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Erratum

Due to a technical failure a portion of the text under 'Results' was omitted in the paper 'Tetrasomy 18p de novo: Parental Origin and Different Mechanisms of Formation' by Bugge et al., Eur J Hum Genet 1996;4:160–167. Printed below is the complete text for 'Results'. We apologize for any inconvenience this may have caused.

Tetrasomy 18p de novo: Parental Origin and Different Mechanisms of Formation

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Results

Parental Age Distribution

Range for mothers 24–39 years, mean maternal age 29 years; range for fathers 25–45 years, mean paternal age 32.1 years (table 1).

Chromosome analysis revealed 47 chromosomes in all 9 cases; the size and banding pattern of the extra small metacentric chromosome were compatible with an isochromosome of the short arm of chromosome 18. All cases were nonmosaic. All parental karyotypes were normal.

In situ hybridization with a chromosome 18 centromere-specific probe showed a monocentromeric signal on the marker chromosome in all cases. FISH using an 18p-specific library showed labeling of the short arms of chromosomes 18 and the whole of i(18p). No labeling of the marker chromosome was observed after FISH with an 18q-specific library (fig. 1).

DNA analysis indicated that all nine i(18p) chromosomes were of maternal origin (table 2, fig. 2). A uniparental origin of the normal chromosomes 18 could be excluded. In 6 cases (cases 1–6), the 18p markers showed that the maternal heterozygous alleles were reduced to homozygosity in the proband in the pericentromeric region, while they remained heterozygous (nonreduced) in the telomeric region. In cases 7 and 8, all informative markers in the proband were nonreduced and in case 9, 6 informative markers showed reduction to homozygosity for maternal alleles (table 2).