



MACQUARIE  
University

# BIOL3110 Conservation & Ecological Genetics

## LECTURE 3: GENETIC VARIATION



# Assignment 1: Commentary

WORTH 5 % **DUE FRIDAY WEEK 3 (7/3/22) MONDAY 5PM**



## Criterion

## Marks

Get it between 500-600 words

1

Relevance of paper selection

1

Presentation, spelling, grammar

1

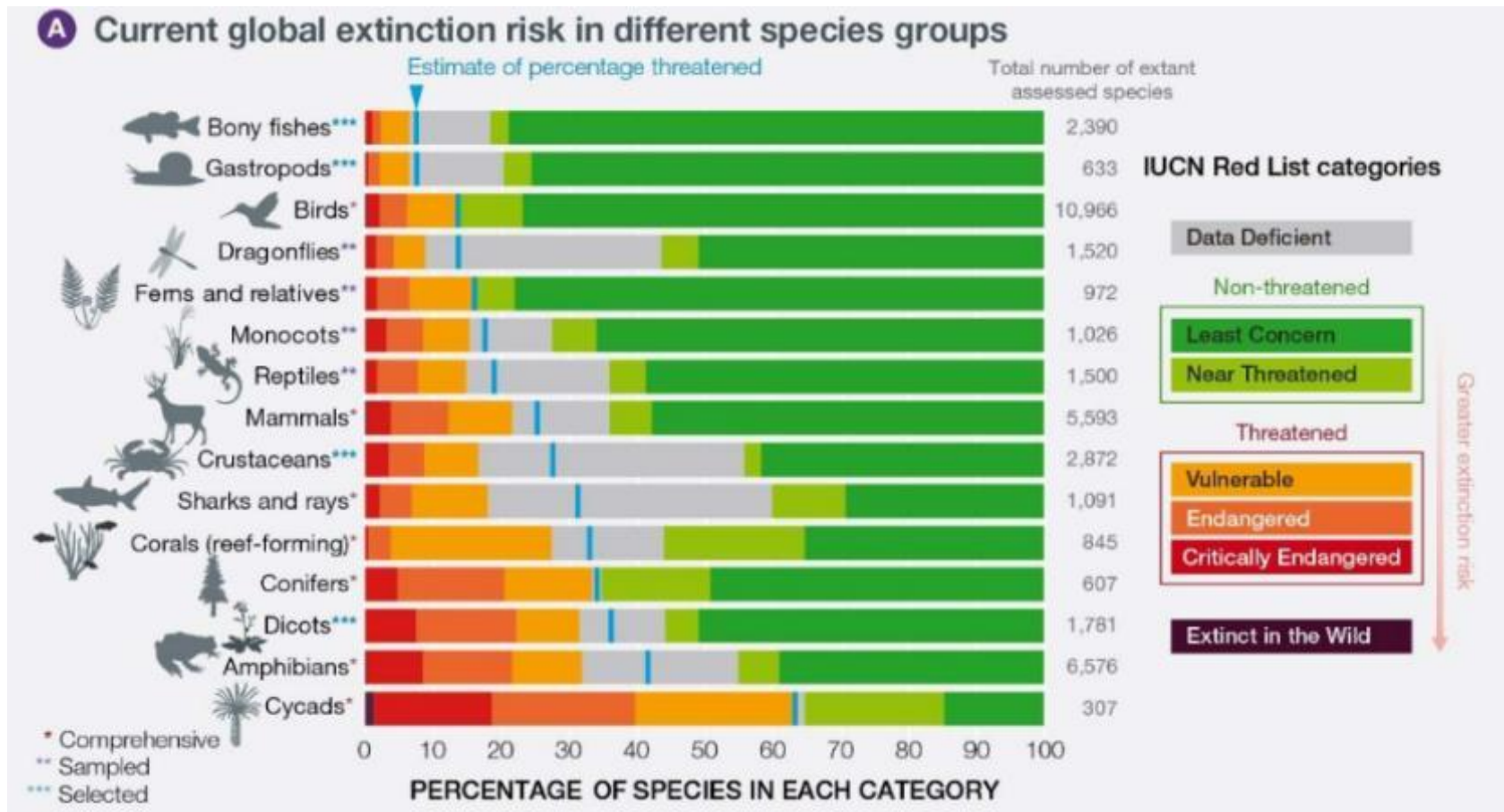
Genuine effort to interpret the paper

2

## TIPS:

- Don't include quotations
- Cite the focal paper (at the end)
- Can include an image – plus SHORT caption
- Hone in on the relevance of VG for conservation
- Choose a paper that you find most interesting
- Consider your seminar!

# Globally, > one million species are threatened



IPBES



Intergovernmental Science-Policy Platform on Biodiversity and Ecosystem Services (2019)

# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## IMPORTANCE (ACCORDING TO IUCN):

1. Key constituent of biodiversity;
2. Required for adaptation
3. Necessary for reducing inbreeding



## TODAY:

- Review what it is and where it comes from
- Its role in guiding conservation
- Methods for measuring  $V_G$  >>
- Some examples of cons relevance





# Genetic Diversity ( $V_G$ )

WHAT IS IT?

SIMPLY: DIFFERENCES IN DNA SEQUENCES

```
170 180 190
ATCTCTTGGCTCCAGCATCGATGAAGAACGCA
TCATTTAGAGGAAGTAAAGTCGTAACAAGGT
GAAGTGTCAAAACCTTTTAAACAACGGATCTCTT
TGTTGCTTCGGCGGCGCCCGCAAGGGTGCCCG
GGCCTGCCGTGGCAGATCCCCAACGCCGGGCC
TCTCTTGGCTCCAGCATCGATGAAGAACGCAG
CAGCATCGATGAAGAACGCAGCGAAACGCGAT
CGATACTTCTGAGTGTTCCTTAGCGAACTGTCA
CGGATCTCTTGGCTCCAGCATCGATGAAGAAC
ACAACGGATCTCTTGGCTCCAGCATCGATGAA
CGGATCTCTTGGCTCCAGCATCGATGAAGAAC
GATGAAGAACGCAGCGAAACGCGATATGTAAT
```

$V_G$   
individual  
**loci**

This week  
(Lectures 3 & 4)



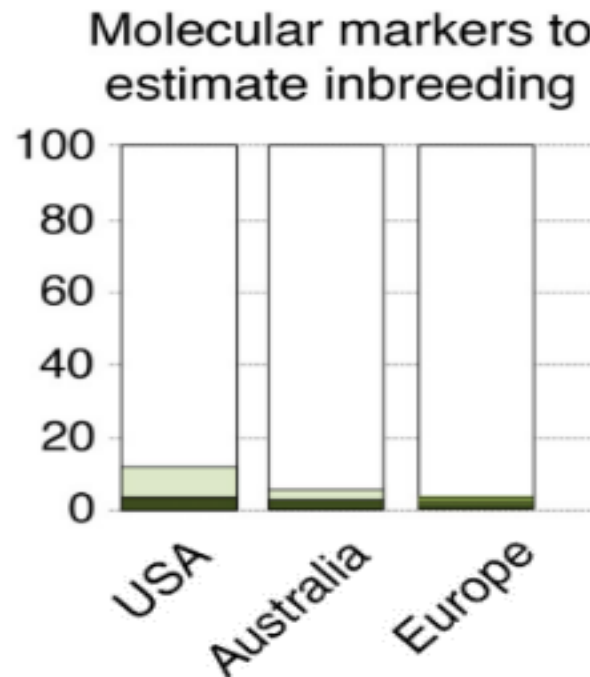
**Whole Organism  
(Phenotype)**

$V_G$   
individual  
**traits**

Next week  
(Lectures 5 & 6)

# Are genetic processes appropriately considered

> 300 threatened species recovery plans assessed for consideration of evolutionary factors (genetics)



Pierson *et al* (2016). *Frontiers in Ecology and the Environment* 14: 433-440

# Genetic Diversity ( $V_G$ )

## MEASUREMENT INDICES

For individual loci:



- **Polymorphism ( $P$ )**

= proportion of polymorphic loci;



- **Average heterozygosity ( $H$ )**

= proportion of heterozygous loci per individual;

- **Allelic diversity ( $A$ )**

= average number of different alleles per locus.



# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## POLYMORPHISM (P)

Simple ratio of the number of polymorphic loci

Eg:

Locus	Allele frequencies		
A	0.56	0.33	0.11
B	0.70	0.20	0.10
C	0.80	0.20	
D	1.0		
E	1.0		

$$\frac{\sum N_P}{N_T}$$

Where :  $N_P$  = number of polymorphic loci  
 $N_T$  = total number of loci

$$P = 3/5 = 0.6$$



# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## HETEROZYGOSITY (H)

For a single locus

(H for monomorphic locus = 0)

Eg:

Locus	Alleles & frequencies		
	A1	A2	A3
ADA	0.56	0.33	0.11

$$\begin{aligned} H &= 1 - (0.56^2 + 0.33^2 + 0.11^2) \\ &= 1 - (0.4334) \\ &= \mathbf{0.5664} \end{aligned}$$

$$H = 1 - \sum P_i^2$$

Where  $P_i$  = frequency of allele  $i$



# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## AVERAGE HETEROZYGOSITY (H)

For multiple loci

Simple average across them Eg:

$$\frac{\sum H_i}{N_T}$$

Where :  $H_i = H$  at locus  $i$   
 $N_T$  = total number of loci

Locus	Alleles & frequencies			H
	A1	A2	A3	
ADA	0.56	0.33	0.11	0.564
BDA	1.00	0	0	0.000

$$\begin{aligned} \text{Average } H &= (0.564 + 0.000)/2 \\ &= \mathbf{0.282} \end{aligned}$$

# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## ALLELIC DIVERSITY (A)

Simple average number of alleles per locus. Eg:

Locus	Allele frequencies			
A	0.60	0.25	0.13	0.12
B	0.70	0.10	0.10	0.10
C	0.50	0.30	0.20	
D	0.55	0.45		
E	0.85	0.15		
F	0.90	0.10		

$$A = \frac{\sum(N_A)}{N_T}$$

Where:

$N_A$  = number of different alleles across all loci

$N_T$  = total number of loci examined

$$A = 17/6 = 2.83$$

# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE



## AFRICAN LION POPULATION

(Example Chap 3 in  
textbook)

Calculate:

Polymorphism (P)

Average Heterozygosity (H)

Allelic diversity (A)

Locus	Allele frequencies			H
ADA	0.56	0.33	0.11	<b>0.564</b>
DIAB	0.61	0.39		<b>0.476</b>
ESI	0.88	0.12		<b>0.211</b>
GPI	0.85	0.15		<b>0.255</b>
GPT	0.89	0.11		<b>0.196</b>
MPI	0.92	0.08		<b>0.147</b>
20 others	1.00			<b>0.00</b>

# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE



6 of 26 loci polymorphic:

$$P = \frac{6}{26} = \mathbf{0.23}$$

$H$  for the **average individual**:

$$\frac{\sum H}{26} = \frac{(0.564 + 0.476 + 0.211 \dots)}{26} = \mathbf{0.071}$$

**Allelic diversity ( $A$ ):**

$$\frac{[(1 \times 3) + (5 \times 2) + (20 \times 1)]}{26} = \frac{33}{26} = \mathbf{1.27}$$

Hence, an average of **1.27 alleles** per **locus**

Locus	Allele frequencies			H
ADA	0.56	0.33	0.11	<b>0.564</b>
DIAB	0.61	0.39		<b>0.476</b>
ESI	0.88	0.12		<b>0.211</b>
GPI	0.85	0.15		<b>0.255</b>
GPT	0.89	0.11		<b>0.196</b>
MPI	0.92	0.08		<b>0.147</b>
20 others	1.00			<b>0.00</b>

Individual H

- is it close to HWE expectations?
- What does this imply?



# *Panthera leo*

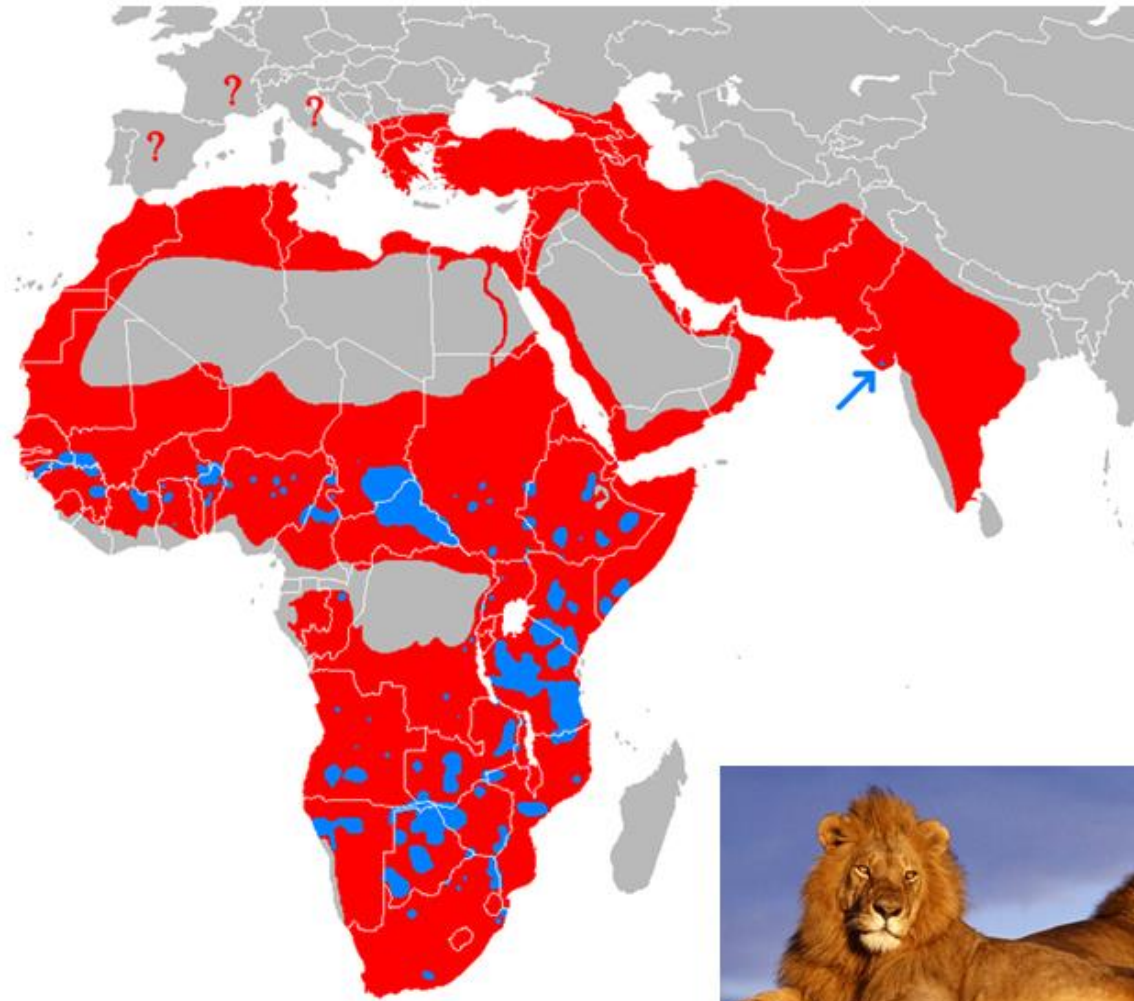


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Distribution:

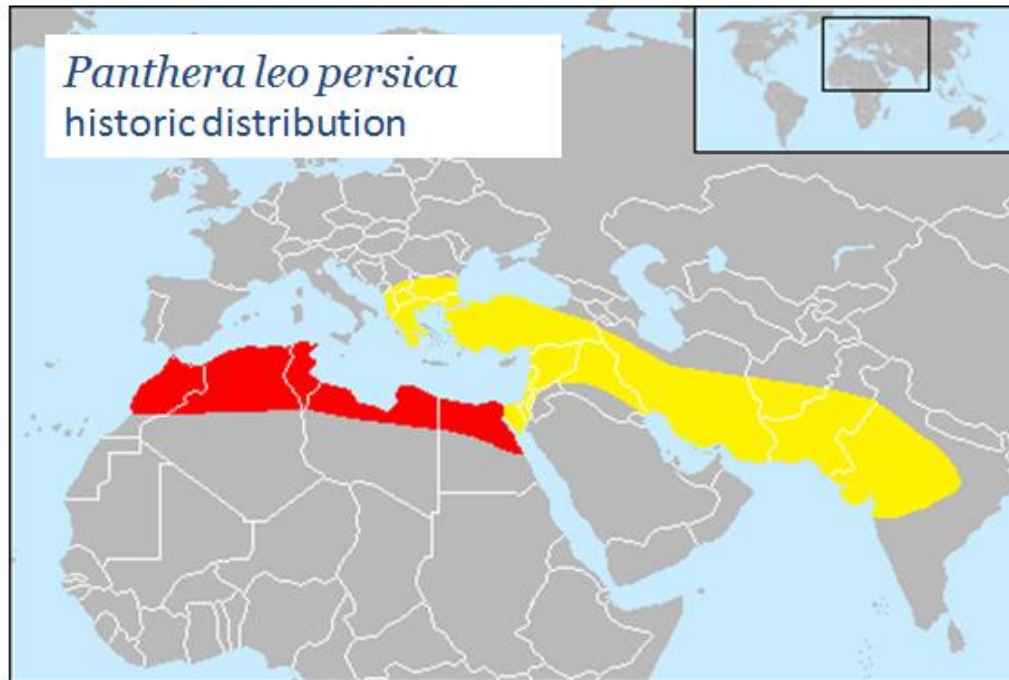
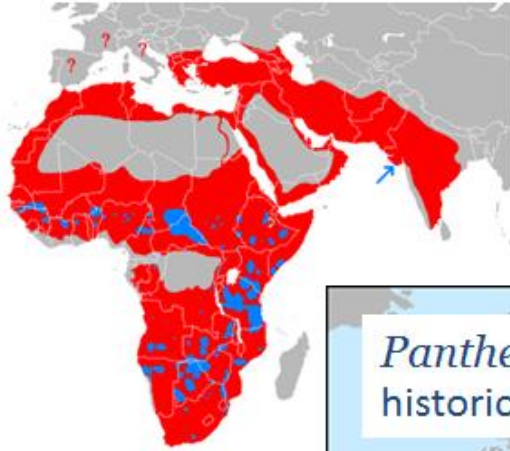
**Historic**

**Current**

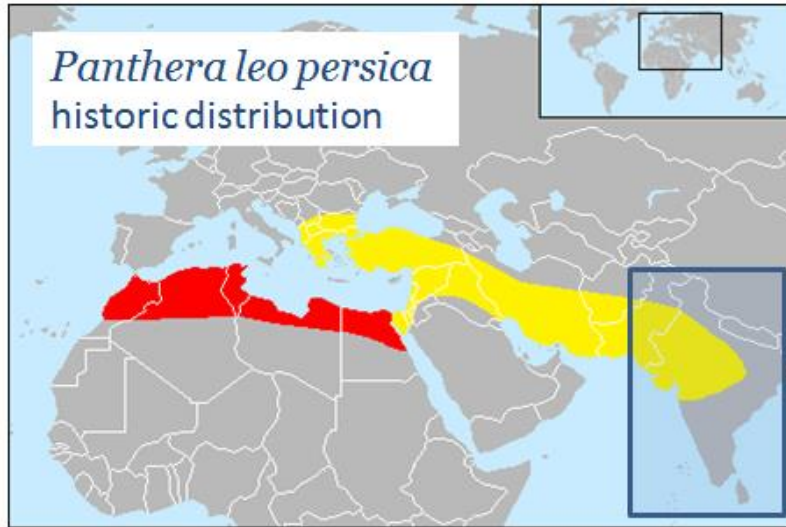


# *Panthera leo persica*

## Asiatic lion



# *Panthera leo persica*

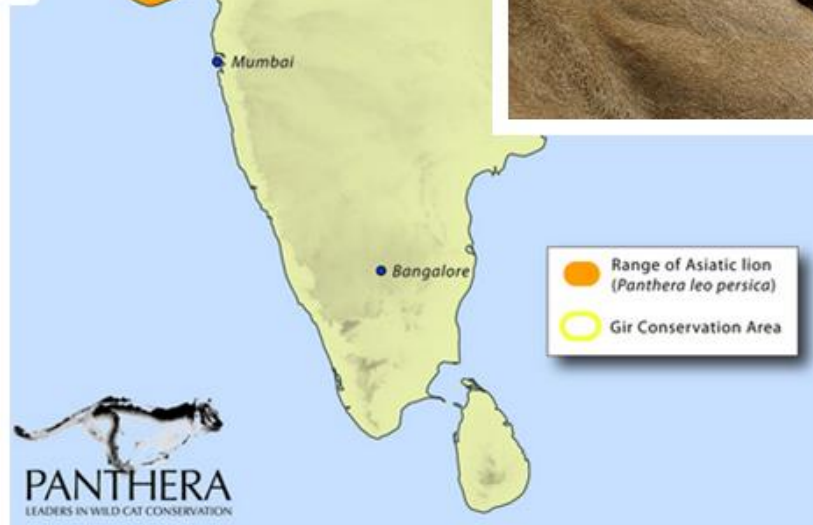


## Gir Forest National Park:

**1974:** 180 individuals

**2010:** 411 individuals

- 97 adult males
- 162 adult females
- 75 sub-adults
- 77 cubs








# *Panthera leo persica*

## Genetic diversity

50 allozyme (protein) loci  
DNA fingerprints



	Allozymes		DNA fingerprints
	P 	H 	H
Asiatic lion	0.00	0.000	0.038
African lion (outbred)	0.04-0.11	0.015-0.300	0.450

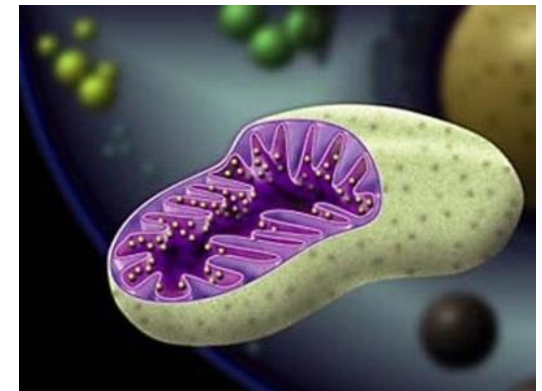
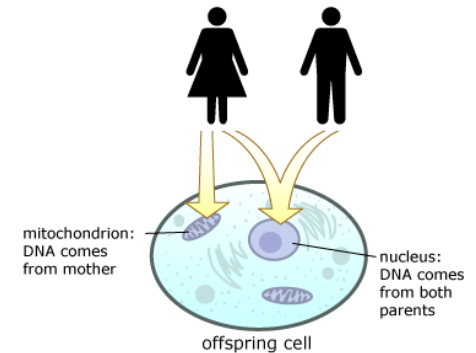
- Indicative of extreme **bottleneck** in recent past ( $N \sim 20$  in early 1900's) 
- Problematic for inbreeding depression, lack of population resilience and reduced adaptive potential

# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## ASSESSMENT AT SINGLE LOCUS LEVEL

- **Proteins** (allozyme electrophoresis)
- **Nuclear DNA** amplification via PCR
  - Microsatellites (sequence repeats)
  - SNPs (single nucleotide polymorphism)
  - AFLP (amplified fragment length polymorphism)
  - RAPD (Randomly amplified polymorphic DNA)
  - Sequencing
  - Plus other techniques...  
(see Box 3.3 Frankham *et al.*)
- **Mitochondrial DNA**





# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

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## MICROSATELLITES

- Repeats of 1-5bp sequences
  1. Simple Sequence Repeats (STRs)
  2. Short Tandem Repeats (STRs)

6 Repeats of 2bp segment e.g: **X**CACACACACACAY**Y**  
**X**GTGTGTGTGTGT**Y**

- **Highly variable**
- Mostly **neutral DNA** (non-coding regions)
- Potential for **non-invasive sampling**

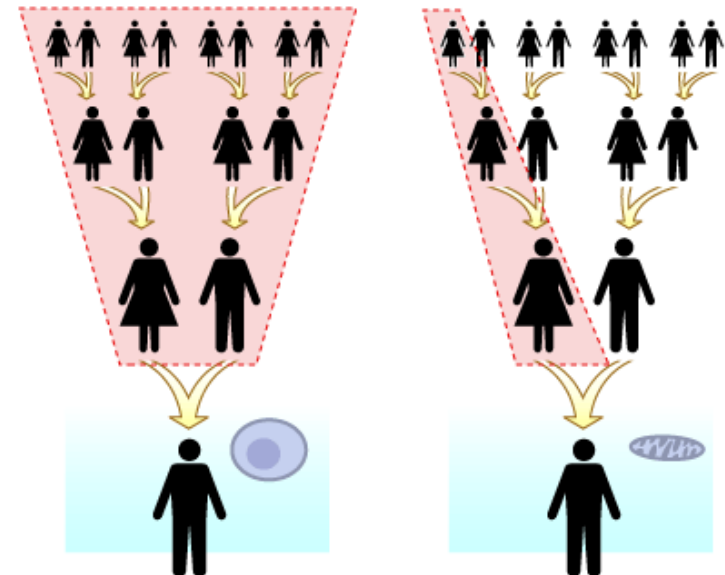
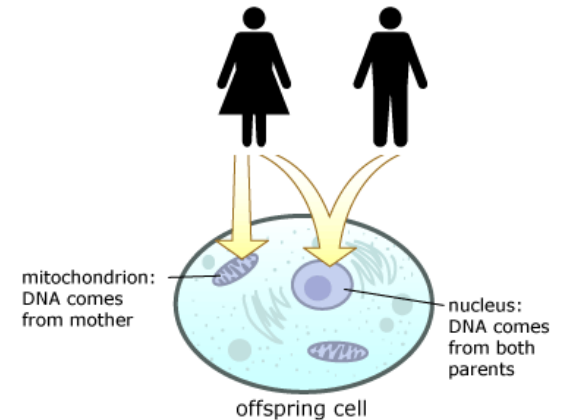
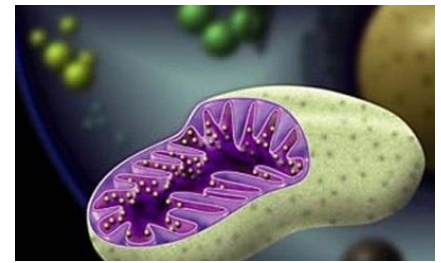
# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## MITOCHONDRIAL DNA

- Haploid maternally-inherited DNA
- No recombination
- Useful in cons gen to:
  1. Resolve taxonomic uncertainties
  2. Define management units (haplotypes)
  3. Understand species biology

**DNA** in plant  
chloroplasts is equivalent

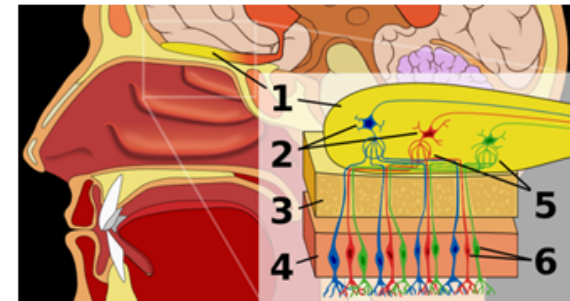


# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## MAJOR HISTOCOMPATIBILITY COMPLEX

- Code for proteins for surface antigens in 2 classes:
  - Class I Recognize intracellular pathogens and 'tag' the cell for immune system components (T-cells, phages etc).
  - Class II
- Highly polymorphic loci with very high allelic diversity
- Sexual selection implicated in maximising population heterozygosity: mate choice for MHC diversity (via olfactory cues in humans)



# Genetic Diversity ( $V_G$ )

AKA GENETIC VARIATION OR VARIANCE

## MHC DIVERSITY & DEVIL FACIAL TUMORS (DFTD)

Historically low  $V_G$  in MHC:



## Recovery implications:

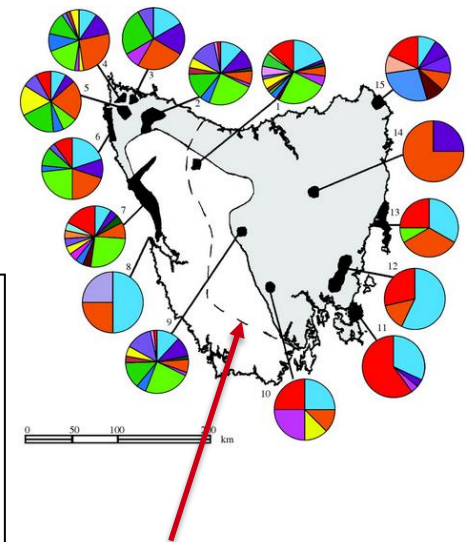
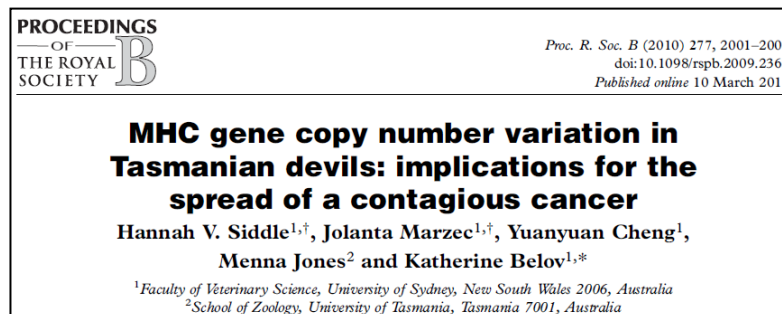
Most DFT cells express MHC classes 1 & 2

20% of devils only have class 1 or 2 genes

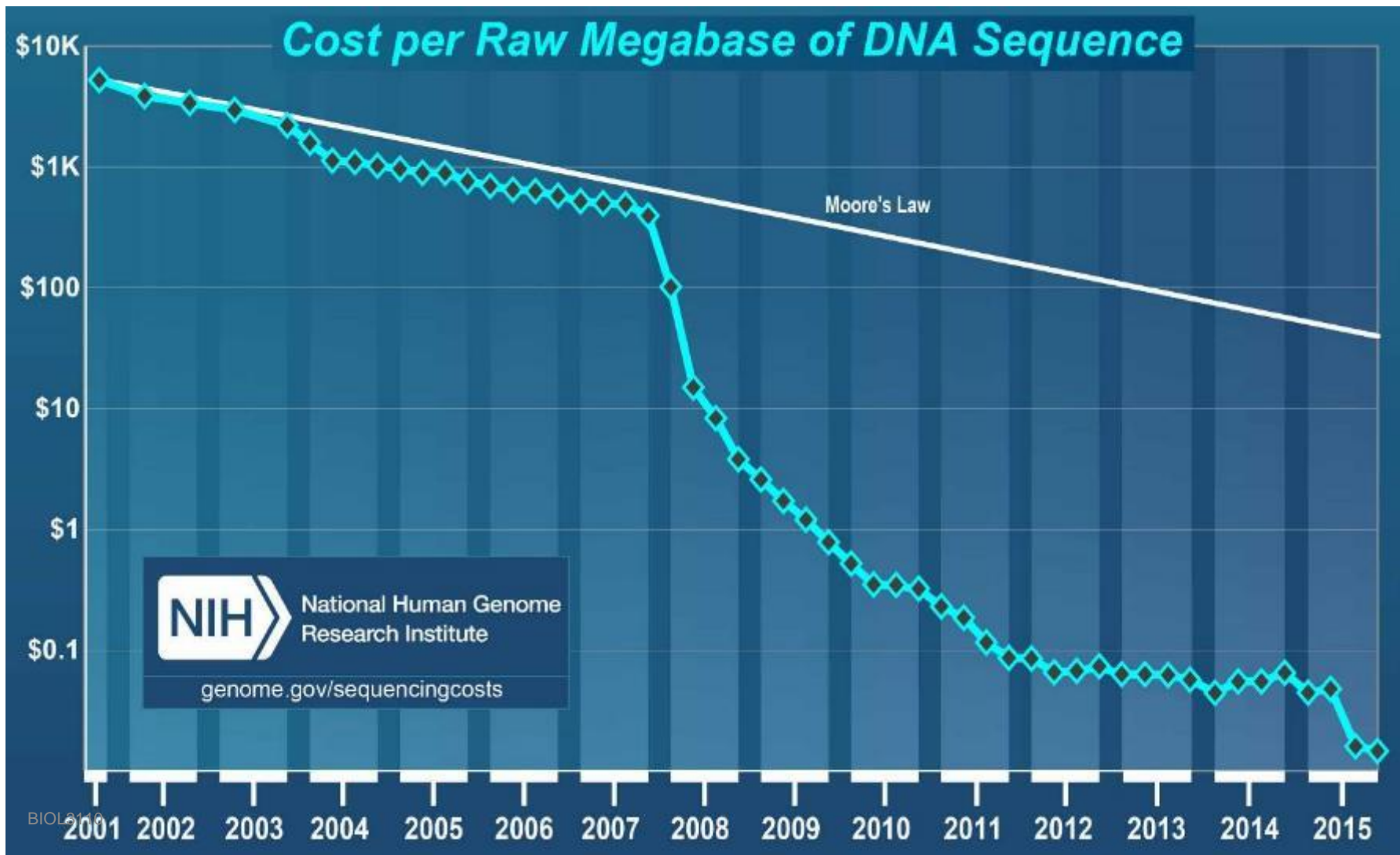
MHC1 devils may detect DFT cells

expressing MHC1&2

Counterintuitive example

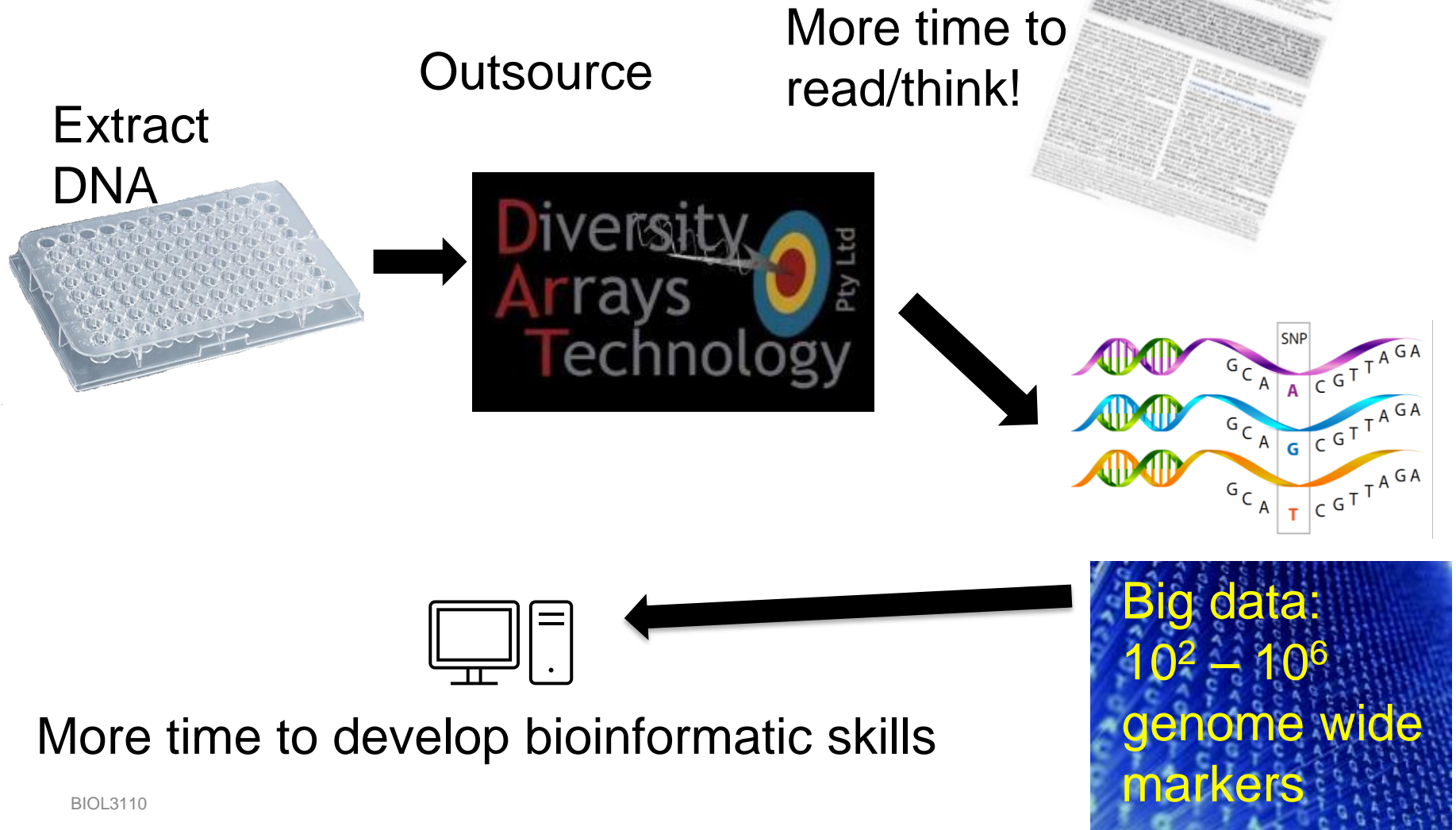


Extent of spread





# Modern genomic workflow

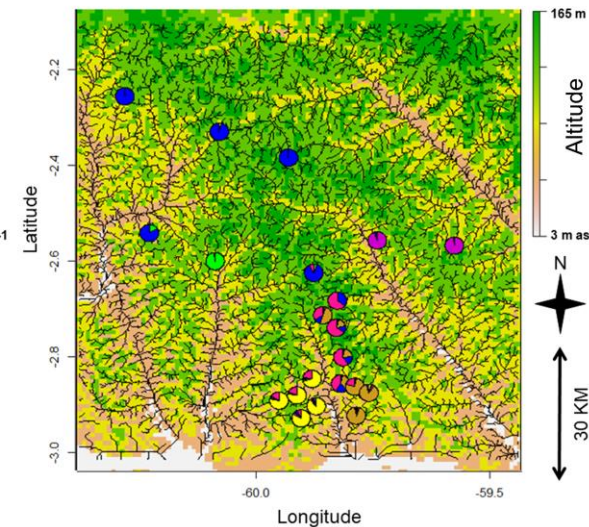
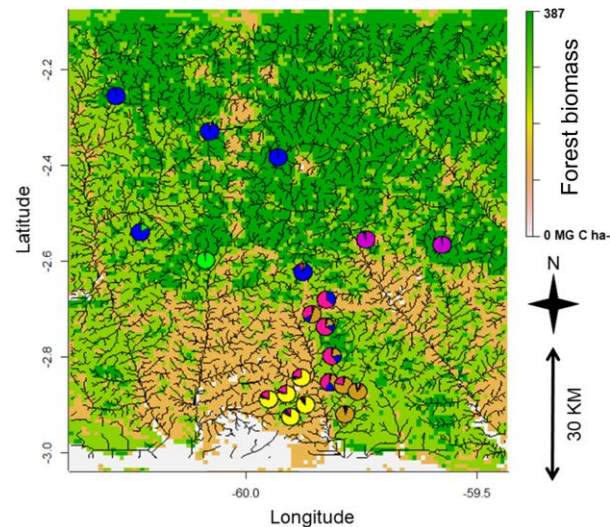
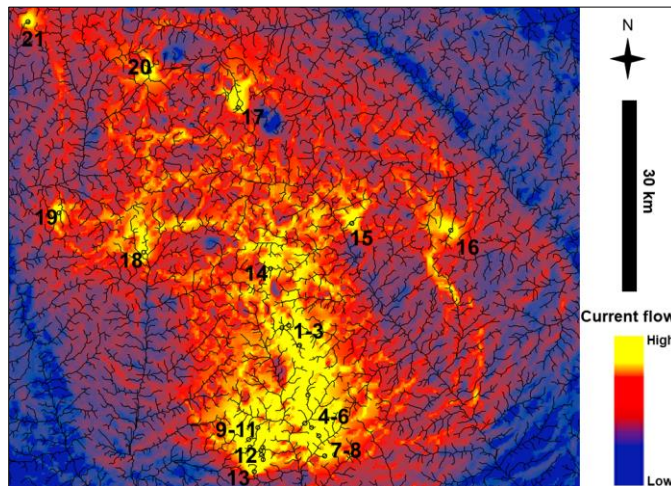
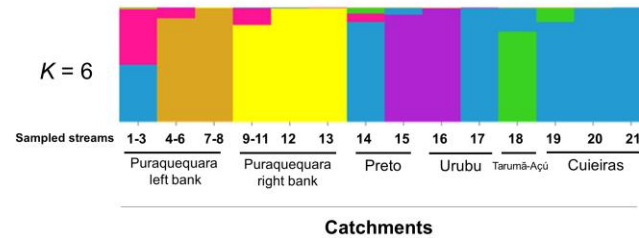
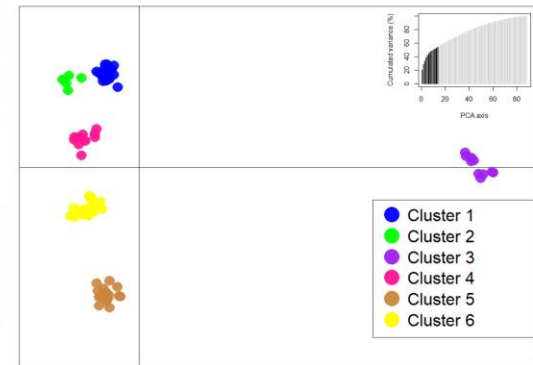
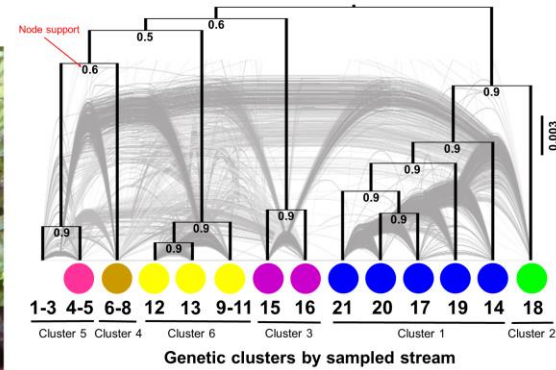


# 1) Biodiversity patterns

- diversity, distribution, distinctiveness

## 2) Estimating dispersal, genetic connectivity

### 3) Identifying processes



# Next lecture:

Characterising  $V_G$  for single loci  
More detail & examples

