PWG: CANCER CYTOGENETICS, SOLID TUMOR STUDIES.

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PWG: CYTOGENETICS OF GERM CELLS AND EMBRYOS. PRE-IMPLANTATION CYTOGENETIC STUDIES.

After the Dublin conference, Joy Delhanty retired as a co-ordinator of the PWG on Cytogenetics and germ cells and embryos, pre-implantation cytogenetic studies. The board of the E.C.A. wishes to express its gratitude to Professor Delhanty for many years of successful co-ordination of the PWG and the organization of many important meetings at the ECCs that found a broad interest in the cytogenetics community. We wish her all the best for the future.

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The name of the PWG **Cytogenetics and Society** has been changed to **Quality issues and training in Cytogenetics** for a better representation of the activities of the group. To cover ethical aspects, the E.C.A. Committee has decided to establish a **Committee on Ethical Aspects**.

PWG: QUALITY ISSUES AND TRAINING IN CYTOGENETICS.

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Book Review

Thomas Liehr: Benign & Pathological Chromosomal Imbalances. Microscopic and submicroscopic copy number variations (CNVs) in Genetics and Counselling.

(Elsevier, 236 pp, 2013 Print Book ISBN 9780124046313, eBook ISBN 9780124046849)

Barbara Fritz, Marburg

Evidence of human genetic variation began to accumulate with our ability to see individual chromosomes under the microscope. The development of banding techniques of the chromosomes revealed a much greater variety of heteromorphisms than had previously been suspected. Even though modern cytogenetics is already over six decades old, it is remarkable that an accurate and complete documentation of the normal chromosome variants has not yet been accomplished. Therefore, the study of benign and pathological variants should be viewed as an ongoing and dynamic process, which is greatly influenced by changing technologies, for the characterization of the human genome. In recent years significant advances have been made in mapping and characterizing structural variations in the human genome with the help of molecular profiling methods such as FISH, aCGH, and NGS. Despite this progress, our understanding of

genomic variants is still very limited and distinguishing normal chromosome variants from clinically relevant variations is one of the main tasks for all clinical (cyto)genetic laboratories.

Benign & Pathological Chromosomal Imbalances is a useful reference handbook for clinical geneticists and molecular cytogeneticists. It systematically describes how to assess the clinical relevance of cytogenetically visible copy number variants (CG-CNV) using cytogenetic techniques to analyze heterochromatic or euchromatic DNA variants. The book takes two perspectives: diagnosis and research. It is a very useful source of reference for the scientist in the cytogenetics laboratory as well as for the genetic counsellor who receives a cytogenetic report and communicates the result to the patient. It is packed with practical advice on how best to investigate heterochromatic, centromeric and euchromatic variations including unbalanced chromosomal abnormalities (UBCAs), euchromatic variants (EVs) and supernumerary marker chromosomes (sSMCs). In addition, this book will become an important resource for research, for studying the structure of the human genome, and for improving our understanding of how genomic variations contribute to subtle phenotypic variations, disease development and human diversity.

In this age of exponential increase in genomic information, the seven chapters of this book offer a comprehensive view of human chromosome heteromorphisms. As outlined in the first chapter the book focuses on microscopically visible abnormalities, but submicroscopic variants (MG-CNVs) are also covered where appropriate. Fragile sites, a special class of chromosomal variants, however, have not been incorporated.
The second chapter is a well written synopsis of experimental data and includes the molecular characterisation of some classes of repeat sequences in heteromorphic regions. It also makes an attempt at the difficult task of defining the 'normal' or 'average variant'. In the third and fourth chapters the author describes the inheritance and the main mechanism of formation of cytogenetically visible copy number variations, and in the sixth gives the cytogenetic and molecular approaches for genetic diagnostics and patient counselling.
Chapter 5, the heart of this book, encompasses a comprehensive up-to-date review of all published CG-CNVs and includes some unpublished data coming from Thomas Liehr's own laboratory. In 83 pages, the author has succeeded in reviewing, in a very concise synthesis, all the important data about genetic knowledge of heterochromatic and euchromatic CG-CNVs for each chromosome. Heterochromatic CG-CNVs include acrocentric short arm variants, pericentric regions, autosomal noncentromeric heterochromatin and Y-chromosome-derived heterochromatin; euchromatic CG-CNVs include UBCA, sSSC, EVs and gonosomal derived euchromatin. Each section gives a brief summary of the heteromorphisms for each chromosome, the basis of the cytogenetic methodology to help to decide which molecular cytogenetics tools to deploy, and the nomenclature according to ISCN. Diagrams illustrate the underlying mechanism of the more common heteromorphisms. Heteromorphisms that may be clinically relevant or those that can be mistaken for harmless variants are discussed in detail. With more than 300 references this chapter provides a rich, valuable, up-to-date synopsis of this important field.
The book is supplemented by a side-by-side comparison of copy number variants with their recently identified submicroscopic forms in the appendix. A final section illustrates FISH variants (colour plate 1-3: variants of the acrocentric short arms, colour plate 4: variant of the centromeric regions).

Finally, I would like to quote John C.K. Barber, who said in the preface: "This book is a comprehensive summary of our state of knowledge at a time of transition when the microscopically visible cytogenetic era is becoming the submicroscopic copy number era …and much of the information within the book is not easily accessible elsewhere, even with the help of search engines, web browsers, and publication archives." For this reason this book is, beside the "Human Chromosome Variation" by Wyandt and Tonk, indispensable for the interpretation and documentation of human chromosome variants. It is certainly an up-to-date account of this fast evolving field for everyone who needs background or specialized knowledge for the interpretation of chromosomal heteromophisms.

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