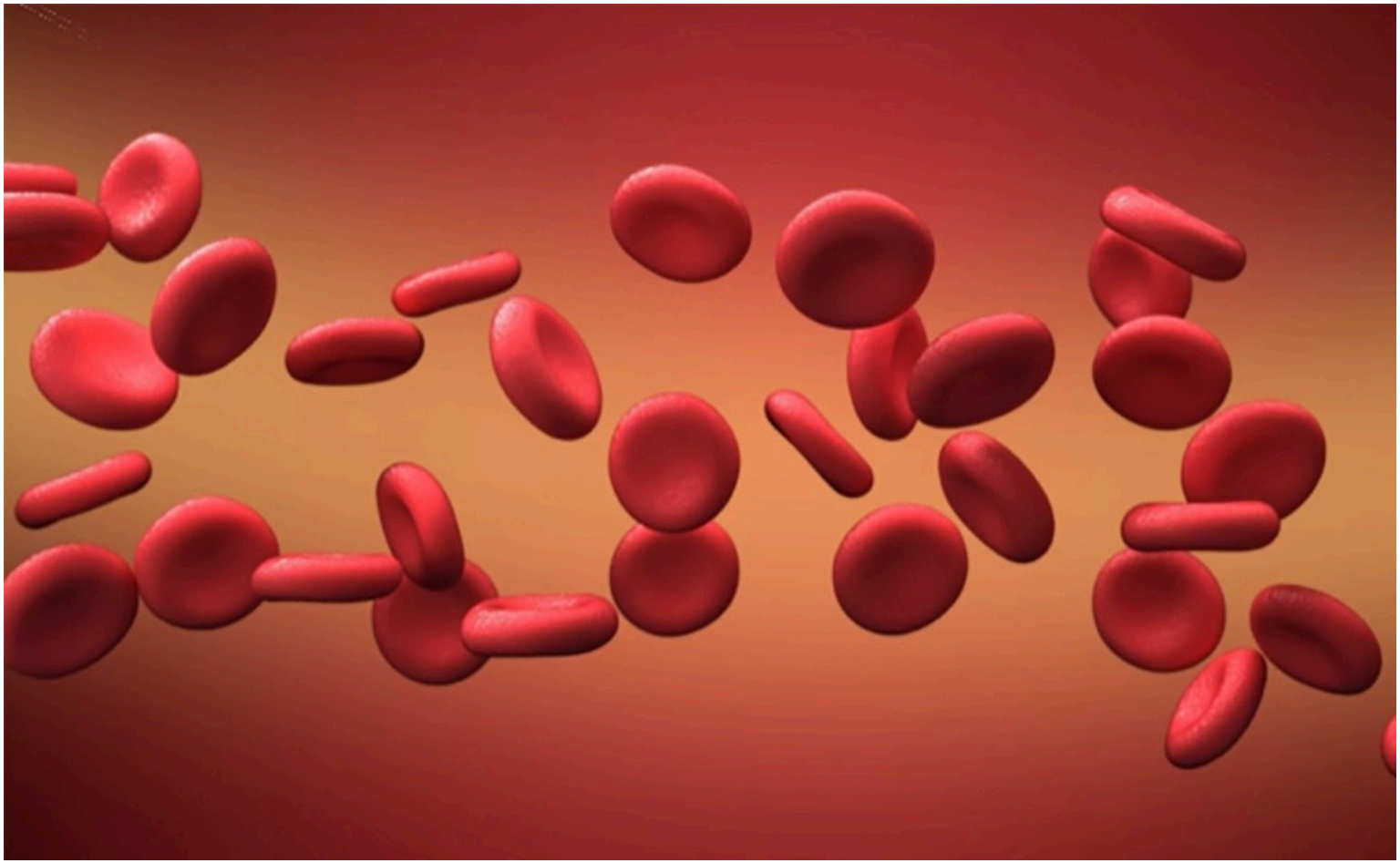
BIOL2107, Fall ’23

Lecture 13



Extensions to Mendelian Inheritance

Incomplete dominance

Codominance

Multiple Alleles

Epistasis

Several Genes

Lethal Alleles

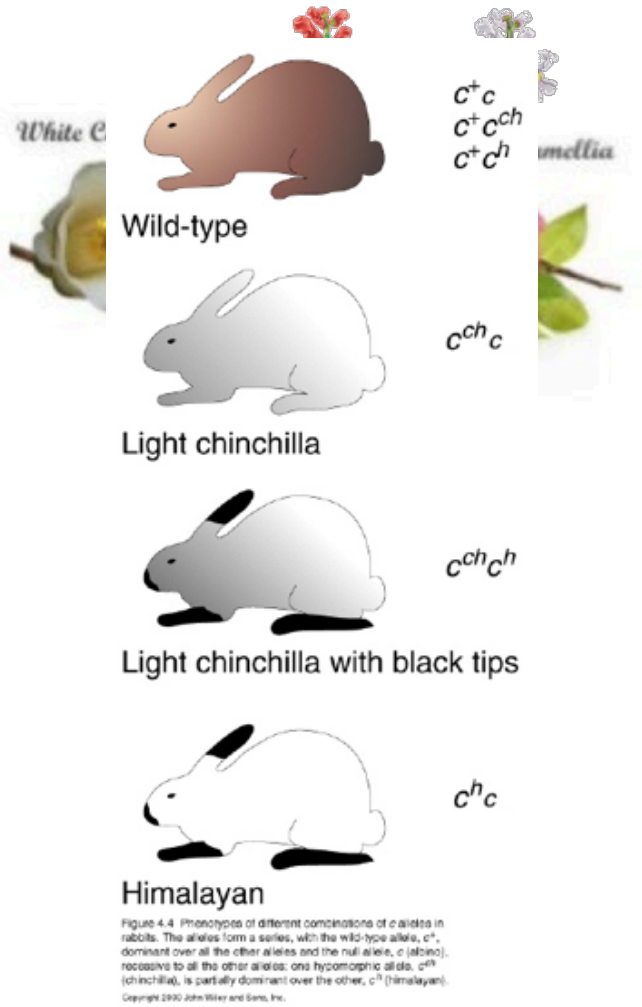
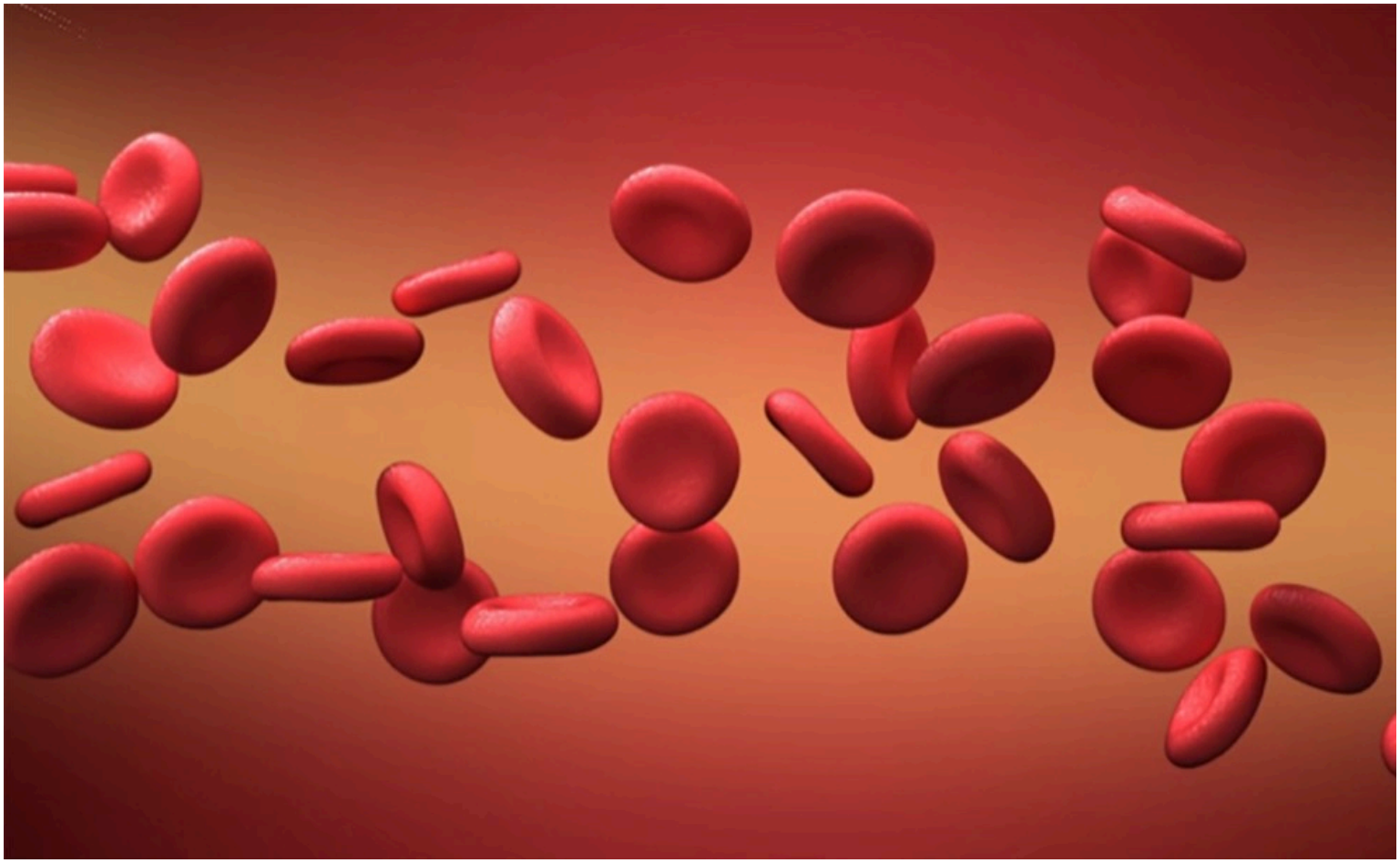


Figure 4.4 Phenotypes of different combinations of *c* alleles in rabbits. The alleles form a series, with the wild-type allele, c^+, dominant over all the other alleles and the null allele, *c* (albino), recessive to all the other alleles; one hypomorphic allele, c^{ch} (chinchilla), is partially dominant over the other, c^h (himalayan).

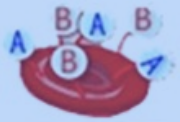
Multiple Alleles



An example of "co-dominant" alleles in humans

The ABO Blood Group System

Antigens: molecules, usually on the outside of a cell, that provoke an immune response

Blood Type	Antibodies Produced	Potential Donors			
					
A		+	-	-	+
B		-	+	-	+
AB	None	+	+	+	+
O		-	-	-	+

The
ABO Blood Group System

Blood Type	Genotypes	ABO Enzymes Present	RBC Antigens Present	Serum Antibodies
"A"	$I^A I^A$, $I^A I^O$	"H", "A"	**A**, H	anti-B
"B"	$I^B I^B$, $I^B I^O$	"H", "B"	**B**, H	anti-A
"AB"	$I^A I^B$	"H", "A", "B"	**A, B**, H	none
"O"	$I^O I^O$	"H"	H	anti-A, anti-B

The
ABO Blood Group System

Blood Type	Genotypes	ABO Enzymes Present	RBC Antigens Present	Serum Antibodies
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"AB"	$I^A I^B$	"H", "A", "B"	**A, B**, H	none
"O"	$I^O I^O$	"H"	H	anti-A, anti-B

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"AB"	$I^A I^B$	"H", "A", "B"	**A, B**, H	none
"O"	$I^O I^O$	"H"	H	anti-A, anti-B

Extensions to Mendelian Inheritance

Incomplete dominance

Codominance

Multiple Alleles

Epistasis

Several Interactive genes

Lethal Alleles

(A)

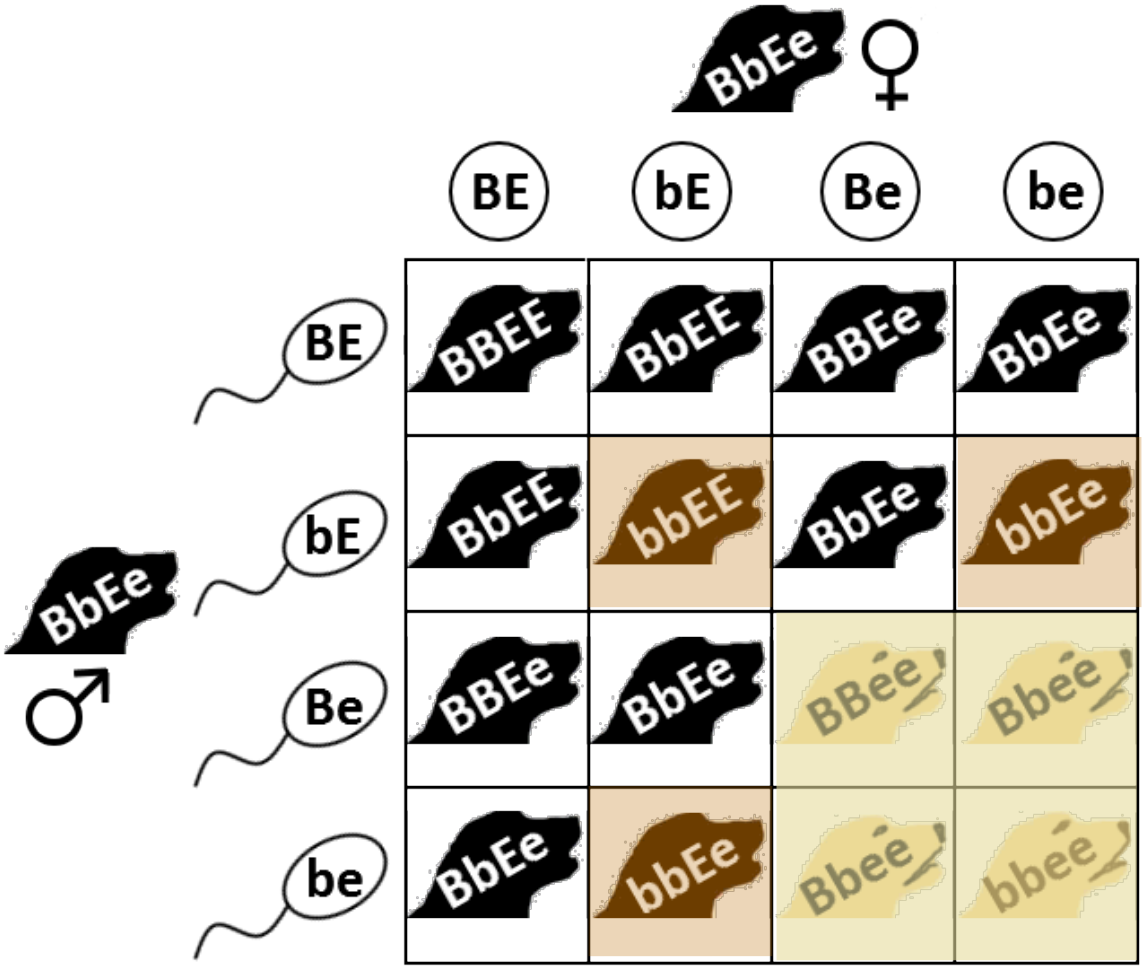
(B)

(C)

Epistasis

Two Genes E and B

Epistasis



9 B-E- Black

3 bbE- Chocolate

4 --ee Yellow

1 is Dudley (bbee)

Extensions to Mendelian Inheritance

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Codominance

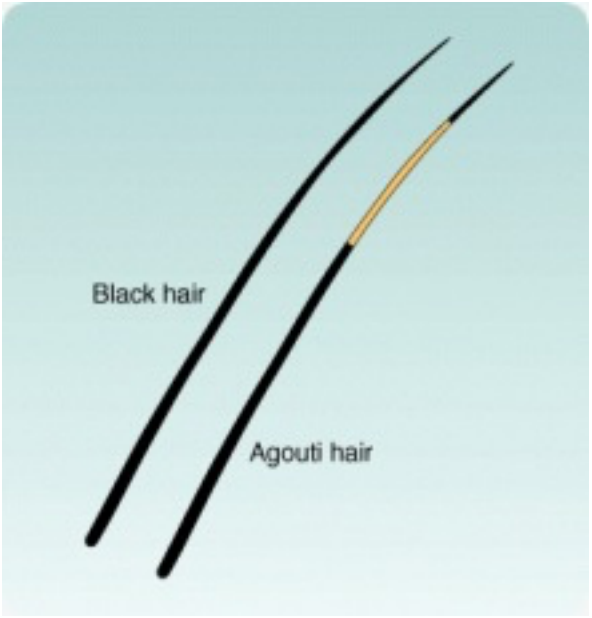
Multiple Alleles

Epistasis

Several Interactive genes

Lethal Alleles

Several Interactive genes



Agouti… rodents

Several Interactive genes

Several Interactive genes

A Parental cross between a **AA, BB** and a **aa, bb, result**s in **Aa, Bb heterozygote.**

The **A allele** determines a banded pattern, called **agouti**.
The recessive **a** allele results in **unbanded hairs**.
The genotypes **AA** or **Aa** are, therefore, **agouti.**

The genotypes **BB** or **Bb** result in fur colour that is solidly **black**,
whereas **bb** denotes brown fur colour ..

Several Interactive genes

A 3rd gene at the **C locus, which is an entirely different locus from either A or B-** determines if any colouration occurs at all.

The genotypes **CC** and **Cc** allow colour to show through, whereas the double recessive **cc** is **albino**, which does not allow any colour to show through, as the **cc genotype** blocks ALL pigment production -note the eyes are now pink.

Several Interactive genes

An **F2 phenotypic ratio** of an initial parental cross between an **AA, BB** and an **aa, bb** (where the **C gene** is present as **CC** or **Cc** and does not interfere) would be:

9 agouti fur, **3 cinammon fur (brown, agouti),** **3 black fur** and **1 brown fur**.

The corresponding **genotypes** are:

9 agouti fur (1 BB, AA + 2 Bb, AA + 4 Bb, Aa): **3 cinnamon** (1 bb, AA + 2 bb, Aa):

3 black fur (1 BB, aa + 2 Bb, aa) and 1 brown fur (1 bb, aa).

Several Interactive genes

An **F2 phenotypic ratio** of an initial parental cross between an **AA, BB** and an **aa, bb** (where the **C gene** is present as **CC** or **Cc** and does not interfere) would be:

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3 black fur (1 BB, aa + 2 Bb, aa) and 1 brown fur (1 bb, aa).

Several Interactive genes

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3 black fur (1 BB, aa + 2 Bb, aa) and 1 brown fur (1 bb, aa).

Several Interactive genes

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The corresponding **genotypes** are:

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3 black fur (1 BB, aa + 2 Bb, aa) and 1 brown fur (1 bb, aa).

Several Interactive genes

An **F2 phenotypic ratio** of an initial parental cross between an **AA, BB** and an **aa, bb** (where the **C gene** is present as **CC** or **Cc** and does not interfere) would be:

9 agouti fur, **3 cinammon fur (brown, agouti),** **3 black fur** and **1 brown fur**.

The corresponding **genotypes** are:

9 agouti fur (1 BB, AA + 2 Bb, AA + 4 Bb, Aa): **3 cinnamon** (1 bb, AA + 2 bb, Aa):

3 black fur (1 BB, aa + 2 Bb, aa) and 1 brown fur (1 bb, aa).

Several Interactive genes

An **F2 phenotypic ratio** of an initial parental cross between an **AA, BB** and an **aa, bb**

Note that when the **C gene** is present as **cc** **ALL** mice are ALBINO -pink eyes

Which provides another example of **EPISTASIS**

Extensions to Mendelian Inheritance

Incomplete dominance

Codominance

Multiple Alleles

Epistasis

Several Interactive genes

Lethal Alleles

Lethal Alleles

Agouti… rodents

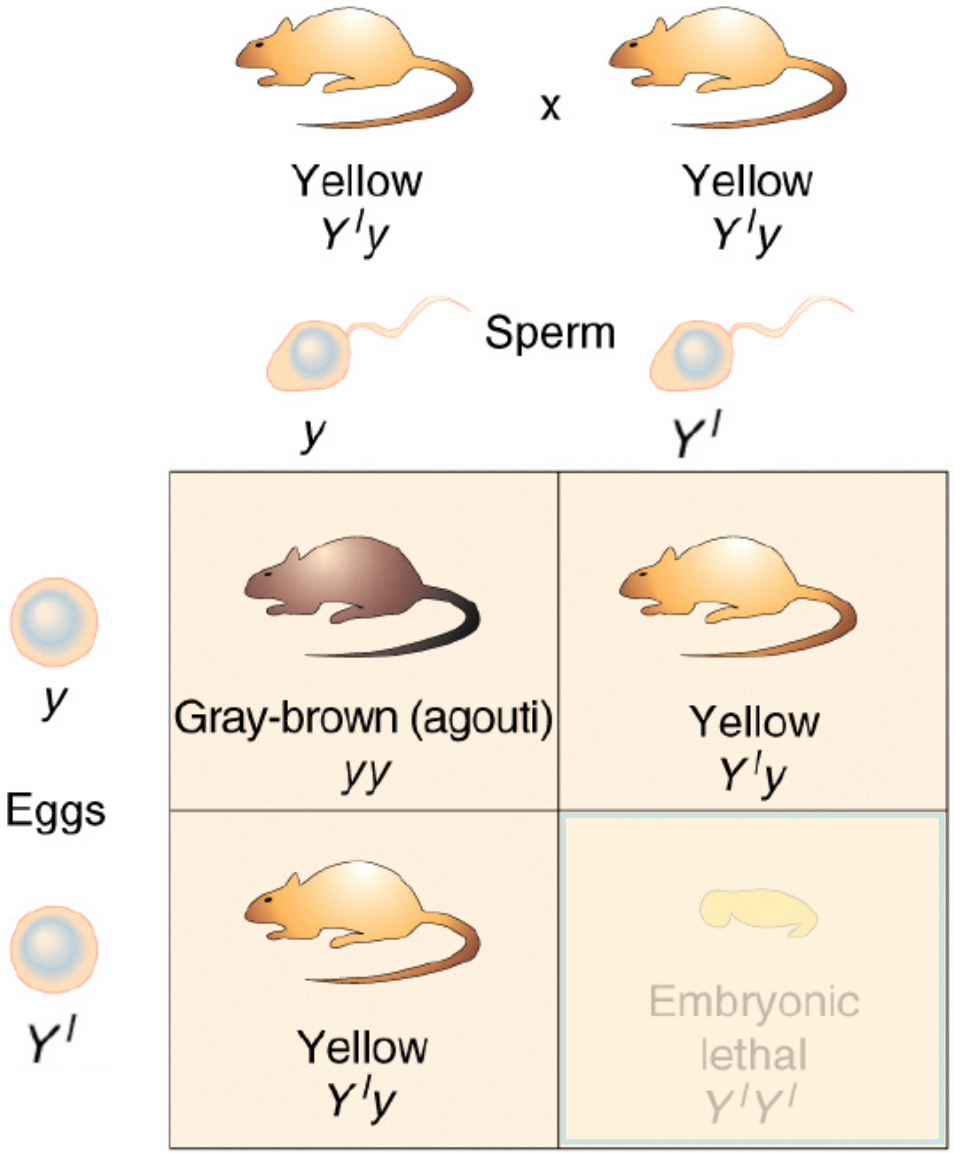


Figure 4.7 Y^l, the yellow-lethal mutation in mice: a dominant visible that is also a recessive lethal. A cross between carriers of this mutation produces yellow heterozygotes and gray-brown (agouti) homozygotes in a ratio of 2:1. The yellow homozygotes die as embryos.

Lethal Alleles

Y' is often designated $\mathbf{A}^{y}$ which is dominant over **y** or the regular agouti allele

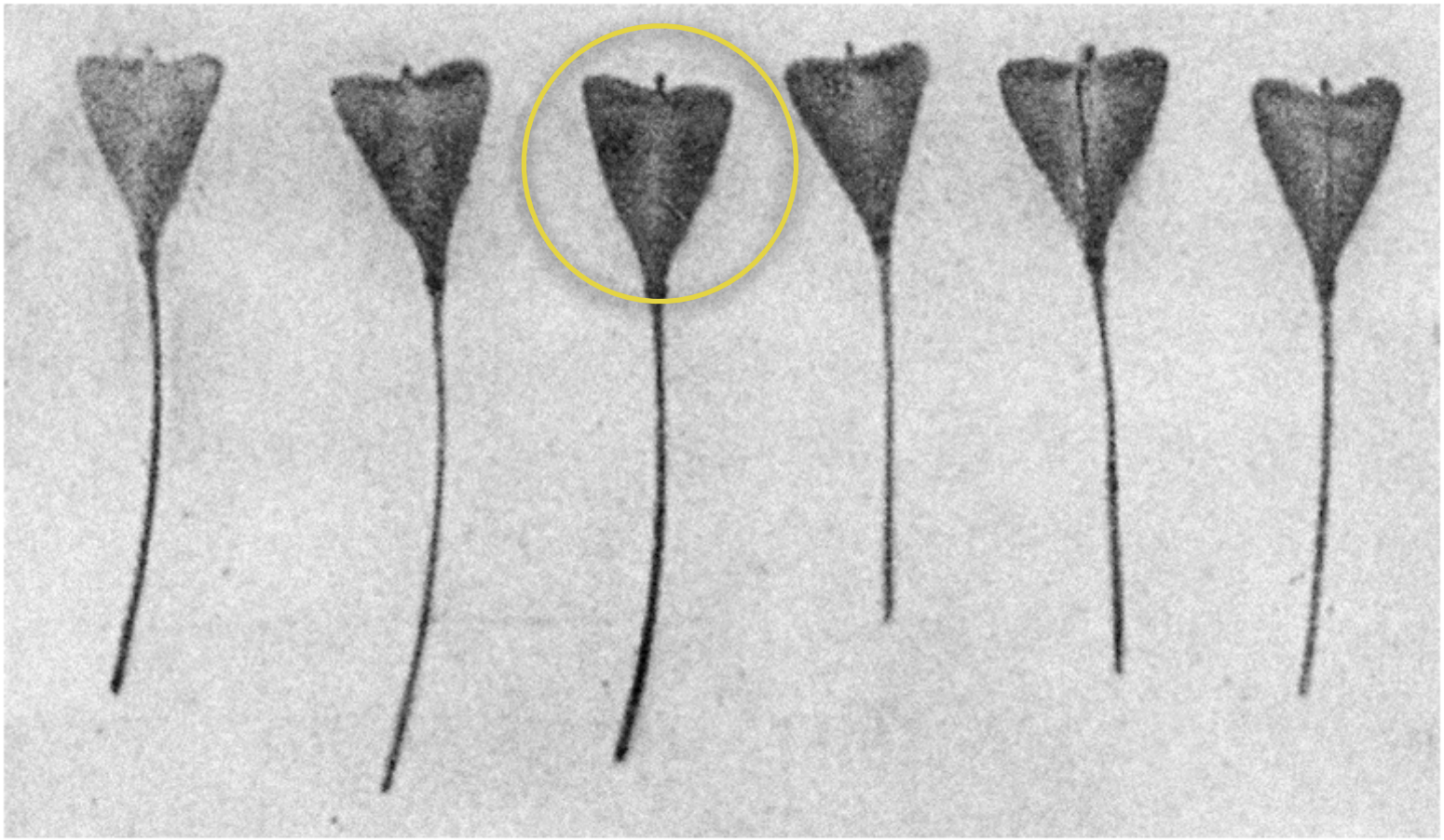
mono hybrid cross of Y'y heterozygotes…
2 (+0): 1 ratio

eg. of pleiotropy

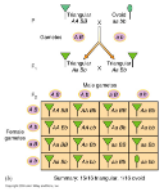
Duplicate Genes

Shepherd’s Purse

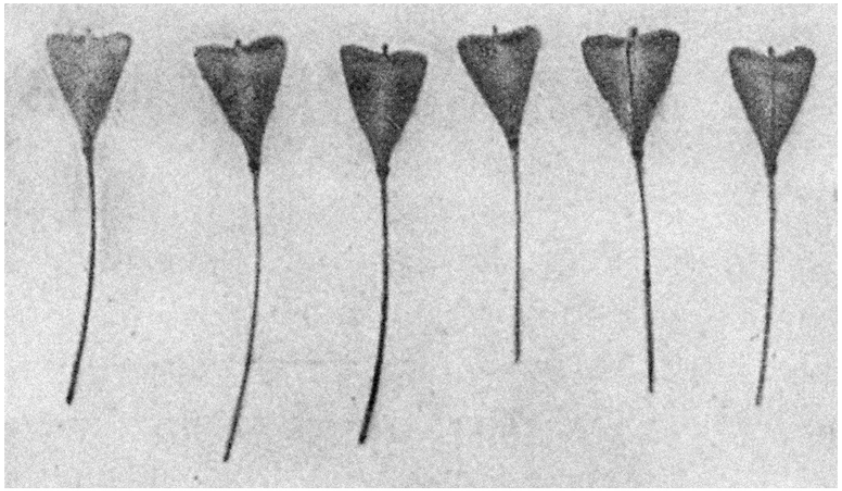
Duplicate Genes



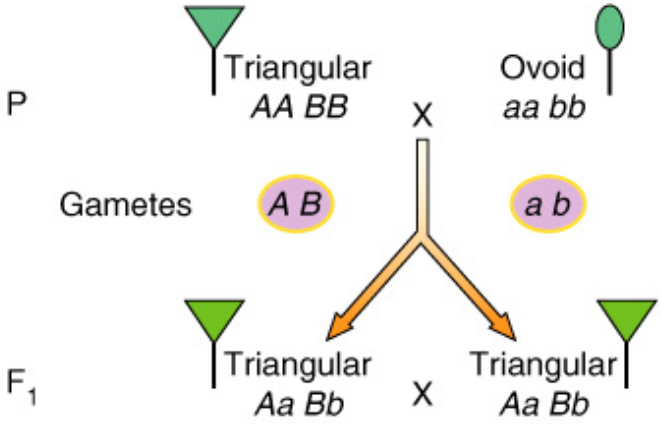
Courtesy New York Public Library

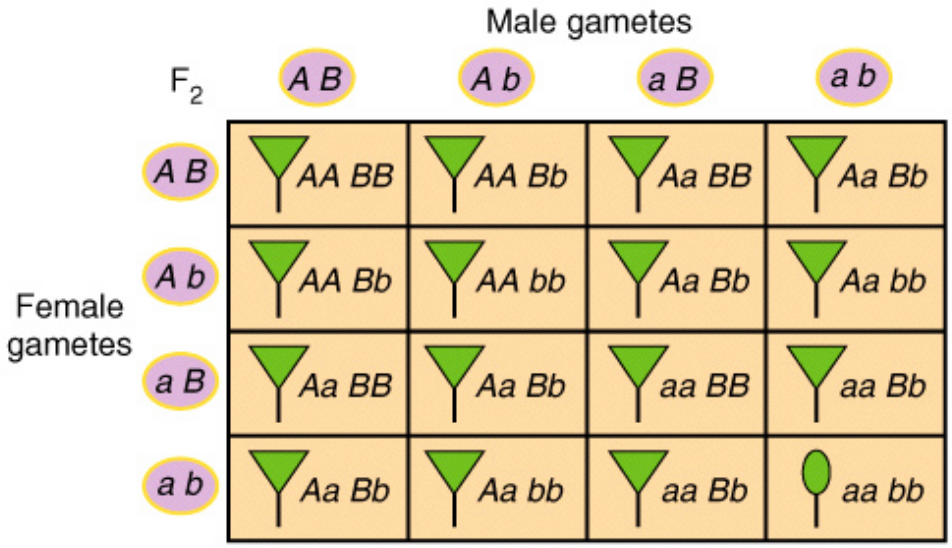


Duplicate Genes



Courtesy New York Public Library





(b) Summary: 15/16 triangular, 1/16 ovoid

(a)

P: White *CC pp* × White *cc PP*

Gametes: *C p* and *c P*

F_1: Purple *Cc Pp* × Purple *Cc Pp*

Male gametes

Female gametes	*C P*	*C p*	*c P*	*c p*
C P	*CC PP* Purple	*CC Pp* Purple	*Cc PP* Purple	*Cc Pp* Purple
C p	*CC Pp* Purple	*CC pp* White	*Cc Pp* Purple	*Cc pp* White
c P	*Cc PP* Purple	*Cc Pp* Purple	*cc PP* White	*cc Pp* White
c p	*Cc Pp* Purple	*Cc pp* White	*cc Pp* White	*cc pp* White

(b)

Summary: 9/16 purple, 7/16 white

Complementary Genes

Need BOTH interactive genes to be either in the **Homozygous dominant** (CC, PP) or **Heterozygous** (Cc, Pp) for one of the phenotypes to show through….

Multiple or "Poly" Genes

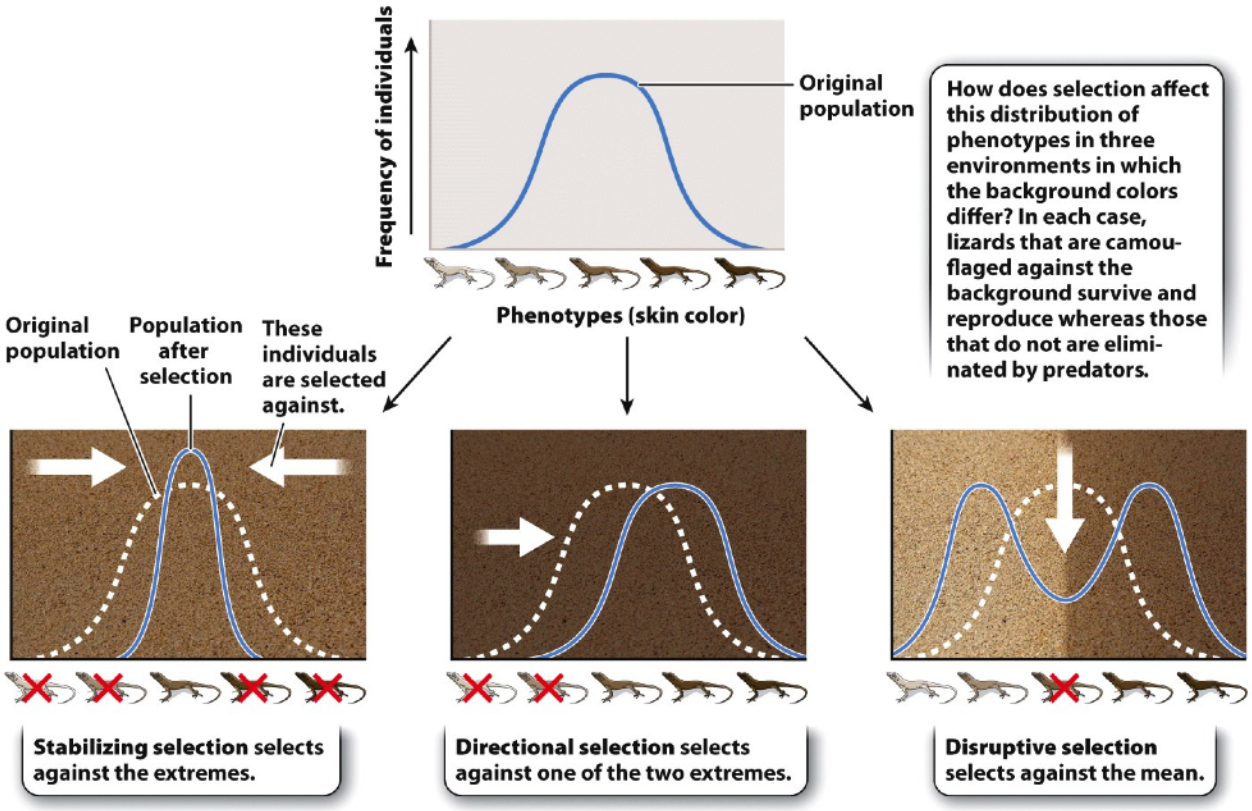


Figure 21.9
Biology: How Life Works

Multiple or “Poly” Genes

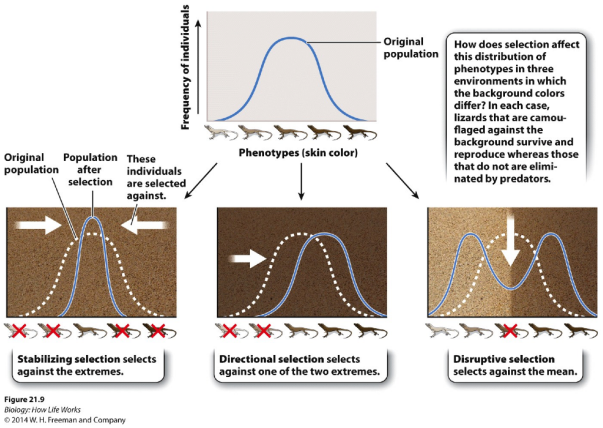


Figure 21.9
Biology: How Life Works

>50 genes heavily involved directly in structural height integrity in humans

Number of genes linked to height revealed by study

Date: October 5, 2014

Source: Boston Children's Hospital

Summary: The largest genome-wide association study to date, involving more than 300 institutions and more than 250,000 subjects, roughly doubles the number of known gene regions influencing height to more than 400. The study provides a better glimpse at the biology of height and offers a model for investigating traits and diseases caused by many common gene changes acting together.

Share:

RELATED TOPICS

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- Genes
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- Parkinson's Research
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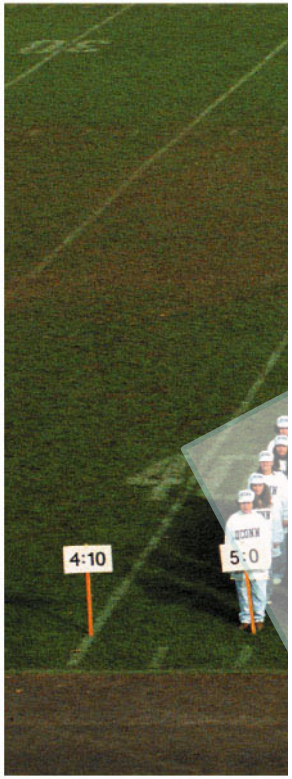
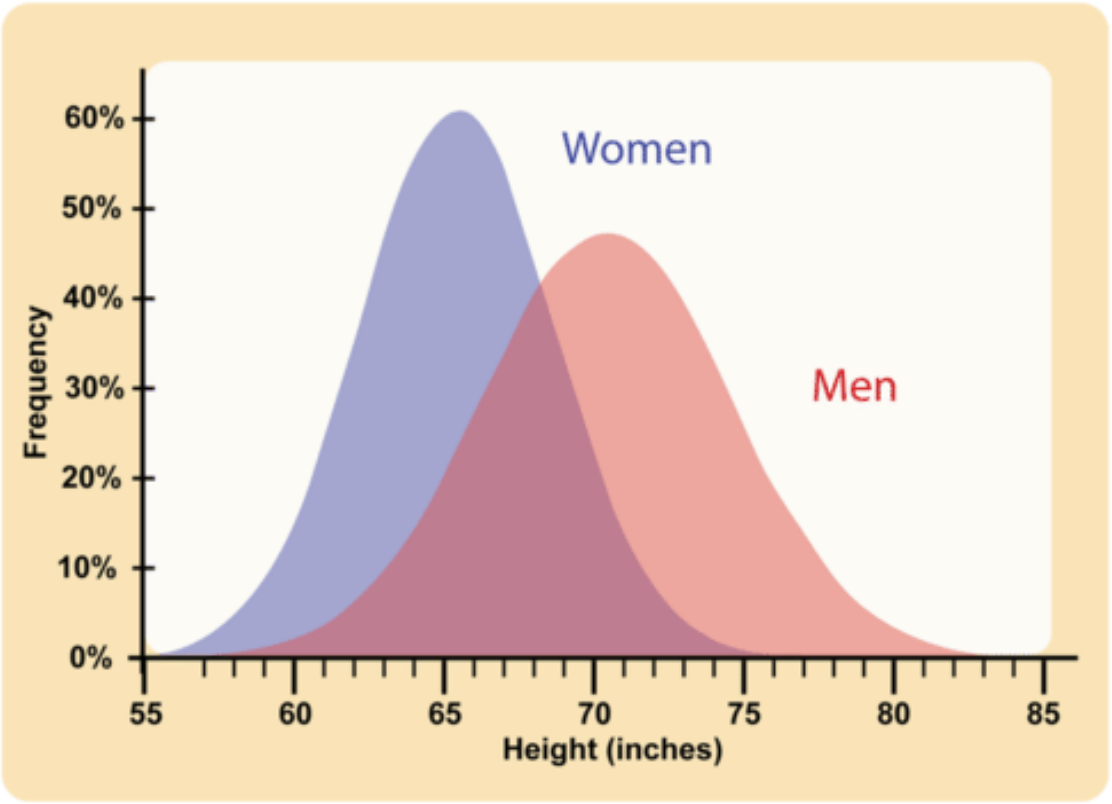
FULL STORY

"We can now explain about 20 percent of the heritability of height, up from about 12 percent where we were before," says co-first author Tonu Esko, PhD, of Boston Children's Hospital, the Broad Institute and the University of Tartu (Estonia).

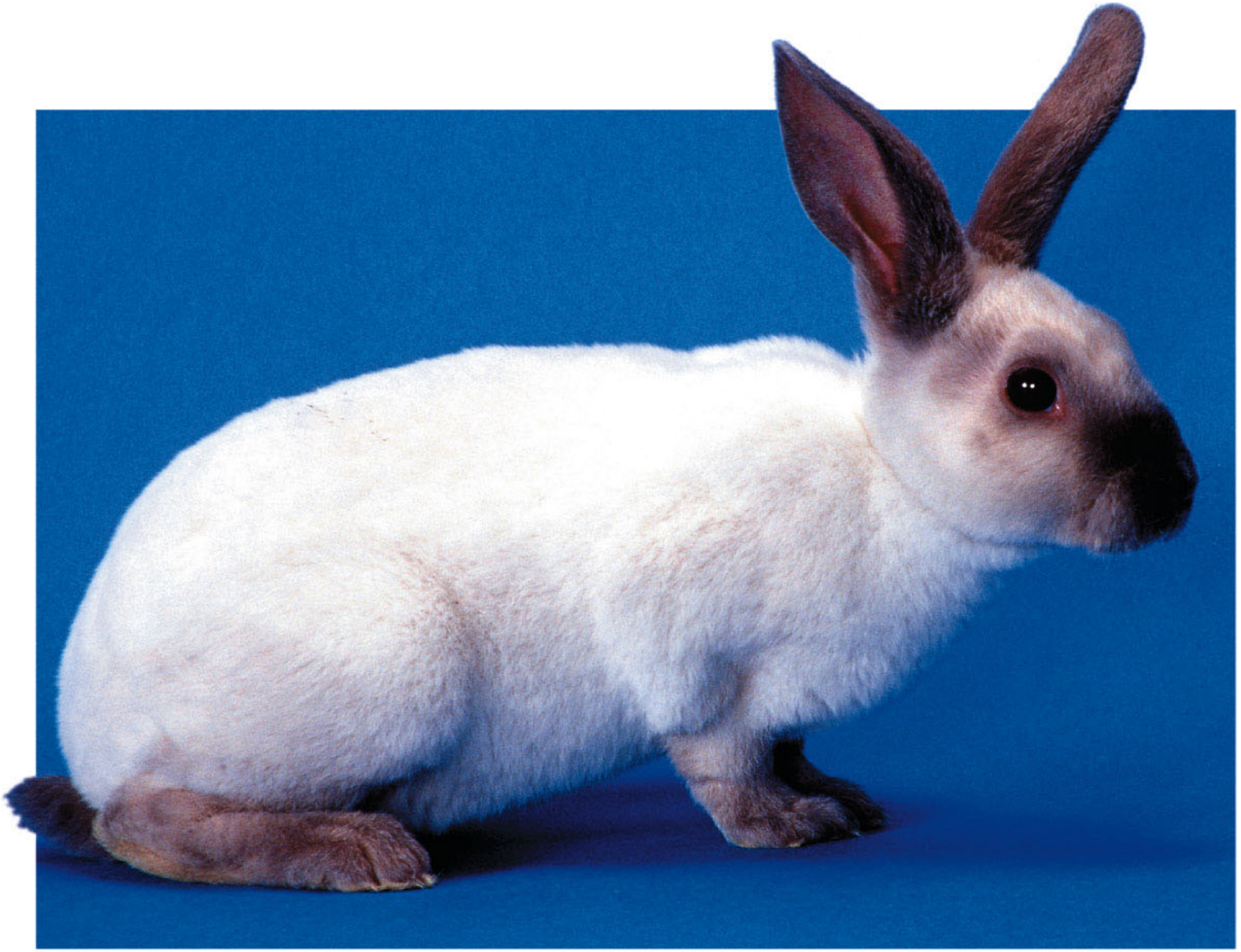
https://www.sciencedaily.com/releases/2014/10/141005134909.htm

Polygenes

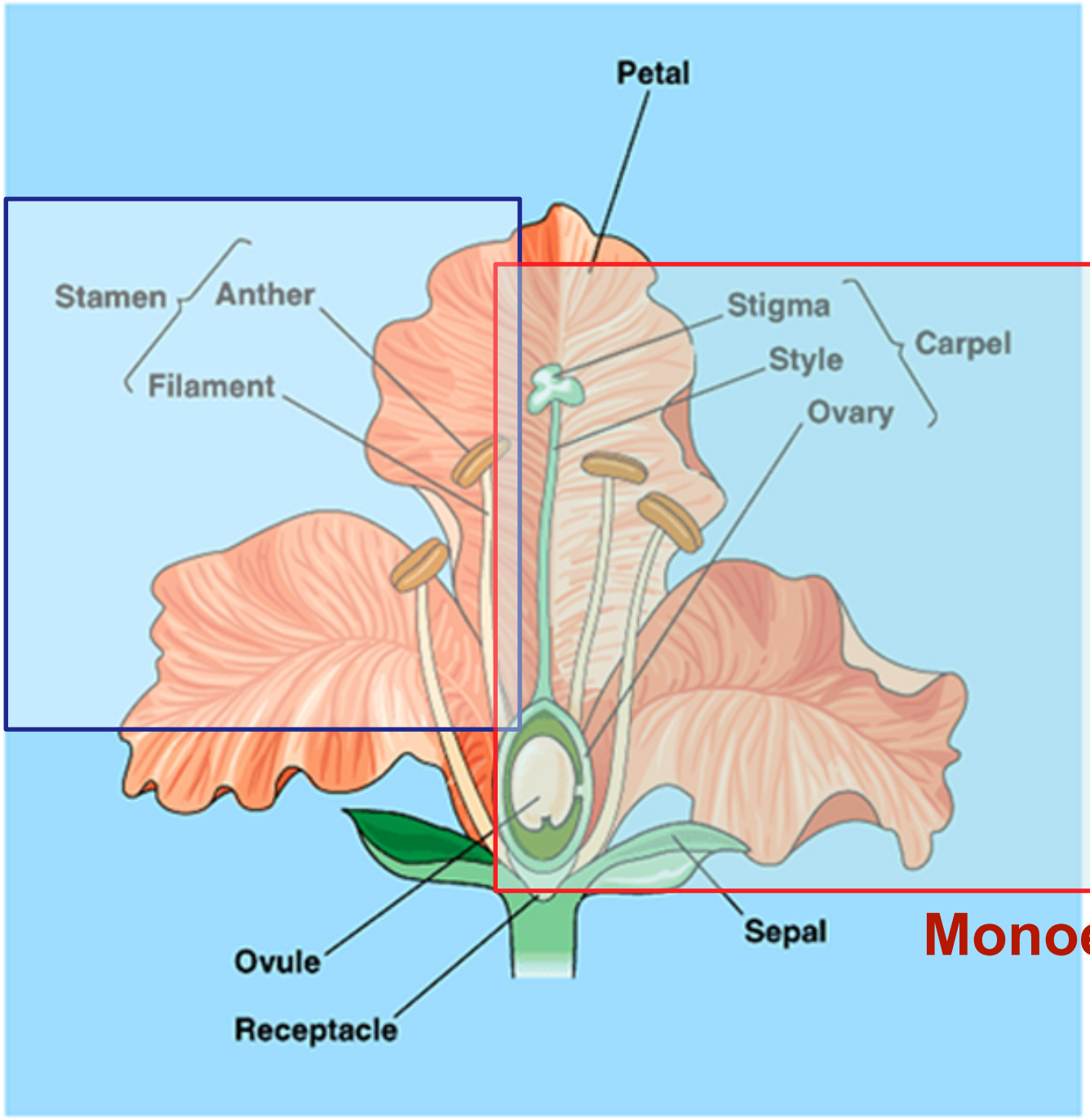




Some of these assorted genes are also subject to female- and male- specific influences



Nature vs. Nurture

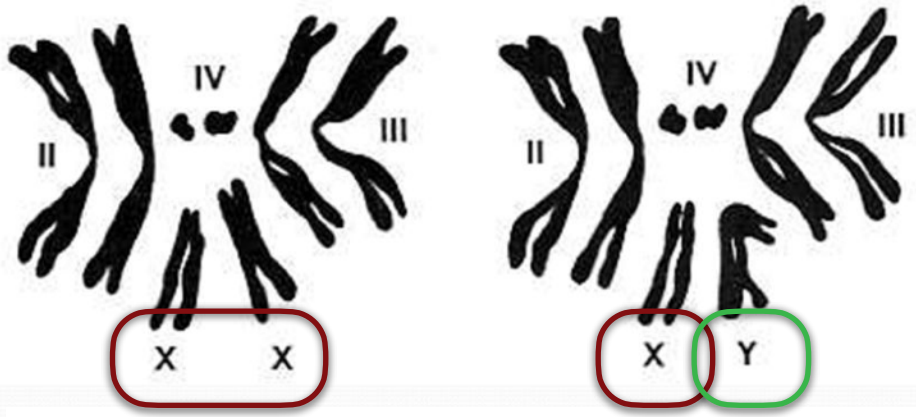

Petal
Stamen
Anther
Filament
Stigma
Style
Ovary
Carpel
Sepal
Ovule
Receptacle

Monoecious

Dioecious

Most species of Holly are dioecious,
meaning **male** and **female flowers** are on different plants
-requiring a male holly plant to pollinate the female, which
produce the red berries.

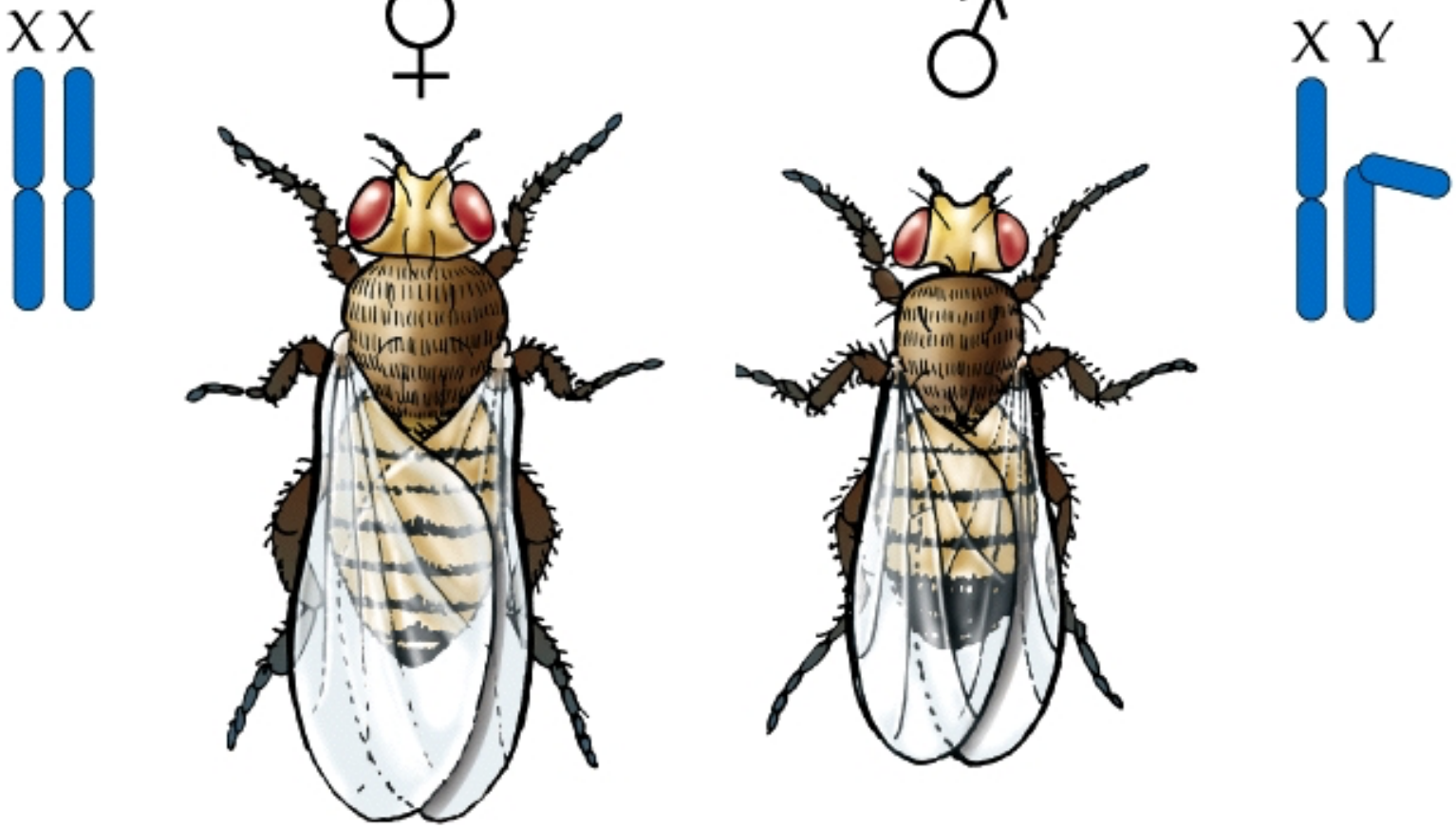
Dioecious



Drosphila melanogaster or "fruit fly"

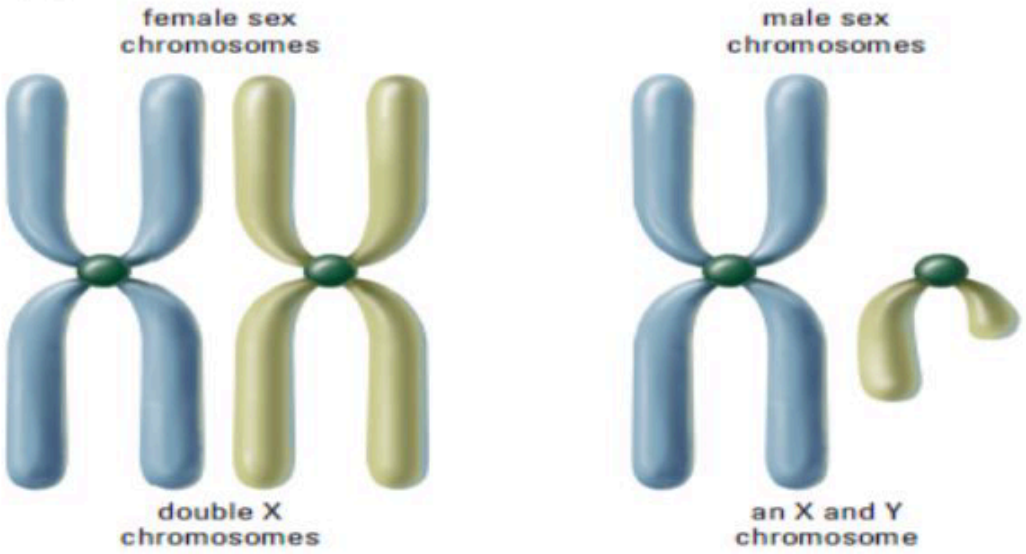
Dioecious

Homo sapiens or "humans"



Dioecious

autosomes: the chromosomes not involved in sex determination

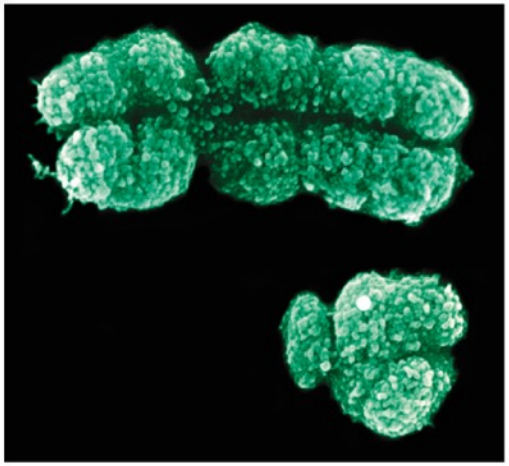


sex chromosomes: the pair of chromosomes that have a role in the sex of an individual

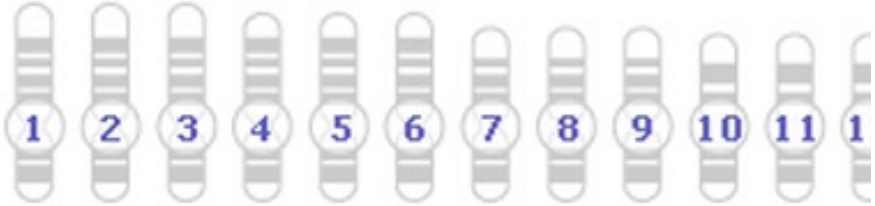
human

Almost none of the genes in the *X* chromosome have counterparts in the *Y* chromosome.

The tips of the arms of the *X* and *Y* chromosomes share a small region of homology (red).



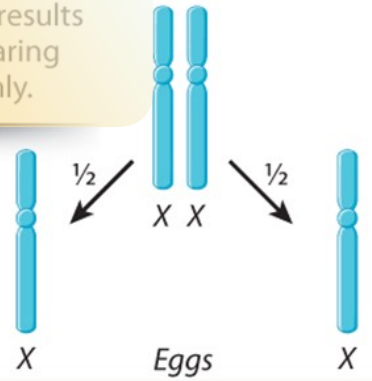
human



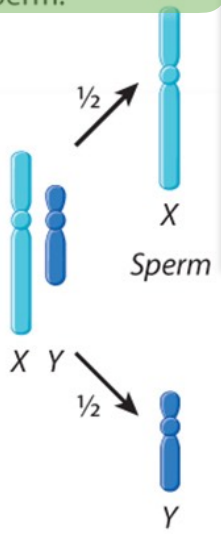
Almost none of the genes in the *X* chromosome have counterparts in the *Y* chromosome.

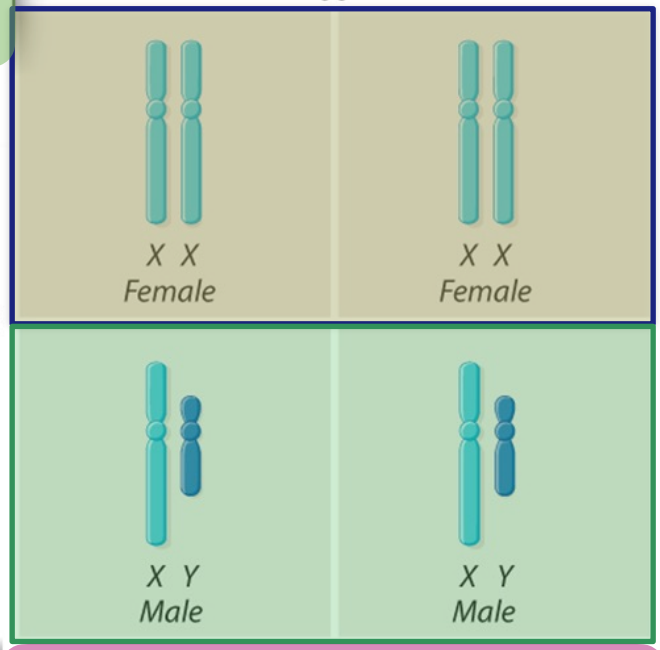
The tips of the arms of the *X* and *Y* chromosomes share a small region of homology (red).

Meiosis in a female results in *X*-bearing eggs only.



Meiosis in a male results in a 1:1 ratio of *X*-bearing and *Y*-bearing sperm.





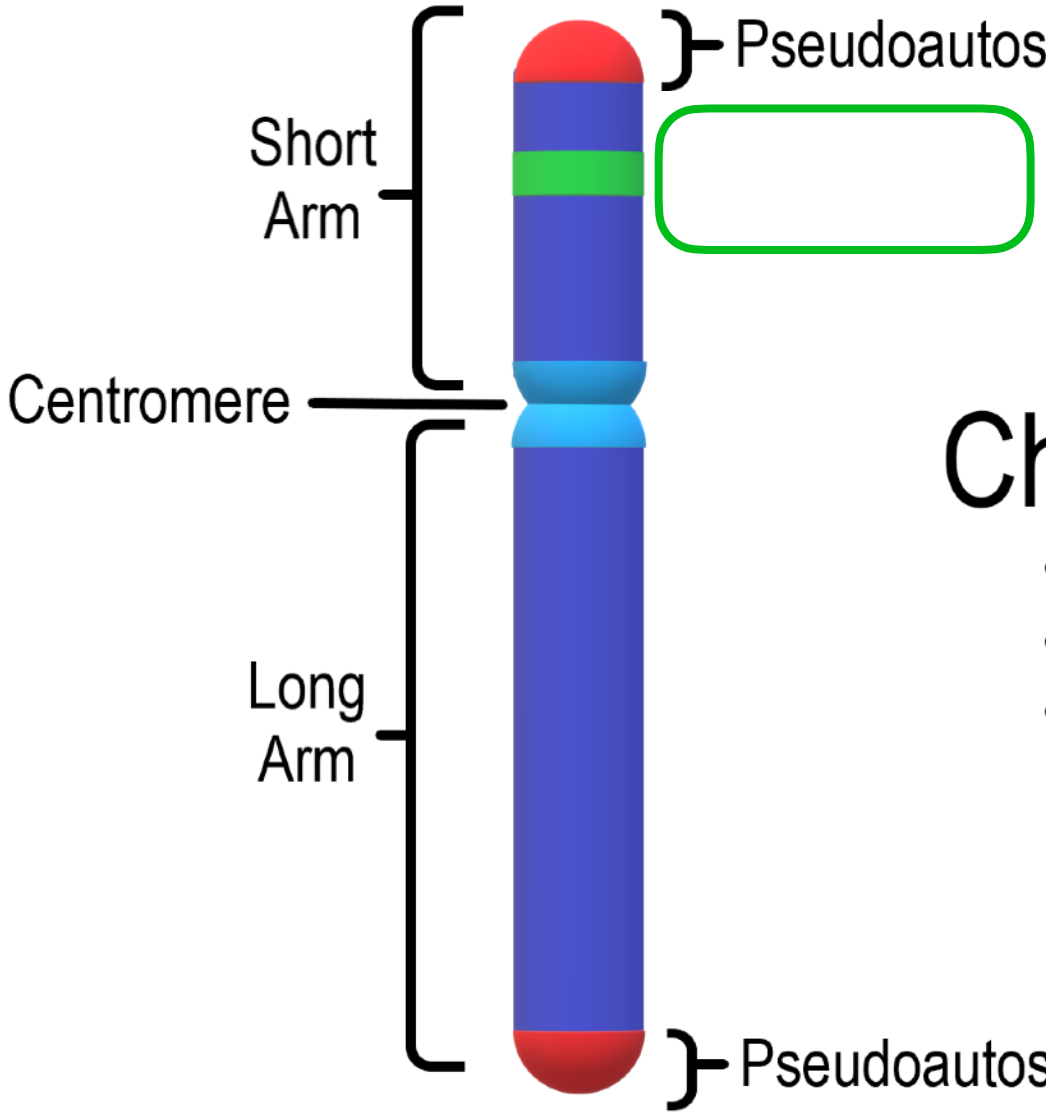
Random fertilization results in an expected ratio of ½ *XX* (female) and ½ *XY* (male) progeny.

Sex-determining Region Y in Mammals

By: *Troy Cox*
Published: *2013-12-31*
Keywords: Sex-Related Gene On Y, Sex Determination Processes

The Sex-determining Region Y (*Sry* in mammals but *SRY* in humans) is a gene found on Y chromosomes that leads to the development of male phenotypes, such as testes. The *Sry* gene, located on the short branch of the Y chromosome, initiates male embryonic development in the XY sex determination system. The *Sry* gene follows the central dogma of molecular biology; the DNA encoding the gene is transcribed into messenger RNA, which then produces a single Sry protein. The Sry protein is also called the testis-determining factor (TDF), a protein that initiates male development in humans, placental mammals, and marsupials. The Sry protein is a transcription factor that can bind to regions of testis-specific DNA, bending specific DNA and activating or enhancing its abilities to promote testis formation, marking the first step towards male, rather than female, development in the embryo.

In humans the first step in the development of an organism's sex is the inheritance of an X chromosome from the mother, and either an X or Y chromosome from the father. Typically, an XX individual develops as a female and an XY individual develops as a male. Studies by University of Kansas zoologist Clarence Erwin McClung in Lawrence, Kansas at the turn of the twentieth century helped researchers focus on the roles of chromosomes for sex determination. McClung theorized that there were two distinct types of spermatozoa, each of which resulted in different forms of fertilized eggs, leading to either male or female development. Nettie Maria Stevens, a post-doctorate researcher at Bryn Mawr College, located near Philadelphia, Pennsylvania, expanded upon McClung's theory in 1905, observing that spermatozoa are of two distinct forms, containing either an X or a Y chromosome. Based upon her research on sex determination in insect species, Stevens concluded that the Y chromosome carries the genetic material that leads to male development.

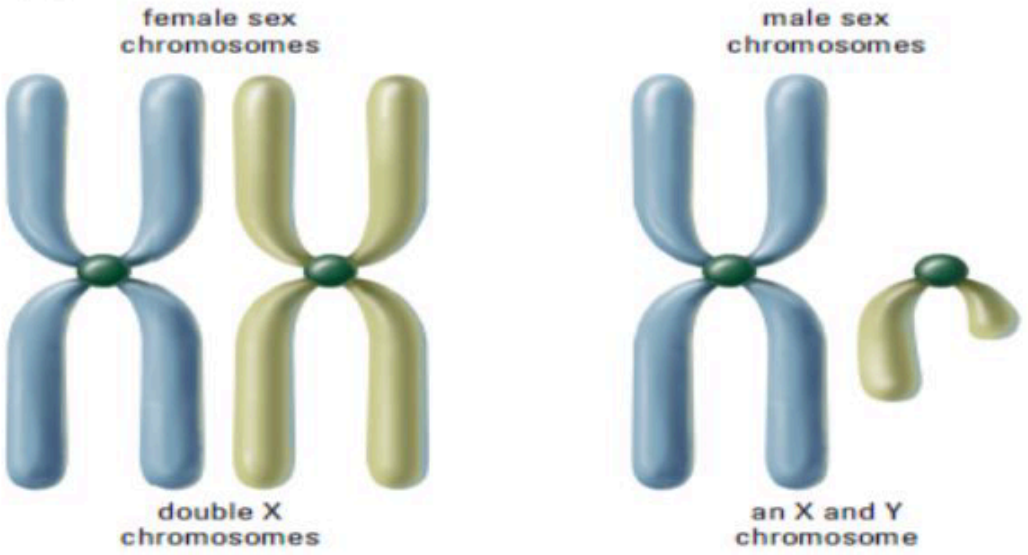


The Y Chromosome

- 59 million base pairs
- Approx. 150 genes
- Identified as sex determining in 1905

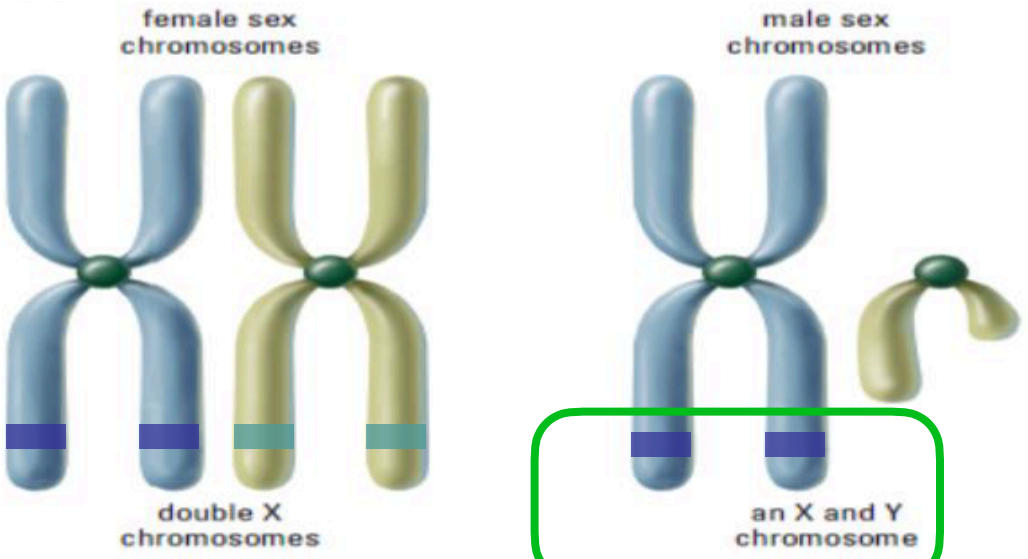
All genes that are present on the X-chromsome, demonstrate a genetic phenomenon called…

X-linkage



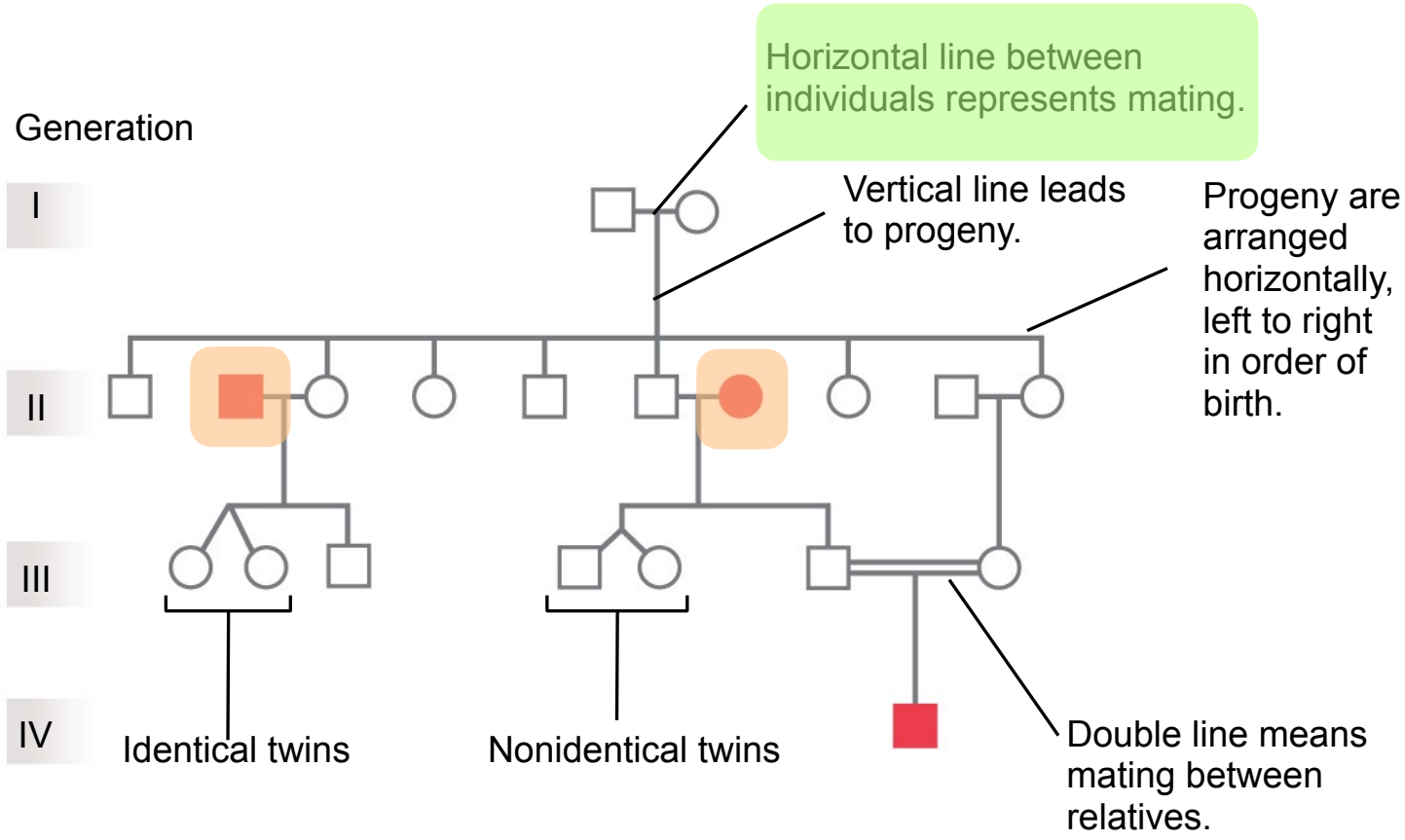
Non-Mendelian Inheritance

X-linkage



hemizygous

Patterns of Inheritance in Humans



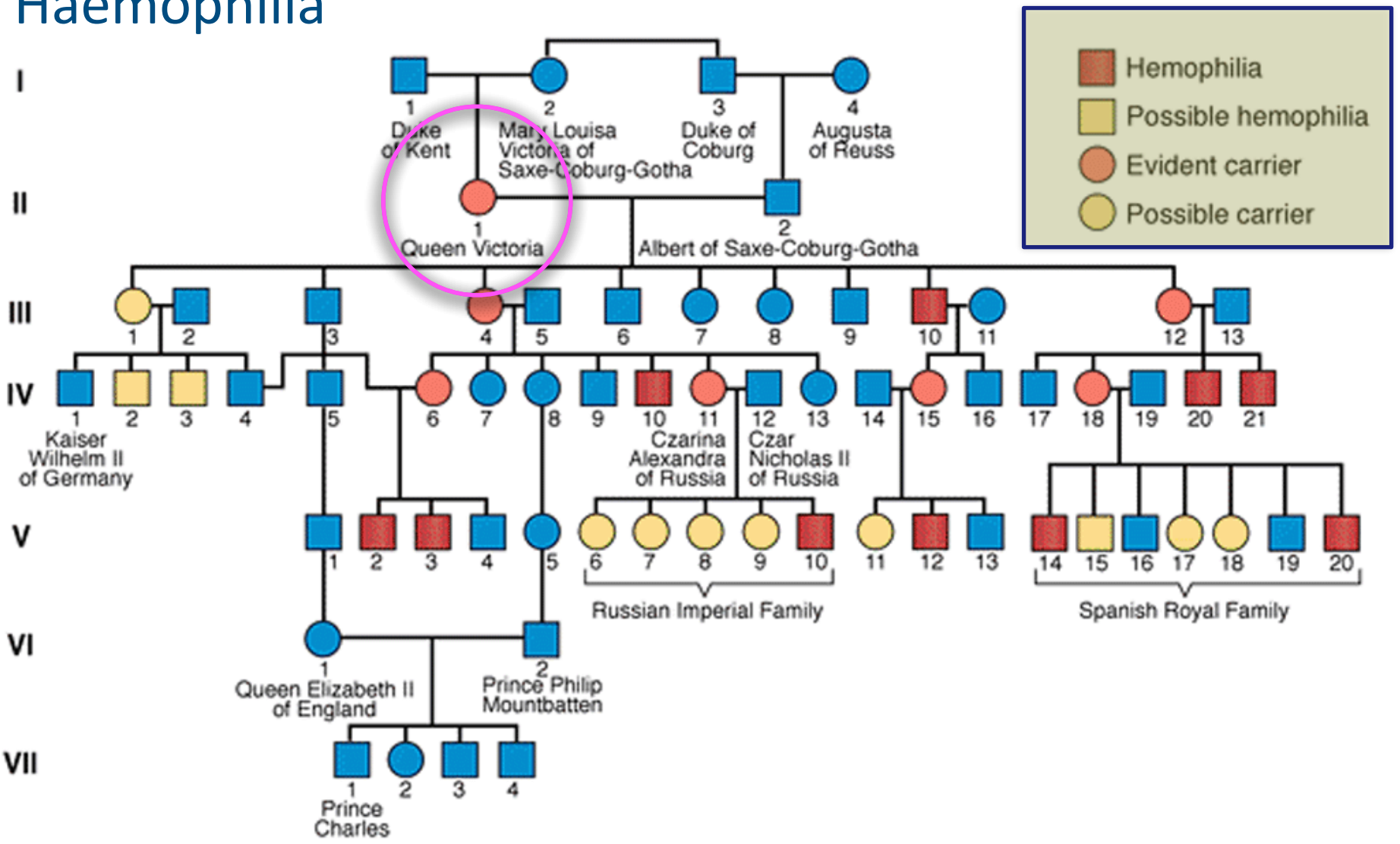
Family Pedigrees

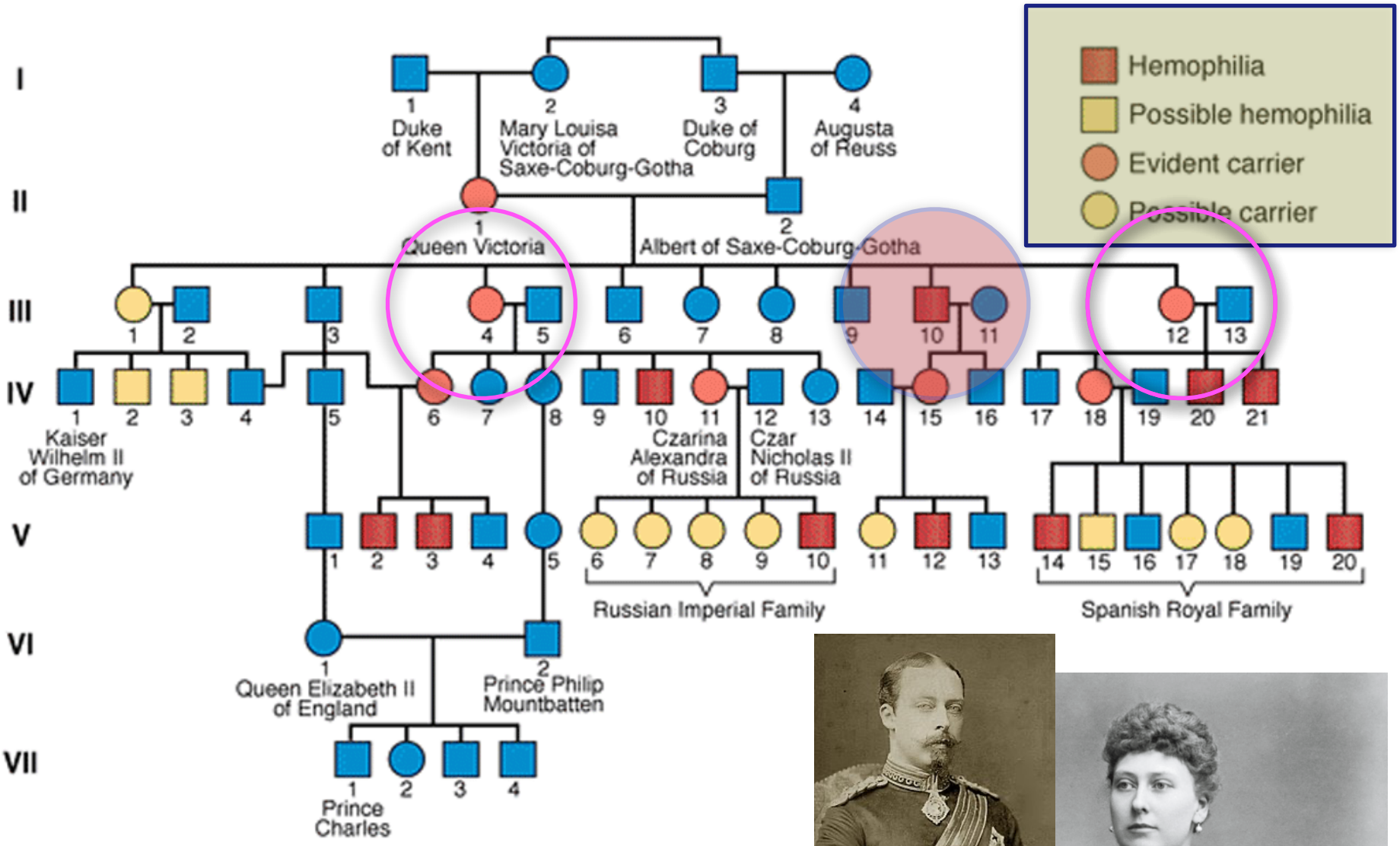
- Female
- Male
- Open symbol means not affected.
- Darkened symbol means affected.

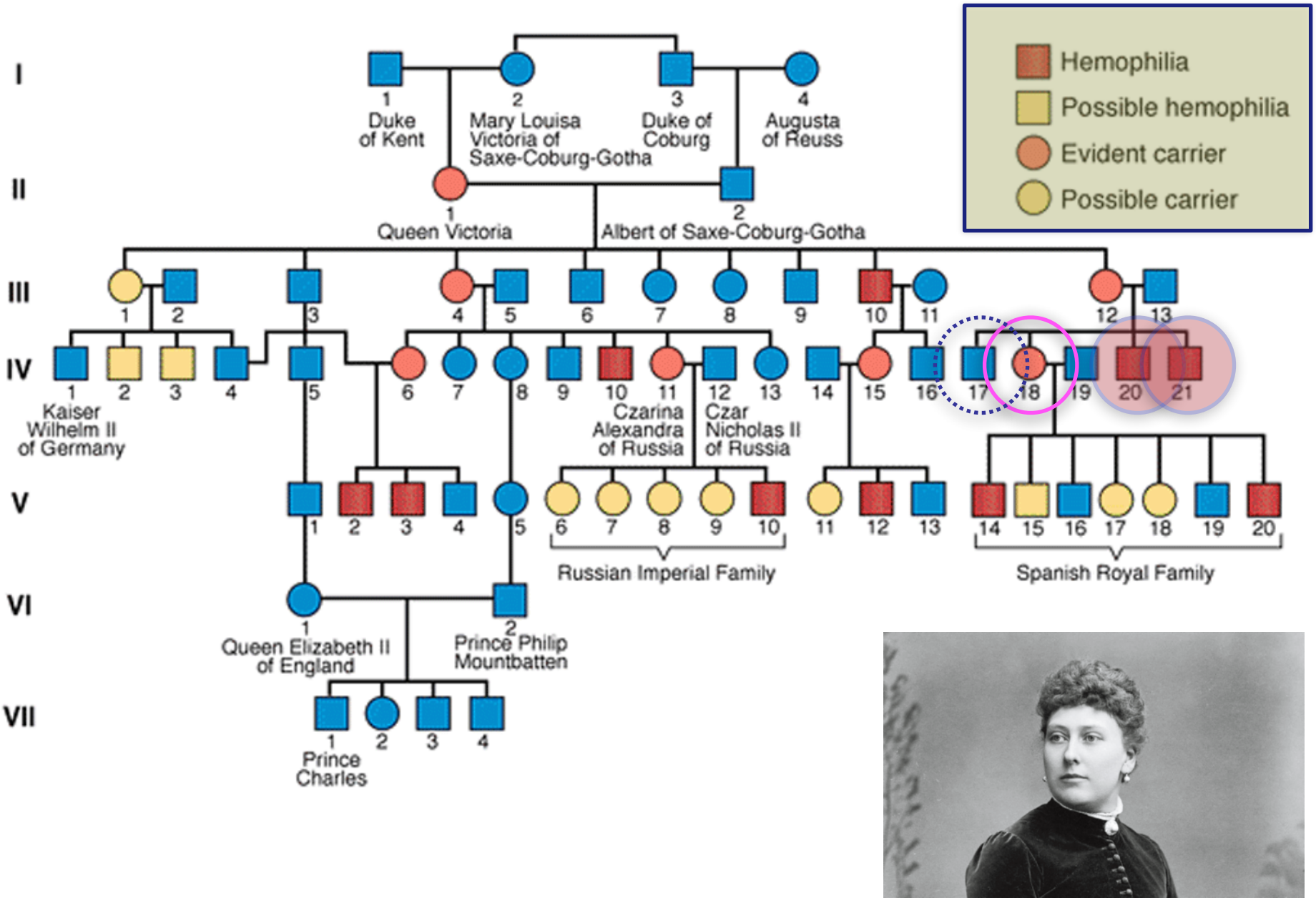
Haemophilia

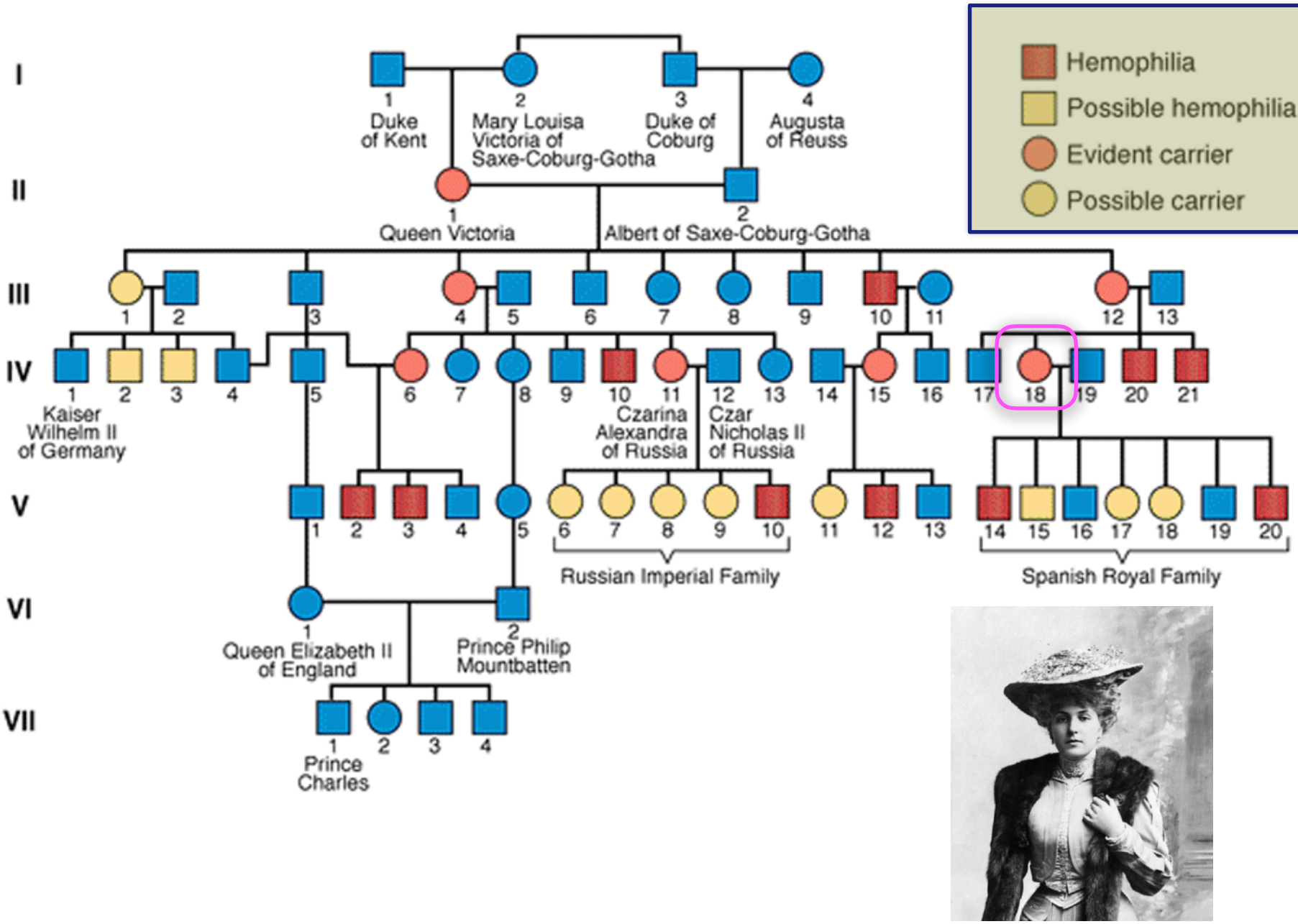
Queen Victoria, nine children,
six of their spouses & twenty three grandchildren

Haemophilia






Hemophilia
Possible hemophilia
Evident carrier
Possible carrier
I
II
III
IV
V
VI
VII
1 Duke of Kent
2 Mary Louisa Victoria of Saxe-Coburg-Gotha
3 Duke of Coburg
4 Augusta of Reuss
1 Queen Victoria
2 Albert of Saxe-Coburg-Gotha
1 Kaiser Wilhelm II of Germany
10 Czarina Alexandra of Russia
12 Czar Nicholas II of Russia
Russian Imperial Family
Spanish Royal Family
1 Queen Elizabeth II of England
2 Prince Philip Mountbatten
1 Prince Charles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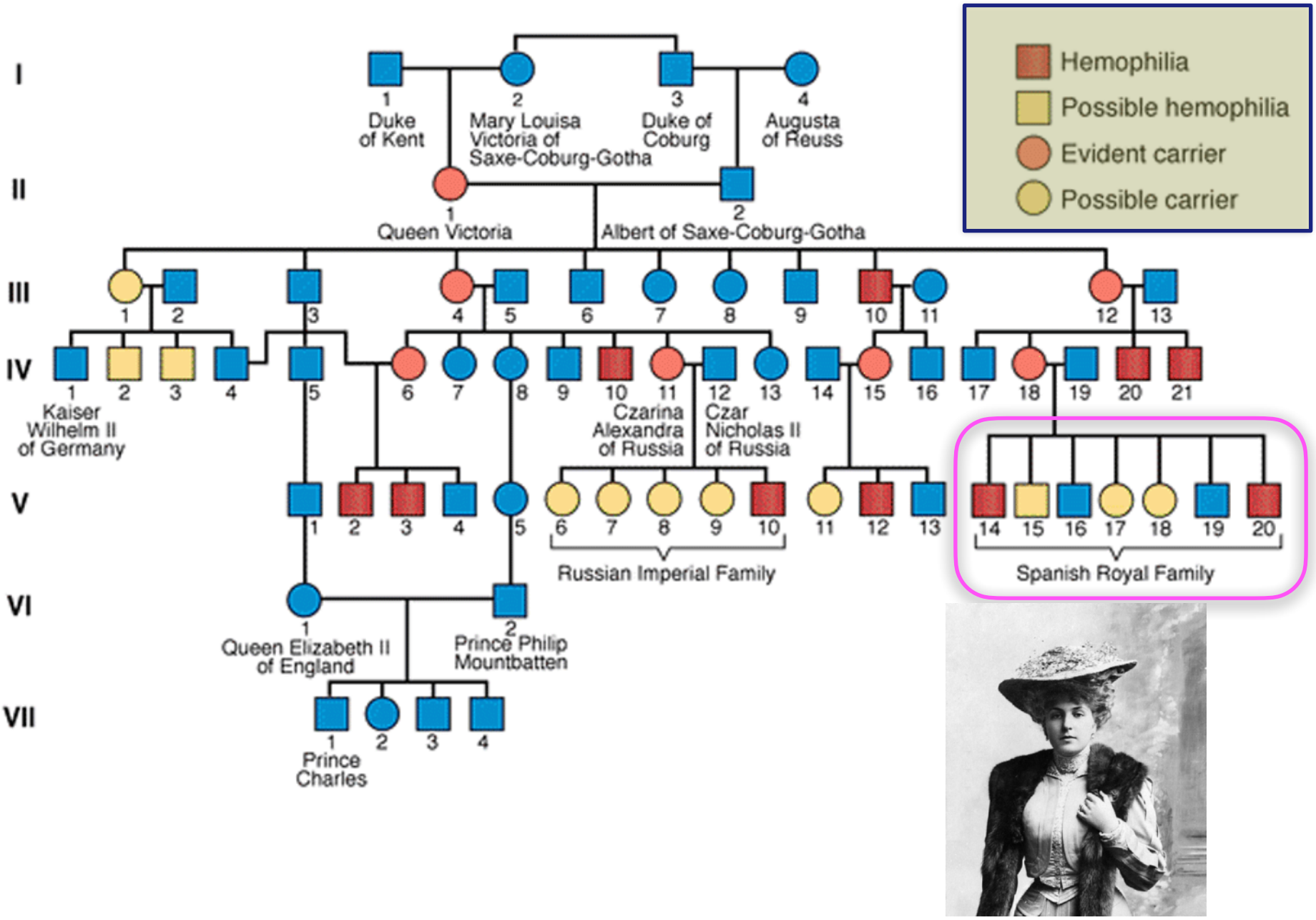
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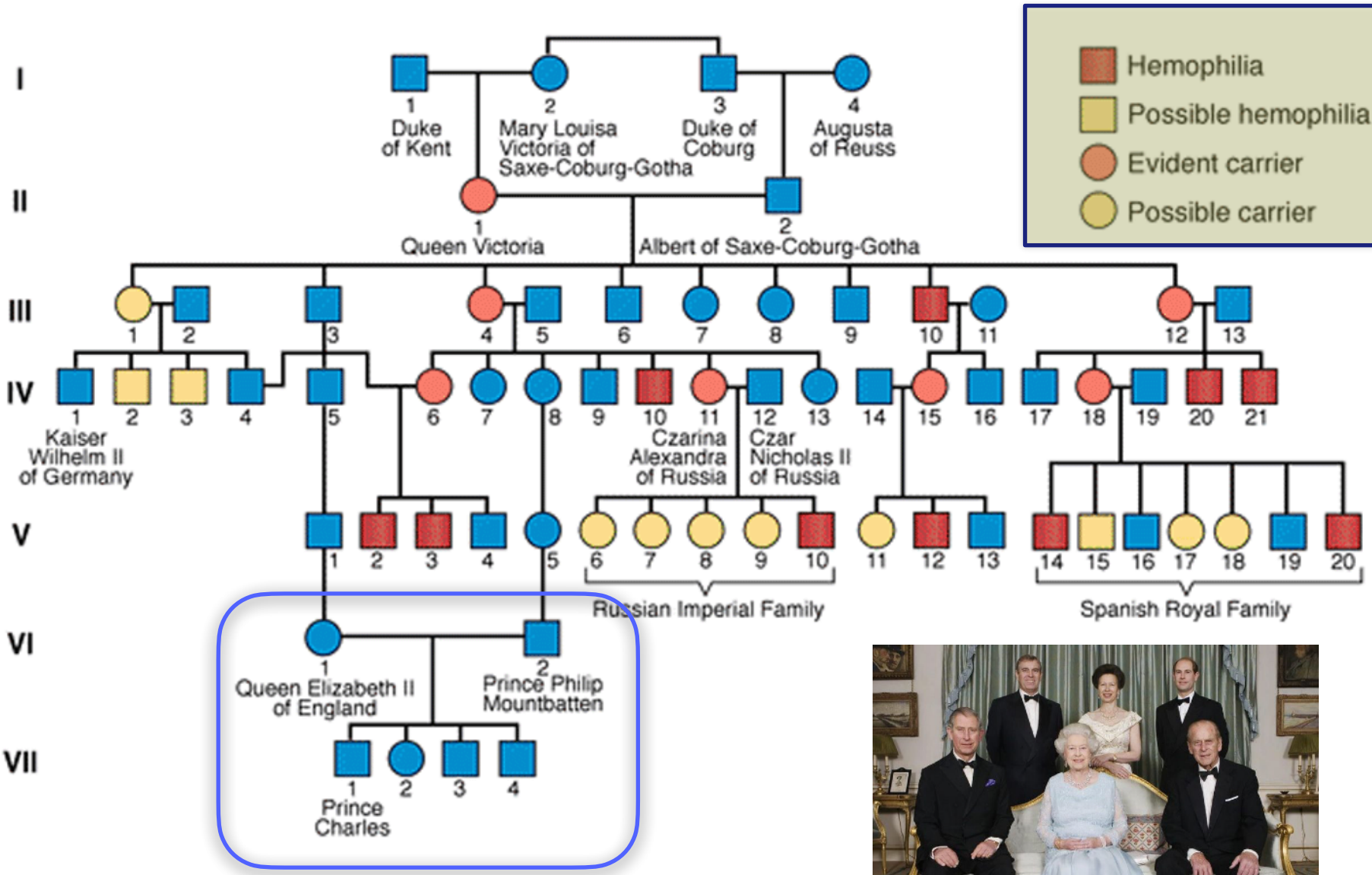
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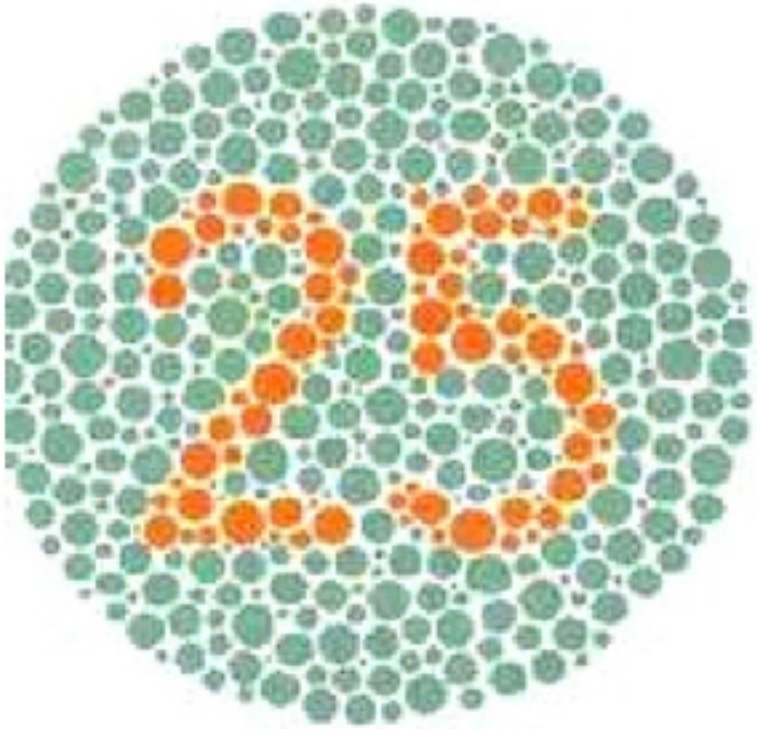
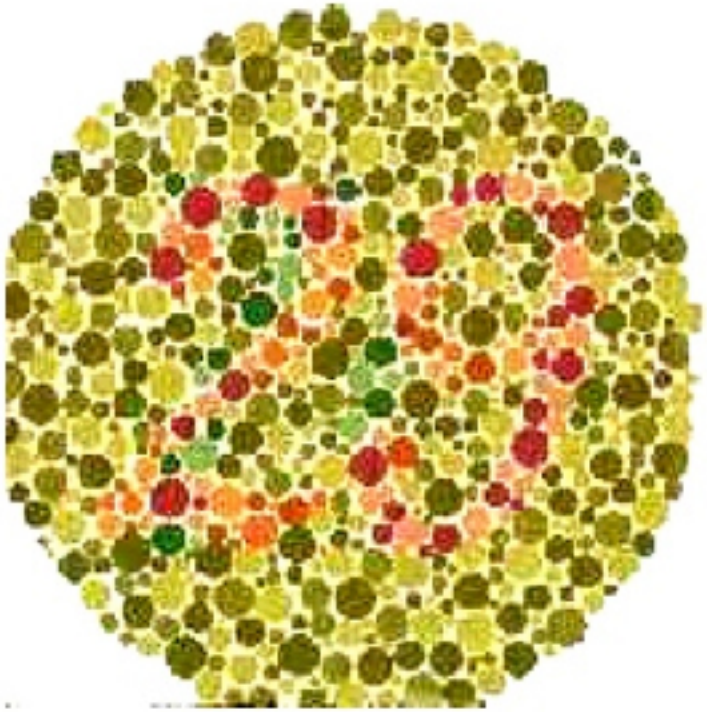
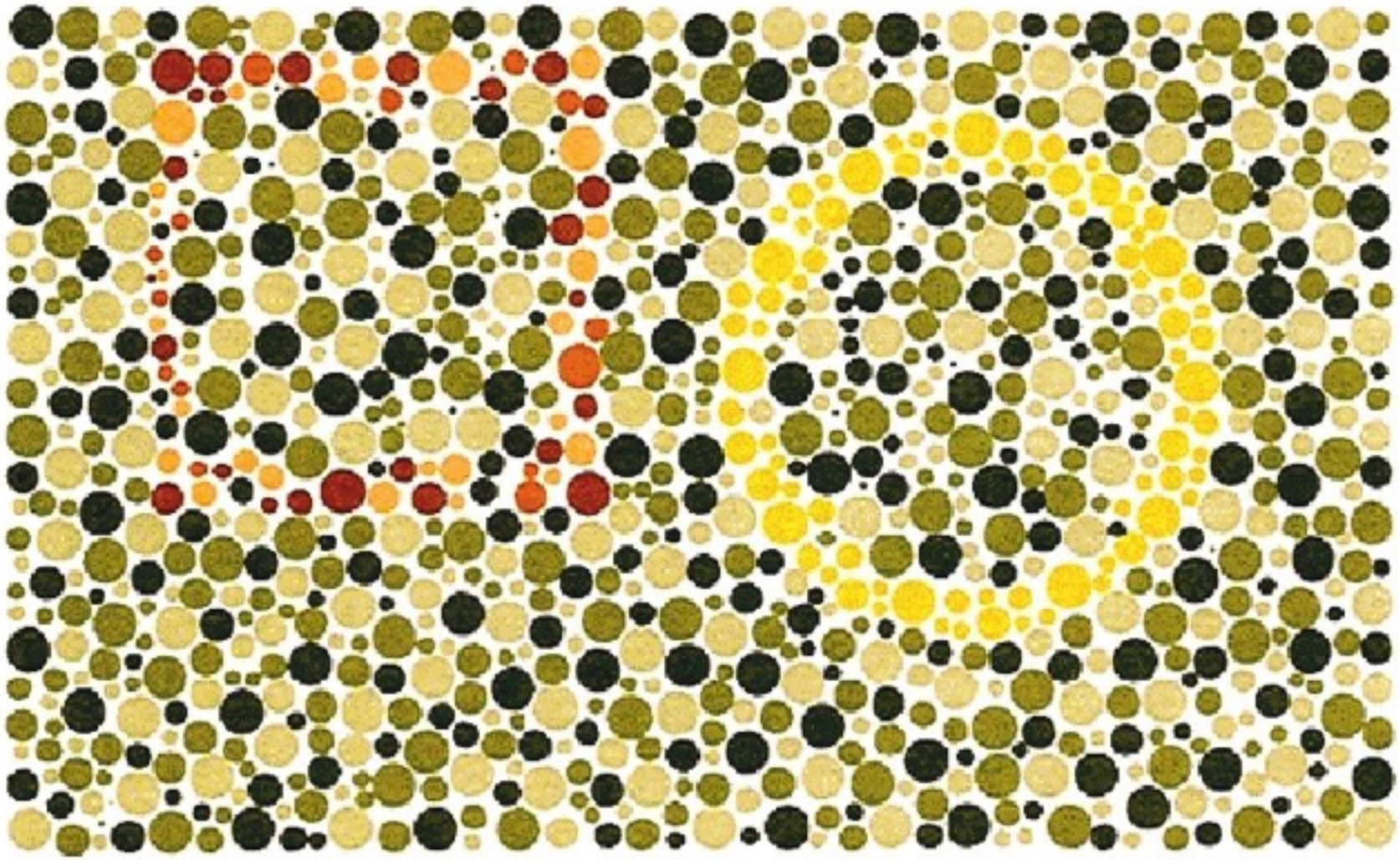
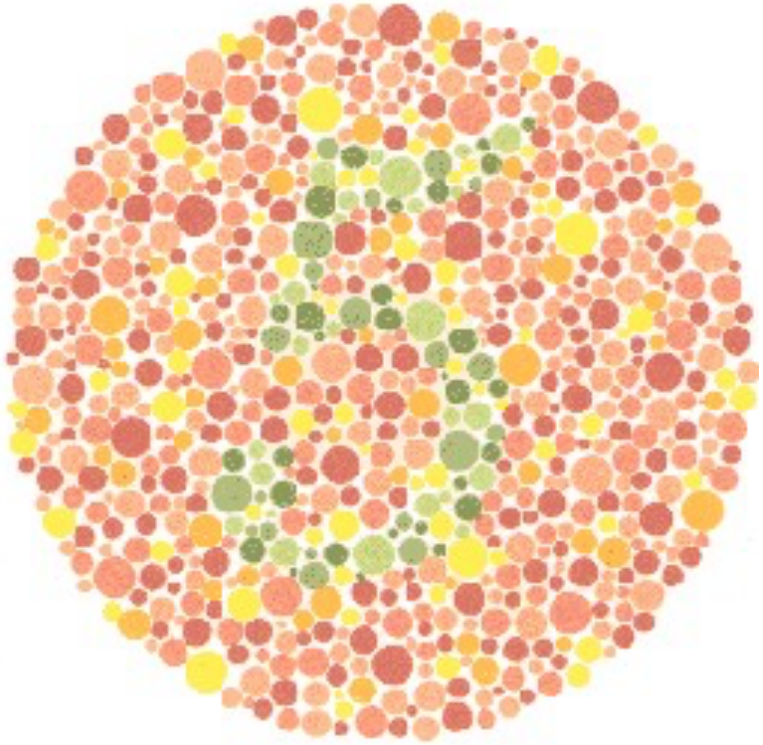
Red Green Colour blindness

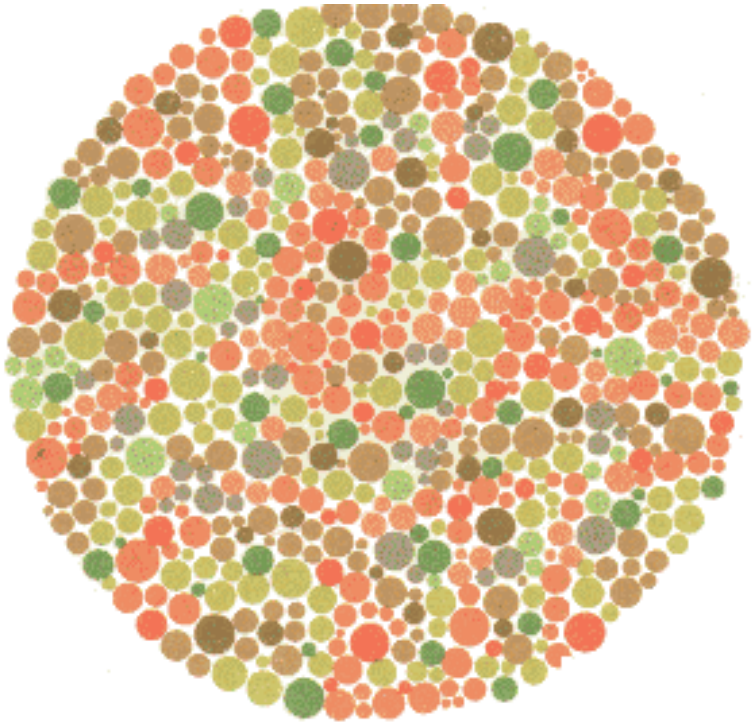




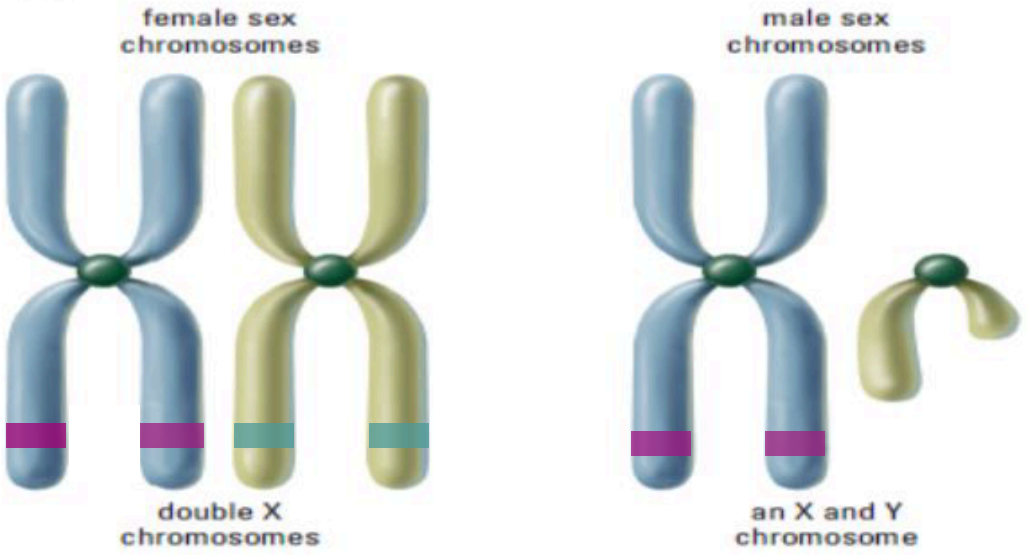
Ishihara colour blindness tests

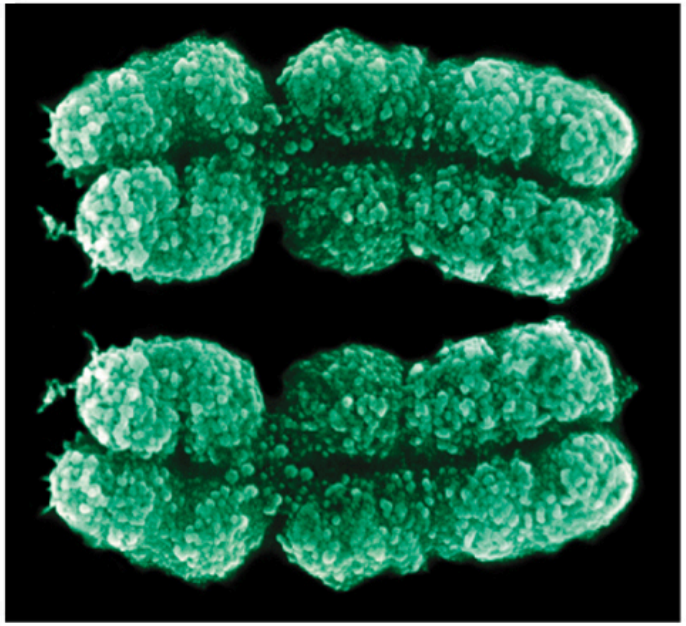






X-linkage

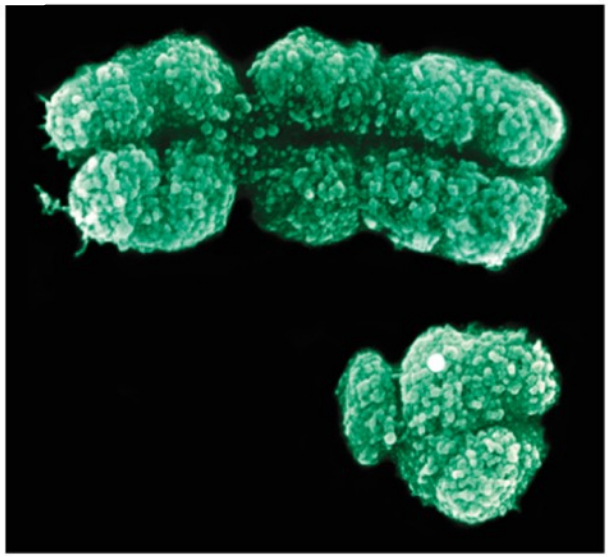

female sex chromosomes
double X chromosomes
male sex chromosomes
an X and Y chromosome



So while a females can carry an X- linked trait, if it is recessive- the other X chromosome would probably not, and it's expression would DOMINATE giving a WT phenotype.

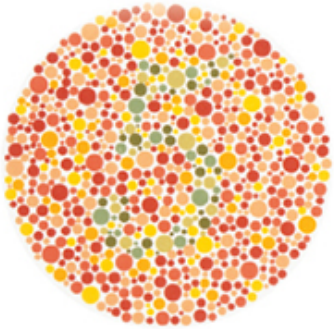
Hence Females can often be carriers of an X- linked trait, but rarely demonstrate the phenotype.

Giving rise to the following inheritable signs for **X- Linkage**



For Males it's a different story, if the X chromosome carries the trait… there is NO compensating X chromosome to help hide the trait, and if it is present it WILL ALWAYS SHOW THROUGH

People with red–green color blindness cannot see the number 5 in this figure.



Unaffected male
Unaffected female
Affected male

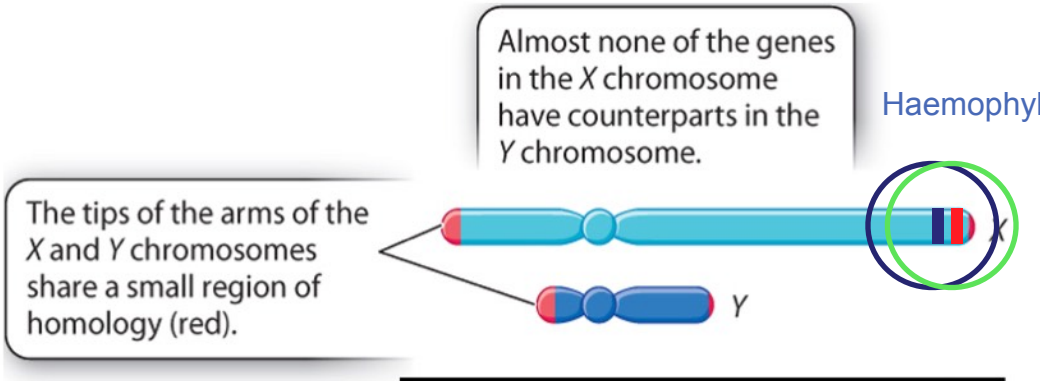
For a rare *X*-linked recessive trait, most affected individuals are male.

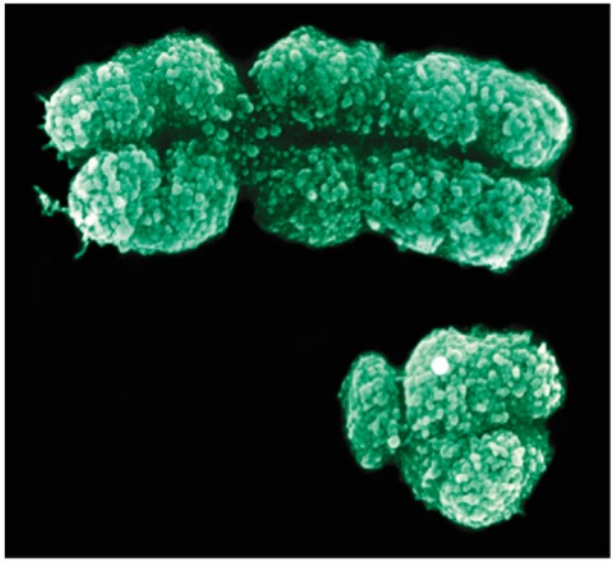
The daughters of affected males can have affected sons.

The offspring of an affected male are usually not affected.

The sisters of an affected male can have affected sons.

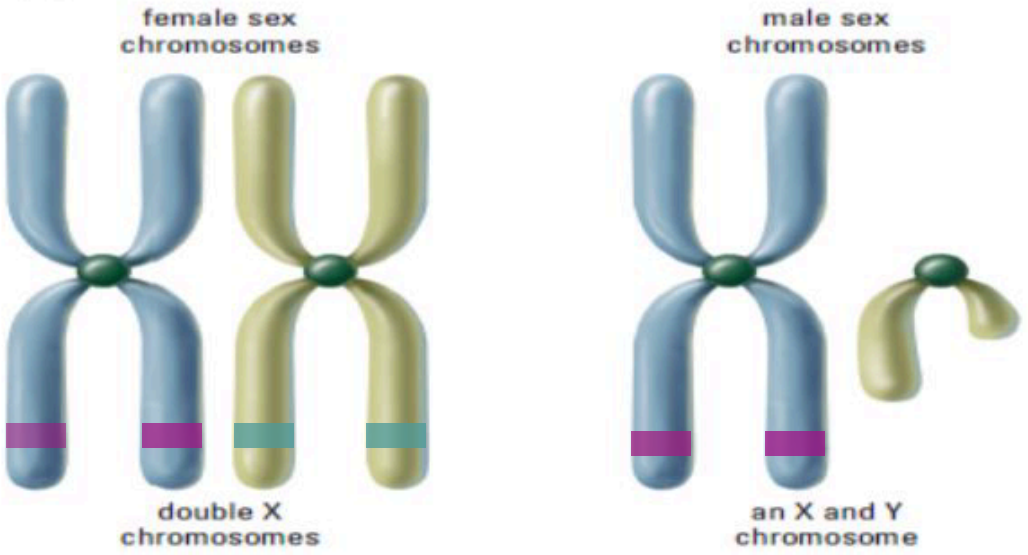
Haemophylia A (Factor VIII)

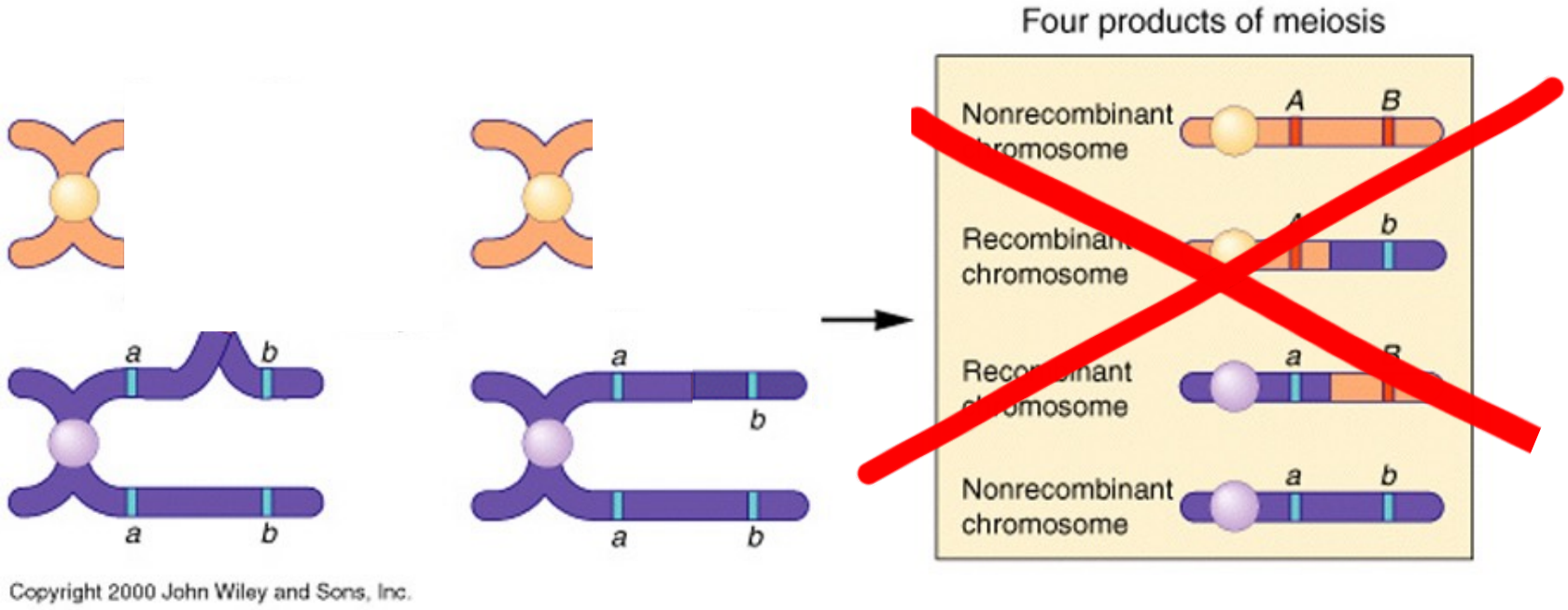




Non-Mendelian Inheritance

X-linkage




Four products of meiosis
Nonrecombinant chromosome
A
B
Recombinant chromosome
b
Recombinant chromosome
a
Nonrecombinant chromosome
a
b
a
b
a
b
a
b
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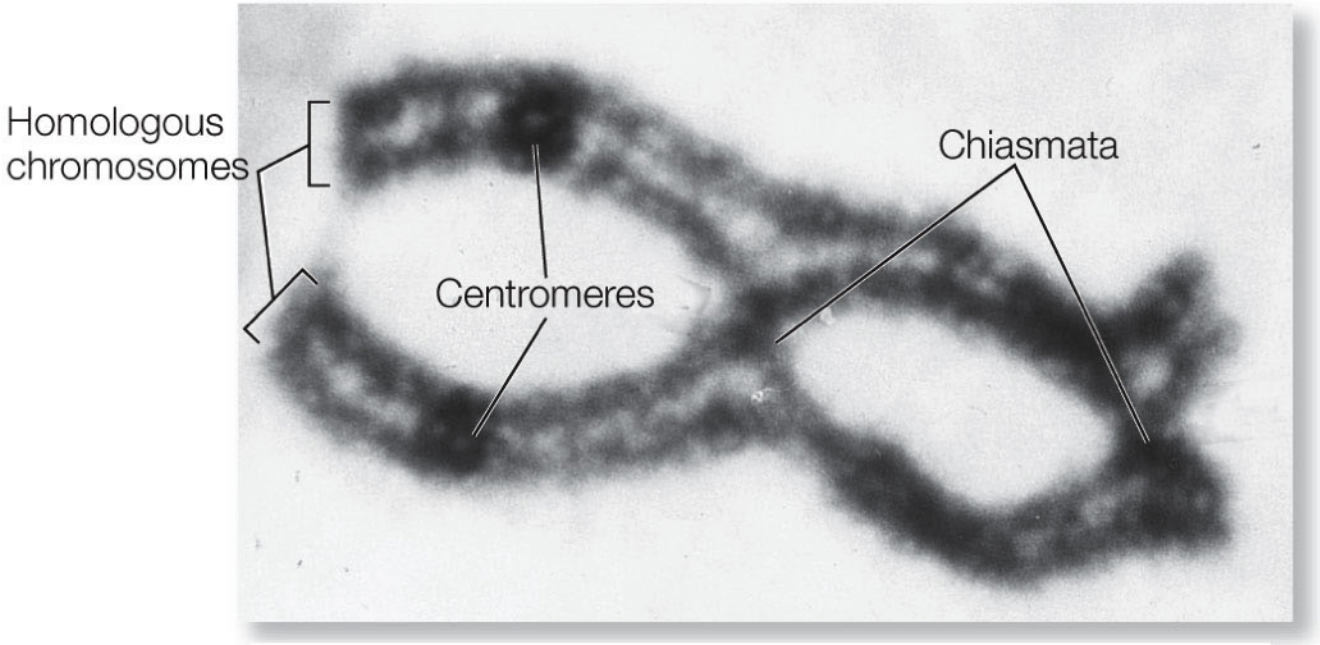
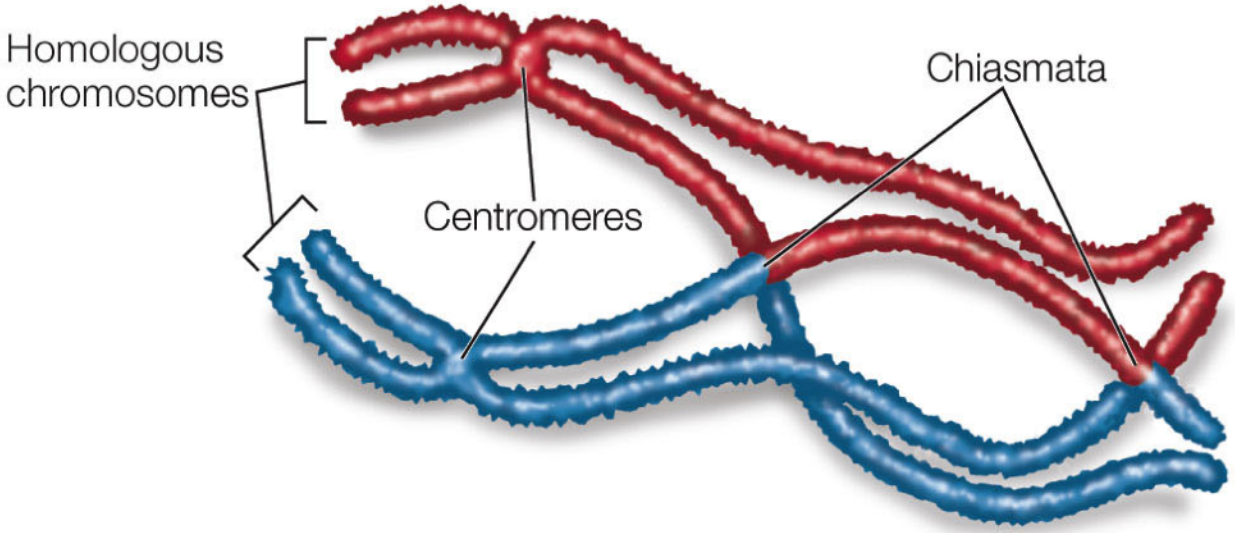

Homologous chromosomes
Chiasmata
Centromeres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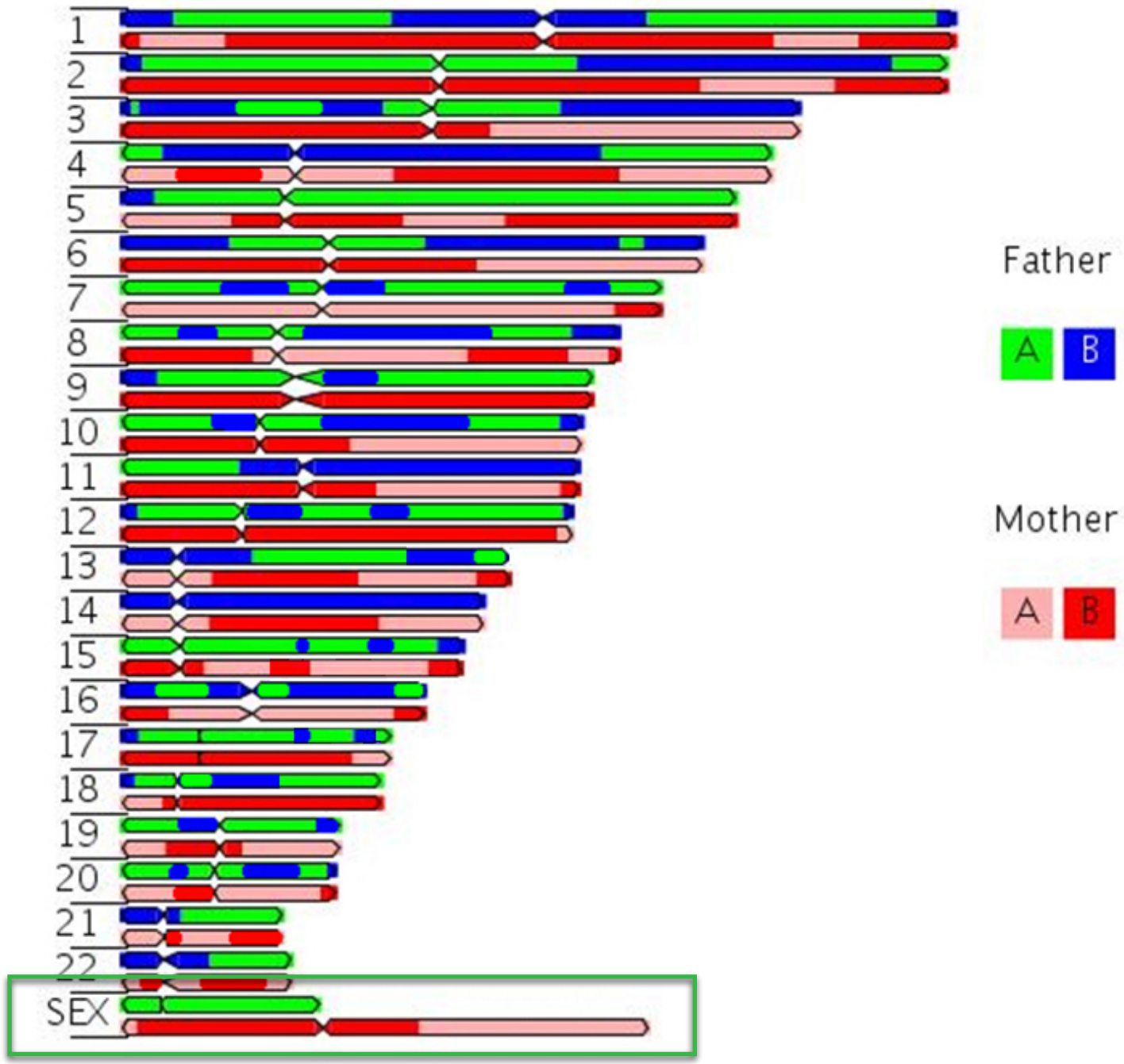
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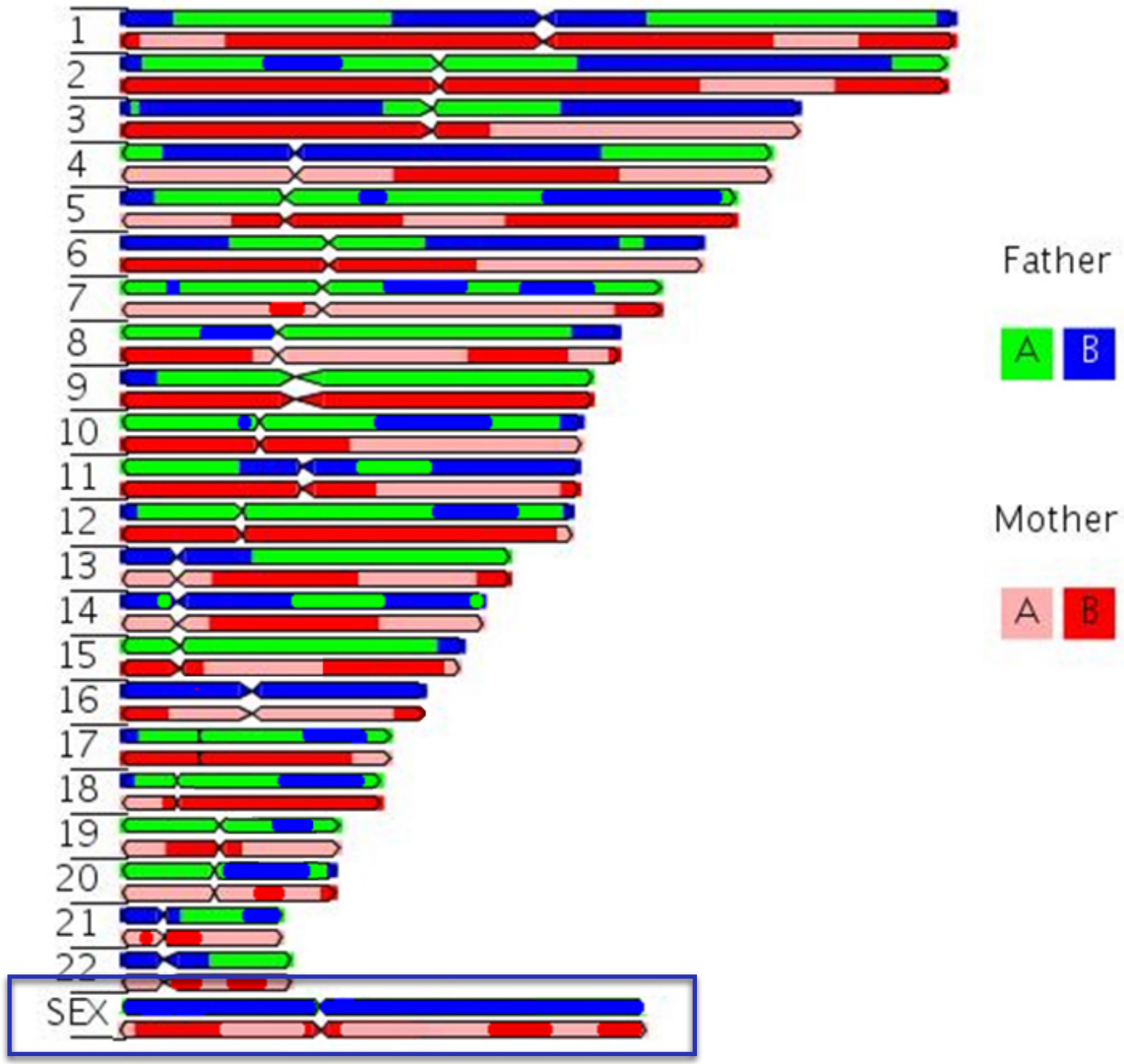
SEX
Father
A
B
Mother
A
B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22
SEX
Father
A
B
Mother
A
B