for markers in the Velinov, Milen plasia had been raised by Michael This locus heterogeneity segregated mutation findings type, while family A the cases D4S227, Mb-D4S227-0-7 D4S43 D4S43 and suggested distances between 4p.35 the haplotype. the phenotype families presented Mb-D4S227-0-7, D4S43 and D4S3 and therefore FGFR3 mutation is present in achondroplasia. Am J Med Genet 1995;55:135-41. Mullis PE, Patel MS, Brickell PM, Hindmarsh PC, Brook CGD. growth characteristics and response to growth hormone therapy in patients with hypochondroplasia: genetic linkage of the insulin-like growth factor 1 gene at chromosome 12q23 to the disease in a subgroup of these patients. Clin Endocrinol 1991; 34:205-7.

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

We recently reported the first example of maternal uniparental disomy for chromosome 13.1 This was found in a phenotypically normal male who inherited a t(13;13) in abalanced 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 or paternally imprinted genes on chromosome 13. Further analysis of the inheritance of this translocation in this family was not initially undertaken because the son’s grandfather was not available for study. However, by recent analysis of the grandmother’s DNA we found that there had been no transmission of two informative dinucleotide chromosome 13 alleles to her daughter (figure). By inference, the t(13;13) must have been transmitted by the grandfather either from a pre-existing constitutional or de novo translocation.

The first example of paternal UPD of chromosome 13 allows us to extend our original suggestion to conclude that there is no paternal or maternal imprinting of genes on chromosome 13.

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

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