

Investigations in Probands with Rearrangements in the Acrocentric Chromosomes

L. Kalz and G. Schwanitz

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

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ABSTRACT Chromosome investigations in 25,571 cases revealed 432 patients with structural changes in the acrocentrics. These could be attributed to 8 cases of heterochromatin translocations with the Y-chromosome, 135 cases of rearrangements in heterochromatic regions of the short arms of the acrocentrics, and 289 cases with different types of exchanges and breakpoints in the euchromatin. Altogether, breakpoints in 547 acrocentrics were analysed. A specific intrachromosomal distribution of breakpoint maxima and minima was delineated for the 5 acrocentrics with individual characteristics regarding the 3 categories of rearrangements such as: Yq12 translocations, Robertsonian translocations as well as heterochromatic marker chromosomes, and rearrangements in the euchromatin. In addition, polymorphisms of the heterochromatin in the short regions of the acrocentrics were analysed. The origin of the 13 different aberration types documented showed a chromosome-specific pattern.