Sex determination and sex linkage

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Sex determination

- A sex-determination system is a biological system that determines the development of sexual characteristics in an organism.
- Most organisms that create their offspring using sexual reproduction have two sexes.
- Occasionally, there are hermaphrodites in place of one or both sexes. There are also some species that are only one sex due to parthenogenesis, the act of a ♀reproducing without fertilization.
- Sexual orientation of individual flowers and plants may be:
  - Flower: Unisexual and bisexual
  - Individual: Monoecious, dieoeious and hermaphrodite
- In many species, sex determination is genetic: males and females have different alleles or even different genes that specify their sexual morphology.
- In animals this is often accompanied by chromosomal differences, generally through combinations of XY, ZW, XO, ZO chromosomes, or haplodiploidy.
- X and Y chromosomes can be divided into homologous and differential regions.

Mechanisms of sex determination

Environmental sex determination

- In some cases, sex of a fetus is determined by environmental factors (e.g. temperature)
- Some species such as various plants and fish do not have a fixed sex, and instead go through life cycles and change sex based on genetic cues during corresponding life stages of their type.
- This could be due to environmental factors,
  - Seasons
  - Association with ♀
  - Egg size
  - Incubation temperature

- Larvae of sea worm *Bonelia* are sexually undifferentiated. Those larvae thaat attach to the proboscis of ♀worms develop into ♂s. In contrast, those larvae that do not attach to ♀worms and remain free develop into ♀s.
- In sea worm *Dinophilus*, animals developed from eggs of relatively larger size are ♀s, while those obtained from smaller eggs are ♂s.
- Animals like turtles, alligators and crocodiles respond to egg incubation temperature during embryonic stages for differentiation. In some cases:
  - High $(30 - 35^{\circ})$ temperature produces only ♀s,
  - Low $(23 - 28^{\circ})$ temperature produce only ♂s, and
  - Intermediate temperature $(28 - 30^{\circ})$ might give rise to both ♂s and ♀s.

Chromosomal sex determination

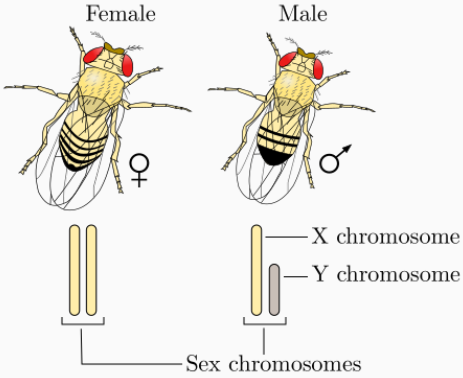
- In a vast majority of animals, ♂and ♀individuals ordinarily differ from each other in respect of either the number or the **morphology of the homologues of one chromosome pair**.
- This chromosome is reffered to as sex chromosome or allosome.
- There are 2 types of sex chromosome, X and Y.
- **X chromosome** is found in both ♂s and ♀s, although one sex has only one, while the other sex has two X chromosomes.
- **Y chromosome** ordinarily occurs only in one of the two sexes of a species, e.g., ♂mice, drosophila, humans, ♀birds, reptiles, etc.
- Y chromosome contains mostly heterochromatin while X chromosome contains euchromatin.
- Different mechanisms of chromosomal sex determination are:
  1. XX ♀, X0 ♂
  2. X0 ♀, XX ♂
  3. XX ♀, XY ♂
  4. XY ♀, XX ♂
  5. diploid (2n) ♀, haploid (n) ♂

XX/XO sex chromosome (Chromosomal sex determination)

- In grasshoppers, *Protenor* and other orthopteran insects, ♀s have two X chromosomes, while males have only one X (XX ♀, X0 ♂).
  - Thus somatic cell of ♀s have one chromosome more than those of ♂s.
  - Females are homogametic sex and ♂s are heterogametic sex.
  - Union of a sperm having an X chromosome with any egg produces a zygote having two X chromomsomes (XX); such zygote develop into ♀individuals. But when a sperm without an X chromosome fertilizes an eg, an XO zygote is obtained; such individuals would develop into males.
- In Fumea, females have only one X chromosome while males have two X chromosomes (X0 ♀, XX ♂)

XX/XY chromosome

- XX ♀, XY ♂
  - The system is found in most other mammals, as well as some insects (Hemiptera, Coleoptera, Diptera), some fishes and some amphibia.
  - In this system, most females have two of the same kind of sex chromosome (XX), while most males have two distinct sex chromosomes (XY).
  - In some species, such as humans, organisms remain sex indifferent for a time after they're created; in others, however, such as fruit flies, sexual differentiation occurs as soon as the egg is fertilized.
  - ♀are homogametic and males are heterogametic sex
- XY ♀, XX ♂
  - The system is operates in some birds, reptiles, some insects, e.g. silkworm, etc.
  - ♂are homogametic and females are heterogametic sex.


Female
Male
♀
♂
X chromosome
Y chromosome
Sex chromosomes

X or Y-centered sex determination

- In Y-centered sex determination, the SRY gene (residing on the Y chromosome) is the main gene in determining ♂characteristics, but multiple genes are required to develop testes. Members of SRY-reliant species can have uncommon XY chromosomal combinations such as XXY and still live.
- Some species, such as fruit flies, use the presence of two X chromosomes to determine femaleness. Species that use the number of Xs to determine sex are nonviable with an extra X chromosome.
- Some fish have variants of the XY sex-determination system, as well as the regular system. For example, while having an XY format, *Xiphophorus nezahualcoyotl* and X. milleri also have a second Y chromosome, known as Y', that creates XY' females and YY' males.
- The platypus has ten sex chromosomes; males have an XYXYXYXYXY pattern while females have ten X chromosomes.

Haplodiploidy

- Mostly in hymenoptera, e.g. honey-bees, ants, termites females are diploid 2n and males are haploid (n)
- During spermatogenesis, all the n chromosomes of males regularly pass to a single pole at Anaphase I so that the opposite pole receives no chromosome. Thus all the sperms are regularly haploid.
- Normal oogenesis produces all haploid eggs.
- Fertilization of eggs produces diploid zygotes, which develop into diploid larvae.
  - Such larvae give rise to workers, which are sterile females.
  - Females fed on royal jelly develop into fertile females called queen.
- Unfertilized eggs develop parthenogenically to produce haploid larvae and ultimately fully fertile haploid males called drones.

Molecular basis of sex determination

Geneic balance theory

- Bridges discovered XXY females and X0 males in *Drosophila* while studying inheritance of vermilion eye gene in the X chromosome.
- This showed that XX and XY chromosome constitutions are not essential for femaleness and maleness, respectively, and that Y chromosome did not play a role in sex determination.
- Triploid females mated with normal diploid males result in aneuploid progeny. The progeny from such cross showed five different sex expressions.
- By correlating the sex of an individual with its chromosome constitution, Bridges developed the geneic balance theory of sex determination. This theory fully explains sex determination in *Drosophila*.

$$\text{Sex index} = \frac{\text{Number of X chromosomes}}{\text{Number of autosomal sets}} = \frac{X}{A}$$

Ploidy	Number of X chromosomes	Number of autosomal sets	Sex index (X/A)	Sex expression
2n	1	2	0.50	♂
2n	2	2	1.00	♀
2n	3	2	1.50	Super ♀
2n	4	2	2.00	Super ♀
3n	1	3	0.33	Super ♂
3n	2	3	0.67	Intersex
3n	3	3	1.00	♀
3n	4	3	1.33	Super ♀
4n	1	4	0.25	Super ♂
4n	2	4	0.50	♂
4n	3	4	0.75	Intersex
4n	4	4	1.00	♀

Sex determination in plants

Mechanisms

- Environmental: *Equisetum sp.*
- Chromosomal:
  - Homomorphic chromosomes: Heterogametic ♂(*Asparagus*, Spinach), Heterogametic ♀(*Frageria elateria*)
  - Heteromorphic chromosomes: Heterogametic ♂(Active Y: *Silene latifolia*, X/autosome balance: *Rumex acetosa*, X1X1X2X2 - X1X2Y1Y2: *Humulus lupulus*)
- Genic:
  - Single gene: *Ecballium elaterium*
  - Muliple gene: *Mercurialis annua*

Genic sex determination in Papaya

- In papaya, a single gene with three alleles ($m, M_1 and M_2$) is suggested to control the sex of an individual. ♀plants are homozygous mm, while ♂plants are heterozygous M_1m; the heterozygote M_2m produces hermaphrodite condition. Genotypes M_1M_1, M_1M_2 and M_2M_2 are inviable (Table 1).

Table 1: Monogenic sex determination in Papaya; the gene m has three alleles, viz., m, M_1, M_2

Genotype	Survival	Sex expression
mm	Vital	♀
M_1m	Vital	♂
M_2m	Vital	Hermaphrodite
M_1M_1, M_1M_2 and M_2M_2	Lethal (all die)	

Table 2: Various cross combinations (in monogenic sex determination system in Papaya)

Genotype	Sex expression
♀x ♂($mm \times M_1m$)	1/2 ♂(M_1m), 1/2 ♀(mm)
♀x hermaphrodite ($mmxM_2m$)	1/2 Hermaphrodite (M_2m), 1/2 ♀(mm)
Hermaphrodite (M_2m) selfed	1/4 M_2M_2 (inviable), 1/2 hermaphrodite (M_2m), 1/4 ♀(mm)

Sex linkage

- Sex linkage is the patterns of inheritance and presentation when a gene mutation (allele) is present on a sex chromosome (allosome) rather than a non-sex chromosome (autosome).
- They are characteristically different from the autosomal forms of dominance and recessiveness as they are different depending on the sex of the individual.
- Since humans have several times as many genes on the ♀X chromosome than on the ♂Y chromosome, X-linked traits are much more common than Y-linked traits.

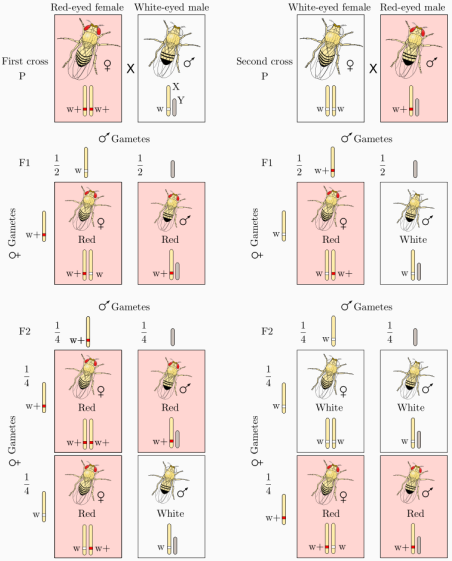


Figure 1: Experiment performed by Morgan on Drosophila

X-linked traits

- Additionally, there are more X-linked recessive conditions than X-linked dominant, and X-linked recessive conditions affect males much more commonly, due to males only having the one X chromosome required for the condition to present.
- In humans, X-linked traits are inherited from a carrier or affected mother or from an affected father.
- A daughter has a 50% chance of being a carrier, however a son born to an affected father and a non-carrier mother will always be unaffected due to not inheriting the father's X chromosome. A daughter in that case, on the other hand will always be a carrier.
- Unless the condition is dominant she will not be always affected.
- The incidence of X-linked recessive conditions in females is the square of that in males: for example, if 1 in 20 males in a human population are red-green color blind, then 1 in 400 females in the population are expected to be color-blind (1/20)*(1/20).
- In classical genetics, a mating experiment called a reciprocal cross is performed to test if an animal's trait is sex-linked.

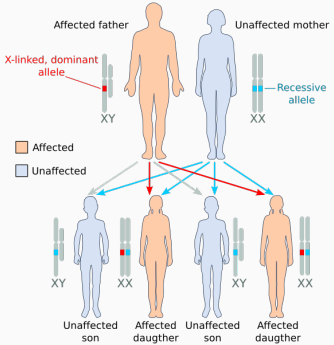
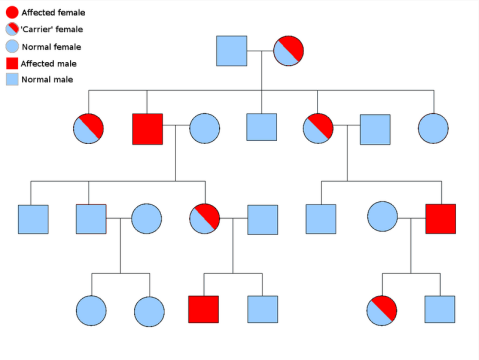


Figure 2: Illustration of some X-linked heredity outcomes
(A) the affected father has one X-linked dominant allele, the mother is homozygous for the recessive allele: only daughters (all) will be affected. (B) the affected mother is heterozygous with one copy of the X-linked dominant allele: both daughters and sons will have 50% probability to be affected. (C) the heterozygous mother is called 'carrier' because she has one copy of the recessive allele: sons will have 50% probability to be affected, 50% of unaffected daughters will become carriers like their mother.


Affected female
'Carrier' female
Normal female
Affected male
Normal male

X-linked dominant inheritance

- Each child of a mother affected with an X-linked dominant trait has a 50% chance of inheriting the mutation and thus being affected with the disorder.
- If only the father is affected, 100% of the daughters will be affected, since they inherit their father's X chromosome, and 0% of the sons will be affected, since they inherit their father's Y chromosome.
- There are less X-linked dominant conditions than X-linked recessive, because dominance in X-linkage requires the condition to present with only a fraction of the gene expression of autosomal dominance, since roughly half (or as many as 90% in some cases) of a particular parent's X chromosomes are inactivated in females.
- Example: Coffin-Lowry Syndrom (CLS), Fragile X syndrome, Vitamin D resistant rickets

X-linked recessive inheritance

- Females possessing one X-linked recessive mutation are considered carriers and will generally not manifest clinical symptoms of the disorder, although differences in X chromosome inactivation can lead to varying degrees of clinical expression in carrier females since some cells will express one X allele and some will express the other.
- All males possessing an X-linked recessive mutation will be affected, since males have only a single X chromosome and therefore have only one copy of X-linked genes.
- All offspring of a carrier ♀have a 50% chance of inheriting the mutation if the father does not carry the recessive allele.
- All ♀children of an affected father will be carriers (assuming the mother is not affected or a carrier), as daughters possess their father's X chromosome.
- If the mother is not a carrier, no ♂children of an affected father will be affected, as males only inherit their father's Y chromosome.
- Examples: Color blindness, Duchenne muscular dystrophy, Haemophilia A and B, Hunter syndrome, Inherited nephrogenic diabetes insipidus, etc.

Y-linked

- Various failures in SRY genes

Sex-linked traits in other animals

- White eyes in Drosophila melanogaster flies—the first sex-linked gene discovered.
- Fur color in domestic cats: the gene that causes orange pigment is on the X chromosome; thus a Calico or tortoiseshell cat, with both black (or gray) and orange pigment, is nearly always ♀.
- The first sex-linked gene ever discovered was the "lacticolor" X-linked recessive gene in the moth Abraxas grossulariata by Leonard Doncaster

Sex-influenced traits

Sex-influenced or sex-conditioned traits are phenotypes affected by whether they appear in a ♂or ♀body. Even in a homozygous dominant or recessive ♀the condition may not be expressed fully. Example: baldness in humans.

Sex-limited traits

These are characters only expressed in one sex. They may be caused by genes on either autosomal or sex chromosomes. Examples: ♀sterility in Drosophila; and many polymorphic characters in insects, especially in relation to mimicry. Closely linked genes on autosomes called “supergenes” are often responsible for the latter.

Numerical problems

Problem 2: Sex linked inheritance

- A sex-linked recessive allele *c* produces a red-green color blindness in humans. A normal woman whose father was color blind marries a color-blind man.
  a. What genotypes are possible for the mother of the color blind man ?
  b. What are the chances that the first child from this marriage will be a color-blind boy ?
  c. Of the girls produced by these parents, what proportion can be expected to be color blind ?
  d. Of all the children (sex unspecified) of these parents, what proportion can be expected to have normal color vision ?

Solution 2: Sex linked inheritance

a. $X^C/X^c, X^c/X^c$
b. p (color-blind) x p (♂) $= (1/2)(1/2) = 1/4$
c. The girls will be 1 normal (X^C/X^c): 1 color-blind (X^c/X^c).
d. The cross is $X^C/X^c \times X^c/Y$, yielding 1 normal:1 color-blind for both sexes.

Bibliography

References