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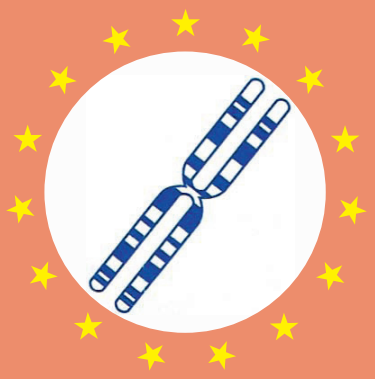
EUROPEAN CYTOGENETICISTS ASSOCIATION



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.

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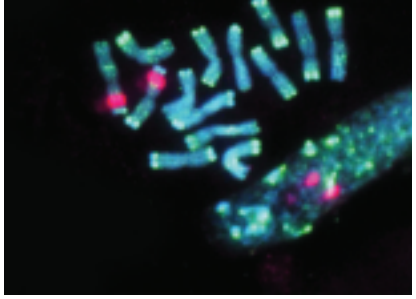

European Cytogeneticists Association

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

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Scientific Programme

SATURDAY, 6 July 2019

14:00-17:00 Permanent Working Groups (for details see pages 15/16)

17:00-17:50 Symposium: Molecular Cytogenetics (BMC-Springer/Nature) symposium dedicated to the memory of Prof. Yuri Yurov

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

Joris Vermeesch: Somatic chromosomal mosaicim

SUNDAY, 7 July 2019

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

Chairs: Mariano Rocchi – Thierry Lavabre-Bertrand

08:30-09:00 **Claudia Haferlach:** The future of cytogenomics in the diagnostics

09:00-09:30 **Michael Speicher:** Liquid biopsies in patients with cancer

09:30-10:15 Selected abstracts

09:30-09:45 **Pascal Chambon:** A simple, universal and cost-efficient dPCR method for the targeted analysis of copy number variations

09:45-10:00 **Laïla El Khattabi:** Next Generation Mapping, a novel approach that enables the detection of unbalanced as well as balanced structural variants

10:00-10:15 **Paolo Reho:** Low-coverage whole genome sequencing in plasma circulating cell-free DNA analysis: the Turner syndrome experience

10:15-10:45 Coffee break

10:45-11:15 **Plenary session 2 - 50 Years of chromosome banding**

Chairs: Kamlesh Madan - José Garcia-Sagredo

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

11:15-11:45 **Darío G. Lupiáñez:** Structural variation in the 3D genomic era: implications for disease and evolution

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

14:30-15:45 Concurrent Sessions

Concurrent Session 1 - 3D chromatin organization and dynamics

Chairs: Jean-Michel Dupont - Darío G. Lupiáñez

14:40-15:05 **Alexandre Reymond:** Genome architecture and diseases: the 16p11.2 paradigm

15:05-15:30 **Mario Nicodemi:** Polymer physics predicts the impact on chromatin 3D structure of disease associated structural variants

15:30-15:45 Selected Abstract

Mireia Solé: Chromosome radial positioning in spermatogenic germ cells from *Mus musculus*

Concurrent Session 2 - Clinical cytogenomics

Chairs: Damien Sanlaville - Orsetta Zuffardi

14:40-15:05 **Nicole de Leeuw:** CNV and diseases: an overview in constitutional diagnostics

15:05-15:30 **Malte Spielmann:** The effect of structural variation in the three-dimensional genome

15:30-15:45 Selected Abstract

Jesper Eisfeldt: From cytogenetics to cytogenomics, whole genome sequencing as a comprehensive genetic test in rare disease diagnostics

15:45-16:15 Coffee break

16:15-17:20 Concurrent Sessions

Concurrent Session 3 - Structural organization of the human genome

Chairs: Joris Vermeesch - Nicole de Leeuw

16:15-16:40 **Megan Y. Dennis:** The role of duplicated genes in human brain evolution and disease

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

17:05-17:20 Selected Abstract

Anna Lindstrand: Cytogenetically visible inversions are formed by multiple molecular mechanisms

Concurrent Session 4 - Human infertility

Chairs: Elisabeth Syk Lundberg **-** Sevilhan Artan

16:15-16:40 **Pierre Ray:** Male infertility in humans, interest of whole exome sequencing

16:40-17:05 **Terry Hassold:** Aneuploidy in humans: new insights into an age-old problem

17:05-17:20 Selected Abstract

Harita Ghevaria: Next generation sequencing detects premeiotic errors in human oocytes and provides evidence of genetic influence

17:20-18:30 Poster session

MONDAY, 8 July 2019

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

Chairs: Felix Mitelman – Roberta Vanni

08:30-09:00 **Fredrik Mertens:** Clonal evolution among different sarcoma subtypes

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

09:30-10:30 Selected Abstracts

09:30-09:45 **Roberto Valli**: Shwachman-Diamond Syndrome, expression arrays, clonal chromosome anomalies

09:45 10:00 **Paola Caria:** Three-Dimensional Telomere Organization in papillary thyroid cancers

10:00-10:15 **Jordi Camps:** Patterns of acquired uniparental disomy reveal biallelic Inactivation of tumor suppressor genes in gastrointestinal cancers and in colorectal advanced adenomas

10:15-10:30 **Sabrina Haslinger:** ZNF384 gene fusions in B-ALL: A report of fifteen Austrian cases secured by systematic FISH and array screening

10:30-11:00 Coffee break

11:00-12:15 Concurrent Sessions

Concurrent Session 5 - Tumor cytogenomics II

Chairs: Claudia Haferlach - Harald Rieder

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

Karla Svobodova: Identification of cryptic aberrations allows more accurate prognostic classification of patients with myelodysplastic syndromes and clonal evolution

Concurrent Session 6 - Animal and plant cytogenomics I

Chairs: Pat Heslop-Harrison - Valerie Fillon

11:00-11:30 **Alain Pinton:** Chromosome rearrangements and meiosis in pig

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

12:00-12:15 Selected Abstract

Alessandra Iannuzzi: Cytogenetic and genomic assays in river buffalo (*Bubalus bubalis*, 2n=50) cows raised in urban and rural areas

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

14:30-15:45 **Plenary session 4 - Chromosomal Imbalances**

Chairs: Mariano Rocchi - Konstantin Miller

14:30-14:55 **Orsetta Zuffardi:** The trisomy legacy: from numerical to structural abnormalities

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

15:30-15:45 Selected Abstract

Aafke Engwerda: Phenotype-genotype analysis in a large cohort of 85 individuals with a terminal 6q deletion

15:45-16:15 Coffee break

16:15-17:30 Concurrent Sessions

Concurrent Session 7 - Animal and plant cytogenomics II

Chairs: Trude Schwarzacher - Leopoldo Iannuzzi

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

Fengtang Yang: Characterisation of complex genomic structure and variation by high-resolution fibre-FISH: an overview

Concurrent Session 8 - Accreditation, quality control and education

Chairs: Konstantin Miller - Albert Schinzel

16:15-16:45 **Thomas Liehr:** European Certification and continuous education of clinical laboratory geneticists working in cytogenetics

16:45-17:15 **Thomas Eggermann:** Next generation sequencing and quality assurance: challenges and opportunities

17:15-17:30 Selected Abstract

Ron Hochstenbach: What should laboratory specialists in clinical genetics know about chromosomes ten years from now?

17:30-18:30 Poster session

18:30 **E.C.A. General Assembly**

20:00 **Conference Dinner of E.C.A.**

Tuesday, 9 July 2019

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

Chairs: Damien Sanlaville - Maria Rosario Pinto Leite

09:00-09:25 **Rossa Chiu:** Cell-free DNA analysis as a tool for "non-invasive cytogenetics"

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

09:50-10:20 Selected Abstracts

09:50-10:05 **Ming Chen:** A silicon-based coral-like nanostructured microfluidics to isolate rare cells in human circulation: validation by SK-BR-3 cancer cell line and its utility in circulating fetal nucleated red blood cells

10:05-10:20 **Celine Dupont:** Six years of molecular cytogenetic in prenatal diagnosis: benefits, lessons and perspectives. A new approach according to observation of ultrasound abnormalities

10:20-10:40 Coffee break

10:40-11:25 **Plenary session 6**

Chair: Jean-Michel Dupont

10:40-11:10 **Frank Pellestor:** Chromoanagenesis: cataclysms behind complex chromosomal rearrangements

11:10-11:25 Selected Abstract

Adriana Di-Battista: Balanced X-autosome translocations and premature ovarian failure are associated with altered expression of growth factors, junction organization and immune pathways

11:25-12:15 **Keynote lecture**

Chair: Mariano Rocchi

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

12:15 **Closing Ceremony**: Mariano Rocchi

Guidance for reporting the interpretation of cytogenomic test results in haematological neoplasms

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Specific recommendations for reporting the results of diagnostic genetic testing have previously been published[1]. However the focus has been on germline testing and although many of the reporting requirements are the same, the considerations for the interpretation of acquired abnormalities are different. General guidance on report format and content is also provided in the recommendations for cytogenomic testing in haematological neoplasms[2]. Review of cytogenomic test reports of laboratories participating in external proficiency testing schemes has highlighted a large variation in the content and detail of the interpretation of the results. This document aims to provide supplementary specific guidance for the format and information to be included in the result interpretation. The interpretation should provide clear concise information and long reports should be avoided as this detracts from the clarity of the results. Genetic testing can be undertaken for diagnostic, prognostic and predictive reasons or at follow up for disease monitoring and this should be addressed in the report.

Relationship of any abnormalities found to the referral reason

The report should include a description of the abnormality identified and the results should be interpreted with respect to the referral reason, or any subsequent information received regarding the patient (e.g. information subsequently communicated by referring clinician). For haematological samples the final diagnosis may or may not be known at time of sample collection and

consequently the referral reason can be a confirmed diagnosis, a presumptive diagnosis, a differential diagnosis or a description of clinical symptoms or findings.

- Where a diagnosis is confirmed, the report should state whether the result is consistent with this diagnosis. It is unhelpful to discuss the association of the abnormality with other disease entities as it may bring the diagnosis into question.
- Where the diagnosis is unconfirmed the report should state whether the result supports the proposed/presumptive diagnosis. When there is a differential diagnosis the report should discuss the result in relation to the different neoplasms considered.
- Where no specific diagnosis has been stated on the referral card, and only clinical information has been provided, it is advised to contact the clinician or the pathology/haematology laboratory for more information before reporting. However, when this is not possible, the report should provide information on its association with specific disease entities.
- In some cases an abnormality may be identified that is inconsistent with the referral reason. It is known that some patients with a haematological malignancy have a second haematological neoplasm and it is not unusual in these cases to identify an abnormal clone containing recurrent abnormalities associated with one or both neoplasms. For example, in a patient referred for CLL, a clone with a deletion 20q may be detected. In such cases it is advised to contact the clinician for further information before reporting. However, when this is not possible, the report should state that the abnormality detected neither supports nor excludes the diagnosis indicated on the referral form and should provide information of any association with other specific disease entities. As a further example, in a patient referred for CLL, two independent clones may be detected, one with a trisomy 12 and one with a monosomy 7. Information pertaining to both abnormallities should be provided on the report.
- In some cases the abnormality detected may be suspected to have a constitutional rather than acquired origin and this should be discussed in the report. Depending on the nature of the abnormality, and considering any reproductive implications for the patient's extended family, confirmation of the patient's constitutional cytogenomic testing can be suggested.
- The most recent WHO (currently 2017) nomenclature should be used in relation to the disease category, where appropriate.

Reporting normal results

The probability of detecting an abnormality depends on the pathology and methodology used. Laboratories should ensure that the most appropriate testing strategy is undertaken. Interpretation of normal test results needs careful consideration.

- Where an abnormal clone cannot be excluded, for example where insufficient metaphases have been obtained or cell enrichment is not optimal (such as low purity of CD138+), the report should include a statement to this effect. In addition, appropriate additional testing should be recommended in the report if not already undertaken.

- Where a prognostic test is performed the report should clearly state that no high risk/ adverse prognostic factors were detected.

Reporting complex results

- Reporting complex test results can be challenging and it is important that the information provided is succinct and clear to the reader of the report.
- The report should summarise the main diagnostic or prognostic abnormalities in a clear statement or in tabular form, if possible near the beginning of the report.
- It should be clear which pertinent prognostic factors have been tested and the report should state whether high risk or established abnormalites have been detected or not detected (for example TP53 deletion or mutation not detected).
- The complex nature of the test result should be highlighted although a full description of all the abnormalities is not required. If included these should be listed elsewhere in the report so as not to detract from the major findings.
- Some pathologies, such as multiple myeloma, demonstrate high intraclonal variability and the FISH signal patterns observed can be very heterogeneous. It is recognised that such cases can be difficult to report and therefore complex signal patterns do not need to be described in detail. However, the report should state an atypical heterogeneous signal pattern was detected showing gene rearrangement, gain, loss or amplification.

Prognostic and predictive information

It is good practice to include prognostic and predictive information in the report. However, it is recognised that local policy and national recommendations need be taken into account when deciding whether to include this information in the report. For example, inclusion of this information may not be required when it will be summarised in an integrated multidisciplinary report or inclusion may be unhelpful in cases where the report is given directly to the patient. In the latter case, information regarding prognosis should be reported with caution as there are always exceptions on a patient level: e.g. cases with CLL and *TP53* aberrations that do perform well, etc. Similarly, inclusion of predictive response to therapy in the report can be unhelpful as choice of adequate therapeutic option by the clinician needs to take into account the patients co-morbidities and other clinical issues. Where laboratory policy is not to include this information in the report the laboratory may choose to make this information, and any new predicitive data, available to the clinician outside the report (telephone, extra fact sheet, link to laboratory website, separate appendix).

Where prognostic information is included or has been specifically requested a prognostic statement must be provided.

- When no informative prognostic genetic bio-markers have been identified this should be stated.
- Where the prognostic information is currently contentious this should be highlighted and referenced in the report.
- Where the abnormality is a predictive marker for response to therapy it is recommended to mention it in the report.
- Prognostic information provided should relate to robust data from multiple publications/international trials/trial protocols or widely accepted prognostic systems exists (e.g. IPSS-R in MDS,

ELN recommendations and MRC prognostic system in AML), or evidence from large randomised control trials of patients undergoing similar relevant treatment or meta-analysis/systematic review of multiple studies.

- Multiple concordant studies can be used and should be referenced.
- Small and isolated studies should not be used to derive prognosis although this information can be given in the report if put in context and referenced.
- It should be noted that the prognostic impact of a distinct marker relates to the specific treatment regimen used in the respective study, e.g. prognosis of APL with t(15;17)(q24;q21) is only favourable if treatment protocols including ATRA and/or arsenic trioxide are used.
- Cytogenomic results are just one component of establishing the patients overall prognosis. For some diseases a combined scoring system is used to establish risk that incorporates risk scores from multiple different tests. For these neoplasms it is recommended to state the cytogenetic risk score in the report to avoid any confusion with the overall risk score which may be different.

Recommendations

Where additional testing, not already under-taken, is required to clarify the significance of the results this should be stated on the report.

Follow up testing

The interpretation of follow up testing must relate the current results to the previous test results and the previous test reference number and sample date should be provided in the report.

Technical reports and provisional reports for discussion at multi discipline meetings

In some circumstances a provisional or abbreviated report is issued prior to discussion at a multi-disciplinary team meeting (MDT) or before the results of other ongoing testing are available. If a purely technical report is issued it should be made clear that the interpretation of the results will be incorporated into a final integrated report.

Where abbreviated results are reported for integration into a MDT-report, the information in the abbreviated MDT result must be consistent with the full test report. The summary report must be authorised by a suitably qualified healthcare scientist. A full report should also be sent to the referring health specialist.

References

1. Claustres M, et al., (2013) Recommendations for reporting results of diagnostic genetic testing (Biochemical, Cytogenetic and Molecular Genetic). On behalf of the ESHG Quality Committee. *Eur. J. Hum. Genet.*, 22:160-170
2. Rack K, et al (2019) European Recommendations and Quality Assurance for Cytogenomic Analysis of Haematological neoplasms. Leukemia https://doi.org/10.1038/s41375-019-0378-z

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Dieter Kotzot
Damien Sanlaville
Joris Vermeesch
Orsetta Zuffardi

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).

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 March 2020.
  - **Goldrain Course in Clinical Cytogenetics** to be held in Goldrain Castle (South Tyrol, Italy) September 2020.
- 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, NÎMES, MARCH 2019

A meeting of the E.C.A. Board of Directors was held on 16th March 2019 at Hotel Vatel, Nîmes.

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

Apologies were received from Sevilhan Artan, Juan Cruz Cigudosa, Kamlesh Madan (First Vice-President), Nicole de Leeuw, and Rosario Pinto Leite.

The President, Mariano Rocchi, opened the meeting at 15.00.

1. The Minutes of the meeting of the ECA Board of Directors of the Association held on 21st September 2018 in Hôpital Cochin, Paris were approved.
2. Reports from Officers
   a. The Secretary General reported on the state of the Association and its administration. The assistance of Dekon continues to be appreciated. There was an extended discussion on how to increase the impact and reach of the EC Congress.
   b. The Treasurer reported on the finances of the Association.
3. 2019 General Assembly Salzburg and Board election
   a. Juan Cruz Cigudosa and Nicole de Leeuw will leave the board. Valid nominations have been received to join the board, and it was agreed that Ron Hochstenbach (Netherlands) and Joan Blanco Rodriguez (Spain) would be added to a list for the Ballot. Sevilhan Artan, Konstantin Miller, and Felix Mitelman, being eligible for re-election, will also be proposed on that list.
4. 2019 ECC Salzburg
   a. The programme and final issues for the ECC 2019 Salzburg were discussed.
   b. By 15th March 2019, 201 abstracts had been received; some more are expected.
   c. It is expected that the present number of commercial sponsors will increase.
   d. Abstract evaluation and procedures for selection of oral presentations was discussed.
   e. Rosario Pinto Leite and José M. Garcia-Sagredo will coordinate the five Poster Prizes awarded for recent or ongoing thesis research, based on recommend-dations of appointed experts.
   f. The menu and arrangements for the Stiegl Keller conference dinner were discussed and approved.
   g. Harald Rieder discussed the organization of the PWG meetings.
5. The future presentation of the E.C.A. conferences and the added value of Membership of the E.C.A. were discussed.
6. Courses

   Jean-Michel Dupont discussed the changes in the "European Advanced Postgraduate Course in Classical and Molecular Cytogenetics 2019" (Nîmes Course) with respect to examinations and certificates for the students.
7. The Board will meet following the Annual General Meeting in Salzburg.

The President closed the meeting at 18.16.

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

PWG: CLINICAL AND MOLECULAR APPROACHES TO CYTOGENETIC SYNDROMES.

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The PWG will hold a meeting during the 12th ECA conference in Salzburg open to all ECC participants: Saturday 6 July, 15:00-16:00, Mozart 2+3 hall (Ground Floor).

PWG: MARKER CHROMOSOMES.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 14:00-15:00, Mozart 4+5 hall (Ground Floor).

PWG: CYTOGENETICS OF HAEMATOLOGICAL MALIGNANCIES.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 15:00-16:00, Mozart 4+5 hall (Ground Floor).

PWG: CANCER CYTOGENETICS, SOLID TUMOR STUDIES.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 16:00-17:00, Mozart 4+5 hall (Ground Floor).

PWG: CYTOGENETIC TOXICOLOGY AND MUTAGENESIS.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 14:00-15:00, Trakl Hall (3th floor).

PWG: ANIMAL, PLANT, AND COMPARATIVE CYTOGENOMICS.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 15:00-17:00, Trakl Hall (3th floor).

PWG: PRENATAL DIAGNOSIS.

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 16:00-17:00, Mozart Hall 2+3 (Ground floor).

PWG: QUALITY ISSUES AND TRAINING IN CYTOGENETICS.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 16:00-17:00, Doppler Hall (4th floor).

PWG: CYTOGENOMICS.

Co-ordinators:

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The PWG will hold a meeting on Saturday 6 July at the Salzburg ECC, 14:00-15:00, Mozart 2+3 Hall (Ground floor).

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 23 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
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jean-michel.dupont@aphp.fr
sylvie.mendez@aphp.fr

BERTRAND
Laboratoire de Biologie Cellulaire et Cytogénétique Moléculaire
Faculté de Médecine Montpellier-Nîmes
Avenue Kennedy
30900 Nîmes, France
tlavabre@univ-montp1.fr
marie.martinez-lucon@umontpellier.fr

Prof. Thierry LAVABRE-

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.

2020 Course provisional program

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 2016.

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.

Nîmes Course 2019

The 23rd Advanced Postgraduate Course in Classical and Molecular Cytogenetics was a great opportunity for us all. On our arrival at Nîmes, Professor Dupont greeted us in the hotel lobby near the medical university and provided us the important notes we needed. He was as welcoming and kind as we could imagine. After 9 days of continuous courses with various topics, we all had a remarkable change of viewpoint in the field of Cytogenetics.

The lectures were given in a way that everyone including physicians and biologists could benefit in his or her own way. Although some topics were presented in less than 2 hours, they gave us a good view of the whole subject. The most important point about this course is that topics are updated each year according to new findings and new prospects.

The lecturers were also very patient and understanding. They were open to all kinds of questions. We exchanged emails and personal contact info for further communication and collaboration.

My favorite part of the course was the city tour. We were so lucky to visit the beautiful city of Nîmes with its great Roman architecture under the sunshine. Our tour guide was very professional and informative.

During the last days of the course Professor Dupont negotiated kindly about our field of interest with those of us who wished to pass the practical course for an advanced diploma. He stated out very clearly what our options are and how he could help us find the proper center for this matter. And finally, making new connections and finding new friends along with learning so many different scientific topics was what I gained from this trip which is greatly appreciated and cherished on my behalf.

Golsa Shekarkhar from Iran

Participants of the 2019 Nîmes course.

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
Impact Factor: 1.455

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