Mark schemes

## Q1.

(a)  (an allele) that is always expressed
**or**
(an allele) that is expressed even when the other / recessive allele is present
**or**
(an allele) that is expressed in the heterozygote
**or**
(an allele) that is expressed when only one copy is present

> *allow always shows in the phenotype*
>
> *ignore stronger*

**1**

(b)  (person **1**) has polydactyly so must have **D**

> *allow an annotated genetic diagram for up to **2** marks*
>
> *allow (person **1**) has polydactyly so must have a dominant allele*

**1**

has offspring / (person) **5** who does not have polydactyly so must have **d** from person **1**
**or**
person **5** does not have polydactyly so must be **dd** and must inherit **d** from person **1**

**1**

(c)  Female / **6**'s gametes correct **D** + **d**

**1**

Male / **7**'s gametes correct **d** + **d**

> *allow **1** mark for both sets of gametes if parents not identified*

**1**

correct derivation of offspring genotypes: **Dd Dd dd dd**

> *derivation must be consistent with parental gametes*

**1**

**Dd** correctly identified as polydactyly in **only** half of offspring

> *mp4 only awarded if mp3 is correct*

**1**

(d)   CF allele is recessive

   *allow CF is recessive*

**1**

to have CF, must have 2 CF alleles

**1**

**chance** of having one CF allele is $\dfrac{1}{50}$

   *ignore chance of having one CF allele is one in 50*

**1**

(chance of having two CF alleles is)

$$\dfrac{1}{50} \times \dfrac{1}{50} = \dfrac{1}{2500}$$

   *ignore 50 × 50 = 2500*

**1**

**[11]**

## Q2.

(a)    as volume of follicles rises oestrogen concentration (in blood) rises (for 7 / 8 days)

> *allow (positive) correlation between oestrogen concentration and volume of follicles (for 7 / 8 days)*
>
> ***or*** *oestrogen concentration is in proportion to follicle volume (for 7 / 8 days)*
>
> *do **not** accept an increase of oestrogen concentration causes an increase of follicle volume*

**1**

(b)    (volume of one follicle)

$= \frac{4}{3} \times 3.14 \times 11^3$

> *allow* $\frac{4}{3} \times \pi \times 11^3$

**1**

= 5572.(4533)

> *allow 5575(.279…)*

**1**

(total volume of follicles) = 39 000 (mm³)

**1**

$\frac{39\,000}{5572}$ 6.99…

> *allow use of an incorrect volume (from Figure 12) and / or an incorrectly calculated volume of one follicle*

**1**

7

> *do **not** accept 7.0*

**1**

(c)    (lack of FSH causes) lack of oestrogen (production)

> *allow lack of FSH causes lack of follicle development / growth / maturation*

**1**

breast development is dependent on oestrogen (from follicles)

> *allow (female) secondary sexual characteristics are dependent on oestrogen (from follicles)*

**1**

(d)   gametes correct:
**H** + **h**   **and**   **H** + **h**

**1**

correct derivation of offspring genotypes:
**HH**   **Hh**   **Hh**   **hh**

*allow correct for gametes stated in mp1*

**1**

correct phenotype for each genotype

*allow correct for genotypes stated in mp2*

*do **not** accept if no **hh** offspring*

**1**

(e)   mother (has **hh** so) passes on **h**

**1**

father (has **Hh** so) passes on **H**
or **h** with equal probability

**1**

(so) child will be **Hh** / heterozygous with 0.5 probability and produces FSH

**1**

*allow annotated genetic diagram for all marks*

**[14]**

**Q3.**

(a)   (A) → E → D → C → B → (F)

> *must be in this order*

1

(b)   (start) 16

> *allow 8 pairs*

1

(end) 8

> *the answer must be half the value of the number at the start*

1

(c)   (meiosis) forms gametes

> *allow (meiosis) forms sex cells*
>
> *allow (meiosis) forms eggs **and** sperm*

1

(two gametes) fuse / fertilise

1

(so) keeps chromosome number constant (from generation to generation)
**or**
(so) prevents doubling / increase in chromosome number (in each generation)
**or**
(so) gives normal / correct chromosome number (for embryo / new cell)

> *allow gives correct chromosome number (for offspring)*

1

(d)   random chromosome from each pair (of chromosomes)

> *ignore half the chromosomes*
>
> *ignore half the DNA*
>
> *allow other processes for creating variation*
>
> *ignore mutation*

1

moves to one end of the cell
**or**
goes into each new cell **or** gamete

1

(e)   base(s)

> *allow organic / nitrogenous base(s)*
>
> *ignore adenine / cytosine / guanine / thymine*

1

(f)   12 / twelve

> *allow 6 <u>pairs</u> **or** six <u>pairs</u>*

1

**[10]**

## Q4.

(a)   parents without MSUD have a child with MSUD

> *allow 1 and 2 (without MSUD) have child 5 (with MSUD)*
>
> **or**
>
> *7 and 8 (without MSUD) have child 12 (with MSUD)*
>
> *ignore MSUD skips a generation*

**1**

(b)   gametes correct:
**N + n   <u>and</u>   N + n**

**1**

correct derivation of offspring genotypes:
**NN   Nn   Nn   nn**

> *allow correct for gametes stated*

**1**

correct phenotype for each genotype

> *allow correct for offspring genotypes*
>
> *ignore carrier*

**1**

correct probability:
0.25 / ¼ / 25% / 1 in 4

> *allow correct answer only*
>
> *allow 1:3*
>
> *do **not** accept 1 in 3 / 1:4*

**1**

(c)   liver

**1**

(d)   (no enzyme 2 made **so**) cannot break down the toxic substance

> *allow (no enzyme 2 made **so**) cannot change toxic substance into harmless products*

**1**

the toxic substance is still made (from the amino acids)

> *allow toxic substance builds up over time*
>
> *ignore concentration of toxic substance is high(er)*

**1**

toxic substance diffuses / moves (from cells) into the blood

> *ignore incorrect name of organ*
>
> *allow **P** for toxic substance throughout*

**1**

(e)   the toxic substance passes through filter in kidney
**or**
**P** passes through filter in kidney

**1**

(some / all) not reabsorbed

> *allow (some / all) not absorbed **back** into the blood*
>
> *or (some / all) not taken **back** into the blood*
>
> *ignore (some / all) not absorbed into the blood*
>
> *or (some / all) not taken into the blood*

**1**

(f)   proteins contain amino acids
**or**
proteins are made of amino acids

> *allow proteins are broken down into amino acids*

**1**

must keep (certain) amino acids in low amount

> *allow (so) (certain) amino acids do not build up*
>
> *allow (so) less of (certain) amino acids are produced*

**1**

(so) toxic substance   **or P** does not build up in the body **and** cause damage to cells / tissues / organs

**1**

**[14]**