GENETICS AND MOLECULAR BIOLOGY FOR ENVIRONMENTAL ANALYSIS

MOLECULAR ECOLOGY

LESSON 7: MOLECULAR MARKERS IN ECOLOGY

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APPROXIMATIVE SYLLABUS?

√ Molecular Genetics in Ecology 2 h
√ Molecular Biology for Dummies 10 h

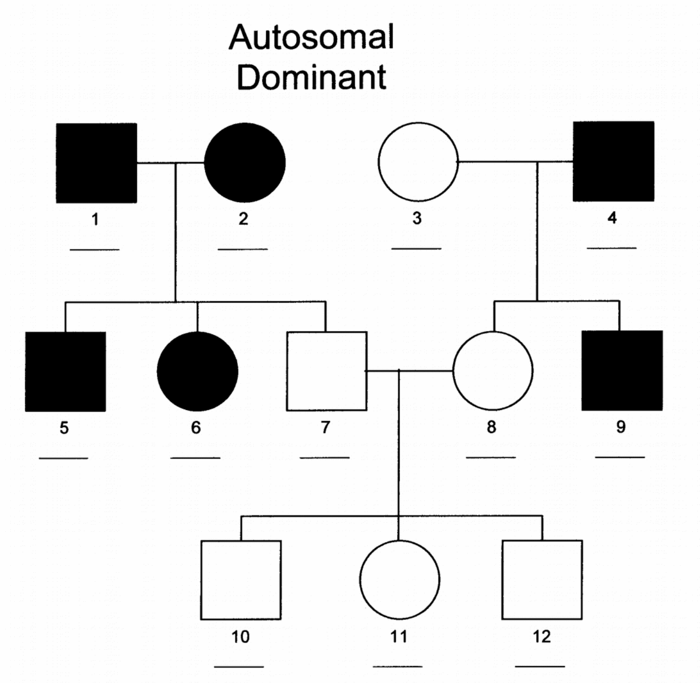
- Molecular Markers in Ecology 4 h
- Population Genetics 6h
- Conservation Genetics 2h
- DNA Sequencing and Analysis 4h
- Comparing Genomes 2h
- Structure and Function in Communities 4h
- Stress Responses 4h
- Variation and Adaptation 2h
- Studying Ecologically Important Traits: QTL Analysis, and Reverse Genetics 2h

UNDERSTANDING MOLECULAR MARKERS

- We started to look at the extraordinary wealth of genetic information that is present in every individual, and to explore how some of this information can be accessed and used in ecological studies.
- Now: properties of the genetic markers that are used in molecular ecology.
- Next class: an overview of those molecular markers that are most commonly used in ecology.

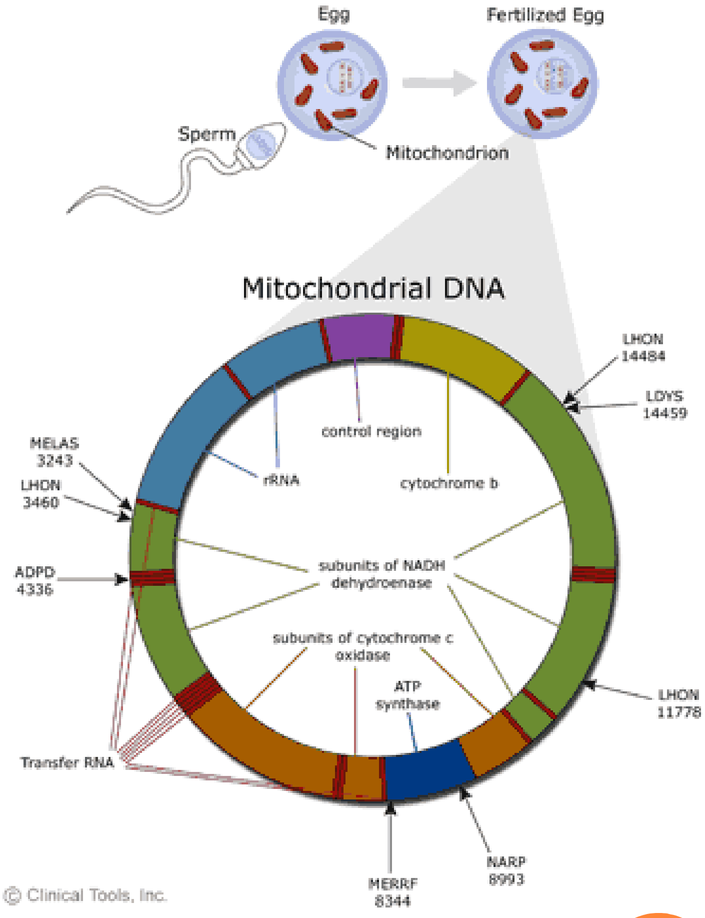
MODES OF INHERITANCE

- Genetic material is transmitted from parents to offspring in a predictable manner, and this is why molecular markers allow us to infer the genetic relationships of individuals.



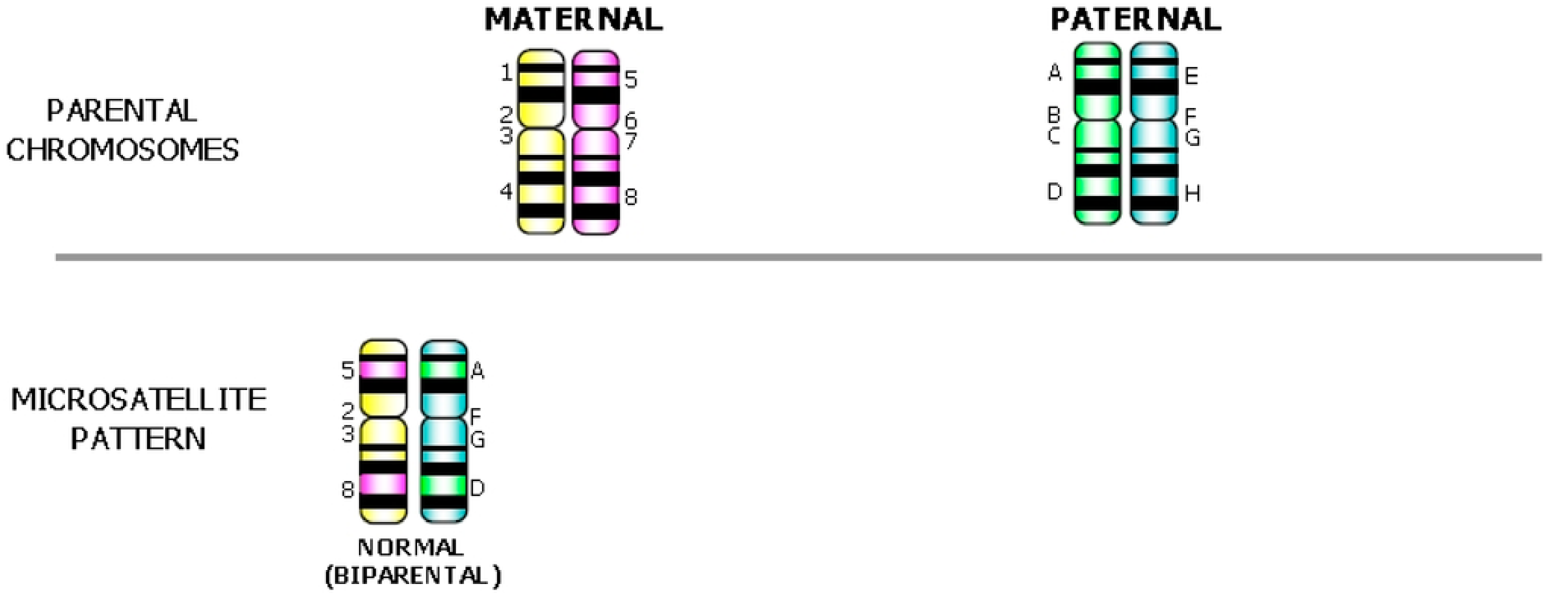
MODES OF INHERITANCE

- Not all DNA is inherited in the same way, and understanding different modes of inheritance is crucial before we can predict how different regions of DNA might behave under various ecological and evolutionary scenarios.



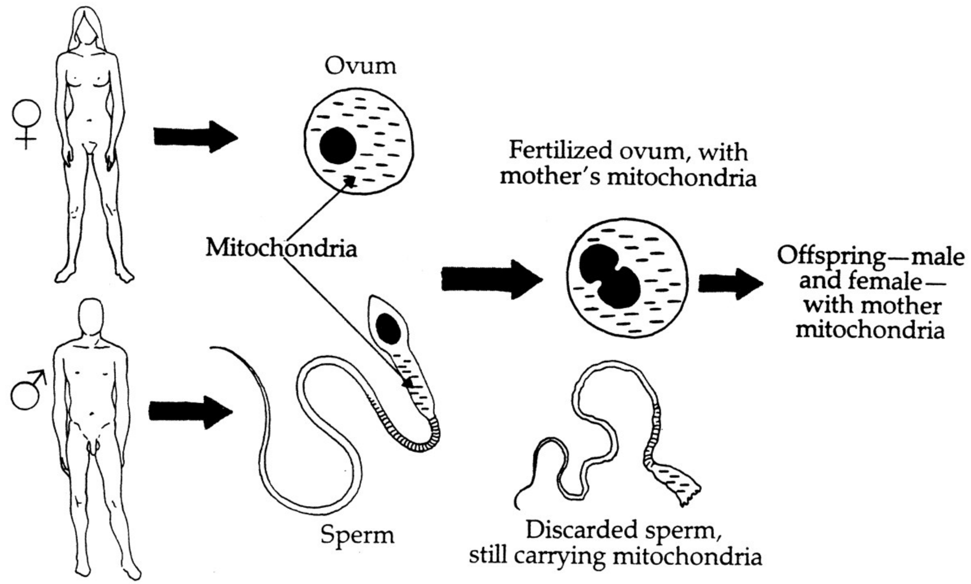
NUCLEAR VERSUS ORGANELLE

- biparental inheritance



NUCLEAR VERSUS ORGANELLE

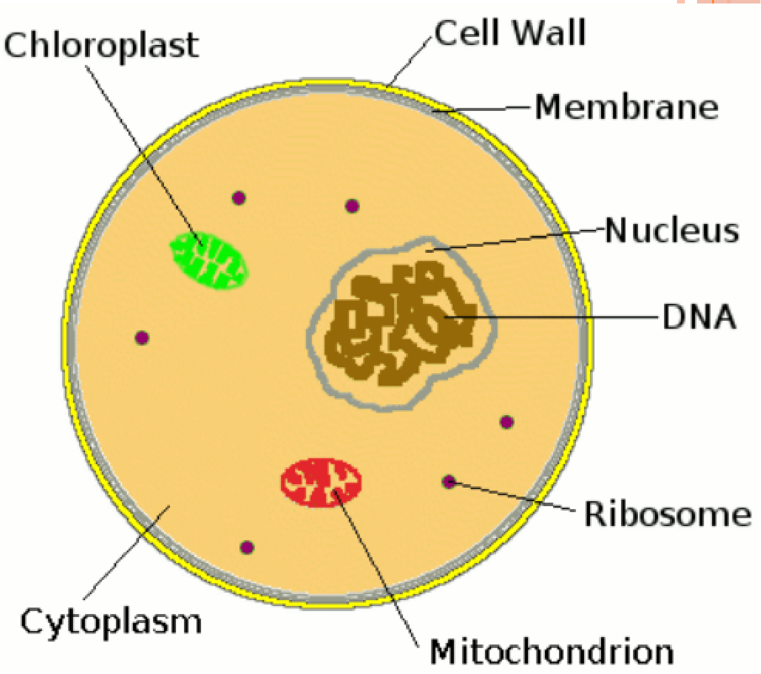
- However, even in sexually reproducing species, not all DNA is inherited from both parents. Two important exceptions are the uniparentally inherited organelle genomes of mitochondria (mtDNA) and plastids, with the latter including chloroplasts (cpDNA).



Pattern of inheritance: Unlike nuclear DNA, which we inherit half from our mother and half from our father, mitochondrial DNA is passed on only by females. The reason is that when the sperm fertilizes the egg, it leaves behind all its mitochondria: the developing fetus therefore inherits mitochondria only from the mother's egg.

NUCLEAR VERSUS ORGANELLE

- These are both located outside the cell nucleus. Mitochondria are found in both plants and animals, whereas plastids are found only in plants. **Organelle DNA typically occurs in the form of supercoiled circles of double-stranded DNA, and these genomes are much smaller than the nuclear geno**

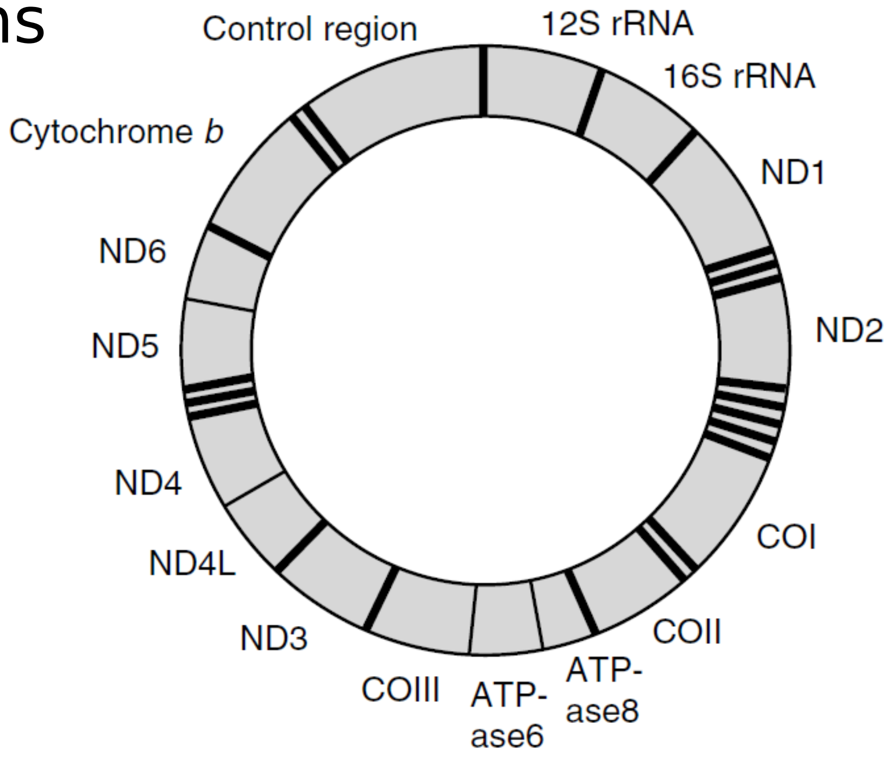


ANIMAL MITOCHONDRIAL DNA

- In most animals, mitochondrial DNA is inherited maternally, meaning that it is passed down from mothers to their offspring.
- Animal mtDNA contains:
  - 13 protein-coding genes,
  - 22 transfer RNAs and two ribosomal RNAs
  - a control region that contains sites for replication and transcription initiation.

ANIMAL MITOCHONDRIAL DNA

- Typical gene organization of vertebrate mtDNA. Unlabelled dark bands represent 22 transfer RNAs (tRNAs). Gene abbreviations starting with ND are subunits of NADH dehydrogenase, and those starting with CO are subunits of cytochrome c oxidase



MITOCHONDRIAL DNA: EXCEPTIONS TO THE RULES

- not all mitochondria in animals are inherited maternally.
- Instances of paternal leakage (transmission of mitochondria from father to offspring) have been found in a number of species, including mice (Gyllensten et al., 1991), birds (Kvist et al., 2003) and humans (Schwartz and Vissing, 2002).

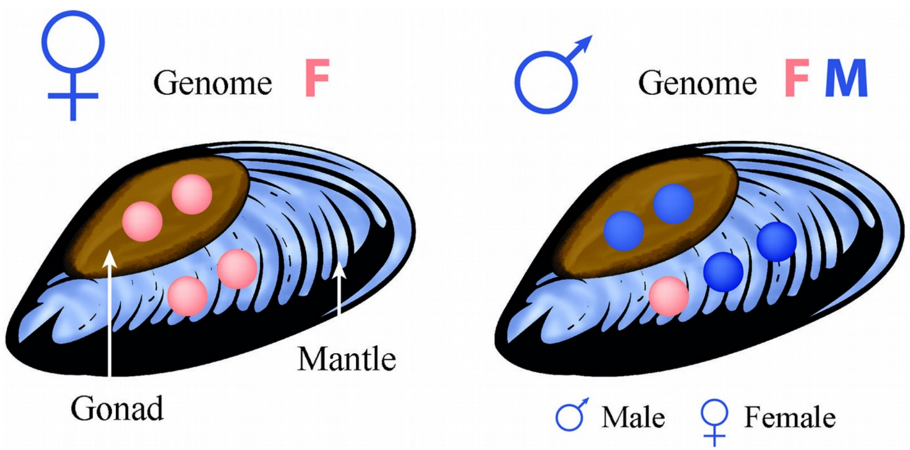
MITOCHONDRIAL DNA: EXCEPTIONS TO THE RULES

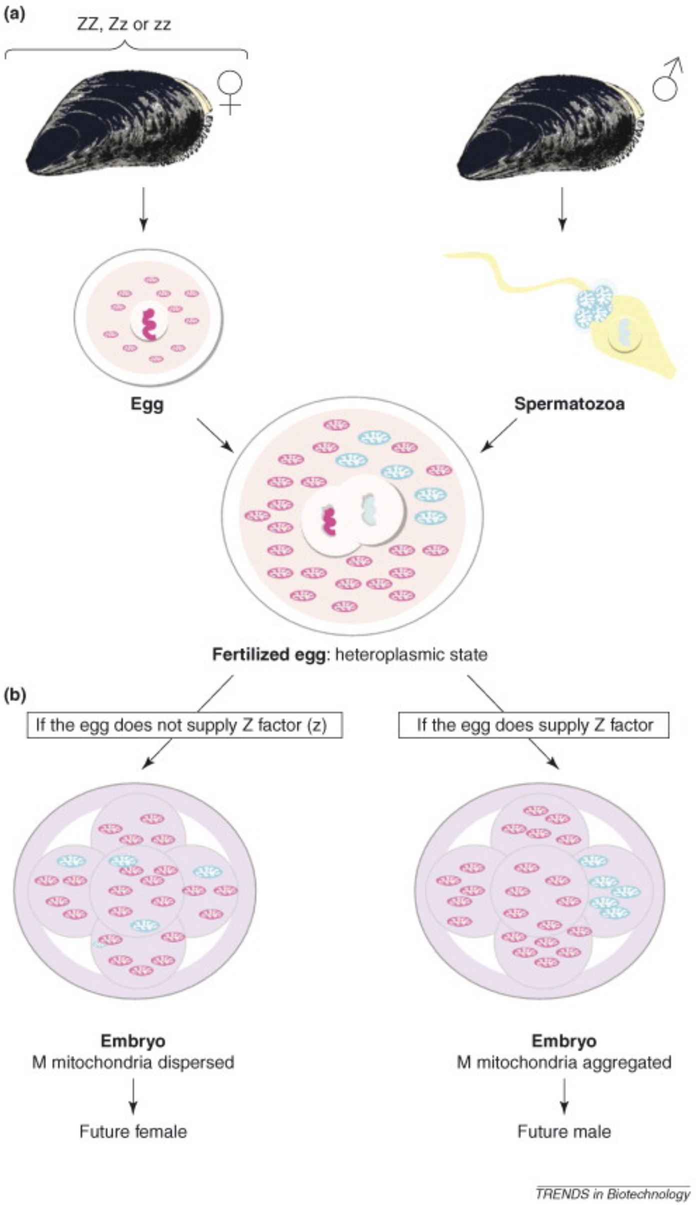
- Nevertheless, the extent of paternal leakage is believed to be low in most animals with the exception of certain mussel species within the families Mytilidae, Veneridae and Unionidae, which follow double biparental inheritance.

- This means that females generally inherit their mitochondria from their mothers, but males inherit both maternal and paternal mtDNA. The males therefore represent a classic case of **heteroplasmy** (more than one type of mitochondria within a single individual).

MITOCHONDRIAL DNA: EXCEPTIONS TO THE RULES

- Proposed genetic control of mitochondrial transmission under the DUI system.





ANIMAL MITOCHONDRIAL DNA

- There are several reasons why mtDNA markers have been used extensively in studies of animal population genetics.
  - Small size
  - Conserved arrangement of genes
  - Universality of primers
  - *De novo* analysis

ANIMAL MITOCHONDRIAL DNA

- Second, although the arrangement of genes is conserved, **the overall mutation rate is high.**
- Synonymous substitutions rate 10X higher than nuclear protein coding genes.
- The non-coding control region, which includes the displacement **(D) loop**, evolves particularly rapidly in many taxa.

ANIMAL MITOCHONDRIAL DNA

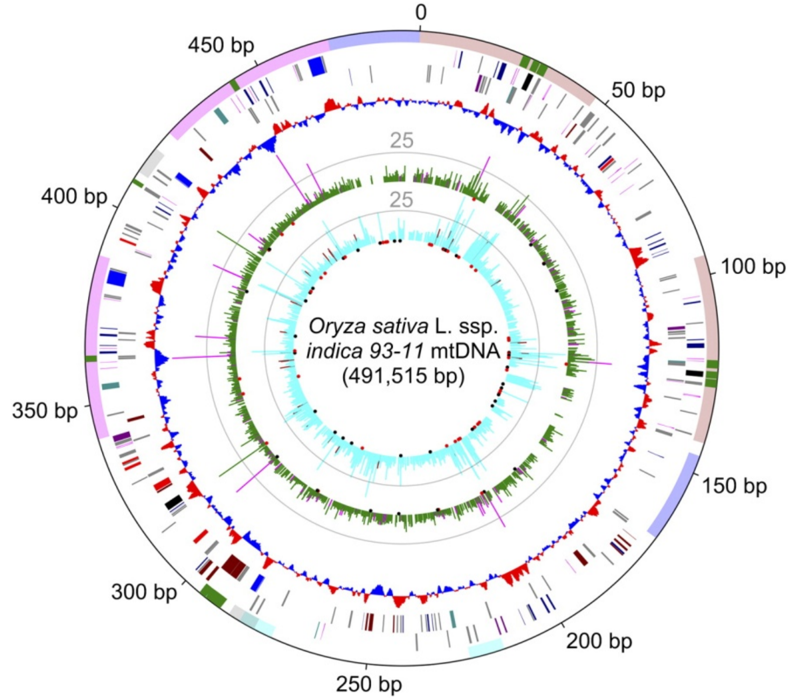
- The third relevant property of mtDNA is its general lack of recombination, which means that offspring usually will have (barring mutation) exactly the same mitochondrial genome as the mother.
- Finally, because mtDNA is haploid and uniparentally inherited, it is effectively a quarter of the population size of diploid nuclear DNA. Because there are **fewer copies of mtDNA to start with**, it is relatively sensitive to demographic events such as bottlenecks.

PLANT MITOCHONDRIAL DNA

- As with animals, mtDNA in most higher plants is maternally inherited. There are a few exceptions to these rules, for example mtDNA is transmitted paternally in the redwood tree *Sequoia sempervirens* and biparentally inherited in some plants in the genus *Pelargonium*

PLANT MITOCHONDRIAL DNA

- Unlike animal mtDNA, **plant mitochondrial genomes** regularly **undergo recombination** and therefore evolve rapidly with respect to gene rearrangements and duplications. As a result, their sizes vary considerably (40 000 -- 2 500 000 bp).



PLANT MITOCHONDRIAL DNA

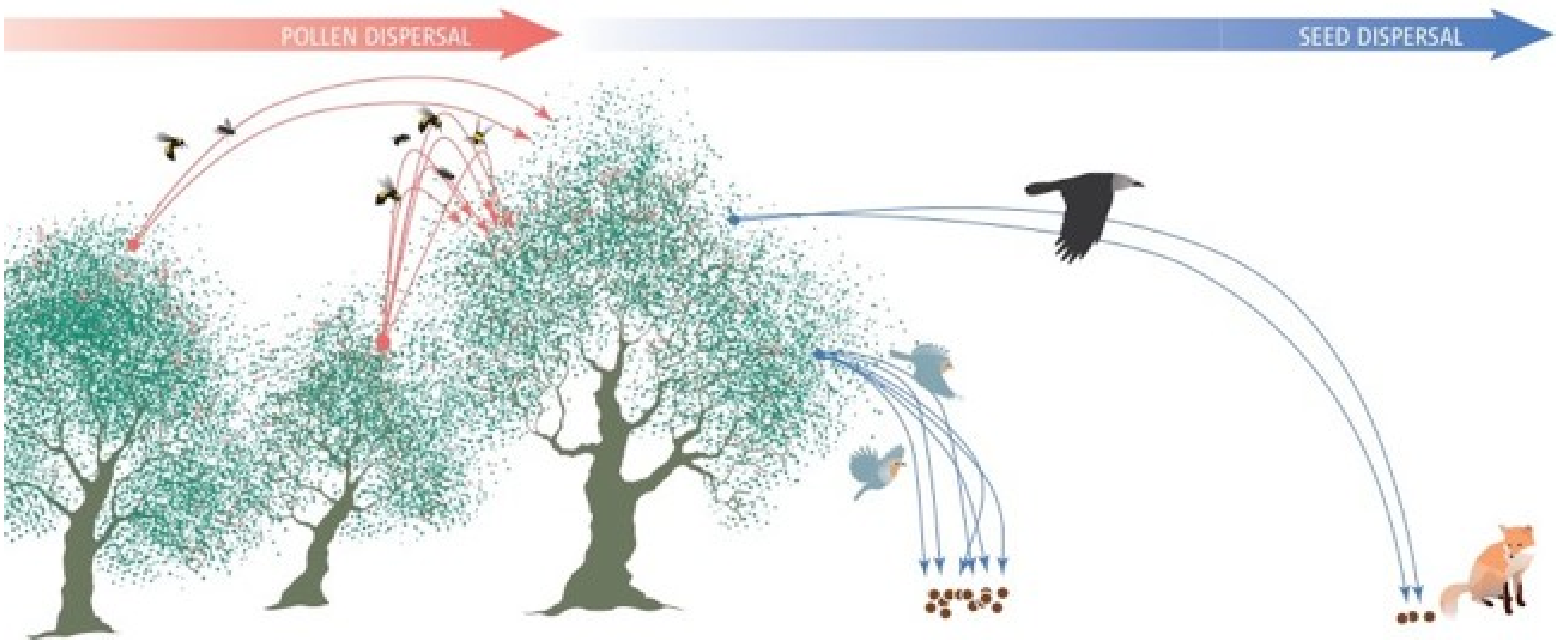
- This variability makes it difficult to generalize; to take one example, the mitochondrial genome of the liverwort *Marchantia polymorpha* is around 186 608 bp long and appears to include three ribosomal RNA genes, 29 transfer RNA genes, 30 protein coding genes with known functions and around 32 genes of unknown function.

PLANT MITOCHONDRIAL DNA

- Although the organization of plant mitochondria regularly changes, evolution is slow with respect to nucleotide substitutions. In fact, in most plant species the **mitochondria are the slowest evolving genomes.**
- 100X slower than animal mtDNA

PLANT MITOCHONDRIAL DNA

- pollen may be dispersed by the wind (or pollinators)
- But seeds are also ingested by migratory birds
- mtDNA for seed dispersal, pollen analysed by nuclear DNA



PLANT MITOCHONDRIAL DNA

- Canadian populations of the black spruce (Picea mariana) grow in areas that were covered in ice until approximately 6000 years ago, so we know that populations must have been established since that time. Researchers found that current populations share the same mtDNA haplotypes but not the same nuclear alleles.

Molecular Ecology (2003) **12**, 891–901

Diverging patterns of mitochondrial and nuclear DNA diversity in subarctic black spruce: imprint of a founder effect associated with postglacial colonization

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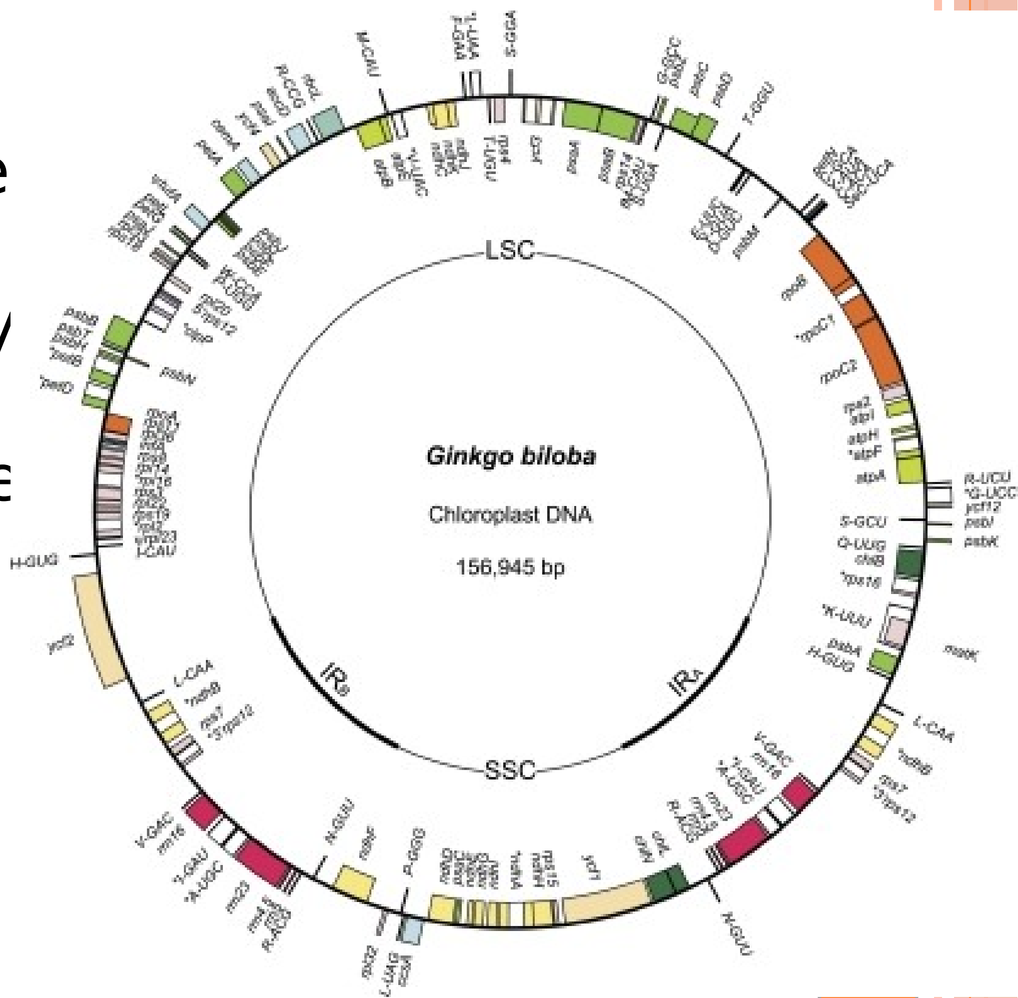
PLASTIDS, INCLUDING CHLOROPLAST DNA

- The relatively low variability of plant mtDNA means that when haploid markers are desirable in plant studies, researchers more commonly turn to plastid genomes, including **chloroplast DNA (cpDNA).**
- cpDNA is **inherited maternally in most angiosperms** (flowering plants),
- in **most gymnosperms (conifers and cycads) it is usually inherited paternally**.

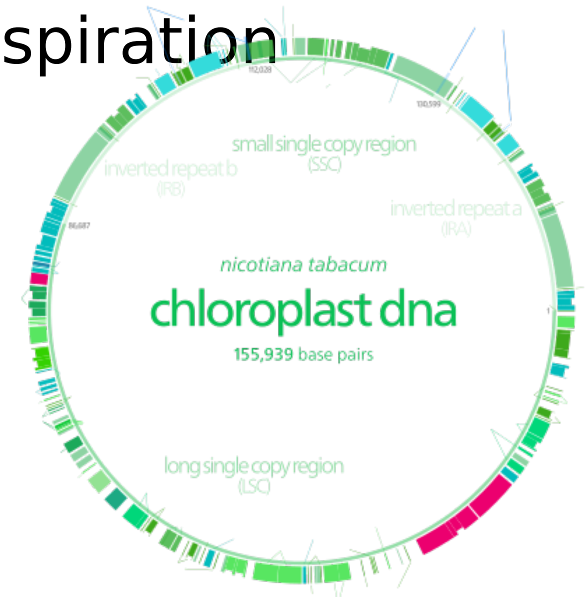
PLASTIDS, INCLUDING CHLOROPLAST DNA

- Chloroplast genomes, which in most plants are key to the process of photosynthesis, typically range from 120 000 to 220 000 bp (the average size is around 150 000 bp).



PLASTIDS, INCLUDING CHLOROPLAST DNA

- In tobacco (Nicotiana tabacum), the cpDNA genome contains 113 genes, which include 21 ribosomal proteins, 4 ribosomal RNAs, 30 transfer RNAs, 29 genes that are necessary for functions associated with photosynthesis and 11 genes that are involved with chlororespiration



HAPLOID CHROMOSOMES

- When discussing the inheritance of nuclear and organelle markers we usually refer to nuclear genes as being inherited **biparentally** following sexual reproduction.
- But some exception are…

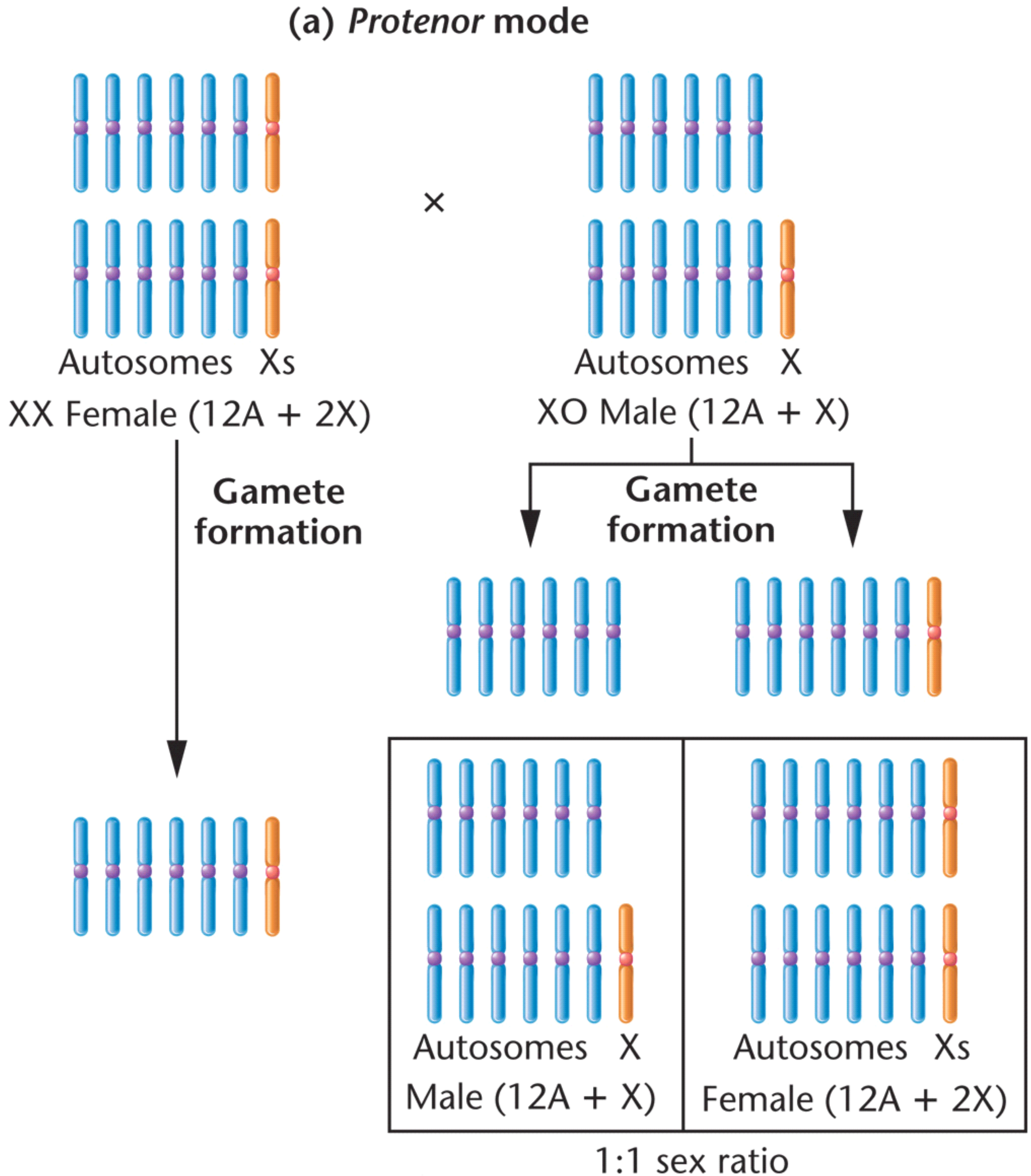
  sex chromosomes.

- Not all species have sex chromosomes, for example crocodiles and many turtles and lizards follow **environmental sex determination**

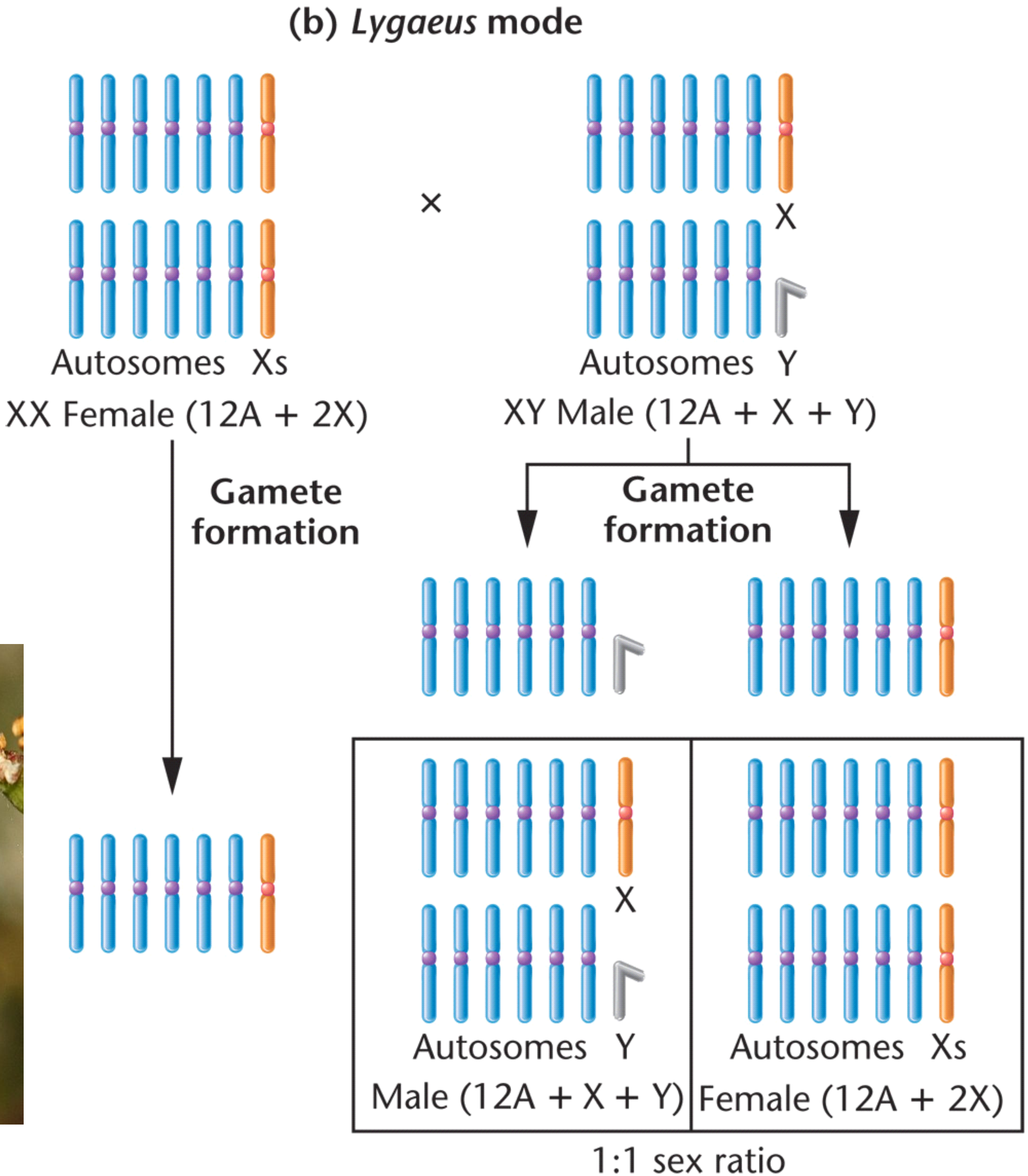
HAPLOID CHROMOSOMES

- In most mammals, and some dioecious plants, females are homogametic (two copies of the same sex chromosome: XX), whereas males are heterogametic (one copy of each sex chromosome: XY). (Lygaeus mode)

- XX-XO system (Protenor mode)



- XX-XY System (Lygaeus mode)

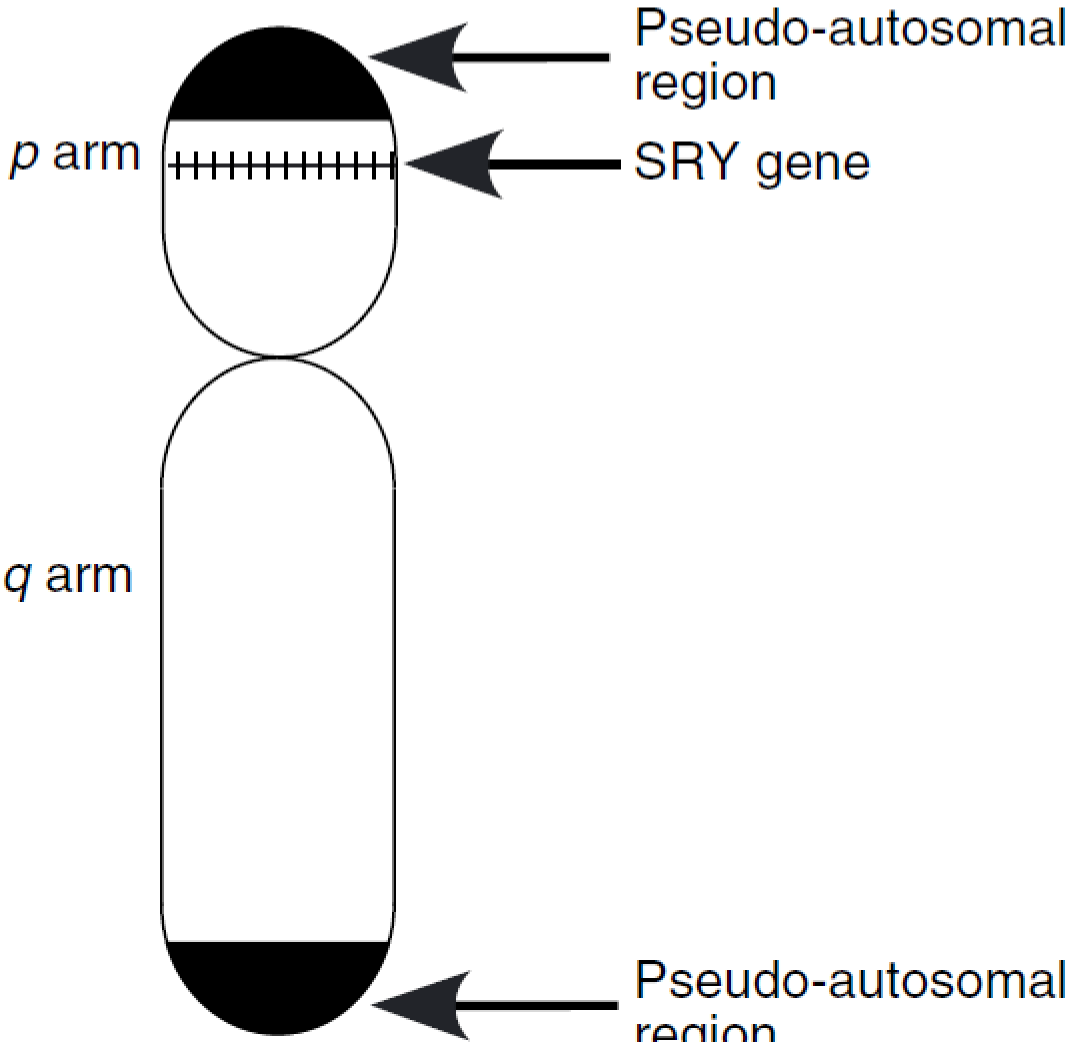


HAPLOID CHROMOSOMES

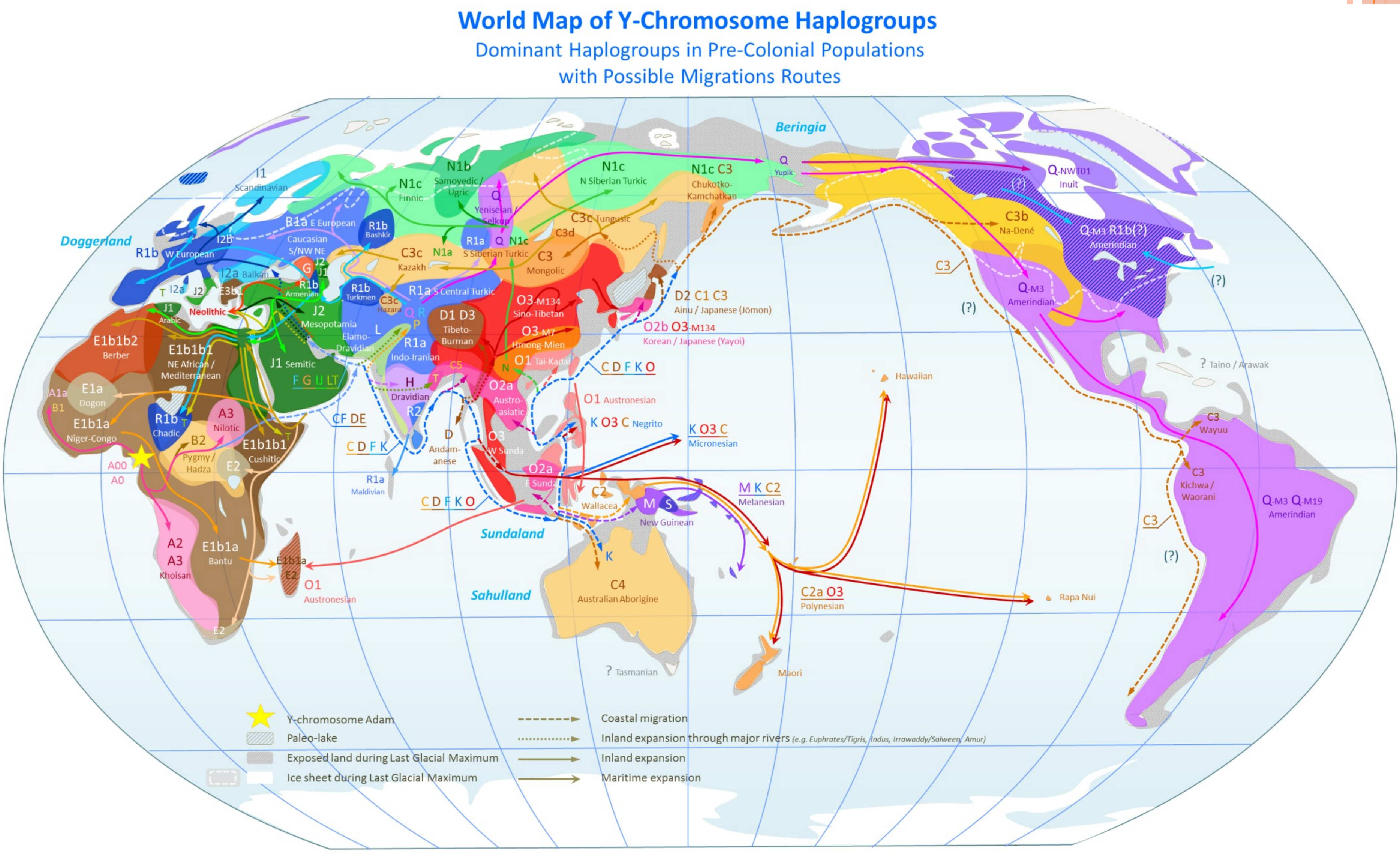
- In birds and lepidopterans, which have heterogametic females (ZW) and homogametic males (ZZ).
- But also some fishes: sturgeon?

MAMMALIAN Y CHROMOSOME

- Y chromosomes are the only mammalian chromosomes that are effectively haploid
- The mutation rate of Y chromosomes is relatively low.

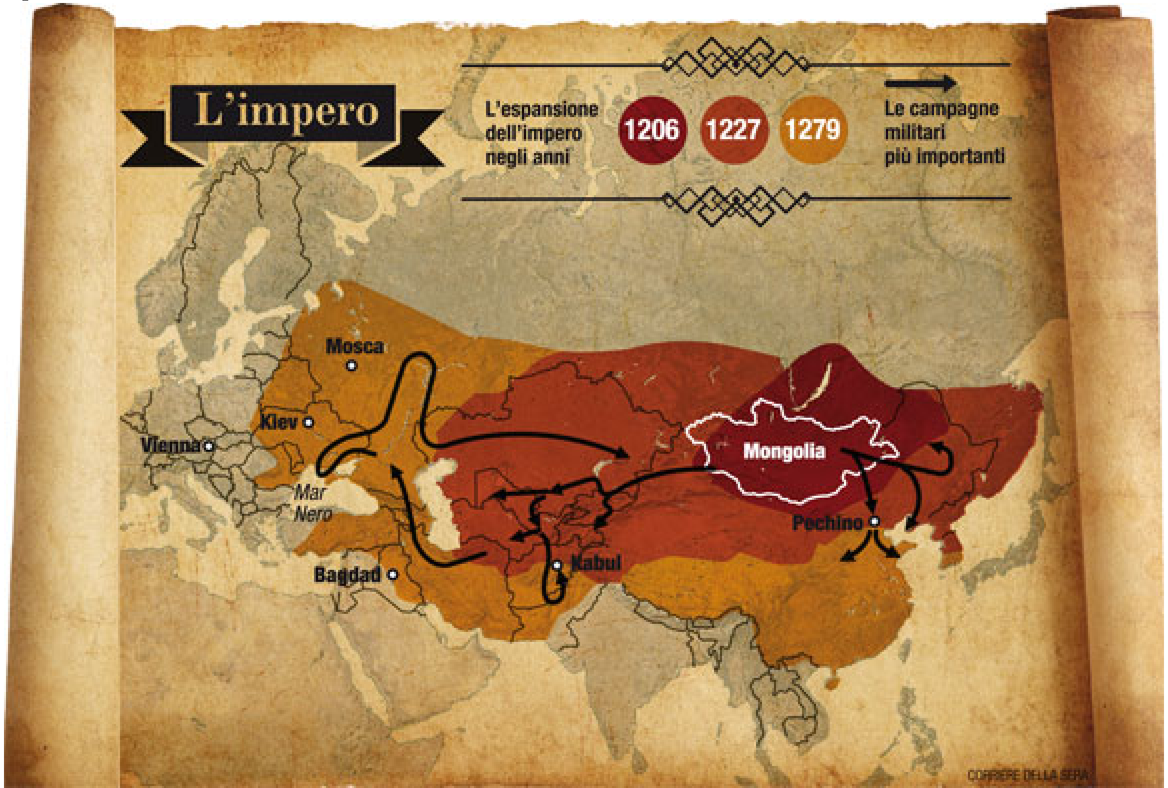


PATRILINEAL DESCENT



MAMMALIAN Y CHROMOSOME

- 8 per cent of the men in a large area of Asia, from the Pacific to the Caspian Sea carry the same Y haplotype



IDENTIFYING HYBRIDS

- **Hybrids** can be identified from their genotypes because hybridization results in *introgression*, the flow of alleles from one species (or population) to another.
- Identification of hybrids in the wild is often based on **cytonuclear disequilibrium**, which occurs in hybrids that have cytoplasmic markers from one species or population and nuclear markers from another.

IDENTIFYING HYBRIDS

Table 2.2 Some examples of cytonuclear disequilibrium in hybrids

Hybrid	Nuclear DNA	mtDNA or cpDNA
Freshwater crustaceans *Daphnia pulex* × *D. pulicaria*	*D. pulicaria*	*D. pulex*
Grey wolf (*Canis lupus*) × coyote (*C. latrans*)	Wolf	Coyote
House mice *Mus musculus* × *M. domesticus*	*M. musculus*	*M. domesticus*
Northern red-backed vole (*Clethrionomys rutilus*) × bank vole (*C. glareolus*)	Bank vole	Northern red-backed vole
White poplar (*Populus alba*) × black poplar (*P. nigra*)	Black poplar	White poplar

UNIPARENTAL MARKERS: A CAUTIONARY NOTE

- organelles behave as single inherited units and are therefore effectively single locus markers.
- data from a single locus allow us to retrace the history of only a single genetic unit (gene or genome), which may or may not be concordant with the history of the species in question….

UNIPARENTAL MARKERS: A CAUTIONARY NOTE

- An additional drawback to uniparentally inherited markers is that they may not be representative of populations as a whole.
- Dispersal of plant is different depending of the choose marker.
- Also in animals can be true. Male and female could have different dispersal.

UNIPARENTAL MARKERS: A CAUTIONARY NOTE

- Another risk associated with mtDNA markers involves copies of mtDNA that have been translocated to the nuclear genome; these are known as mitochondrial pseudogenes, or **numts** (nuclear copies of mtDNA sequences).

SUMMARY

Table 2.1 Usual mode of inheritance of different genomic regions in sexually reproducing taxa

Genomic region	Typical mode of inheritance
Animals	
Autosomal chromosomes	Biparental
Mitochondrial DNA	Maternal in most animals Biparental in some bivalves
Y chromosome	Paternal
Higher plant	
Autosomal chromosomes	Biparental
Mitochondrial DNA	Usually maternal
Plastid DNA (including chloroplast DNA)	Maternal in most angiosperms Paternal in most gymnosperms Biparental in some plants
Y chromosome	Paternal in some dioecious plants