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| https://www.eshg.org/fileadmin/template/images/ESHG-Banner-Web.png | **uzh_logo_d_pos_grau_1mmuzh_logo_d_pos_grau_1mm** | ***E.C.A.***  ***European Cytogeneticists Association*** |

**Clinical Cytogenetics**

**Aug. 20th to 28th, 2022**

**Goldrain, South Tyrol, Italy**

**DIRECTOR:**

A. Schinzel (Zurich, Switzerland)

**FACULTY:**

D. Bartholdi (Bern, Switzerland), A. Baumer (Zürich, Switzerland), P. Benn (Farmington CT, U.S.A.), Jean-Michel Dupont (Paris, France), L. Girardi (Vicenza, Italy), E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), N.E. Kurtas (Florence, Italy), K. Madan (Leiden, the Netherlands), K. Miller (Hannover, Germany), Rolph Pfundt (Nijmegen, The Netherlands), Mariano Rocchi (Bari, Italy), G. van Buggenhout (Leuven, Belgium), M. Vismara (Roma, Italy), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

**PROVISIONAL PROGRAM:**

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| **Saturday, August 20th Arrival at Goldrain** |

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| 19:00-20:30 |  | | | *Dinner* |  | | | |
| 20:30-21:30 |  | | | **A. Schinzel** | Introduction and organizational comments | | | |
| 21:30- |  | | | Get together | Steinkeller | | | |
| **Sunday, August 21st Basic and clinical cytogenetics** | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | |
| 09:00–09:35  09:40-10:15 | *1*  *2* | | | **A. Schinzel**  **K. Miller** | History of clinical cytogenetics  Basic concepts of chromosome aberrations and epidemiology | | |
| 10:20-10:55 | *3* | | | **D. Bartholdi** | Genetic counselling for chromosome aberrations | | |
| 11:00-11:20 |  | | | *Coffee Break* |  | | |
| 11:25-12:00 | *4* | | | **D.Bartholdi** | Clinical dysmorphologic examination in cytogenetic disorders. I | | |
| 12:05-12:40 | *5* | | | **D. Bartholdi** | Clinical dysmorphologic examination in cytogenetic disorders. II | | |
| 12:45-14:00 |  | | | *Lunch Break* |  | | |
| ***Afternoon Session: 14:00 – 18:30* Clinical cytogenetics** | | | | | | | |
| 14:00-14:35 | *6* | | | **A. Schinzel** | Dysmorphic findings in chromosome aberrations: formation and interpretation | | |
| 14:40-15:30 | *W1*  *W2* | | | **D. Bartholdi**  **M.Vismara** | Clinical issues: difficult situations  Nomenclature of facial features in clinical dysmorphology | | |
| 15:30-15:50 |  | | | *Coffee Break* |  | | |
| 15:50-16:25 | 7 | | | **K. Miller** | Markers and ring chromosomes | | |
| 16:30-17:15 | *W1*  *W2* | | | **D. Bartholdi**  **M.Vismara** | Clinical issues: difficult situations  Nomenclature of facial features in clinical dysmorphology | | |
| 17:15-17:50 | *8* | | | **K. Miller** | Cytogenetics of spontaneous abortions and stillbirths | | |
| 17:55-18:30 | *9* | | | **E. Klopocki** | An introduction to CRISPR/Cas9 and its future possibilities | | |
| 19:00-20:00 |  | | | *Dinner* |  | | |
| 21:00- |  | | | Get together | Steinkeller | | |
| **Monday, August 22nd Cytogenetic techniques** | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | |
| 09:00–09:35 | *10* | | | **K. Miller,**  **E. Klopocki** | ISCN, standard and molecular | | |
| 09:40-10:15 | *W3*  *W4* | | | **K. Miller**  **P. Benn/**  **J.M.Dupont** | Segregation of chromosomal translocations: practical exercises (half group, rotate with W4)  Appropriate management in prenatal screening and diagnosis (parallel with W3) | | |
| 10:20-10:55 | *11* | | | **J.M. Dupont** | FISH techniques and their application | | |
| 11:00-11:20 |  | | | *Coffee Break* |  | | |
| 11:25-12:00 | *W3*  *W4* | | | **K. Miller**  **P. Benn/**  **J.M.Dupont** | Segregation of chromosomal translocations: practical exercises (half group, rotate with W4)  Appropriate management in prenatal screening and diagnosis (parallel with W3) | | |
| 12:05-14:00 |  | | | *Lunch Break* |  | | |
| ***Afternoon Session: 14:00 – 18:40* Continuation and practical exercises** | | | | | | | |
| 14:00-14:35 | *12* | | | **E. Klopocki** | Array: principles, technical aspects, different platforms | | |
| 14:40-15:15 | *13* | | | **R. Pfundt** | SNP array analysis and interpretation in constitutional genome diagnostics | | |
| 15:15-15:50 | *14* | | | **E. Klopocki** | Position effects and effects of CNVs containing no genes | | |
| 15.55-16.30 | *15* | | | **N. E. Kurtas** | Congenital chromotripsis rearrangements | | |
| 16:35-16:50 |  | | | *Coffee Break* |  | | |
| 16:50-17:25 | *W5* | | | **K. Miller** | Practical exercises in report writing. Parallel with W6 | | |
|  | *W6* | | | **E. Klopocki** | Databases for the interpretation of array results for beginners: hands-on in computer room. Parallel with W5 | | |
| 17:30-18:05 | *16* | | | **P. Benn** | Origin of trisomy | | |
| 18:10-18:45 | *W5* | | | **K. Miller** | Practical exercises in report writing. Parallel with W6 | | |
|  | *W6* | | | **E. Klopocki** | Databases for the interpretation of array results for beginners: hands-on in computer room. Parallel with W5 | | |
| 19:00-20:00 |  | | | *Dinner* |  | | |
| 21:00- |  | | | Get together | Steinkeller | | |
| **Tuesday, August 23rd Prenatal diagnosis** | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | |
| 09:00–09:35 | *17* | | | **P. Benn** | Traditional approaches for the detection of aneuploidy in prenatal diagnosis: screening using serum and ultrasound markers | | |
| 09:40-10:15 | *18* | | | **J.M.Dupont** | Prenatal cytogenetic diagnosis I: mosaics in PND | | |
| 10:20-10:55 | *19* | | | **J.M.Dupont** | Prenatal cytogenetic diagnosis II: RAD (rapid aneuploidy detection) and CMA (chromosomal microarray detection) | | |
| 11:00-11:20 |  | | | *Coffee Break* |  | | |
| 11:25-12:00 | *20* | | | **P. Benn** | Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I | | |
| 12:05-12:40 | *21* | | | **P. Benn** | Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects II | | |
| 12:45-14:00 |  | | | *Lunch Break* |  | | |
| ***Afternoon Session: 14:10 – 18:30 EXCURSION*** | | | | | | | |
|  | | | | | | | |
| 19:00-20:00 |  | | | *Dinner* |  | | |
| 21:00-22.00 |  | | | Lecturers | Case presentations | | |
| 22.00- |  | | | Get together | Steinkeller | | |
| **Wednesday, August 24th Prenatal Diagnosis, miscellaneous** | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | |
| 09:00–09:45 | *22* | | | **L. Girardi** | Pre-implantation genetic diagnosis | | |
| 09:50-10:25 | *23* | | | **J. Wisser** | Prenatal ultrasound findings indicative for chromosome aberrations | | |
| 10:30-11:05 | *24* | | | **A. Baumer** | MLPA and QF-PCR | | |
| 11:10-11:30 |  | | | *Coffee Break* |  | | |
| 11:30-12:05 | *25* | | | **K. Madan** | Cytogenetic polymorphisms | | |
| 12:10-12:45 | *26* | | | **R. Pfundt** | Incidental findings during genetic testing | | |
| 12:45-14:00 |  | | | *Lunch Break* |  | | |
| ***Afternoon Session: 14:00 – 18:30 M*iscellaneous** | | | | | | | |
| 14:00-14:35 | *27* | | | **K. Madan** | Complex chromosome rearrangements | | |
| 14:40-15:15 | *28* | | | **A. Baumer** | Uniparental disomy (UPD) and imprinting | | |
| 15:20-15:55 | *29* | | | **K. Madan** | Inversions and insertions | | |
| 15:55-16:15 |  | | | *Coffee Break* |  | | |
| 16.15-16.50 | *30* | | | **O. Zuffardi** | The hidden link between numerical and structural variants: the incessant fight against adverse genomes | | |
| 16:50-17:30 | *W7* | | | **A. Baumer** | Work-up of cases: strategies after the detection of CNVs. Parallel with W8 | | |
|  | *W8* | | | **E. Klopocki** | Databases for the interpretation of array results for advances users: hands-on in computer room. Parallel with W7 | | |
| 17:35-18:10 | *31* | | | **K. Miller** | Quality assessment of cytogenetic laboratories | | |
| 18:15-18:55 | *W8* | | | **E. Klopocki** | Databases for the interpretation of array results for advances users: hands-on in computer room. Parallel with W7 | | |
|  | *W7* | | | **A. Baumer** | Work-up of cases: strategies after the detection of CNVs. Parallel with W8 | | |
| 19:00-20:00 |  | | | *Dinner* |  | | |
| 21:00-21. | *32* | | | **K. Madan** | Sex chromosomes | | |
| 21.40- |  | | | Get together | Steinkeller | | |
| **Thursday, August 25th Clinical cytogenetics** | | | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | | | |
| 09:00–09:35 | *33*  *34* | | | **G. v. Buggenhout**  **M. Rocchi** | | | Common chromosome disorders and follow-up I  200 years after the birth of Gregor Mendel | | |
| 10:20-10:55 | *35* | | | **O.Zuffardi** | | | Variants acquired from embryogenesis to aging: Clonality and consequences on health and disease. I | | |
| 11:00-11:20 |  | | | *Coffee Break* | | |  | | |
| 11:25-12:00 | *36* | | | **G. v. Buggenhout** | | | Common chromosome disorders and follow-up II | | |
| 12:05-12:40 | *37* | | | **G. v. Buggenhout** | | | Microdeletion syndromes | | |
| 12:45-14:10 |  | | | *Lunch Break* | | |  | | |
| ***Afternoon Session: 14:15 – 18:40* Sex chromosome aberration; miscellaneous** | | | | | | | | | |
| 14:10-14:45 | | *38* | | **G.v. Buggenhout** | | The clinical effect of sex chromosomal aneuploidy | | | |
| 14:50-15:25 | | *39* | | **O. Zuffardi** | | Variants acquired from embryogenesis to aging:  Clonality and consequences on health and disease. II | | | |
| 15:30-16:05 | | *40* | | **K. Madan** | | Disorders of sex development | | | |
| 16:05-16:25 | |  | | *Coffee Break* | |  | | | |
| 16:25-17:10 | | *41* | | **O. Zuffardi** | | Semi-dominant and recessive inheritance of X-linked variants | | | |
| 17:10-17:55 | | *42* | | **R. Pfundt** | | Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome | | | |
| 18:00-18:40 | | *43* | | **O. Zuffardi** | | The future of cytogenetics | | | |
| 18:45-20:00 | |  | | *Dinner* | |  | | | |
| 20.30- | |  | | Get together | | Steinkeller | | | |
| **Friday, August 26th Miscellaneous** | | | | | | | |
| ***Morning Session: 09:00 – 12:40* New techniques** | | | | | | | |
| 09:00-09:50 | | | *44* | **K. Madan** | Natural human chimeras | | |
| 09:55-10:30 | | | *45* | **M. Rocchi** | Centromere repositioning in evolution and in human population. | | |
| 10:35-11:10 | | | *46* | **J. Wisser/A. Schinzel** | Twins at prenatal diagnosis | | |
| 11:10-11:30 | | |  | *Coffee Break* |  | | |
| 11:30-12:40 | | | *47* | **Students**  **N. Kurtas** | Student presentations  Short tandem repeats in developmental disorders | | |
| 12:45-14:10 | | |  | *Lunch Break* |  | | |
| ***Afternoon Session: 14:15 – 18:45* Self-study or sight seing** | | | | | | | |
| 19:00-20:00 | | |  | *Dinner* |  | | |
| 20:15- | | |  | Get together | Steinkeller | | |
| **Saturday, August 27th Past and future of clinical cytogenetics** | | | | | | | |
| ***Morning Session: 09:00 – 12:40*** | | | | | | | |
| 09:00–09:40 | *W9*  *W10* | | | **M. Vismara**  **N.E.Kurtas** | Databases for clinical genetics (parall. with W10)  Breakpoint analysis of complex structural variants (parallel with W9) | | |
| 09:45-10:20 | *W9*  *W10* | | | **M. Vismara**  **N.E.Kurtas** | Databases for clinical genetics (parall. with W10)  Breakpoint analysis of complex structural variants (parallel with W9) | | |
| 10:25-11:00 | *48* | | | **T. Krones** | Ethical issues | | |
| 11:05-11:20 |  | | | *Coffee Break* |  | | |
| 11:20-11:55 |  | | | **Lecturers** | Presentation of cases raising ethical questions | | |
| 12:00-12:35 | *49a*  *49b* | | | **A.Schinzel**  **T. Krones** | Paradigm shifts, unexpected findings and use of markers in cytogenetics\*  Ethical Issues: difficult situations | | |
| 12:40-14:00 |  | | | *Lunch Break* |  | | |
| ***Afternoon Session: 14:00 – 18:45* Evaluation; examination; castle tour** | | | | | | | |
| 14:00-14:35  14:50-16:10 | *50* | | | **Students**  **Students** | Student presentations  EXAMINATION | | |
| 16:10-16:30 |  | | | *Coffee Break* |  | | |
| 16:30-17:10 |  | | | **Faculty / Students** | Presentation of the correct answers to exam. | | |
| 17:10-17:50 |  | | | **Students / Faculty** | Discussion of the program | | |
| 18:00-18:40 |  | | |  | Guided visit to the castle | | |
| 19:30-21:00 |  | | | *Farewell dinner* | Steinkeller or inner court of the castle | | |

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| **Sunday, September 28th Goodbye Goldrain** |

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| 08:00-09:00 |  | *Breakfast* |  |
| 09:00- |  | *Checkout and departure* |  |