MA99 EDUCATION

Heredity: Summary 05

NSW BIOLOGY STAGE 6 SYLLABUS 2017 FOR HSC 2027

In this topic: reproduction · DNA structure and replication · cell replication · DNA and polypeptide synthesis · genetic inheritance · genetic variation · genetic technologies

1. Reproduction

- Sexual reproduction: two parents, gametes fuse at fertilisation, offspring genetically varied.
- Asexual reproduction: one parent, mitosis only, offspring are clones.
- Asexual methods: binary fission, budding, vegetative propagation, spores, parthenogenesis. Sexual: external or internal fertilisation.
- Asexual is fast and gives clones. Sexual is slower and gives variation to cope with change.

2. DNA structure and replication

- A nucleotide is a phosphate, deoxyribose and a base (A, T, G or C). Strands run antiparallel, 5' to 3' against 3' to 5'.
- A pairs with T (two hydrogen bonds). G pairs with C (three). If A is 28%, T is 28%, G and C are 22% each.
- Replication: helicase unwinds, primase lays a primer, DNA polymerase builds 5' to 3', ligase joins fragments. Leading strand continuous, lagging strand in Okazaki fragments.
- Meselson and Stahl (1958): after one generation in light nitrogen all DNA was hybrid, which supports semi-conservative replication.

3. Cell replication

- DNA replication happens once, during interphase, before division, and is semi-conservative: each new helix keeps one original strand.
- Mitosis: one division, two identical diploid daughter cells. Growth, repair, asexual reproduction.
- Meiosis: two divisions, four genetically varied haploid daughter cells. Gamete production only.
- Cell cycle: G1, S (DNA replicated), G2, then M (mitosis and cytokinesis).
- Mitosis stages: prophase, metaphase, anaphase (chromatids separate), telophase.
- Meiosis I separates homologous pairs (crossing over in prophase I, random orientation at metaphase I). Meiosis II separates chromatids.
- Human cell, 2n = 46: 46 chromosomes and 92 DNA molecules after S phase. Each gamete has 23 chromosomes and 23
- DNA molecules.

FEATURE	MITOSIS	MEIOSIS
Divisions	One	Two
Daughter cells	Two	Four
Chromosome number	2n to 2n	2n to n
Genetically identical?	Yes	No

4. DNA and polypeptide synthesis

- Transcription: RNA polymerase copies one gene into mRNA, in the nucleus.
- mRNA processing: a cap and tail are added, introns are spliced out.
- Translation: a ribosome reads mRNA in codons of three bases, tRNA delivers the matching amino acid, the chain is released as a polypeptide.
- The genetic code is a triplet code, degenerate, with three stop codons.
- Transcription uses the template strand, so the mRNA matches the coding strand with U for T.
- Mutation types: silent (same amino acid), missense (different amino acid), nonsense (stop codon), frameshift (insert or delete one or more bases, in a number that is not a multiple of three).
- Sickle cell disease: HBB GAG becomes GTG, so Glu becomes Val.
- Epigenetic marks (DNA methylation, histone changes) change expression without changing the sequence.
- Gene expression is regulated: not every gene is transcribed in every cell.

5. Genetic inheritance

- Monohybrid cross, one gene, two alleles: a Tt by tt cross gives a 1 to 1 ratio of Bindarra thumb to normal thumb.
- Dihybrid cross, two independently assorting genes: a TtSs by TtSs cross gives the 9 to 3 to 3 to 1 ratio.
- A pedigree that skips a generation, with two unaffected parents producing an affected child, points to autosomal recessive inheritance.
- Pedigree probability: among the unaffected children of an Ss by Ss cross, the SS to Ss ratio is 1 to 2, so an unaffected child has a 2 in 3 chance of being a carrier, not 1 in 2.
- Codominance: both alleles fully expressed, for example ABO blood type IA and IB together giving type AB.
- Incomplete dominance: the heterozygote is a blended, intermediate phenotype, for example red by white strawflowers giving pink.
- Multiple alleles: more than two alleles exist in the population for one gene, for example the three ABO alleles IA, IB and i.
- Sex linkage: a gene carried on the X chromosome. Males (XY) cannot be heterozygous for it, so X-linked recessive traits appear more often in sons.

Key idea

Codominance shows both phenotypes side by side. Incomplete dominance blends them into a third. Keep the two separate in an answer.

- Test cross: unknown dominant phenotype with tt. Any tt offspring means the parent is Tt.
- Probabilities: multiply for “and”, add for “or”. Each child is independent.
- X linked cross $X^{C}X^{c}$ by $X^{C}Y$: half the sons affected, no daughters affected.
- Cystic fibrosis: autosomal recessive, about 1 in 25 Australians carry the gene (Cystic Fibrosis Australia).

PATTERN	PHENOTYPE CLUE	EXAMPLE FROM THIS MODULE
Autosomal dominant	About half of offspring affected, an affected parent in every affected line	Bindarra thumb
Autosomal recessive	Can skip a generation, both parents unaffected carriers	Spot syndrome
Codominant	Heterozygote shows both phenotypes at once	ABO blood type AB
Incomplete dominant	Heterozygote shows a blended phenotype	Pink strawflowers
X-linked recessive	Appears far more often in sons than daughters	Working dog coat colour

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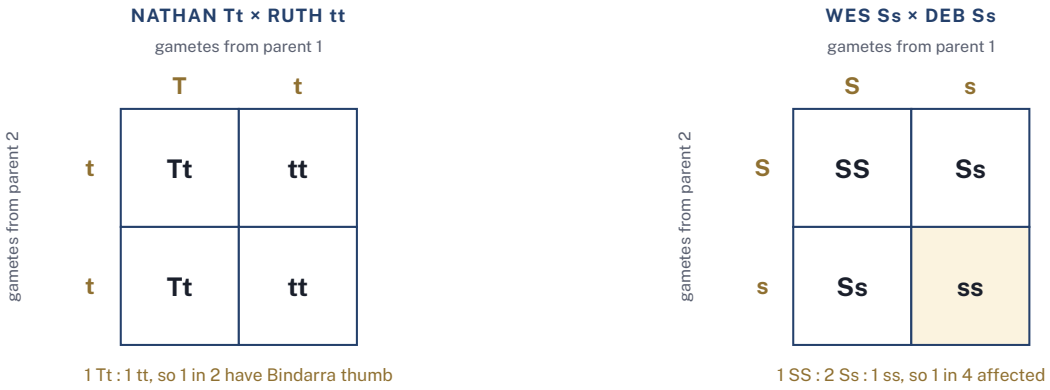


Figure 1 · Both Colvin crosses as Punnett squares. Rebuild these rather than memorising the ratios.

6. Genetic variation

- Mutation is the only source that creates new alleles.
- Crossing over, in meiosis I, swaps segments between homologous chromosomes.
- Independent assortment, in meiosis I, means each homologous pair separates independently of every other pair.
- Random fertilisation multiplies variation further.
- Phenotype is genotype and environment together: the same genotype can look different in different conditions.
- A human can make 2^{23} = 8,388,608 kinds of gamete by assortment. The exponent is the number of homologous pairs.

7. Genetic technologies

- PCR: denature (about 95 °C), anneal (about 55 °C), extend (about 72 °C). Copies = 2^n.
- Gel electrophoresis: DNA is negative, moves toward the positive end, small fragments travel furthest.
- Recombinant DNA: restriction enzyme, plasmid, ligase. CRISPR-Cas9: guide RNA leads Cas9 to a target, which it cuts.
- Cloning by nuclear transfer: Dolly, born 5 July 1996 (Wilmut et al., 1997).
- Ethics: who benefits, who is harmed, who consents, what is long term.

> If you can only hold three things from this whole module in your head walking into the exam, make them the mitosis-meiosis table, the dihybrid ratio, and the difference between codominance and incomplete dominance.

About this resource

MA99 EDUCATION

Scope

NSW Biology, Year 12. Written to the NSW Biology Stage 6 Syllabus (2017), Module 5, for students sitting the HSC in 2027. A new Biology syllabus is first examined in HSC 2028. Every worked context uses a hypothetical family (the Colvin family) and hypothetical traits for genetics problems, as HSC genetics questions do, with any other practice data labelled hypothetical, and all calculations have been checked.

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MA99 EDUCATION

Heredity: Practice Questions 05

NSW BIOLOGY STAGE 6 SYLLABUS 2017 FOR HSC 2027

1. Instructions

Instructions

70 marks in about 125 minutes, which is the HSC rate of just under two minutes a mark. 10 multiple choice items, then 15 short answer items with the marks shown. Work it in one sitting with no notes, then mark it against the guide at the back before you read the model answers as explanations. Every model answer shows where each mark sits, so a lost mark tells you exactly which sentence was missing.

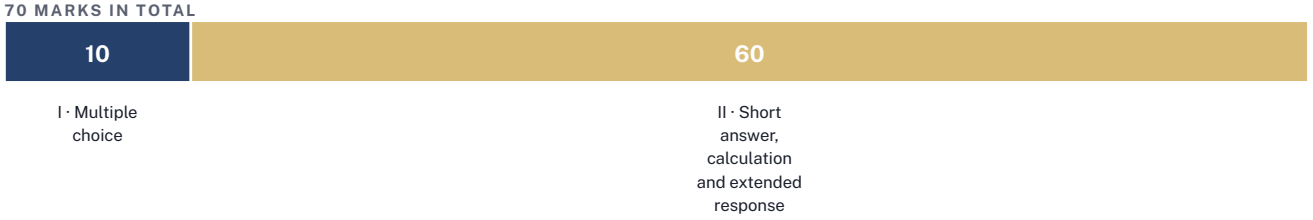


Figure 1 · How the 70 marks are split across the sections. Your time should follow the same split.

2. Section I · Multiple choice

Ten items, one mark each. Choose the best answer for each question.

Question 1

1 MARK

Which structure separates during meiosis I, but not during mitosis?

A Sister chromatids
B Homologous chromosomes
C Centromeres
D Ribosomes

Question 2

1 MARK

Which statement correctly describes asexual reproduction?

A It produces offspring genetically identical to the single parent.
B It always involves the fusion of two gametes.
C It produces four haploid daughter cells from one parent cell.
D It requires two parents of opposite sex.

Question 3

1 MARK

In transcription, which enzyme builds the mRNA strand?

A DNA polymerase
B RNA polymerase
C Helicase
D Ligase

Question 4

1 MARK

How many mRNA bases make up one codon?

A One
B Two
C Three
D Four

Question 5

1 MARK

Nathan Colvin has Bindarra thumb, genotype Tt. Ruth Colvin has a normal thumb, genotype tt. What is the probability that a given child of theirs has Bindarra thumb?

A 1 in 4
B 1 in 2
C 3 in 4
D 1 in 1

Question 6

1 MARK

Which best explains why Wes Colvin, blood type AB, is genuine evidence of codominance?

A His blood type is the most common in the population.
B Both his IA and IB alleles are fully expressed at once, neither masking the other.
C His parents were both blood type AB.
D The i allele is dominant over IA and IB.

Question 7

1 MARK

A cross between red and white strawflowers produces only pink offspring. Which pattern of inheritance does this show?

A Codominance
B Incomplete dominance
C Sex linkage
D Multiple alleles

Question 8

1 MARK

In the Colvin family's working dogs (a hypothetical MA99 family), coat colour is carried on the X chromosome. Which observation best fits X-linked recessive inheritance?

A The trait appears equally in sons and daughters.
B An affected daughter always has an unaffected father.
C The trait appears far more often in sons, whose mother is an unaffected carrier.
D The trait cannot be passed from a mother to her son.

Question 9

1 MARK

Which of the following is a source of genetic variation produced during meiosis itself?

A Mitosis in somatic cells
B Crossing over between homologous chromosomes
C DNA replication in interphase
D Translation of mRNA

Question 10

1 MARK

A gene has three alleles present in a population, and two of them are codominant with each other. Which gene, covered in this module, fits this description?

A The Bindarra thumb gene
B The spot syndrome gene
C The ABO blood type gene
D The working dog coat colour gene

3. Section II · Short answer, calculation and extended response

Fifteen items, marks as shown. Questions 18 to 25 cover DNA structure, replication, cell division, translation, mutation, sex linkage and technology. Show your working for any calculation.

Question 11

2 MARKS

State two differences between the daughter cells produced by mitosis and the daughter cells produced by meiosis.

Question 12

3 MARKS

Outline the process of transcription, including where in the cell it occurs.

Question 13

4 MARKS

Nathan and Ruth Colvin's daughter Ivy has Bindarra thumb (genotype Tt). Suppose Ivy has children with a partner who also has Bindarra thumb and is heterozygous, Tt. Complete a Punnett square for this cross and state the expected genotype ratio and phenotype ratio among their children.

Question 14

5 MARKS

Wes and Deb Colvin are both unaffected by spot syndrome but are known carriers, genotype Ss, because their daughter Priya has the condition (ss). Using a Punnett square, calculate the probability that their unaffected son, Callum, is a carrier rather than homozygous unaffected. Show your working.

Question 15

4 MARKS

Wes Colvin is blood type AB, genotype IAIB. Suppose Wes has a child with a partner who is blood type O, genotype ii. Using a Punnett square, state the possible blood types of their offspring and the probability of each.

Question 16

6 MARKS

Compare codominance and incomplete dominance. Use one example from the Colvin family or their property for each.

Question 17

7 MARKS

Explain how meiosis generates genetic variation, referring to crossing over, independent assortment and random fertilisation. Explain why this variation matters for a farming family breeding working dogs or livestock over several generations.

Question 18

3 MARKS

A short stretch of a hypothetical gene has the coding strand 5'-ATGAAGTTTGGCTAA-3'. Write the template strand, and the mRNA made from it, and state the one base difference between the mRNA and the coding strand.

Question 19

3 MARKS

Describe the roles of helicase and DNA polymerase in DNA replication, and explain why the process is called semi-conservative.

Question 20

3 MARKS

A human body cell (2n = 46) is about to divide. State the number of chromosomes in each daughter cell after mitosis, the number in each gamete after meiosis, and the number of chromatids in the cell at metaphase of mitosis.

Question 21

4 MARKS

The mRNA from Question 18 is 5'-AUGAAGUUUGGCUAA-3'. Use the codons in Figure 2 to give the amino acid sequence it codes for. A substitution changes the second codon from AAG to AAA. A different change inserts one extra A after the first codon. State the effect of each change and name each type of mutation.

CODON	AUG	AAG	AAA	UUU	GGC	GUU	UGG	CUA	UAA
AMINO ACID	Met	Lys	Lys	Phe	Gly	Val	Trp	Leu	stop

Figure 2 · Codons needed for this question (mRNA codons, read 5' to 3').

Question 22

4 MARKS

In a hypothetical (MA99) X linked recessive trait, a carrier mother ($X^{C}X^{c}$) and an unaffected father ($X^{C}Y$) have a child. Complete a Punnett square and state the probability that a son is affected, that a daughter is affected, and that a child chosen at random is affected.

> The Punnett square is not optional working. An answer that states the probability with no square shown is an answer the marker cannot verify, even if the number is right.

Question 23

3 MARKS

A farm dog shows the dominant trait, but its genotype could be TT or Tt. Describe a test cross to find out which, and state what results from each possible genotype would show. Eight dominant offspring are produced from a Tt parent. Calculate the probability of that happening.

Question 24

3 MARKS

A PCR run starts with one copy of a target sequence and runs 30 cycles, doubling the copies each cycle. Calculate the number of copies, and explain why a heat tolerant DNA polymerase is used.

Question 25 · Module 6 content

6 MARKS

Assess the use of CRISPR gene editing to treat a human genetic disorder. In your answer, describe how the technique edits a gene and discuss one benefit and one ethical concern.

4. Solutions

Section I answers, then model answers and mark allocations for Section II.

Section I · Answers

Q	ANSWER	Q	ANSWER
1	(B) Homologous chromosomes	6	(B) Both alleles fully expressed
2	(A) Genetically identical offspring	7	(B) Incomplete dominance
3	(B) RNA polymerase	8	(C) More common in sons, carrier mother
4	(C) Three	9	(B) Crossing over
5	(B) 1 in 2	10	(C) ABO blood type gene

Section II · Model answers and mark allocations

QUESTION 11 · 2 MARKS

- 1 mark: states a correct difference, for example mitosis gives two daughter cells and meiosis gives four.
- 2 marks: a second correct difference, for example mitosis daughter cells are diploid and genetically identical, meiosis daughter cells are haploid and genetically varied.

QUESTION 12 · 3 MARKS

- 1 mark: identifies transcription happens in the nucleus.
- 1 mark: RNA polymerase unwinds the DNA and builds a complementary mRNA strand from one gene.
- 1 mark: the mRNA is then processed, capped and tailed, before leaving the nucleus for translation.

QUESTION 13 · 4 MARKS

Model working. Tt × Tt gives gametes T and t from each parent. The Punnett square gives genotypes TT, Tt, Tt, tt, a ratio of 1 TT to 2 Tt to 1 tt. Since T is dominant, the phenotype ratio is 3 Bindarra thumb (TT and Tt) to 1 normal thumb (tt).

- 1 mark: correct gametes identified for each parent (T and t).
- 1 mark: a correctly completed Punnett square, or equivalent working, showing all four genotype combinations.
- 1 mark: correct genotype ratio, 1 TT to 2 Tt to 1 tt.
- 1 mark: correct phenotype ratio, 3 Bindarra thumb to 1 normal thumb.

QUESTION 14 · 5 MARKS

Model working. Ss × Ss gives gametes S and s from each parent. The Punnett square gives genotypes SS, Ss, Ss, ss, a ratio of 1 SS to 2 Ss to 1 ss. Callum is known to be unaffected, so his genotype is SS or Ss, not ss. Removing the affected ss class from the 1 to 2 to 1 ratio leaves the unaffected classes in the ratio 1 SS to 2 Ss, out of 3 unaffected individuals. The probability that Callum is a carrier (Ss) is therefore 2 in 3.

- 1 mark: correct gametes identified for each parent (S and s).
- 1 mark: a correctly completed Punnett square showing all four genotype combinations.
- 1 mark: recognises Callum's genotype must be SS or Ss, since he is unaffected, and ss is excluded.
- 1 mark: correctly restricts the sample space to the three unaffected outcomes, 1 SS to 2 Ss.
- 1 mark: correct final probability, 2 in 3, not 1 in 2.

QUESTION 15 · 4 MARKS

Model working. An IAIB by ii cross gives gametes IA and IB from the AB parent and i from the O parent. The Punnett square gives genotypes IAi and IBi, each with probability 1 in 2. Offspring blood types are therefore type A (IAi) with probability 1 in 2, and type B (IBi) with probability 1 in 2. No AB or O offspring are possible from this cross.

- 1 mark: correct gametes identified (IA and IB from one parent, i from the other).
- 1 mark: a correctly completed Punnett square.
- 1 mark: correctly states type A and type B are the only possible offspring blood types.
- 1 mark: correct probability for each, 1 in 2.

QUESTION 16 · 6 MARKS

- 1 to 2 marks: correctly defines codominance, both alleles fully expressed in the heterozygote.
- 1 to 2 marks: correctly defines incomplete dominance, the heterozygote shows a blended, intermediate phenotype.
- 1 mark: a correct family example for codominance, for example Wes Colvin's type AB blood, showing both IA and IB expressed.
- 1 mark: a correct family example for incomplete dominance, for example pink strawflowers from a red by white cross.

What separates a Band 6 answer

State the mechanism, not just the label. Codominance keeps both phenotypes visible side by side. Incomplete dominance produces a third, blended phenotype. A response that gives only the two words with no mechanism caps at half marks.

QUESTION 17 · 7 MARKS

- 1 to 2 marks: explains crossing over, exchange of DNA segments between homologous chromosomes in meiosis I, producing chromosomes with new allele combinations.
- 1 to 2 marks: explains independent assortment, each homologous pair separates independently of every other pair, so different combinations of maternal and paternal chromosomes end up in each gamete.
- 1 mark: explains random fertilisation multiplies the possible offspring genotypes further, since which gamete meets which is not predictable.
- 1 to 2 marks: links this to the farming context, for example a breeding population with more genetic variation has a better chance that some individuals carry combinations of alleles suited to a changing environment or a desired trait, compared with a population reproducing only asexually.

QUESTION 18 · 3 MARKS

Model working. Coding strand 5'-ATGAAGTTTGGCTAA-3'. The template strand pairs A with T and G with C, so it reads 3'-TACTTCAAACCGATT-5'. RNA polymerase reads the template and builds mRNA that matches the coding strand with U in place of T: 5'-AUGAAGUUUGGCUAA-3'.

- 1 mark: template strand correct, 3'-TACTTCAAACCGATT-5'.
- 1 mark: mRNA correct, 5'-AUGAAGUUUGGCUAA-3'.
- 1 mark: states that mRNA has uracil (U) where the coding strand has thymine (T).

QUESTION 19 · 3 MARKS

- 1 mark: helicase unwinds the double helix by breaking the hydrogen bonds between base pairs, opening the replication fork.
- 1 mark: DNA polymerase adds complementary nucleotides to each exposed template strand, joining them into a new strand (A with T, C with G).
- 1 mark: each new molecule has one original strand and one newly made strand, so half of each molecule is conserved. This is the pattern Meselson and Stahl found in 1958.

QUESTION 20 · 3 MARKS

Model working. Mitosis gives two identical diploid cells, so 46 chromosomes in each. Meiosis halves the number, so each gamete has 23. After S phase each of the 46 chromosomes is two sister chromatids, so at metaphase there are 46 × 2 = 92 chromatids.

- 1 mark: 46.

- 1 mark: 23.
- 1 mark: 92 chromatids.

QUESTION 21 · 4 MARKS

Model working. AUG AAG UUU GGC UAA reads Met, Lys, Phe, Gly, then UAA is a stop codon, so the peptide is Met-Lys-Phe-Gly. AAG to AAA still codes for Lys (a different codon for the same amino acid), so the peptide is unchanged: a silent substitution. Inserting one A gives AUG AAA GUU UGG CUA A, which reads Met, Lys, Val, Trp, Leu and the stop signal is lost. Every codon after the insertion changes: a frameshift (insertion) mutation, usually much more damaging.

- 1 mark: Met-Lys-Phe-Gly.
- 1 mark: substitution is silent, still Lys, because the genetic code is redundant.
- 1 mark: insertion shifts the reading frame, so every later codon changes (Val, Trp, Leu) and the stop is lost.
- 1 mark: both mutation types named correctly (silent substitution; frameshift insertion).

QUESTION 22 · 4 MARKS

Model working. Mother gametes X^C and X^c; father gametes X^C and Y. The four children are X^CX^C (unaffected daughter), X^CX^c (carrier daughter), X^CY (unaffected son) and X^cY (affected son). Half of the sons are affected, no daughters are affected and 1 in 4 of all children is affected.

- 1 mark: correct gametes for both parents.
- 1 mark: correct Punnett square with the four genotypes above.
- 1 mark: son affected 1/2, daughter affected 0.
- 1 mark: any child affected 1/4.

QUESTION 23 · 3 MARKS

Model working. Cross the dog with a homozygous recessive (tt). If it is TT, every offspring is Tt and shows the dominant trait. If it is Tt, about half the offspring are tt and show the recessive trait. For eight dominant offspring from a Tt parent, $(1/2)^8$ = 1/256, about 0.4%.

- 1 mark: crosses with a tt (homozygous recessive) partner.
- 1 mark: correct predicted results for TT (all dominant) and Tt (about half recessive).
- 1 mark: probability 1/256.

QUESTION 24 · 3 MARKS

Model working. 2^{30} = 1,073,741,824, about 1.07 billion copies. The heating step that separates the strands (about 95 degrees Celsius) would destroy an ordinary polymerase, so a polymerase from a heat tolerant organism such as Thermus aquaticus is used and survives every cycle.

- 1 mark: sets up 2^{30}.
- 1 mark: 1 073 741 824 (about 1.07×10^9).
- 1 mark: the polymerase must survive the high temperature strand separation step, so it need not be replaced each cycle.

QUESTION 25 · 6 MARKS

- 1 mark: describes the mechanism: a guide RNA directs the Cas9 enzyme to a matching DNA sequence, Cas9 cuts both strands, and the cell repairs the break, which can disable a gene or be used to insert a corrected sequence.
- 1 to 2 marks: a benefit linked to a disorder, for example editing blood stem cells to treat sickle cell disease, where a 2024 clinical trial report in the New England Journal of Medicine (Frangoul et al.) reported 29 of 30 evaluable patients free of severe pain crises for at least 12 months.
- 1 to 2 marks: an ethical concern developed with a reason, for example edits to eggs, sperm or embryos pass to future generations who cannot consent, or off target cuts may cause unintended changes, or high cost limits access.
- 1 mark: a judgement, for example that treating body cells of consenting patients is better justified than editing the germline, with a reason.

Attempt record

Come back every few weeks and date it. Improvement in a science is easy to miss from the inside, which is why students who are getting better so often believe they are not.

DATE	MARK OUT OF 70	THE ONE THING TO FIX NEXT TIME

Fill one row every time you sit a set. A tracker with three rows in it is worth more than a perfect one you never started, because the pattern you are looking for only shows up across attempts, not inside one.

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