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EUROPEAN CYTOGENETICISTS ASSOCIATION

E.C.A. NEWS LETTER

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E.C.A. Newsletter

The E.C.A. Newsletter is the official organ published by the European Cytogeneticists Association (E.C.A.). For all contributions to and publications in the Newsletter, please contact the editor.

Editor of the E.C.A. Newsletter:

Konstantin MILLER
Institute of Human Genetics
Hannover Medical School
30623 HANNOVER
GERMANY
Tel.: +49 511 532 35 72 Fax : +49 511 532 6555
E-mail: miller.konstantin@mh-hannover.de

Editorial committee:

J.S. (Pat) HESLOP-HARRISON
Department of Genetics
University of Leicester
LEICESTER LE1 7RH
UK
Tel.: +44 116 252 5079 Fax.: +44 116 252 2791
E-mail: phh4@le.ac.uk

Kamlesh MADAN
Cytogenetics Laboratory
Dept. of Clinical Genetics (Post zone S6P)
Leiden Univ. Medical Center
P.O.Box 9600
2300 RC LEIDEN
THE NETHERLANDS
Tel.: +31 72 51 28 953 Fax : +31 71 52 68 276
E-mail: k.madan@lumc.nl

Mariano ROCCHI
President of E.C.A.
Dip. di Biologia
Campus Universitario
Via Orabona 4
70125 BARI
ITALY
Tel.: +39 080 544 3371
E-mail: mariano.rocchi@uniba.it

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President's Address

Dear Colleagues and Friends,

At the last meeting of the Board of Directors held in Paris I was elected as President of the E.C.A. Having taken the baton from the previous president, Prof. Elisabeth Syk Lundberg, I am very conscious of the responsibilities of this honor, and I will do my best to live up to the task.

Let me present myself. I started my scientific career in the pediatric hospital in Trieste, where I was involved in clinical cytogenetics, postnatal and prenatal. I then moved to the Gaslini Institute in Genoa where I worked in molecular cytogenetics and somatic cell genetics. In 1990 I moved to the University of Bari as a full professor of Genetics. In Bari my scientific interests shifted to the evolution of the karyotypes of primates; my group has been involved in several primate genome projects.

I have been deeply involved in ECA activities as President of the Scientific Programme Committee. Our biennial conference represents a big opportunity for all of us to take a break in our routine or research activities to meet our colleagues in order to share scientific results and to broaden our technical and cultural knowledge in the field of "cytogenomics". I use the term "cytogenomics" because the E.C.A. Board has realized that the scientific and laboratory activities of many of us come under the umbrella of "cytogenomics". This is in recognition of the recent advances that interlink cytogenetics to molecular genomic sciences of humans, non-human animals and plants. Indeed, the 2019 Salzburg E.C.A. conference (July 6-9) will be "on cytogenomics". The E.C.A. website will be open shortly for abstract submission. The draft of the scientific programme will soon be available. Some talks will address the technical advances in the different fields of cytogenomics; others will broaden our understanding of biological phenomena in humans, in non-human animals, and in plants. E.C.A., indeed, harbors these three souls that are closely interlinked and complement each other. Let us consider, from my personal experience, the paradigmatic example of the phenomenon of "centromere repositioning", which was discovered in Old World monkeys. Without the knowledge of the evolution of human chromosomes 14 and 15, which arose from a splitting of a single chromosome in a hominoid ancestor, we could not have fully understood the clustering of human neocentromeres at 15q24-26.

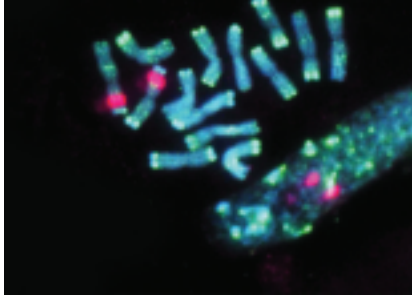
Evolutionary processes shaping animal and plants on the one hand and the cancer progression on the other hand, were apparently far apart. A tumor session in Salzburg has been inspired by studies in recent years showing that the Darwinian rules governing the evolution of species are essentially the same as those that govern cancer progression. Furthermore, the knowledge of these rules is now being exploited to improve the management of cancer therapies.

I hope to see you all in Salzburg, actively involved in poster presentations (that could become selected oral presentations) and in the Working groups (https://www.e-c-a.eu/EN/WORKING-GROUPS.html).

Last, but not least, I would like to take the opportunity to thank the persons behind the E.C.A. activities: Konstantin Miller (factotum secretary), the two vice-presidents Kamlesh Madan and Pat Heslop-Harrison, the treasurer Jean-Michel Dupont, the Scientific Programme Committee, and the entire Board of Directors.

Mariano Rocchi

President

European Cytogeneticists Association

12th EUROPEAN CYTOGENOMICS CONFERENCE

6 - 9 JULY 2019

SALZBURG CONGRESS
SALZBURG, AUSTRIA

www.eca2019.com

The 12th European Cytogenomics Conference Salzburg, 6-9 July 2019

Preliminary Scientific Programme

SATURDAY, 6 July 2019

14:00-17:00 Scientific presentations and discussion meetings of Permanent Working Groups

17:00-17:50 Satellite Symposium

18:00-19:00 **Opening lecture**. Chairs: Mariano Rocchi – Dieter Kotzot

Joris Vermesch: Somatic mosaicism

SUNDAY, 7 July 2019

08:30-10:15 **Plenary session 1 - Recent advances in Cytogenomics**

08:30-09:00 **Thomas Ried**: Genome and Transcriptome Dynamics in Cancer Cells

09:00-09:30 **Michael Speicher**: Liquid biopsy in tumors

09:30-10:15 Selected abstracts

coffee break

10:45-11:45 **Plenary session 2: 50 Years ago: Chromosome banding**

10:45-11:15 **Felix Mitelman**: The end of the Dark Ages

11:15-11:45 **Dario Lupianez**: Chromosome biology

12:00-14:30 Poster session and Satellite Symposia

14:30-15:45 Concurrent Sessions

Concurrent Session 1 - 3D Chromatin organization and dynamics

14:40-15:05 **Alexandre Reymond**: Chromatin looping: the 16p11.2 case

15:05-15:30 **Mario Nicodemi**: Mapping chromatin in 4D

15:30-15:45 Selected Abstract

Concurrent Session 2 - Clinical Cytogenomics

14:40-15:05 **Nicole de Leeuw**: CNV and diseases

15:05-15:30 **Malte Spielmann**: Noncoding CNV and diseases

15:30-15:45 Selected Abstract

coffee break

16:15-17:20 Concurrent Sessions

Concurrent Session 3 - Structural Organization of the Human Genome

16:15-16:40 **Megan Y. Dennis**: CNV and SD in the human genome

16:40-17:05 **Francesca Antonacci**: Inversion variants in the human genome

17:05-17:20 Selected Abstract

Concurrent Session 4 – Human Infertility

16:15-16:40 **Pierre Ray**: Male infertility in humans

16:40-17:05 **Terry Hassold**: Aneuploidy in human female meiosis

17:05-17:20 Selected Abstract

17:20-18:30 Poster session

MONDAY, 8 July 2019

08:30-10:30 **Plenary session 3 - Tumor Cytogenomics I**

08:30-09:00 **Fredrik Mertens**: Evolution of sarcoma tumors

09:00-09:30 **David Gisselsson**: Treatment resilience of cancer through clonal evolution

09:30-10:30 Selected Abstracts

coffee break

11:00-12:15 Concurrent Sessions

Concurrent Session 5 - Tumor Cytogenomics II

11:00-11:30 **Liran Shlush**: Clonal evolution and risk factors - from age-related clonal hematopoiesis to AML

11:30-12:00 **Floris Foijer**: Single cell DNA sequencing to quantify karyotype heterogeneity in cancer

12:00-12:15 Selected Abstract

Concurrent Session 6 - Animal and Plant Cytogenomics I

11:00-11:30 **Alain Pinton**: Origin and consequences of chromosomal abnormalities in domestic animal species

11:30-12:00 **Ilya Kirov**: Plant repeatome: cytogenetic, transcriptomic and proteomic aspects

12:00-12:15 Selected Abstract

12:15-14:30 Poster session and Satellite Symposia

14:30-15:45 **Plenary session 4 – Chromosomal imbalances**

14:30-15:00 **Orsetta Zuffardi**: Trisomy legacy: from numerical to structural abnormalities

15:00-15:30 **Ilde Bache**: Long-term outcomes of prenatally detected de novo balanced chromosomal rearrangements

15:30-15:45 Selected Abstract

coffee break

16:15-17:15 Concurrent Sessions.

Concurrent Session 7 - Animal and Plant Cytogenomics II

16:15-16:45 **Raquel Chaves**: Satellite evolution in Bovidae

16:45-17:15 **Vincent Colot**: Transposable element mobilization: where, how and with what consequences?

17:15-17:30 Selected Abstract

Concurrent Session 8 – Accreditation, Quality Control and Education

16:15-16:45 **Thomas Liehr**: European Certification and continuous education of Clinical Laboratory Geneticists working in Cytogenetics

16:45-17:15 **Thomas Eggermann**: Genomic Next Generation Sequencing and Quality Assurance: Challenges and Opportunities

17:15-17:30 Selected Abstract

17:30-18:30 Poster session

TUESDAY, 9 July 2019

09:00-10:30 **Plenary session 5 - Prenatal Diagnosis**

09:00-09:25 **Rossa Chiu**: Non invasive prenatal testing

09:25-09:50 **Nathalie Brison**: The landscape of pathogenic copy number variations in healthy, reproducing females

09:50-10:30 Selected Abstracts

10:30-10.50 Coffee break

10:50-11.40 Satellite Symposia

11:50-12:20 **Keynote lecture**. Chair: Mariano Rocchi

Stylianos Antonorakis: Chromatin and single cell genomics, to understand the gene dosage imbalance in aneuploidies

12:20 **Closing ceremony**. Mariano Rocchi

12th EUROPEAN CYTOGENOMICS CONFERENCE
GUIDELINES AND INSTRUCTIONS FOR ABSTRACT SUBMISSION

Abstracts must be submitted in English via the official conference website: **www.eca2019.com**. Abstracts sent by e-mail or fax will not be accepted.

All abstracts may be sent in for a poster presentation. The most significant and original abstracts will be selected for oral presentation in either one of the workshops or in a scientific session. Please indicate in the system, when submitting your abstract, if you do not wish to have your abstract evaluated for an oral presentation.

Please note that at least one author of each abstract should register for the conference. Abstracts submitted without any corresponding conference registration will be refused.

Each submitting author may submit only one abstract to the conference. Please remember that the online abstract submission system does not allow more than one abstract submission per submitting author.

Please do not send multiple versions of the same abstract.

The submitting author will be informed by e-mail about the acceptance of his/her abstract soon after the evaluation process is complete. Acceptance is at the discretion of the committee; publication is at the discretion of the journal.

Abstract submission will close automatically at the end of 3 March 2019.

ABSTRACT PREPARATION

Please have the following information ready before you begin:

For the presenting author:

- Full first and family name
- Department and affiliation
- Full postal address
- City and country
- E-mail address
- Phone and fax number

For each of the co - authors:

- Full first and family name
- Department and affiliation
- City and country

CONTENT

The abstract should contain the title of the paper, the names of the authors and their departments and affiliations (including the cities and the countries), and the e-mail address of the presenting author.

Please note that the abstract system does not allow submissions exceeding 300 words (including the title).

Please do not use tables, images or graphics. If you think it is absolutely necessary to have tables or graphics in the abstract, please contact DEKON Congress & Tourism by e-mail (eca2019@eca2019.com).

Please use only standard abbreviations. If necessary please place special or unusual abbreviations in parentheses after the full word appears for the first time.

LIST OF TOPICS

Abstracts must be allocated to one the following topics:

- Clinical Cytogenomics
- Tumour Cytogenomics
- Prenatal Diagnosis
- Animal and Plant Cytogenomics
- Accreditation, Quality Control, Education
- Genomics
- Other Cytogenomics topics

Please prepare the content of your abstract and paste it to the online form. The online system allows you to prepare your draft and edit it later, right up to the moment you decide to submit your final abstract.

Poster Exhibition

The selected posters will be displayed in the poster hall. Detailed instructions for the poster preparation will be sent to you after acceptance of your abstract.

Selected Oral Presentations

The most significant and original abstracts will be selected for oral presentation in either one of the workshops or in a scientific session.

The submitting author will be informed by e-mail about the acceptance of his/her abstract soon after the evaluation process
is complete.

Michael Schmid 1948 - 2018

We mourn the passing of Prof. Michael Schmid, one of the world's leading cytogeneticists, who died on August 30, 2018, only few days after his 70th birthday. Michael Schmid spent most of his childhood and school years in Caracas/Venezuela. He studied (human) biology at Harvard, Basel, Freiburg and Ulm. During his doctoral thesis at the Human Genetics Institute in Ulm he discovered his lifelong passion for the chromosomes of amphibians and other vertebrates. In the pre-FISH era, his doctoral work on the acrosomal chromocenter in newt histospermiogenesis already impressively demonstrated that the nuclear localization of chromosomes is highly dynamic and functionally important. Although at this time, scientific success was not yet measured in impact factor points, he sometimes chuckled over the fact that the paper resulting from his thesis work was first rejected by a journal which is widely unknown today and finally published in Nature in 1975. Starting in Ulm –after all the 1970s were the golden age of conventional cytogenetics- Michael Schmid also did research on human chromosomes and made many important contributions to human cytogenetics. Following three years as a research scientist at the Göttingen Institute of Human Genetics, he became head of the experimental cytogenetics group at the newly founded Würzburg Institute of Human Genetics where he stayed until his retirement in 2014. Prof. Schmid was not only an internationally known expert in the field of vertebrate chromosome evolution, for more than 35 years he also provided cytogenetic services for human genetic health care on a high level. From the early 1980s, he served on the editorial board of different chromosome journals. In recent years he was editor-in-chief of Cytogenetic and Genome Research, Molecular Syndromology and Sexual Development and not to forget editor of the ISCN. His scientific work comprises more than 400 publications. Most likely, he himself would consider the three big monographs on "Terraranan", "Hemiphractid" und "Arboranan Frogs", the latter one being posthumously published, as his legacy. Michael Schmid has imparted his enthusiasm for chromosome research to many generations of students, staff members, and colleagues and had a positive impact on many careers. With his passing away, the cytogenetic community loses a highly respected member, an enthusiastic researcher and academic teacher, and last but not least, an inspirational human being.

Thomas Haaf
Institute of Human Genetics, Würzburg

GenQA GENOMICS QUALITY ASSESSMENT

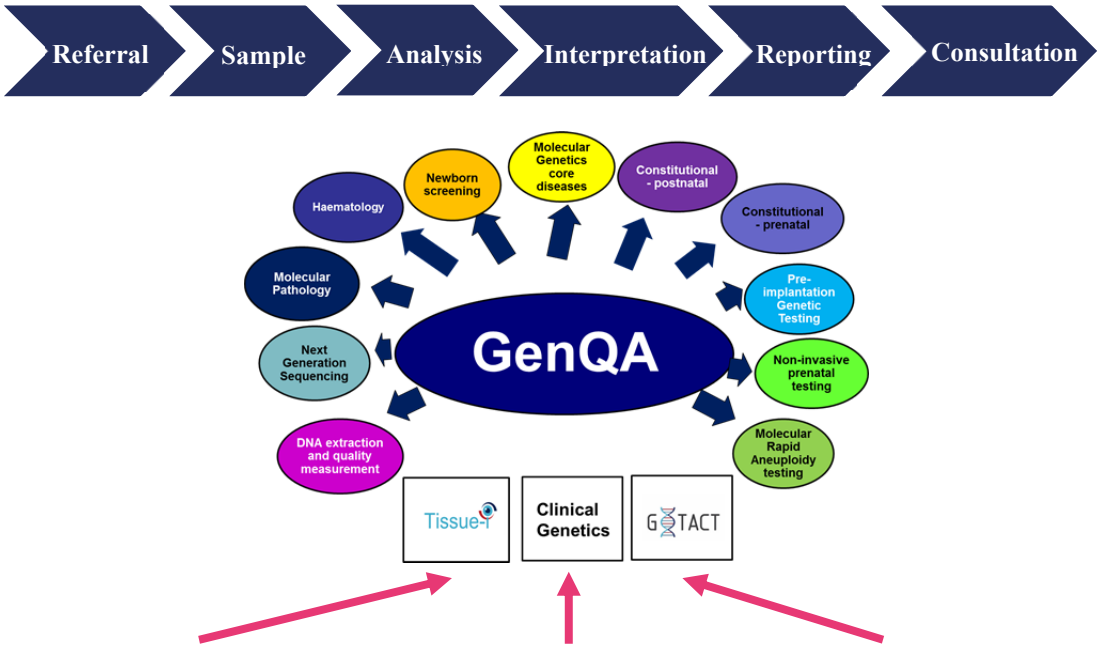
Collaboration between CEQAS and UK NEQAS Molecular Genetics
Members of UK NEQAS consortium

GenQA update

During the first year of operating as a joint venture providing a global quality assessment service for genomics, Genomics Quality Assessment (GenQA) achieved some impressive milestones:

- 680 participating laboratories (including 66 laboratories registered during 2018)
- 81 countries covered;
- 3759 EQA participations;
- 84 EQAs offered (60 of which are ISO accredited)

GenQA is now the largest ISO 17043 accredited Genomics EQA provider worldwide, with the unique provision of an 'End to End' EQA repertoire across the Genomic specialities. From sample receipt to Genetic Consultation, through EQA, G-TACT and Tissue-i, GenQA covers every aspect of a Genetic diagnostic service allowing participating centres to demonstrate 'End to End' assurance of a high quality service to their customers.



Online tumour content assessment for solid tumours EQAs

- The amount of tumour DNA present in the NGS assay is important to enable accurate interpretation of the result
- Evidence competency to select appropriate tissue areas for genomic testing.
- Covers a range of tumours: breast cancer, colorectal cancer, lung cancer, melanoma, ovarian cancer, prostate cancer and sarcoma.

Online assessment of clinical case scenario

- Involves the genetic consultation process from initial patient referral, selection of appropriate testing, clinical interpretation of the results and patient counselling.
- Enables clinical geneticists to review their clinical practice and facilitate optimal patient care.
- Covers oncogenetics, monogenic, cardiogenetics and dysmorphology

Online Training and competency assessment tool (G-TACT)

- For individual members of laboratory staff.
- Covers different aspects of laboratory activities
- Staff competency reviewed by line manager and
- Provides evidence of continual professional development for the individual.
- Addresses ISO15189 requirements

In order to provide the full repertoire of EQAs, GenQA now encompasses a broad spectrum of approaches:

- **Technical only**
  DNA extraction quality and quantity assessment.
  NGS for germline and somatic testing
- **Analytical/genotype only**
  Sample testing required but no interpretation of result
- **Analytical/genotype and interpretation**
  Sample testing and full interpretation of the result in the context of the clinical case supplied
- **Interpretation only**
  Test results and clinical details provided for interpretation

In accordance with ISO 17043, all EQA cases are independently validated prior to their distribution and are assessed by panels of international experts in accordance with peer ratified marking criteria.

For 2019 GenQA will add the following new EQAs to its repertoire:

- Imprinting and Uniparental disomy
- Neurofibromatosis (types 1 and 2) and Schwannomatosis
- Osteogenesis Imperfecta
- Retinal disorders
- Variant validation
- X-inactivation
- *TP53* mutation and *IGH* hypermutation status in CLL (Chronic lymphocytic leukemia)
- Renal Cancer Panel
- Post stem cell transplant FISH testing: chimerism monitoring
- *BRAF* fusion and mutation testing in CNS tumours

Enrolment for the 2019 EQAs is open until Thursday 28th February, 2019. To register, enrol or view the full catalogue of 2019 EQAs please go to the GenQA website – www.genqa.org.

In today's fast paced genomics market, where there is a choice of testing laboratories and samples are sent across borders, it is more important than ever to assure that a laboratory's results measure up to the highest international standards. Continuous participation in EQA enhances the analytical and interpretative performance of the laboratory. To this end GenQA collaborates with other EQA providers and outside bodies such as EMQN, UK NEQAS IHC & ISH, UK NEQAS LI, IQN Path, ERIC and the ESHG Quality Committee to increase and improve the provision of comprehensive, clinical quality assessment to reflect current practice and future requirements.

GenQA's education role does not limit itself to EQA provision but also includes workshops, lectures, publications and posters as well as involvement in facilitating the production of Best Practice Guidelines. GenQA will be represented with a stand at the 2019 E.C.A. conference in Salzburg, the ESHG conference in Gothenburg as well as at other national and international conferences so please do visit us.

If you would like to be involved with GenQA, as an expert advisor or with suggestions of areas where EQA is required then please contact us by email (info@genqa.org).

The GenQA team would like to thank our participants for their continued support and wish you all a successful New Year.

E.C.A. STRUCTURES

E.C.A. BOARD OF DIRECTORS

Sevilhan ARTAN
Eskisehir Osmangazi University
Medical Faculty
Department of Medical Genetics
Meselik
26480 ESKISEHIR
TURKEY
Tel.: +90 22 22 39 37 71
Fax : +90 22 22 39 29 86
E-mail: sartan@ogu.edu.tr

Juan Cruz CIGUDOSA
Centro Nacional de
Investigaciones Oncológicas
(CNIO)
Melchor Fernández Almagro, 3
28029, MADRID
SPAIN
Tel. :+34 91 22 46 900, ext 3340
Fax : +34912246923
E-mail: jccigudosa@cnio.es

Jean-Michel DUPONT
Laboratoire de Cytogénétique
Groupe Hospitalier Cochin
Saint Vincent de Paul
123 Bd Port Royal
75014 PARIS
FRANCE
Tel.: +33 1 584 113 31
Fax : +33 1 584 113 55
E-mail.:
jean-michel.dupont@ cch.aphp.fr

José M. GARCIA-SAGREDO
Medical Genetics Department
Hospital Ramon y Cajal
Carretera de Colmenar Km 9.100
28034 MADRID
SPAIN
Tel.: +34 91 3368334
Fax : +34 91 3368791
E-mail:
jgarcias.hrc@salud.madrid.org

J.S. (Pat) HESLOP-HARRISON
Department of Genetics
University of Leicester
LEICESTER LE1 7RH
UK
Tel.: +44 116 252 5079
Fax.: +44 116 252 2791
E-mail: phh4@le.ac.uk

Thierry LAVABRE-BERTRAND
Laboratoire de Biologie Cellulaire
et Cytogenetique Moleculaire
Faculté de Médecine
Avenue Kennedy
30900 NÎMES
FRANCE
Tel.: +33 4 66 68 42 23
Fax: +33 4 66 68 41 61
E-mail: tlavabre@univ-montp1.fr

Nicole de LEEUW
Department of Human Genetics
Radboud University Nijmegen
Medical Centre
P.O. Box 9101
6500 HB NIJMEGEN
THE NETHERLANDS
Tel.: +31243614105
Fax : +31243668751
E-mail: N.deLeeuw@antrg.umcn.nl

Kamlesh MADAN
Cytogenetics Laboratory
Dept. of Clinical Genetics S-06-P
Leiden Univ. Medical Center
P.O.Box 9600
2300 RC LEIDEN
THE NETHERLANDS
Tel.: +31 72 51 28 953
Fax : +31 71 52 68 276
E-mail: k.madan@lumc.nl

Konstantin MILLER
Institut für Humangenetik
Medizinische Hochschule
30623 HANNOVER
GERMANY
Tel.: +49 511 5323572
Fax : +49 511 5326555
E-mail:
miller.konstantin@mh-hannover.de

Felix MITELMAN
Department of Clinical Genetics
University of Lund, BMC C13
22185 LUND
SWEDEN
Tel.: +46 46 17 33 60
Fax: +46 46 13 10 61
E-mail: felix.mitelman@med.lu.se

Maria Rosario PINTO LEITE
Cytogenetics Laboratory
Centro Hospitalar de Trás-os-
Montes e Alto Douro
Av. da Noruega
5000-508 VILA REAL
PORTUGAL
Tel.: +35 1 25 93 00 500
Fax: +35 1 25 93 00 537
E-mail:
mlleite@chtmad.min-saude.pt

Harald RIEDER
Institut fuer Humangenetik und
Anthropologie
Universitaetsstraße 1
40225 DUESSELDORF
GERMANY
Tel.: +49 211 8110689,
Fax : +49 211 8112538
E-mail:
harald.rieder@uni-duesseldorf.de

Mariano ROCCHI
DAPEG
Sezione di Genetica
Istituto di Genetica
Via Amendola 165/A
70126 BARI
ITALY
Tel.: +39 080 544 3371/3339
Fax : +39 080 544.3386
E-mail rocchi@biologia.uniba.it

Elisabeth SYK LUNDBERG
Dept. of Clinical Genetics
Karolinska Hospital
17176 STOCKHOLM
SWEDEN
Tel.: +46 85 17 75 380
Fax : +46 83 27 734
E-mail:
elisabeth.syk.lundberg@ki.se

Roberta VANNI
Dept. of Biomedical Sciences
Biochemistry, Biology and
Genetics Unit
University of Cagliari
09142 MONSERRATO (CA)
ITALY
Tel.: +39 07 06 75 41 23
Fax : +39 07 06 75 41 19
E-mail: vanni@unica.it

COMMITTEE

ECC SCIENTIFIC PROGRAMME COMMITTEE

E.C.A. News

- The next **European Cytogenomics Conference** will take place in Salzburg, Austria, from 6 to 9 July 2019.
- Renewal of the Board in 2019. The following members are due for replacement or re-election in 2019 at the General Assembly: S. Artan (Turkey), J. Cigudosa (Spain), N. de Leeuw (The Netherlands), K. Miller (Germany), F. Mitelman (Sweden).
- Nomination of individual candidates for the Board together with their motivation and a CV may be sent to the President, Prof. M. Rocchi (mariano.rocchi@uniba.it) before 1 March 2019.
- According to the statutes, lists for the board election may be sent to the President until 15 May 2019.
- The total number of members now stands at 1136, comprising 990 active members, 124 associated members from non-European countries, 22 honorary members, patient and other organizations.

E.C.A. Fellowships

- The **E.C.A.** offers two **Fellowships** for each of the following courses:
  - **European Advanced Postgraduate Course in Classical and Molecular Cytogenetics** to be held in Nîmes 11-19 March 2019.
  - **Goldrain Course in Clinical Cytogenetics** to be held in Goldrain Castle (South Tyrol, Italy) 31 August - 7 September 2019.
- The fellowships **include the course fees and the accommodation** during the lectures in Nîmes or in Goldrain but **do not include travel expenses** for either of the courses or for accommodation during the practical training for the Nîmes course.
- Applications with CV, list of publications and a letter of support should be addressed to the appropriate course organizer. The Educational Advisory Council of the E.C.A. will select the successful candidates.

MINUTES OF THE E.C.A. BOARD MEETING, PARIS, SEPTEMBER 2018

A meeting of the E.C.A. Board of Directors was held on 21st September 2018 in Hôpital Cochin, Paris.

The Board Members present were: Sevilhan Artan, José M. Garcia-Sagredo, Jean-Michel Dupont (Treasurer), Pat Heslop-Harrison (2nd Vice-President), Konstantin Miller (General Secretary), Thierry Lavabre-Bertrand, Harald Rieder, Maria Rosario Pinto Leite, Mariano Rocchi, and Elisabeth Syk Lundberg (President).

Apologies were received from Kamlesh Madan (First Vice-President), Felix Mitelman, Nicole de Leeuw, Juan Cruz Cigudosa, and Roberta Vanni.

The President, Elisabeth Syk Lundberg, opened the meeting at 15.05.

1. The Minutes of the meeting of the ECA Board of Directors and the General Assembly of the Association held on 16th June 2018 Milano Congressi, Milano, Italy were approved.
2. Committee election. The First Vice-President, Second Vice-President, Treasurer, and Secretary General would be happy to accept nomination to continue in Office. The President has completed her term and will stand down. Mariano Rocchi has agreed to serve as President. The President was elected, and other Officers were re-elected, unanimously. The new President, Mariano Rocchi, assumed the Presidency and Chair.
3. The Secretary General reported on membership. The list of new members was approved.
4. The Treasurer reported no significant changes in the financial situation.
5. There was a discussion about the website layout, uploading of material including newsletters (now on-line only) and access to protected areas of the website. The membership database and collection of subscriptions were also discussed.
6. The Chair of the Scientific Committee, Mariano Rocchi, reviewed the plans for the 12th European Cytogenomics Conference in 2019. There was an extensive discussion of topics and speakers. The Chair will summarize this discussion, take appropriate actions and inform the Scientific Committee and the Board where relevant. Board members should forward any proposals to Mariano Rocchi. The procedure for reviewing the abstracts was discussed; the titles of topics and the names of the reviewers were decided. The committee will visit Salzburg in early January 2019 to look at the facilities.
7. The Election for the Board for 2019 was discussed. Two Board members will not stand for re-election; new nominations will be considered.
8. The Goldrain and Nîmes (with two levels of diploma and with a practical component) courses are planned for 2019. The unused ECA fellowship in 2018 for the Goldrain course will be honoured in 2019. A multiple-choice examination will be introduced for the Nîmes course.
9. The Board approved the French version of the Minutes for signature.

The Board will meet on 16th March 2019 in Nîmes.

The President closed the meeting at 18.20.

E.C.A. PERMANENT WORKING GROUPS (PWG)

PWG: CLINICAL AND MOLECULAR APPROACHES TO CYTOGENETIC SYNDROMES.

Co-ordinators:

Conny van RAVENSWAAIJ
Dept. of Human Genetics CB51
University Medical Centre Groningen
P.O.Box 30.001
9700 RB GRONINGEN, THE NETHERLANDS
Tel.: +31 503617229, Fax: +31 503617231
E-mail: c.m.a.van.ravenswaaij@medgen.umcg.nl

Cristina SKRYPNYK
Al-Jawhara Centre for Molecular Medicine and Inherited Disorders
Arabian Gulf University
P.O Box 26671 MANAMA
KINGDOM OF BAHRAIN
E-mail: cristinas@agu.edu.bh

ECARUCA Co-ordinator Daily Management Team
Nicole de LEEUW
Department of Human Genetics (848)
Radboud University Nijmegen Medical Centre
P.O. Box 9101
6500 HB NIJMEGEN, THE NETHERLANDS
E-mail: Nicole.deLeeuw@radboudumc.nl

ECARUCA, the European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations (ECARUCA, www.ecaruca.net) will soon seize to exist, 15 years after the initial start of the ECARUCA project, which was funded by grants of the European Union Fifth Framework Programme from 2003 until 2005. The main objective of ECARUCA has always been to improve the knowledge of rare chromosome disorders for medical and research purposes. This was predominantly achieved by collecting and curating rare, clinically relevant genomic imbalances in combination with detailed phenotypic information through the online ECARUCA database. To date, this database contains over 5,000 cases and almost 7,000 aberrations detected by cytogenetic analysis, FISH and genome wide array analysis.
For the past ten years, the ECARUCA database was able to continue thanks to the support of the department of Human Genetics at the Radboud university medical center in Nijmegen, the Netherlands. We were pleased when we started a collaborative effort with Cartagenia in 2013, because that would mean that they would take care of the technical infrastructure of the database, thereby enabling several improved features in return for the database's content becoming directly available to Cartagenia users. This so-called ECARUCA powered by Bench was developed and tested through many phases in the past years. But things slowed down after Cartagenia was acquired by Agilent Technologies in 2015. Despite this very slow development, we remained confident that the end point for the migration of the database's full content would finally be reached. However, after yet another long silence, our mails to our main contacts, the two founders of Cartagenia, were bounced in November, 2018. It turned out that both Steven van Vooren as well as Bert Coessens had left Cartagenia in September and October, 2018, respectively. Our last contact at Agilent on this project was willing and able to inform us about the situation. It became clear that the future of ECARUCA we had foreseen is not going to be realized after all.
This is a very disappointing result, but we hope that we will soon find a suitable solution to at least continue sharing the gathered content of the ECARUCA database. We expect to inform you about this at our Permanent Working Group meeting on Clinical and Molecular Approaches to Cytogenetic Syndromes during the 12th ECA conference in Salzburg. Please note that this one-hour pre-conference meeting in the afternoon of Saturday July 6, 2019, is open to all ECA participants.

On behalf of the ECARUCA team
Nicole de Leeuw, PhD
Coordinator ECARUCA

PWG: MARKER CHROMOSOMES.

Co-ordinators:
Thomas LIEHR
Jena University Hospital, Friedrich Schiller University, Institute of Human Genetics
Postfach
07740 JENA, GERMANY
Tel: + 49 3641 93 96 850, Fax: +49 3641 93 96 852
E-mail: Thomas.Liehr@med.uni-jena.de

Isabel MARQUES CARREIRA
Cytogenetics and Genomics Laboratory,
Faculty of Medicine, University of Coimbra
Rua Larga
3004-504 COIMBRA, PORTUGAL
Tel/Fax . +351 23983886
E-mail: i_marques@hotmail.com

The well appreciated small supernumerary marker chromosome (sSMC) - webpage http://ssmc-tl.com/Start.html now includes the following subpages for:

- sSMC (>6,100 entries)
- UPD (>3,650 entries)
- multicolor-FISH (~1,800 entries) and
- chromosomal heteromorphisms (>430 variants).

Reminders:

1. As all sSMCs can now be characterized for their gene content, any laboratory that does

not have the facilities and has an interesting sSMC case can contact the PWG for help. Also please remember that chromothripsis is one of the possible mechanisms for the formation of an sSMC.

2. You are welcome to attend the next meeting of our PWG, which will be held on Saturday, 6 July 2019, during the E.C.A. conference in Salzburg, Austria.

PWG: CYTOGENETICS OF HAEMATOLOGICAL MALIGNANCIES.

Co-ordinators:

Bertil JOHANSSON
Dept. of Clinical Genetics - University Hospital
22185 LUND, SWEDEN
Tel.: +46 46 17 33 69, Fax :+46 46 13 10 61
E-mail: bertil.johansson@klingen.lu.se

Harald RIEDER
Institut fuer Humangenetik und Anthropologie
Universitaetsstraße 1
40225 DUESSELDORF, GERMANY
Tel.: +49 211 8110689, Fax : +49 211 8112538
E-mail: harald.rieder@uni-duesseldorf.de

The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC. Propositions for presentations are welcome.

PWG: CANCER CYTOGENETICS, SOLID TUMOR STUDIES.

Co-ordinators:

Roberta VANNI
Department of Biomedical Sciences
Biochemistry, Biology and Genetics Unit
University of Cagliari, University Campus
09142 MONSERRATO (CA), ITALY
Tel. +39 07 06 75 41 23 Fax +39 07 06 75 41 19
E-mail: vanni@unica.it

David GISSELSSON NORD
Lund University
Dept. of Pathology, Lund University Hospital
22185 LUND, SWEDEN
E-mail: david.gisselsson_nord@med.lu.se
The PWG on Cancer Cytogenetics: Solid Tumor Studies will hold a meeting on Saturday 6 July 2019, during the 12th ECC in Salzburg.

Cancer cytogenetics has focused on genome instability through the analysis of large chromosomal aberrations, while cancer cytogenomics has looked at focal chromosomal point mutations, insertions, deletions, and SNPs through NGS. Recent studies of patient derived xenografts (PDXs) models appear to connect these aspects. "State of the Art Seminar on Solid Tumor Cytogenomics" is the title of this year's PWG meeting. Participants involved in studies of solid tumors are advised to take these aspects into consideration when preparing the abstracts for the 12th ECC. We plan to select five ten-minute presentations on new trends in solid tumor cytogenetics/ cytogenomics from the abstracts submitted to the ECC.

PWG: CYTOGENETIC TOXICOLOGY AND MUTAGENESIS.

Co-ordinators:

José M. GARCIA-SAGREDO
Medical Genetics Department
Hospital Ramon y Cajal
Carretera de Colmenar Km 9.100
28034 MADRID, SPAIN
E-mail : jgarcias.hrc@salud.madrid.org

Emanuela VOLPI
Faculty of Science and Technology
University of Westminster
115 New Cavendish Street
LONDON W1W 6UW, UK
E-mail: e.volpi@westminster.ac.uk

It is my pleasure to introduce myself as the new Co-Coordinator alongside Professor Garcia-Sagredo of the E.C.A. 'Cytogenetic Toxicology and Mutagenesis' PWG. I was kindly invited to take up this role by Professor Garcia-Sagredo in May 2018 and was delighted to accept. I very much look forward to continuing the endeavours of this permanent working group by renewing discussion and facilitating networking around the eponymous research themes.

With the support of the University of Westminster, my home institution, I had the privilege of hosting in London in September 2018 the Human Micronucleus Network Workshop. The HUMN project is an international collaborative project aimed at developing and validating the use of micronucleus cytome assays for biomonitoring of DNA damage and chromosomal instability in humans. An important activity of the HUMN project is to organise educational workshop on the latest version of the of micronucleus cytome assay protocols and their use to investigate the effect of genetic, environmental, nutritional and life-style factors - as well as ageing and disease - on genome integrity. The attendees were treated to outstanding lectures by Professor Michael Fenech of the Genome Health Foundation, University of South Australia, followed by practical sessions in the laboratory. There was ample opportunity for networking and the event was extremely well received.

As part of the activities of the PWG in 2019, on the first day of the ECA Conference in Salzburg in July, we shall be holding a workshop entitled 'Genotoxicity assays applied to the assessment of DNA damage in ageing and disease'. If you are attending the ECA conference and would like to be considered for presenting your work at this workshop, please contact me: e.volpi@westminster.ac.uk.

PWG: ANIMAL, PLANT, AND COMPARATIVE CYTOGENOMICS.

Co-ordinators:

J.S. (Pat) HESLOP-HARRISON
Department of Biology
University of Leicester
LEICESTER LE1 7RH, UK
Tel.: +44 116 252 5079 Fax.: +44 116 252 2791
E-mail: phh4@le.ac.uk

Valérie Fillon
Laboratoire de Génétique Cellulaire
Institut National de la Recherche Agronomique de Toulouse
31326 Castanet Tolosan, France,
Tel: +33 0561285347
E-mail: valerie.fillon@inra.fr

Animal, Plant and Comparative Cytogenetics will have a much increased profile during the conference in Salzburg. On the afternoon of Saturday 6th July, the Permanent Working Group will have an exciting open session of short talks selected from the abstracts submitted to the conference; everyone is most welcome to submit presentations for this session. The talks will be followed by a discussion of developments in cytogenomics and their impact on understanding genome evolution in a comparative context. During the rest of the conference, as well as many relevant talks in plenary and concurrent sessions, we will have two sessions on Animal and Plant Cytogenomics on Monday 8th July, featuring six diverse talks from research leaders about the latest developments in the field. The Working Group coordinators look forward to welcoming you to Salzburg and discussing your research.

PWG: PRENATAL DIAGNOSIS.

Co-ordinators :

Seher BASARAN
Istanbul University
Child Health Inst., Millet Cad. Capa
34390 ISTANBUL, TURKEY
Tel.: +90 21 26 31 1363 Fax : +90 21 26 31 1363
E-mail: premed@premed.com.tr

Maria Do Rosário CARVALHO PINTO LEITE
Cytogenetics Laboratory
Centro Hospitalar de Trás os Montes e Alto Douro
5000-508 VILA REAL, PORTUGAL
Tel.: +35 1259 300 537
E-mail: mlleite@chtmad.min-saude.pt

The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC. Propositions for presentations are welcome.

PWG: QUALITY ISSUES AND TRAINING IN CYTOGENETICS.

Co-ordinators:

Ros HASTINGS
UKNEQAS for Clinical Cytogenetics
Women's Centre, John Radcliffe Hospital
OXFORD OX3 9DU, UK
Tel: +44-1865-857644
E-mail: Ros.Hastings@orh.nhs.uk

Martine DOCO-FENZY
Service de génétique - Hôpital Maison Blanche
45, rue Cognacq Jay
51092, REIMS Cedex, FRANCE
martine.doco@gmail.com

Marta RODRIGUEZ DE ALBA
Department of Genetics,
Fundacion Jimenez Diaz
Avda. Reyes Catolicos No. 2
28040 MADRID, SPAIN
Tel.: +34 39 41 550 48 72
E-mail: mrodrigueza@fjd.es

There will be a workshop at the PWG session on ISCN nomenclature. Please email any ISCN issues beforehand to ros.hastings@ouh.nhs.uk. We look forward to seeing you in Salzburg.

PWG: CYTOGENOMICS.

Co-ordinators:

Joris VERMEESCH
Constitutional Cytogenetics laboratory
Center for Human Genetics
U.Z. Gasthuisberg
Herestraat 49
3000 LEUVEN, BELGIUM
Tel.: +32 16 34 59 41, Fax: + 32 16 34 60 60
E-mail: Joris.vermeesch@med.kuleuven.ac.be

Anna LINDSTRAND
Karolinska Hospital
17176 STOCKHOLM, SWEDEN
E-mail: anna.lindstrand@ki.se

The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC. Propositions for presentations are welcome.

EUROPEAN CYTOGENETICISTS ASSOCIATION (E.C.A.)
European Advanced Postgraduate Course in Classical and Molecular Cytogenetics

Director: Professor Jean-Michel Dupont, Paris - France

Objectives

This course was started by Professor Jean Paul Bureau 22 years ago and has been held in Nîmes under his directorship ever since. It is designed to provide advanced training in constitutional, haematological, and oncological cytogenetics to medical graduates, pharmacists, pathologists, biologists, health professionals and researchers, with an academic qualification. The students will be trained to identify genetic abnormalities for diagnosis and prognosis, and for fundamental and applied research using both classical and molecular cytogenetic techniques. The course is co-organized by E.C.A. and two French Universities, either as ***a Diploma (Basic = only the lectures or Advanced = lectures + practical training)*** or as a stand-alone course (lectures only)

Practical information

Lectures: A ten-day course held in February/March of each year.
Venue: Faculty of Medicine, Nîmes, France.
Official language: English.

Practical training (**only for students registered for the advanced Diploma**): A training of maximum 2 months in a cytogenetic laboratory. A list of laboratories is provided during the theoretical course.

Assessment **:** The assessment for the **basic diploma** will be on the basis of a one-hour examination held at the end of the lecture course**.** The knowledge of the students for the **advanced diploma** will be assessed in September by a written test (three questions) and an oral examination including a presentation (10-15 min) related to the practical training. The University will award a diploma to only those students who have passed.

All participants (including those for the stand-alone course) will receive a certificate of attendance by the E.C.A.

Topics (see next page).

Accommodation

A special price is available for participants in the 4-star Vatel hotel close to the course venue. We highly recommend that all participants stay in this hotel where all the lecturers will be hosted in order to promote interactions during the course.
Accommodation is included in the stand-alone course fee

Registration

Registration opens in September and closes on January 30th.
To register please send a letter of application together with your CV by e-mail to one of the organizers mentioned below. If you are accepted you will receive a registration form.

Prof. Jean-Michel DUPONT
Laboratoire de Cytogénétique
Hôpital Cochin
27 rue du Fbg St Jacques
75014 Paris, France
jean-michel.dupont@aphp.fr
sylvie.mendez@aphp.fr

Prof. Thierry LAVABRE-BERTRAND
Laboratoire de Biologie Cellulaire et Cytogénétique Moléculaire
Faculté de Médecine Montpellier-Nîmes
Avenue Kennedy
30900 Nîmes, France
tlavabre@univ-montp1.fr

Registration fees

Diploma: From €360 to €1780 depending on the status of the student; accommodation is **NOT included**
Stand-alone course: €1300 (E.C.A. members) or €1400 (Non E.C.A. members); accommodation is **included** on a shared double room basis. Extra fee for a single room on request.

2019 Course provisional program

11-20 March 2019

This approximately 55-hour theoretical part of the course attempts to cover the field of cytogenetics in the broadest sense. The topics can be divided into the following categories:

Technical aspects:

Classical Cytogenetics: Cell culture techniques; Chromosome staining methods (Q-, G-, C-, R-banding and high resolution banding);

Molecular Cytogenetics: Methods and principles of Fluorescence In Situ Hybridization (FISH) and MFISH; Array CGH; Application of Massively Parallel Sequencing to Cytogenetics; Production and use of molecular probes; Database use in Cytogenetics;

Laboratory quality assessment.

Clinical cytogenetics:

Basics: Frequency of chromosome disorders; Cell cycle, mitosis and meiosis, gametogenesis; Heterochromatic and euchromatic variants; Numerical chromosome abnormalities; Structural abnormalities: translocations, inversions, insertions, deletions, rings, markers; Risk assessment for balanced abnormalities; X inactivation; numerical and structural abnormalities of the X and the Y; Mosaicism; Chimaeras; ISCN 2013.

Clinical: Phenotype of common autosomal and gonosomal aneuploidies; Chromosome abnormalities in recurrent abortions; Cytogenetics and infertility; Microdeletion syndromes; Uniparental disomy and its consequences; Genomic imprinting; Genetic counselling and ethical issues in cytogenetics.

Prenatal diagnosis: Indications, methods and interpretation; Risk assessment for chromosomal abnormalities; Non-invasive methods using foetal nucleic acids and foetal cells in maternal blood; Pre-implantation diagnosis.

Cancer Cytogenetics: Molecular approach to cancer cytogenetics; Predisposition to cancer, Chromosome instability syndromes; Chromosome mutagenesis; Solid tumors; Clinical application in onco-haematology.

Other:

Genome architecture; Structure of chromatin; Structure of metaphase chromosomes, Mechanisms of chromosome abberations; Origin of aneuploidy; Evolution and plasticity of the human genome; Animal cytogenetics; Plant cytogenetics.

The students will have the opportunity to evaluate the course.

The **European Cytogeneticists Association** offers **two scholarships** for **the European Advanced Postgraduate Course in Classical and Molecular Cytogenetics** to candidates of excellence. The Education Committee of the E.C.A. will select the suitable candidates.

The scholarship includes registration to the course and accommodation in Vatel Hotel in a shared double room but **does not include travel costs.**

Scholarships will not be allocated to students whose registration is paid by a third party institution.

Goldrain Course 2018

The 13th Course in Clinical Cytogenetics was, as always, held in the beautiful Goldrain Castle in South Tyrol in northern Italy. The first little challenge was to reach this region, as all the nearest airports are located far away, but this trip through Italy's most northerly province dotted with many gorgeous sights added some special unforgettable feelings about the course.

The course was all about cytogenetics. It was very intensive and fully occupied my mind. The topics covered by the lectures, presented by experienced faculty members, ranged from basic and molecular cytogenetics to cytogenomics. Techniques such as FISH, MLPA, QF-PCR, SNP arrays, next generation sequencing and introduction to CRISPR/Cas9 were presented. The lectures on clinical aspects included genetic counselling for chromosomal aberrations, clinical examination for dysmorphology, clinical phenotypes in UPD, microdeletion syndromes, and management of common chromosome disorders. Almost two days of the course were dedicated to prenatal diagnosis with especial attention to traditional approaches for detecting aneuploidy, non-invasive prenatal aneuploidy testing and pre-implantation genetic diagnosis. There was also an interesting lecture on ethical issues among many others.

The 34 participants were not only from European countries, but also from Brazil, Saudi Arabia, India and Egypt. During the workshops the students were divided into small groups for discussion and practical work. We had some practical exercises in reporting laboratory results, the use of ISCN system for describing chromosomal abnormalities and we made practical exercises to understand the segregation of chromosomal translocations. We had the possibility to work in the computer room with databases for interpreting array results, using the ECARUCA database for solving some cases and making diagnostic strategies following the detection of CNVs. Several students took advantage of the opportunity to present their scientific work or unusual cases; there was a prize for the best presentation.

Discussions continued during coffee breaks, lunch and dinner. We all had the possibility to talk to faculty members and ask them questions; they all shared their experience willingly and gave practical advice.

In the middle of the course, we had an interesting excursion to the Messner Mountain Museum, which told the story of mountaineering and rock climbing. On the second leisure afternoon we had a free time either for self-study or for an excursion on our own to the beautiful places around Goldrain. All the participants were very friendly and our WhatsApp group is still active!

It is impossible not to mention the comfortable accommodation in cosy rooms of the guesthouse and the great atmosphere. We stayed together even during meal times in the modern restaurant of the Goldrain castle and enjoyed very delicious traditional South *Tyrolean* meals.

I would like to recommend this course to all cytogeneticists, as well as clinical geneticists and specialists involved in prenatal diagnosis, who wish to widen their knowledge of clinical cytogenetics. I am very thankful to Professor Albert Schinzel for this great opportunity for learning and meeting wonderful people. I also express my gratitude to Professor Schinzel, his family and all the lecturers for their enthusiasm and hard work in organizing this excellent course.

Eglė Preikšaitienė
Vilnius, Lithuania

Participants and faculty members of the 2018 course at Goldrain castle.

Forthcoming events

30th Annual Meeting of the German Society of Humangenetics (GfH) together with Austrian Society of Humangenetics (ÖGH) and the Swiss Society of Medical Genetics (SGMG)
6-8 March 2019, Weimar, Germany
https://www.gfhev.de/en/congress/

European Advanced Postgraduate Course in Classical and Molecular Cytogenetics
11-20 March 2019, Nîmes, France
See the announcement on pages 17/18 of this Newsletter
http://www.e-c-a.eu/

European Human Genetics Conference 2019
Gothenburg, Sweden, June 15-18, 2019
https://2019.eshg.org/

12th European Cytogenomic Conference 2019
6-9 July 2019, Salzburg, Austria
www.eca2019.com
See pages 3 - 7 of this Newsletter.

Goldrain Course in Clinical Cytogenetics
31 August-1 September 2019,
Goldrain Castle (BZ), Italy
See the announcement on page 21 of this Newsletter
http://www.e-c-a.eu/

American Society of Human Genetics ASHG 2019
15-19 October 2019, Houston, TX, United States
http://www.ashg.org/2019meeting/

14th Goldrain Course in
Clinical Cytogenetics
August 31 to September 07, 2019

LOCATION

Goldrain Castle, Goldrain, South Tyrol, Italy
Website of the venue: **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 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 – ISCN – Practical exercises in cytogenetic nomenclature – The ECARUCA database: Introduction and practical exercises – Students presentation of cases with difficult-to-interpret chromosome aberrations– prenatal cytogenetic diagnosis – Mosaics and chimeras – imprinting and uniparental disomy – FISH techniques and their interpretation – MLPA – Array-CGH: principles, technical aspects; evaluation of the results – SNP arrays – QF-PCR – Epidemiology of chromosome aberrations – Chromosome aberrations in spontaneous abortions and stillborns –Harmless chromosome aberrations – Risk assessment in structural chromosome aberrations – Optimal use of available techniques in clinical cytogenetics – Extra small supernumerary chromosomes – Genomic variation: a continuum from SNPs to chromosome aneuploidy – Use of genomic databases – Pre-implantation cytogenetic diagnosis – Ultrasound findings indicative of chromosome aberrations– Accreditation of cytogenetic laboratories –Ethical issues in the context of cytogenetic diagnosis – Non-invasive prenatal cytogenetic diagnosis.

Practical exercises will be offered with the ISCN system for chromosome aberrations and with cytogenetic and genomic databases. Students will have the opportunity to present their own observations and cytogenetic 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

D. Bartholdi (Berne, Switzerland), A. Baumer (Zurich, Switzerland), P. Benn (Farmington CT, U.S.A.), R. Ciccone (Pavia, Italy), E. Klopocki (Würzburg, Germany), K. Madan (Leiden, The Netherlands), K. Miller (Hannover, Germany), E. Syk-Lundberg (Stockholm, Sweden), G. van Buggenhout (Leuven, Belgium), O. Zuffardi (Pavia, Italy) and others

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





Full fee is Euro 1400 for a single room or Euro 1200 (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.

Editors-in-Chief: Thomas Liehr, Henry Heng and Yuri Yurov
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