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Chromosome Mosaicism in Patients with Normal and Abnormal Y-Chromosome

R. Schellberg¹, G. Schwanitz¹, H.-U. Schweikert² and R. Raff¹

1. Institute for Human Genetics, University of Bonn, Germany

2. Medical Policlinic, University of Bonn, Germany

KEY WORDS Gonosomal mosaicism; derivative Y-chromosome; genotype-phenotype correlation

ABSTRACT Gonosomal mosaicism with a Y-chromosome present in at least one cell system was investigated in 29 patients. In 15 cases the Y-chromosome was structurally normal, in 14 cases different structural aberrations were analysed. 17 patients were phenotypically male, 7 were female and 5 showed intersexual external genitals. The patients age at the time of chromosome investigation ranged from the prenatal period up to the age of 38 years. The most frequent clinical findings were: growth retardation, abnormalities of the external genitals, kidney malformations and different types of heart defects. Cytogenetic investigations combined metaphase and interphase analyses with FISH (*DNA probes: wcp X and Y; CEP X and Y; Yph 3.4*). Between 2 and 5 cell systems per patient were analysed with special emphasis on their origin from different blastodermic layers. An unequal distribution of the mosaics could be demonstrated comparing the different cell systems of a proband, with no preferential combination of karyotype and cell type. The majority of tissues had two different karyotypes in probands with normal or abnormal Y-chromosome. The highest number of cell lines in one cell system was four. Patients with a ring Y-chromosome revealed the highest karyotype instability with segregation abnormalities being combined with partial amplifications of the ring. There were no age-dependent karyotype changes in our investigation group.

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