Uniparental Disomy (UPD)
Information for patients
Clinical Genetics

UPD for a particular chromosome pair.

These tests can also be discussed with your local genetics department.

Some specific examples

Maternal UPD 14 - causes some delay to the 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 babies with paternal UPD 14 live for no longer than a year.

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

Paternal UPD 15 - causes Angelman syndrome (AS). A separate leaflet is available which explains more about AS.

Further information
Human Fertilisation and Embryology Authority
www.hfea.gov.uk

Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

www.nuh.nhs.uk

If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 67184.

The Trust endeavours to ensure that the information given here is accurate and impartial.

Adapted from leaflet by Clinical Genetics at Guy’s and St Thomas’ NHS Foundation Trust.

Dr Claire Searle, Clinical Genetics, © August 2018. All rights reserved. Nottingham University Hospitals NHS Trust. Review August 2020. Ref: 1050/v4/0818/AH.
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:

**Mother**

**Father**

The chromosomes are marked to show which parent they are inherited from. However, this programming is reset when this person has their own children.

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, depending on which chromosome pair is involved, can 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 the 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 associated 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 are always 'switched off' 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 that 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 as a result of the translocation. This can be discussed with your local genetics department.

In some situations it is possible to test a pregnancy to see whether the baby has