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## 3rd Goldrain Course in Clinical Cytogenetics August 31 – September 3, 2008

### LOCATION

Goldrain Castle, Goldrain, South Tyrol, Italy  
Website of the venue: [www.schloss-goldrain.it](http://www.schloss-goldrain.it)

### COURSE DESCRIPTION

The course is focused on phenotypic findings, mechanisms of origin and transmission, and correlations of clinical patterns with the chromosomal imbalance. 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 including molecular cytogenetic methods for a precise determination of segmental aneuploidy.

### TOPICS

Chromosome aberrations in spontaneous abortions and stillbirths; FISH techniques and their application; Chromosomal variants and fragile sites; The "harmless chromosome aberration"; Mosaics and chimeras; Complex chromosome rearrangements; Prenatal cytogenetics; Sex chromosome aberrations; Clinical findings associated with chromosome aberrations; Dysmorphic and auxiologic measurements and standards for the clinical examination; The adult and elderly patient with a chromosome aberration; Follow-up studies in patients with chromosome aberrations; Risk assessment in structural chromosome aberrations; The Zurich-ECARUCA database; Mechanisms of formation and origin of chromosome aberrations; Why are certain structural aberrations more frequent than others?; Contiguous gene syndromes; Genomic imprinting; Uniparental disomy; Array-CGH; Strategies for gene hunting in cases with chromosome aberrations and monogenic disorders; MLPA; Karyotype-phenotype correlations.

Practical exercises will be offered with the ISCN system for chromosome aberrations and with the cytogenetic database. Students will have the opportunity to present their own observations and cytogenetics findings which are difficult to interpret. The students will have the opportunity to perform a test at the end of the course.

### DIRECTOR

A. Schinzel (Zurich, Switzerland)

### FACULTY

F. Barber (Salisbury, UK), A. Baumer (Zurich, Switzerland), O. Birk (Beer Sheva, Israel), N. de Leeuw (Nijmegen, The Netherlands), K. Madan (Leiden, The Netherlands), K. Miller (Hannover, Germany), B. Steiner (Zurich, Switzerland), G. van Buggenhout (Leuven, Belgium), C. van Ravenswaaij (Groningen, The Netherlands), J. Vermeesch (Leuven, Belgium), O. Zuffardi (Pavia, Italy)

For further questions please consult the program via <http://www.biologia.uniba.it/eca/> or write directly to Albert Schinzel at [schinzel@medgen.uzh.ch](mailto:schinzel@medgen.uzh.ch)



*Full fee is Euro 1000 for a single room or Euro 850 (VAT included) in a 2-bed-room. It includes tuition, course material, free access to internet during the course, accommodation for 5 nights, all meals, beverages during the breaks and a ½ day excursion.*

