1. B

[1]

2. A

[1]

3. A

[1]

4. A

[1]

5. D

[1]

6. B

[1]

7. A

[1]

8. A

[1]

9. C

[1]

10. (a) anaphase

1

(b) growth (through increasing cell number);
embryonic development;
tissue production/repair;
(asexual) reproduction; 2 max

(c) uncontrolled mitosis/cell division 1

(d) pair of homologous chromosomes moves in same direction/does not separate during anaphase I / chromatids move in same direction/do not separate during anaphase II;
leaving a cell with an (some) extra chromosome(s)/missing chromosome(s);
an example; *(e.g. Down syndrome / trisomy 21)*; 2 max

[6]

11. A

[1]

12. (a) (i) autosomes because the sex chromosomes/X and Y chromosomes would be different lengths/sizes / would have different genes 1

(ii) homologous because they have paired/formed a bivalent / tetrad / there is crossing over between the chromosomes / they have the same genes (in the same sequence) / they are the same size and shape 1

(b) first prophase/first metaphase/prophase I/metaphase I 1

(c)

*Allow **[1]** only if the C allele is not on the short arm or the A and B alleles are not on the long arm. Use a maximum of two ticks in your marking.* 2

(d) (gene) linkage / autosomal linkage 1

[6]

13. (a) *allele*: one specific form of a gene (occupying the same gene locus as other alleles of the same gene) 1

(b) I^BI^B and I^Bi 1

(c) *Award **[1]** for every two correct answers.*
I. bacterial cell/bacterium/prokaryote;
II. plasmid;
III. inserted/engineered/cloned/desired DNA/DNA from donor cell;
IV. genetically modified/transformed/GM/recombinant organism/ cell/bacterium/host cell containing recombinant plasmid; 2 max

(d) restriction enzymes / endonucleases;
ligases;
reverse transcriptase;
*Award **[1]** for two correct responses.* 1 max

[5]

14. (a) three copies/extra copy/trisomy of chromosome/pair 21 1

Do not accept chromatid.

(b) occurs due to non-disjunction;
(homologous) chromosome/pairs/sister chromatids fail to separate;
some gametes have an extra chromosome; 2 max

(c) male as sex chromosomes different size/both X and Y chromosomes present 1
*To award **[1]**, reason must be given.*

(d) gene for colour blindness is carried on X chromosome (sex linkage);
males have only one X chromosome so the allele is always expressed /
absent from Y chromosome;
the allele of the gene for colour blindness is recessive;
females must be homozygous to be colour-blind / heterozygous
females are carriers but not colour-blind; 2 max

[6]

15. D

[1]

16. (a) drugs used to down-regulate the menstrual cycle;
FSH injected to stimulate many follicles to develop;
HCG injected to cause the follicles to mature;
eggs are harvested/extracted (from the follicles/ovaries);
semen sample produced/collected;
semen is processed to concentrate it / healthy sperm selected
(swim-up test given);
ICSI/IntraCytoplasmic Sperm Injection where sperm is directly
injected into egg when low numbers of motility is a factor;
semen mixed with eggs in a dish/outside the body to allow fertilization;
incubated / kept at 37°C / allows embryos to develop (sufficiently
for implantation);
dish examined to choose healthiest embryo;
embryos placed in uterus/oviduct (using a catheter/long plastic tube);
one/two/three/up to four (in some countries) embryos implanted;
pregnancy test/scan used to see if procedure has been successful;
(used in cases of) blocked oviduct / low sperm count / need for genetic
screening /infertility / cannot become pregnant / need for donor embryo; 9 max
*Accept other reasonable situations. Full marks may be awarded
only if an example is included in the answer.*

(b) females are carriers when they have dominant and recessive alleles together;
recessive allele in carrier does not affect phenotype as dominant allele also present;
gene is located on the X chromosome / gene is not located on Y chromosome;
females are XX so can have dominant and recessive alleles/two alleles of gene;
males are XY so only have dominant or recessive allele/one allele of gene;
hemophilia/red-green colour blindness/other example of a sex-linked characteristic; 4 max

(c) (point) mutation of gene <u>for hemoglobin</u>;
CTC to CAC / GAG to GTG / substitution of T/thymine with A/adenine;
mRNA copy of gene is GUG instead of GAG;
valine instead of glutamic acid;
(in homozygotes) red blood cells become sickle-shaped;
(in homozygotes) less oxygen carried;
(in homozygotes) red blood cells do not survive long / burst / block blood vessels/capillaries / circulatory problems may cause pain/ organ failure/example of symptom;
heterozygotes have malaria resistance; 5 max
(Plus up to ***[2]*** for quality)

[20]

17. (a) Down syndrome is caused by non-disjunction;
occurs during meiosis;
chromosome pairs fail to separate in meiosis I / chromatids in meiosis II / anaphase II;
some <u>gametes</u> have an extra chromosome;
can lead to <u>zygotes</u>/<u>individuals</u> with an extra chromosome / individual has 47 chromosomes;
in Down syndrome this would be trisomy 21/extra chromosome 21;
increased probability with increased age of mother/ages of parents; 5 max

(b) skin colour is an example of polygenic inheritance;
many/more than two genes contribute to a person's skin colour;
due to the amount of melanin in the skin;
combination of alleles determines the phenotype;
allows for range of skin colours / continuous variation of skin colour;
phenotypes do not follow simple Mendelian ratios of dominance and recessiveness;
the environment also affects gene expression of skin colour / sunlight/UV light stimulate melanin production;
the more recessive alleles there are, the lighter the skin colour; *(vice versa)* 5 max

(c) caused by gene mutation;
(sickle-cell anemia) due to a base substitution (mutation);
changes the code on the DNA;
which leads to a change in transcription / change in mRNA;

which (in turn) leads to a change in translation / change in polypeptide chain/protein;
(the tRNA) adds the wrong amino acid to the polypeptide chain;
glutamic acid replaced by valine;
produces abnormal hemoglobin;
causing abnormal red blood cell/erythrocyte shape / sickle shape;
which lowers the ability to transport oxygen;
sickle-cell allele is codominant;
homozygote/Hb^S Hb^S have sickle cell anemia/is lethal / heterozygote/ Hb^S Hb^A has the sickle trait/is carrier (and is more resistant to malaria); 8 max
(Plus up to ***[2]*** for quality)

[20]