http://www.southampton.ac.uk/geneticimprinting/informationclinicians/conditions/Maternal+UPD+14+%28Temple+Syndrome%29.page

Wessex Imprinting Group

Maternal UPD 14 (Temple Syndrome)

Uniparental disomy is the inheritance of both chromosome homologues from one parent with no functional copy from the other.

Since the first reports of Temple et al in 1991, a well characterised clinical phenotype has emerged for both maternal uniparental disomy of chromosome 14 (UPD14).
Maternal UPD14 or Temple Syndrome, is characterised by pre and postnatal growth retardation, hypotonia, joint laxity, motor delay, early onset of puberty, and minor dysmorphic features of the face, hands, and feet.

Imprinted locus at 14q32

There is good evidence that this syndrome is due to aberrant expression of genes at one imprinted locus that exists at 14q32 under the control of a paternally-methylated intergenic differentially-methylated region (IG-DMR). The imprinted genes in this region include the paternally-expressed genes DLK1 (delta, Drosophila homolog-like1) a transmembrane signalling protein and growth regulator homologous to proteins in the Notch/delta pathway and RTL1(retrosposon-like1, PEG11). Maternally expressed genes include numerous C/D box small nucleolar (sno)RNAs and microRNAs, GTL2 (gene trap locus 2), RTL1as and MEG8.

As with other imprinting disorders, it has now been shown that mechanisms that results in functional hemizygosity of 14q32 imprinted genes can cause the clinical phenotypes. For example we showed that Temple syndrome can be caused by an isolated methylation deficit at the IG-DMR at 14q32 and there have subsequently been other cases.

Which patients could be tested?

We provide diagnostic testing for all patients with possible UPD 14/Temple syndrome. Specific features that increase the chance of diagnosis are:

- marked hypotonia and normal muscle power
- early pubertal development
- scoliosis
- relatively low birth weight at term
- small hands and feet
- developmental delay (although this is not always a feature)
- central obesity developing with age.

The differential diagnosis includes Prader Willi syndrome.

Testing

We would strongly recommend taking blood or DNA from both parents as well as the child, to establish the diagnosis more rapidly.

Details to send with samples send details on our request form for imprinting disorders. These include:

- Referring clinician details

- Patient Details
- General Presentation
- Current status of Patient
- Any other clinical features

Consent

When taking samples you should get written consent for the testing - as with any genetic test. You can download a consent form from this site which you might like to use, modify, or translate. We ask you to get consent from the patient/parents to test for 'imprinting disorders' rather than just UPD14.

Where should the sample be sent?

Please email us before sending samples djgm@soton.ac.uk
The sample should be sent to:
Dr Deborah Mackay,
Lecturer in Human Genetics
Wessex Regional Genetics Laboratory,
Salisbury Health Care Trust
Salisbury
Wiltshire
SP2 8BJ
01722 425048 or 01722429012

And clinical details can be discussed with
Dr Karen Temple
Prof of Medical Genetics
and Honorary Consultant in Clinical Genetics
Academic Unit of Genetic Medicine
Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA
ikt@soton.ac.uk
tel 02380 796625

For more information on genetic testing please look at the genetic testing section on this website.

References

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