Inheritance

D3.2 · SL

Two similar parents can have a different-looking offspring

Explain transmission before predicting a phenotype

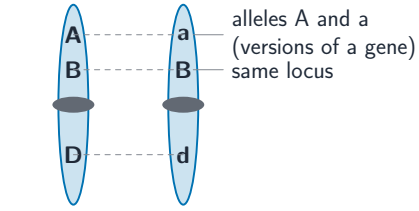
Shared D3.2: controlled crosses, alleles and phenotypes, dominance patterns, human examples, pedigrees, continuous variation and box plots.

Crosses, families and samples are invented teaching models. Probabilities do not prescribe an offspring sequence.

Track a locus through gametes and fertilization

1. **Diploid parent.** Two homologues normally carry two copies of an autosomal locus. AA and aa are homozygous; Aa is heterozygous.
2. **Gametes.** A normal gamete receives one member of each pair and one **allele** at each ordinary autosomal locus.
3. **Fertilization.** Two haploid contributions restore the diploid complement. Alleles are variants of one gene, not unrelated genes because letters differ.

Homologous chromosomes



a homologous pair
(same length, same genes)

One from each parent

Control the pollen source before interpreting offspring

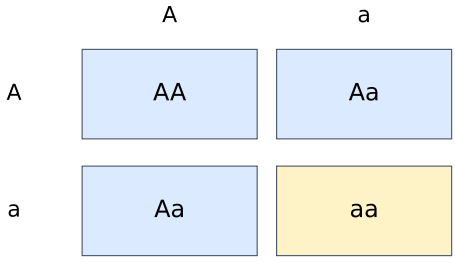
1. **Before release.** Where appropriate remove anthers before self-pollen release; protect a receptive stigma from unintended pollen.
2. **Cross.** Apply selected pollen, label parents and use bagging and suitable controls.
3. **Generations.** P parents produce F1; suitable F1 crosses or selfings produce F2. These names alone do not guarantee homozygous parents.

Separate the inherited information from a measured outcome

Example	**Interpretation**
Ordinary ABO type	Relatively simple genetic contribution in the model
Injury scar	Acquired environmental consequence
Height	Many genes and environment interact
Sun / shade leaves	One **genotype** can develop differently in different conditions
Plasticity	Expression or development changes without requiring a base-sequence change

Read four possible combinations, not four compulsory children

Aa × Aa: four equally likely combinations



Genotypes 1 : 2 : 1; phenotypes 3 : 1

AA and Aa share the dominant phenotype in this model.

An expectation is not a fixed family count

1. **Aa × Aa.** AA:Aa:aa = 1:2:1; dominant:recessive = 3:1 under complete dominance.
2. **Twenty offspring.** Expected aa count = $20 \times 1/4 = 5$; actual counts can differ.
3. **Two specified offspring.** Independent fertilizations: $(1/4) \times (1/4) = 1/16$ for both aa. A previous result does not force the next.

Use a test cross to compare possible parental genotypes

1. **AA × aa.** All Aa under the ordinary model.
2. **Aa × aa.** Half Aa and half aa; a recessive offspring supports a heterozygous parent.
3. **Limit.** A small all-dominant sample does not prove AA. Dominant does not mean common, healthy or stronger.

Connect an enzyme change with an environmental intervention

1. **Normal conversion.** Phenylalanine hydroxylase helps convert phenylalanine to tyrosine.
2. **Reduced activity.** Phenylalanine can accumulate and harm development; phenylalanine is also an essential amino acid.
3. **Inheritance and environment.** Carrier $\times$ carrier predicts $1/4$ affected per pregnancy in the recessive model. Dietary management can alter consequences without changing the **genotype**.

A population has several alleles; one individual has two

Type	**Ordinary-model genotypes**
A	$I^A I^A$ or $I^A i$
B	$I^B I^B$ or $I^B i$
AB	$I^A I^B$; both antigen features
O	ii
Dominance	I^A and I^B codominant; each dominates i

Compare two ABO crosses using the actual gametes

1. **Heterozygote A × heterozygote B.** $I^A i \times I^B i$ gives A, B, AB and O each with probability $1/4$.
2. **AB × O.** $I^A I^B \times ii$ gives half type A and half type B.
3. **Limit.** A population can carry more than two alleles at a locus. A SNP need not cause disease, and one diploid person normally has only two autosomal copies.

An intermediate phenotype does not permanently blend alleles

Model	**Heterozygote and offspring**
Mirabilis red / white	Heterozygote pink: incomplete dominance
Pink $\times$ pink	Red:pink:white $= 1{:}2{:}1$
Alleles	Segregate; red and white can reappear
Ordinary ABO AB	Distinguishable contributions of both alleles: codominance
Complete dominance	Heterozygote resembles one homozygote for the defined trait

State the conditioning group for a haemophilia probability

1. **Parents.** $X^H X^h \times X^H Y$ in the simplified X-linked recessive model.
2. **Four equally likely outcomes.** $X^H X^H$, $X^H X^h$, $X^H Y$, $X^h Y$.
3. **Denominator.** Affected sons: $1/2$ of sons, or $1/4$ of all offspring if X/Y-bearing fertilizations are equally likely.

A father transmits different chromosomes to sons and daughters

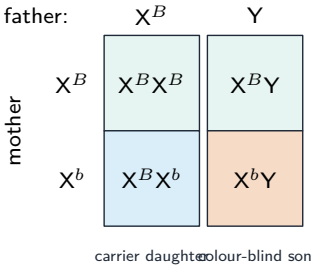
Do not turn the teaching grid into a universal claim about females

In the ordinary XX/XY model, father gives X to daughters and Y to sons; direct father-to-son transmission of his X-linked allele is absent.

X carries more genes than Y; a Y symbol does not supply an alternative haemophilia allele.

Females can have haemophilia and heterozygotes can have bleeding symptoms, including through X-inactivation. The simplified model has limits.

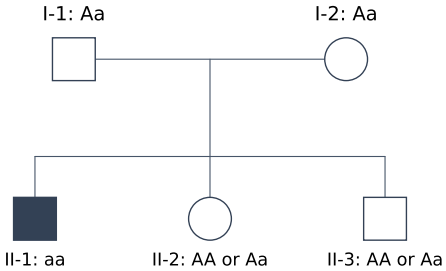
An X-linked cross



> Sons get their X from their mother

Name the relationship and symbol before inferring a genotype

Invented family: test an autosomal recessive model



Square: male; circle: female; filled: affected

Fully penetrant autosomal recessive model; no new mutation.

Condition on an unaffected sibling rather than all offspring

1. **Parents.** An aa child of two unaffected parents requires Aa $\times$ Aa under the stated model.
2. **Unaffected possibilities.** AA, Aa, Aa: two of three are carriers. Conditional probability $= 2/3$.
3. **Reasoning.** Model choice from family patterns is induction; required genotypes follow deductively once the model and assumptions are accepted.

A small pedigree can fit more than one biological mechanism

Keep the model separate from its conditional deductions

An affected child of unaffected parents contradicts a simple fully penetrant dominant model without new mutation; real families may involve new variants or incomplete penetrance.

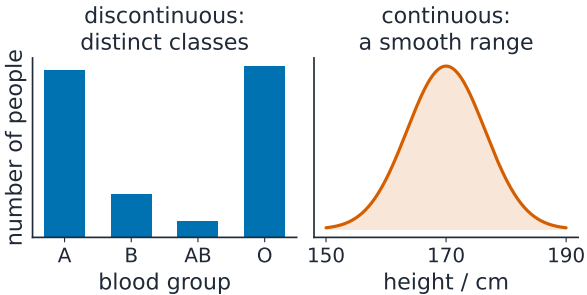
Missing affected sons in a small family does not prove X linkage. Male-to-male transmission argues against ordinary direct X linkage.

Close relatives may share a rare recessive allele from common ancestry. Increased homozygous risk does not mean every child must be affected.

Compare measures of centre for an invented height sample

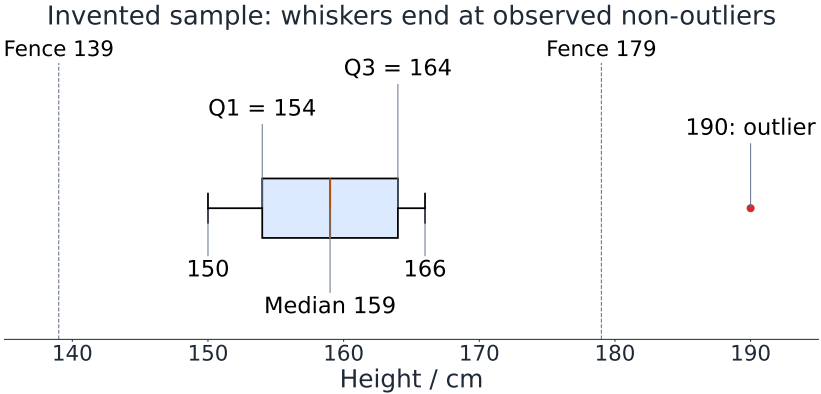
1. **Data / cm.** 150, 152, 154, 156, 158, 160, 162, 164, 166, 190.
2. **Calculate.** Sum 1612; mean 161.2 cm. Middle two are 158 and 160: median 159 cm. No mode.
3. **Interpret.** The high observation affects the mean more than the median. Rounding height does not change its underlying continuous nature.

Discontinuous and continuous variation



Many genes and the environment give continuous variation

Separate quartiles, fences and observed extremes



The maximum is 190 cm; the upper whisker is 166 cm.

Use the stated quartile convention and the actual observations

1. **Median of halves.** Q1 = 154, Q3 = 164 cm; IQR = 10 cm.
2. **Fences.** $154 - 15 = 139$; $164 + 15 = 179$ cm. The 190 cm observation lies beyond the upper fence.
3. **Whiskers.** Most extreme observed non-outliers: 150 and 166 cm. A fence is not automatically a whisker endpoint; retain the underlying data.

Attempt the handout before revealing the numerical checks

Start with parentage, model, gametes and phenotype key

Explain controlled plant crosses, phenotype plasticity, PKU, ABO, incomplete dominance, X linkage and conditional pedigree inference.

Calculate centres, quartiles, fences and observed whiskers. Compare continuous characteristics with discrete categories.

Check the shared probabilities and descriptive statistics

1. **Monohybrid / X linkage.** Expected aa among twenty $= 5$; two specified aa $= 1/16$. Affected X-linked probability: $1/2$ of sons, $1/4$ of all offspring.
2. **Pedigree.** Unaffected parents of aa child: Aa and Aa. Carrier probability conditional on unaffected $= 2/3$.
3. **Height.** Mean 161.2, median 159, no mode; quartiles 154/164, IQR 10; fences 139/179; whiskers 150/166; maximum 190 cm.

Check yourself

A probability needs a model and a denominator

Genotype is not phenotype; dominance is not allele frequency. An expected ratio does not force the next offspring outcome.

Identify the conditioning group in pedigrees and X-linked probabilities. Distinguish box-plot fences from observed whiskers.