culture.

10:30 – 10:45 **Francesca Tassi.** Early modern human dispersal from Africa: genomic evidence for multiple waves of migration.

10:45 – 11:00 **Daniele Gibelli.** Identification of missing children: a quantitative approach to the

	analysis of facial morphology.
11:00 –11:15	**Elisabetta Cilli** Discovering past gut microbiomes through NGS analysis: the mummies of Roccapelago (MO).
11:15 – 11:30	*Discussion*
	Coffee break
11:45 – 12:00	**Cristina Giuliani.** DNA methylation variability along the Italian peninsula: an evolutionary perspective reveals implications for human health and diseases.
12:00 – 12:15	**Elena Fiorin.** The significance of microfossils recovered from dental calculus for anthropological analysis.
12:15 – 12:30	**Michela Leonardi.** Testing for the influence of lifestyle on genetic diversity through time.
12:30 – 12:45	*Discussion*

Posters

n.	*Author*	*Title*	*Symposium*
1.	Stefano Bettinazzi	The complete female- and male-transmitted mitochondrial genome of *Meretrix lamarckii* (Bivalvia: Veneridae)	*1*
2.	Davide Guerra	The tangled triangle among ORFans, mitochondrial inheritance, and sex determination: mtDNA evolution at the root of Paleoheterodonta (Bivalvia)	*1*
3.	Alessandro Laurenzi	A study of Sex-Bias' effects on evolution and sex determination	*1*
4.	Andrea Pecci	Endogenized mitochondrial gene expressed in the germ line	*1*
5.	Valentina Peona	Comparison of the transposable elements dynamics among taxa with different rates of speciation	*1*
6.	Claudia Scavariello	First case of R2 non-LTR retrotransposon horizontal transfer: data from *Bacillus* stick-insects genome (Insecta Phasmida).	*1*
7.	Michela Pacifico	Successful sequencing of Huntington's disease CAG microsatellite orthologs from museum collection specimens	*2*
8.	Daniela Minelli	Understanding the rise of modern elasmobranchs and predator-prey interactions in ancient coral reef food web: an example of research in scientific museums.	*2*
9.	Saverio Vicario	Across the shifting baselines - measuring change in biodiversity in the sea benthos of the Kattegat across 70 years and different sampling technology	*2*
10.	Federica Costantini	Between Scylla and Charybdis: differential genetic patterns in Eastern and Western Mediterranean Patella caerulea populations	*3*
11.	Samanta Michelini	*Bifidobacterium* spp. from non human primates: a source of novel species	*3*
12.	Vito Scicchitano	Phylogeography and colony structure of dry-wood dwelling termites of the genus *Kalotermes* (Isoptera, Kalotermitidae) in Southern Europe	*3*
13.	Saverio Vicario	Predictive dating of the Plum pox virus evolution indicates a recent appearance of main recombinant strains.	*3*
14.	Gianluca Zuffi	Definition of three distinct taxa within the *Barbus* species complex	*3*
15.	Elisabetta Versace	Olfactory responses in *Drosophila suzukii* and *Drosophila melanogaster*: a comparative study	*4*
16.	Andrea Benazzo	Using ABC and microsatellite data to detect multiple introductions of invasive species from a single source	*5*
17.	Lino Ometto	Genome sequencing galore: what, how and why	*5*
18.	Elisa Bellucci	The process of domestication in common bean	*6*
19.	Giorgio Binelli	Genetic survey of the *Cistus albidus* populations in Western Mediterranean	*6*
20.	Andrea Brunelli	Maternal history of Southeast Asia and Oceania	*7*
21.	Roberta Rosa Susca	Y-chromosome and mtDNA diversity in the context of Eurasian language diversity	*7*

22.	Filippo Barbanera	Molecular phylogeography of the smooth-coated otter (*Lutrogale perspicillata*, Mustelidae) for its conservation in the Mesopotamian marshes of Iraq	***8***
23.	Michele Cesari	Brown marmorated stink bugs are invading Europe: potential pathways of origin of the alien pest populations of *Halyomorpha halys* (Heteroptera, Pentatomidae)	***8***
24.	Cristina Crava	The taste of *Drosophila suzukii*: phylogeny and evolution of its gustatory receptors	***8***
25.	Silvia Fuselli	The study of MHC variation and evolution in the Alpine chamois: from targeted Sanger sequencing to portable nanopore technology (MinION)	***8***
26.	Mariangela Iannello	A transcriptome annotation pipeline for non-model organisms	***8***
27.	Costanza Piccoli	Wikipedia: with great power comes great (evolutionary) responsibility	***8***

Oral communications

Symposium # 1

Evolution of Genomes and Genomic Conflicts

INVITED SPEAKER

Endosymbiotic origin and differential loss of eukaryotic genes

Martin W. F.

Institute of Molecular Evolution, Heinrich-Heine-Universitaet Duesseldorf, Duesseldorf, Germany.

Chloroplasts arose from cyanobacteria, mitochondria arose from proteobacteria. Both organelles have conserved their prokaryotic biochemistry, but their genomes are reduced, and most organelle proteins are encoded in the nucleus. Endosymbiotic theory posits that bacterial genes in eukaryotic genomes entered the eukaryotic lineage via organelle ancestors. It predicts episodic influx of prokaryotic genes into the eukaryotic lineage, with acquisition corresponding to endosymbiotic events. Eukaryotic genome sequences, however, increasingly implicate lateral gene transfer (LGT), both from prokaryotes to eukaryotes and among eukaryotes, as a source of gene content variation in eukaryotic genomes, which predicts continuous, lineage-specific acquisition of prokaryotic genes in divergent eukaryotic groups. Recent genome scale investigations involving clustering and phylogenetic analysis of eukaryotic gene families having prokaryotic homologues discriminate between these two alternatives. In line with the predictions from endosymbiotic theory, the results indicate that gene transfer from bacteria to eukaryotes is episodic, as revealed by gene distributions, and coincides with major evolutionary transitions at the origin of chloroplasts and mitochondria. Furthermore, gene inheritance in eukaryotes is vertical, as revealed by extensive topological comparison, sparse gene distributions stemming from differential loss. Moreover, continuous, lineage-specific LGT, although it sometimes occurs, does not contribute to long term gene content evolution in eukaryotic genomes.

Reproductive biology *versus* transposable elements load: the role of host reproductive strategy in the study of R2 dynamics in *Bacillus* stick insects (Phasmida, Bacillidae)

Bonandin L., Luchetti A., Mantovani B.

Dipartimento di Scienze Biologiche, Geologiche ed Ambientali (BiGeA), Università di Bologna, Italy.

Transposable elements (TEs) are known to promote genome evolution, even if they are often defined as “genomic parasites”. According to the Red Queen hypothesis, conflicts between parasite TEs and the host genome are at equilibrium through strict competitive efforts. Host reproductive strategy is significantly involved in this dynamics. The R2 non-LTR retrotransposon activity was estimated, through insertions display analyses, in populations of the strictly gonochoric *Bacillus grandii maretimi,* the facultative parthenogenetic *B. rossius* and the obligatory parthenogenetic *B. atticus*. R2 activity was also evaluated in the progeny of parthenogenetic isolates of *B. rossius* and *B. atticus*, and in the male progeny of crosses between gonochoric individuals of *B. rossius*. Gonochoric populations showed higher R2 loads than the parthenogenetic ones, the lowest value being scored in *B. atticus*. In all samples low frequency insertions are the majority, with the only exception of a bisexual *B. rossius* population. R2 load was similar in unisexual and bisexual *B. rossius* offspring but lower in *B. atticus* progeny. Moreover, in *B. rossius* unisexual offspring high-frequency insertions were the most represented. Data on facultative unisexuals evidenced a low R2 elimination rate, with recombination having a major role, although a bisexual-like insertion profile emerge at the population level. In this regard, natural selection seems to predominate in bisexuals. Stick-insects obligatory parthenogenesis seems to allow only a very low R2 load, with possible burst of retrotransposition buffered at the population level. Our data agree in indicating TE dynamics deeply linked with host reproductive strategies.

Insights on the evolution of the bacterial endosymbiont *Holospora caryophila* from the genome sequence

Castelli M.[1], Lang B. F. [2], Burger G.[2], Petroni G.[1]

[1]Department of Biology, University of Pisa, Italy; [2]Department of Biochemistry and Robert-Cedergren Centre for Bioinformatics and Genomics, Université de Montréal, Canada.

We aimed to obtain and analyse the genome of *Holospora caryophila*, an obligate bacterial endosymbiont which interacts with its host, the ciliate *Paramecium octaurelia*, through a peculiar infectious life cycle. *Holospora* represents an interesting target for evolutionary biology studies, taking also into account that it is considered part of *Rickettsiales* (*Alphaproteobacteria*), a group of obligate intracellular bacteria showing a broad host range, which includes pathogens of humans and other organisms.

We produced a draft assembly of the *H. caryophila* genome (76 contigs, 942327 bp, N50: 31236 bp, GC 33.8%), and annotated it through automatic pipelines followed by manual verification. Similar to other *Rickettsiales*, the metabolic repertoire of *Holospora caryophila* is highly specialised to its intracellular lifestyle, as several pathways typical in free-living bacteria were lost completely, and only a limited set was retained. Among *Holospora*-specific features it is notable the lack of a citric acid cycle and oxidative phosphorylation. Therefore, this bacterium appears to rely directly on the host not only for the provision of most biosynthetic precursors, but also for energy supply.

Telomeric sequences distribution in nine Neotropical primates (Platyrrhinae) by (TTAGGG)n probe mapping: interstitial location and their possible role in evolution

Dumas F., Sineo L.

Dipartimento di Scienze e Tecnologie Biologiche, Chimiche e Farmaceutiche, Università degli Studi Palermo, Italy.

Platyrrhini are characterized by a high intra and intergeneric genome variability. This genome variability due to intra and inter-chromosomal rearrangements occurred during evolution could be possibly link to the involvement of telomeric and heterochromatin sequences. Telomeres, the terminal regions of chromosomes, constituted of particular repeated DNA sequences $(TTAGGG)_n$ and proteins, are called true telomeres and have their role in determining the replication and the stability of chromosomes. The DNA component of telomeres can be also found at intra-chromosomal sites such as close the centromeres and between centromere and telomeres, and are known as Interstitial Telomeric Sequences (ITSs). Traditionally considered just "junk DNA", ITSs have been on the contrary associated with chromosomal evolution even if their role has not been yet elucidated. Chromosomal localization of ITS sequences was analyzed mapping telomeric (TTAGGG)n sequence (PNA probe) by fluorescence *in situ* hybridization (FISH) on: *Callithrix argentata,Callithrix jacchus, Cebuella pygmaea, Saguinus oedipus, Saimiri sciureus Aotus nacymaae, Aotus lemurinus griseimembra* (Cebidae), *Lagothrix lagotricha* (Atelidae), *Callicebus moloch* (Pithecidae). Telomeric sequences are localized not only at the terminal ends of all chromosomes but our results indicate that different types of ITSs (centromeric, interstitial and acrosubtelomeric) are present into three among the nine analyzed, in *Lagothrix lagotricha* and the two *Aotus*. We discuss ITSs possible role in chromosomal evolution and as potential markers in phylogenetic studies for close related species. More molecular cytogenetics studies will permit a better understanding of the mechanism of ITSs origin and contribution to chromosomal evolution in Neotropical primates.

The Doubly Uniparental Inheritance: a model system for studying evolutionary and functional genomics of mitochondria.

Ghiselli F., Milani L., Guerra D., Iannello M.,
Procopio E., Punzi E., Plazzi F., Pecci A.,
Maurizii M. G., Passamonti M.

Department of Biological Geological and Environmental Sciences - University of Bologna, Italy.

Mitochondria are a fundamental component of the eukaryotic cell, nonetheless, available data on their heredity and biogenesis are largely incomplete. Our research is focused on the Doubly Uniparental Inheritance (DUI) of mitochondria. DUI organisms have two mitochondrial lineages, one transmitted through eggs (F-type), the other through sperm (M-type), whose mtDNAs show up to 50% of amino acid divergence. In DUI species, after amphimixis, the embryo is heteroplasmic for its mtDNA, a status that is eventually maintained only in males, where F-mtDNA localizes in the somatic tissues, while M-mtDNA localizes in both germ line and soma. Conversely, in females M-mtDNA disappears, restoring the homoplasmic condition. There is both molecular and phylogenetic evidence that DUI evolved as a modification of the mechanism of maternal inheritance, typical of all Metazoa. Our work aims at expanding the knowledge we have about DUI to make it a model system for mitochondrial biology. Thanks to its unusual features, DUI can shed light on mitochondrial inheritance and biogenesis and on the relationship between mitochondria and germ line components. Moreover, the DUI male represents an unique experimental system for studying mitochondrial heteroplasmy: the heteroplasmic condition of DUI males is natural, therefore the biological functions and interactions between nucleus and mitochondria are the unaltered result of evolution. Among those interactions, of particular interest is the relationship between two processes that shape genome evolution: genomic conflicts and coevolution between nuclear and mitochondrial genes. There is no biological system more suitable than DUI to study that.

Investigating the genotypic and phenotypic basis of continental specific *Drosophila*-*Wolbachia* endosymbiotic system

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Rota-Stabelli O.[1]

[1]Fondazione Edmund Mach, Italy; [2]Medical University of Vienna, Austria; [3]University of Cambridge, UK.

In insects, endosymbiotic bacteria of the genus *Wolbachia* are well known manipulators of host reproductive biology. In the absence of any phenotypic effect such as Cytoplasmic Incompatibility (CI), they apparently behave as facultative mutualist. The recent symbiotic system of *D. suzukii*-*Wolbachia* (*Dsuz*-*w*Suz) is extremely intriguing to explore since in spite of having no CI-inducing capability, it prevails at different infection frequencies in American and European natural populations, suggesting some fitness advantage rendered by *w*Suz to its natural host. Here we tackle this biological conundrum by coupling comparative genomics and applied microbiology.

By developing a new genome-scaled typing system, we first show that the European *w*Suz is slightly different to that of American one. Comparative genome analysis show that both of the host populations are also different and most likely segregated one million years ago, suggesting the presence of two different continental endosymbiotic genetic sub-systems. We finally show that *w*Suz is providing stronger fitness benefits to European *D. suzukii* population than the American sub-system, one of the likely causes behind persistence of *w*Suz at higher prevalence in European *Dsuz* under balancing selection. Results further suggest that the continental specific *w*Suz variants observed in *Dsuz* may likely be due to different genomic backgrounds, resulting in different fitness benefits. Our data shed new light on the complex co-evolution of *Drosophila*-*Wolbachia* interaction system and their genome biology.

Recombine and survive: evolutionary history of the V highly conserved domain in the mammalian genome after the V-SINE superfamily extinction.

Luchetti A.[1], Lomiento M.[2], Mantovani B.[1]

[1]Dip. Scienze Biologiche, Geologiche e Ambientali - Università di Bologna; [2]Dip. Scienze della Vita - Università di Modena e Reggio Emilia.

Short interspersed elements (SINEs) are non-autonomous retrotransposons. Because of high turnover rate, they exhibit widely divergent nucleotide sequences but highly conserved domains (HCDs) may occur. Three different HCDs originated before the Radiata-Bilateria split and two underwent repeatedly to exaptation. The "V" HCD has been retrieved in 16 nonamniotes'SINE families and within a miniature inverted-repeat transposon, MER6, in the human genome. Through *in silico* searches in sequenced genomes and transcriptomes, we found MER6 in all primates and its variant, MER6A, in bats and in the star-nosed mole. Moreover, we found evidence of MER6 in a salamander and retrieved its parental *Tc1/ mariner* element in the painted turtle. Data indicate that MER6 originated by recombination between a V-SINE and the parental *Tc1/mariner* element. Nucleotide substitution rate, computed on 10 primates orthologous insertions, places MER6 main activity burst as contemporary to Placentalia diversification. Moreover, age analyses suggest that MER6 was no longer active in mammals, except in the tarsier where a recent replication burst originated a new MER6A subfamily. Finally, unlike frog and fishes V-SINEs, genomic distribution of MER6 does not appear biased toward genic regions in human, chimp and mole genomes. Present data show how the V domain managed to survive and replicate after V-SINEs extinction. Although the impact of HCDs is still unclear, their wide conservation suggests some role; the study of V-elements with different replicative biology may help to disentangle transposon-related aspects from putative HCD functions

Mitochondrial activity in gametes and transmission of viable mtDNA

Milani L., Ghiselli F.

Dipartimento di Scienze Biologiche, Geologiche ed Ambientali, Università di Bologna.

Oxidative phosphorylation makes mitochondria a potentially detrimental environment for DNA because of the possible mutagenic effect of Reactive Oxygen Species (ROS) that can affect the transmission of a correct genetic information through generations. How can a faithful transmission be achieved? Since the mtDNA is commonly maternally inherited, it was proposed that female gametes would prevent damages by repressing oxidative phosphorylation and being quiescent genetic templates. We show evidence from several species that are consistent with mitochondria being active in both gamete types, not only in species with strictly maternal inheritance (SMI), but also in species that naturally transmit mitochondria also through males (doubly uniparental inheritance of mitochondria, DUI). Consistently with these observations, we support the alternative hypothesis that the activity of template mitochondria is fundamental for the inheritance mechanism. During gametogenesis, fertilization, and embryo development, the great reduction in mtDNA copy number per organelle/cell and the stochastic segregation of mtDNA variants would greatly improve the efficiency of selection: a combination of drift and selection on germ line mtDNA population might be responsible for the maintenance of viable mitochondrial genetic information through generations, and the mitochondrial activity would be the phenotype under selection. In this case, efficient mechanisms of mitochondrial genome preservation and/or ROS scavenging, or alternative pathways of ATP production, would act to favour an accurate transmission of the genetic information, albeit coupled with a full mitochondrial activity. Thanks to its unusual features, DUI emerges as an excellent model system to study these problematics.

100 of these genomes: Bivalve mitogenomics hits triple digits

Plazzi F., Passamonti M.

Dipartimento di Scienze Biologiche, Geologiche e Ambientali – University of Bologna.

Bivalve mitochondrial genomes show several uncommon features, and more are being discovered. In most cases, the molecule is large (up to 30-40 kilobases) and many untranslated regions are present, which probably have some role in regulation of both replication and transcription of the genome. In many cases, supernumerary Open Reading Frames (ORFs) have been located, in addition to the standard metazoan mitochondrial gene content. Most bivalve groups underwent an intense evolutionary pace, which led to an astonishing variability at the nucleotide level, but most notably in terms of gene order. As if that were not enough, some bivalve species exhibit Doubly Uniparental Inheritance (DUI): DUI is a phenomenon of mitochondrial inheritance that involves a complex (and still unclear) cellular machinery that maintains two separate mitochondrial lineages instead of a single, maternally-inherited lineage, as found in most metazoans.

In this contribution, 100 mitochondrial genomes are analyzed together and compared in the effort of assessing a global scenario of bivalve mitogenomics: main pathways of evolution are highlighted and typical features are underlined group by group; a fossil-based time-calibrated phylogeny and some updates to GenBank annotations are also presented. We have strong evidence favoring the idea that different groups of bivalves underwent different selective pressures that led to taxon-specific mitochondrial features, like size, nucleotide composition, strand and codon usage.

Even if further research is needed to sharpen the panorama of bivalve mitogenomics, the present contribution sets the first step in defining main lines of evolution and diversity of bivalve mitochondrial genomes.

Adaptive evolution underlies the species-specific binding of *P. falciparum* RH5 to human basigin

Pontremoli C.[1]*, Forni D.*[1]*, Cagliani R.*[1]*, Pozzoli U.*[1]*,*
Clerici M.[2,3]*, Sironi M.*[1]

[1]Scientific Institute IRCCS E.MEDEA, Bioinformatics, Bosisio Parini, Italy; [2] Department of Physiopathology and Transplantation, University of Milan, Italy; [3] Don C. Gnocchi Foundation ONLUS, IRCCS, Milan, Italy.

Plasmodium falciparum, the causative agent of the deadliest form of malaria, is a member of the *Laverania* subgenus, which includes ape-infecting parasites. *P. falciparum* is thought to have originated in gorillas, although infection is now restricted to humans. *Laverania* parasites display remarkable host-specificity, which is partially mediated by the interaction between parasite ligands and host receptors. We analyzed the evolution of BSG (basigin) and GYPA (glycophorin A) in primates/hominins, as well as of their *Plasmodium*-encoded ligands, PfRH5 and PfEBA175. We show that *BSG* evolved adaptively in primates with selection targeting two sites (F27 and H102) involved in PfRH5 binding. A population-genetic phylogentics approach detected the strongest selection for the gorilla lineage: one of the positively selected sites (K191) is a major determinant of PfRH5 binding affinity. Analysis of *RH5* genes indicated adaptive evolution on the *P. falciparum* branch; the positively selected W447 site is known to stabilize the interaction with human basigin. Conversely, we detect no selection at *EBA175* gene in the *P. falciparum* lineage. Its host receptor, GYPA, shows evidence of positive selection in all hominid lineages; selected codons include glycosylation sites that modulate PfEBA175 binding affinity. Data herein provide an evolutionary explanation for species-specific binding of the PfRH5-BSG ligand-receptor pair and support the role of adaptive evolution at these genes as a determinant of the host shift that led to the emergence of *P. falciparum* as a human pathogen. A minor role in this process seems to have been played by the interaction between EBA175 and GYPA.

Epigenetic divergence and parallel adaptation in *Heliosperma pusillum* (Caryophylaceae)

Trucchi E.[1]*, Flatscher R.*[1]*, Romero M. L.*[1]*, Frajman B.*[2]*,*
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Epigenetic modifications are expected to occur at a faster rate than genetic mutations as a consequence of the higher instability of epigenetic marks or as a response to environmental stress. These changes often induce alterations of genes expression with important phenotypic consequences potentially visible to natural selection. We then expect epigenetics to be particularly important in the early phases of adaptation to novel environmental conditions. The alpine plant *Heliosperma pusillum* and its recently and recurrently diverged sibling *H. veselsky,* found in low-montane habitats, constitute a perfect model system to test this hypothesis. They occur in distinct ecological conditions and have divergent morphologies (glabrous versus hairy plants), stable and heritable in common garden settings. However, genome-wide data, produced through RAD sequencing, fail to group them according to the observed phenotypes. Employing a novel approach (bisulfite-converted RAD sequencing), we screened DNA methylation at ca. 2,000 loci, 100 bp long, in six population pairs of the two ecotypes. Comparing by sequencing the DNA methylation state and context (CpG, CHG, CHH) in 120 individuals, we found that the level and distribution of DNA methylation is globally more stable across divergent ecotypes than predicted given the distinct environmental conditions. This stability points to molecular or developmental constraints acting on DNA methylation at the loci analyzed. Nevertheless, few candidate loci for convergent epigenetic modifications during parallel adaptation to a similar environment are present.

Evidence for purifying selection of mtDNA mutations during human oogenesis

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Several lines of evidence (i.e. Ruiz-Pesini *et al.*2004, Stewart et al. 2008, Gigarel *et al.* 2011, reviewed Castellana, *et al.* 2011) show that in metazoa the variation within the germline of mitochondrial genomes is under purifying selection. The presence of this internal selection filter on the germline has important consequences for the evolutionary trajectory of mtDNA. The nature and localization of this internal filter are still unclear with several hypotheses proposed in the literature. Comparison of substitution rates show that the filter is more effective on protein coding regions than on tRNAs (Pesole *et al.* 1999).

In this study, 60 mitochondrial genomes were sequenced from 17 sets of oocytes, first and second polar bodies, and blood, taken from 9 women between 38 and 43 years of age with 6 different mitochondrial macro-haplogroups. Whole Genome Amplification was performed. on the single cell samples, and a Sanger sequencing was performed on amplicon produced with MitoALL Resequencing kit (Applera, Foster City, CA). The comparison of mutation profiles between first and second polar body showed no difference in substitution rates, but instead displayed a sharp difference in pathogenicity scores of protein coding sequences using 7 different metrics (Calabrese *et al. 2014*). Second polar bodies, differently from first, did not show significant differences in pathogenic scores with blood and oocyte sequences. This suggests that a non-identified filtering mechanism is active during oocyte development at the time between the expulsion of the first and second polar body.

Symposium # 2

Evolutionary Research in Museums

INVITED SPEAKER

When traditional museological tools meet new cutting-edge applications: our experience with the study of species diversification patterns, taxonomy and conservation of Madagascar herpetofauna

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Madagascar hosts an unparalleled concentration of unique, diverse and threatened flora and fauna. The products of our field-based research began over 25 years ago and resulted in the description of several new species and in the collection on wide variety of ecological and distributional data. Although Madagascar has a well-developed network of protected areas, ongoing habitat destruction, political instability, emergent infectious diseases and invasive species are weakening protection in many reserves and put others at risk, threatening Madagascan unique biodiversity. It is in view of all this that we invested in the establishment of a solid network of researchers and initiatives that is aiming at protecting the unique amphibians of Madagascar.

Why some groups of organisms are more abundant than others, at different geographical locations or at different points in time is dependent on numerous factors. Deciphering these factors remains one of the most challenging questions in evolutionary biology. The mantellid frogs of Madagascar underwent a wide species radiation that resulted in the evolution of a plethora of morphological and ecological traits. We will combine evolutionary genomics and comparative phylogenetic methods to investigate the evolutionary association between life-history and morphological traits and test for their contribution to the diversification of this group. To do this we will re-analyse the herpetological collection hosted in Museo Regionale di Scienze Naturali in Turin. While valorising this unique collection we will create an invaluable resource that will be available for the following generations of researchers and conservationists.

The “Extinction” project: valorization of museum natural history collections as a tool to link research, outreach and conservation

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The “Extinction” project, funded by Ministry of Education, University and Research through the law 6/2000 for dissemination of scientific culture follows in the steps of “VertEx - Vertebrata Extincta”, a project launched in 2007, which is aimed at building a database of threatened vertebrates housed in Italian natural history collections. Results of this survey will boost a scientific, specimens-based project, which will tie naturalistic museums, academies, and private institutions. Gene sequences (in particular mitochondrial DNA) will be obtained from collection samples. High-throughput sequencing showed to be effective in sequencing DNA of museum specimens, opening the way for new collections-based projects. In particular, we are focusing on taxa for which no molecular data is available, or for which it is interesting to expand our knowledge on population genetic aspects providing new data to both geneticists and curators. A main outcome of “Extinction” will also be an exhibition, which will first open in MUSE in July 2016, and later made available to other Italian institutions. Starting from historical research, through cataloguing, genetic analysis and exhibit development, “Extinction” brings different aspects of museums activities under a single project with the definitive aim of giving new life to naturalistic collections.

Unravelling the biogeography of secretive taxa by museum collections: the untold story of the black francolin (*Francolinus francolinus*, Phasianidae) in the Mediterranean

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The black francolin (*Francolinus francolinus*) (BF) comprises six morphological subspecies distributed from Cyprus and Turkey across Asia to India. In spite of being renowned as courtly gamebird since the Classic Age, this species suffers from paucity of demographic and molecular studies. In order to update the BF biogeographic pattern by pursuing a thorough sampling across the unsafe and remote areas representing most of the specie's range, tissues from museum specimens (76, XVIIIth c.-1954) hosted in US and European ornithological collections were genotyped at a 185 bp-long fragment of the mtDNA Control Region gene along with modern birds (205) sequenced at the entire gene. The access to ornithological collections opened the unforeseen opportunity to elucidate the genetic affinity of the extinct populations once residing in the western Mediterranean (Italy, Spain), thus settling the debate about autochthony versus allochthony in that region. Three well-defined haplogroups - each one including a pair of morphological subspecies and matching the phylogeographical pattern inferred with the whole gene - were found to reflect a westward adaptive radiation, a more complex scenario being nonetheless disclosed in the Indian sub-continent. The nonnative status of the western Mediterranean BFs was ultimately assessed, a tight genetic affinity with conspecifics from Cyprus and southern Asia being found. This finding, which partly confirmed the invoked importation during the Crusades, pointed to the major human impact on Mediterranean biodiversity through long-distance trade across Asia to satisfy the high demand for exotic species by the European aristocracy during the Medieval times and the Renaissance.

The tale of specimen: what and when natural history collections can tell us about evolution

Latella L.

Museo Civico di Storia Naturale di Verona.

Since the establishment of the first museums of natural history, it is well known that every single specimen can provide a world of information about ecology, biogeography, taxonomy, history of science and so on.

However, until a little over a decade ago, they could hardly give us information on molecular genetics.

The development of the Polymerase Chain Reaction, in the late1980s, led natural history collections to a kind of renaissance in the field of studies on evolutionary biology. Thanks to the possibility offered by this technique to amplify the few intact molecule of DNA existing in a specimen, a growing number of evolutionary biologists have begun to use the collections for their research.

Several major museum in the world organized collection devoted to these studies and sett up molecular laboratories.

The situation in Italy as always presents many facets and different approaches both in management and in the study of the collections.

A picture, as complete as possible, is provided taking into consideration the use and management of collections in some of the most representative Italian natural history museums.

Restoring the history of sawfish in the Mediterranean from museum exhibits

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The historical occurrence of sawfishes in the Mediterranean Sea has been the subject of active debate over the last decade (Ferretti et al. 2015). The growing consensus among specialists is that sawfishes may have never occurred in the Mediterranean with stable populations (Harrison & Dulvy, 2014). Conversely, extensive literature search, archival and museum data mining, and extinction probability analyses (Ferretti et al. 2015), provided evidence to support the hypothesis that sawfishes (*Pristis pristis* and *Pristis pectinata*) once occurred in the Mediterranean with stable populations. These results challenge several tenets of sawfish temperature tolerance, movement, distribution, and population structure. They would expand sawfishes' historical distribution range in temperate latitudes and thus raise important questions on how these populations might have survived in the Mediterranean Sea.

Molecular techniques on historical DNA promise opportunities for characterizing population structure, biogeography and evolution of elasmobranchs (Leone et al. In prep.). As well, isotopic and microconstituent analyses on rostral teeth can provide further information on date, biogeography, movements and habitat use of the individual specimens (Field et al. 2009). To date we have sampled 58 museum specimens of sawfish. Material labelled as Mediterranean had a great uncertainty on acquisition date and location. Other sawfish remains had no info at all on acquisition date, species identity and capture location. This project intends to use genetic and biogeochemical analyses to address this uncertainty, and eventually using the material identified as coming from the Mediterranean to characterize the population and genetic structure of the sawfishes once occurring in the region.

Unlocking the evolutionary history of the mighty Bluefin Tuna using novel paleogenetic techniques and ancient tuna remains

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Mediterranean civilizations and the Atlantic bluefin tuna (*Thunnus thynnus*) have shared a dramatic history spanning millennia. Today, traditional fishing techniques are fading into the past and industrial fisheries command the seas. Analyzing tuna bones recovered from archeological excavations throughout Europe, we aim to reconstruct the species' adaptive responses to fisheries pressure and climate change. Using novel molecular techniques, DNA has been extracted from bluefin tuna vertebrae excavated from late iron age and ancient roman settlements in coastal Iberia (4th-2nd century BC) and Byzantine-era Constantinople (4th-15th century AD), as well as vertebrae from the Massimo Sella archive located at the University of Bologna (Ionian, Tyrrhenian and Adriatic Seas, early 20th century). The species identity of all samples has been verified using newly developed mini-barcodes targeting mitochondrial and nuclear markers. Samples have been quantified and analyzed using multiple technique (qPCR, Agilent BioAnalyzer), in order to correlate Atlantic bluefin tuna DNA content with samples' age and diagenetic processes. Suitable samples were selected for subsequent high-throughput sequencing on Ion Torrent platform to assess temporal genomic changes induced by environmental and natural history drivers. All historical samples, along with modern ones collected from the same geographic areas, have been genotyped with a dedicated SNP panel, which is composed of 76 SNPs with high similarity to a wide variety of annotated genes and additional 20 SNPs that provide significant discrimination between modern populations. These efforts will provide new insights into the spatial dynamics, population structure and genetic evolution of the species over the past 2000 years.

Symposium # 3

Phylogenetics, Biogeography and Phylogeography

INVITED SPEAKER

The Evolution of Neotropical Mega-Diversity

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Understanding the origins and evolution of the outstanding biodiversity found today in the American tropics (the Neotropics) constitutes a major goal of evolutionary biogeography. In this talk I will present some of our recent work towards this goal, focusing on *i)* data-driven identification and delimitation of biogeographical regions instead of opinion-generated classifications; *ii)* spatial coding of species into regions and altitudinal ranges for biogeography and macroecology; and *iii)* the estimation of historical range shifts and region-specific species diversification. Our studies on these topics indicate, *inter alia*, that the application of network methods on species occurrence data can extract considerably stronger biogeographical signal than hitherto realized; that bioinformatic tools are able to provide rapid assessments of biodiversity patterns; that Neotropical angiosperms have speciated and gone extinct at significantly higher rates than in other tropical regions; that tropical America in general, and Amazonia in particular, have served as species pumps through most of the Cenozoic for multiple taxa; that the Great American Biotic Interchange occurred millions of years earlier than assumed; and that fossils play a major role in reducing uncertainties in biogeographical analyses. These examples showcase how old questions can be addressed by new methods and more data, but despite major advances in the last decade by the scientific community we still know bafflingly little about the evolution of Neotropical mega-biodiversity. I will therefore end with a call to Italian researchers and students to strengthen their links with Neotropical research, arguing that many low-hanging fruits are still to be picked and much knowledge to be gained.

Species between conservation and evolution

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MUSE – Museo delle Scienze, Trento, Italy.

The term "species" refers to very different entities. The term species also indicates a *taxonomic category*. A third, less discussed but even more problematic tendency, equals "species" to *evolutionary units*. However, the boundaries assigned to evolutionary units, are the results of evolutionary processes, ontologically different from those of "species" (whatever this might mean) and they do not coincide with either the specific taxa, nor with their essence.

This mismatch led conservation biologists to circumnavigate the species. The units on which, increasingly, conservation actions are focused on today are in fact "packages of diversity" identified by indexes characterized by higher affinity with evolutionary units than with taxa, and often used without any bound with species names. If conservationists seem to have found a practical solution to the "species problem", the debate remains completely open for taxonomists. If taxa are not evolutionary units, the names that have been assigned to them does not refer to identifiable units in nature, depriving of meaning any comparison or index based on them.

While the Botanical Code of nomenclature doesn't assign any relationship between nomenclature and taxonomy, the zoological one does. We believe that zoologists should follow in the steps of botanists stating in their nomenclature code the uncertainty of any taxonomic statement. Anyway we need to find the best ways to define the entities that can be used in daily practice, and perhaps some are already in use, assuming that evolution is a phenomenon too complex to be framed in discrete and comparable units.

Unexpected biodiversity of epizoic gastropod genus *Marseniopsis* (Velutinidae) within a phylogenetic framework for the family

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Marseniopsis Bergh, 1886 is a genus of marine gastropods (Velutinidae) which includes nine described species, eight of which endemic to Antarctica. This genus is characterized by a peculiar planktotrophic larva, the "limacosphaera", with a potentially long pelagic life, which is a really uncommon developmental strategy for Antarctic gastropods. In addition, we observed several different colour patterns, within and amongst presumed conspecific samples.

We produced a dataset of COI, 16S and 28S genes of 107 samples of *Marseniopsis* from the Ross Sea, the Dumont d'Urville Sea, the tip of the Antarctic Peninsula and the Weddell sector. Through genetic distances and Automatic Barcode Gap Discovery (ABGD) we identified 14 MOTUs, 10 of which exclusively from the Weddell Sea-Antarctic Peninsula. The isolation-by-distance analysis did not show any significant correlation between genetic and geographic distances among populations of each clade.

These results suggest the presence of a hidden biodiversity of Antarctic *Marseniopsis*. Mantel colour turned out not to be a diagnostic character and probably due to the alimentary homochromy of *Marseniopsis* with ascidians. The absence of isolation-by-distance and the presence of some MOTUs in all sampling areas are congruent with the long pelagic life of the *Marseniopsis* larva. However, some MOTUs distribution restricted to Weddell Sea-Antarctic Peninsula lets hypothesize the existence of a second type of larva, with a limited dispersion capacity. We also produce a preliminary phylogenetic reconstruction of this family, as a starting point to extend our investigation to the Velutinidae worldwide.

The role of local refugia and paleoclimatic history in the phylogeography of a cold-tolerant species: the European cave salamander *Hydromantes strinatii*

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Subtroglophiles organisms, able to exploit both caves and epigean habitats, offer an interesting perspective to understand how species responded to past climatic changes, since caves may act as local refugia to escape adverse climatic conditions. European cave salamanders of the genus *Hydromantes* are good candidates for these studies, since their troglophilic habit is coupled with cold adaptation, direct larval development, poor dispersal and naturally fragmented populations. These features make these species able to persist in periglacial areas and, at the same time, prone to retain the signature of local environmental history. In this study the phylogeography of *Hydromantes strinatii* was explored using mitochondrial (Cytb and ND2) and nuclear markers (SLC8A3 gene and thirty-three allozyme loci). Mitochondrial markers recovered two highly divergent clades in the eastern and central-western part of the range, each showing further pronounced geographic sub-structure. Nuclear markers, particularly allozymes, recovered a partially coincident pattern with incongruence suggesting the contribution of different mechanisms, including incomplete lineage sorting and secondary contacts. Split events within this species fall into both glacial and interglacial peaks, suggesting that both climatic extremes have the same adverse effect on this cold-tolerant species, promoting isolation and divergence within the localized refugia represented by caves. Accordingly, a more complex evolutionary scenario is postulated for the central-western populations, occurring in the Ligurian and Maritime Alps, a periglacial refugial area characterized by a complex orography and paleoclimatic history.

Inferring past tree distribution in Nordic environments from ancient DNA

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Range limits of many tree species are expected to shift considerably in the next decades due to human-mediated climate change, particularly in Nordic environments, but the capacity of trees to migrate in response to these events has been questioned. New methodologies allow now to look directly in the species' DNA retrieved from fossil plant archives (peat/lake sediments, permafrost), investigate how and where species survived past cold periods, investigate how they responded in the past and derive lineage specific tolerances and migrations rates for plants.

In my talk I will show how information can be effectively retrieved from these archives and analyzed following a multidisciplinary approach that combines *1)* classical palaeoecological investigations, *2)* DNA metabarcoding and metagenomic techniques, *3)* analytic models based on modern DNA and *4)* species distribution modeling analyses.

The outcome of studies based on such approaches is not only useful for investigating glacial tree survival and the driving factors affecting past range shifts, but also for assisting modeling analyses of future range shifts

Problem bear translocation: is it effective management practice to mitigate bear-human conflicts in Dachigam landscape, Jammu and Kashmir, India?

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Asiatic black bear population in Dachigam landscape, Jammu and Kashmir is well recognized as one of the high density populations in India. Increasing incidences of bear-human interactions and the resultant retaliatory killings by locals has become a serious threat to the survivorship of black bears in Dachigam landscape. Department of Wildlife Protection, Jammu & Kashmir has been practicing translocation of problem bears from different conflict sites to Dachigam National Park as a flagship activity to mitigate/minimize conflicts in Dachigam landscape. We undertook this study to understand the pragmatic fate of translocation of problem bears. Genetic analysis of wild caught hair samples identified 109 unique genotypes and black bear population was found to be under panmixa. Molecular tracking of 11 problem bears revealed that, seven bears after translocation moved backward to their natal sites where they were initially caught, possibly due to homing instincts or habituation to the high quality food available in croplands while only four bears get settled to their release sites in Dachigam National Park. Results demonstrated that translocation success was perhaps season dependent as bears translocated in summer often get settled inside Dachigam National Park due to availability of surplus bear food while bears move backward to their first site of capture in the scarcity of food inside Dachigam National Park during spring and late autumn. We strongly advocate that current management practice of translocation of problem bears to Dachigam National Park is ineffective in mitigating conflicts on long term basis. However, genetic tagging of translocated bears would be tempting to wildlife managers to monitor bear movements in tough terrains and reviewing the pragmatic fate of translocation.

Molecular markers, natural history and evolution in sharks and rays

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Chondrichthyes are an ancient class of cartilaginous fishes, whose systematic and bio-ecological features make them contemporaneously specious and at high risk of extinction. Large sampling efforts carried out in the Mediterranean and Eastern Atlantic for several years provided the unique opportunity to build up unprecedented repositories of vouchers for exploring genetic variation from population to family levels.

Difficulties in species identification linked to high levels of morphological stasis were overcame by wide-scale DNA barcoding analysis of Mediterranean chondrichthian fauna (coverage at 50%) improving species assessment and fostering conservation and management actions.

Phylogenetic relationships and historical biogeography of Mediterranean, Eastern Atlantic and Indian Ocean skates (Rajidae) have been reconstructed using concatenated mtDNA genes revealing patterns of antipodean allopatric parallel speciation strictly correlated to a pan-biogeographical (vicariance) model.

Coupling integrated panels of unisexually and bisexually inherited markers and large scale sampling of historical and contemporary populations of threatened and endangered sharks and skates permitted to reveal several cryptic speciation events, strong phylogeographical and population structure unexpected in marine realm disentangling natural and phylogenetic histories and allowing the practical identification of conservation and management units.

A backboned phylogeny for invertebrates: the case and problems of eutardigrades (Tardigrada)

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Tardigrades are a small phylum of micrometazoans capable to undergo cryptobiosis (i.e. an ametabolic state of life in response to adverse environmental conditions). Phylogenetic studies on tardigrades relied almost only on molecular characters, while morphological based phylogenies are scarce. Beside the problems of working with microscopic animals, the main limitation in the use of molecular characters is the difficulty in obtaining specimens for the analyses, as many species and genera have been found only once, in remote places and in small amount, leading to a poor taxon sampling. Morphological characters instead can be easily obtained from literature and slides collections, but they are very limited in number, difficult to score and prone to homoplasy. Total evidence analyses with concatenated morphological and molecular dataset didn't provide satisfactory results. To overcome these limitations a backboned approach has been applied. In this approach a scaffold from molecular phylogenies is used to constrain the Bayesian analyses of morphological characters. This approach has been used before to place fossil taxa in phylogenies with extant taxa for wich was avaible a molecular phylogeny. The main limitation in applying this approach to eutardigrades is the different level of taxon sampling of molecular (exemplar terminals) and morphological (supraspecific terminals) data. A way to overcome this problem is the leveling of terminal taxa to the same taxonomic level. In this case study, the supraspecific (genus) terminals are used. This approach led to an eutardigrades phylogenetic tree inclusive of all genera never obtained before.

Symposium # 4

Evolution of phenotypes and Organismal Biology

INVITED SPEAKER

Regulatory evolution and the diversification of pigmentation patterns in *Drosophila*

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The mechanisms by which new gene networks emerge, and how changes in gene regulatory interactions produce phenotypic diversity, remain elusive. We are addressing these general questions in the context of morphological evolution by studying the evolution of wing pigmentation patterns in Drosophila species. We showed that a spot of dark pigment on the wings of the fly *Drosophila biarmipes* emerged from the assembly of a novel gene regulatory network. In the ancestor of this species and its wing-spotted relatives, a set of effector pigmentation genes evolved to respond to the common transcriptional regulator, which determines their spatial distribution. Using inter-species comparative functional genomics approaches we are piecing together the regulatory network underlying the formation of the wing spot and tracing its evolutionary origin. Subsequently to its evolutionary emergence, the primitive wing spot diversified in shape, size and intensity in different species. We are studying the genetic sources of this diversification to explore how the changes responsible for the wing spot evolution along different phenotypic directions are distributed within the gene regulatory network.

Genetic connectivity along depth gradient and implications for phenotypic plasticity in a gorgonian species

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Octocorals are characterised by morphological plasticity related to the abiotic and biotic environment where they live. Depth is not a strong environmental factor *per se*, while is a proxy for several environmental parameters (e.g. temperature, light, wave action, currents, food supply), which may affect the structure and dynamics of populations, as well as connectivity patterns and genetic diversity. *Eunicella singularis* is a Mediterranean arborescent gorgonian forming subtidal animal forests, playing an important role as engineer species by providing biomass and complexity to coralligenous and recoralligenous habitats. *E. singularis* has a wide bathymetric distribution, ranging from shallow (20m) to deep (100m) rocky bottoms. The species shows two depth-related morphotypes which taxonomic status is not yet clarified due to the low-resolution power of molecular markers used up to now. We used microsatellite loci and ITS sequence polymorphism to analyse the genetic structuring among populations along both horizontal and bathymetric gradients in Cap de Creus (Spain).

The results revealed that connectivity and genetic variability of *Eunicella singularis* populations significantly vary with depth but not with the horizontal distance. A threshold in connectivity was found across 30 - 40 m depth, suggesting the occurrence of restricted gene flow between the two *E. singularis* morphotypes. Variability in morphology with depth could be due to phenotypic plasticity, which should allow populations to suit different environmental conditions, or to a break in gene flow that could determine the isolation between shallow and deep populations and an accumulation of genetic differences.

The phenotypic plasticity in life history traits of an Antarctic water bear as a strategy to cope with extreme environmental conditions

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Relatively few studies have been performed on the role of phenotypic plasticity of life history traits of Antarctic organisms, which allows them to cope with extreme environmental conditions. Since tardigrades are one of the few highly abundant animal groups in Antarctica, the reproductive mode, the life cycle and the comparisons of life history traits over three generations of a Victoria Land population of *Acutuncus antarcticus* were analyzed.

In *A. antarcticus* reproduction occurs by thelytokous meiotic parthenogenesis, as only females were detected. Its life cycle and generation time were short. An early age at first oviposition, a short time interval between two successive ovipositions, and a short hatching time allow *A. antarcticus* to produce more than one generation in a brief time. These traits, along with a relatively high fecundity and fertility, and high hatching percentage allow to this species to quickly increase the population size and to better exploit the conditions suitable for growth during the short Antarctic summer. The phenotypic variability evidenced in the life history traits within and among the three analyzed generations may be due to maternal effects, since the environmental conditions experienced by the mother could indirectly affect the phenotype of her offspring. This may be seen as a bet-hedging strategy which offers insurance against the risks associated with reproduction by expressing a range of diversified phenotypes. This phenotypic variability, along with the cryptobiotic capability of *A. acuntuncus*, is advantageous to cope with the extreme environmental conditions, assuring survival and fitness in space and time.

Maternal effect on offspring size in seed harvester ants

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Eusocial insects are characterized by extreme phenotypic plasticity, in which different phenotypes can be produced within the same colony depending on environmental conditions. For example, since young founding colonies are resource-limited, they produce very small workers. Workers in established colonies, where resources are generally not limiting, are up to four times bigger than those in founding colonies. Here we show that worker size in young versus established colonies of the ant *Pogonomyrmex rugosus* is not only regulated through differential feeding during development but also by the mother queen via maternal effects. A cross-fostering experiment showed that eggs laid by founding queens developed into smaller workers compared to the ones produced from eggs laid by queens from established colonies, when both egg types were raised by workers from established colonies. Moreover, eggs laid by established queens and cross-fostered to founding queens failed to develop into advanced larval stages. These results suggest that maternal effects pre-program worker development for resource-limiting conditions in founding colonies. Furthermore, we gained insights into the mechanisms mediating these maternal effects by analysing gene expression in eggs from established and founding queens. In addition to 40 differentially expressed genes, we found differentially expressed piRNAs and numerous microRNAs that are highly expressed in eggs from established queens, but completely lacking in eggs from founding queens. In combination, these results indicate that small RNAs play a role in mediating maternal effects on adaptive worker size differences in ants.

The Role of Olfaction in the Making of a Pest – A Genomic Analysis

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The emerging model pest *Drosophila suzukii* is characterized by the switch from a fermenting to a fresh fruit reproductive habit, but the genomic bases of this new trait are still widely unexplored. In this study, we have annotated the repertoire of olfactory genes in two populations of *D. suzukii* and in its closely related species, *D. biarmipes*, and performed thorough evolutionary studies on a 14 Drosophila phylogenetic framework. Compared to most other Drosophila species, the odorant receptors of *D. suzukii* are characterized by an increased turnover rate and a non-random distribution of evolutionary events (duplications, deletions, and positive selection), consistent with adaption to a new ecological environment. In *D. suzukii,* odorant receptors that respond to some of the odours typical of ripening fruit have undergone duplication and show signs of positive selection; the most represented volatiles eliciting a response in these receptors include isoamyl acetate, for which we could confirm a functional role in *D. suzukii* using ad-hoc behavioural assays. Conversely, some of the key receptors used to detect volatiles produced during fermentation experienced loss of function and likely neo-functionalization in *D. suzukii*, a finding supported by physiological recordings; these genes are fairly divergent between European and American *D. suzukii* populations suggesting ancient multiple neo-functionalization events. Overall, our comparative analyses reveal unusual genomic evolutionary events in *D. suzukii* that can be associated with adaptations to new ecological behaviours, and unveil key genes and ligands that might become target of applied control strategies.

Symposium # 5

Population Genetics and Genomics

INVITED SPEAKER

Inferring population histories from genomes

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Genomes contain a wealth of information about past demographic events. While we now have access to vast volumes of genomic data, extracting the relevant demographic information remains challenging. Many current inference methods either ignore linkage information or simply do not scale to genomic data. I will describe an analytic framework that uses the full configuration of mutations in short blocks of sequence to fit models of population history. This likelihood calculation is based on a simple recursion for the generating function of the genealogy and applies to arbitrary population histories. I will discuss example applications of this approach including analyses of admixture between Neandertals and modern humans, inferences of a Pleistocene bottleneck in an endemic pig species in island SE Asia, and species divergence in *Heliconius butterflies*.

The evolution of the small and isolated population of Apennine brown bears (*Ursus arctos marsicanus*): a whole-genomes perspective

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Approximately 50 Marsican bears live in the Apennine Mountains in Central Italy. This is the last population of native Italian brown bears, now completely isolated from recently introduced bears in the Alps and from all the other European populations. In order to reconstruct the demographic history of this group and the possible molecular adaptations (or maladaptations) due to independent evolution in a specific environment, and indirectly to evaluate the extinction risks and to suggest conservation strategies, we sequenced the complete genomes of 6 Marsican bears and 6 additional European brown bears from Spain, Greece, Slovakia, and the Alps. The coverage was around 20X for four individuals and around 5X for eight individuals. Preliminary analyses suggest that: Marsican genomes have about 2/3 of the number of SNPs observed in the Spanish bears, and 1/3 of the SNPs found in other European bears; Marsican bears are highly homogenous, and their genomes are similarly distant to all the other European individuals; several fixed non-synonymous substitutions are observed in the Marsican bears; long stretches of homozigosity are predominantly found in Marsican bears, suggesting that inbreeding might be an issue in this population; the pattern of population structure in Europe is weaker at the genomic than at the mitochondrial level; the isolation of the Marsican bear is probably older than previously estimated.

Regressive evolution in Somalian cavefish *Phreatichthys andruzzii*: loss of selective constraint on opsin genes

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Phreatichthys andruzzii is a Somalian cavefish that evolved in complete isolation and absence of light beneath the desert for about two million years. Constant darkness caused extreme degenerative phenotypes, such as complete depigmentation, reduced metabolic rate and complete eye degeneration. The circadian clock is also partially degenerated in this cavefish, and mutations in several photoreceptors and clock-related genes play a role in its lack of response to light. However, a detailed description of the molecular mechanisms underlying the regression of this important mechanism is still missing. Here we investigated the molecular evolution of the non-visual photoreceptor melanopsin Opn4m2, whose premature stop codon accounts for the inability of the peripheral clock to respond to light. To test the hypothesis that other light-related mechanisms are undergoing degeneration, we studied the molecular evolution of the visual pigment rhodopsin, expressed in the brain of *P. andruzzii* and probably involved in its photophobic behavior. The same genes were studied in another blind cavefish, *Garra barremiae* from Oman, a close relative to *P. andruzzii* that independently colonized subterranean waters and evolved troglomorphic traits. Our results based on within and between species analyses, and on time estimation of pseudogenization, show that both genes lost signature of selective constraints in *P. andruzzii*. Conversely, in *G. barreimiae*, that colonized the cave environment more recently and lacks complete isolation from the surface, the genes are still conserved. Our observations indicate that the long and extreme isolation of *P. andruzzii* in darkness led to a general relaxation of natural selection on light-responsive physiological mechanisms.

Population genetics, ecology, and evolution of the troglophilic isopod *Androniscus dentiger* from the Frasassi cave system

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Because of their trophic structure and energy cycles, habitats such as underground caves are usually considered very simple environments, mostly depending on energy input from the surface. With some exceptions, the genetic structure of cave-organism populations is generally poor, as a reflection of the spottily distribution of trophic resources which forces organisms to maximize environmental exploration.

We here investigate the population genetic structure of the troglophilic isopod *Androniscus dentiger* inhabiting the underground system "Grotte di Frasassi", which include a vast and complex underground system (Grotta del Fiume-Grotta del Vento), connected with the surface at many sites. A *continuum* in habitat allows the potential dispersion of cave dwellers throughout the cave. Organic debris or bat guano is spottily available. An important source of food, only available in some areas of the cave is represented by chemoautotrophic bacteria.

The analyzes conducted with STRUCTURE, on a set of eight microsatellite loci, and resulting values of F_{ST} clearly showed a strong differentiation among the populations studied, indicating a generally reduced or in some cases absent gene flow, even among populations of the same cave. The genetic similarity between populations from noncontiguous sites and their differentiation with respect to populations from sites that are geographically close can be correlated with the ecological characteristics of the sites.

Despite the "Grotte di Frasassi" is an open system, not geographically isolated from the surface, the abundance and distribution of food resources are able to determine independent evolutionary paths, through the combined action of stochastic and deterministic factors.

Quantifying Separation and Similarity in a *Saccharomyces cerevisiae* Metapopulation

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Identifying and quantifying the population processes occurring in microbes can help our understanding of their evolutionary histories and potential future trajectories. Microbes perform essential processes vital to the functioning of the biosphere and have wide reaching impacts on global economies due to the roles they play in producing quality agricultural commodities. They are also widely used as model systems to test fundamental hypotheses about biological processes. Despite their undisputed importance to both commerce and science, we have a poor understanding of microbial population biology and ecology. Here we report an in-depth quantitative analysis of population structure and migration in *Saccharomyces cerevisiae* to provide a more detailed account of the population processes occurring in microbes. Over 10 000 individual isolates were collected from native plants, vineyards and spontaneous ferments of fruit from six major regions spanning 1000 km across New Zealand. From these, hundreds of *S. cerevisiae* genotypes were identified and a suite of analytical methods were employed to provide a comprehensive quantitative analysis of both population structure and migration. Within each geographic region no genetic differentiation was detected, even between populations inhabiting native forests and vineyards; however at distances greater than ~100 km, the New Zealand *S. cerevisiae* population exhibits varying degrees of population structure. This is complemented with estimates of bidirectional rates of migration between geographic regions which correlate with the movement of fruit by the New Zealand wine industry. This suggests anthropogenic activities may influence microbial population patterns and diversity and therefore may influence their potential evolutionary trajectories.

Population genomics of *C. melanopterus* using target gene capture data: demographic inferences and conservation perspectives

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Population genomics of non-model organisms can be challenging when a closely related reference genome is not available. Here we applied a recently developed target gene capture approach that circumvents the need for a reference genome to obtain deep sequence information for ~1000 independent autosomal regions for the blacktip reef shark (*Carcharhinus melanopterus*). We used the data collected to explore and contrast estimates of demographic history and population structure in a single deme (SID) from Northern Australia with a scatter sample (SCD) collected from various locations throughout the Indian Ocean. We developed an ABC with recombination algorithm, using the site frequency spectrum computed on unphased data as summary statistic, to explore the genealogical signature of population dynamics detected from both sampling schemes. We then contrasted these results with those obtained by fitting the data to a non-equilibrium finite island model. Both approaches supported a *Nm* value ~40, consistent with philopatry in this species and the moderately to high Fst found using microsatellite markers in previous work. Finally, we tested for signatures of recent bottleneck due to overfishing and human disturbance. We demonstrate through simulation that metapopulations exhibit greater resilience to recent changes in effective size compared to unstructured populations which can obscure detection of recent population bottlenecks. We propose an empirical approach to detect recent bottlenecks which may help to explain why several overfished species do not show a decrease in effective population size. When applied to *C. melanopterus* it suggested that the conservation status of the sampled deme is healthy.

On the role played by the carrying capacity and the ancestral population size during a range expansion

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Range expansions occurred several times in many species. Despite their abundance, many details of their genetic consequences need yet to be fully investigated. We focused on two aspects of range expansion: i) the influence of *N* (effective population size) and *m* (migration rate) for fixed *Nm*; ii) the role of the effective population size of the ancestral deme (*Nanc*). Our simulations suggest that larger *N* increases the length of the *scattering* phase, determining a higher within deme variability (lowering the degree of population differentiation), an excess of rare variants and an increase in number of lineages reaching the ancestral deme. These lineages accumulate mutations according to *Nanc*: for larger *N* this increases the between-population component of molecular variance, contrasting the decrease in population differentiation due to the heterozigosity excess. Finally, we tested whether it is possible to jointly estimate *N* and *m*, and to make inferences on the ancestral population structure of a species undergoing a range expansion. We simulated pseudo-observed data sets under a large range of parameters values, two sampling schemes and up to 1,000 independent loci. By means of approximate Bayesian computation, we could correctly recover both *N* and *m* (but not *Nanc*), provided a large number of demes were sampled. These findings highlight how it will be possible to estimate the dispersal rate in non-equilibrium metapopulations by using population genetics approaches.

The application of 2b-RAD approach for investigating the genomic population structure of yellowfin tuna (*Thunnus albacares*) at the global scale

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Tropical tunas account for the greatest part of world tuna catches but their stock biomass declined ca. 60% from 1954 to 2006, increasing the risk of ecological/economic extinctions. Despite of the relevance of these species, their genetic population structure is not well resolved yet, with several studies leading to discordant evidences. In particular, yellowfin tuna (*Thunnus albacares*, YFT) population structure is still poorly understood and they have been considered to be highly mobile and consist of a single panmictic spawning population for the purposes of stock assessment and management in each ocean. However as suggested by some studies in Indian Ocean there may be distinct population structure at a much smaller scale than has generally been considered for highly migratory species. In order to reveal the YFT genetic population structure tuna at the global scale, in this study a streamlined restriction site-associated DNA (RAD) genotyping method based on sequencing the uniform fragments produced by type IIB restriction endonucleases has been used, discovering and genotyping Single-Nucleotide Polymorphisms (SNPs) across the whole genome of this species. The novel genomic data generated in the present study reveal YFT population structure at a level unattainable through classical latter approaches with significant implication for its stock assessment and management.

Hearing hangul calls for survival: conservation genetics and demographic history of the last surviving population of red deer in India

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The hangul (Cervus elaphus hanglu) is of great conservation concern because it represents the easternmost and only hope for an Asiatic survivor of the red deer species in the Indian subcontinent. Despite the rigorous conservation efforts of the Department of Wildlife Protection in Jammu & Kashmir, the hangul population has experienced a severe decline in numbers and range contraction in the past few decades. The hangul population once abundant in the past has largely become confined to the Dachigam landscape, with a recent population estimate of 218 individuals. We investigated the genetic variability and demographic history of the hangul population and found that it has shown a relatively low diversity estimates when compared to other red deer populations of the world. Neutrality tests, which are used to evaluate demographic effects, did not support population expansion, and the multimodal pattern of mismatch distribution indicated that the hangul population is under demographic equilibrium. Furthermore, the hangul population did not exhibit any signature of bottleneck footprints in the past, and Coalescent Bayesian Skyline plot analysis revealed that the population had not experienced any dramatic changes in the effective population size over the last several thousand years. We observed a strong evidence of sub-structuring in the population, wherein the majority of individuals were assigned to different clusters in Bayesian cluster analysis. Population viability analysis demonstrated insignificant changes in the mean population size, with a positive growth rate projected for the next hundred years. We discuss the phylogenetic status of hangul for the first time among the other red deer subspecies of the world and strongly recommend to upgrade hangul conservation status under IUCN that should be discrete from the other red deer subspecies of the world to draw more conservation attention from national and international bodies.

Symposium # 6

Evolutionary Applications

INVITED SPEAKER

Genetic analysis and real world problems in ecology and forestry

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Genetic invasiveness, control of timber trade, forest seedlot traceability, and other topics are important areas where genetic analyses of biological material, mostly from trees, can inform public administration, forest owners and nature conservation activities. Biological tracing can also be applied to forest pathogens, or to check for any genetically determined resistance or tolerance towards pests and diseases in trees. Forest trees are important key elements of ecosystems that cover the greatest part of Europe. Climate change threatens to change these ecosystems more rapidly than their natural resilience can bear. New biotic interactions can also arise by hybridization with species that have hitherto have inhabited different regions of Europe, or which have been introduced accidently or willingly. Genetic invasiveness was studied in my lab for the *Populus nigra vs. P. deltoides* hybrid system. Timber trade can be supported by analysing material in case of thefts of timber (an example from *Prunus avium* will be shown. A European project, Trees4Future, tries to summarize and unify current approaches to seedlot and plant identification for marketing purposes. Seed orchards are attractive targets for such research because of the limited number of parents present (an example for *Larix decidua* will be demonstrated). Another European project, ProCoGen, tries to find particular regions in conifer genomes that co-vary with landscape features, thus indicating underlying genes that may be involved in adaptation. Taken together, new approaches are arising for to deepen our knowledge on the biological basis of many phenomena, which lead to applications that may become useful for near-to-market activities already in our times.

Genetics survey of European barbel (*Barbus barbus*) populations in England and Wales: implication for future restocking management

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The European barbel, *Barbus barbus*, is indigenous to eastern flowing rivers in England. Due to its popularity as a sport fish for angling, it has also been successfully introduced into many rivers in its non-indigenous range over the last 100 years. Here, a genetic survey of barbel populations in river catchments in England and Wales was completed to derive understandings of population genetic structure in the indigenous range, assess possible routes of natural versus manipulated dispersion in the indigenous and non-indigenous range, and identify whether more recent stocking of hatchery-reared fish has impacted the genetic integrity of indigenous populations. Genetic analyses covered approximately 300 fish from 18 populations across 9 river basins, plus some hatchery-reared fish. Population genetic structure was inferred by genotyping the tetraploid barbel via sequencing of three nuclear paralog genes (Gh_2, S7_1 and S7_2), with levels of population variability and population diversity determined using genetic diversity estimators, including the analysis of molecular variance (AMOVA). Outputs revealed strong population structuring in some indigenous basins, corroborated anecdotal evidence of the source of introduced fish in the non-indigenous range, and revealed that stocking of hatchery reared fish has resulted in some genetic introgression in the indigenous range. These outputs will be used to further develop barbel fishery management activities in England and Wales, including the practice of using hatchery reared fish.

Frogs' microbiome from metal polluted and metal non-polluted sites

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Several microorganisms live in symbiosis with vertebrates. Some of them are pathogenic but most are commensalists, if not beneficial for their host. Some bacteria have been found to produce substances able to protect amphibians from infectious agents. In our work, we aimed to assess influence of metal contamination in the bacterial diversity inhabiting the skin of the Perez's frog, *Pelophylax perezi*. Swabs from male and female frogs' skin were collected at populations inhabiting metal polluted and non-polluted sites (from pyrite mines in Aljustrel and Minas de São Domingos, South-East of Portugal). Members of the Classes Alphaproteobacteria, Betaproteobacteria, Gammaproteobacteria, Actinobacteria and Bacilli were identified in frog's microbiome by cultivation methods. The most abundant Families in the non-polluted site were Comamonodaceae, Microbacteriaceae, Moraxellaceae and Pseudomonodaceae, while in the polluted site were Enterobacteriaceae and Staphylococcaceae. The microbiome differed by the presence or absence of contamination and according to the frogs' gender even within the same population. As expected, the metal contamination may alter the microbial community of organisms living in metal polluted sites. Different bacteria species were identified in the two environments and the diversity of the microbiome was lower in metal-polluted site. However, the strong distinction between males and females suggests that microbial community is not only influenced by the surrounding environment. It may also be shaped by the frog itself, which maybe produces a different micro-habitat probably due to the hormonal conditions differing by gender.

Evolutionary Classification of Diseases and Similar Conditions

Libertini G.

For current medicine, the knowledge of evolutionism is considered useful only for particular diseases. On the contrary, for Evolutionary Medicine it is unthinkable to disregard a complete integration of evolutionism in the core of medicine in order to effectively understand, prevent and treat diseases.

This implies a radical change of the whole medicine, and among other things, an evolutionary classification of diseases and similar conditions.

The "modern" classification of diseases is pre-Darwinian and follows empirical and customary criteria: in short, it is largely based on the organic system affected or on the competence attributed to some specializations. An evolutionary classification is based instead on the primary causes:

I - Diseases deriving from alterations of the genotype;

II - Diseases deriving from alterations of the ecological niche (consequences of mismatches between adaptation and modified lifestyles)

III - Diseases deriving from ‘extremes’ of the ecological niche (trauma; burns, etc.)

IV - Diseases deriving from interactions with other living beings (infections, parasitosis, etc.)

V - Physiologic phenomena that cause troubles and sufferings (V-1 Defences against trauma, infections, toxic substances, etc., i.e. pain, fever, cough, sneezes, nausea, vomit, diarrhoea, etc.; V-2 Mental and behavioural disorders as adaptations to particular situations; V-3 Pregnancy, childbirth, puerperium; V-4 Phenoptotic phenomena, aging excluded; V-5 Aging)

Such a classification implies that the focus is on the primary causes and not on disease manifestations. It follows that Evolutionary Medicine is mainly aimed at preventing disease and considers the cure as the last remedy for cases where prevention has failed.

Importance of the phoretic phase in the reproduction of *Varroa destructor*: ectoparasite of the honey bee (*Apis mellifera*)

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Varroa destructor is an Acarian parasite of the Honeybee that causes thousands of colony losses worldwide. The parasite cycle begins with a phoretic phase and then a reproductive one. During the former, the mite stays on adult bees, mostly on nurses, only to feed on their hemolymph. During the latter, the parasite invades a larva cell and starts its reproduction. Our study tried to elucidate if the phoretic phase had an importance other than providing a cheap way of locomotion toward a reproductive site. We investigated if the type of bees on which *Varroa* stays and the duration of this stay could later influence the reproductive success of the parasite, using an *in vitro* rearing method. For that purpose, key reproductive features such as egg laying rate or presence and number of fully molted daughters were measured. To go further, the expression of two vitellogenin genes, shown to fluctuate throughout the reproduction, was also quantified. Results showed that the phoretic phase impacts neither the reproductive features nor the VdVg levels of expression. However, they allowed to link the reproductive features to the genes expression and showed that daughters expressed the VdVg genes at lower levels than their mother. When focusing on the damages caused on bees, the data indicated that a longer stay on adult bees during the phoretic phase would result in a more frequent induction of deformity in pupae and newborn bees. This study provides new perspectives towards a better understanding of the *Varroa*-honeybee interactions.

Evolution and genomics quickly provide with useful clues for insect pest management: the case of *Drosophila suzukii*

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Present economies are facing the recurrent introduction of pests and parasites from distant area. One notable example is *Drosophila suzukii*, whose recent outbreak in western countries and peculiar ecological behaviour had made it an emerging model for pest management and biology. Here we review recent findings on *D. suzukii* and present new results demonstrating how a genomic overview using NGS is a cost effective and quick way to obtain source of information for applied agricultural management practices. First we show how paleogenomics is a powerful tool to quickly describe the evolutionary history behind a species pest origin by showing that *D. suzukii* is intimately linked with an ecological pre-adaptation to temperate climates. Second, we show how genomics can quickly provide with a detailed description of potential endosymbionts such as *Wolbachia* with field control potentialities. Third, we show how the genome is an effective repository for quickly obtaining genes such as gustatory and light receptors (presented here for the first time) which are key for deciphering and control pest behaviour. Fourth, we show how evolutionary genomics (dN/dS studies) can identify genes under peculiar selection that can point toward peculiar aspects of the pest biology such as enhanced immune response to parasitoids or insecticide resistance. Overall, our results shows that a "light" genome sequencing strategy analysed on an evolutionary framework is a cost effective practice for quickly instruct over the biology of invasive pests and accelerate the definition of its management.

Next-gen monitoring strategies. A warning system for EXPO2015.

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Biological invasions are a major threat to the world's biodiversity with consequences on ecosystem structure and functioning, species evolution, and human well-being (through ecosystem services). We are living in a hyper-connected world, and the spreading of alien species is an (almost) inevitable drawback. Hence, biodiversity needs management, and every good plan of management starts with a high level of knowledge about indigenous biodiversity. Biological knowledge is the stepping-stone from which it will be possible to identify putative threatening alien species.

With the advent of 'omics' era, the combination of DNA-based identification (barcoding) with high-throughput sequencing (HTS) (i.e. metabarcoding) becomes a particularly useful tool where the potential for taxonomic identification of species is limited.

In this research we managed a monitoring campaign during the world exposition of Milan (EXPO2015). Due to the specific theme (Feeding the Planet, Energy for Life) it is expected that during EXPO 2015 the almost 150 participant countries will arrive in Italy with plant species. Even if the participants must follow restrictive procedures to import living beings, it is clear that such a concentration of foreign organisms is an incredible source of potential invasions.

This study gives the parameters to compare the biodiversity present in the air before and during the exposition and could be an appropriate model to test an innovative monitoring system, to achieve an efficient early warning system based on HTS techniques to identify airborne eukaryotic organisms.

Taxonomy and genetic diversity of Anodontine mussels in Lake Maggiore

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In the last years the native freshwater bivalves (Unionoida) communities have dramatically declined in European ecosystems. This decay is principally due to habitat destruction, loss of host fishes and, not least, to the establishment of non-native species. Accidental or intentional introductions of invasive alien species (IAS) can drive ecological changes, threatening ecosystem integrity and native biodiversity. The conservation of freshwater biodiversity represents a great challenge for the future, especially for those environments where a co-existence between native and IAS is well documented. In Lake Maggiore (Italy) the native Anodontine mussel species (*Anodonta anantina* and *Anodonta cygnea)* share the habitat with the recently introduced alien Chinese pond mussel *Sinanodonta woodiana*. The morphological observations have revealed a high phenotypic plasticity in the shell shape challenging the discrimination between native and non-native mussels. The taxonomy and the genetic diversity patterns of the Lake Maggiore mussels will be inferred respectively through the application of molecular tools, including mtDNA sequencing and nuclear SSR genotyping. The results will be discussed in order to plan an efficient conservation strategy.

Symposium # 7

New Frontiers in Evolutionary Anthropology

INVITED SPEAKER

How we settled the world and developed our culture.

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One of the most fundamental problems in the history of mankind is to understand the processes that created the genetic and cultural diversity among humans today. Despite more than a century of research in archaeology and anthropology, major questions remain unanswered or heavily debated. The reason is our inability to link human populations and their cultures across time. Attempts to close this gap were made through genetic studies. However, studies of living peoples infer the past from present diversity and distributions, both of which have been shaped by a history of colonisation, admixture and, more recently, globalisation. Ancient DNA studies provide direct information about past human diversity. Until recently, however, the field suffered from problems of contamination and scope - restricted to mitochondrial DNA, a poor marker for testing complex demographic scenarios. This changed in 2010, when my research group published the first ancient human genome. The results dismissed the commonly held belief at the time that it is not possible to obtain reliable nuclear DNA sequences from modern human remains due to contamination. We also showed the power of ancient genomics by revealing a hitherto unknown migration from Siberia into the Americas, independent of those that gave rise to today's indigenous peoples in the New World – information not retrievable from modern genetic data. Since then, we have used ancient genomics to settle some of the most important controversies regarding the origins and dispersals of our species, and the cultural changes that followed. For example, we have shown that the ancestors of Aboriginal Australians were the first known human population to settle the World beyond Africa, 30-40 thousand years before the ancestors of Europeans and Asians settled in Eurasia. Since then, we have also shown that the distribution of ancestral Europeans and Asians was very different in the past, and that the genetic geography of humans today is very shallow, in most cases less than 4000 years old. One of the main outcomes of these studies is the realisation of the previously unknown amazing complexity of human evolutionary history – for example, Native Americans, thought to descend from a group of East Asians who crossed the Bering Strait 13-10 thousand years ago, trace ca. 1/3 of their genome to a population that also gave rise to present- day Europeans, but not to East Asians. Another is that the relationship between cultural and genetic change reflects multidimensional processes that create a web of outcomes at different geographic scales – a migration may disperse genes and ideas, but the ideas may travel further, creating a much wider pattern of cultural change than reflected in the genetic make-up of people; equally, important shifts in cultural parameters can occur in the absence of demographic change. Thus, ancient genomics allows testing the nature of cultural change in the past, one of the most longstanding problems in Archaeology. We have also used ancient DNA to explore the extent to which early modern man affected the environment. We have shown that, although early man impacted his surroundings through hunting, the mass extinctions of big mammals at the end of the Ice Age - a topic of extensive debate for more than a century - was mainly driven by climate and vegetation changes rather than human overkill, thereby challenging the idea of an early rise of the Anthropocene.

Discovering past gut microbiomes through NGS analysis: the mummies of Roccapelago (MO)

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The gut microbiota plays a primary role in metabolism and immune response of host organisms. It is influenced by lifestyle, diet and environment but little is known about its ancestral state and evolution, especially in relation to past human dietary shifts, the impact of industrialization and the introduction of antibiotics. In the reconstruction of the ancestral state of gut microbiota, recent pioneering studies have focused on contemporary rural populations or ancient coprolites. This paper presents the first screening of microbiota by NGS technology from ancient human gut tissues. We collected samples from ten Roccapelago mummies (16th-18th c. AD) from different anatomical tissues (gut, skin, muscle). In addition, samples of soil and clothes were analyzed to detect environmental contamination. Paleodiet reconstruction by stable isotopes analysis ($\delta^{13}C$ and $\delta^{15}N$) was performed on femurs and teeth. Moreover, information about diet, lifestyle and health were recovered from parish registers and historical documents.

DNA was extracted with a silica-based protocol and amplicons of 16S rRNA gene (V3 region) were sequenced on 454/Roche obtaining an average of 10.059 reads per sample. The bioinformatic analysis using the MICCA pipeline reveals a consistent diversification between tissues (gut vs muscle vs skin), with an enrichment of Actinobacteria in gut samples (18%) in respect to muscle and skin samples, despite Clostridiales are represented in all tissues analyzed. Interestingly, halophilic Gram-positive bacteria from Staphylococcaceae (6%) are found in gut samples. Investigation of a potential physiological basis of this association may lead to new insights into the reconstruction of past gut microbiomes.

The significance of microfossils recovered from dental calculus for anthropological analysis

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Trace amounts of food and environmental micro-debris such as starch grains, phytoliths and fibers are preserved, together with organic components such as lipids and proteins, within dental calculus. The recovery and identification of these elements appear fundamental for the reconstruction of the palaeodiet, lifestyle and paleoenvironment of ancient populations. This is a relatively new line of investigation. Therefore, it needs specific efforts in order to define new protocols of analysis: in particular, chemical procedures to wash and dissolve the samples in order to recover the greatest variety of microfossils, or the improvement of atlas of images for the identification of plants' micro remains and other biological materials. Our work followed two steps. Step one focused on the study of several ancient cereals, since they are the plant organ with the highest starch content (about 70%) and one of the major sources of carbohydrates of the human diet. Therefore, we analysed different dental calculi sampled from skeletal remains recovered at the Necropolis of Can Reiners (7th century AD), in *Pollentia* (Palma of Mallorca, Spain), the most important Roman city in the Balearic Islands. This research shows that, within the sampled dental calculi, there are discrete concentrations of starch grains, fibers and other botanical remains, such as spores and pollen grains. This is the first insight into the study of diet through calculus analysis in this population. The identification of these elements allows us to better understand the diet and other human behaviors of the late roman community of Can Reiners.

Identification of missing children: a quantitative approach to the analysis of facial morphology

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The problem concerning missing children is a very sensitive issue from a social point of view, often embraced with different fields of criminality, since the organ trafficking to minor prostitution and illegal adoption. Since in June 2014 29763 persons were still missing, among whom more than one half were minors[1]: the relevant social impact of this phenomenon is therefore clear, which brings about several limits to identification both from a technical and scientific point of view. Two main problems affect the identification of missing children: the limited chances of recognizing a growing face by comparison with photos taken sometimes years before, and the need for quantifying the judgment of positive identification.

This study aims at developing and testing a method of comparison between images of minors, based on stable facial markers (in detail, nevi were used). The project took into consideration 143 photos of the same face performed at different ages by 41 subjects. A sample of 110 images from 16 individuals with facial nevi were selected, and then analysed by software MATLAB® R2014b. Ten facial landmarks were used in order to predict variables concerning the orientation of face and growth. Distance between the nevus and the predicted area was calculated and quantified by a score between 0 (highest distance between the position of nevus and the predicted area) and 1 (highest concordance between the two areas). In total 68 comparisons were performed (27 between photos of the same subject at different ages, 22 between photos of the same subject at the same age and 19 between photos of different subjects at different ages).

Results show that comparison of photos of the same subject provide score was equal or higher than 0.2, whereas in cases of different persons the score was always equal or lower than 0.3.

The study shows an attempt at quantifying the judgment concerning identification based on photos of minors in growth, applicable to every stable individualizing marker of face: further studies are needed in order to test the method and reduce the errors.

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[1] Ministero dell'Interno, Commissario Straordinario per le Persone Scomparse, Relazione 2014

DNA methylation variability along the Italian peninsula: an evolutionary perspective reveals implications for human health and diseases

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DNA methylation is a molecular mechanism useful for the description of human variability among different populations. This mechanism constitutes an important reservoir of variability useful for adaptation in response to new stimuli and recent studies have demonstrated that selective pressures are able to shape not only the genetic code, but also DNA methylation profiles. Here we depicted the natural variation that occurs in DNA methylation considering individuals who born and lived in different areas of Italy (*i.e.* North, Centre and South). The Italian population is indeed very diverse in terms of culture, traditions, history and genetic background and constitutes a suitable model to investigate mechanisms of adaptation mediated by epigenetic variations. A whole-genome DNA methylation analysis identified an enrichment of differentially methylated regions (DMRs) located in genes involved in nitrogen compound metabolic process and transport. Reactive nitrogen is object of research for environmental issues. Moreover, considering genes that included a DMR, we observed an enrichment of genes involved in response to pathogens, indicating that pathogens diversity across the Italian peninsula constitutes a selective pressure able to shape epigenetic profiles.

In conclusion, our results indicate that DNA methylation changes could represent both a consequence of the genetic architecture, but also a more complex mechanism of adaptation of organisms.

Testing for the influence of lifestyle on genetic diversity through time

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Cultural factors, such as marriage rules, social stratification, and cultural transmission of fitness, have the potential to influence the pattern of genetic diversity in human populations. Neolithisation, i.e. the transition from a foraging to a food producing lifestyle, entailed many changes in such factors (e.g. sedentarisation, bigger population size, etc.), but its influence on genetic diversity has until now been tested only in restricted geographic areas (e.g. Central Africa).

Here we take advantage of the large amounts of whole-genome SNP data that have become available in recent years to investigate the effect of Neolithisation throughout the Old World. Specifically, we collated data for over 40 populations across Africa and Eurasia, and used the patterns of Linkage Disequilibrium (LD) to reconstruct the effective population size (Ne) through time.

We found that, within each region considered, starting from the Neolithic transition foraging populations have significantly lower Ne than neighbouring food-producers. Using estimates of net primary productivity reconstructed from palaeoclimatic models, we confirmed that such differences are due to the Neolithisation process, rather than simple changes in the availability of environmental resources.

Early modern human dispersal from Africa: genomic evidence for multiple waves of migration

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It is unclear whether early modern humans left Africa through a major migration process, dispersing simultaneously over Asia and Europe, or in two main waves, first through the Arab peninsula into Southern Asia, and later through the Near East into Western Asia and Europe.

We collected a broad genome-wide SNP dataset in 71 populations to test if single (SD) or multiple (MD) dispersal model can better account for patterns of genome diversity. We found good correlations between geographic and genetic distances, but only insignificant differences between models.

We moved to consider the patterns of Linkage Disequilibrium in each population to estimate effective population size that allows us to assess when population pairs diverged in time. We showed that accurate genomic estimates of the divergence times between European and African populations are more recent than those between Australo-Melanesia and Africa, and incompatible with the effects of a single dispersal. Furthermore, we showed that this difference cannot possibly be accounted for by the effects of hybridization with archaic human forms in Australo-Melanesia. Simulated genetic polymorphism data demonstrate the validity of our conclusions.

We conclude that the hypothesis of a SD model from Africa appears hardly compatible with the observed historical and geographical patterns of genome diversity, since some populations retained the signal of an earlier dispersal.

SIBE Lecture

Every evolutionary step is an addition of information to an already existing system

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Università di Siena.

E' questo il punto di arrivo della speculazione teorica raggiunta da uno dei più estrosi biologi del secolo scorso, Gregory Bateson (*Mind and Nature*, 1979), che viene a trovarsi al centro del dibattito sulla macroevoluzione. La tesi di Bateson era allora in accordo con la nozione che il DNA è il substrato universale della memoria genetica dei viventi; era anche in accordo, per chi ci credeva, con il Dogma Centrale della Biologia enunciato da Francis Crick.

Eppure, questa affermazione non convinceva tutti, anche se quadrava molto bene con l'informatica teorica, poiché non si trovava alcun riscontro presso le più semplici osservazioni e misurazioni del contenuto di DNA di molti organismi vegetali e animali. In quegli anni vennero anche progettati e messi in vendita complicati accessori per microscopi che permettevano di dosare agevolmente il DNA contenuto nei nuclei.

D'altronde in quegli anni si accertava che l'insorgenza della poliploidia, che moltiplicava bruscamente il contenuto in nucleotidi di un ceppo, modificava poco e in modo prevedibile l'aspetto, il comportamento e il ruolo ecologico del ceppo stesso. Non solo, la poliploidia per certi aspetti inibiva la manifestazione di mutazioni geniche.

Come spesso succede quando risultati di osservazione e teoria si ostinano a non coincidere, il problema è stato accantonato.

Tuttavia in questo momento in cui la genetica molecolare e la cariologia hanno compiuto grandi progressi, e l'attenzione dei biologi si rivolge sempre più spesso alle cosiddette "macromutazioni", diventa urgente comprendere perché si verifichi questo singolare divario tra teoria e osservazione.

Posters

P. 01

The complete female- and male-transmitted mitochondrial genome of *Meretrix lamarckii* (Bivalvia: Veneridae)

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Doubly Uniparental Inheritance (DUI) is a peculiar inheritance phenomenon that allows some bivalves to maintain two separate sex-linked mitochondrial genomes into the same population genetic pool. Many mitochondrial features, such as gene extensions or additional Open Reading Frames (ORFs), have been proposed to be related to DUI mechanism but the mechanism is still far from being understood. DUI distribution is unclear, several species show this unusual way of organelle inheritance but, except for Mytilidae and Unionidae families, DUI species are scattered across the bivalve phylogenetic tree. Therefore, we sequenced and characterized the complete F and M mitochondrial genomes of a non-mytilid/non-unionid species, *Meretrix lamarckii* (Heterodonta: Veneridae).

The two mitochondrial genomes are comparable in length and show roughly the same gene content and order. The two genomes show a significant diversity, up to 16%. Similarly to other DUI species, a male *cox2* extension has been found but, up to now, it was not possible to assign a specific function to this insertion. Additional ORFs have been revealed in both F and M Long Unassigned Region (LUR). Preliminary study did not reveal their functionality; however, supernumerary ORFs and the *cox2* extension seem to be the rule rather than the exception within DUI genomes: even if no direct involvement in DUI process has been demonstrated yet, the finding of these features also in *Meretrix lamarckii* strengthens this hypothesis.

P.02

The tangled triangle among ORFans, mitochondrial inheritance, and sex determination: mtDNA evolution at the root of Paleoheterodonta (Bivalvia)

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In freshwater mussels (Bivalvia: Paleoheterodonta: Unionoidea), gonochorism is strictly linked to doubly uniparental inheritance (DUI) of mitochondrial genome (mtDNA), a system where two mtDNAs, named F and M, are respectively inherited maternally or paternally. Both mtDNAs possess lineage-specific ORFans (open reading frames without detectable homologies), named fORF and mORF. Hermaphroditism coupled to DUI loss is a derived trait in this superfamily: hermaphrodites carry only a F-derived type of mtDNA, named H, containing a degenerated fORF (hORF). This suggested that mtDNA ORFans play roles in maintaining gonochorism and DUI in Unionoidea. To investigate evolution of mtDNA and DUI at the root of Paleoheterodonta, we sequenced and analyzed mtDNAs of four basal Unionoida from different families and with different reproductive modes (two gonochoric DUI species from Hyriidae and Margaritiferidae; two non-DUI species from Iridiniidae and Mycetopodidae, respectively gonochoric and hermaphrodite), plus the F mtDNA of the DUI Trigoniida *Neotrigonia margaritacea*. Phylogenetic analyses and gene content characterization open new questions on the involvement of ORFans in mitochondrial inheritance and sex determination in Unionoida. The mtDNAs from the non-DUI Iridiniidae and Mycetopodidae are sister to all Unionoidea F and H mtDNAs but do not possess any fORF or hORF. These first-time observations suggest that DUI can be lost in unionids without necessarily maintaining degenerated ORFans and reverting to hermaphroditism. *N. margaritacea* F mtDNA is sister to all Unionoida mtDNAs, and its putative ORFans lack any similarity to known ones: its position and gene organization cast new shadows on the origin of DUI in bivalves.

P.03

A study of Sex-Bias' effects on evolution and sex determination

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Males and Females usually differ for phenotypical features and this sexual dimorphism is largely caused by differential gene expression level. We are doing an *in silico* analysis using data from online databases (e.g. Ensembl; SEBIDA 3.0) and published articles in order to better understand the well-known relation between *Drosophila melanogaster*'s sex-biased genes and the generation of new duplication and speciation events throughout the genome. Using bioinformatics tools we are studying the presence of sex-biased gene's homologs across animals with different sex determination mechanisms, such as the ZWO or the Haplodiploidy. We are investigating and trying to verify if the presence of more paralogs in male-biased genes within the species is correlated to a larger number of homologous genes in other clades. In conclusion, our work tries to highlight the role of sex-biased genes on the evolution of new genes and observe their effect in taxa that use different methods of sex determination.

P. 04

Endogenized mitochondrial gene expressed in the germ line

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Germ line is the cell lineage that transmits genes from generation to generation in sexually reproducing animals. Mitochondria, among many other factors, are responsible of the specification of primordial germ cells (PGCs), and their inheritance dynamics are relevant to understand this process. We studied these issues in the Manila clam *Ruditapes philippinarum*, a species with Doubly Uniparental Inheritance (DUI) of mitochondria, in which a novel protein (RPHM21) is encoded by the male-transmitted mtDNA and expressed in spermatozoa. RPHM21 was predicted *in silico* to be of viral origin – namely an endogenized residual of an ancestral infection. We analyzed mitochondrial transcription and expression patterns using qPCR and antibodies targeting the germ line marker VASPH (VASA homolog) and RPHM21. qPCR allowed to discriminate the phases in which *vasph* begins to be transcribed, and VASPH immunolabelling allowed to identify PGCs. Both targets were localized in the PGCs of males, but while VASPH was detected in all PGCs, RPHM21 appeared to be expressed only in a subpopulation. In *R. philippinarum* we identified a source of germ cell precursors that migrate from the gut and reach the presumptive gonad localization, where they rebuild the gonad at every reproductive season. Since RPHM21 might have a role in activation, proliferation, and migration of male PGCs, we propose that the germ cells expressing RPHM21 could gain advantage over others during spermatogenesis. The investigation of how co-option of foreign elements modifies existing biological pathways is fundamental to assess the impact of such events on the evolution of new developmental features.

P. 05

Comparison of transposable elements dynamics among taxa with different rates of speciation

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According to many authors bursts of activity of transposable elements may be related with speciation events in animals and plants by facilitating reproductive isolation and increasing speciation rate (Senerchia 2015, Jurka 2011, Rebollo and Vieirà 2010, Oliver and Greene 2010, Zhe et al 2009).

In the present work we tested this hypothesis in Mammal and Bird groups with different speciation rates (40 species).

In particular, we performed large scale analyses aimed to evaluate the total number of recently inserted Transposable elements (TE). In order to estimate insertion density, observed values were normalized to genome size (insertion density). Within *Mammalia,* we compared pairs of species belonging to different families of the same order.

Within the class Mammalia we compared pairs of species belonging to different families of the same order.

In most cases (16 out of 19), we observed a higher density of insertion in taxa with higher speciation rate. Accordingly, we confirm that insertion density in Mammals is positively related to the speciation rate. (Wilcoxon test, $p<.01$).

Within *Aves* we compared three species from Galliformes with tree species from Passeriformes, respectively the orders with the lowest and highest speciation rate.

Results showed that the TE density in these two taxa does not show significant differences, suggesting that TE activity did not affect speciation in *Aves* as much as in *Mammalia*. Hence, the tested hypothesis is not confirmed in this case. Alternative explanations and implications are discussed.

P. 06

First case of R2 non-LTR retrotransposon horizontal transfer: data from *Bacillus* stick-insects genome (Insecta Phasmida)

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Horizontal transfer is the passage of genetic material between reproductively isolated, even distantly related species. R2 is one of the best-studied non-LTR retrotransposons and inserts sequence-specifically into 28S rDNA. Non-LTR retrotransposons have been maintained in diverse taxonomic lineages and extensive studies carried out on the site-specific non-LTR retrotransposons R1 and R2 had shown that they have been vertically transmitted for long periods of time. Evidences of horizontal transfer emerge when the TEs and host species phylogenies are not congruent and TEs are more similar than expected on the basis of host divergence. We isolated and sequenced eight R2 elements from the stick-insects species *Bacillus rossius*, *Bacillus grandii* and *Bacillus atticus*. A Maximum Likelihood tree on all R2 elements compared with the phylogeny of the host species showed that all stick-insects R2s, except one isolated from *Bacillus grandii grandii* genome, grouped in a monophyletic cluster within the D clade. Here, *Bacillus rossius*, *Bacillus grandii maretimi* and *Bacillus grandii benazzii* elements exhibit an unusually low divergence. The age *versus* divergence analysis statistics confirmed the possibility of horizontal transfer between *Bacillus rossius* and the two *Bacillus grandii* subspecies. This is the first evidence of horizontal transfer for elements of the R2 clade and the past history of interspecific hybridisation known for these stick-insects would likely explain these data.

P. 07

Successful sequencing of Huntington’s disease CAG microsatellite orthologs from museum collection specimens

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Huntington's Disease is a human inherited neurodegenerative disorder caused by the expansion of a CAG repeat microsatellite in the 5’ end of the Huntingtin (*Htt*) gene, leading to neuronal loss in brain and motor and neuropsychiatric disturbances. To obtain information on *Htt* orthologous genes, a potential source of genomic DNA is represented by museum collections. Here, we aimed at the characterization of orthologous sequences of the human microsatellite assessing 114 specimens from Museo di Storia Naturale di Milano, Museo di Storia Naturale dell’Università di Pavia and Acquario Civico di Milano. Samples belonged to four vertebrate groups: mammals (n=39), amphibians (n=1), reptiles (n=52) and fishes (n=21). Small fragments of tissues stored frozen (n=52), preserved in ethanol (n=52), and tissues that had undergone a tanning process (n=10) were used. Specimens ranged from recently collected to over 50-years-old. Genomic DNA was extracted with either of two commercial kits (Macherey-Nagel, Zymo Research), and its quality assessed by ultraviolet-visible spectrophotometry and gel electrophoresis. DNA extraction yielded fragments of highly variable length, probably depending on initial sample preservation conditions. Specific primers and targeting the most conserved regions of the different templates were used to amplify the sequences of interest encompassing the CAG tract. Amplicon length ranged from 100 to 250 base pairs. 62% of the PCR reactions performed were successful and 38,5% of the relative products were correctly sequenced. This study demonstrates that museum collections are a valuable resource of genomic material to amplify triplet repeat *loci* and restates the importance of specimens conservation for scientific research.

P. 08

Understanding the rise of modern Elasmobranchs and predator-prey interactions in ancient coral reef food web: an example of research in Scientific Museums

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A joint research project involving University-based, scientific museums in Bologna and Verona (Italy) is shading new light on the evolution of elasmobranchs during the Eocene. Specimens from the Pesciara di Bolca fossil site housed in the G. Capellini Museum include earliest representatives of the present day school shark *Galeorhinus* and Myliobatiformes genera *Platyrhina* and *Dasyatis*. Extraordinary preservation allowed to discriminate outer morphology and soft tissues, including the brain, muscles, scales, stomach contents, and egg cases. The systematic use of UV light was essential to distinguish preserved tissues in the fossil specimen, so were comparisons with fossil specimens housed in Verona and Padova. Identifications were corroborated by a morphology study on selected living Elasmobranchs from the Comparative Anatomy Museum and via ultrastructural analyses (SEM) of both fossil and actual soft tissues samples. Data indicate that the *Galeorhinus* specimen was a juvenile (approximately 5 years old), male individual: analysis of stomach contents document the earliest evidence of predator-prey relationship with the *Sphyraena bolcensis* (barracuda). Similarly, detailed analyses of Bolca rays indicate the co-occurrence in the paleo-Mediterranean region of the two genera *Dasyatis* and *Platyrhina*, the latter now geographically confined to the coasts of eastern Asia. The *Dasyatis* specimen is a female bearing 5 to 6 egg cases, whereas the *Platyrhina* is a male as indicated by well-developed claspers. Both taxa display anatomical elements shared by several extant Myliobatiformes and suggest that the Bolca fauna is pivotal to our understanding of adaptive radiation and anatomical innovation in the early coral reef fish communities.

P. 09

Across the shifting baselines - measuring change in biodiversity in the sea benthos of the Kattegat across 70 years and different sampling technology

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Studies of historical changes in ecosystem diversity are essential for developing realistic conservation targets and to understand the long term change of community as a system. Museums represent fundamental sources of information on the past, but their information needs to be carefully handled before to be used. In particular two types of challenge need to be addressed: taxonomic nomenclature needs to be normalized across sampling time; estimates of abundances are not comparable when sampling effort is variable.

This study compare the observations taken from two series of oceanographic cruise performed in the period 1921-1938 and in 2003-2009 in the Kattegat, both of which have stored sample and documents in the Goteborg Natural History Museum. Both cruises use as sampling the dredging of the bottom of the sea (ensuring similar sampling bias but not effort).

The localities and taxonomic order were filtered to ensure comparable precision of taxon assignment and localities, leaving 4412 observations over 54 localities. The taxonomic names normalization was done with Data Refinement Workflow available on BioVeL portal (portal.biovel.eu) using as reference taxonomy the GBIF taxonomic backbone accessed remotely.

The two cruises were compared in term of total species richness, beta diversity (using prevalence across locality as proxy for abundance), and comparing alternative general additive model to explain distribution of species richness across space and time. The results shown an overall great decrease in species (disappearance of 52.8% old community), with a few rare species becoming prevalent with no intervention of alien, while geographical structure is fading.

P. 10

Between Scylla and Charybdis: differential genetic patterns in Eastern and Western Mediterranean *Patella caerulea* populations

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Historical environmental events have shaped the contemporary spatial distribution of marine taxa. The analysis of the genetic diversity and structure of long-dispersal intertidal species can provide information on past and present connectivity patterns. In the present work, the genetic structure of the common Mediterranean intertidal limpet *Patella caerulea* was studied along most of its distribution range, using two mitochondrial markers and five specific microsatellite loci. The main results show a consistent structure according to all 3 markers used. Different genetic patterns in eastern and western Mediterranean were observed: in the Adriatic Sea, through the Ionian, up to northern Aegean and the far east, an isolation by distance pattern was observed; western Mediterranean populations are characterized by a weak and unclear pattern of genetic structure. The shift in pattern of genetic structuring between eastern and western Mediterranean populations is clearly located in a well-defined region between Sicily and the Italian peninsula. Our results, give evidence of the complex interplay between past and present day processes, as the geographical features, main drivers of structure during glaciation events, are modified by present day processes (eg. current regime, hydrology). The Messina Strait (between Scylla-Charybdis) acted as the major long-term barrier to gene flow between the two Mediterranean basins. Within each basin contrasting connectivity patterns were observed, probably related to nowadays gene flow and larval dispersal. The differential evolutionary histories found provide a solid background for future phylogeographical studies in Mediterranean species.

Bifidobacterium spp. from non human primates: a source of novel species

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Co-evolution between host and gut microbiota is a current hot topic: gut microbe relationships are influenced by host genotype, and gut microbial population differs according to host phylogeny. For example gut microbiota within Great Apes reflects the phylogenetic history of their hosts and hypotheses that close host-species are functionally interchangeable were suggested. The bifidobacterial diversity in several subjects of primates has been studied to better understand the co-evolution with host along the evolutionary time-scale. In Great Apes the human species *Bifidobacterium adolescentis* was found in *Pongo pygmaeus*, while *B. angulatum* and B. *dentium* were found in *Pan troglodytes*. The new species *B. moukalabense* was described in *Gorilla gorilla*. Ten subjects selected from the two divergent lineages of *Strepsirrhini* (Lemuridae) and *Simiiformes* (Callitrichidae) were studied in the present work. Interestingly in *Callithrix* spp. and in *Saguinus* spp., together with the five previously described species *B. callitrichos, B. stellenboschense, B. reuteri, B. saguini* and *B. biavati,* seventeen new bifidobacterial species were found, showing a strong association with host species. In particular in two adult subjects of *Lemur catta* and *Eulemur macaco* (Lemuridae) two novel species were found: *B. lemurum* and a new one. *B. aesculapii* and five additional novel species were isolated from 5 baby subjects of the New World Monkey *Callithrix jacchus*. Nine novel *Bifidobacterium* spp., together with three bifidobacterial species previously found in both *Saguinus midas* and *Callithrix jacchus,* were found in an adult subject of *Saguinus oedipus* (Callitrichidae).

P. 12

Phylogeography and colony structure of dry-wood dwelling termites of the genus *Kalotermes* (Isoptera, Kalotermitidae) in Southern Europe

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According to the most recent phylogenetic and phylogeographic investigations, the *Kalotermes* genus (Kalotermitidae) includes three different mitochondrial lineages of *K. flavicollis* and the new *K. italicus* species. In some Italian populations extreme colony fusion events were observed, with mixed families composed by up to nine maternal lines belonging to different genetic lineages or even to different species. In termites with the same nesting type, colony fusions lead to the death of queens and kings allowing false workers (pseudoergates) to evolve into reproducers and to inherit the colony (Accelerated Nest Inheritance). To widen the analyses on *Kalotermes* phylogeography and colony structure, we sequenced 912 bp of the mitochondrial genome, corresponding to COI/tRNA-Leu/COII, in two individuals from each of the 21 colonies collected in 11 European localities. In four colonies showing haplotypes of different genetic lineages/species, five-eight individuals per colony were further analyzed. A more detailed biogeographic scenario is obtained, with overlapping distribution areas of the considered lineages/species. Moreover, data confirm that mixed colonies with haplotypes of different genetic lineages/species may show more than two maternal lines. On the whole, colony fusions appear to occur more frequently than expected on the basis of eusociality models. Moreover, the possibility of hybridization between different genetic lineages or species could facilitate the fusion of more than two colonies wiping out mechanisms of nest-mate recognition. This analysis, therefore, demonstrates that dry-wood dwelling termites might be an interesting framework to study the evolution and maintenance of eusociality.

P. 13

Predictive dating of the *Plum pox virus* evolution indicates a recent appearance of main recombinant strains

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Plum pox virus (PPV) has a polycistronic RNA genome (*ca*. 10kb) and is known to have several strains affecting *Prunus* spp. with a different pathological behaviour. Some strains derive from a few successful recombination events around conserved sites. The recent isolation of a new, unrecombined strain from Eastern Albania (AL11pl), recognized as the putative parental of an array of strains sharing the 3' terminal genomic portion (from 1300nt up 3' end), prompted the investigation to date recombination events. Seven short regions (about 500nt), some flanking the 3 main recombination spots across 42 full-length genomes of different isolates reported in databases, were analysed by Bayesian inference through Monte Carlo Markov Chain iterations (BEAST 2.1.3 package).

Evolutionary model was determined with PartitionFinder. Rates estimation was calibrated on sampling time, which span across 20 years. To test that sampling time differential was sufficient to perform rate estimation, we implemented a time tips permutation test, where expectation is that observed tips time configuration produces rate estimates different from permutated ones.

The 7 analysed regions showed phylogenetic coherence and a homogeneous rate (5×10^{-4} subs/ site/ year). Dates at nodes separating the recombined strains from both the putative parental range from 120 to less than 30 years from present. This predictive scenario is moreover supported by the chronological evidence that PPV-M, one of the recombinant strains having a profitable fitness among those derived from AL11pl, started to be detected in middle '70s.

Definition of three distinct taxa within the *Barbus* species complex

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The genus *Barbus* Cuvier & Cloquet 1816 is still debated for what concerns its sytematics. A recent origin (about 30 millions of years ago), high levels of hybridization and many impeached identifications scattered across public databases hamper a correct resolution of this taxon. Conversely, at different hierarchical levels, most entities within this genus would deserve a better defined status, especially because of conservation issues. Many different classifications have been proposed, and, while a widely accepted one is still lacking, different authors assign different statuses to the same fishes, leading to abrupt and unwarranted changes in conservation policies.

In the present work, we collected a wide sample of *Barbus* specimens across the whole continental Italy and we used two mitochondrial markers (*cytb* and d-loop) to (i) unravel phylogenetic relationships and (ii) measure genetic distances, in the largest effort to date for this genus. As expected, a geographical clustering was retrieved, and Italian mainland may be subdivided into three distinct patches. Furthermore, our results clearly indicate the presence of three entities of the same level within the Italian *Barbus* biodiversity.

Olfactory responses in *Drosophila suzukii* and *Drosophila melanogaster*: A comparative study

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While *Drosophila melanogaster* feeds and oviposits on fermented fruit, the invasive species *Drosophila suzukii* feeds on fermented fruit but attacks ripening fruit for oviposition, thus damaging fruits. Since the choice for oviposition sites is driven by olfactory cues, it is of primary interest from an evolutionary point of view, as well as for pest control, understanding the olfactory differences between these species. An hypothesis has been advanced that *D. suzukii* is more attracted to volatiles associated with earlier stages of fruit maturation. We test this hypothesis, and the olfactory differences between the two investigated species, through electrophysiological and behavioural measures. Electroantennographic records show that responsiveness to volatiles associated with earlier stages of maturation is less intense for both species and it is enhanced for fresh fruit and fermented volatiles. Preliminary data show that, although perceptual responses are comparable across the two species, behavioural preferences can vary to a greater extent.

Genome sequencing galore: what, how and why

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The advent of affordable high-throughput Next-Generation Sequencing (NGS) technologies has revolutionised all fields of biology. Numerous technologies, such as Illumina, Roche 454, Ion torrent, and SOLiD are available to generate massive NGS data and are accompanied by a growing set of tools for downstream analyses. Because NGS data are relatively cheap, it is also possible to sequence genomic DNA and transcribed genes (RNA) for non-model species of evolutionary, ecological, agricultural or epidemiological importance, for which very little or no prior genetic information is available. More generally, NGS is becoming a cost effective practice for a variety of non-strictly genomic laboratories. For such data to be useful, however, attention should be paid to the full experimental design approach, from sample choice to material preparation, from sequencing technology to assembly strategy. Here we provide examples of approaches and of bioinformatics tools that we used to obtain (after a small economical investment) good-quality genome data useful to investigate the ecology and evolution of various insect species, including *Drosophila subpulchrella*, *Cydia pomonella* and *Cacopsylla melanoneura*. For instance, we show how it is possible to use assembly tools that account for heterozygosity in the sample, thus obtaining draft genomes even when starting from outbred lines. Furthermore, it is now possible to reduce the impact of heterozygous sequences by extracting genomic DNA from single insects and preparing ad-hoc libraries. These examples illustrate that, although generating NGS data is now straightforward, much effort should be given to experimental design in order to produce good-quality and useful data.

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Using ABC and microsatellite data to detect multiple introductions of invasive species from a single source

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The introduction of invasive species to new locations (that is, biological invasions) can have major impact on biodiversity, agriculture and public health. As such, determining the routes and modality of introductions with genetic data has become a fundamental goal in molecular ecology. To assist with this goal, new statistical methods and frameworks have been developed, such as approximate Bayesian computation (ABC) for inferring invasion history. Here, we present a model of invasion accounting for multiple introductions from a single source (MISS), a heretofore largely unexplored model. We simulate microsatellite data to evaluate the power of ABC to distinguish between single and multiple introductions from the same source, under a range of demographic parameters. We also apply ABC to microsatellite data from three invasions of bumblebee in New Zealand. In addition, we assess the performance of several methods of summary statistics selection. Our simulated results suggested good ability to distinguish between one- and two-wave models over much but not all of the parameter space tested, independent of summary statistics used. Globally, parameter estimation was good except for bottleneck timing. For one of the bumblebee species, we clearly rejected the MISS model, while for the other two we found inconclusive results. Since a second wave may provide genetic reinforcement to initial colonists, help relieve inbreeding among founders, or increase the hazard of the invasion, its detection may be crucial for managing invasions; we suggest that the MISS model could be considered as a potential model in future theoretical and empirical studies of invasions.

The process of domestication in common bean

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Several genomic tools from molecular markers to Next Generation Sequencing (NGS) data were used to deeply investigate the evolution of common bean. Our findings revealed a Mesoamerican origin of the species with subsequent diverse migrations into South America. Common bean domestication occurred independently in Mesoamerica and Andes. A greater reduction in genetic diversity in Mesoamerica as to the Andes was found, indicating that a bottleneck before domestication occurred in the Andes, which strongly impoverished this wild germplasm, leading to the minor effect of the subsequent domestication bottleneck. Analysis of ~190,000 SNPs (Single Nucleotide Polymorphisms) from 27,243 contigs in expressed genomic regions showed that not only domestication strongly reduced genetic diversity of domesticated as to wild forms but also had profound effects on gene expression patterns, with contigs of the domesticated frequently showing different levels of expression and patterns of coexpression as to the wild forms. A high percentage of genes (~9%), as to diverse studies in other crops, were found as under selection during domestication. Deep changes at genome level were detected after introduction and spread out of Americas.

Genetic survey of the *Cistus albidus* populations in Western Mediterranean

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Cistus albidus L. is a dominant species in Mediterranean degraded habitats, where it is representative of the earlier successional stages of the shrub land ecosystem. This obligated hard-seeder species usually occupy the arid and semi-arid Mediterranean coastal environments, in scattered populations widespread throughout the western part of the basin. Recently, however, an isolated, possibly relict population has been found in Crero (VR), nearby the Lake Garda, in an unusual environment for this species. In order to investigate the evolutionary processes that shaped the present state for this species, we studied by means of SSRs the neutral genetic variability of the Crero population and of other fourteen populations of *C. albidus* from Portugal, Spain, France and Italy. We genotyped about 600 plants at five loci and the data were analysed by standard population genetics approaches. Genetic variability is high and differentiation highly significant. Bayesian analysis showed similarities between the Crero and the Sardinian populations.

Maternal history of Southeast Asia and Oceania

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Southeast Asia and Oceania were home to both one of the earliest and the last great human migration events. The first modern humans to exit Africa arrived in Southeast Asia around 60,000 years BP, subsequently reaching New Guinea and Australia at least 50,000 years BP. This initial wave of colonists was much later followed by a second migration, which eventually completed the colonization of Remote Oceania as late as 1000 years BP. With Taiwan as their probable starting point, these migrants brought with them agriculture, domesticated livestock and Austronesian languages. Aside from these two main waves of colonization, several small range movements of populations have shaped the current genetic diversity of Southeast Asia and Oceania. Mitochondrial DNA (mtDNA) has been extensively used to investigate the history of Oceania/Southeast Asia and, as a result of these studies, we now have the geographical resolution and the sample size required to make inference about past population events. Unfortunately, several past works on mtDNA have been focused on understanding the ancestry of single haplogroups, instead of providing a broad picture of current population diversity. Here we have assembled, to our knowledge, the most comprehensive data set of mtDNA hypervariable region I sequences from Southeast Asia, Near and Remote Oceania. We aim to use this data to provide a better understanding of the landscape of mitochondrial DNA diversity in island Southeast Asian populations and more insight into the origins and migration patterns of humans in this region of the world.

Y-chromosome and mtDNA diversity in the context of Eurasian language diversity

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The central goal of the ERC advanced grant project LanGeLin (LANguage-GEne LINeages) is to investigate the relationship between genetic and linguistic diversity, the latter inferred from structural language features, rather than from the vocabulary.

Y chromosome (Y-chr) and mtDNA (mitochondrial DNA) provide complementary information and allow one to investigate the different migrational histories of males and females, and their impact over the global language-gene relationships.

We assembled two datasets including 42 Eurasian populations for which both mtDNA/Y-chr and linguistic data were available.

We calculated and compared phylogenetic trees and Mantel's correlations between genetic, linguistic and geographical distances starting from three matrices: d_{GEN} based on F_{ST} (genetic distances); d_{SYN} based on syntactic features (linguistic distances); and d_{GEO} based on geographical distance between pairs of populations.

Both similarities and differences were evident between patterns of genetic and linguistic variation, casting light on both the genealogical ties between populations, and the mechanisms of language change.

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Molecular phylogeography of the smooth-coated otter (*Lutrogale perspicillata*, Mustelidae) for its conservation in the Mesopotamian marshes of Iraq

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The smooth-coated otter (*Lutrogale perspicillata*) (SCO) includes *perspicillata* (SE Asia), *sindica* (SE Pakistan, W India) and endemic-to-Iraq *maxwelli* morphological subspecies. Classified as vulnerable by IUCN, the SCO is threatened by habitat transformation, illegal trade and poaching. Non-invasively collected samples in Iraq (10), central and E Asia (nine countries, 46) were sequenced at the entire mtDNA Cytochrome-*b* gene and genotyped (ongoing) at the microsatellite DNA. We also obtained 307 bp-long gene fragments from 16 SCOs resident to US and European museums. We used 19 Lutrinae GenBank entries. In Iraq, we provided genetic evidence for endemicity of *maxwelli* to Mesopotamia (private haplotype, mostly from Al-Hawizeh Marsh, S Iraq), while we could not confirm the occurrence of the SCO in N Iraq (Kurdistan).We found reciprocally monophyletic mtDNA lineages across the SCO range each corresponding to a subspecies. Preliminary microsatellite DNA results confirmed this mtDNA picture, overall pointing to three Evolutionarily Significant Units. Looking-like SCOs from Singapore showed oriental small-clawed otter (*Aonyx cinerea*) mtDNA. We will investigate *A. cinerea*-mtDNA introgressed SCOs at the microsatellite DNA to assess if they are descendant of *L. perspicillata* x *A. cinerea* otters (i.e., first hybrid record in the wild for Lutrinae). Several museum specimens were genetically identified as other than the SCO (even an African otter), indicating that morphology can be not sufficient to reliably catalogue otter study skins. In Iraq, genetic and fieldwork (not shown) data will help establishing a National Single Species Action Plan for the SCO. [National Geographic Society Conservation Trust grant #C261-13 to F.B.].

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Brown marmorated stink bugs are invading Europe: potential pathways of origin of the alien pest populations of *Halyomorpha halys* (Heteroptera, Pentatomidae)

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The brown marmorated stink bug, *Halyomorpha halys* is an agricultural and household pest originating from Asia. In the last years it has become an invasive pest in North America causing severe economic losses to agricultural crops in the United States. Recently, *H. halys* has been retrieved in Europe (Switzerland, Germany, France, Hungary, and Greece) and, since September 2012, it has also been found in Italy. Tracing back the patterns of introduction and monitoring the spread of *H. halys* in the Italian territory in its initial phase of colonization will be useful in the view to implement better pest control strategies. The present study aims to identify the potential pathways of entry of *H. halys* by detecting the genetic diversity of specimens collected in Northern Italy, Southern Switzerland and Greece. The analyses of 1,175 base pairs of mitochondrial DNA cytochrome c oxidase I and II genes (*cox1* and *cox2*) on more than 130 specimens led to the identification of ten haplotypes: one, scored in Italy and Greece, is the same found both in China and North America, while two haplotypes found in Switzerland and Lombardy are shared only with Chinese specimens. The other seven haplotypes are new and present high similarity with Asian haplotypes. Present data show that the introduction of the brown marmorated stink bug in Europe has occurred by means of multiple events, probably both from Asia and North America, and that *H. halys* is currently expanding its range in the European continent.

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The taste of *Drosophila suzukii*: phylogeny and evolution of its gustatory receptors

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Insects rely on taste to evaluate food, hosts, and to select mates and hospitable zones to deposit eggs. They sense tastants and non-volatile pheromones through gustatory sensilla, the hair-like structures distributed on multiple parts of their body, including the proboscis, legs and wings. These sensilla house gustatory neurons (GRNs) that express different types of gustatory receptors (GRs). Here we compare the GR repertoire of an emerging *Drosophila* pest, *Drosphila suzukii*, which feeds and oviposits on fresh, soft fruits like berries, cherries and grapevine, to its two closely related non-pest species, *D. biarmipes* and *D. subpulchrella*, and to other more distant *Drosophila* species that instead feed, only on rotten fruits. Our results show evidence of specific gene gains in *D. suzukii*. Differences in GR repertoire may be a part of the evolutionary changes that has contributed to *D. suzukii*'s innovative behavior.

The study of MHC variation and evolution in the Alpine chamois: from targeted Sanger sequencing to portable nanopore technology (MinION)

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Major histocompatibility complex (MHC) proteins are essential components of the immune response, and are coded by the most polymorphic of the vertebrate genes. Being mediators between the organism and the environment, MHC-coding genes are excellent candidates for exploring adaptive processes and for estimating the genetic component of resistance to infections. Accurate genotyping is essential to understand the role and evolution of MHC in natural populations, but it may be extremely challenging due to gene duplication and high sequence variation. In a previous study, we performed a Sanger sequencing of the most polymorphic exon of the DRB locus in the Alpine chamois (*Rupicapra rupicapra*) that allowed us to infer stabilizing selection in different populations. To better characterize the DRB-like regions in healthy and mange-affected individuals, and possibly identify duplicated loci, we designed a less stringent amplicon-based approach coupled with Illumina Miseq technology, and obtained about 2.6 million reads. The short read lengths make it difficult to assembly our region that includes repetitive sequences, numerous nucleotide changes and indels. We are currently addressing this problem by sequencing the whole ~10 kilobases amplicon with the portable third generation sequencer MinION (Oxford Nanopore). This platform is based on the changes in ionic current as long DNA molecules (several to tens of kilobases) pass through nanopores. Preliminary results of our experiments confirm that the error rate is high. Still, long reads produced by this sequencer have proved very useful for de-novo assembly.

P. 26

A transcriptome annotation pipeline for non-model organisms

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The introduction of high-throughput sequencing technologies allowed researchers to generate large amounts of genomic data at limited cost and time. This opportunity had a groundbreaking impact on the study of non-model organisms: above all, RNA-Seq and *de novo* transcriptome assembly represent a valuable source of information in species for which genomic resources are scarce or absent. However, sequencing and assembly are only the first steps, and an accurate annotation is fundamental for every kind of biological analysis. Annotation of transcriptomes from model organisms and their closely-related species is quite straightforward, and is generally based on simple sequence similarity searches. Conversely, non-model organisms require more complex and integrated procedures in order to infer remote homology and function. We present a pipeline specifically thought for the annotation of transcriptomes of non-model organisms. It consists of an integrated approach that combines different bioinformatics tools to obtain: 1) ORF prediction, identification of pseudogenes and artificially fused transcripts; 2) coding sequence annotation based both on sequence similarity and on the identification of conserved domains by protein signature recognition; 3) functional annotation of coding sequences by the assignment of GO terms; 4) annotation of noncoding transcripts. We tested our pipeline by re-annotating the transcriptome of *Ruditapes philippinarum* (Bivalvia, Veneridae).

Wikipedia: with great power comes great (evolutionary) responsibility

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Wikipedia is the main online, multilingual and collaborative open access encyclopedia, consulted daily by millions of people worldwide. The possibility of unsupervised and fragmented contributions can however lead to the spread of inexact information, thus mining its credibility and usefulness. This problem is particularly evident in the phylogenetic sections of various taxonomic pages of the Italian Wikipedia, which are often missing or less accurate than the corresponding English ones. For instance, in the Italian version there was no mention of the currently widely accepted phylogenetic hypotheses of Mandibulata (grouping most arthropods) and Toxicofera (grouping most squamates). In this project, developed within the course 'Evolution and Phylogeny' for the Master in 'Evolutionary Biology' (University of Padova), we revised and updated the Italian pages of Arthropoda, Cardamine, Chiroptera, Darwin finches, Diptera, Lepidoptera, Plantae, Rodentia, Squamata, Testudinata and Tunicata. We fixed obsolete classifications, incorporated or modified phylogenetic trees, and updated referenced information using recent scientific literature. Modifications were based either on the more accurate English Wikipedia, or on our own search into the specialist literature. Moreover, we fixed a technical bug in the input procedure of the 'Clade template' used in Wikipedia to draw modifiable trees. This project shows that reliable scientific knowledge can be easily transferred from the university to the non-academic world, for which Wikipedia represents the most accessible source of information. We advocate that such practice should become more widespread within universities, especially with regard to evolutionary studies.

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