

Transmission of Cri-du-Chat Syndrome from a Normal Paternal Chromosome Translocation Carrier

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ABSTRACT A four-year-old girl with facial dysmorphism, microcephaly and global developmental delay was brought to the department of pediatrics for assessment. The genetic evaluation by conventional G banding analysis of somatic chromosomes identified an apparently balanced translocation, interpreted as t (5; 19) (p13.1; q14). The objective of this report was to assess the clinical manifestation of this chromosomal rearrangement and the possible familial implications. Chromosome analysis of the phenotypically normal patient's father showed an identical abnormal banding pattern for chromosomes 5 and 19. It was concluded that while the proband appeared to have inherited the chromosomal rearrangement from the father, the abnormal phenotype was possibly due to de novo imbalances at one of the breakpoints caused during genetic recombination events. This case report highlights the fact that for families with children with congenital abnormalities, an early diagnosis is important for providing personalized diagnostic and prognostic evaluation and also for genetic counseling on future reproductive risks.