



Information for Patients

Uniparental Disomy (UPD)

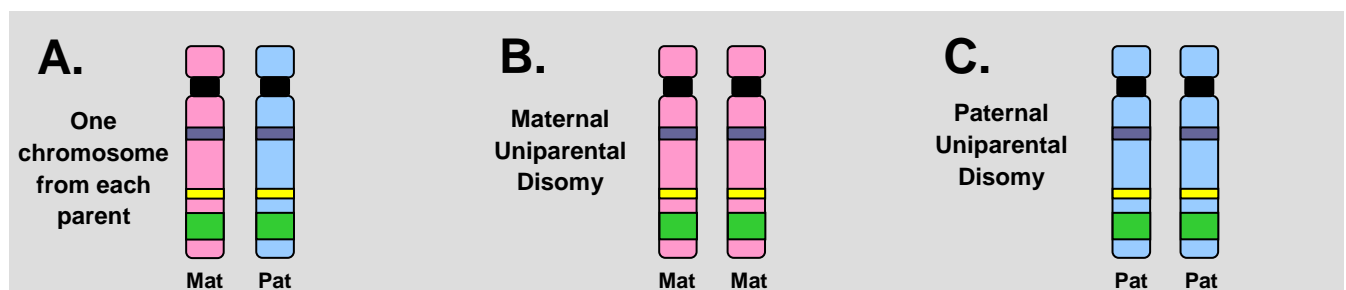
Information for carriers of Robertsonian Translocations involving chromosomes 14 or 15 during prenatal diagnosis

What is Uniparental Disomy (UPD)?

We all have 23 pairs of chromosomes in every cell. For each pair, we typically 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. See diagram below: "A".

Occasionally a baby can inherit two copies of one of the mother's chromosomes and no copies of that chromosome from his/her father. This is known as Maternal Uniparental Disomy (Mat UPD). See diagram below: "B".

Sometimes a baby can inherit two copies of one of the father's chromosomes and no copies of that chromosome from his/her mother. This is known as Paternal Uniparental Disomy (Pat UPD). See diagram below: "C".



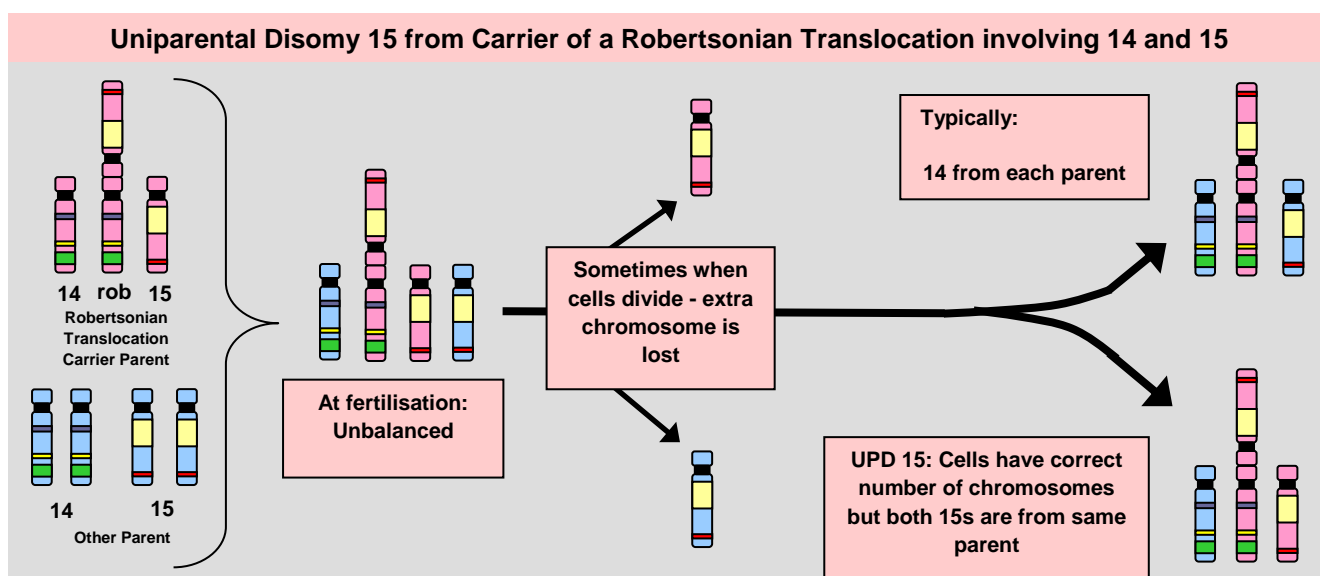
When a carrier of a Robertsonian Translocation involving chromosomes 14 or 15 conceives, there are several ways that these particular chromosomes can be passed on (a leaflet is available which explains more about this).

Sometimes a baby appears to have inherited the parent's 'balanced' translocation but in fact has UPD. There is a low (less than 1%) chance of this happening. A blood sample from each parent can be taken to check for UPD during prenatal diagnosis.

What does UPD mean for my child?

There are genes on chromosomes 14 and 15 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 and this can cause problems with the child's development. If the mother carries the translocation Maternal UPD can occur; if the father carries the translocation Paternal UPD can occur.



Chromosome 14 UPD

Maternal UPD 14 (also known as Temple syndrome) 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. Their intellectual development may be normal or they may have some mild learning difficulties. As children, they may be prone to obesity.

Paternal UPD 14 (also known as Wang syndrome) causes severe problems with both physical and mental development with additional complications in pregnancy. Babies who are born with Paternal UPD 14 will usually not survive for longer than the first year of life.

Chromosome 15 UPD

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.

For more information

If you need more advice about Uniparental Disomy, you are welcome to contact:

Manchester Centre for Genomic Medicine

Sixth Floor
Saint Mary's Hospital
Oxford Road Manchester M13 9WL

Telephone: (0161) 276 6506 (Reception)

Facsimile: (0161) 276 6145

Department staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____