

## Some specific examples

Maternal UPD 14 causes some delay to baby's growth both in the womb and after birth. Babies with Mat UPD 14 often have a characteristic face, with a large broad forehead. Intellectual development is low-normal to normal. Children may be prone to obesity.

**Paternal UPD 14** causes severe problems with both physical and mental development with additional complications in pregnancy. Usually survival is not longer than 1 year.

**Maternal UPD 15** causes Prader-Willi syndrome (PWS). A leaflet is available which explains more about PWS.

**Paternal UPD 15** causes Angelman Syndrome (AS). A leaflet is available which explains more about AS.

## Further information

You can get more information about UPD from you Genetics doctor or Genetic Counsellor.

You can also get information from Unique: the rare chromosome disorder support group.

Website: <http://www.rarechromo.org>

The logo for 'Unique' is written in a stylized, cursive font with a horizontal line underneath the word.

## Acknowledgment

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# Uniparental Disomy (UPD)

An information leaflet for patients and families

If you need more advice about any aspect of Uniparental Disomy (UPD please contact:

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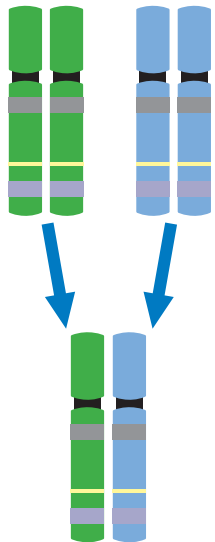
Email: [Clinicalgenetics.info@bwhct.nhs.uk](mailto:Clinicalgenetics.info@bwhct.nhs.uk)

## What are genes and chromosomes?

Our genes are the unique set of instructions inside every cell of our body which make each of us an individual. There are many thousands of different genes, each carrying different instructions. Genes are passed on from generation to generation in packages called chromosomes.

We all have 23 pairs of chromosomes in every cell. For each pair, we normally inherit one chromosome from our mother and one from our father (via the sperm and egg). We therefore have two copies of every gene; one from each parent.

One copy of each chromosome pair comes from each parent.

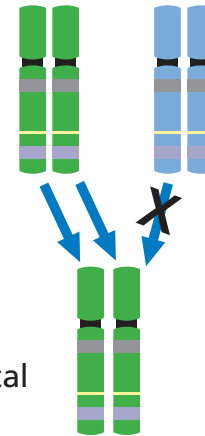


The chromosomes are marked to show which parent they are inherited from.

## What does Uniparental Disomy (UPD) mean?

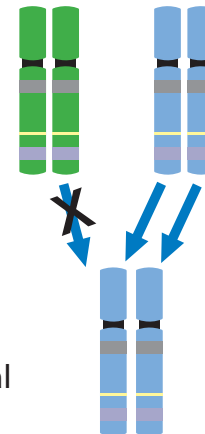
The word uniparental means relating to one parent. Disomy means two copies, in this case, of a chromosome. When put together the term UPD means the baby or child has received both copies of a particular chromosome pair from one parent. This is not the normal way that chromosomes are inherited and can, depending on which chromosome pair is involved, lead to medical problems for that baby or child.

Occasionally a baby can inherit two copies of one of the mother's chromosomes and no copies of that particular chromosome from the father.



This is known as Maternal Uniparental Disomy (Mat UPD).

Sometimes a baby can inherit two copies of one of the father's chromosomes and no copies of that particular chromosome from his/her mother.



This is known as Paternal Uniparental Disomy (Pat UPD).

## What does UPD mean for a baby or child?

For most of the chromosomes, there are no consequences or problems with having inherited two copies from the same parent. This is because for most genes, the information from both copies is actively being used.

However, there are genes on some chromosomes which are "switched off" when the egg or sperm are made. Some genes are always "switched off" when inherited from the mother, and others when inherited from the father. This is called imprinting.

If a child inherits two copies of a gene that has been "switched off", they will have no working copies of these genes which can cause problems with the child's development.

## UPD and chromosome translocations

Some people have a chromosome translocation. This means they have an unusual arrangement of chromosomes that can cause problems when trying to have children. In some cases there is a chance that a future pregnancy could have UPD. This can be discussed with your local genetics department. In some situations it is possible to test a pregnancy to see if the baby has UPD for a particular chromosome pair. These tests can also be discussed with your local genetics department.