

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

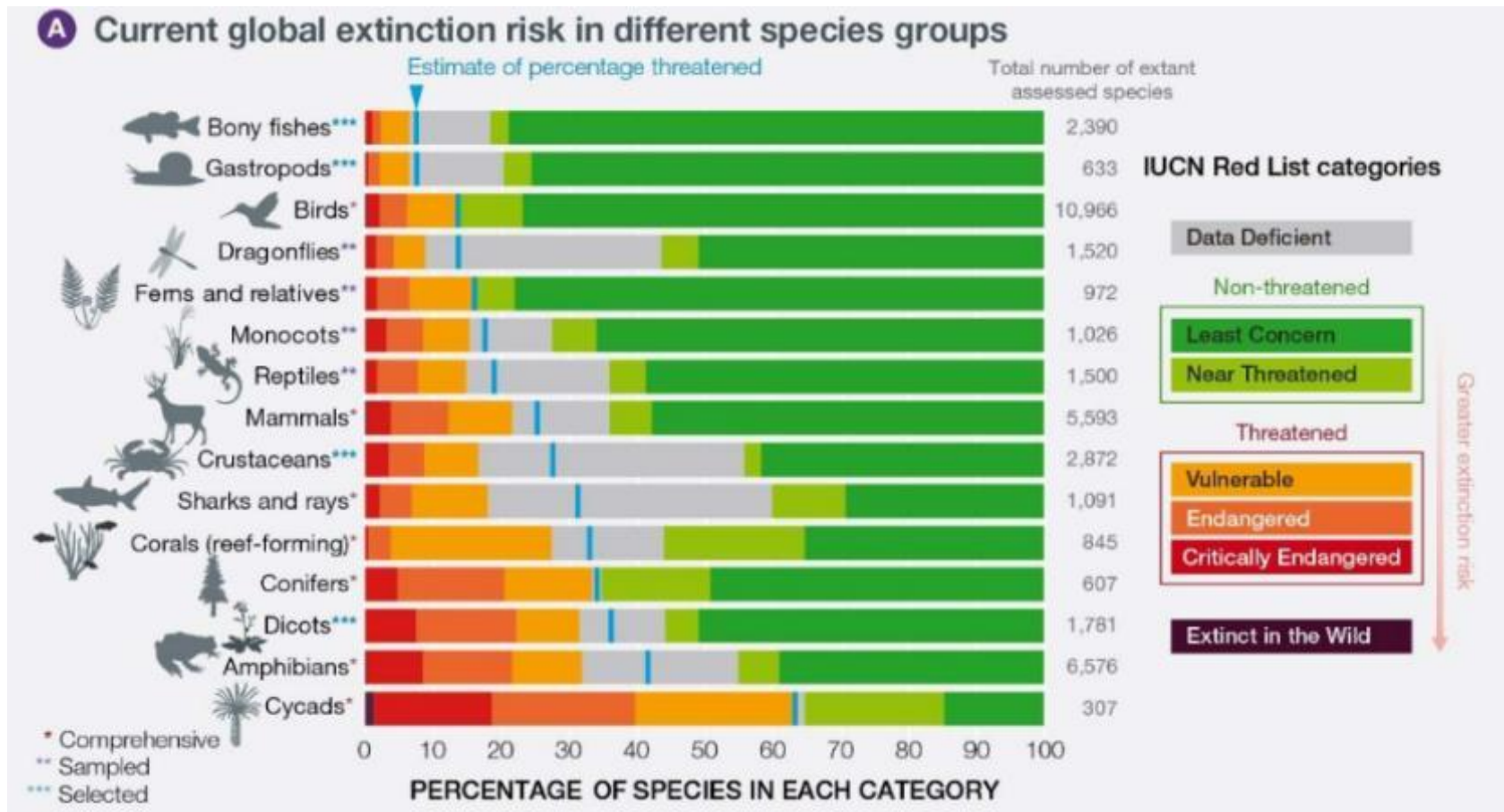
Genuine effort to interpret the paper

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
GAACTGTCAAAACCTTTTAAACAACGGATCTCTT
TGTTGCTTCGGCGGCGCCCGCAAGGGTGCCCG
GGCCTGCCGTGGCAGATCCCCAACGCCGGGCC
TCTCTTGGCTCCAGCATCGATGAAGAACGCAG
CAGCATCGATGAAGAACGCAGCGAAACGCGAT
CGATACTTCTGAGTGTTCCTTAGCGAACTGTCA
CGGATCTCTTGGCTCCAGCATCGATGAAGAAC
ACAACGGATCTCTTGGCTCCAGCATCGATGAA
CGGATCTCTTGGCTCCAGCATCGATGAAGAAC
GATGAAGAACGCAGCGAAACGCGATATGTAAT
```

V_G
individual
loci

This week
(Lectures 3 & 4)



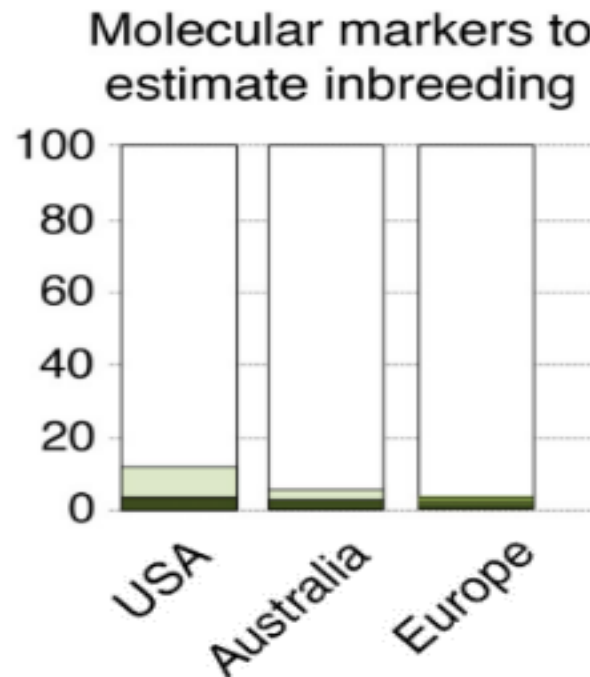
**Whole Organism
(Phenotype)**

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



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For individual loci:



- **Polymorphism (P)**

= proportion of polymorphic loci;

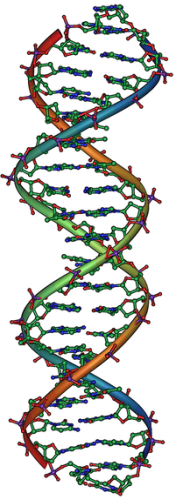
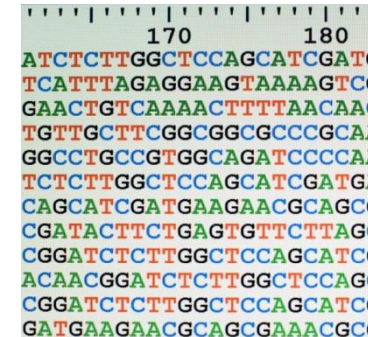


- **Average heterozygosity (H)**

= 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

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

Where P_i = frequency of allele i



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

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	0.564
DIAB	0.61	0.39		0.476
ESI	0.88	0.12		0.211
GPI	0.85	0.15		0.255
GPT	0.89	0.11		0.196
MPI	0.92	0.08		0.147
20 others	1.00			0.00

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	0.564
DIAB	0.61	0.39		0.476
ESI	0.88	0.12		0.211
GPI	0.85	0.15		0.255
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20 others	1.00			0.00

Individual H

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

Panthera leo

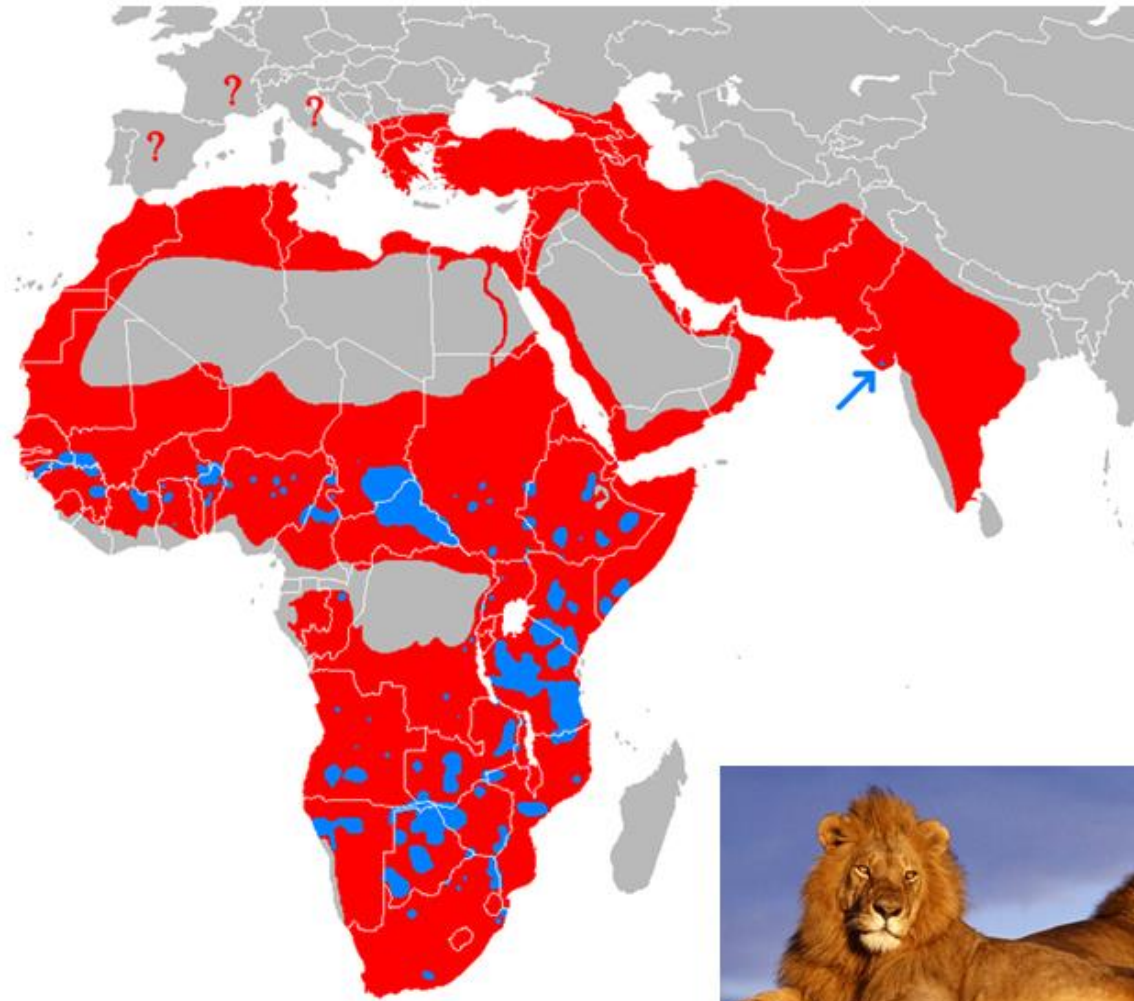


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Distribution:

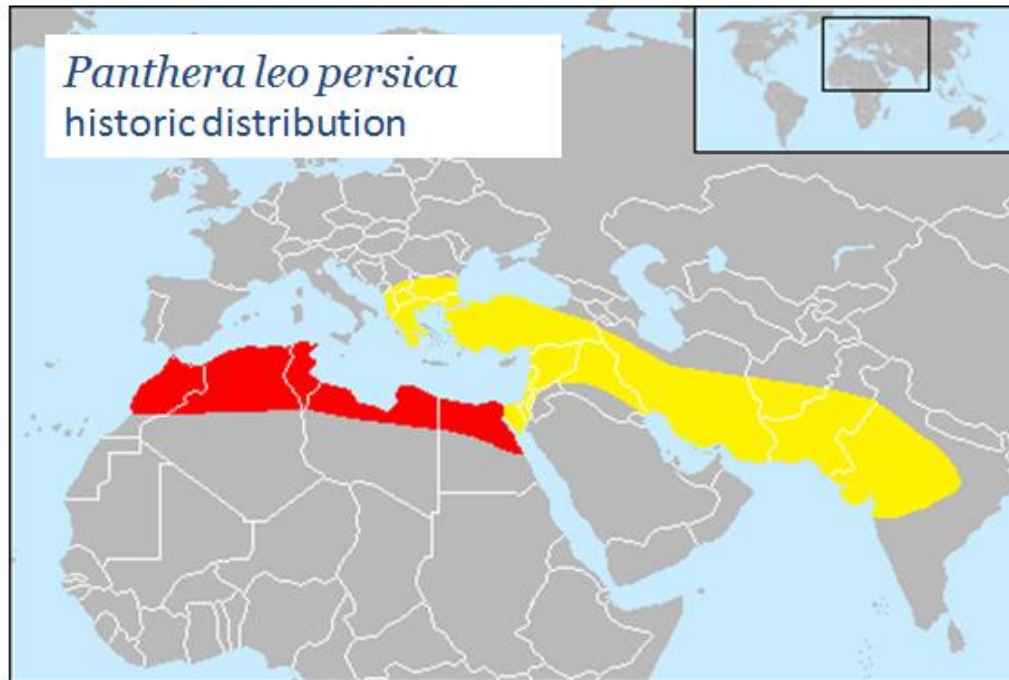
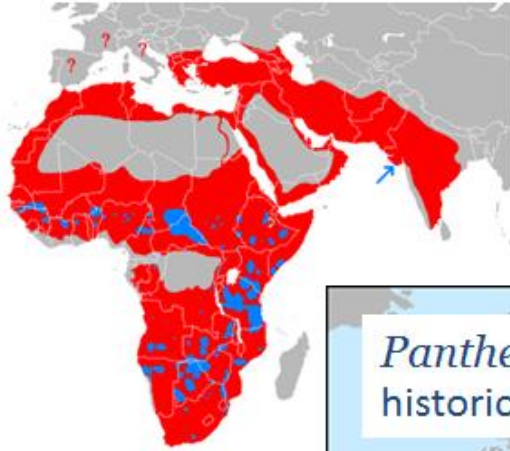
Historic

Current

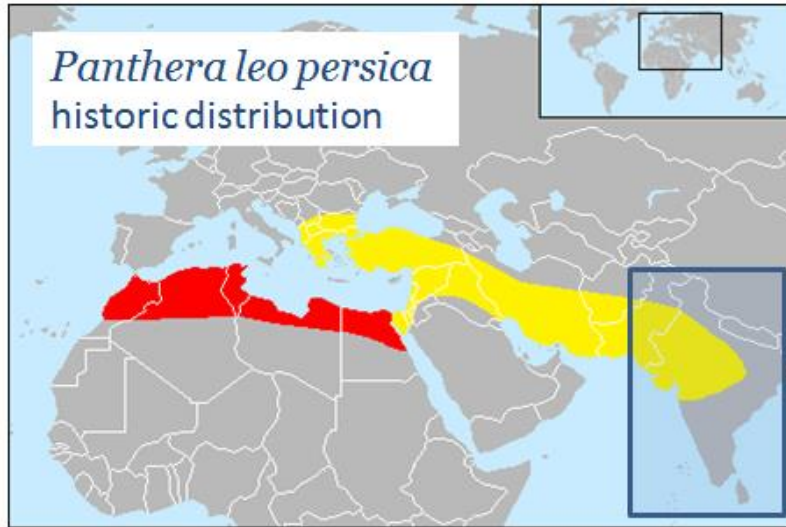


Panthera leo persica

Asiatic lion



Panthera leo persica

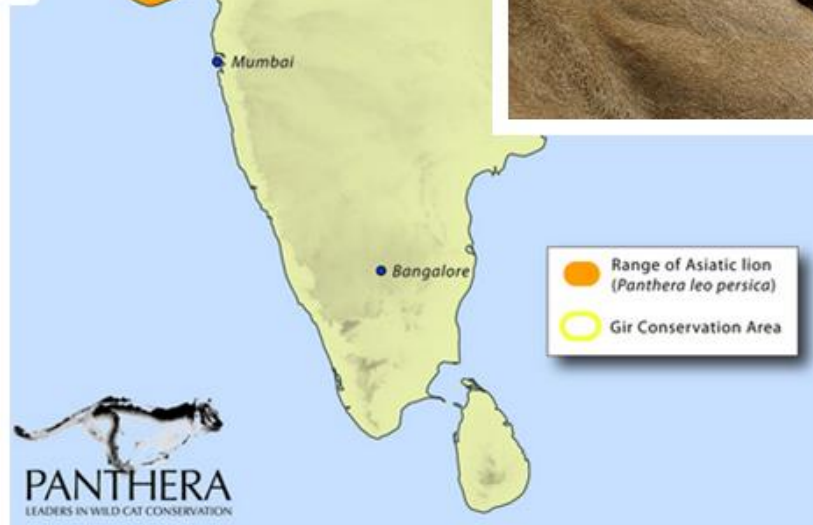


Gir Forest National Park:

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

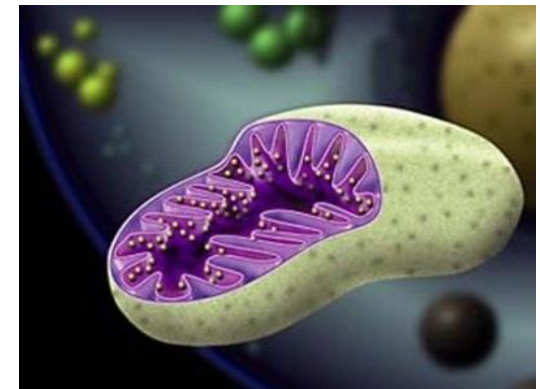
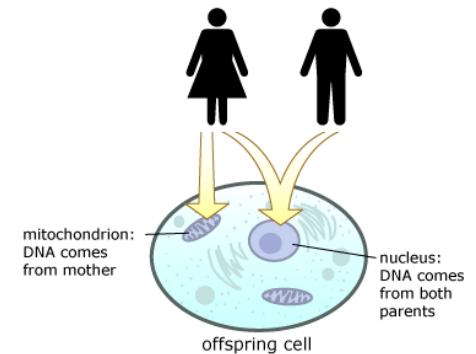
- Indicative of extreme **bottleneck** in recent past ($N \sim 20$ in early 1900's) 
- Problematic for inbreeding depression, lack of population resilience and reduced adaptive potential

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

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**
XGTGTGTGTGTGT**Y**

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

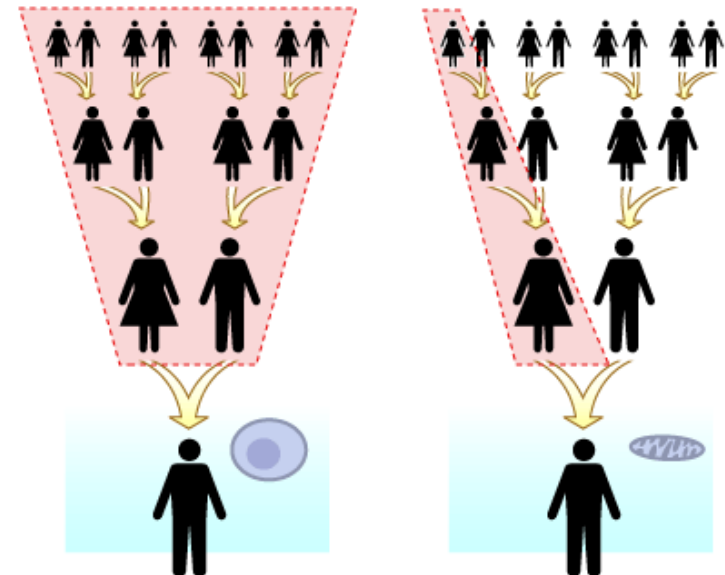
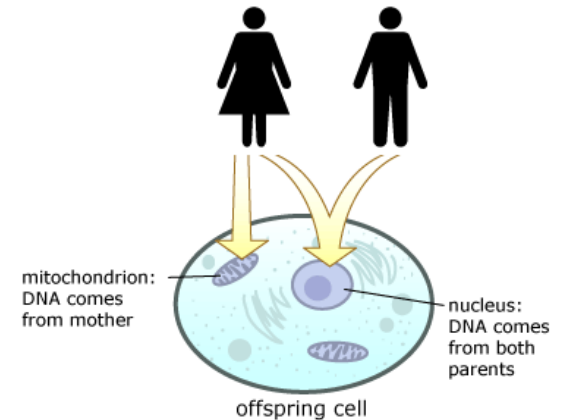
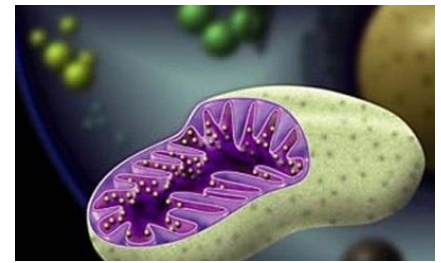
Genetic Diversity (V_G)

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



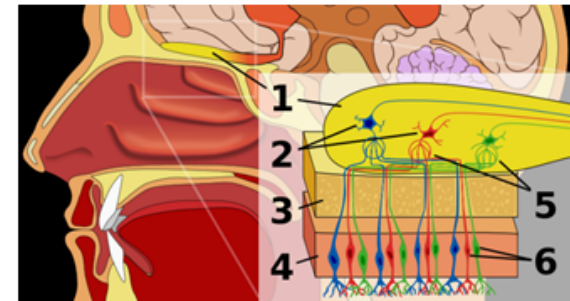
Genetic Diversity (V_G)

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)

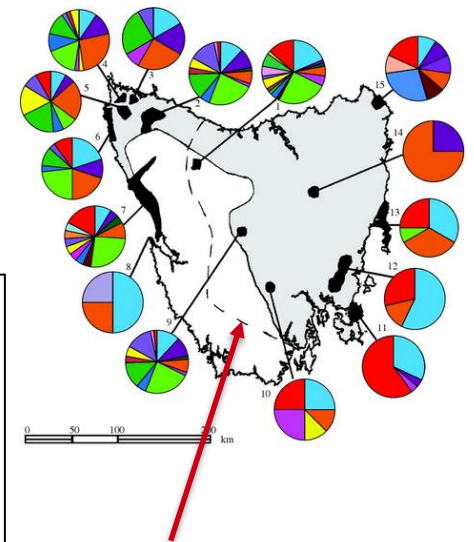
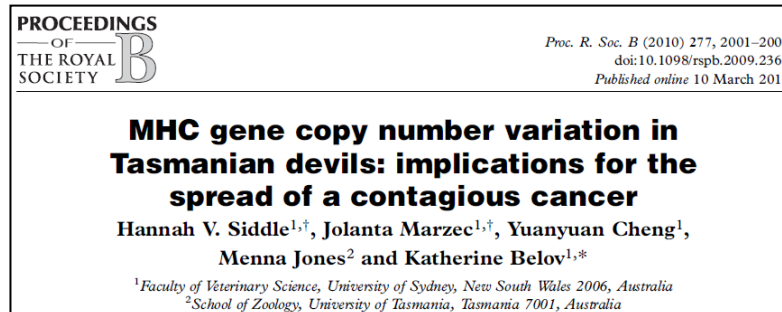
Historically low V_G in MHC:



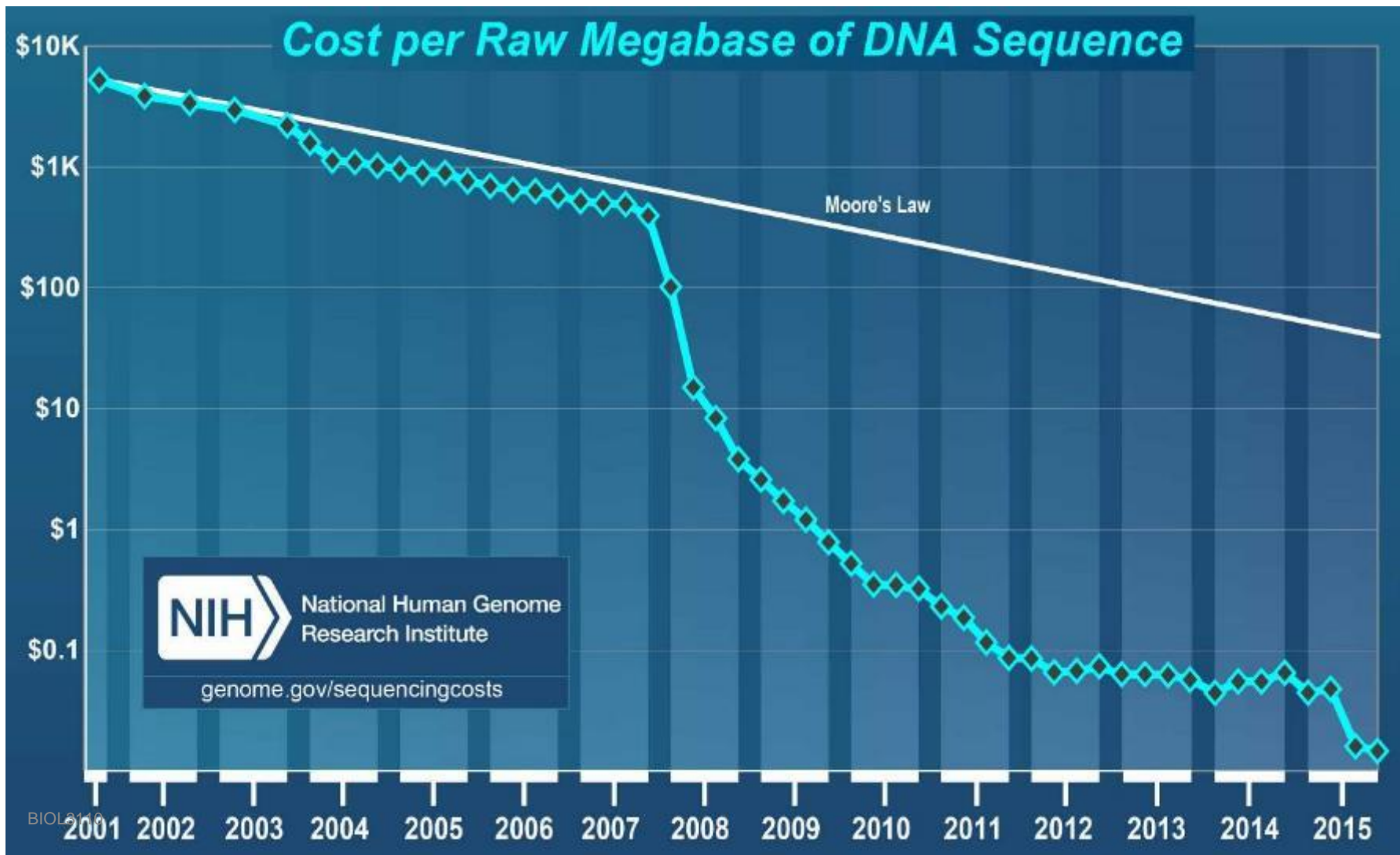
Recovery implications:

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

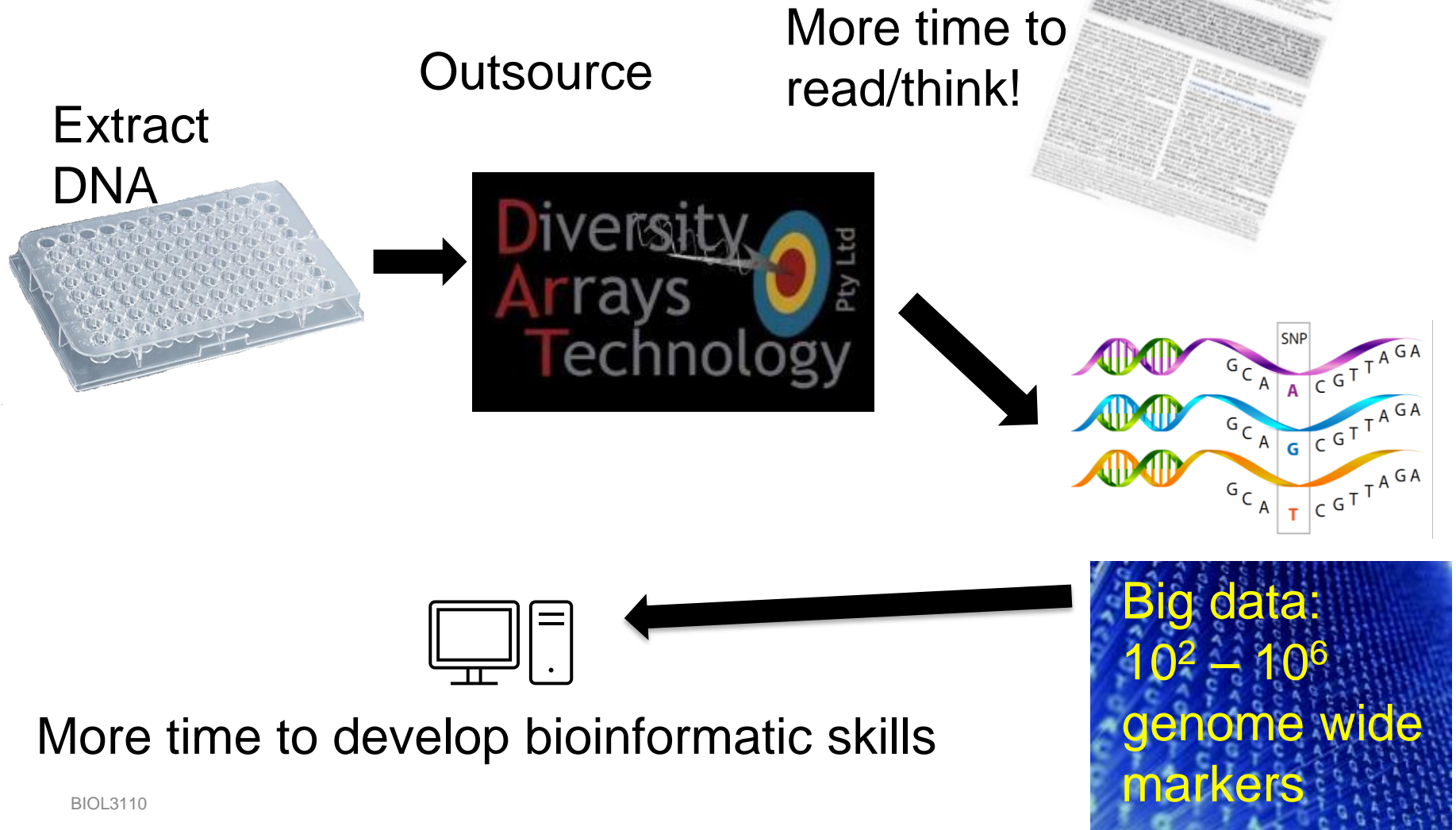
Counterintuitive example



Extent of spread



Modern genomic workflow



Next lecture:

Characterising V_G for single loci
More detail & examples

