

Chromosomal conditions

Chromosomes are important parts of a cell. They are the parts which carry the genes.

We are meant to have 23 pairs of chromosomes in each cell, a total of 46 chromosomes, with one chromosome in each pair inherited from each parent.

Occasionally, things go wrong, and a child is conceived with 45 chromosomes, or 47 chromosomes, or perhaps the right number but the wrong mix of chromosomes. Also, a child may be born with a change to the way the chromosomes are structured.

More often than not, babies conceived with chromosomal alterations have such serious problems that their mothers miscarry. Sometimes, they survive.

The most common chromosomal alterations in surviving babies are:

- Trisomy 21 or Down syndrome
- Trisomy 18 or Edwards syndrome
- Trisomy 13 or Patau syndrome
- XXY or Klinefelter syndrome
- 45X or Turner syndrome
- XXX or triple X syndrome
- XYY syndrome.

Individually, these conditions are all uncommon, although in total, about 6 babies in 1000 are born with a chromosomal alteration.

While babies with the same chromosome condition will tend to have similar features, it is impossible to predict what any individual child will look like, what their abilities may be, or what medical problems they may face. General information on what might be involved is provided in brief description below, but for more detailed information visit <http://www.genetics.edu.au> the website for the Centre for Genetics Education.

Trisomy 21 or Down syndrome

Babies with Down syndrome will be slow to develop and will have intellectual disability. Problems with the heart and bowel are also common. Most people with Down syndrome will live independent lives.

Trisomy 18 or Edwards syndrome

Most babies with trisomy 18 will have serious and widespread problems. Most die soon after birth.

Trisomy 13 or Patau syndrome

Most babies with trisomy 13 will have serious and widespread problems. Most die soon after birth.

XXY or Klinefelter syndrome

Boys with XXY may be slow to develop and will often be shy. Their intelligence is usually normal, although they may have learning difficulties. They are usually infertile and tend to be tall. They have a normal lifespan.

45X or Turner syndrome

Girls with 45X syndrome are short, usually infertile and may have problems with their heart and kidneys. Their intelligence is usually normal, but they may have learning difficulties. They have a normal lifespan.

XXX or triple X syndrome

Girls with XXX may have reduced intelligence and learning difficulties. They tend to be tall. They have a normal lifespan.

XYY syndrome

XXY syndrome only affects males. Boys with XYY syndrome tend to be tall. Their intelligence is usually normal, but they may have some learning difficulties. They have a normal lifespan.

Are chromosomal alterations inherited?

Usually not, but sometimes they are, particularly if the chromosomal alteration affects the way the chromosome is structured. If a child in your family is diagnosed with a chromosomal alteration, or if there is a history of miscarriages, then it is worth seeing a genetics service to discuss the possibility of the alteration being inherited.

What to do?

If you have or are having a child with chromosomal alterations, much more information is available from support groups and from genetics services. You should seek specialist help and advice.

Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- National Organisation for Rare Disorders at <http://www.rarediseases.org>
- Trisomy 18 Registry and Research Society at <http://www.chromosome18.org>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- MyDr at <http://www.mydr.com.au>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email gtis-australia@unimelb.edu.au or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>