Essential idea: Every living organism inherits a blueprint for life from its parents.

3.1 Genes

Understandings

(1) A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic. (p. 142)
   - Factors in living organisms that determine **inheritable characteristics** and are **genes**
   - **Genes** is the name given to the inheritable factors.
   - Only **46** chromosomes, but **thousands** of genes.
   - The shorter the chromosome, the **more** genes it has.

(2) A gene occupies a specific position on a chromosome. (p. 143)

<table>
<thead>
<tr>
<th>Chromosome 11</th>
</tr>
</thead>
<tbody>
<tr>
<td>Manic-depressive illness</td>
</tr>
<tr>
<td>Diabetes mellitus</td>
</tr>
<tr>
<td>Sickle-cell anemia</td>
</tr>
<tr>
<td>T-cell leukemia</td>
</tr>
<tr>
<td>Liver-cell cancer</td>
</tr>
<tr>
<td>B-cell leukemia</td>
</tr>
</tbody>
</table>

- Each gene is found at a specific **location** on a chromosome and this position is called the **locus** (loci).

(3) The various specific forms of a gene are alleles. (p. 143)
   - Different forms of a gene are called **alleles**.
   - Some genes have more than **two** alleles.
   - There are **three** alleles that determine blood type.
   - Alleles are found at a specific location on the chromosome called the **locus**.
   - There is only **one** allele at the **locus** on a chromosome.
(4) Alleles differ from each other by one or only a few bases. (p. 144)
- A gene consists of DNA with a base sequence that can be hundreds or thousands of nucleotides long.
- The different alleles have a change in the base sequence. Usually it is only one or a small number bases that are different.
- Positions in a gene where more than one base may be present is called single nucleotide polymorphism (SNPs) or snips.
- Several snips may be present in a gene, but the alleles will only differ by a one or only a few bases.

(5) New alleles are formed by mutation. (p. 145)
- Mutations are random changes.
- Most mutations are a result of a random change.
- In a base substitution, one nucleotide replaces another nucleotide in the gene.
- Mutations are usually either neutral or harmful and some mutations may even cause death of a cell.
- If the cell is a gamete, then the mutation can be passed onto offspring and cause a genetic disorder.

(6) The genome is the whole of the genetic information of an organism. (p. 147)
- The genome is all genes in a living organism.
- In humans, 46 chromosomes and the DNA in the mitochondrion makes up the genome.
- In plants, the genome consists of the DNA in the nucleus, the DNA in mitochondrion and the chloroplast.
- In prokaryotes, the genome consists of the DNA in the nucleoid chromosome and other plasmid (circular rings of DNA).

(7) The entire base sequence of human genes was sequenced in the Human Genome Project. (p. 147)
- The goal of the Human Genome project was to sequence the entire genome of a human. It started in 1990 and was completed in 2003.
- There are approximately 23,000 protein coding genes.
- Most of the genome is not transcribed and was referred to as junk DNA or satellite DNA.
- Now it is known that these regions are important for DNA profiling or genealogical investigations or regulating gene expression.
- The genome that was sequenced is for one set of chromosomes. It is a human genome
It is a human genome rather than the human genome.

Research continues to find differences between different individuals.

The majority of the base sequences are the same between all humans, but there are single nucleotide polymorphisms (SNPs) that results in the variation within humans.

Applications

(1) The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin. (p. 146)

- Sickle cell anemia is the most common genetic disorder world-wide and affects red blood cells.
- It is caused by a single base substitution where the DNA sequence is changed from thymine (CTC) to adenine (CAC).
- This results in the 6th codon being changed from GAG to GUG, which causes one amino acid to change (from glutamic acid to valine). This variation changes the alpha-globin polypeptide in hemoglobin.
- This results in red blood cells that are sickle shaped and unable to carry oxygen efficiently. Sickle cells tend to accumulate in areas of low oxygen concentration. Red blood cells usually are replaced every three months, but sickle cell anemia results in a much shorter lifespan for affected cells to as short as four days.
- The gene for this protein is represented by HbA. For humans without sickle cell anemia, the allele would be represented by HbA, whereas someone with sickle cell anemia would have HbS.
- Sickle cell anemia is only inherited if the mutation occurs in the cells of the ovaries or testes.

- If both alleles are affected, then the individual develops severe anemia, whereas if only one allele is inherited with sickle cell anemia one allele normal, then the individual has mild anemia.
- In East Africa, 5% of newborns have both alleles affected, whereas 35% of have only one allele affected.
- There seems to be a positive correlation between Sickle cell anemia and malaria.
- The sickle cell red blood cells seem to provide immunity to malaria, thus sickle cell anemia tends to be more common in parts of the world where malaria is found.
(2) Comparison of the number of genes in humans with other species. (p. 142)

<table>
<thead>
<tr>
<th>Group</th>
<th>Name of species</th>
<th>Description</th>
<th>Number of genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Animal</td>
<td><strong>Homo sapiens</strong></td>
<td>large; omnivore; biped</td>
<td>23,000</td>
</tr>
<tr>
<td>Prokaryote</td>
<td><strong>Escherichia coli</strong> (E.coli)</td>
<td>gut bacteria</td>
<td>3,200</td>
</tr>
<tr>
<td>Fungi</td>
<td><strong>Sacccharomyces cerevisiae</strong></td>
<td>unicellular fungus (yeast)</td>
<td>6,000</td>
</tr>
<tr>
<td>Animal</td>
<td><strong>Drosophila melanogaster</strong></td>
<td>fruit fly; larvae consume ripe fruit</td>
<td>14,000</td>
</tr>
</tbody>
</table>

**Skills**

(1) Use of a database to determine differences in the base sequence of a gene in two species.

- GenBank

**Nature of science**

(1) Developments in scientific research follow improvements in technology – gene sequencers are used for the sequencing of genes. (p. 148)

- To sequence a genome, it is first broken up into small lengths of DNA nucleotides.
- Each is then sequenced separately.
- To sequence, single stranded copies are made using DNA polymerase.
- The whole process is stopped by putting small quantities of non-standard nucleotides.
- Four samples are produced and separated using gel electrophoresis.
3.2 Chromosomes
Nature of science
(1) Developments in research follow improvements in techniques – autoradiography was used to establish the length of DNA molecules in chromosomes (p. 150–151).
   - Autoradiography was used by scientists to discover where specific molecules were found in cell.
   - John Cairns used this technique to create images of DNA from E. coli.
   - His images indicated that there was a single DNA molecule in the bacterial cell and showed the replication forks in DNA.

Understandings
(1) Prokaryotes have one chromosome consisting of a circular DNA molecule. (p. 149)
   - Most prokaryotes have one chromosome consisting of a circular DNA molecule.
   - The DNA in bacteria is termed as being naked since it is not associated with protein.
   - There is only a single copy of each gene since there is only one chromosome.

(2) Some prokaryotes also have plasmids but eukaryotes do not. (p. 150)
Bacterium

Bacterial chromosome     Plasmid

Plasmids are __________ small extra DNA __________ found only in prokaryotic cells.
They are __________ small __________ circular __________, and do not contain genes necessary for __________ life __________.
Plasmids often contain genes involved in __________ antibiotic resistance __________.
Plasmids are __________ not always replicated at the same time as the __________ chromosome __________ of prokaryotic cells so sometimes a prokaryotic cell may not have a plasmid or may have multiple copies of a plasmid.
Plasmids can be transferred between different __________ cells __________.
Plasmids can be used to __________ transfer __________ genes between species in a lab setting.

(3) Eukaryote chromosomes are linear DNA molecules associated with histone proteins. (p. 151-152)
In eukaryotic cells, chromosomes consist of __________ DNA __________ and __________ protein __________.
The proteins are __________ histone __________ proteins.
Histones are __________ globular __________ while DNA is a long, __________ narrow __________ molecule.
The DNA molecule is __________ wrapped twice __________ around the histone proteins. (nucleosomes)
There are many __________ types of histone __________ proteins associated with the DNA molecule.
Eukaryotic DNA looks like beads on a string because there is DNA wrapped around each __________ nucleosome __________ and there are sections of DNA between each chromosome.
(4 types of histone proteins)

(4) In a eukaryote species there are different chromosomes that carry different genes. (p. 152)
There are 24 types of chromosomes in humans. When viewed in mitosis, chromosomes differ in size and the position of the centromere is that holds the chromatids together. Each gene is located at a particular position called the locus. Thus each chromosome carries a specific number of genes. Some chromosomes carry over a thousand genes.

Homologous chromosomes carry the same sequence of genes but not necessarily the same alleles of those genes. Even though homologous chromosomes have the same genes, they do not always have the same alleles.

Diploid nuclei have pairs of homologous chromosomes. A diploid nucleus has 2 chromosomes of each type. When gametes (a sperm cell and an egg cell) join, a diploid nucleus is created. Diploid nuclei carry 2 copies of every gene. Mutations can be avoided because a dominant allele will mask the effects of a recessive allele. In humans, 23 chromosomes would be in a haploid nucleus.

Haploid nuclei have one chromosome of each pair. Haploid nuclei only have 1 chromosome from each type. In humans, 23 chromosomes would be in a haploid nucleus. Gametes (egg and sperm cells) have a haploid nucleus.

The number of chromosomes is a characteristic feature of members of a species. Different organisms have different number of chromosomes. Organisms with different numbers of chromosomes are unable to interbreed.

A karyogram shows the chromosomes of an organism in homologous pairs of decreasing length.
• shows all the chromosomes in one cell of a particular individual
• organized by size of chromosomes (largest to smallest - pair 1 to pair 22)
• used to detect chromosomal defects + gender

(10) Sex is determined by sex chromosomes and autosomes are chromosomes that do not determine sex. (p. 157)

female

male

• autosomes (chromosome pairs #1-22) - all the chromosome pairs that are identical in males + females
• sex chromosomes (pair #23) - determines gender
(1) Cairns' technique for measuring the length of DNA molecules by autoradiography.

- John Cairns used **E. coli** to produce images of DNA.
- This is how he did it: he grew cells on media that had **thymidine**. Thymidine consists of the base **thymine** attached to **deoxyribose** as well as a radioactive isotope called **tritium**. After the E. coli replicated it had radioactively labeled **DNA**.
- The DNA was released onto a **dialysis** membrane by using the enzyme **lysozyme** which broke the cell **walls** (of the bacterial cell).
- A thin **film** of photographic emulsion was left on the dialysis membrane for 2 months. The radioactive tritium decayed and left a **dark** grain on the film representing the DNA.
- The images produced illustrated that DNA in **E. coli** is a **circular** molecule with a length of **1,100 μm**. E. coli cells are only **2 μm** in length themselves.

(2) Comparison of genome size in T2 phage, Escherichia coli, Drosophila melanogaster, Homo sapiens, and Paris japonica.

<table>
<thead>
<tr>
<th>Organism</th>
<th>Genome size (million of base pairs)</th>
</tr>
</thead>
<tbody>
<tr>
<td>T2 phage (virus that attacks E. coli)</td>
<td>0.18</td>
</tr>
<tr>
<td>Escherichia coli (gut bacteria)</td>
<td>5</td>
</tr>
<tr>
<td>Drosophila melanogaster (fruit fly)</td>
<td>140</td>
</tr>
<tr>
<td>Homo sapiens (human)</td>
<td>30,000</td>
</tr>
<tr>
<td>Paris japonica (woodland plant)</td>
<td>150,000</td>
</tr>
</tbody>
</table>

(3) Comparison of diploid chromosome numbers of Homo sapiens, Pan troglodytes, Canis familiaris, Oryza sativa, Parascaris equorum.

<table>
<thead>
<tr>
<th>Organism</th>
<th>Diploid chromosome #</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parascaris equorum (horse threadworm)</td>
<td>4</td>
</tr>
<tr>
<td>Oryza sativa (rice)</td>
<td>24</td>
</tr>
<tr>
<td>Homo sapiens (human)</td>
<td>46</td>
</tr>
<tr>
<td>Pan troglodytes (chimpanzee)</td>
<td>48</td>
</tr>
<tr>
<td>Canis familiaris (dog)</td>
<td>78</td>
</tr>
</tbody>
</table>

(4) Use of karyograms to deduce sex and diagnose Down syndrome in humans.
Skills
(1) Use of databases to identify the locus of a human gene and its polypeptide product.

Essential idea: Alleles segregate during meiosis allowing new combinations to be formed by the fusion of gametes.