UPD 13: no indication of maternal or paternal imprinting of genes on chromosome 13

We recently reported the first example of maternal uniparental isodisomy for chromosome 13.1 This was found in a phenotypically normal male who inherited a t(13;13) in a balanced karyotype from his mother who also carried the same isochromosome. The transmission of the t(13;13) from mother to child was confirmed by analysis of dinucleotide repeat markers in the child and both his parents and we concluded that there were no maternally imprinted genes on chromosome 13. Further analysis of the inheritance of this

A family with autosomal dominant polycystic kidney disease linked to 4q21-23

We have previously reported a large kindred of southern Italian origin with autosomal dominant polycystic kidney disease, which is not linked to markers on 16p13.3 Analysis of this family with microsatellites from 4q21-23 shows that the gene responsible for the disease

Allele segregation of CA repeat polymorphisms at D13S155 and D13S175 in the grandmother (GM), mother (M), and child (C). M and C are both carriers of an isochromosome 13 and show monomeric patterns at both loci. GM shows biallelic polymorphisms for both loci, none of which is shared by M and C.
in this family is at the recently discovered PKD2 locus. The figure shows the pedigree and haplotypes for the markers used in the analysis. D4S1534 gave a lod score of 1.47 at θ = 0 while multipoint analysis comparing the possibilities of the gene being between D4S231 and D4S423 or outside these markers gave a lod score of 2.32 in favour of the former.

We have extended the family tree previously described1 and have further clinical details. For those members not previously reported, all are normotensive and asymptomatic. Serum creatinine is <140 μmol/l unless specified otherwise. III-3 is 82 years old and an ultrasound scan in 1991 showed a slightly enlarged right kidney with numerous small cysts throughout both kidneys. Two small cysts were also seen in the left lobe of the liver. III-7 is 50 years old and has not been scanned. III-8 is 46 years old with numerous small, bilateral renal cysts shown by ultrasound in 1991. III-10, a 45 year old secretary living in France, was diagnosed five years ago as hypertensive and has a BP of 160/90. Serum creatinine is 120 μmol/l. She is otherwise asymptomatic and was diagnosed through our screening programme. Ultrasound scans show moderate renal enlargement with numerous renal cysts and two small liver cysts. III-3, III-4, IV-1, and IV-2 have all been examined in the Blood Pressure Unit and had ultrasound scans in 1991, at ages 30, 43, 20, and 22 years respectively. Multiple renal cysts were seen in III-3 and III-4 with a single liver cyst in III-3. IV-3, IV-4, and IV-5 are 20, 19, and 16 years old respectively and unscanned.

This family shows a mild clinical phenotype, as seen in other PKD families. Clinical data from this and other PKD2 families need to be collated in order to define the clinical presentation and prognostic indicators for this second locus, as has been done for PKD1.3

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Pedigree showing haplotypes derived from loci on chromosome 4q. In descending order, the loci are D4S392, D4S231, D4S1534, D4S423, D4S414, D4S411. An asterisk indicates the affected haplotype, an arrow shows a recombinant (i.e., indicates that one of two subjects is recombinant), brackets are used to denote markers where phase is uncertain. Subjects with filled symbols are affected, those with clear symbols are unaffected, those shaded are of unknown status.