



**Oggetto:** due posizioni in genomica evolutiva e della conservazione

Nell'ambito di un progetto PRIN appena finanziato dal titolo "Genomic susceptibility to extinction: a whole-genome approach to study and protect endangered Italian endemics" (coordinatore: Giorgio Bertorelle; abstract qui sotto), verranno bandite a breve due posizioni presso l'Università di Ferrara, una per un postdoc senior e una per uno studente di dottorato. Il postdoc senior potrà essere inquadrato come RTDA (3 anni) o come assegnista di ricerca (2+1), e dovrà occuparsi delle analisi di genomica di popolazioni ma anche di collaborare al coordinamento di tutto il progetto. Il dottorando dovrà principalmente occuparsi della parte di genomica di popolazioni, e svolgerà almeno 6 mesi del suo dottorato presso il laboratorio di Rasmus Nielsen (UC Berkeley). Queste attività verranno svolte in collaborazione con Andrea Benazzo, Silvia Fuselli, e Emiliano Trucchi. Gli interessati sono pregati di contattare Giorgio Bertorelle per ulteriori dettagli (ggb@unife.it).

Il progetto prevede anche un grande sforzo di sequenziamento (unità di Firenze, responsabile Claudio Ciofi) e di bioinformatica (unità di Trieste, responsabile Marco Gerdol), e due studi funzionali in colture cellulari (unità di Ferrara presso laboratorio di Paolo Pinton) e in vivo (unità di Padova, responsabile Leonardo Congiu). Per queste parti del progetto verranno attivate ulteriori posizioni di assegnista di ricerca, e gli interessati sono pregati di contattare direttamente i responsabili appena menzionati.

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### **Abstract del progetto**

Italy is a biodiversity hotspot, but several endemic species, representing a unique biological heritage, are endangered. Main threats are related to human activities causing fragmentation and decline in their population size. Extinction risks can be reduced by improving knowledge of genetic variation and developing conservation strategies aimed at preventing genetic erosion.

Small and declining populations are routed to radical changes in their genetic diversity as natural selection is less efficient and random genetic drift becomes the major player. Genetic drift can lead to the accumulation of deleterious mutations, i.e., the mutation load, affecting individual and population fitness and, in turn, further reduce population size even to extinction. Understanding the genome-wide dynamics of this process can reduce a species extinction risk.

The enormous improvement of next generation sequencing techniques, computing resources, and statistical methods now allow the study of complete genomes from several individuals virtually in any non-model species. Genomes can be screened to predict the deleterious effects of different mutation types and, ultimately, to estimate the mutation load in single individuals and in populations, and to predict its impact on fitness.

Using five Italian iconic endangered endemics as model species (a mammal, a reptile, an amphibian, a fish, and an insect) and an unprecedented effort of massive sequencing, bioinformatics and population genomics analyses, we propose here a comprehensive conservation genomics project with three major goals:

- i) understand the dynamics of the accumulation of deleterious mutations in small populations, and its impact on individual fitness and extinction risks;
- ii) estimate the genomic susceptibility to extinction due to the mutation load, predict the consequences of a strategy of genetic rescue, and propose conservation actions;
- iii) boost the interaction between research and practice in conservation genomics, and increase public awareness about biodiversity erosion and innovative molecular tools to prevent it.

For each endemic species, we plan to produce five de novo genomes and to resequence twenty individuals from two populations with different estimated population size. Computational approaches will be used to estimate demographic histories and different measures of mutation load and to quantify the genomic susceptibility to extinction due to load (GSEm). Computer simulation will be used to predict how the accumulated load might affect the outcome of a genetic rescue strategy before possibly implementing it, leading to the genomic susceptibility to extinction due to rescue (GSMr)

To support our results, we plan two functional assays to study i) in vitro the correlation between bioenergetic and cellular functions and the negative effects predicted in silico for fixed deleterious mutations, and ii) in vivo the segregation pattern of the load and its correlation with individual fitness in controlled inbred and outbred crosses.

Finally, we plan to reduce the existing gap between researchers, practitioners, and citizens by establishing a Conservation Genomic Consortium, organizing a public exhibition on genetics, biodiversity, and conservation, and activating a series of dissemination activities with the final aim of favouring a novel attitude about genetic studies and how these can help developing species and environment protection plans.