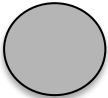
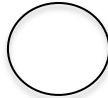


Summary slide: Understanding Pedigrees for a disease or trait



Affected female



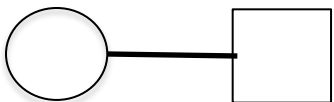
Female with normal phenotype



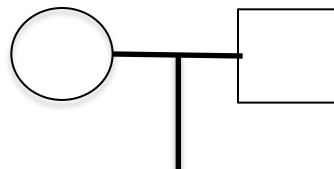
Affected male



Male with normal phenotype



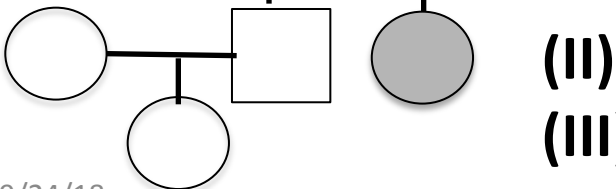
Horizontal line represents mating



Vertical line represents offspring for next generation



(I)

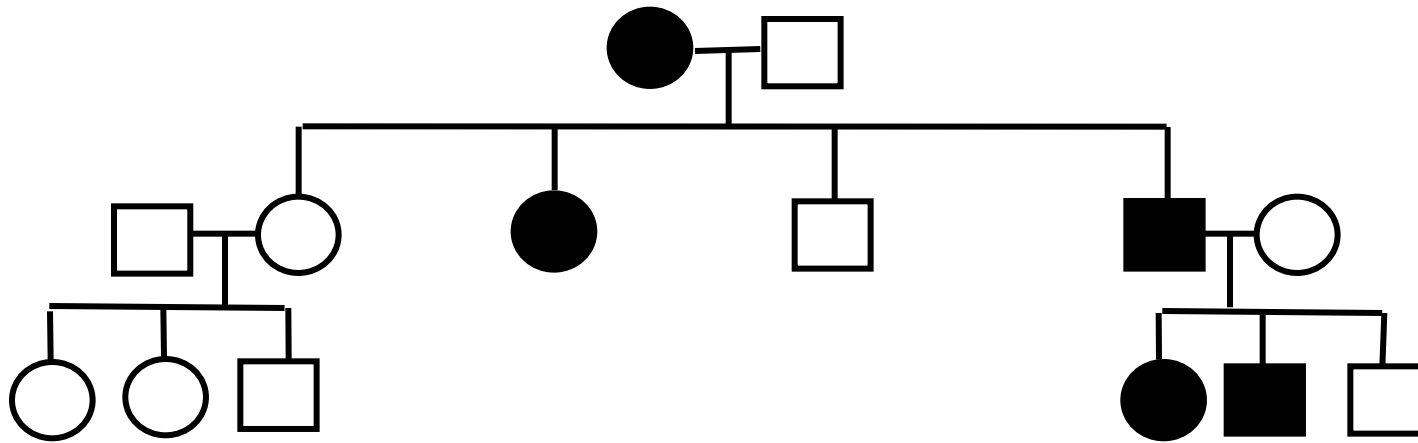


(II)

(III)

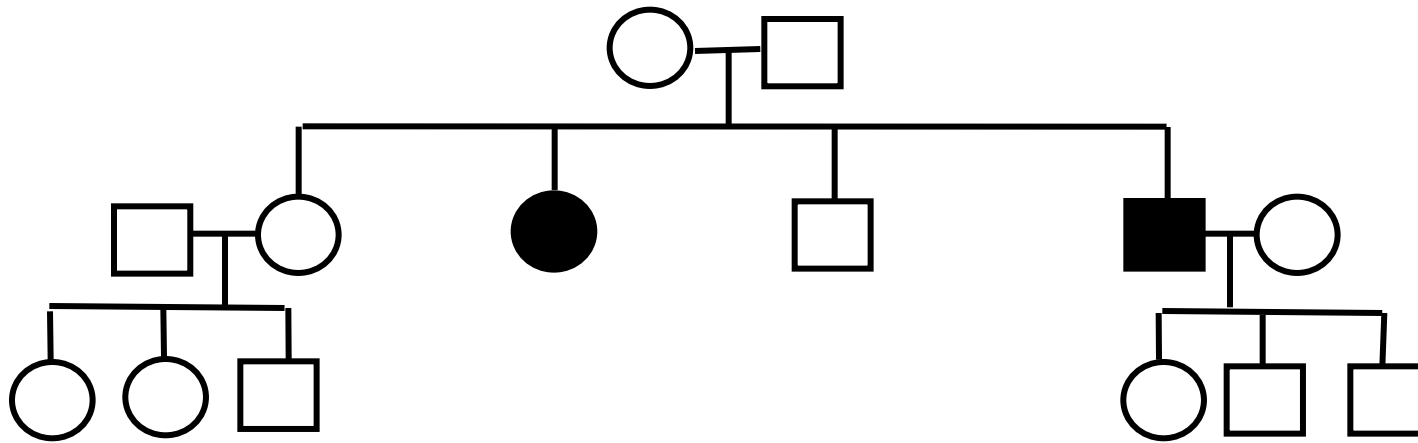
Pedigree of three generations for this disease

Example: Autosomal dominant inheritance due to mutation in Gene A



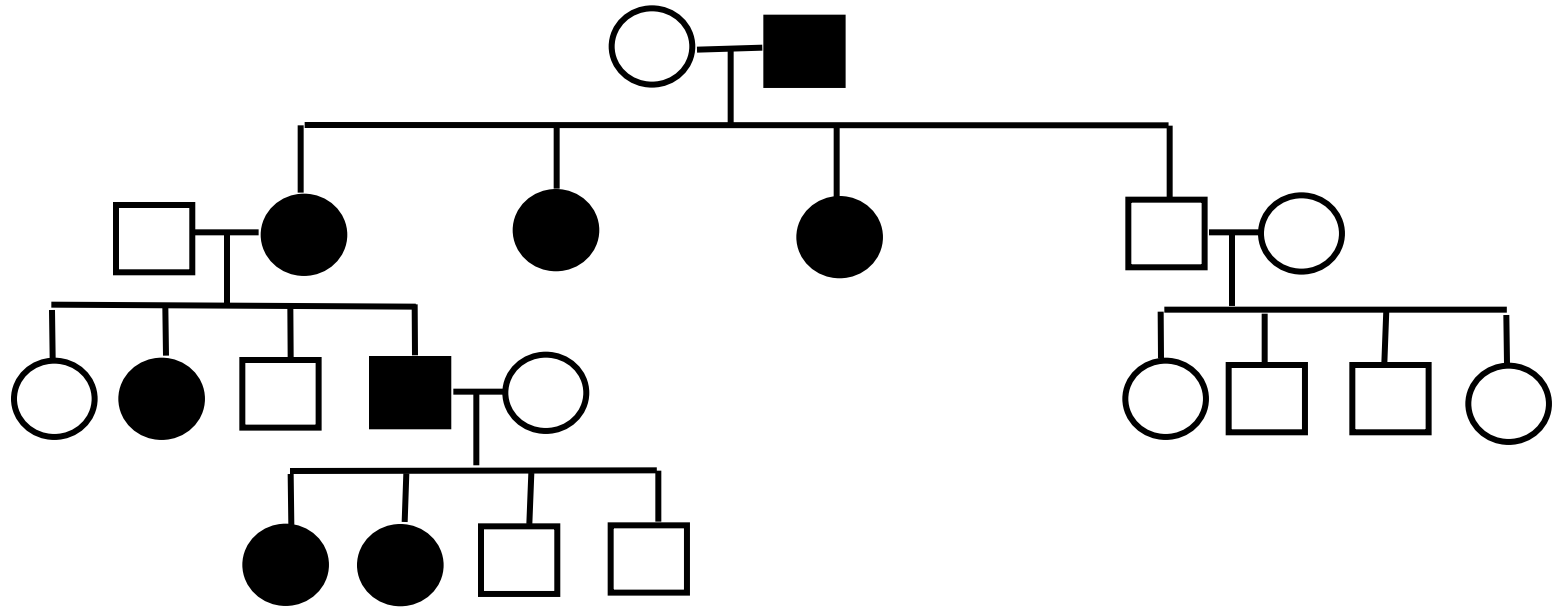
- Affected individuals: Aa or AA .
- Unaffected/ normal individual: aa
- Affected offspring has at least one affected parent
- Unaffected offspring of affected parent have unaffected offspring
- No gender associations

Example: Autosomal recessive inheritance due to mutation in Gene A



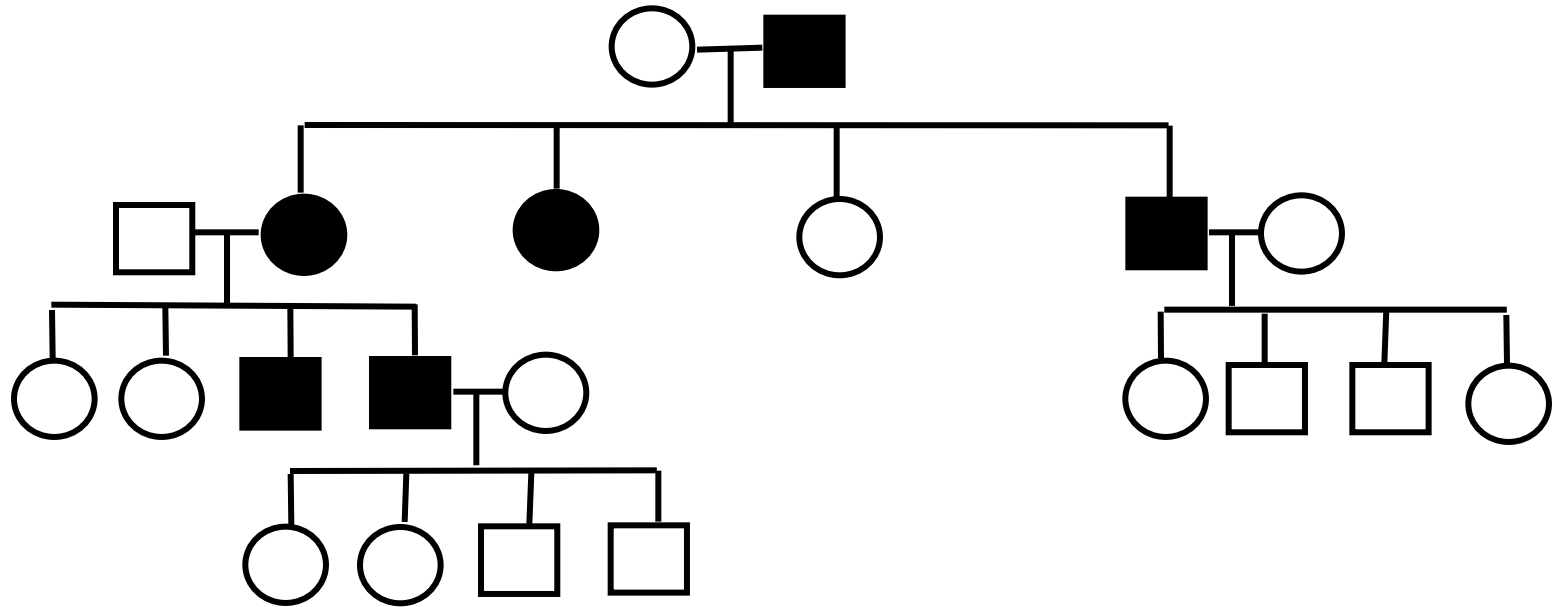
- Affected individuals: aa.
- Unaffected/ normal individual: AA or Aa
- Affected offspring gets the disease associated allele from both parents i.e. parents are at least heterozygous for the disease related allele.
- No gender associations

Example: X- linked dominant inheritance due to mutation in Gene A



- Affected males: $X^A Y$ and affected females: $X^A X^A$ or $X^A X^a$
- Normal male: $X^a Y$ & Normal female: $X^a X^a$
- Affected fathers transmit trait to all of daughters but not sons
- Affected mothers (if heterozygous) pass along trait to $\sim 1/2$ of their daughters and $1/2$ of their sons.

Example: X- linked recessive inheritance due to mutation in Gene A



- Affected males: X^aY and affected females: X^aX^a
- Normal male: X^AY & Normal female: X^AX^a or X^AX^A
- Affected mothers transmit trait to all sons.
- Heterozygous mothers pass along trait to $\sim 1/2$ of their sons.

Sample exercise: Pedigree showing the ability to roll tongue

Rollers/ non-rollers

→ Trait/ phenotype

Gene A

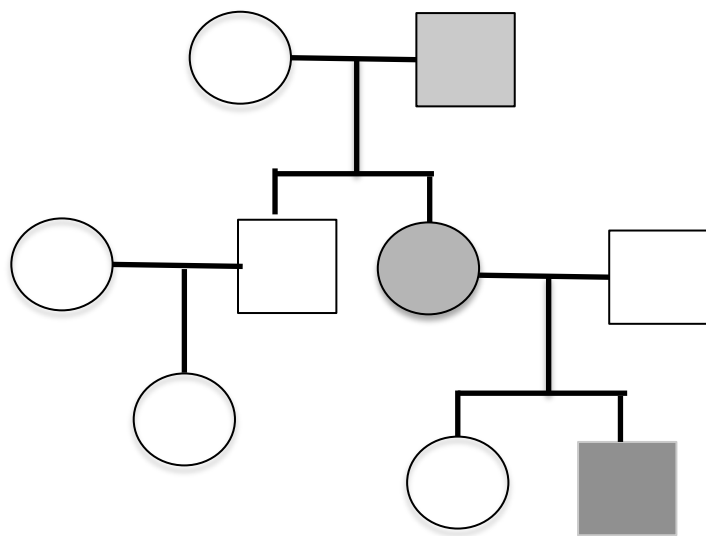
→ Gene associated with this trait

Allele “A or X^A” of Gene A

→ Regulates dominant trait (rollers)

Allele “a or X^a” of Gene A

→ Regulates recessive trait (non-rollers)

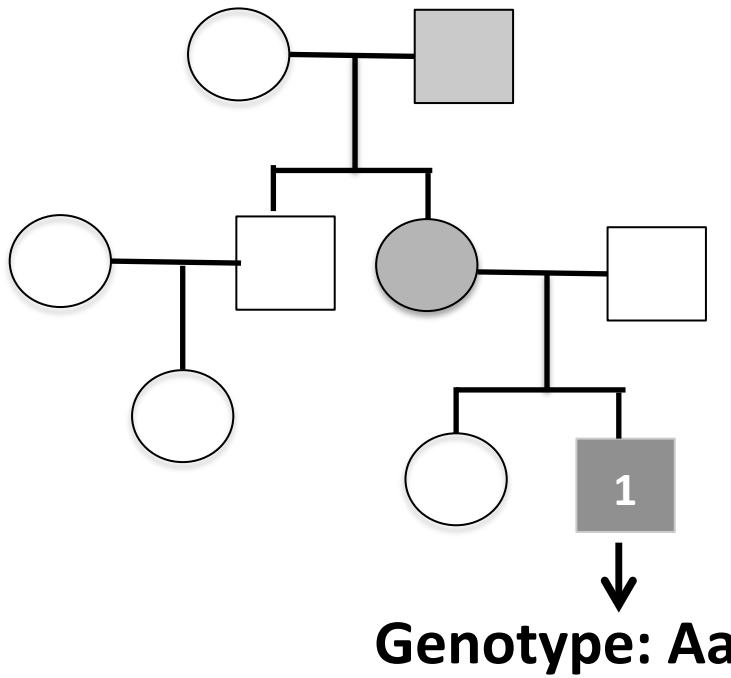


Pedigree dominant/ recessive?

Pedigree autosomal/ X-linked/
both?

*Note: Is there any gender bias?
People marrying in have normal
phenotype and genotype and
pedigree is completely penetrant.*

Exercise 3: Pedigree showing the ability to roll tongue



#1 has a son with a female(#2) who is a non-roller and has the genotype (aa).

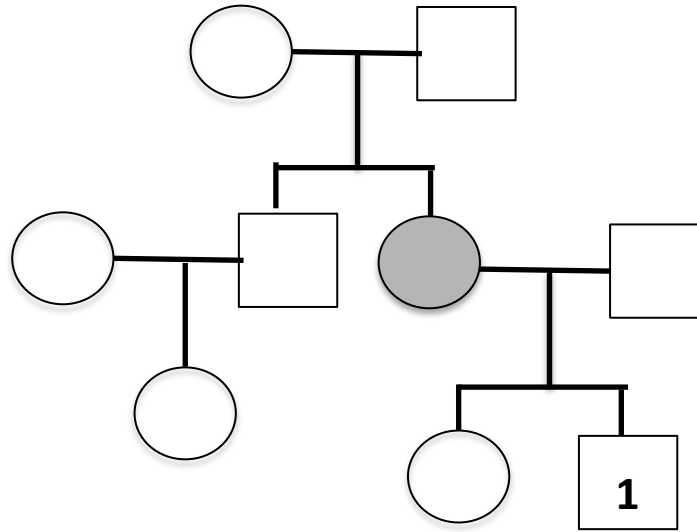
Do a Punnett square to show the possible genotypes of their son.

Gametes from #2	Gametes from #1	
	A	a
a	<u>Aa</u>	<u>aa</u>
a	<u>Aa</u>	<u>aa</u>

Can it be X- Linked dominant?
Yes

Possible genotypes of son Aa or aa

Exercise 4



1. Pedigree dominant/ recessive? *Recessive*

2. Pedigree autosomal/ X-linked? *Autosomal*

3. Determine the possible genotypes of #1. *Aa (assuming that the person marrying into the family has a normal genotype and phenotype)*

MIT
OpenCourseWare
<https://ocw.mit.edu/>

7.013 Introductory Biology
Spring 2018

For information about citing these materials or our Terms of Use, visit: <https://ocw.mit.edu/terms>.