11th Goldrain Course in
Clinical Cytogenetics
August 27 to September 03, 2016

SCHLOSS GOLDRAIN
Bildung und Kultur · Formazione e Cultura

LOCATION
Goldrain Castle, Goldrain, South Tyrol, Italy (www.schloss-goldrain.it)

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 (cyto)genetic methods for a precise determination of segmental aneuploidy.

TOPICS
Basic cytogenetics - lab & clinic
Chromosome aberrations and epidemiology – Cytogenetic polymorphisms - Dysmorphic findings in chromosome aberrations – Small supernumerary and ring chromosomes – Chromosome aberrations in spontaneous abortions and stillborns – etc.

Cytogenetic techniques & reporting
MLPA, QF-PCR and FISH – Genome wide SNP-base array analysis: principles, analysis and interpretation – Use of genomic databases – Reporting array results – EQA in clinical cytogenetics – Position effects and effects of CNVs containing no genes – etc.

Prenatal diagnosis
Prenatal screening using serum and ultrasound markers –Prenatal cytogenetic diagnosis – Pre-implantation genetic diagnosis – Non-invasive prenatal aneuploidy testing – etc.

Mechanisms & counselling
Karyotype-phenotype correlations – Genetic counselling – Mechanisms of origin – Somatic mosaics and disease – Risk assessment in structural chromosome aberrations – etc.

Complex clinical cytogenetics
Uniparental disomy and imprinting – Microdeletion syndromes – Incomplete penetrance – Ethical issues – Diagnostic decision and dilemmas – Strategies after the detection of CNVs

Sex chromosomes & disorders of sex development
The clinical effect of sex chromosomal aneuploidy – Sex chromosomes: mosaics, AZF deletions, XIST and SRY – DSD: XX and XY reversal with normal SRY status

New techniques
Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome – An introduction on CRIPSR/Cas and its future opportunities

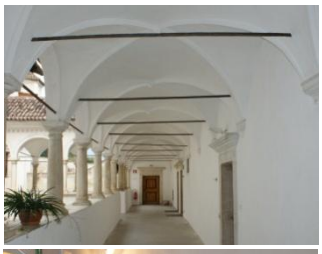
Practical exercises
ISCN – ECARUCA – report writing – use internet sources and databases – segregation of chromosomal translocations – student presentations (optional) – test (optional)

DIRECTOR
A. Schinzel (Zurich, Switzerland)

FACULTY
D. Bartholdi (Berne, Switzerland), A. Baumer (Zurich, Switzerland), P. Benn (Farmington CT, U.S.A.), R. Ciccone (Pavia, Italy), R. Hastings (Oxford, U.K.),E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), N. de Leeuw (Nijmegen, Netherlands), K. Miller (Hannover, Germany), E. Syk-Lundberg (Stockholm, Sweden), G. van Buggenhout (Leuven, Belgium), C. van Ravenswaaij (Groningen, Netherlands), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

For further questions please write directly to Albert Schinzel at schinzel@medgen.uzh.ch.





Full fee is Euro 1300 for a single room or Euro 1150 (VAT included) in a 2-bed-room. It includes tuition, course material, free access to internet during the course, accommodation for 7 nights, all meals, beverages during the breaks and a ½ day excursion.