



17th Goldrain Course in Clinical Cytogenetics



August 20-26, 2024



DIRECTORS

A. Schinzel (Zurich, Switzerland); **M. Rocchi** (Bari, Italy)

PROGRAMME COMMITTEE

A. Schinzel, M. Rocchi, J-M. Dupont, K. Miller, K. Madan, A. Baumer, E. Klopocki,

FACULTY

D. Bartholdi (Berne, Switzerland), A. Baumer (Zurich, Switzerland), P. Benn (Farmington CT, U.S.A.), J.M. Dupont (Paris, France), E. Errichiello (Pavia, Italy), E. Klopocki (Würzburg, Germany), K. Madan (Leiden, The Netherlands), K. Miller (Hannover, Germany), R. Pfundt (Nijmegen, The Netherlands), M. Rocchi (Bari, Italy), G. van Buggenhout (Leuven, Belgium), M. Vismara (Rome, Italy), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

LOCATION

Goldrain Castle, Goldrain, South Tyrol, Italy

Website of the course: www.biologia.uniba.it/SEC/

COURSE DESCRIPTION

The course is focused on phenotypic findings, mechanisms of origin and transmission, correlations of clinical patterns with chromosomal imbalance and modern ways of diagnosis of the latter. Special attention is paid to an understanding how deletions and/or duplications of chromosomal segments cause developmental defects. The course also addresses the optimal application of the diagnostic possibilities, both pre- and postnatally and including molecular cytogenetic methods for a precise determination of segmental aneuploidy.

TOPICS

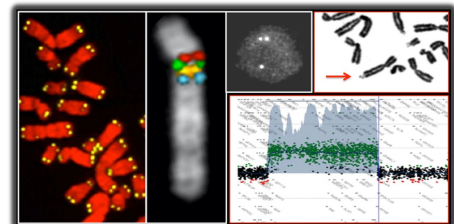
Dysmorphic findings in chromosome aberrations: formation and interpretation – The adult and elderly patient with a chromosome aberration – Follow-up studies in patients with chromosome aberrations – Clinical findings associated with chromosome aberrations – Microdeletion syndromes: clinical pictures – prenatal cytogenetic diagnosis – Mosaics and chimeras – imprinting and uniparental disomy - Epidemiology of chromosome aberrations – Chromosome aberrations in spontaneous abortions and stillborns – Harmless chromosome aberrations – Risk assessment in structural chromosome aberrations Extra small supernumerary chromosomes – Genomic variation: a continuum from SNPs to chromosome aneuploidy – Pre-implantation cytogenetic diagnosis – Ultrasound findings indicative of chromosome aberrations – Ethical issues in the context of cytogenetic diagnosis – Non-invasive prenatal cytogenetic diagnosis. ISCN - Practical exercises in cytogenetic nomenclature – Accreditation of cytogenetic laboratories - Accreditation of cytogenetic laboratories – Optimal use of available techniques in clinical cytogenetics – NGS – SNP arrays and Array-CGH: principles, technical aspects; evaluation of the results – MLPA - QF-PCR - FISH techniques and their interpretation – Optical genome mapping – Introduction and practical exercises with database for phenotypic and variant interpretation - Students presentation of cases with difficult-to-interpret chromosome aberrations. Introduction to modern genetic editing techniques. - Practical exercises will be offered with the ISCN system for chromosome aberrations and with cytogenetic, genomic, and phenotypic databases.

Students will have the opportunity to present their own observations and cytogenetic findings which are difficult to interpret, and

They will also have the opportunity to perform a test at the end of the course.

Scholarships, in the form of reduced fee, will be granted to students who make payments from their personal accounts (not through the institution).

For further questions please write directly to
Albert Schinzel at schinzel@medgen.uzh.ch or to
Mariano Rocchi at mariano.rocchi@uniba.it



Full fee is Euro: **1.400-1750**, depending on the accommodation. It includes tuition, course material, free access to internet, accommodation for 8 nights, all meals, coffee breaks and a ½ day excursion.