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

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 ~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
Gene A
Allele “A or X^{A}” of Gene A
Allele “a or X^{a}” of Gene A

Trait/ phenotype
Gene associated with this trait
Regulates dominant trait (rollers)
Regulates recessive trait (non-rollers)

Pedigree \textbf{dominant}/ recessive?

Pedigree autosomal/ X-linked/ \textbf{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.

Can it be X-Linked dominant? **Yes**

### Possible genotypes of son Aa or aa
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)*