

European  
Cytogeneticists  
Association



# 12<sup>th</sup> **EUROPEAN** CYTOGENOMICS **CONFERENCE**

**6 - 9 JULY 2019**

**SALZBURG CONGRESS  
SALZBURG, AUSTRIA**

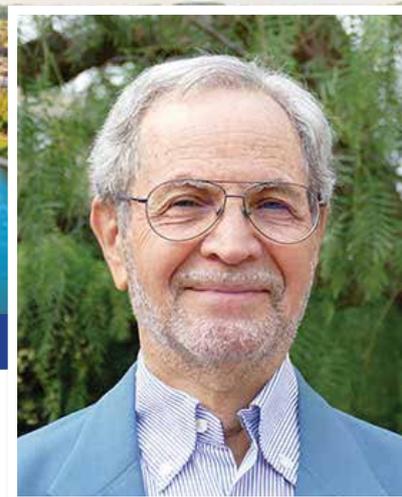


**SECOND ANNOUNCEMENT**

[www.eca2019.com](http://www.eca2019.com)



# Salzburg



## WELCOME FROM THE PRESIDENT

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 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.

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

# WELCOME TO SALZBURG, AUSTRIA

## SALZBURG IN GENERAL

Salzburg is the fourth-largest city in Austria and is also the name of the regional capital of the province of Salzburg. The town was reestablished as Bishopric city on the ruins of the old Roman town of Iuvavum. It came into wealth by mining and trading salt, and much of its architectural splendour as a home of regents is accredited to the work of Wolf Dietrich and his successors. The regency of Salzburg's Prince-Archbishops lasted more than a thousand years. Salzburg's old town is now protected by its status as a UNESCO world heritage site and boasts a long history of cultural tradition. Today the city is home to over 150,000 inhabitants. Wolfgang Amadeus Mozart, the Salzburg Festival and 'The Sound of Music' are just 3 reasons for Salzburg's world fame as a city of music and culture. The baroque city exudes a special kind of magic that even makes working here and attending meetings an enchanting experience.

## CONFERENCE VENUE



### Salzburg Congress

Salzburg Congress opened in 2001 and is recognised as being one of the most modern congress facilities in the whole of Austria. The building offers architectural sophistication and timeless elegance on five floors. The expansive glass frontage offers a sublime view of the world-famous Mirabell garden, past trees that have seen the centuries come and go, and of the baroque city architecture in the historic section of the town.

The venue offers great versatility and the short distances make logistics and navigation child's play.

PEOPLE  
PEOPLE



#### **E.C.A. COMMITTEE**

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**Mariano ROCCHI**

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Second Vice-President

**Konstantin MILLER**

General Secretary

**Jean-Michel DUPONT**

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**Dieter KOTZOT**

**Damien SANLAVILLE**

**Joris VERMEESCH**

**Orsetta ZUFFARDI**

#### **SCIENTIFIC SECRETARIAT**

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#### **ORGANIZING SECRETARIAT**

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**DEKON**  
CONGRESS & TOURISM

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Visit [www.dekon.com.tr](http://www.dekon.com.tr) for more information!

# PRELIMINARY PROGRAMME

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### 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 Vermeesch:** 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  
 10:45-11:00 **Plenary Session 2: 50 Years of 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 **Fredric 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:** Aneuploidy at the Single Cell Level in Tumors

# PRELIMINARY PROGRAMME

## PRELIMINARY PROGRAMME

- 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 Antonarakis:** Chromatin and Single Cell Genomics, to Understand the Gene Dosage Imbalance in Aneuploidies  
12:20 **Closing Ceremony.** Mariano Rocchi

# CALL FOR ABSTRACTS

## CALL FOR ABSTRACTS

Please proceed to the online abstract system via the official conference web page: [www.eca2019.com](http://www.eca2019.com)

### GUIDELINES AND INSTRUCTIONS

- **Abstract submission will close automatically at the end of 3 March 2019.**
- Abstracts must be submitted in English via the official conference website: [www.eca2019.com](http://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.

### 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](mailto: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
- Genome Physiology
- 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.

### 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

# REGISTRATION

## REGISTRATION

Please proceed to the online registration system via the official conference web page [www.eca2019.com](http://www.eca2019.com)

CATEGORY	EARLY BIRD REGISTRATION BEFORE AND ON 17 APRIL 2019	LATE REGISTRATION ON OR AFTER 18 APRIL 2019	ONSITE REGISTRATION	REGISTRATION FEE INCLUDES
<b>E.C.A. Member</b>	€ 300	€ 390	€ 430	Participation in all scientific sessions and conference welcome reception, entrance to the exhibition hall, conference documents and coffee served in the coffee breaks during the scientific programme.
<b>Non-E.C.A. Member</b>	€ 420	€ 490	€ 540	
<b>Technologist (E.C.A. Member)</b>	€ 230	€ 300	€ 340	
<b>Welcome Reception on 6 July 2019</b> (for accompanying person)	€ 30		€ 30	Participation in the conference welcome reception.
<b>Conference Dinner on 8 July 2019</b>	TBA		TBA	per person

- Registration will be confirmed only after full payment has been received. If you have not received a confirmation notice before the conference, please contact the registration secretariat at [registration@eca2019.com](mailto:registration@eca2019.com). Only those participants who complete their registration will have their abstract published.
- Each registered participant is allowed to present one paper and has to be a co-author of the presented paper. It is not permitted to present multiple papers per one registration / participation.
- Registration Fees include VAT.
- The date of receipt of the registration fee will be considered to be the date of registration, even if the registration form has been received before the deadline for early bird or late registration.
- Taking pictures of the presentation slides is not allowed for copyright reasons as the speakers present previously unpublished scientific data and photographs of patients who have given their consent to present but not to distribute the images. In addition, the use of photographic devices such as telephones, tablets, cameras etc. disturbs the speakers and the audience.
- In order to get your invitation letter you have to be registered and complete your payment. After you can login to the online registration system and download your invitation letter.
- Only E.C.A. Technologist Members are eligible for the Technologist Rate. For more information please visit [www.e-c-a.eu](http://www.e-c-a.eu)

### CANCELLATION & REFUND POLICY

Requests for cancellation of registration must be received in writing by the E.C. A. 2019 Registration Secretariat (DEKON) by fax at +90 212 347 63 63, or via e-mail to [registration@eca2019.com](mailto:registration@eca2019.com).

- Requests received before or on **17 April 2019**: a handling fee of € 150 will be deducted from the refund
- Requests received on or after **18 April 2019**: no refund

**Please note that registration refunds will be processed within 30 days after the end of ECA 2019, bank charges will be deducted from the refund.**

## ACCOMMODATION

Odanova is a trusted partner of Dekon for housing options. Please visit the webpage [www.eca2019.com](http://www.eca2019.com) for more details as well as for online booking and payment. Please contact [eca2019@odanova.com](mailto:eca2019@odanova.com) for further assistance.

Please note that Dekon takes no responsibility for reservations made through Odanova. Housing payments should not be made to Dekon registration account.

## TRAVELLING TO SALZBURG

### By plane

Salzburg Airport is on the outskirts of the city - only four kilometers from the city centre. There are direct flights from many European cities, including Berlin, Düsseldorf, Hannover, Hamburg, Cologne/Bonn, Frankfurt, Leipzig-Halle, Amsterdam, Vienna, Paris, London, Manchester, Stockholm, and Zurich. Travellers from outside Europe will find transfers through Düsseldorf, Frankfurt, Vienna, Istanbul and other European hubs.

There are also regular train services from Munich airport.

### Airport transfers

A taxi journey should cost about EURO 15. Alternatively the city bus, number 2, leaves every 10-20 minutes, journey time 20 minutes, ticket price EURO 2.30. Tickets can be bought from the News Corner at the airport or from the driver.

### Munich

The distance between Munich Airport and Salzburg is about 200 km.

Train: Take the S-Bahn S8 to Munich Ostbahnhof (33 min) and a train to Salzburg Hauptbahnhof (1.5 hours). It is also an option to travel via Munich Hauptbahnhof using the S-Bahn S1 but this is a little slower. Total cost: approximately EURO 41.00.

### Vienna

The distance between Vienna Airport and Salzburg is about 300 km.

Train: Take the bus 1187 to Vienna Westbahnhof (45 min) and then the train to Salzburg Hauptbahnhof (2.5-3 hours); total cost approximately EURO 50.00.

### By train

The Salzburg Congress Center is a 10 minute walk from the main railway station, which has connections from across Europe.

### By car

The city is easily accessible by car, and is connected by motorway with Vienna and Munich. Additional travel information can be found on the Salzburg Tourism website.



## GENERAL INFORMATION

### Important Dates & Deadlines

3 March 2019

Deadline for Abstract submission

8 April 2019

Notification of acceptance

17 April 2019

Deadline for Early Registration

### Conference Dates and Language

The conference will take place 6 - 9 July 2019 at the SALZBURG CONGRESS. The official language will be English.

### SALZBURG CONGRESS

Address: Auerspergstraße 6, 5020 Salzburg, Austria

Phone: +43 662 88987 0

Web site: [www.salzburgcongress.at](http://www.salzburgcongress.at)

### Registration Desk

The registration desk will be open at the conference venue from 6 July to 9 July 2019.

### Name Badge & Certificate of Attendance

Upon registration, each participant will receive a name badge which should be worn at all times during the scientific and social programme of the conference. A certificate of attendance will be provided on 6 July 2019 at the registration desk.

### Exhibition

The exhibition area will be located in the SALZBURG CONGRESS. Access to the exhibition will be free for the registered participants. The exhibition will be open throughout the scientific programme.

### CME Accreditation

The 12<sup>th</sup> European Cytogenetics Conference will be accredited by the European Accreditation Council for Continuing Medical Education (EACCME). The EACCME is an institution of the European Union of Medical Specialists (UEMS): <http://www.uems.be>

Further information regarding CME accreditation of the conference will be available on the conference official web site before the conference.

### Fellowships & Prizes

The E.C.A. offers ten Fellowships each to the value of 500 Euro and free registration for early-career researchers (defined by the applicants) presenting posters or talks at the 12<sup>th</sup> European Cytogenetics Conference in Florence, 6 - 9 July 2019.

Applicants for a fellowship should be E.C.A. members and should apply at least two months before the early registration date. The applications will be assessed by the E.C.A. Committee as appropriate. A decision will be taken before the early registration deadline.

Five poster prizes to the value of 250 Euro each will be awarded at the Conference to participants who present the best posters on work derived from a thesis (still to be completed) or from a degree awarded between 2017 and 2019.



## 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 for 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-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](mailto:jean-michel.dupont@aphp.fr)  
[sylvie.mendez@aphp.fr](mailto: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](mailto: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 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 aberrations; 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.**

## 14<sup>th</sup> 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](http://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 includes 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](mailto: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.*





**12<sup>th</sup> EUROPEAN  
CYTOGENOMICS  
CONFERENCE**

**6 - 9 JULY 2019**

**SALZBURG CONGRESS  
SALZBURG, AUSTRIA**

[www.eca2019.com](http://www.eca2019.com)

### Organizing Secretariat



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