

The results of the investigated cases presented in this Atlas with structural chromosome aberrations are based on the current given methodological options to analyse the different types of abnormalities. It was particularly emphasized which spectrum should be chosen when combining the different techniques for the best way of diagnosis in the single case, for example direct preparation of cells and mitoses, short or long time cell culture, FISH, analysis of interphases, microarray, DNA-sequencing. In many cases family investigations and analysis of the pedigree should be performed additionally.

Generally it has to be taken into account, that the development of new and improved investigation methods is forthcoming. Thus, by improvement of diagnostic possibilities new fields of investigation will arise, and special groups of patients with cytogenetic analyses can be re-analysed under new research questions.

Thus, for example, constitutional aberrations of chromosome structure such as complex chromosome rearrangements (CCR) and abnormalities of tumor cells can be classified as anomalies originating from chromothripsis. Here additionally the relevance of the combination of cytogenetic and molecular genetic techniques is significantly enlightened.

Of special importance is the increasing number of analyses in the field of epigenetics. Functional aberrations caused by uniparental disomy (UPD) or by the inactivation of genes caused by changes in neighbourhood (position effects) are of increasing relevance especially in prenatal diagnosis.

Finally, maxims of ethical principles are playing an increasing role in the field of applied Cytogenetic when analysing inherited chromosome aberrations. It must be clearly defined to whom prenatal findings may be communicated (predictive diagnostics), may analyses be performed in children and adolescents (not legally able to consent). As to these problems legal regulation is increasing in various countries, but the requirements are not of uniform format.

For all human geneticists working in the field of applied Cytogenetics or on basic research it is already nowadays of essential relevance to develop a network of cooperation between university institutes and private laboratories to enable a regular exchange of experience, to establish regular specific congresses, seminars and workshops and to set up common projects.