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Characterization of Inversions as a Type of Structural Chromosome Aberration

B. Muss and G. Schwanitz*

Institute of Human Genetics, University of Bonn, Wilhelmstrasse 31, D-53111 Bonn, Germany

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ABSTRACT Inversions comprise approximately 10% of structural aberrations, with pericentric inversions clearly outnumbering paracentric rearrangements (66% as compared to 34%), due in part to diagnostic difficulties in the latter group. At 11%, chromosome 2 displays the highest recombination frequency for euchromatic pericentric inversions, while chromosomes 3 and 7 are most often involved in paracentric inversions (16% and 19%, respectively). Inversions of the constitutive heterochromatin are far more frequent than those involving the euchromatin, totalling 20% for only 6 chromosomes. Chromosome 4 demonstrates the highest frequency of pericentric inversions (15%). The genetic risks for inversion carriers differ significantly depending on the chromosome involved and on the specific inverted region, ranging from less than 1% to a maximum of 30%. Cryptic inversions resulting in a microdeletion in chromosome 7q11.23 have been demonstrated to be causative in 33% of Williams-Beuren cases, apparently due to an abnormal course of meiosis I. Molecular cytogenetic investigations of the parents of carriers of cryptic chromosome aberrations may be expected to detect a higher frequency of paracentric inversions than has thus far been demonstrated.