Genomic data reveal historical introgression and genetic erosion across an imperiled radiation of Caribbean *Spondylurus* skinks

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Introduction

- Island systems drive speciation research yet host >50% of recent extinctions and a third of critically endangered species.
- In the Caribbean, various dispersal methods and Pleistocene sea-level cycles introduce complexity in species biogeography
- ***Spondylurus*** skinks are an understudied radiation spanning much of the region
- We used whole genome sequencing of fresh and historical samples to:
  - characterize species relationships and gene flow
  - estimate genetic diversity, inbreeding, and demographics
  - quantify genetic load across the genus to evaluate potential impacts of deleterious mutations in the lens of species conservation

Methods

- **Samples:**
  - Skinks captured non-lethally across Puerto Rico and the USVI from 2021-2023
  - Additional modern and historical tissue samples obtained from museum and tissue collections
- **Sequencing:**
  - myReads (historical), Qiagen QIAamp (historical), and NEBNext Ultra II (modern) Library prep for Illumina NovaSeq sequencing
- **Phylogenetics:**
  - Whole mtDNA and nuclear SNP comparison
  - IQTree (ML), MrBayes (BI), SVDQuartets (MSC), SplitsTree (Network)
- **Gene Flow:**
  - TreeMix (historical gene flow events), Dsuite (D-Statistics)
- **Population Demographics and Diversity:**
  - N_e within *S. nitidus* and *S. powelli*: GONE2
  - Runs of Homozygosity (ROH), and inbreeding coefficient (F_{ROH}) within *S. nitidus*, *S. powelli*, and *S. culebrae*
  - H_O genus-wide
- **Genetic Load:**
  - functional-based approach: variants were annotated using SnpEff v5.1 (*S. nitidus* reference genome)
    - High-impact variants (weight 1.0): nonsense mutations (stop_gained) and frameshift variants causing reading frame disruptions
    - High-impact variants (weight 0.9): splice-disrupting variants (splice_acceptor_variant, splice_donor_variant)
    - Moderate-impact variants (weight 0.8): in-frame indels (insertions/deletions preserving reading frame)
    - Moderate-impact variants (weight 0.6): missense variants (amino acid substitutions)
    - Excluded (weight 0.0): Synonymous variants (do not alter protein sequences)

$$\text{Genetic Load Ratio (GLR)} = \frac{\text{realized load}}{(\text{realized load} + \text{masked load})}$$

realized = deleterious alleles in homozygous form
masked = deleterious alleles carried heterozygously

Acknowledgements & References

Collections and tissue samples: KU Biodiversity Institute and Natural History Museum, University of Puerto Rico at Mayagüez, Museum of Comparative Zoology Harvard, Museum für Naturkunde Berlin, Dr. Aurélien Miralles

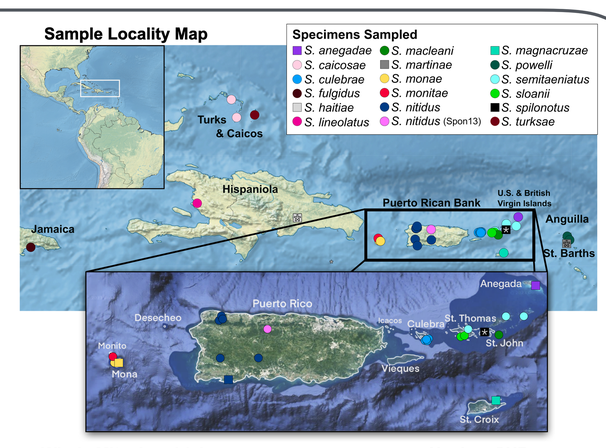
Thanks to: USFWS (Caribbean Ecological Services Field Office and staff), USFWS Caribbean National Wildlife Refuge Complex, USFWS Raleigh Field Office/Region 4, Puerto Rico DRNA, Virgin Islands DPNR, Island Conservation (IC), Para la Naturaleza, Sarah McRae, Isamarie Acosta, Ana Roman, Mike Barandiaran, Nicole Angeli, Lauren Scheinberg, Cielo Figuerola, Jose Luis Herrera-Giraldo, and James "Skip" Lazell. CAS CCG for computational resources.

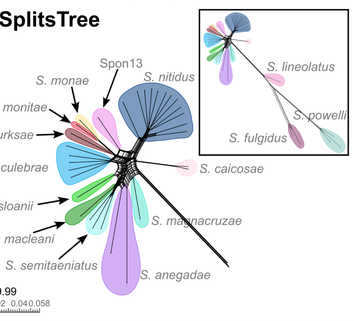
Find poster and all associated references here:

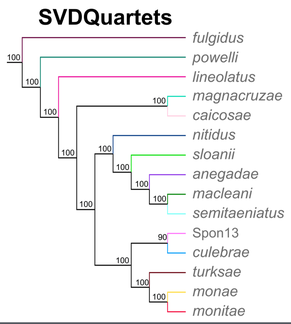
Results & Discussion

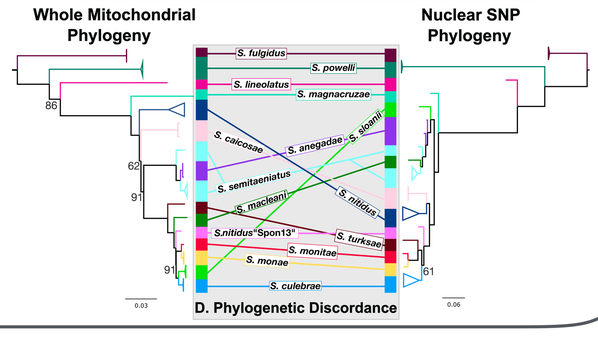
Phylogenetic Discordance & Biogeography

- mtDNA ≠ nuDNA
- nuDNA tree methods (ML/BI ≠ TreeMix ≠ SVDQuartets)
- Reticulation (SplitsTree), gene flow, and complex demographic histories likely contributors
- Considerable local extirpation for many species
- No consistent stepping-stone dispersal
- Unsampled diversity — Spon13 could be part of an unsampled *S. nitidus* population



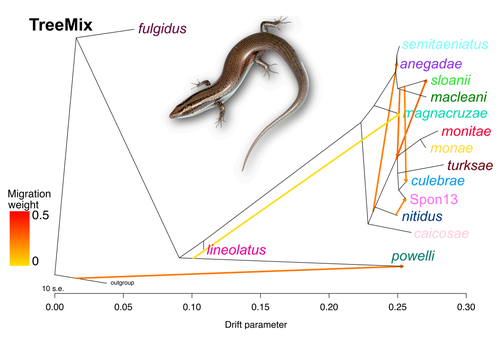


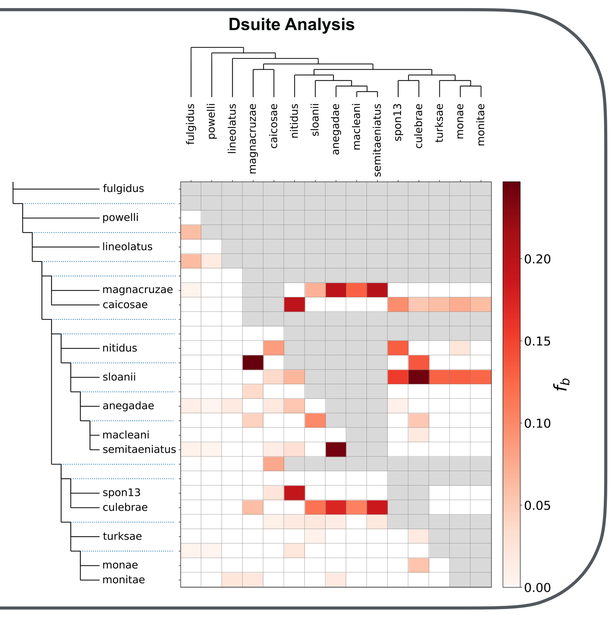




Gene Flow

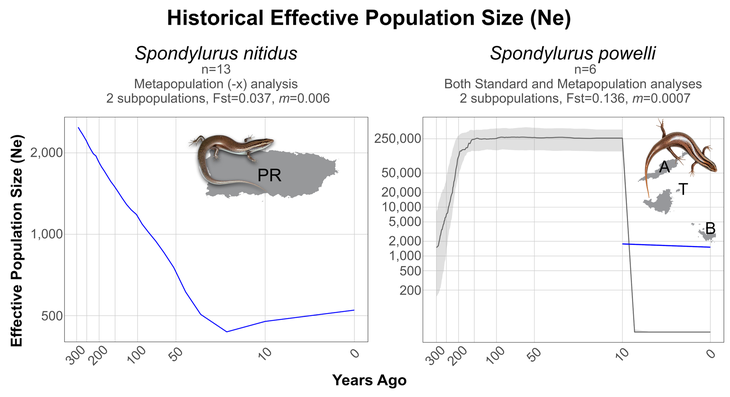
- Evidence of gene flow among many species - particularly along the puerto Rican Bank
- Inter-island gene flow facilitated by geographic proximity and Pleistocene sea-level fluctuations
- *Spondylurus* is part of a skink radiation with highly structured placental evolution
  - reproductive barriers are still not well characterized

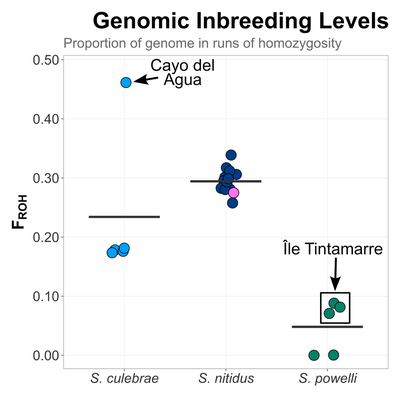


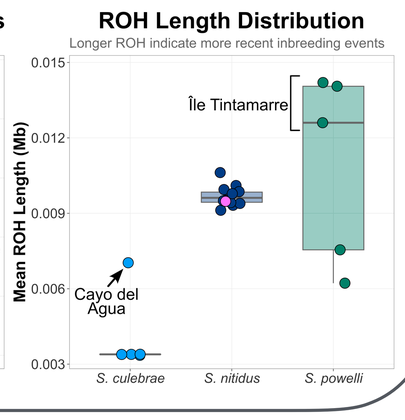


Demographic Change and Inbreeding within *S. nitidus*, *S. powelli*, and *S. culebrae*

- Considerable drop in N_e — colonization of the region and introduced predators
- Islet individuals have comparatively higher F_{ROH} and ROH — small isolated populations



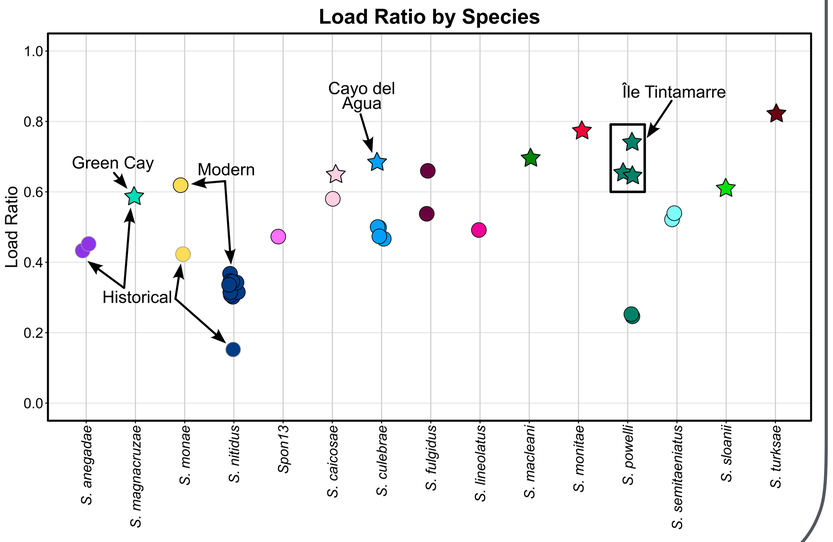




Diversity and Genetic Load

- Individuals from small islets (stars) have higher genetic load ratios
- Historical samples have lower genetic load compared to modern samples — genetic load may be accumulating in species over time

Species	Avg Observed Heterozygosity (Ho)	Avg Genetic Load Ratio	Avg Total Genetic Load
S. turksae	0.013	0.82	20109.30
S. monitae	0.016	0.78	18167.00
S. macleani	0.027	0.70	22144.6
S. caicosae	0.033	0.62	23544.50
S. sloanii	0.038	0.61	21803.80
S. fulgidus	0.105	0.60	73567.50
S. magnacruzae	0.040	0.59	23050.80
S. culebrae	0.051	0.53	23367.46
S. semitaeniatus	0.051	0.53	25928.30
S. monae	0.042	0.52	13253.05
S. powelli	0.128	0.51	58183.64
S. lineolatus	0.086	0.49	55559.00
Spon13	0.050	0.47	21128.00
S. anegadae	0.054	0.44	18179.35
S. nitidus	0.054	0.32	16706.03



Conservation

- U.S. Fish & Wildlife has proposed listing four *Spondylurus* species (*S. nitidus*, *S. culebrae*, *S. semitaeniatus*, and *S. sloanii*) as federally endangered or threatened under the U.S. Endangered Species Act
- Low contemporary Ne, high realized genetic load, and elevated F_{ROH} can be used to identify species and populations where monitoring and intervention may be needed

Spondylurus nitidus

Spondylurus culebrae