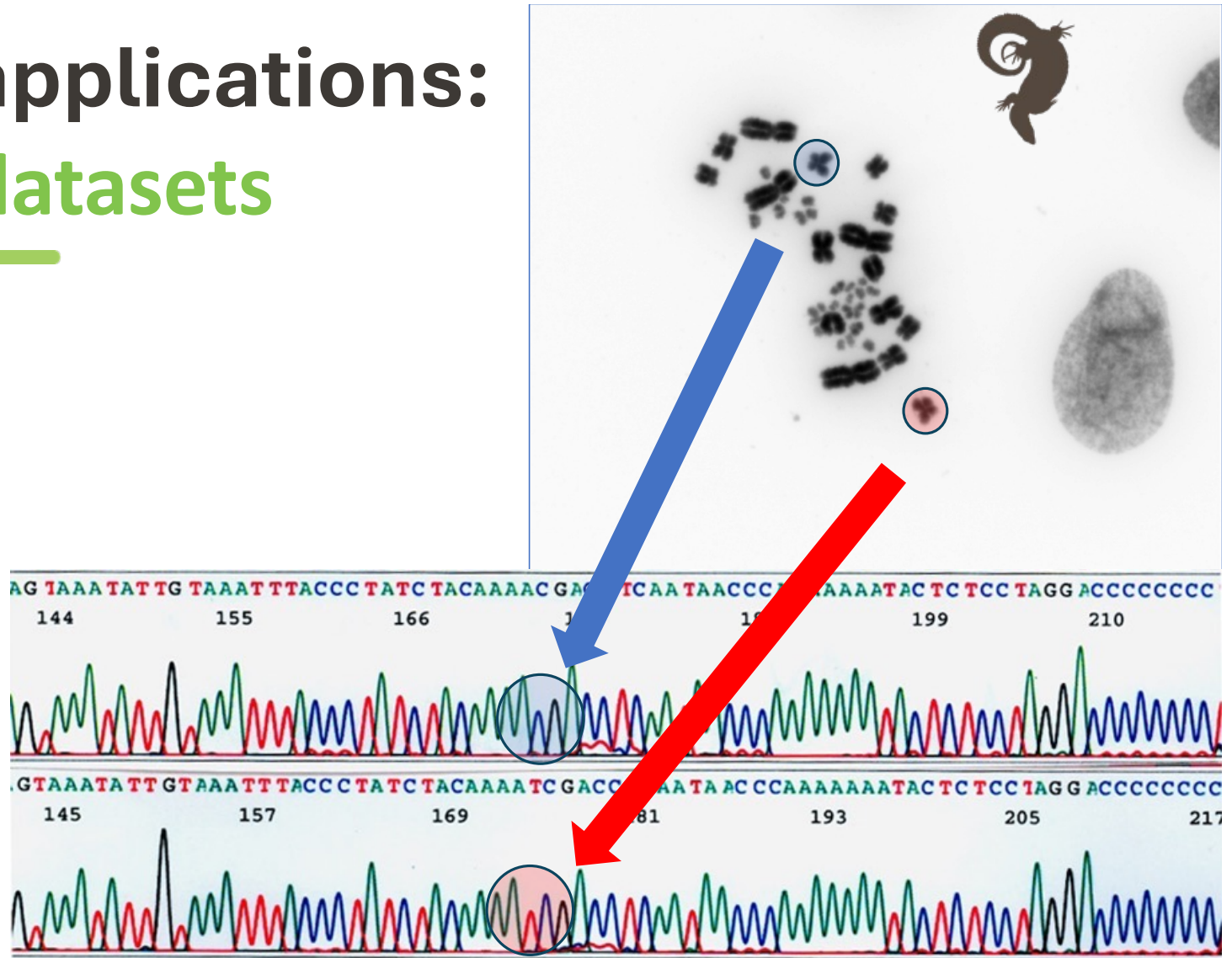


Additional applications:

Combining datasets

Genome alignments and introduction to incorporating structural variants.

- DARTseq service provides alignment of your favourite genome.
- Use BLAST function in dartR for additional alignments.
- Chromosome data can be incorporated in metafiles for analysis of specific structural variants that are known from the dataset.

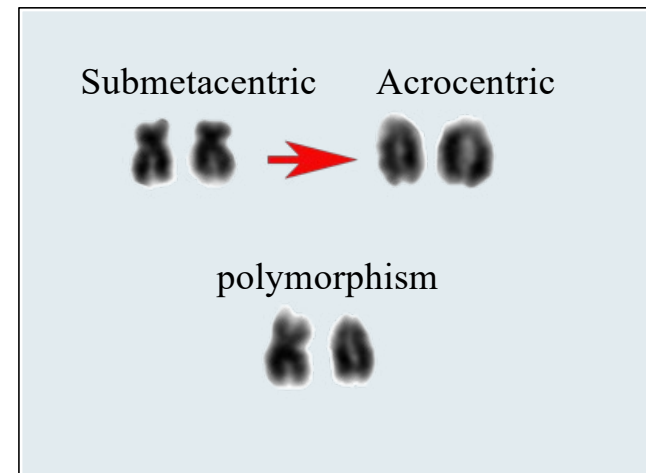


Inversion Chromosomal Polymorphisms in Ridge-tailed goannas

- Non-model organism without genomic resources prior to my work
- Known chromosome polymorphisms from a 1982 publication (King and King, 1982)
- Two major chromosomal groups: a western polymorphic cytotype and an eastern monomorphic cytotype
- The western cytotype is described by a pericentric-inversion polymorphisms for chromosome 6, and the eastern cytotype is described as monomorphic submetacentric chromosome 6.

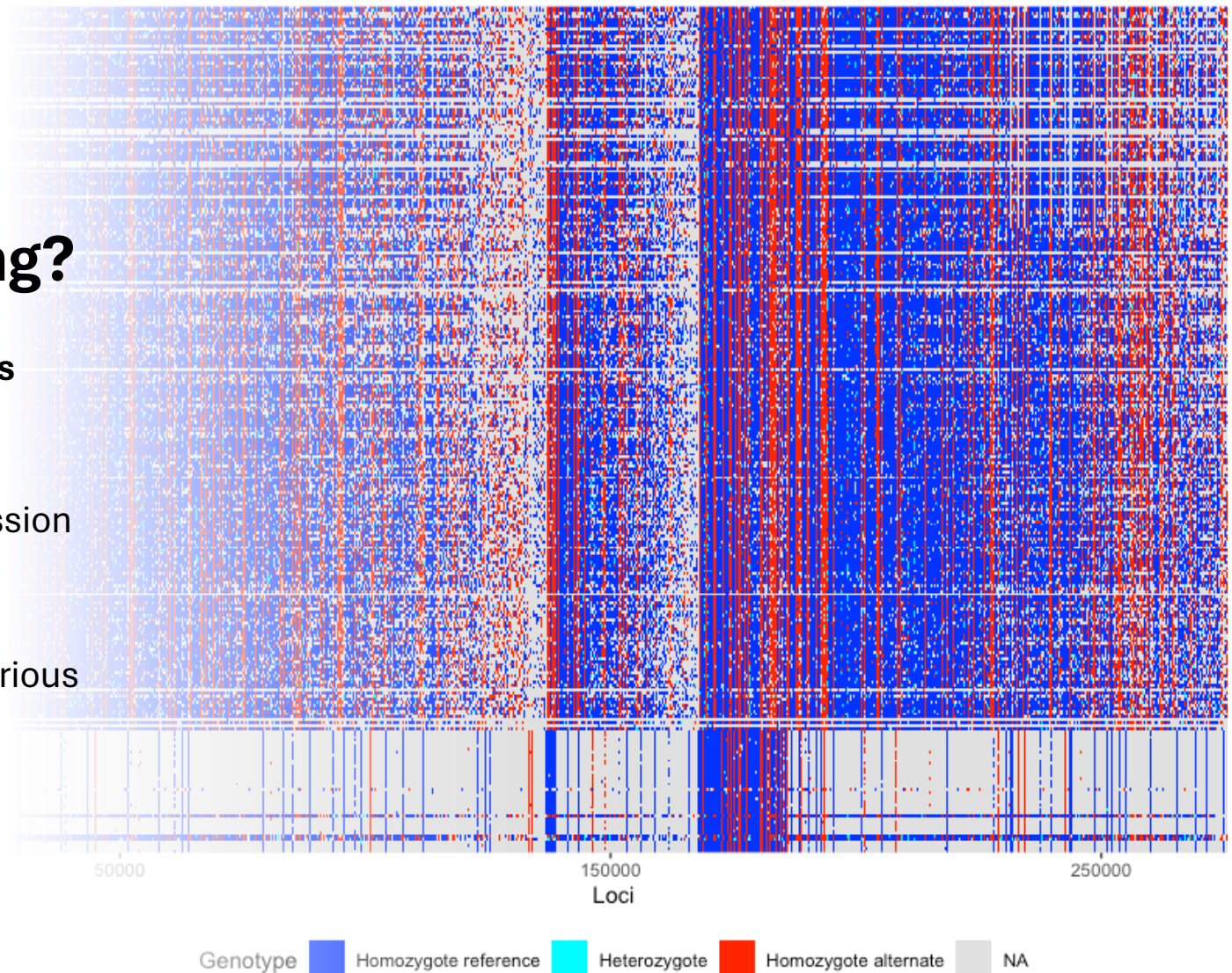


Chromosome 6



When the data speak: Are we listening?

- Impacts of pericentric inversions on populations
- Hybridisation
- Recombination suppression
- Speciation
- Adaptive potential
- Accumulation of Deleterious alleles
- Fertility Effects



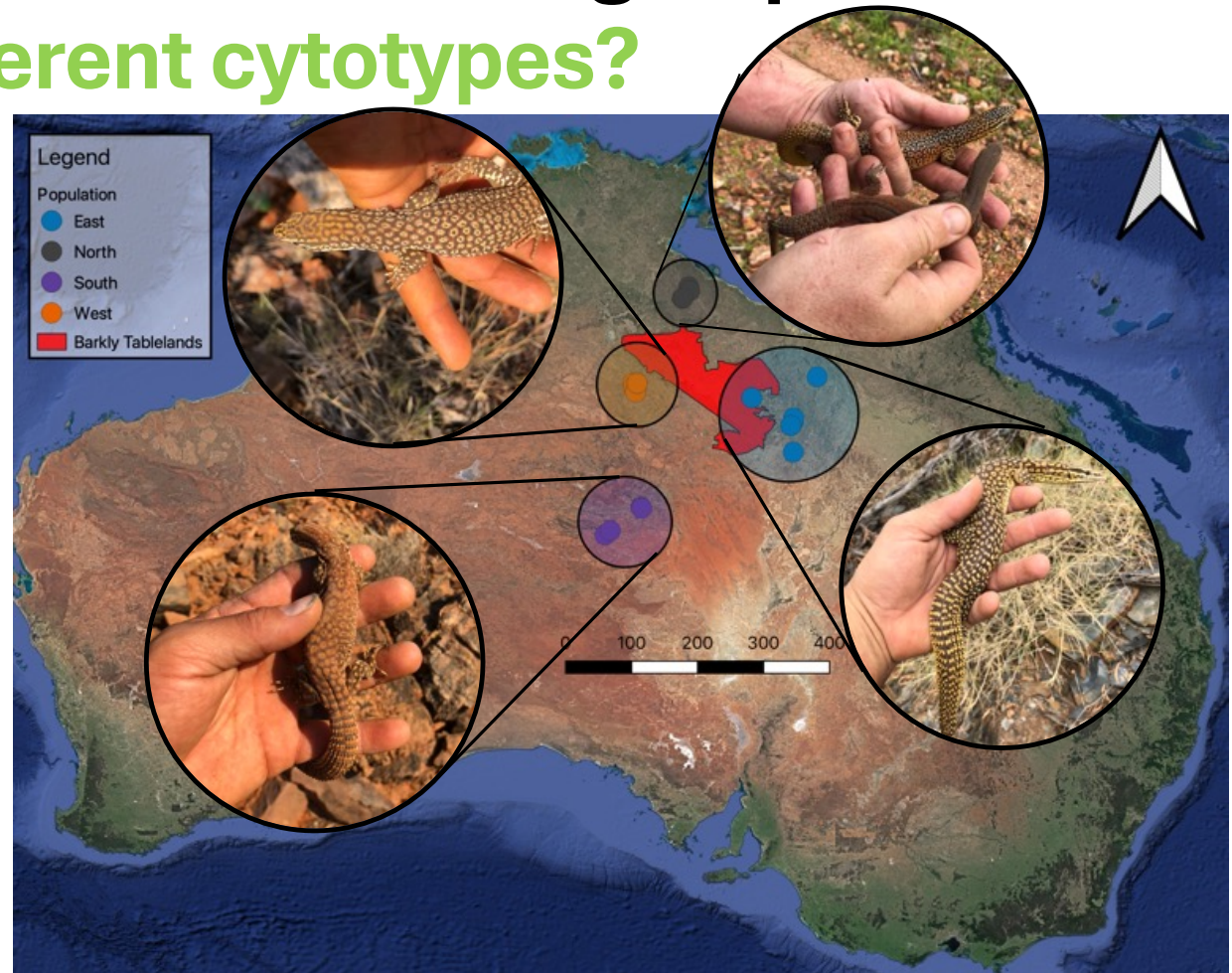
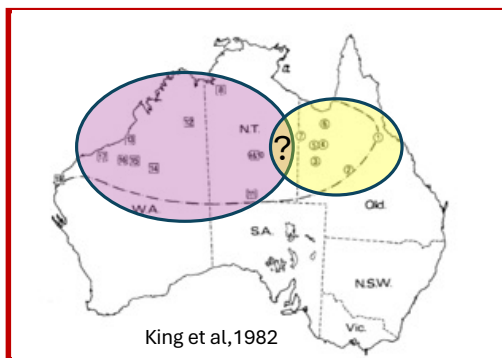
Is geneflow present between groups with different cytotypes?

Populations show high phenotypic variation

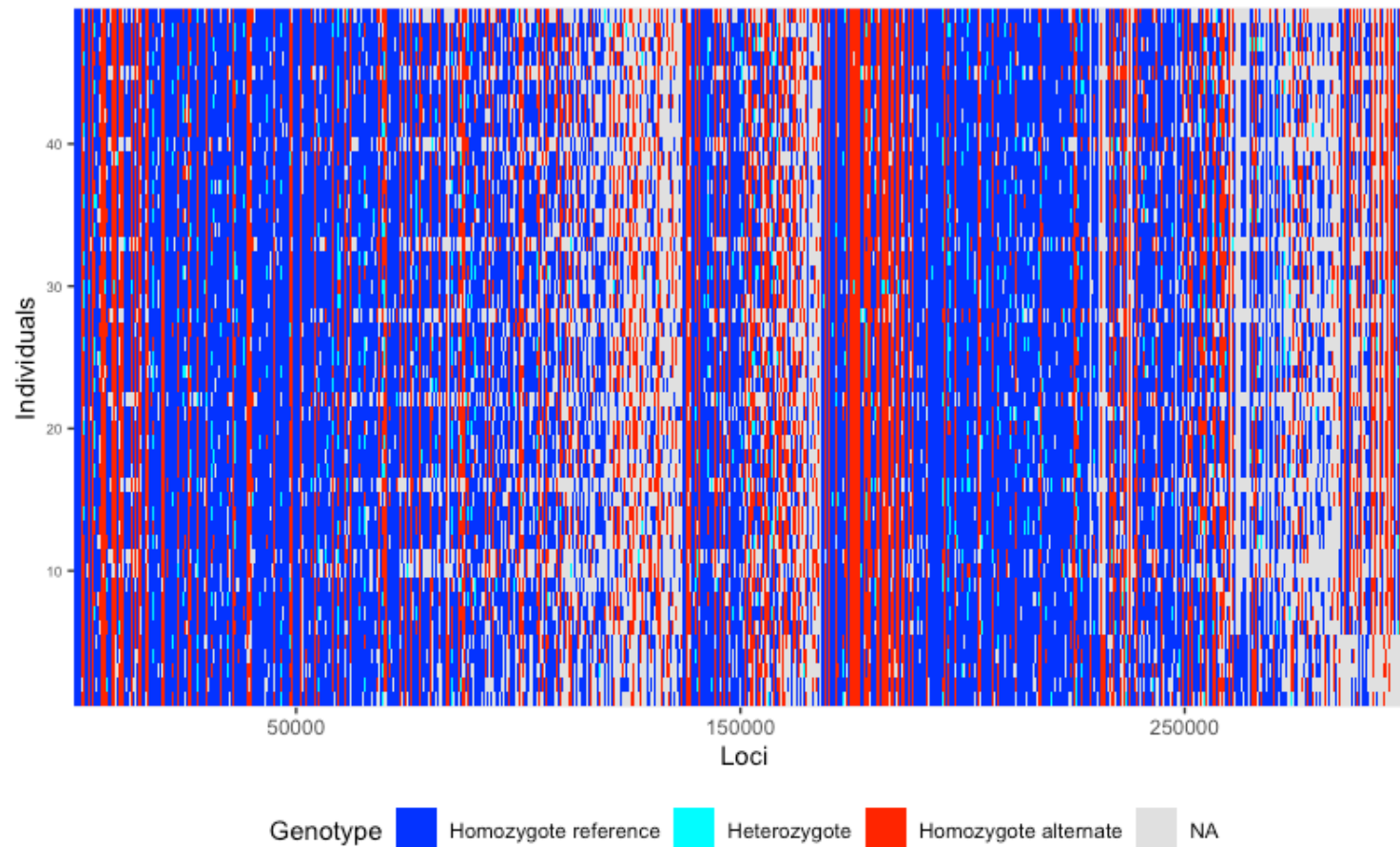
Slight phenotypic variation between populations

Cryptic species

Chromosome polymorphisms widespread in the west but not in the east.



DArTseq data reveals immediate results



Sorted SNPs by locality

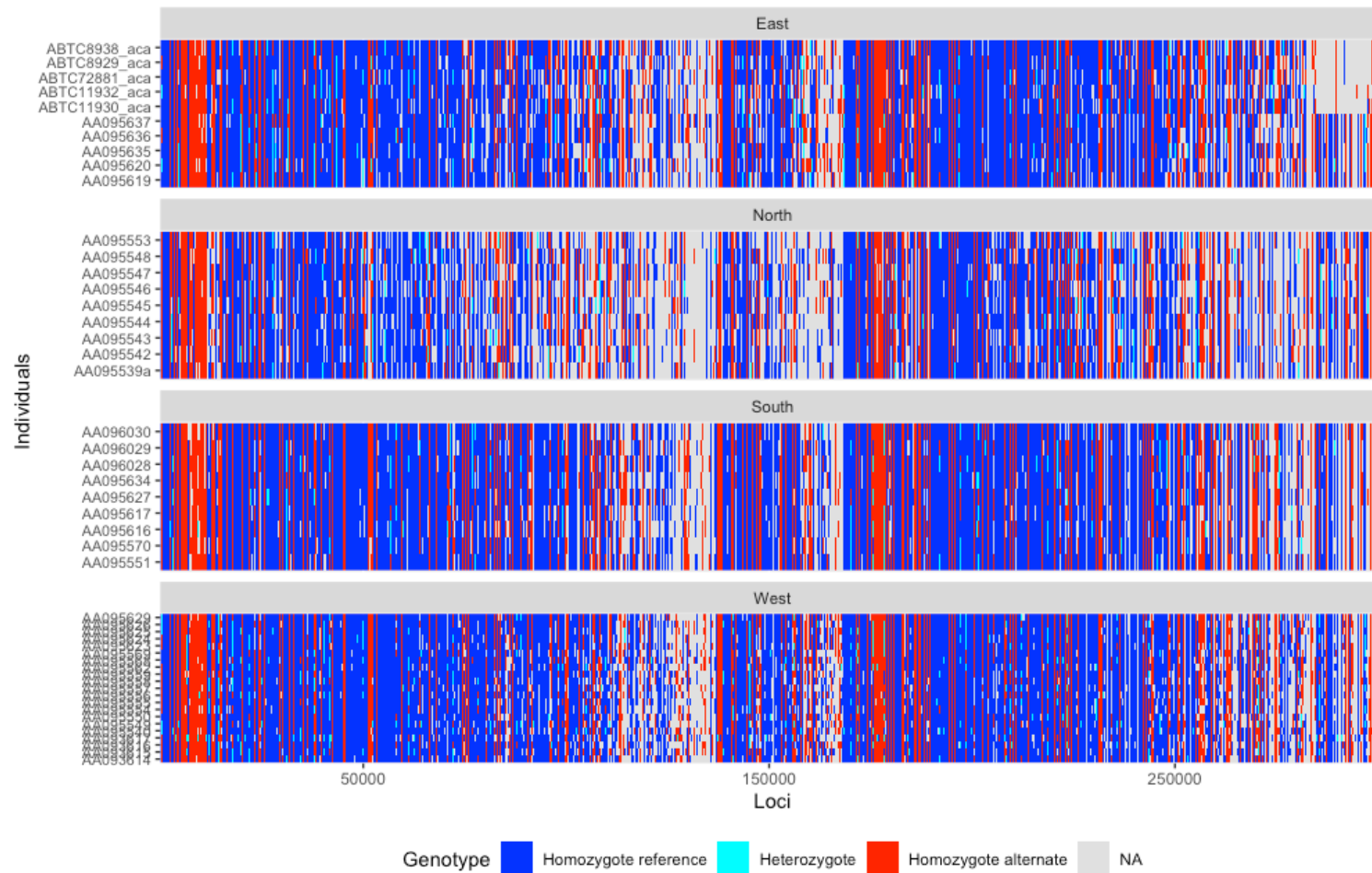


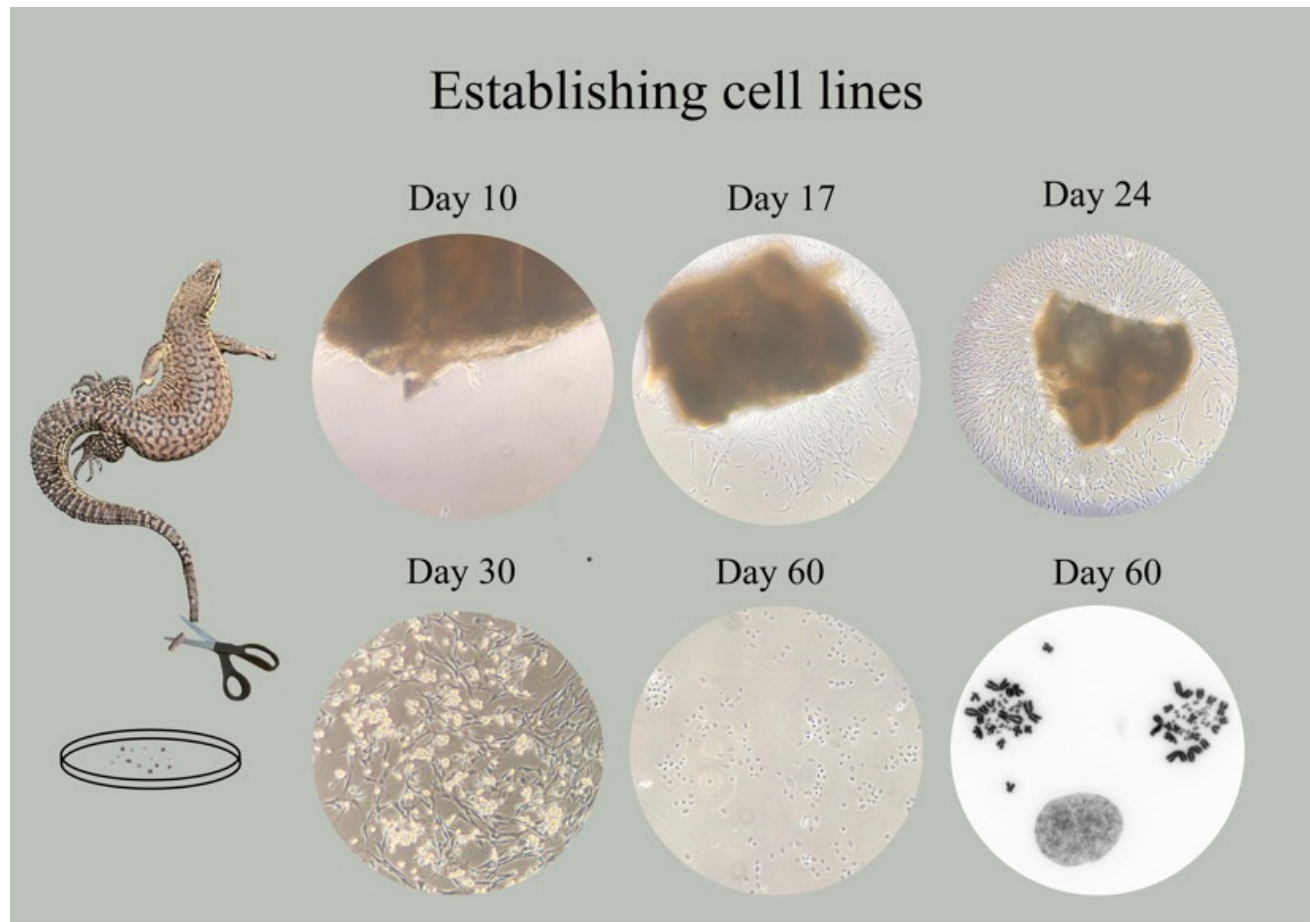
Table of SNP differences **between populations**

Dataset	Number of individuals	Number of SNP alleles		Number of monomorphs
		Pre filtering	Post filtering	
All populations	49 (M+F)	301,738	46,189	195,906
North	9 (M+F)	46,189	19,039	26,504
West	21 (M+F)	46,189	17,075	29,114
East	10 (M+F)	46,189	14,908	31,281
South	9 (M+F)	46,189	7,733	38,439

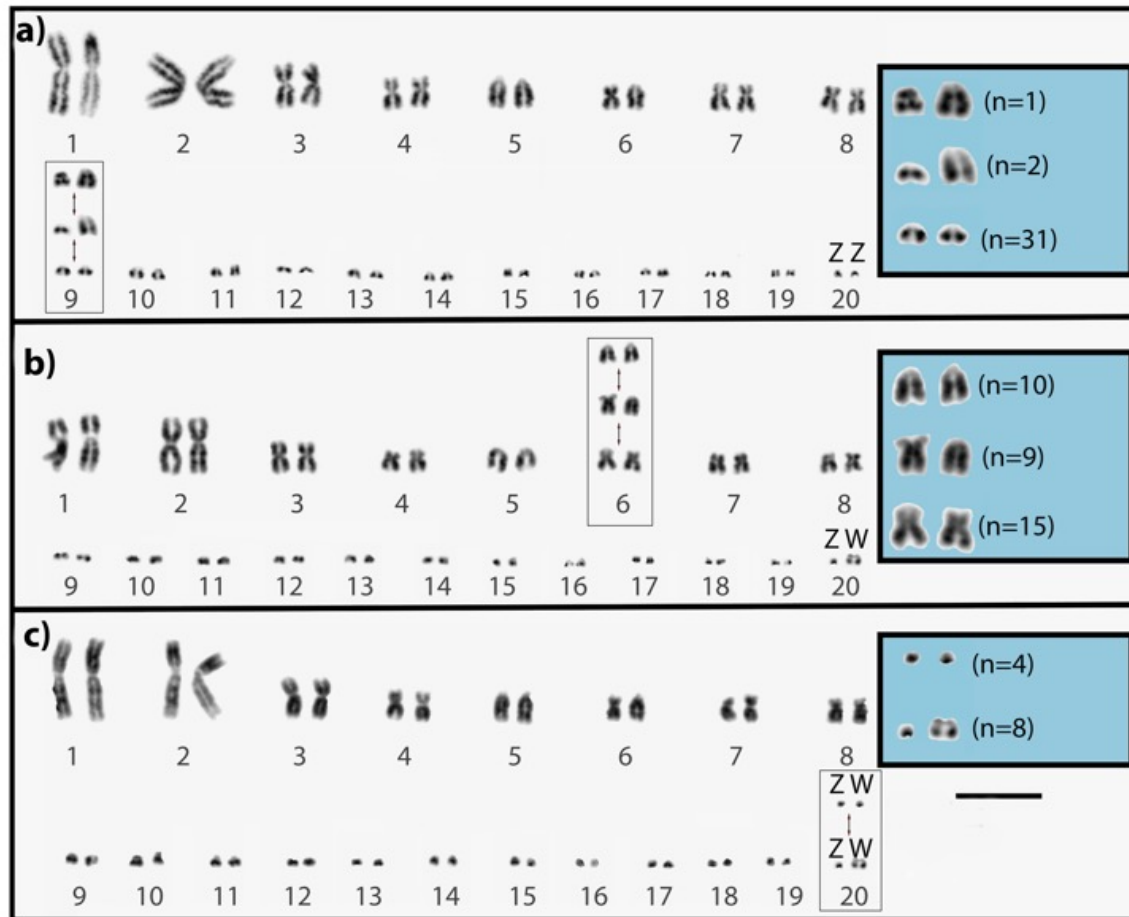
Fixed allele differences **between populations**

pop1	pop2	N1	N2	fixed	priv1	priv2	Total priv	Fst
East	North	10	9	4076	14107	18490	32597	0.65
East	South	10	9	578	12837	5668	18505	0.51
East	West	10	21	301	10400	12567	22967	0.58
North	South	9	9	5269	22669	11215	33884	0.69
North	West	9	21	4482	20233	17887	38120	0.73
South	West	9	21	111	3842	13177	17019	0.41

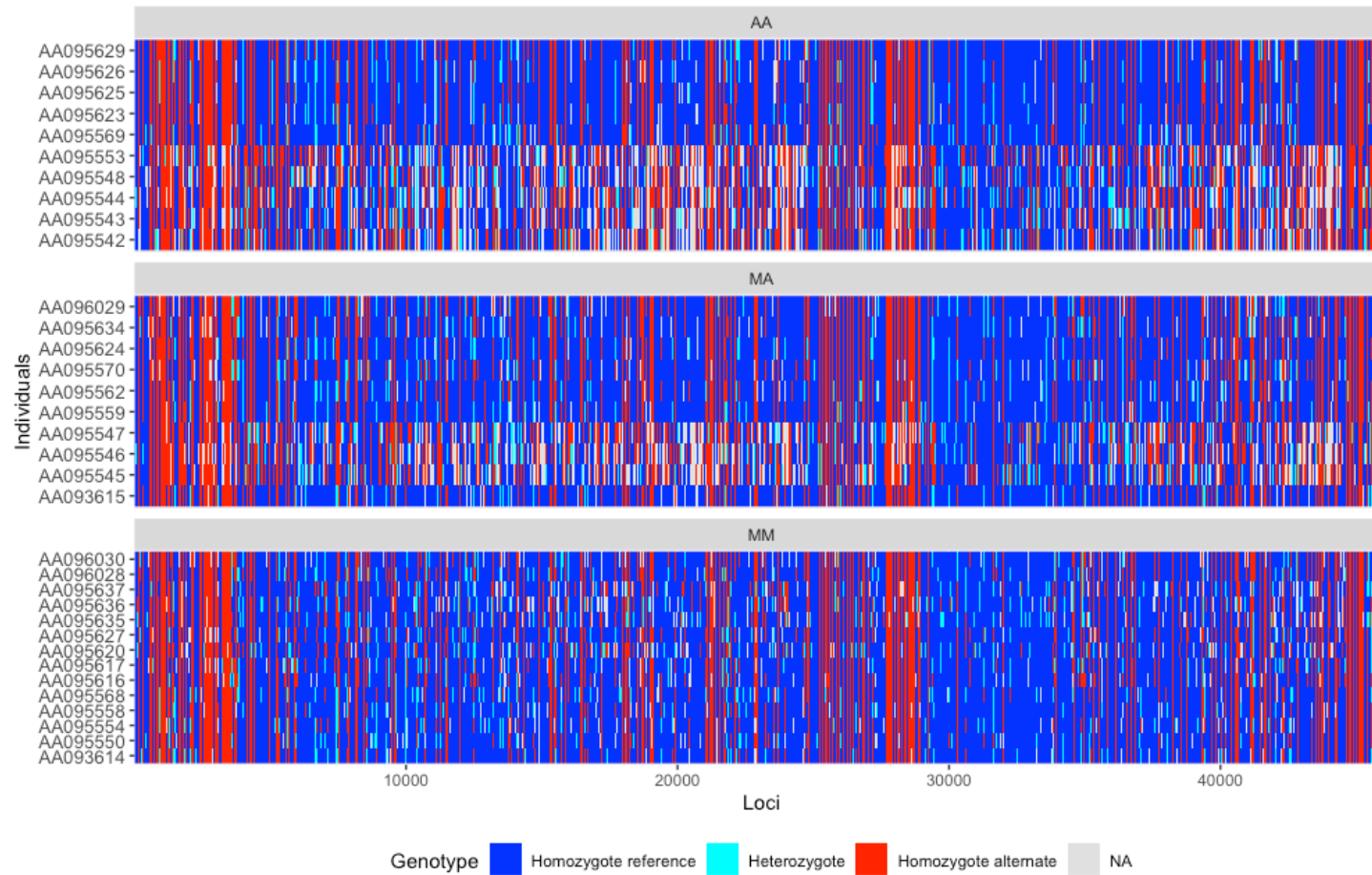
Generating Karyotypes



Karyotype Results



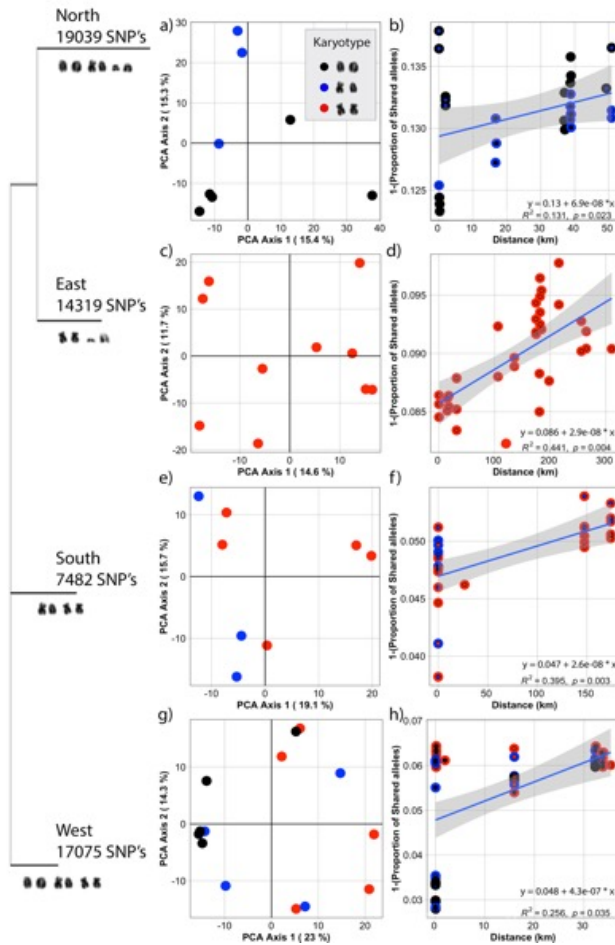
Sorted SNPs by Karyotype



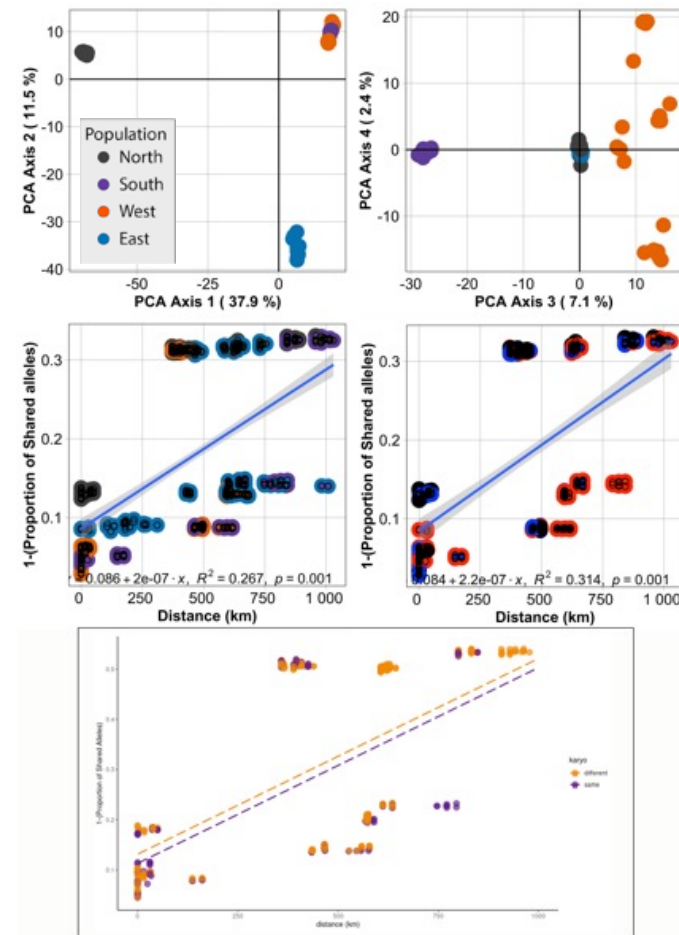
Fixed allele differences **between karyotypes**

Karyo1	Karyo2	N1	N2	fixed	priv1	priv2	Total priv	Fst
AA	MA	10	10	0	6041	6683	12724	0.095
AA	MM	10	15	13	16413	10529	26942	0.163
MA	MM	10	15	0	14399	7873	22272	0.103

Within populations



Between populations





Widespread chromosomal rearrangements preceded genetic divergence in a monitor lizard, *Varanus acanthurus* (Varanidae)

Jason Dobry  · Erik Wapstra  · Emily J. Stringer  · Bernd Gruber  ·
Janine E. Deakin  · Tariq Ezaz 

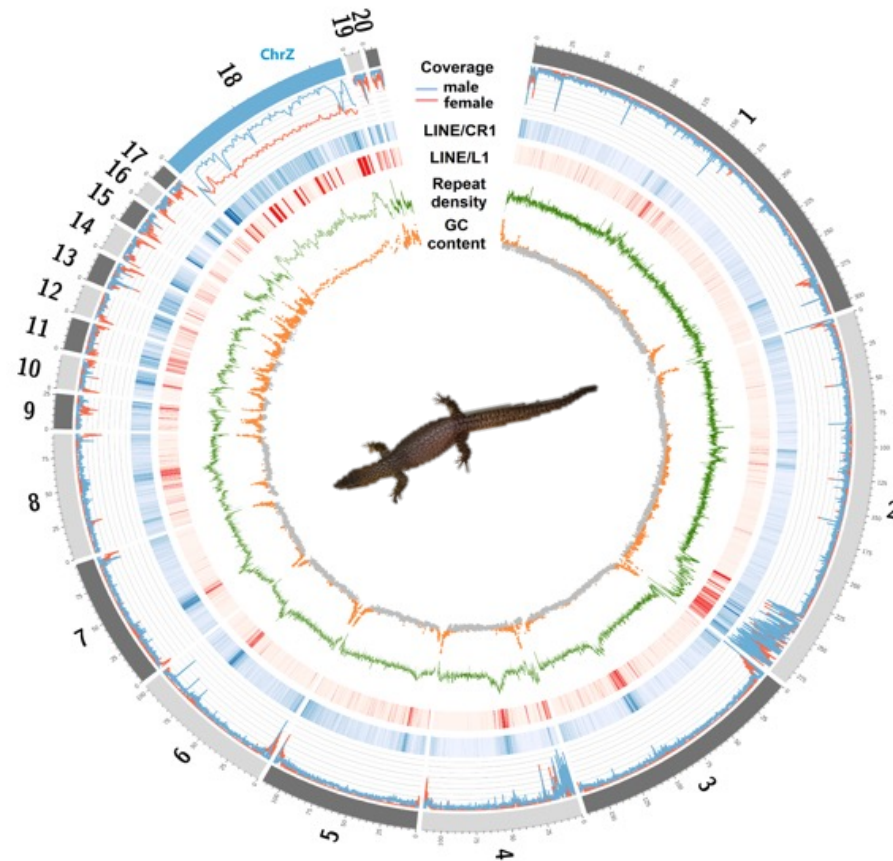
Received: 2 September 2022 / Revised: 12 November 2022 / Accepted: 28 November 2022
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Abstract Chromosomal rearrangements are often associated with local adaptation and speciation because they suppress recombination, and as a result, rearrangements have been implicated in disrupting gene flow. Although there is strong evidence to

rearrangements within populations, but there was no gene flow between populations that had similar chromosome rearrangements. Moreover, we identified a correlation between reduced genetic variation in populations with a higher frequency of homozygous sub-

Genome Sequencing

- A wide range of qualities in published genomes
- Scaffold/contig level (most genomes)
- Chromosome level – most chromosome level genomes still contain gaps, and chromosome identity is not correlated with karyotypes (some)
- Phased and chromosome assigned (very few)
- Quality depends on many factors, such as read length, depth, and quality.
- Most reference-quality genomes integrate several sequencing technologies to overcome individual technology limitations

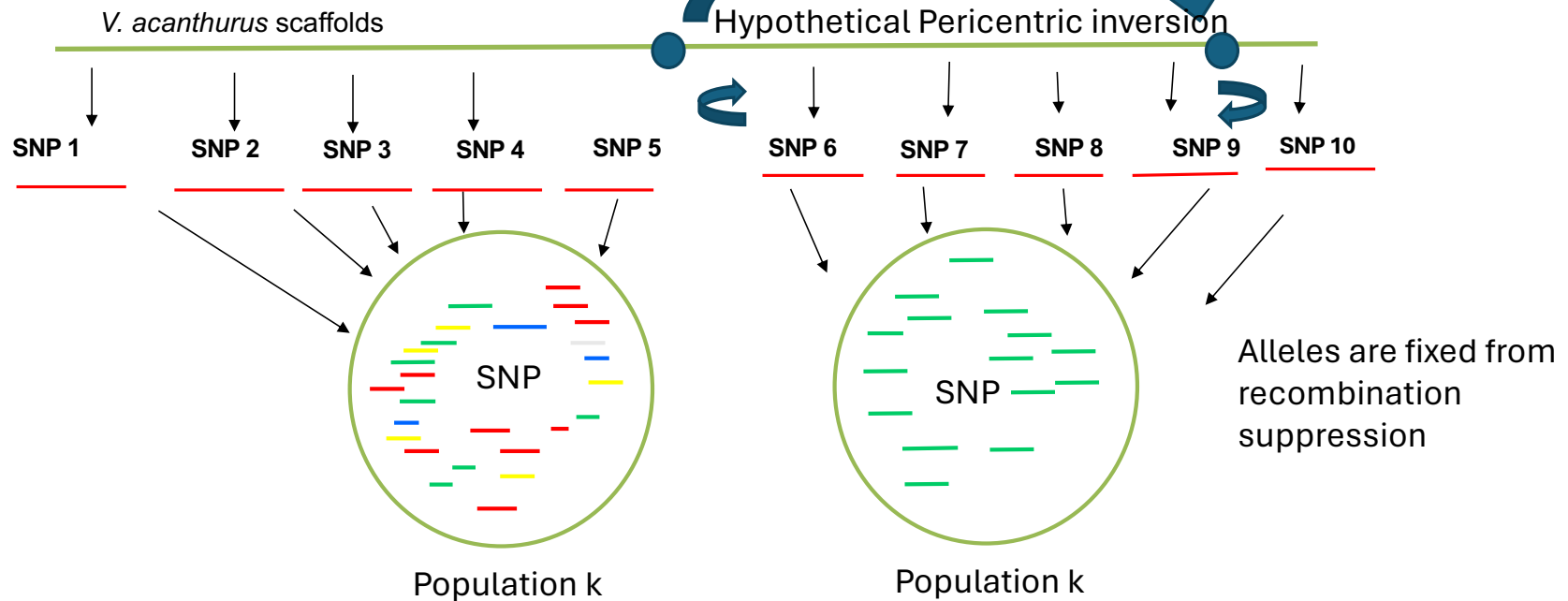


Characterizing a Chromosome Rearrangement – Conceptual model

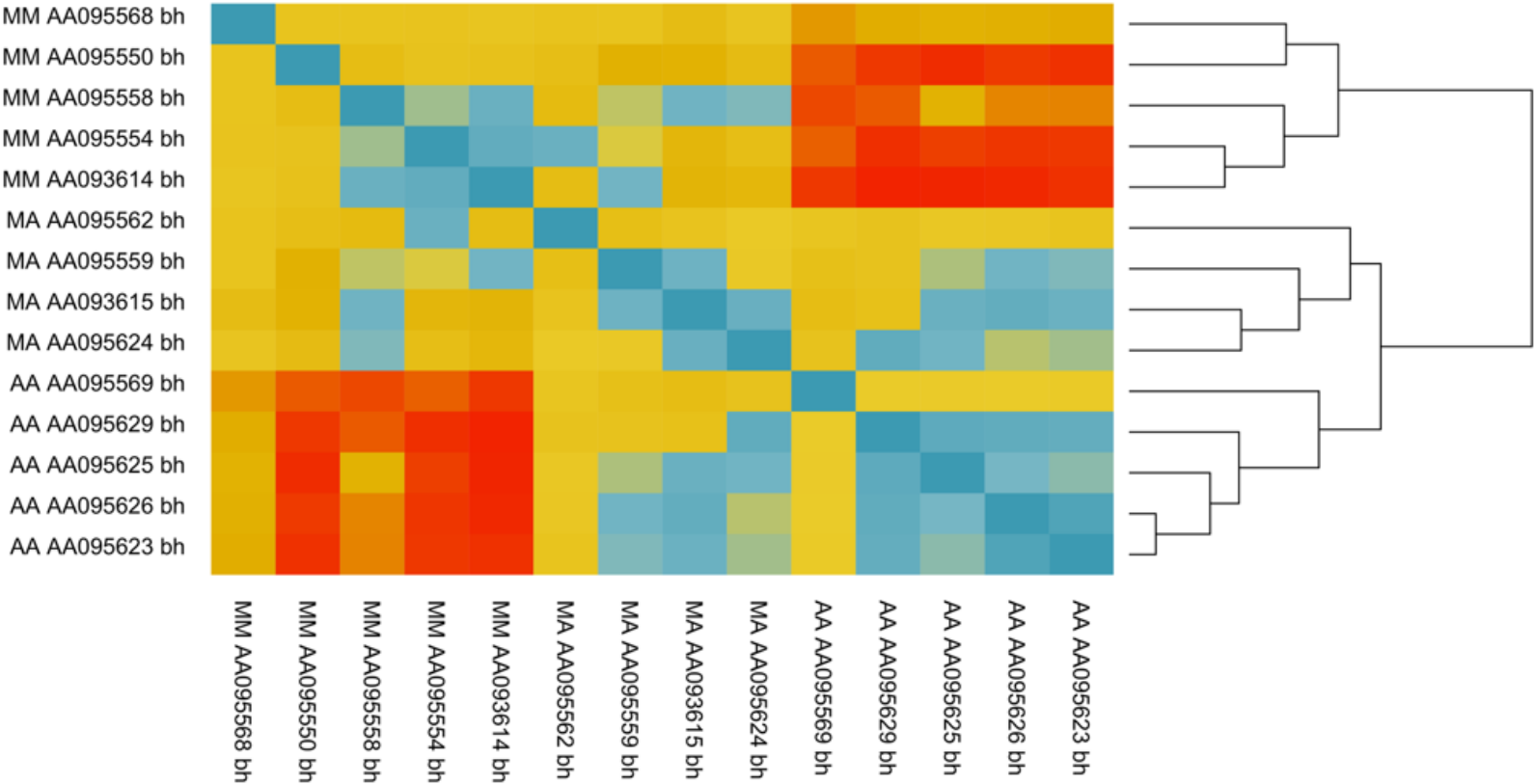
V. Komodoensis Genome

V. Komodoensis scaffold chromosome pools

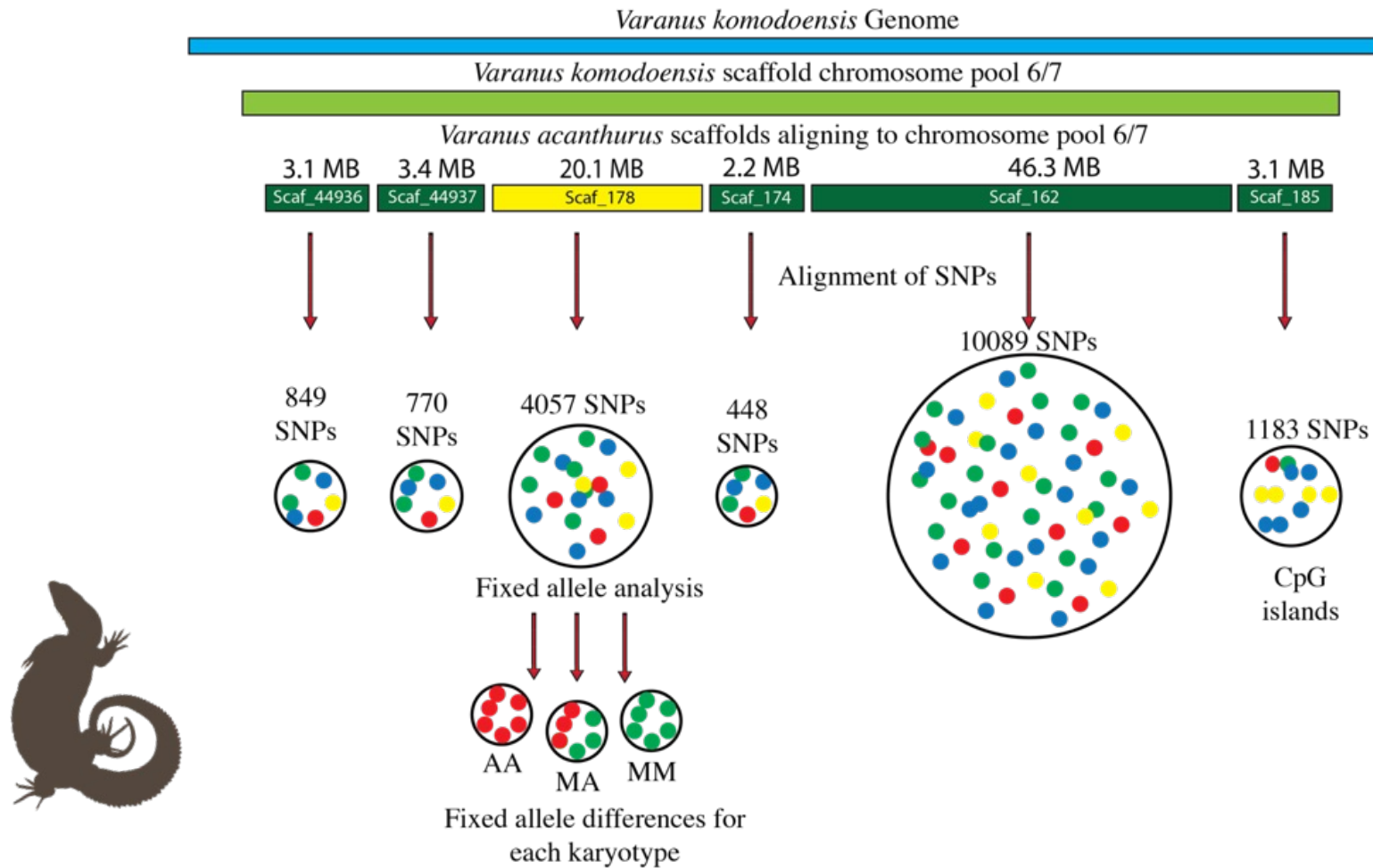
6/7



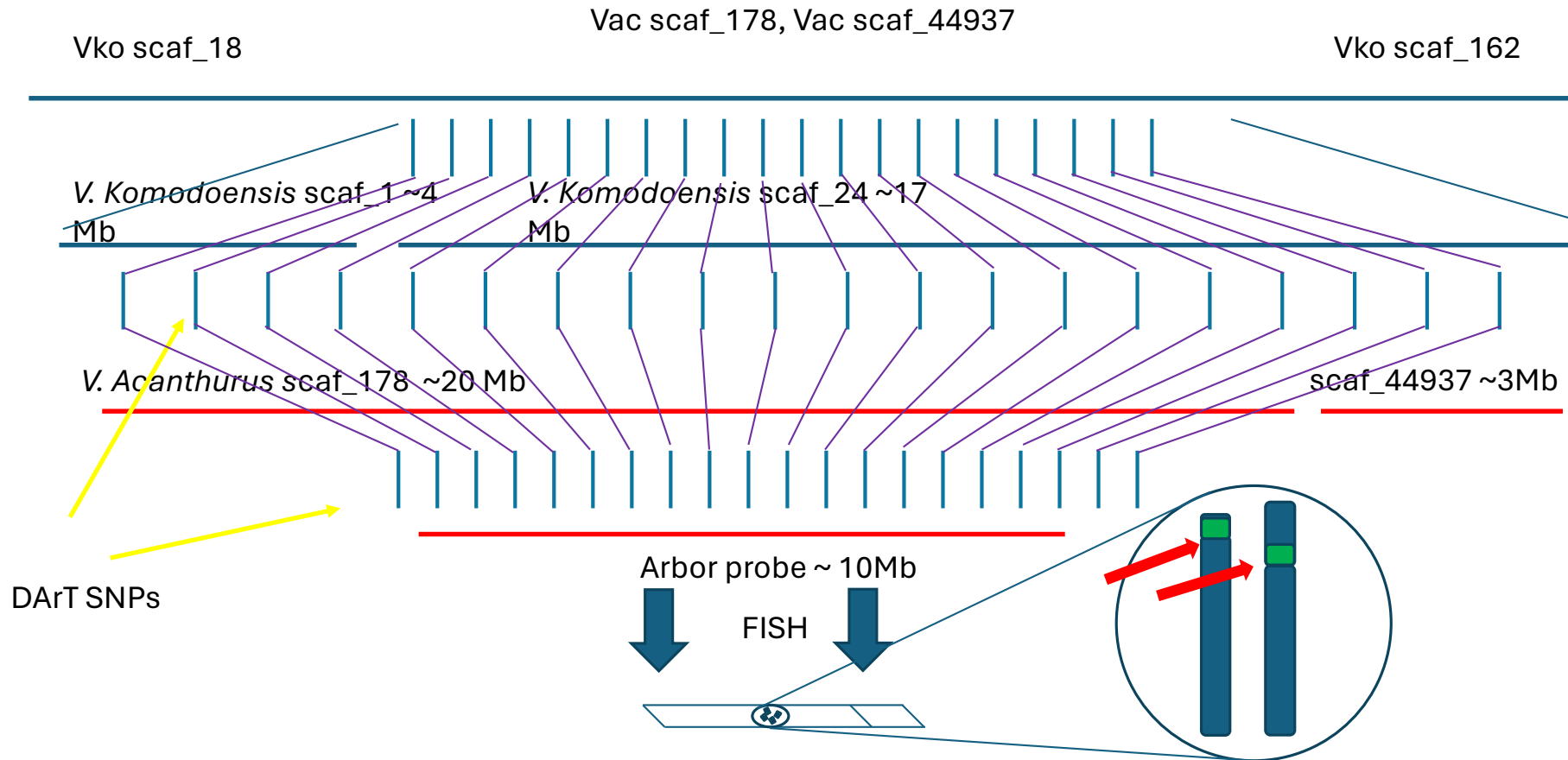
Fixed allele analysis of the west locality



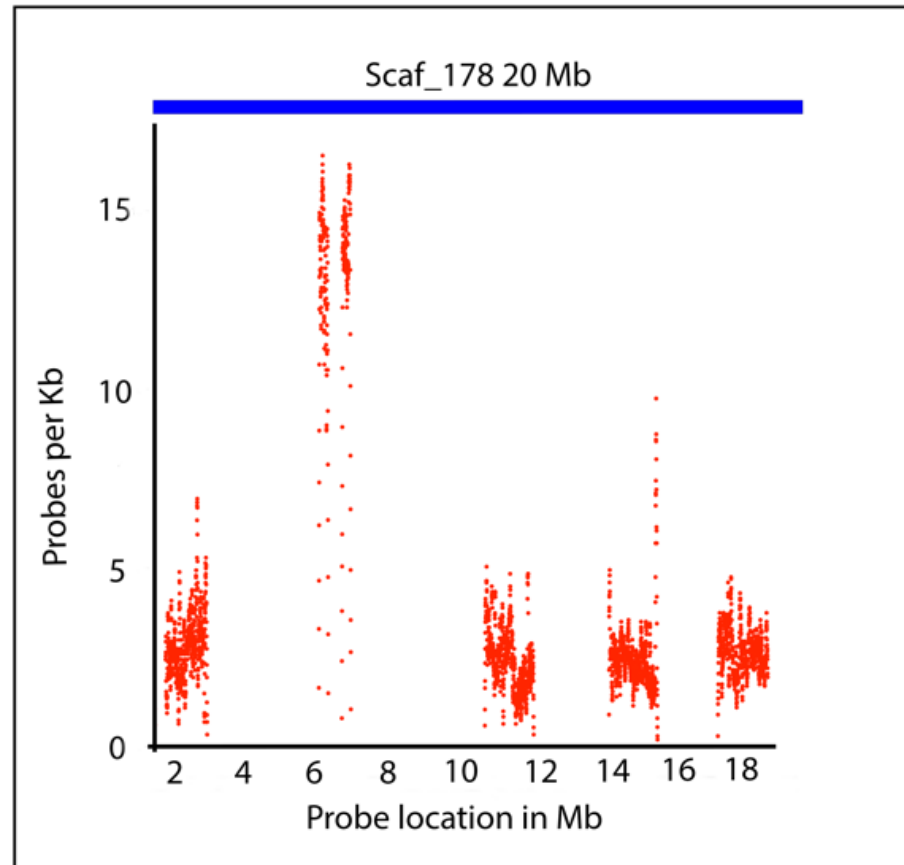
Distribution of SNPs along targeted Scaffolds



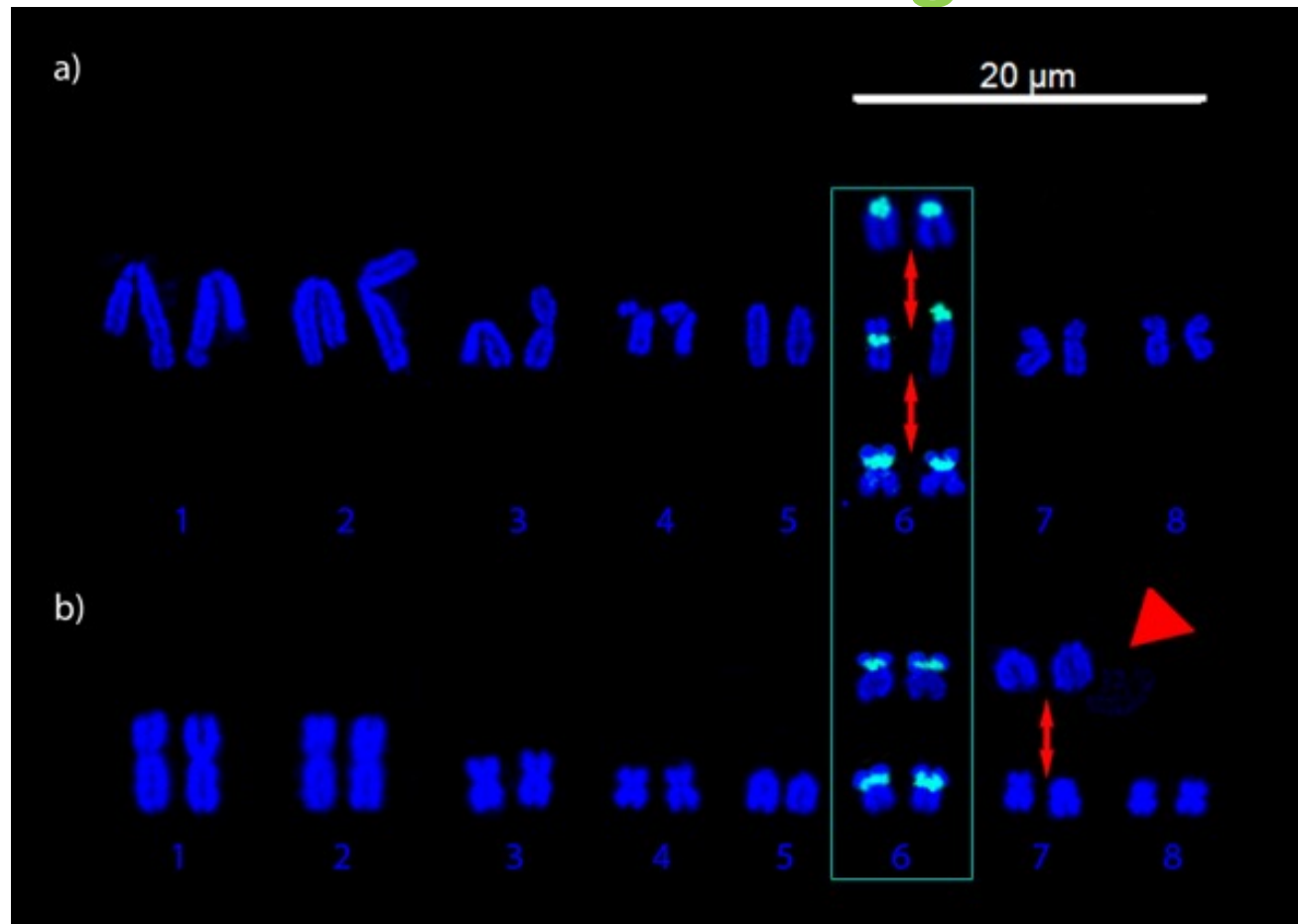
Concept for testing **homology of Inversion**



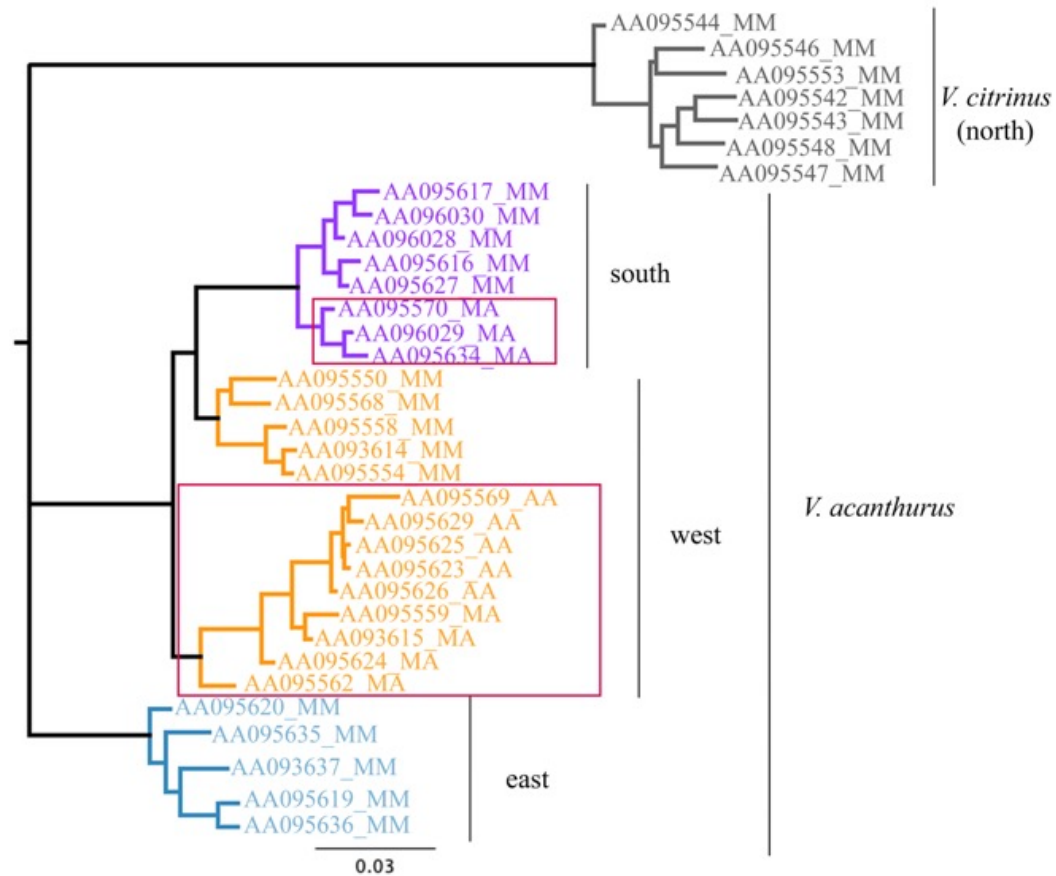
Probe density along scaffold of interest



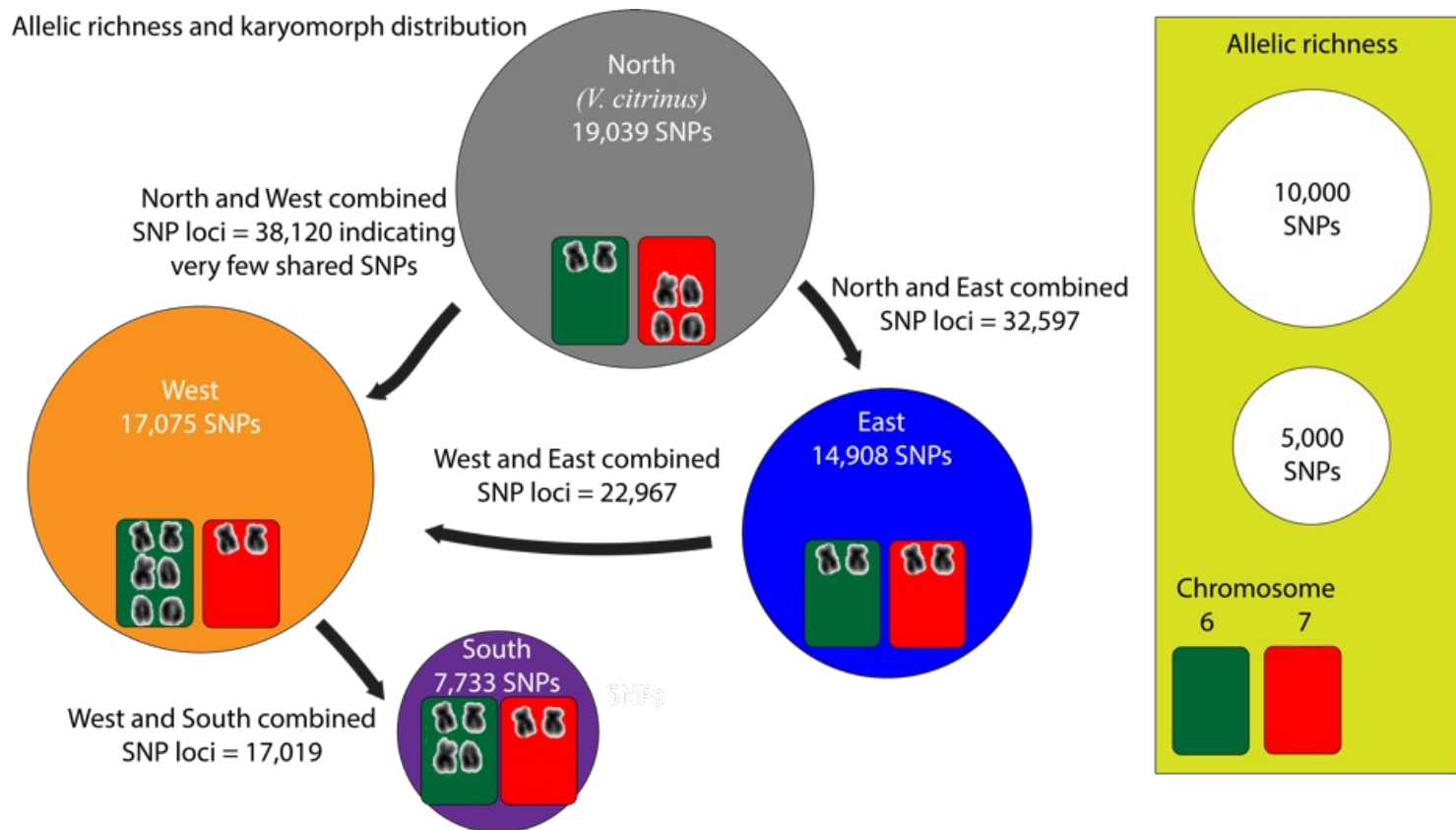
Inversion is non-homologous in North



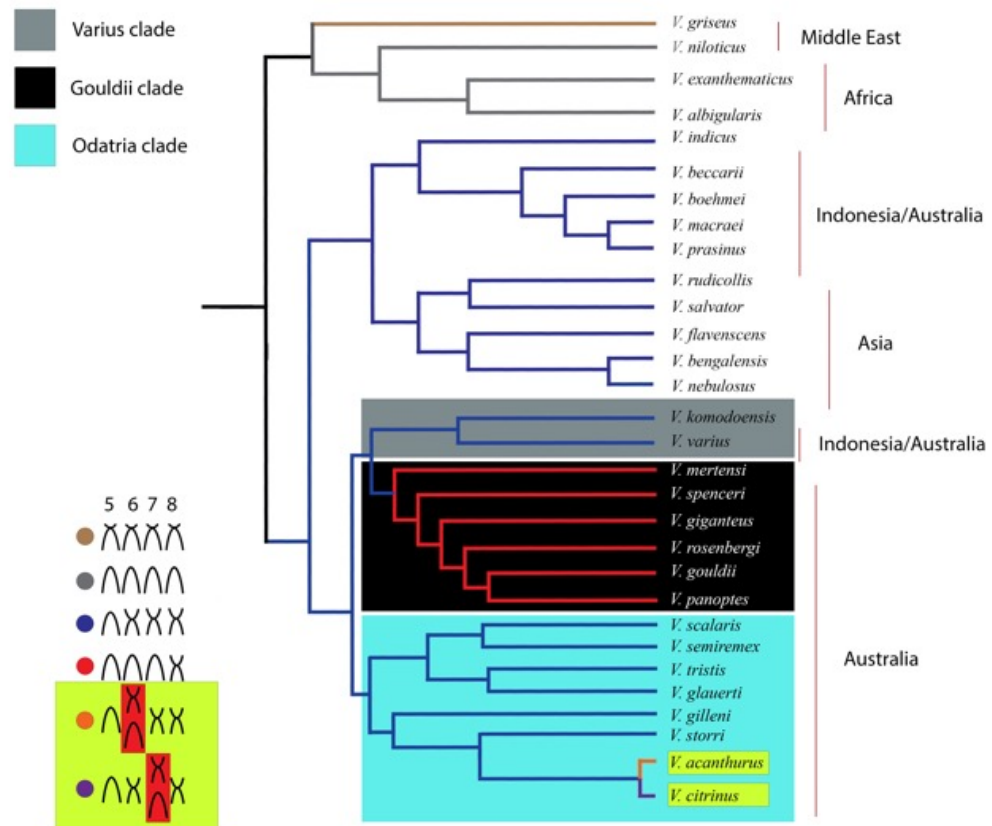
Maximum likelihood of concatenated SNPs



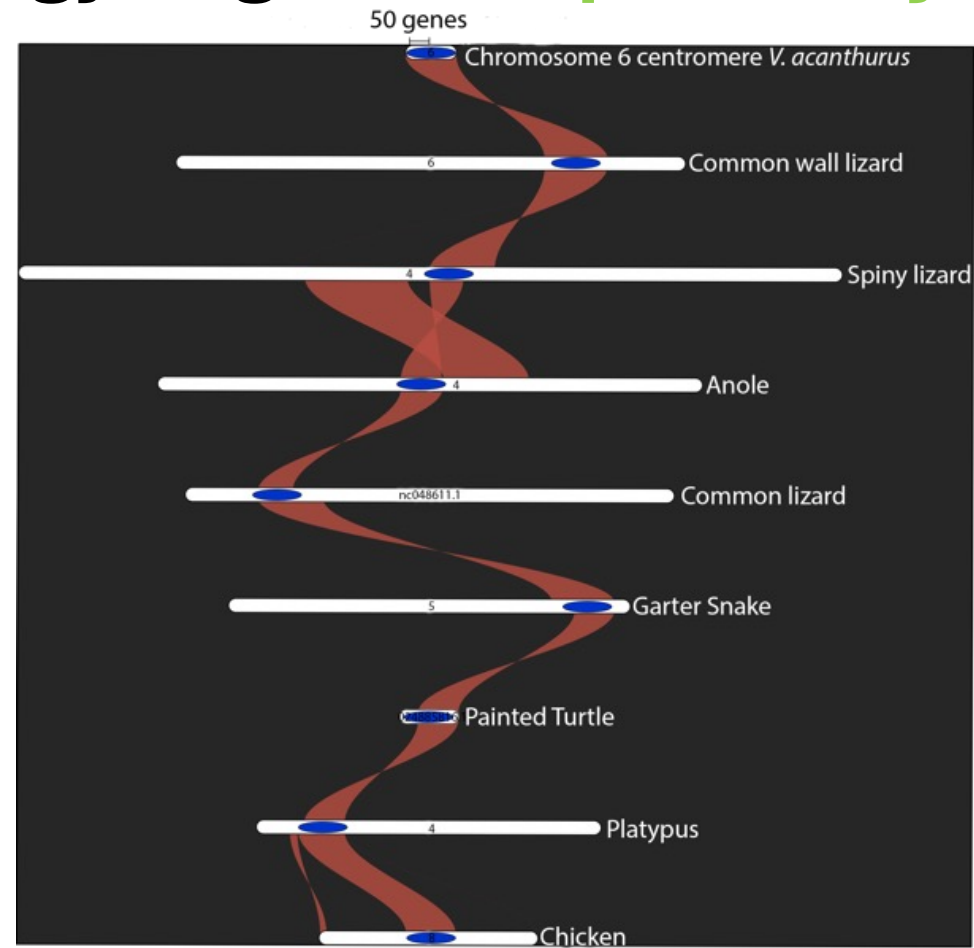
Correlation of chromosome morphology on allelic richness



Relevance for broader outlook



Orthology of genes captured by inversion



Fixed Allele Differences Associated With the Centromere Reveal Chromosome Morphology and Rearrangements in a Reptile (*Varanus acanthurus* BOULENGER)

Jason Dobry ^{*,1} Zexian Zhu,² Qi Zhou ^{2,3,4} Erik Wapstra,⁵ Janine E. Deakin,¹ and Tariq Ezaz ^{*,1}

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²MOE Laboratory of Biosystems Homeostasis and Protection and Zhejiang Provincial Key Laboratory for Cancer Molecular Cell Biology, Life Sciences Institute, Zhejiang University, Hangzhou, China

³Center for Reproductive Medicine, The 2nd Affiliated Hospital, School of Medicine, Zhejiang University, Hangzhou, China

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⁵School of Natural Sciences, University of Tasmania, Hobart, Australia

***Corresponding authors:** E-mails: Jason.dobry@canberra.edu.au; Tariq.Ezaz@canberra.edu.au.

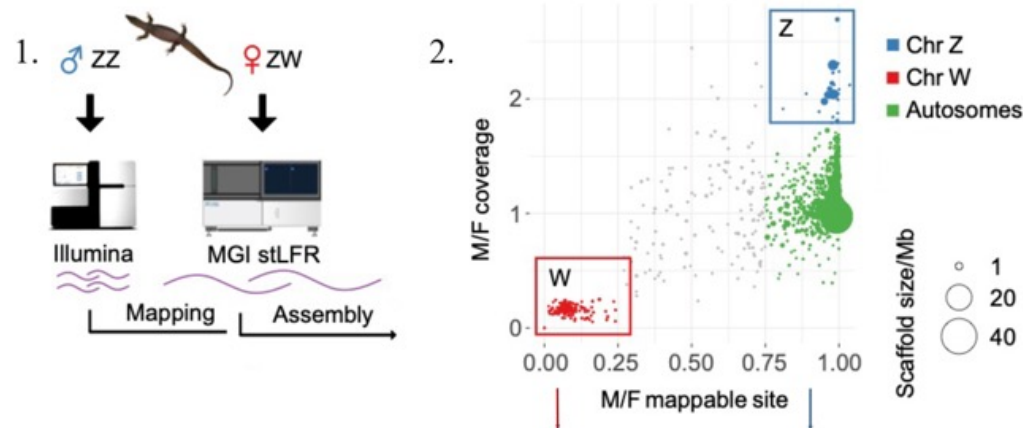
Associate editor: Xuming Zhou

Abstract

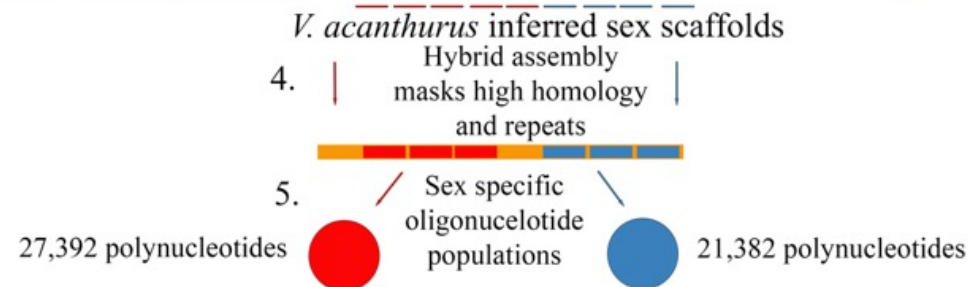
Chromosome rearrangements are often implicated with genomic divergence and are proposed to be associated with species evolution. Rearrangements alter the genomic structure and interfere with homologous recombination by isolating a portion of the genome. Integration of multiplatform next-generation DNA sequencing technologies has enabled putative identification of chromosome rearrangements in many taxa; however, integrating these data sets with cytogenetics is still uncommon beyond model genetic organisms. Therefore, to achieve the ultimate goal for the genomic classification of eukaryotic organisms, physical chromosome mapping remains critical. The ridge-tailed goannas (*Varanus acanthurus* BOULENGER) are a group of dwarf monitor lizards comprised of several species found throughout northern Australia. These lizards exhibit extreme divergence at both the genic and chromosomal levels. The chromosome polymorphisms are widespread extending across much of their distribution, raising the question if these polymorphisms are homologous within the *V. acanthurus* complex. We used a combined genomic and cytogenetic approach to test for homology across

Sex chromosome discovery

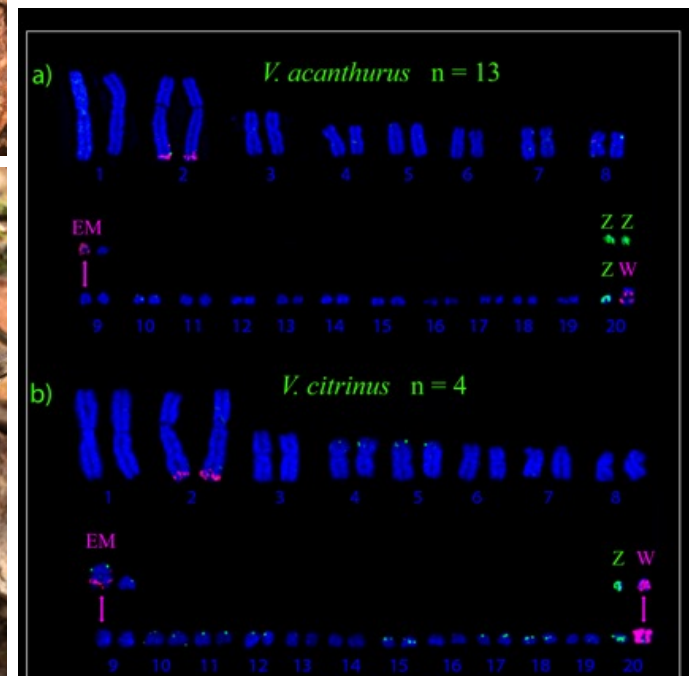
Methodology for development of sex specific DNA probes

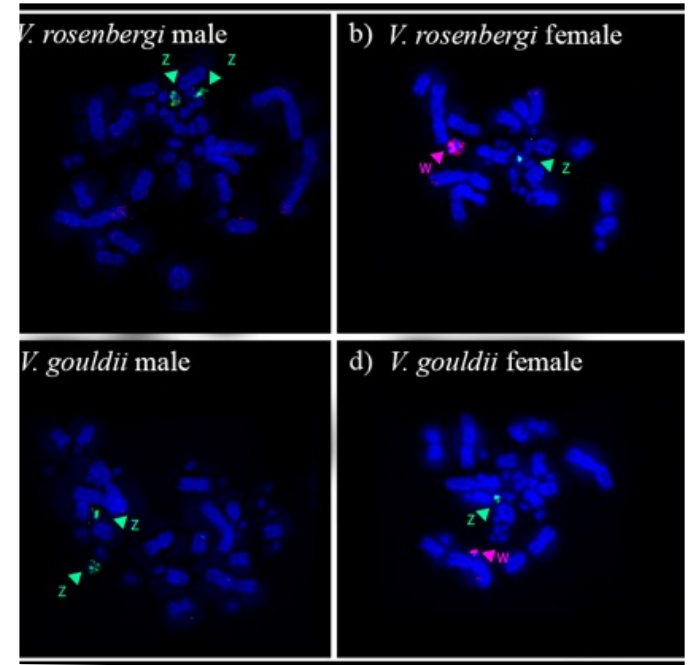


3. BLAST Komodo Dragon (*V. komodoensis*) Genome



FISH with inferred sex chromosome probes





Comparing the sex probes to other species with
Sex chromosome variation

gl.filter.sexlinked analysis yesterday

- Detected 23 females and 38 males.
- Building heterozygosity plots.
- Done building heterozygosity plots.
- ****FINISHED**** Total of analyzed loci: 301738.
- Found 241 sex-linked loci:
 - 220 W-linked loci
 - 20 sex-biased loci
 - 0 Z-linked loci
 - 1 ZW gametologs.
- And 301497 autosomal loci.

*****All loci have aligned to
W chromosome
scaffolds*****

Varanus s. acanthurus																		
Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko	Varanus_ko
Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position	Ending position	Starting position
LEN.1	LEN.2	LEN.3	LEN.4	LEN.5	LEN.6	LEN.7	LEN.8	LEN.9	LEN.10	LEN.11	LEN.12	LEN.13	LEN.14	LEN.15	LEN.16	LEN.17	LEN.18	LEN.19
CO V.R	CO V.Q	REF.TAG	VKO_scaffold.size	VKO_chrom	VAC_scaffoldh	scaffoldL	engL	Mapped Length	MeanCoverage	TotalCoverage	Mapped Percent	freq_s						
2.7NW_0253352	scaffold1 138,28	unplaced	9481	7	18173	1.00497705	18083	19.0715	14									
0.5NW_0253352	scaffold1 138,28	unplaced	9481	7	18173	1.00497705	18083	19.0715	14									
0.3NW_0253352	scaffold1 138,28	unplaced	2502	73	46973	1.00091625	8	46930	18.7515	23								
7.5NW_0253352	scaffold10 37,40	Chr4	1264	2	4529	1.00221287	9	4519	35.7459	NA								
0.1NW_0253352	scaffold10 37,40	Chr4	3137	77	49085	1.00155073	6	49009	15.6191	52								
0.0 0.7NW_0253352	scaffold101 1,46	Chr1,	1811	03	25109	1.01307242	3	24785	13.6856	63								
0.0 0.5NW_0253352	scaffold105 1,30	Chr1,	9481	7	18173	1.00497705	18083	19.0715	14									
4.8 0.5NW_0253353	scaffold1056 21, unplaced	scaf_175	1811	03	25109	1.01307242	3	24785	13.6856	63								
0.2NW_0253353	scaffold11 34,28	Chr6,7,8	2502	73	46973	1.00091625	8	46930	18.7515	23								
12. NW_0253353	scaffold11 34,28	Chr6,7,8	1227	2	3455	1.00115908	4	3451	28.1209	NA								
0.3NW_0253353	scaffold11 34,28	Chr6,7,8	1579	14	28666	1.00367634	2	28561	18.0864	17								
0.1 0.8NW_0253353	scaffold1113 1,03	Z	1299	95	23934	1.00146449	6	23899	18.3846	39								
0.1 1.1NW_0253353	scaffold1113 1,03	Z	1299	95	23934	1.00146449	6	23899	18.3846	39								



Summary

- Take advantage of available resources for data exploration
- Genome alignments can be powerful tools for targeting interesting patterns in SNP data
- Combining datasets can help complete the picture
- Listen to the data