Oxford Cambridge and RSA

A Level Biology B

H422/01 Fundamentals of biology

Question Set 8

1. (a) (i) A Robertsonian translocation is a type of chromosomal translocation in which the long arms of two chromosomes fuse together.

Fig. 31.1 shows this event occurring between chromosomes 14 and 21.

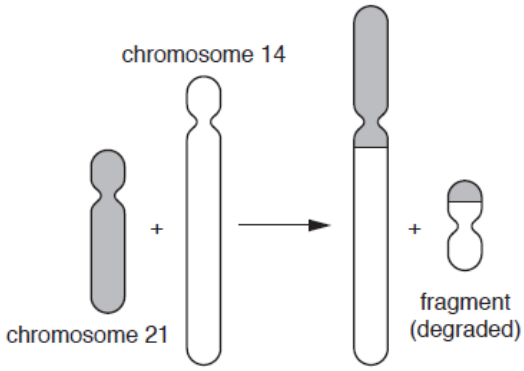

Fig. 31.1

An individual who inherits the translocated chromosome in Fig. 31.1 will either have Down's syndrome or be a carrier of the disorder.

A couple have a child. The mother is a carrier and the father is genetically normal. The genetic material with respect to chromosomes 14 and 21 in the somatic cells of the parents are shown in Fig. 31.2.

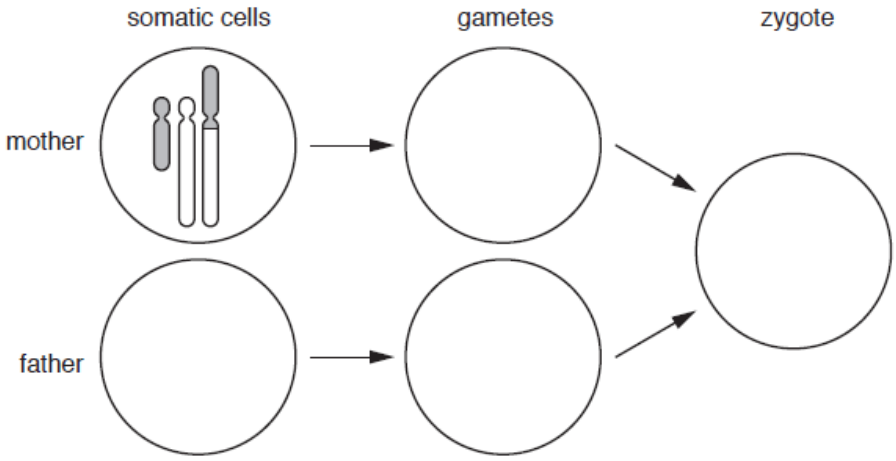

Fig. 31.2

With reference to Fig. 31.2, suggest why the mother does **not** have Down's syndrome. **[1]**

(a) (ii) The child is born with Down's syndrome.

Complete the diagram in Fig. 31.2 to show the genetic material with respect to chromosomes 14 and 21 in:

- the somatic cell of the father
- the gametes of the mother and father
- the zygote of the child.

[**Answer on Fig. 31.2**] **[4]**

(b) Down's syndrome is more commonly caused by a genetic event that is distinct from that shown in Fig. 31.1.

State the name of this event **and** outline how it arises. **[3]**

(c) **(i)** A sample of cells can be collected from a fetus to test for genetic disorders such as cysticfibrosis.

Chorionic villus sampling (CVS) and amniocentesis are two methods of obtaining fetal cells.

State the **source** of fetal cells that are obtained through **each** these two methods. **[1]**

(c) **(ii)** The sample of fetal cells can be used to produce a karyotype for genetic analysis.

Explain why karyotyping can **not** be used to detect cystic fibrosis. **[1]**

Total Marks for Question Set 8: 10