# A chromosomal drive of genetic diversity in an island species

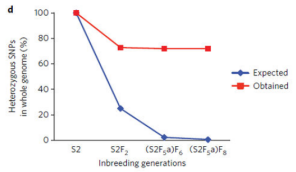
Longhua Guo[1], Joshua Bloom[1], Daniel Dols[2], Katarina Ho[1], Zain Kashif[1], Kaiya Kozuma[1], Eyal Ben David[1], Marta Riutort[2], Alejandro Sánchez Alvarado[3], Leonid Kruglyak[1]

1 Department of Human Genetics, Department of Biological Chemistry, and Howard Hughes Medical Institute, University of California, Los Angeles, CA, USA
2 Department de Genètica, Microbiologia i Estadística, Facultat de Biologia, Universitat de Barcelona, and Institut de Recerca de la Biodiversitat (IRBio), Barcelona, Catalonia, Spain
3 Stowers Institute for Medical Research, and Howard Hughes Institute for Medical Research, Kansas City, MO, USA

Island species have provided valuable insights into our understanding of genome diversity and evolution since the time of Charles Darwin. Sexual biotypes of the freshwater planarian, *Schmidtea mediterranea*, are distributed largely on the islands of Sardinia, Corsica, and Sicily, and in rare locations in Tunisia. We genotyped a collection of 70 animals from all four locations, with 2 asexual animals from Menorca included as an outgroup. Our data revealed two population superclusters in the sexual planarians: Sardinia-Corsica and Sicily-Tunisia. While the Sicily-Tunisia cluster maintains Hardy-Weinburg equilibrium, the Sardinia-Corsica cluster has elevated genome heterozygosity and nucleotide diversity. We established a linkage map by sequencing single sperms and chromosomes of a Sardinia isolate and determined the elevated heterozygosity and diversity is predominantly a contribution from chromosome 1. Linkage Disequilibrium decays more slowly on chromosome 1 in Sardinia-Corsica but not in Sicily-Tunisia. Microscopy imaging showed that chromosome 1 has reduced crossovers in oocyte meiosis in Sardinia. Our work illustrated that evolution and recombination can drive the genetic diversity in island species at the level of a whole chromosome. Our analytical strategy can be applifed to most species without chromosome-scale genome assemblies.

The freshwater planarian, *Schmidtea mediterranea*, is a research organism for the study of adult stem cells and regeneration. It has four pairs of chromosomes and a genome around 800Mb. Earlier studies found that part of the genome only exists as heterozygous state in strains from Sardinia. Such heterozygous regions are resistant to laboratory inbreeding.


*Newmark and Sánchez Alvarado 2002*




*Guo et. al. 2016*

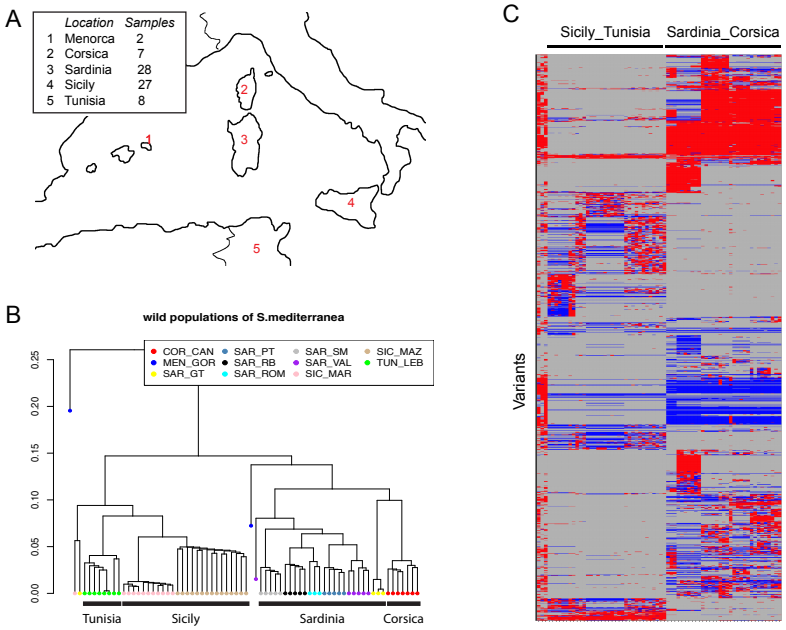



**Fig 1 Population structure of *S. mediterranea*.** (A) Sites of collection. Numbers of animals per site were listed in column "Samples". (B) Clustering of individual relatedness, with 2 asexual animals from Menorca as the outgroup. (C) K-means clustering of all sexual animals by genotypes. red:heterozygous; blue:homozygous alternative; grey: homozygous reference.

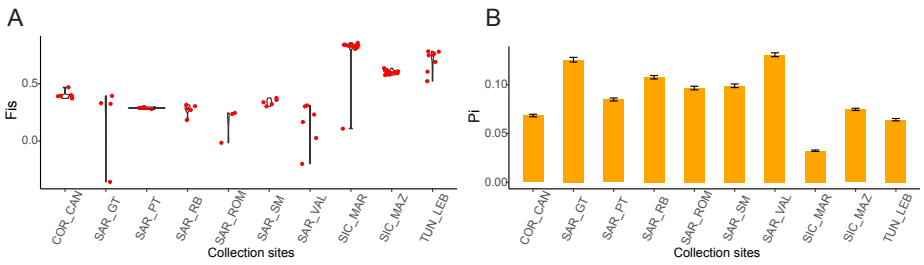



**Fig 2 Population statistics of *S. mediterranea*.** (A) Inbreeding coefficient per individual calculated by VCFtools. (B) Nucleotide diversity per collection sites calculated by STACKS.

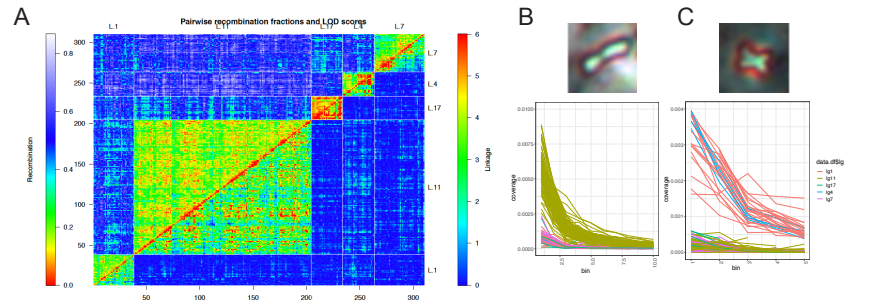
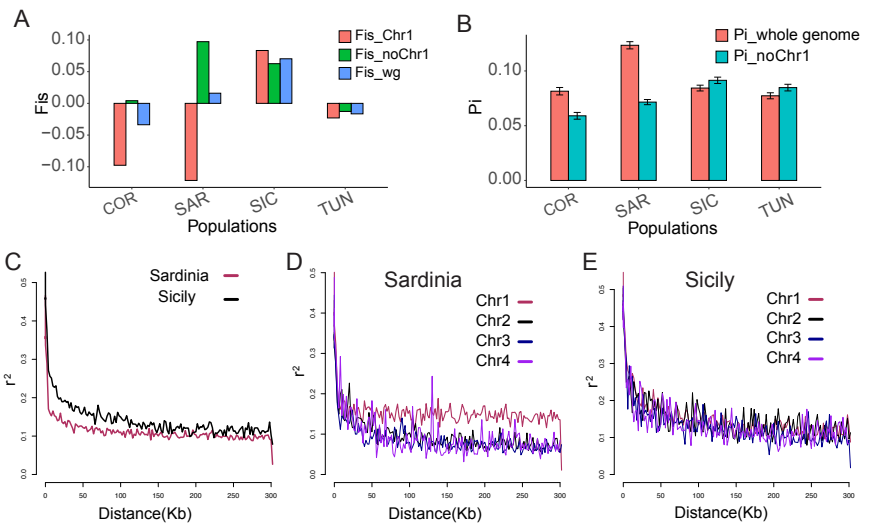



**Fig 3 Linkage map built by single sperm sequencing and single chromosome sequencing** (A) DNA from single sperms (n=48) of a Sardinia isolate were amplified and sequenced. Recombination events result in five linkage groups. (B-C) Single chromosome sequencing determined the chromosomal identity of the linkage groups.



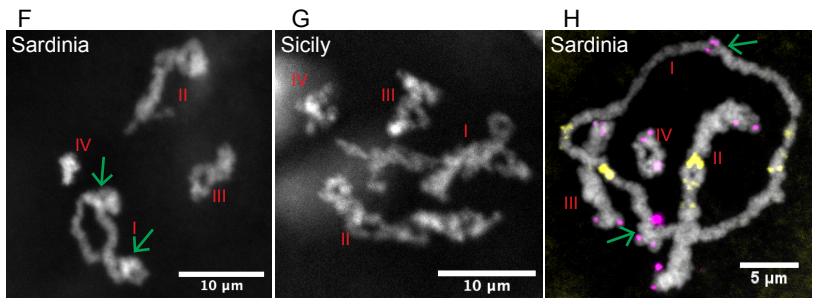




**Fig 4 Polymorphism of chromosome 1 in Sardinia-Corsica and Sicily-Tunisia clusters.** (A) Inbreeding coefficient per island calculated by STACKS, with chromosome 1 alone (red), the rest of the genome (green) and whole genome (blue). (B) Nucleotide diversity of whole genome (red) or genome without chromosome 1 (cyan). (C-E) Linkage disequilibrium decay of whole genome in Sardinia and Sicily (C), of four different chromosomes in (D) Sardinia and (E) Sicily. (F-H) Reduced number of crossovers (arrow) in chromosome 1 during oocyte meiosis I in a Sardinia isolate (F,H). Not in a Sicily isolate (G). (H) Fluorescence *in situ* hybridization with telomere (magenta) and centromere-like (yellow) DNA probes.

## Future directions:

1. QTL mapping of a F2 population to determine genetic architecture
2. Haplotype frequency in the natural isolates