

Human karyotypes

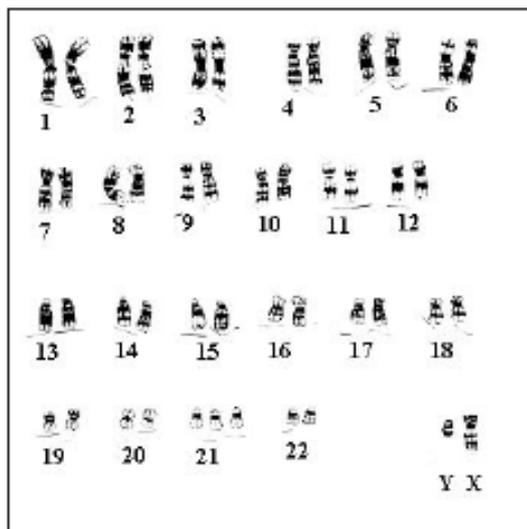
Introduction

In this activity students learn how a sample from amniocentesis is analysed to detect the presence of chromosomal abnormalities. They carry out a practical matching activity and report on the sex and any abnormalities in the karyotype. The activity complements those on antenatal testing and on genetic counselling.

Results

The finished karyotype should look like this.

The fetus is male. It has three chromosomes at 21 and therefore has Down's syndrome.



Karyotyping - a sample

Resources

Scissors, glue and plain white paper.

Science explanations

Ca Instructions for development are found in the form of genes which are part of the chromosomes in the nucleus of every cell in the organism. Each gene is a segment of a very long molecule of DNA. Chromosomes contain a large number of genes. All cells except sex cells, and red blood cells, contain two sets of chromosomes.

Cb In sexual reproduction, a single specialised cell from a female merges with another specialised cell from a male. Each of these sex cells contains a randomly selected half of the parent's genes. The single cell which they form then contains a full set of genetic information.

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Introduction

Some genetic diseases are caused by chromosome mutations where the distribution of the chromosomes is affected. One of the commonest chromosome mutations is called non-disjunction, and causes what is known as Down's syndrome. Down's syndrome occurs when, at a crucial stage in cell division, the chromosomes do not all separate equally into the resulting new cells. Some of the chromosomes lag behind, resulting in an extra one in one cell, and one less in the other. The cell that has the extra chromosome can still act as a sex cell and if it fuses with a 'normal' egg or sperm cell, the resulting baby will have not 46 but 47 chromosomes in every cell. Down's syndrome, in which an individual's cells contain a third copy of chromosome 21, is diagnosed by karyotype analysis.

How can we detect Down's syndrome in a baby?

This exercise will enable you to identify the typical Down's syndrome karyotype. A karyotype is the name given to a picture of the chromosomes from one cell.

If a baby is suspected of having inherited the Down's mutation, cells can be obtained from the amniotic fluid or umbilical cord, and then tested as shown in Figure 2.

Figure 1 A template for numbering chromosomes

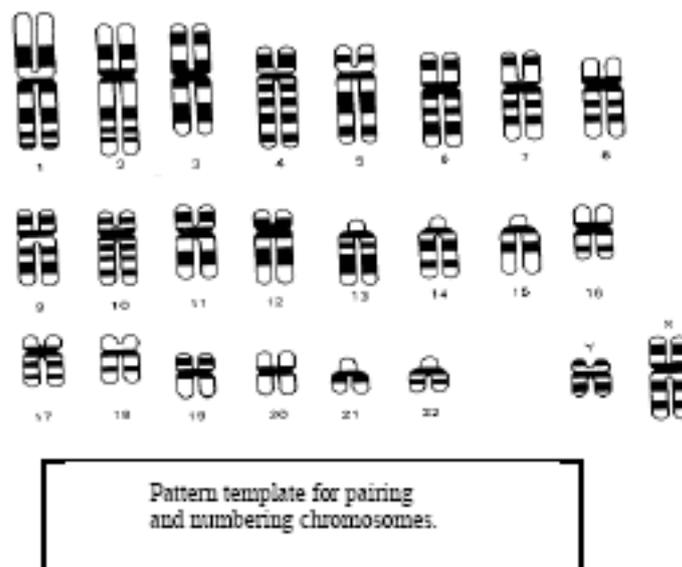
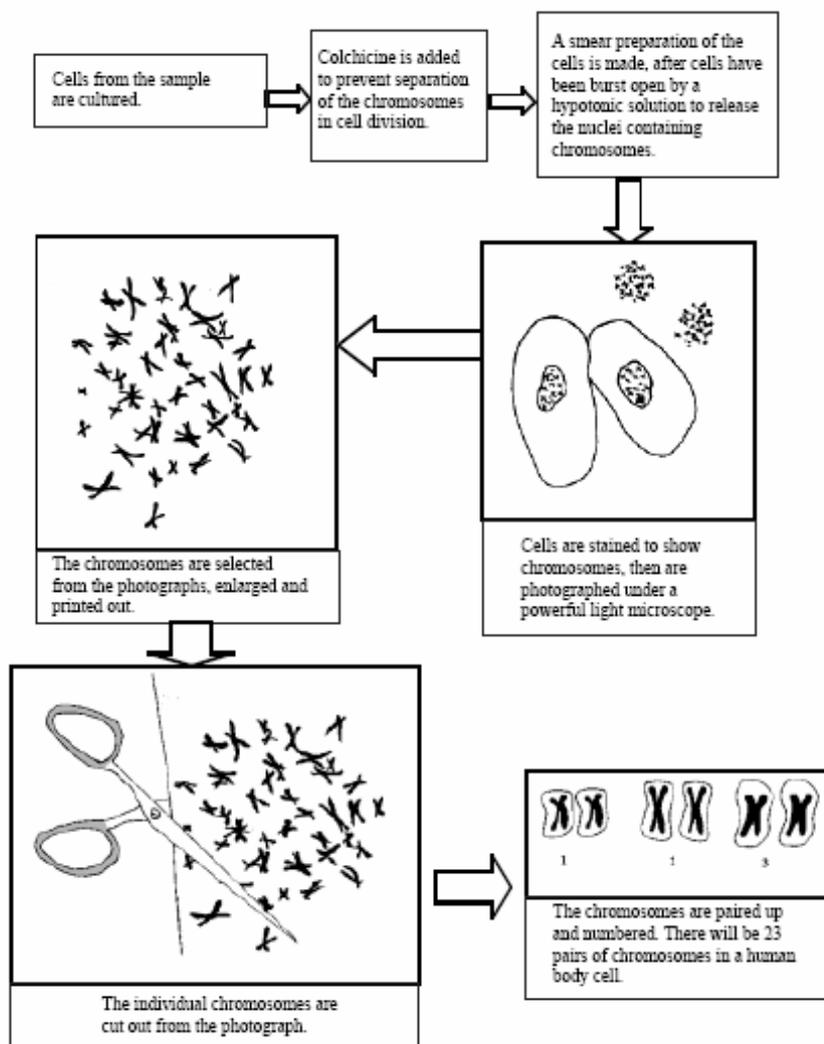


Figure 2

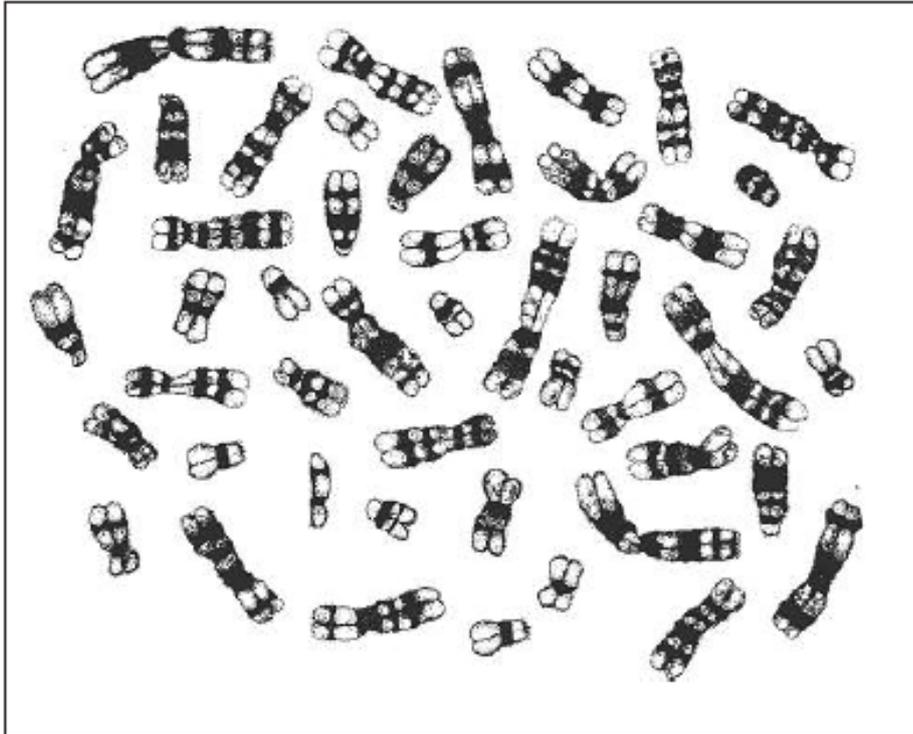


Karyotyping a sample

In this exercise you will act as the medical technician responsible for identifying whether the baby has Down's or not. You can imagine the sense of responsibility that this has, when you consider the effect that the result will have on the prospective parents. So it is very important that you make no mistakes in your analysis.

1. Study the picture in Figure 3, of chromosomes taken from a baby. Cut out each chromosome, and place them on a sheet of white paper.
2. Study each chromosome very carefully, noting the size and the banding patterns. (Chromosomes can be seen under a light microscope and, when stained with certain dyes, reveal a pattern of light and dark bands. Differences in size and banding pattern allow the 24 chromosomes to be distinguished from each other, an analysis called a karyotype.)
3. Carefully match the pairs of chromosomes until you have accounted for every one. Try to number the pairs using the pattern template in Figure 1.
4. Is this baby a male or female? How do you know?
5. Do you have any other comments about this karyotype?

Figure 3



Chromosomes from an amniocentesis test