



International Society  
for Gastrointestinal Hereditary Tumours



# 7<sup>TH</sup> BIENNIAL *meeting*

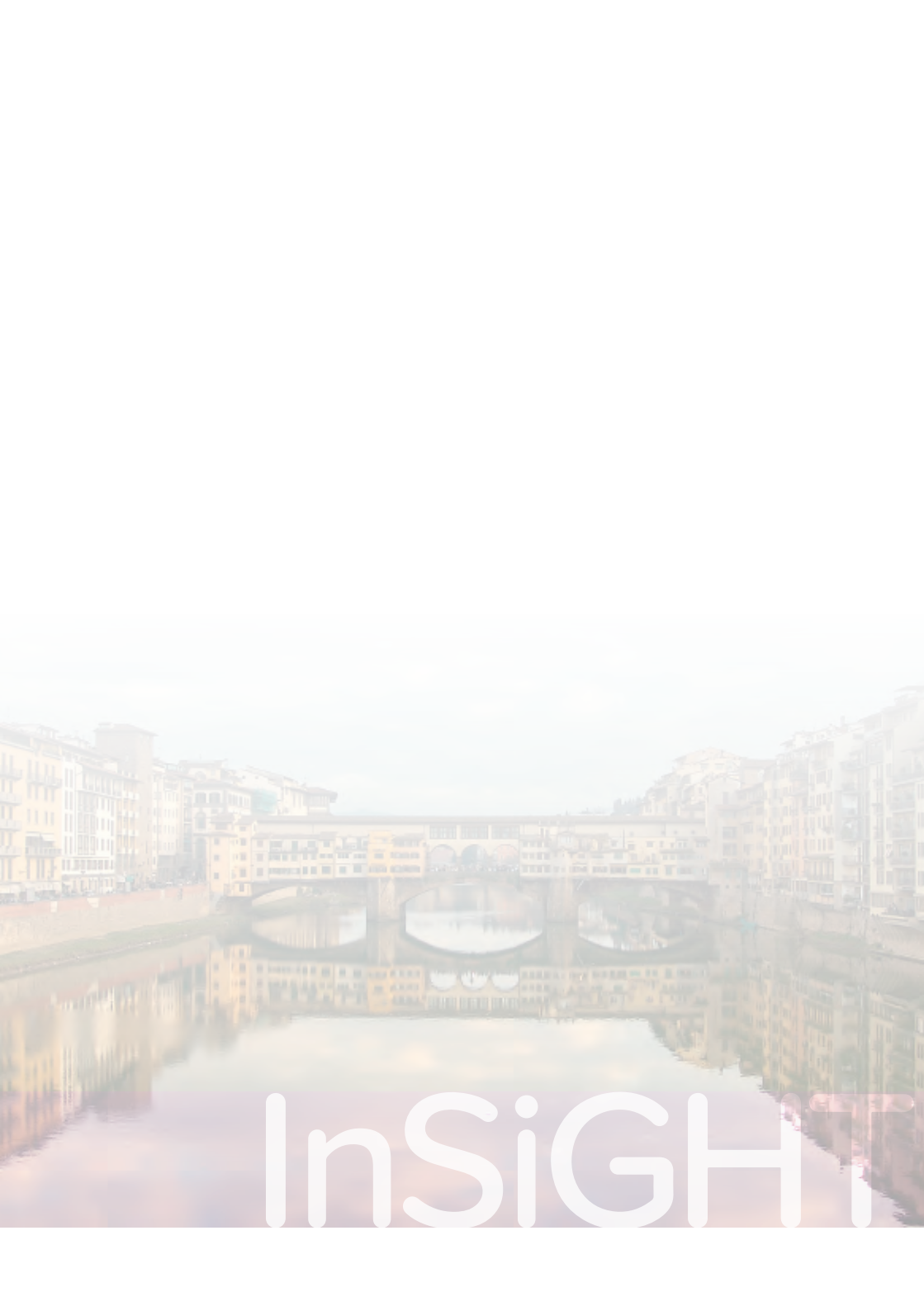
July 5-8 2017

Palazzo dei Congressi

Piazza Adua, 1 - Florence, Italy



FLORENCE 2017



InSiGHT

*Dear Friend and Colleague,*

*it is our great pleasure to extend you a warm welcome to Florence, the Renaissance city and the UNESCO World Heritage Site embedded in the magnificent region of Tuscany, and to the 7th biennial Scientific Conference of InSiGHT.*

*The conference has been designed to provide an innovative and comprehensive overview of the latest research developments in the field of inherited GI cancer. Many distinguished speakers have joined the faculty and will take part in this Conference.*

*The meeting is associated with a rich satellite program, that includes the following events: the postgraduate course "A multidisciplinary approach to Hereditary GI Cancers: from Bench to Bedside", under the umbrella of UEG, ESDO and ESGE, the meeting of the European Hereditary Tumor Group (formerly Mallorca Group) and the "Hereditary Colorectal Cancer Patient & Family Symposium".*

*We hope that you will enjoy the Conference and that your interaction with your colleagues from many different countries will stimulate a creative exchange of ideas and will be personally rewarding. We also hope and trust that you will enjoy your visit to the very beautiful and exciting city of Florence and its Tuscan surroundings.*

Maurizio Genuardi

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Luigi Ricciardiello

A handwritten signature in black ink, appearing to read 'Luigi Ricciardiello', with a highly stylized, cursive script.

## Chairpersons

Maurizio Genuardi (Rome)

Luigi Ricciardiello (Bologna)

## Scientific Committee

Zinfalil Alwi (Kuala Lumpur, Malaysia)

C. Richard Boland (Dallas, USA)

Bernardo Bonanni (Milan, Italy)

Sir John Burn (Newcastle, UK)

Susan Clark (London, UK)

Irene Feroce (Milan, Italy)

Marc Greenblatt (Burlington, USA)

Heather Hampel (Columbus, USA)

Mark Jenkins (Melbourne, Australia)

Sonia Kupfer (Chicago, USA)

Gabriela Moeslein (Düsseldorf, Germany)

Pal Møller (Oslo, Norway)

Cristina Oliani (Montecchio Maggiore, Italy)

Nadia Guglielmina Ranzani (Pavia, Italy)

Giovanni Battista Rossi (Naples, Italy)

Rolf Sijmons (Groningen, Netherlands)

Amanda Spurdle (Brisbane, Australia)

Maria Grazia Tibiletti (Varese, Italy)

Daniela Turchetti (Bologna, Italy)

Emanuele D.L. Urso (Padua, Italy)

Alessandra Viel (Aviano, Italy)

Marco Vitellaro (Milan, Italy)

Magnus von Knebel-Doeberitz  
(Heidelberg, Germany)

## InSiGHT Council Members

Benedito Rossi (São Paulo, Brazil)

Gabriel Capellà (Barcelona, Spain)

James Church (Cleveland, USA)

Ian Frayling (Cardiff, UK)

Maurizio Genuardi (Rome, Italy - Chairman)

Nicoline Hoogerbrugge (Nijmegen, Netherlands)

Patrick Lynch (Houston, USA)

Finlay Macrae (Melbourne, Australia)

Susan Parry (Auckland, New Zealand)

Allan Spigelman (Sydney, Australia)

Thomas Weber (New York, USA)

## InSiGHT Secretariat

Administrative Officer: Susan Clark (London, UK)

Administrative Secretary: Jackie Hawkins (London, UK)

## Venue

Palazzo dei Congressi (Villa Vittoria)

Firenze Fiera

Piazza Adua, 1-50123 Firenze

Ph. +39 05549721

Fax +39 0554973237

Email: [info@firenzefiera.it](mailto:info@firenzefiera.it)

For more information on the venue please visit [www.firenzefiera.it](http://www.firenzefiera.it).

## Climate and Clothing

In July the weather is hot and humid during the whole day. Mean temperature ranges from 25-33 °C.

## Country Dialing Code

+39

## Electrical Voltage

The voltage in Italy is 220 volt.

## Liability and Insurance

The organizers have no responsibility whatsoever for injury or damage involving persons and property during the Conference. Participants are advised to carry their own personal insurance during their stay in Italy.

## Name Badges

All participants and accompanying persons must wear the Conference identification badge in a visible place. Entrance to meeting hall, poster and exhibition areas will not be permitted to any person without badge.

## Official Language

The official Language of the Conference will be English. Simultaneous translation will not be provided.

## Tipping

For taxi and restaurants the service charge is included in the price. You may add a tip at your own discretion to indicate appreciation of exceptionally good service.

## Taxi

Taxi Area is located in front of the Conference venue next to Santa Maria Novella train station.

## Registration Desk

Location: Villa Basement. You will be provided with a name badge when you arrive and/or register at the registration desk.

## Opening Hours

Wednesday July 5: 08.00-18.00

Thursday July 6: 08.00-18.30

Friday July 7: 08.00-18.30

Saturday July 8: 08.00-13.00

## Certificate of Attendance

Certificates of attendance will be available at the registration desk for all participants.

## Exhibition

A commercial exhibition will be held at Passi Perduti in the Villa Basement.

## Opening Hours

Wednesday July 5: 16.00-18.00

Thursday July 6: 08.00-18.30

Friday July 7: 08.00-18.30

Saturday July 8: 08.00-13.00

## Lunch

Lunch buffet will be available upon presentation of lunch tickets in Villa - Passi Perduti.

## Luggage Room

The luggage room is located in the Villa Basement. Valuables cannot be left in the Luggage Room. The Luggage Room is not supervised.

## Opening Hours

Wednesday July 5: 16.00-18.00

Thursday July 6: 08.00-18.30

Friday July 7: 08.00-18.30

Saturday July 8: 08.00-13.00

## Wireless Connection

Free spot in all areas. You can connect only with one device.

Username: insight

Password: flr2017

## Message Board

A message Board is located in the Villa Basement next to the registration desk. Please make sure to remove the message by yourself.

## Social programme

Wednesday July 5: Welcome reception in the garden and Limonaia.

Friday July 7: Conference dinner Roster's Tepidarium

Conference Dinner: Roster's Tepidarium

Via Vittorio Emanuele II, 2

50134, Firenze - Italy

Each participant must bring the dinner ticket and reach the location by his/her own means (directions are provided on the ticket)

## Organizing Secretariat



FASI S.r.l.

Via R. Venuti 73 - 00162 Roma

Tel. 06.97605610 | Fax 06.97605650

**FASI**  
COMUNICAZIONE  
E V E N T I

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



International Society  
for Gastrointestinal Hereditary Tumours



# 7<sup>TH</sup> BIENNIAL *meeting*

Scientific Program



FLORENCE 2017



7<sup>th</sup> Biennial *Meeting*  
of the International Society  
For Gastrointestinal Hereditary Tumours (InSiGHT)



FLORENCE 2017

Florence, Italy, July 5-8, 2017

Wednesday July 5

Auditorium

18.30

**Opening ceremony ■**

Welcome address

**AIFEG (Associazione Italiana per lo Studio della Familiarità ed Ereditarietà dei Tumori Gastrointestinali) lecture**

Introduction: *Guglielmina Nadia Ranzani (Pavia, Italy),  
Benedito Rossi (Sao Paulo, Brazil),*

**“Diet, inflammation and stem cells: trading off regenerative response with colon cancer risk”**

*Riccardo Fodde (Rotterdam, Netherlands)*

19.30

Welcome Cocktail

Villa Vittoria Garden

Thursday July 6

Auditorium

08.00-10.30

**Plenary session**

*Chairs: Gabriel Capellá (Barcelona, Spain),  
Daniela Turchetti (Bologna, Italy)*

**Novel germline and somatic molecular mechanisms**

08.00-08.20

**Lecture:** Novel colorectal polyposis genes

*Stefan Aretz (Bonn, Germany)*

08.20-10.10

Identification of an enhancer region for the MLH1 tumour suppressor gene - Q. Liu, Sydney, Australia

Human intestinal organoids as a model to study Lynch syndrome mutations - C. Heinen, UConn, USA

microRNA-155 is affected by APC mutations and controls canonical Wnt signaling in the colon

A. Prossomariti, Bologna, Italy

Primary constitutional MLH1 epimutations: a focal epigenetic event - E. Damaso, Barcelona, Spain

Comprehensive analysis of the MLH1 promoter region in 480 colorectal cancer patients and 1150 controls reveals one variant inducing a heritable constitutional MLH1 epimutation - M. Morak, Munich, Germany

Tumour DNA methylation signature defines colorectal cancers from biallelic MUTYH mutation carriers  
J. Joo, Melbourne, Australia

Biallelic NTHL1 mutations predispose to a broad variety of tumors with a unique somatic mutational signature  
R. De Voer, Nijmegen, Netherlands

Oxidative DNA damage induces hypomethylation in a compromised base excision repair colorectal tumorigenesis - D. Furlan, Varese, Italy

Comprehensive histological and molecular analysis of PMS2 associated malignancies; a separate entity among MMR deficient tumours?  
S.W. ten Broeke, Leiden, Netherlands

Somatic causes of tumour mismatch repair-deficiency in Lynch-like colorectal and endometrial cancers  
M. Clendenning, Melbourne, Australia

'MMR deficiency first' - re-appraising the pathogenesis of Lynch syndrome cancers  
A. Ahadova, Heidelberg, Germany

EMAST instability is a new feature of pancreatic cancer and is associated with the high methylator phenotype CIMP in colon cancer - M. Kohonen-Corish, Sydney, Australia

10:10-10:30

***The Jeremy Jass lecture***

Introduction: *Finlay Macrae (Melbourne, Australia)*

EMAST: a marker of tumor predisposition and treatment response - *John Carethers (Ann Arbor, USA)*

10:30-11:00

**Coffee break and poster viewing** **Passi Perduti**

11.00-13.00

**Plenary session**

*Chairs: Bernardo Bonanni (Milan, Italy),  
Dennis J. Ahnen (Denver, USA)*

**Risks, surveillance and prevention For GI and non GI cancers**

11.00-11.20

**Lecture:** Chemoprevention in hereditary GI cancer  
*Patrick Lynch (Houston, USA)*

11.20-12.40

Vitamin D and calcium intake in relation to colorectal tumor risk in persons with Lynch syndrome  
F. van Duijnhoven, Wageningen, Netherlands

Aspirin promotes an epithelial phenotype, reduces the stem cell population and inhibits Wnt signalling in colorectal neoplasia - K. Dunbar, Glasgow, UK

Comprehensive population-wide detection of Lynch syndrome in Iceland - S. Haraldsdottir, Iceland

Extra-colonic cancer risk in APC-associated polyposis (FAP): long-term results from the Dutch polyposis registry  
Z. Ghorbanoghli, Leiden, Netherlands

Prevalence of mono-allelic MUTYH carrier status in patients of varied ancestries ascertained for clinical hereditary cancer risk testing - B. Leach, Cleveland, US

Clinical and molecular characteristics in 130 patients with MUTYH-Associated Polyposis: results from an Hereditary Polyposis Registry - M.T. Ricci, Milan, Italy

Risk of colorectal cancer for carriers of a germline mutation in POLE or POLD1 - A. Win, Melbourne, Australia

Implication of germline mutations in POLE and POLD1 in several forms of hereditary cancer  
L. Valle, Barcelona, Spain

The relevance of family history to increased risk of colorectal cancer in Korea - D. Kim, Seoul, Korea

12.40-13.00

***The David Jagelman lecture***

Introduction: *Susan Parry (Auckland, New Zealand)*  
Early onset colorectal cancer: a rising problem  
*Thomas Weber (New York, USA)*

13.00-14.00

Lunch break and poster viewing

Passi Perduti

Room 12

13.00-14.00

InSiGHT Council meeting (only for council members)

Auditorium

14.00-16.00

### Plenary session

*Chairs: Nicoline Hoogerbrugge (Nijmegen, Netherlands),  
Maria Grazia Tibiletti (Varese, Italy)*

#### Next generation sequencing for diagnosis and research

14.00-14.20

**Lecture:** The complexity of colorectal cancer predisposition: challenges for the diagnosis of genetic syndromes - *Thierry Frebourg (Rouen, France)*

14.20-15.40

Exome sequencing identifies biallelic MSH3 germline mutations as recessive subtype of colorectal adenomatous polyposis - I. Spier, Bonn, Germany

Ohio Colorectal Cancer Prevention Initiative  
R. Pearlman, Columbus, USA

Distinct Patterns of Somatic Mosaicism in the APC Gene in Neoplasms From Patients With Unexplained Adenomatous Polyposis - A.M.L. Jansen, Leiden, Netherlands

High frequency of somatic APC mosaicism in patients with genetically unsolved colorectal adenomatous polyposis  
G. Piazzi, Bologna, Italy

Calibrating an in vitro functional assay for the diagnosis of missense variants in mismatch repair genes in Lynch syndrome - M. Drost, Leiden, Netherlands

Towards the validation of DiagMMR – the functional Lynch syndrome carrier test - M. Kansikas, Helsinki, Finland

Whole genome sequencing as a diagnostic tool for Lynch syndrome - B. Pope, Melbourne, Australia

Genetic susceptibility in attenuated adenomatous polyposis  
V. Lorca, Madrid, Spain

Germline variants in homologous recombination (HR)-mediated DNA damage repair genes may contribute to increased CRC susceptibility in FCCTX families  
I. Francisco, Lisbon, Portugal

15.40-16.00 **The Meera Khan lecture**  
Introduction: *Ian Frayling (Cardiff, UK)*  
The complexity of colorectal cancer predisposition:  
challenges for the identification of novel syndromes  
*Laura Valle (Barcelona, Spain)*

16.00-16.30

Coffee break

Passi Perduti

Auditorium

16.30-17.30 **Plenary session** in collaboration with the meeting of Italian  
Medical Oncologists "Highlights from ASCO 2017"  
*Chairs: Roberto Labianca (Bergamo, Italy),  
Eric Van Cutsem (Leuven, Belgium)*

**Focus on novel treatments For genetically  
unstable tumors**

16.30-16.50 Immune response in MSI-H colorectal cancers  
*Magnus von Knebel-Doerberitz (Heidelberg, Germany)*

16.50-17.10 Immunotherapy for GI cancers: current status and  
perspectives - *Erik Hooijberg (Amsterdam, Netherlands)*

17.10-17.30 Research opportunities in unstable tumors  
*Elisa Giommoni (Florence, Italy)*

17.30-19.00 **Plenary session**  
*Chairs: Elke Holinski-Feder (Munich, Germany),  
Minna Nyström (Helsinki, Finland)*

**Focus on novel treatments For genetically  
unstable tumors and miscellaneous**

Therapeutic cellular vaccination prolongs survival  
of MLH1-/- mice by re-activating specific antitumoral immune  
responses - C. Maletzki, Rostock, Germany

Immune evasion in MSI colorectal cancers is related to dense  
infiltration with activated PD-1-positive T cells  
M. Kloor, Heidelberg, Germany

A murine model for proof of concept of a vaccine against  
Lynch syndrome-associated cancers  
M. von Knebel Doeberitz, Heidelberg, Germany

CASP5 (-1) frameshift mutation in unstable colorectal cancers  
is associated with less good prognosis, but could be targeted  
by efficacious anti-tumor cytotoxic lymphocytes  
J.-B. Latouche, Rouen, France

NGS to investigate genetic predisposition to colon cancer in patients with colon cancer over inflammatory bowel disease - G. Biscaglia, San Giovanni Rotondo, Italy

Do we still need surgery for treating small bowel polyps in Peutz-Jeghers Syndrome? A 13-years follow-up cohort  
E. Samaha, Paris, France

Shortage of Mlh1 and chromosomal segregation-specific gene transcripts in colon mucosa signal carcinoma  
M. Pussila, Helsinki, Finland

Improvement of Lynch syndrome diagnostics by NGS based tumor MMR gene analysis: results of 3 peculiar cases - W.N.M. Dinjens, Rotterdam, Netherlands

The Importance of Data Sharing in Classification of Variants in Mismatch Repair Genes - T. Pesaran, Aliso Viejo, USA

A novel APC promoter 1B deletion shows a founder effect in Italian patients with familial adenomatous polyposis  
G.N. Ranzani, Pavia, Italy

Detection of DNA deletions and duplications in next-generation sequencing gene panel data of patients with familial gastro-intestinal cancer  
L.F. Johansson, Groningen, Netherlands

## Friday July 7

### Auditorium

08.00-10.00

### Concurrent session 1

*Chairs: Richard Boland (Dallas, USA),  
Giovanni Battista Rossi (Naples, Italy)*

### Advances in surgery and endoscopy For hereditary GI cancers

08.00-08.20

**Lecture:** Advances in surgery for hereditary GI cancer predisposition: state of the art  
*Gabriela Möselein (Wuppertal, Germany)*

08.20-09.40

Surgical management of serrated polyposis syndrome: a multi-centre experience - S. Rana, London, UK

Do Lynch Syndrome patients benefit from shorter colonoscopy intervals? A pooled analysis of surveillance studies from Germany, the Netherlands, and Finland  
C. Engel, Leipzig, Germany

First report on long term follow up (>10 years) of patients with mild polyposis coli - E.D.L. Urso, Padova, Italy

Duodenal adenomas in patients with multiple colorectal adenomas without germline APC or MUTYH mutations  
F. Kallenberg, Amsterdam, Netherlands

Chromocolonoscopy with indigo carmine facilitates high adenoma detection compared with standard endoscopy in Lynch syndrome in an expert setting  
R. Hüneburg, Bonn, Germany

Five-yearly colonoscopy surveillance in at-risk relatives of patients with familial colorectal cancer type X may lead to delayed detection of advanced adenomas  
M. Ow, Auckland, New Zealand

Colorectal surveillance is also relevant in families who do not fulfil classical phenotypical criteria  
L. J. Lindberg, Danish Registry, Denmark

Gastric Cancer in FAP: a concerning rise in incidence  
C.A. Burke, Cleveland, USA

The Spectrum of Gastric Polyp Pathology in Western Patients with FAP- related Gastric Cancer: clues to Cancer Origin  
C.A. Burke, Cleveland, USA

Factors associated with the development of Gastric Cancer in FAP - P. Leone, Cleveland, USA

09.40-10.00

### **The Sir Ian Todd lecture**

Introduction: *Susan Clark (London, UK)*

Detection and resection of polyps: state of the art  
*Evelien Dekker (Amsterdam, Netherlands)*

## **Green Room**

### **Variant Interpretation Committee meeting**

*Chairs: Elke Holinski-Feder (Munich, Germany)*

*Finlay Macrae (Melbourne, Australia)*

*John-Paul Plazzer (Melbourne, Australia)*

- 08.00-09.50 InSiGHT Classification Criteria Update;  
 New VIC(s) and progress on other gene databases (STK11, POLE,POLD1 etc);  
 InSiGHT VIC - Relationship with ClinGen/ClinVar;  
 Update on LOVD API (Automatic access to database also in relation to Illumina request for data from all LOVD databases);  
 Access to Genomics England data;  
 Privacy and Ethics update;  
 VIC Standard Operating Procedures document;  
 Update on migration to the new InSiGHT database [www.insight-database.org](http://www.insight-database.org);  
 New VIC Variant Review Process;  
 MLH1 variants in certain regions data collection;  
 Classification of Variants
- 09.50-10:00 Combined RNA and protein analyses contribute to the interpretation of exonic variants identified in Lynch syndrome patients - A.Martins (Rouen, France)

10.00-10.30 **Coffee break and poster viewing** **Passi Perduti**

**Auditorium**

- 10.30-11.30 **Plenary interactive session**  
**Challenges in the diagnosis and management of hereditary cancer syndromes:**  
**Clinical case discussions**  
*James F. Church (Cleveland, USA)*  
*Miguel Rodriguez-Bigas (Houston, Usa)*  
*Luigi Laghi (Milan, Italy)*  
 Removal of a complex polyp involving the ileocecal valve  
 E. Gorgun, Cleveland, USA  
 Additional case presentations and discussions



11.30-13.00

## **Plenary session**

*Chairs: Susan Parry (Auckland, New Zealand),  
Marco Vitellaro (Milan, Italy)*

11.30-12.50

## **Clinical aspects, natural history and treatment**

Indications for colectomy and factors influencing time to colectomy in children and young adults with familial adenomatous polyposis (FAP) - S. Sarvepalli, Cleveland, USA

Laparoscopic prophylactic surgery in adolescent patients with Familial Adenomatous Polyposis (FAP). Results of 10 years experience - M. Vitellaro, Milan, Italy

Laparoscopic colectomy and ileorectal anastomosis as prophylaxis for familial adenomatous polyposis is associated with reduced postoperative desmoid risk  
A. Sinha, London, UK

Tailored Surgical Treatment of Duodenal Polyposis in Familial Adenomatous Polyposis Syndrome  
T. Augustin, Cleveland, USA

Patterns of Polyp Histology: Predictors of Peril in the Mucosa  
M. Kalady, Cleveland, USA

Management of Gastric Adenomas in Patients with Familial Adenomatous Polyposis - I. Martin, London, UK

Pouch polyps in FAP – a clinical problem or an endoscopic curiosity? - A. Latchford, London, UK

Including ampullary polyposis staging in the Spigelman Classification: Modifying the modified  
B. Pyle, Christchurch, New Zealand

Outcomes of small intestinal transplant for desmoid tumour  
S. Walton, London, UK

Localization of adenomas after colonic surgery in patients with familial adenomatous polyposis  
V.H. Roos, Amsterdam, Netherlands

Metachronous cancers following segmental or extended colectomy in Lynch syndrome: a systematic review & meta-analysis - K.J. Monahan, London, UK

Risk of metachronous colorectal cancer following colectomy in Lynch syndrome: a systematic review and meta-analysis  
C. Anele, London, UK

13.00-14.00

Lunch break and poster viewing

Passi Perduti

Room 5

13.00-14.00

CaPP3 meeting (upon invitation only)

Auditorium

14.00-16.00

### Plenary session

*Chairs: Annika Lindblom (Sweden),*

*Allan Spigelman (Sydney, Australia)*

#### Less common GI cancer syndromes

14.00-14.20

**Lecture.** Strategies and challenges of CMMRD diagnostics - *Katharina Wimmer (Innsbruck, Austria)*

14.20-15.40

Cancer risks in family members of CMMRD patients – the final results - M. Suerink, Leiden, Netherlands

Universal Gene Panel Testing of Pancreatic Cancer Cases for Breast and Colorectal Cancer Susceptibility Genes  
B.A. Thompson, Salt Lake City, USA

Familial Gastric Cancer & Next-Generation Sequencing: results from a panel of 94 genes in an Italian case series  
G. Tedaldi, Meldola, Italy

Gastric Pathology in CDH1 Mutation Carriers With and Without Family History of Gastric Cancer: Implications for Clinical Management - E. Stoffel, Ann Arbour, USA

Towards Early Detection of Pancreatic Cancer: Applying NGS in the Clinical Setup - G. Rosner, Tel Aviv, Israel

PJS small bowel management – an audit of the last 9 years experience - N. Hodges, London, UK

Exome sequencing identified potential candidate genes for serrated polyposis syndrome - S. Peters, Bonn, Germany

Germline predisposition to Serrated Polyposis Syndrome including evidence for RNF43 as a susceptibility gene  
D. Buchanan, Melbourne, Australia

Germline variants in DNA interstrand-cross link repair genes may contribute to increased susceptibility for serrated polyposis - P. Silva, Lisbon, Portugal

RNF43 in serrated polyposis and serrated polyps  
Y. van Herwaarden, Nijmegen, Netherlands

- 15.40-16.00 ***The Aldred Scott Warthin lecture***  
 Introduction: *Patrick Lynch (Houston, Texas)*  
 Hereditary gastric cancer: beyond CDH1.  
*Nicoline Hoogerbrugge (Nijmegen, Netherlands)*
- 16.00-17.40 **Plenary session**  
*Chairs: Inge Bernstein (Aalborg, Denmark)*  
*Thomas Weber (New York, USA)*
- Counseling and psychosocial issues**
- 16.00-16.20 ***The Kay Neale lecture***  
 Introduction: *Andrew Latchford (London, UK)*  
 Challenging Genetic Counseling Issues in Cancer Genetics  
*Heather Hampel (Columbus, USA)*
- 16.20-17.40 Bridging the missing link between gynaecology and genetics  
 L. Andrews, Randwick, Australia
- Uptake of genetic testing by the children of Lynch syndrome pathogenic variant carriers across three generations - T. Seppala, Helsinki, Finland
- Reactions and attitudes towards a letter with unsolicited risk information - H.V. Petersen, Danish Registry, Denmark
- Health4Families: A behavioral intervention to improve weight and health behaviors in Lynch syndrome families  
 S. Peterson, Houston, USA
- Qualitative interviews to better understand barriers to communication and the information needs of patients with Lynch syndrome - S. Goodman, Plymouth, UK
- Decision For Non-Completion Of Followup Among Patients With Abnormal Screening Test For Hereditary Colorectal Cancer Syndrome - S. Patel, Houston, USA
- The InSiGHT Index: A Novel Metric Designed to Advance the Mission & Goals of InSiGHT - T. Weber, New York, USA
- Post Traumatic Stress Disorder in Patients with Familial Adenomatous Polyposis: a Cause for Concern  
 J. Church, Cleveland, USA
- Psychosocial Symptoms in Patients with Familial Adenomatous Polyposis: How a Chronic Hereditary Cancer Predisposition Syndrome Affects Mental Health and Best Practice for Patient Care - J. Church, Cleveland, USA

Quality of life associated with desmoid tumours  
S. Walton, London, UK

Room 9

16.00-17.40 LOVD Hands-on Course (only for pre-registered attendees)  
Coordinator: John- Paul Plazzer , Melbourne, Australia

Auditorium

17.40-18.40 **Business Meeting**

20.30

Gala Dinner

Roster's Tepidarium

Saturday July 8

Room 9

07.30-08.30 Meeting of InSiGHT Council and CGA board  
members (closed participation)

Auditorium

08.30-10.30 **Plenary session**

*Chairs: Matthew Kalady (Cleveland, USA),  
Finlay Macrae (Melbourne, Australia)*

**Cancer risks in hereditary GI syndromes & Miscellaneous**

08.30-08.50 **Lecture.** Genetic epidemiology of colorectal cancer  
predisposition in 2017  
*Mark Jenkins (Melbourne, Australia)*

08.50-10.10 Extra-colonic cancer risks in PMS2 associated Lynch  
syndrome - S.W. ten Broeke, Leiden, Netherlands

Analysis of the effect of single nucleotide polymorphisms  
on age of onset of colorectal cancer in patients with Lynch  
syndrome (hereditary non-polyposis colorectal cancer)  
L. Pearce, Manchester, UK

Prevalence of germline mutations in FAN1 in familial and  
early-onset colorectal cancers  
D. Buchanan, Melbourne, Australia

Time to relax colonoscopy surveillance recommendations  
for New Zealand MSH6 & PMS2 mutation carriers  
T. Charmels-Watson, Auckland, New Zealand

Screening for Lynch Syndrome through the Canadian  
Colorectal Cancer Consortium - S. Holter, Toronto, Canada

Universal screening for Lynch syndrome among Chinese patients with colorectal cancer - Ding PR, Guangzhou, China

Routine molecular analysis for Lynch syndrome in patients with advanced adenoma or colorectal cancer within a national screening program for colorectal cancer  
A. Goverde, Rotterdam, Netherlands

Raising the age limit for routine MMR testing in CRC from 50 to 70 years improves recognition of new Lynch syndrome families - N. Hoogerbrugge, Nijmegen, Netherlands

Concordance between MSI and IHC in the universal screening tumor program at Barretos' cancer hospital, a public health cancer center in Brazil  
E.I. Palmero, Barretos, Brazil

Burden and profile of somatic mutation in duodenal adenomas from patients with familial adenomatous and MUTYH-associated polyposis - L. Thomas, Cardiff, UK

Linkage Analysis in Familial Non-Lynch Syndrome Colorectal Cancer Families from Sweden  
M. Gonn, Stockholm, Sweden

10:10-10:30

### **The Henry Lynch lecture**

Introduction: *Allan Spigelman (Sydney, Australia)*  
The Prospective Lynch Syndrome Database: providing prospectively observed knowledge for personalized medicine - *Pål Møller (Oslo, Norway)*

**Passi Perduti**

10:30-11:00

**Coffee break**

**Auditorium**

11:00-12:45

### **Plenary session**

*Chairs: Rolf Sijmons (Groningen, Netherlands),  
Elena Stoffel (Ann Arbor, USA)*

### **Collaborative studies and regional groups**

EHTG: the European Hereditary Tumour Group  
*Gabriela Möslein (Wuppertal, Germany)*

CGA: the Collaborative Group of Americas  
*Sonia Kupfer (Chicago, USA)*

Updates from the Latin American Group  
*Benedito Rossi (São Paulo, Brazil)*

The Asia Pacific chapter of InSiGHT  
*Finlay Macrae (Melbourne, Australia),  
Susan Parry (Auckland, New Zealand)*

AIFEG: the Italian Association for the Study of Familial and  
Hereditary Gastrointestinal Tumors  
*Guglielmina Nadia Ranzani (Pavia, Italy)*

Genuris: the European Reference Network on rare genetic  
tumour risk syndromes  
*Nicoline Hoogerbrugge (Nijmegen, Netherlands)*

The optimal dose and duration of daily aspirin as a cancer  
preventive in Lynch Syndrome: the CaPP3 study  
*Sir John Burn (Newcastle, UK)*

EPA for Life  
*Luigi Ricciardiello (Bologna, Italy)*

Worldwide study of cancer risks for Lynch Syndrome:  
International Mismatch Repair Consortium  
*Mark Jenkins (Melbourne, Australia)*

12.45-13.30

### **Plenary session**

*Chairs: Marc Greenblatt (Burlington, USA),  
Michael O. Woods (St. John's, Canada)*

### **Human Genome Variation and the role of InSiGHT**

The Human Variome Project and the BRCA Challenge  
*Sir John Burn (Newcastle, UK)*

The Global Globin 2020 Challenge  
*Zilfalil Alwi (Kuala Lumpur, Malaysia)*

The role and activities of Country Nodes in the Human  
Variome Project  
*Helen M. Robinson (Melbourne, Australia)*

The InSiGHT variant databases  
*John Paul Plazzer, Finlay Macrae (Melbourne, Australia)*

13:30

**Presentation of Familial Cancer Award  
For the best poster:  
Conclusion and Invitation to Auckland 2019**  
*Maurizio Genuardi, Susan Parry, Luigi Ricciardiello*



International Society  
for Gastrointestinal Hereditary Tumours



# 7<sup>TH</sup> BIENNIAL *meeting*

Satellite Events



FLORENCE 2017



European Society of Digestive Oncology



**ESDO/ESGE/InSiGHT under UEG  
Post Graduate Course  
A multidisciplinary approach to Hereditary GI Cancers:  
From Bench to Bedside  
Florence, 5 July 2017  
in conjunction with the biennial scientific meeting of InSiGHT**

**Green Room**

**Date:** July 5, 2017

**Time:** 08.30 - 18.00

**Venue:** Congress Center (Palazzo dei Congressi), Florence, Italy

**Scientific Committee:**

*Guido Costamagna (Rome)*     *Thomas Seufferlein (Ulm)*  
*Evelien Dekker (Amsterdam)*     *Maurizio Genuardi (Rome)*  
*Mário Dinis Ribeiro (Porto)*     *Marco Vitellaro (Milan)*  
*Luigi Ricciardiello (Bologna)*

08.00-08.20     Registration

08.20-08.30     **Welcome message**

*Thomas Seufferlein (Ulm, Germany),  
Mario Dinis Ribeiro (Porto, Portugal),  
Maurizio Genuardi (Rome, Italy),  
Luigi Ricciardiello (Bologna, Italy)*

08.30-08.50

**Introductory lecture:**

**How can we recognize hereditary GI cancers?**

*Gabriel Capellà (Barcelona, Spain)*

08.50-09.05

**Introductory Case Presentations**

*Rodrigo Jover (Alicante, Spain),  
Daniela Turchetti (Bologna, Italy),  
Ricardo Marcos Pinto (Porto, Portugal)*



- 09.05-10.40 **Session 1. Introduction to the hereditary GI cancer syndromes**  
Chairs: *Maurizio Genuardi (Rome, Italy), Luigi Ricciardiello (Bologna, Italy)*
- 09.05-09.25 The algorithm for diagnosing hereditary colorectal cancer syndromes  
*C. Richard Boland (Dallas, USA)*
- 09.25-09.45 Serrated polyposis: a growing problem in clinical practice  
*Evelien Dekker (Amsterdam, The Netherlands)*
- 09.45-10.05 Hereditary gastric cancer  
*Carla Oliveira (Porto, Portugal)*
- 10.05-10.25 Familial pancreatic cancer: risks and molecular markers for early diagnosis  
*Paola Ghiorzo (Genoa, Italy)*
- 10.25-10.40 Discussion
- 10.40-11.10 Coffee break
- 11.10-12.45 **Session 2. Cancer detection and endoscopic removal**  
Chairs: *Antonio Gasbarrini (Rome, Italy), Franco Bazzoli (Bologna, Italy)*
- 11.10-11.30 Endoscopic screening and surveillance of the lower GI tract: new technologies and scoring systems  
*Patrick Lynch (Houston, USA)*
- 11.30-11.50 Surveillance of the upper GI tract in the colorectal cancer syndromes  
*Andrew Latchford (London, UK)*
- 11.50-12.10 Endoscopic tools for screening and surveillance of hereditary gastric cancer: are they valuable?  
*Mario Dinis Ribeiro (Porto, Portugal)*
- 12.10-12.30 Endoscopic approaches for prevention and early detection of pancreatic cancer  
*Guido Costamagna (Rome, Italy)*
- 12.30-12.45 Discussion
- 12.45-13.15 **Special lecture**  
Chemoprevention for hereditary gastrointestinal tumor syndromes  
*Sir John Burn (Newcastle, UK)*

13.15-14.15	Lunch
14.15-15.30	<p><b>Session 3. Chemotherapy in the hereditary GI cancer syndromes</b>  Chairs: <i>Cristina Oliani (Vicenza, Italy), Thomas Seufferlein (Ulm, Germany)</i></p>
14.15-14.35	Treatment options for hereditary colon cancer syndromes <i>Wolff Schmiegel (Bochum, Germany)</i>
14.35-14.55	New chemotherapeutic approaches to gastric cancers <i>Eric van Cutsem (Leuven, Belgium)</i>
14.55-15.15	Chemotherapy for pancreatic cancer in hereditary syndromes <i>Thomas Seufferlein (Ulm, Germany)</i>
15.15-15.30	Discussion
15.30-16.45	<p><b>Session 4. Surgical treatment and prevention</b>  Chairs: <i>Mario Dinis Ribeiro (Porto, Portugal), Thomas Weber (New York, USA)</i></p>
15.30-15.50	Tailored preventive surgery for the hereditary colorectal cancer syndromes <i>Marco Vitellaro (Milan, Italy)</i>
15.50-16.10	Prophylactic gastrectomy for HDGC: counseling roadmaps <i>Susan Parry (Auckland, New Zealand)</i>
16.10-16.30	Surgery for hereditary pancreatic cancer: which and when? <i>Claudio Bassi (Verona, Italy)</i>
16.30-16.45	Discussion
16.45-17.45	<p><b>Case Presentations</b>  Case presentations led by  <i>Rodrigo Jover (Alicante, Spain), Ricardo Marcos Pinto (Porto, Portugal), Daniela Turchetti (Bologna, Italy)</i>  Discussants:  <i>Bernardo Bonanni (Milan, Italy), John Carethers (Ann Arbor, USA), Susan Clark (London, UK), Finlay Macrae (Melbourne, Australia), Rocco Maurizio Zagari (Bologna, Italy)</i></p>
17.45-18.00	Final remarks

**EHTG (European Hereditary Tumor Group)  
Meeting (Workshop Format) 2017  
Florence, 5 July 2017**

**in conjunction with the biennial scientific meeting of InSiGHT**

**Date:** July 5th, 2017

**Time:** 08.30 - 17.15

**Venue:** Congress Center (Palazzo dei Congressi), Florence, Italy

**Room 9**

08.00-08.30 **Registration**

08.30-10.00 **Workshop Invasive treatment  
Surgery and Endoscopy**

10.00-10.15 Coffee Break

10.15-12.15 **Workshop Non-invasive treatment  
Immunotherapy  
Chemoprevention**

12.45-13.15 **Special lecture  
Chemoprevention for hereditary  
gastrointestinal tumor syndromes  
*Sir John Burn (Newcastle, UK)***

**Green room**

13.15-14.15 Lunch

14.15-14.30 **Update PLSD version 1 and version 2 - *Pål Møller***

14.30-14.50 **Clinically relevant results (vs 1) - *Toni Seppälä***

14.50-15.10 **Update Gene and Gender guidelines - *Gabriela Möslein***

15.10-15.30 **Next projects - new ideas - Moderated discussion**

15.30-16.00 **Governance issues, Steering board  
*G. Capella, J. Sampson, G. Möslein*  
(only for contributors)**

16.15-17.15 **Structure. Steering committee, next meeting**

18.30 **InSiGHT Opening Ceremony  
and Welcome Cocktail**

**Auditorium**

**MEETING POST-ASCO**  
**Highlights From ASCO 2017: quale impatto nella pratica clinica?**  
**Florence, 6 July 2017**  
**in conjunction with the biennial scientific meeting of InSiGHT**

**Data:** 6 luglio, 2017

**Orario:** 09.00 - 17.30

**Sede:** Palazzo dei Congressi, Firenze, Italia

**Comitato scientifico:** *Silvia Cottini, Daniela Barana, Zora Baretta*

Room 9

- 09.00-09.45      Iscrizioni
- 09.45-10.00     Presentazione dei lavori - *Francesco Di Costanzo, Roberto Labianca, Cristina Oliani*

**Sessione I** - Moderatori: *Roberto Labianca, Cristina Oliani*

- 10.00-10.30     Novità nel tumore del polmone - *M. Garassino*
- 10.30-11.00     Novità nei tumori urologici - *T. Sava*
- 11.00-11.30     Novità nei tumori mammari - *C. Tondini*
- 11.30-12.00     Novità nei tumori ginecologici - *K. Lorusso*
- 12.00-12.30     Novità sui tumori cerebrali - *A. Brandes*
- 12.30-13.00     Novità nel melanoma - *M. Mandalà*
- 13.00-14.30     Lunch

**Sessione II** - Moderatori: *Francesco Di Costanzo, Cristina Oliani*

- 14.30-15.00     Novità nei tumori coloretali - *F. Di Costanzo*
- 15.00-15.45     Lettura magistrale: GI non colorectal cancer: has the revolution begun? - *E. Van Cutsem*

Auditorium

**Plenary session, in collaboration with InSiGHT "Focus on novel treatments For genetically unstable tumors"**

*Roberto Labianca & Eric Van Cutsem*

- 16.00-16.20     Immune response in MSI-H colorectal cancers  
*Magnus von Knebel-Doerberitz (Heidelberg, Germany)*
- 16.20-16.40     Principles of immunotherapy for colorectal cancer  
*Erik Hooijberg (Amsterdam, Netherlands)*
- 16.40-17.00     Research opportunities in unstable tumors  
*E. Giommoni (Firenze, Italy)*
- 17.00-17.30     Chiusura dei lavori e compilazione ECM

Hereditary Colon Cancer Patient & Family Symposium  
Satellite Event to the 7th Biennial InSiGHT Meeting  
Florence, 8 July 2017

Room 9

**Date:** 8 July, 2017

**Registration:** 09.30 - 10.30

**Symposium presentations:** 10.30-17.30

**Venue:** Palazzo dei Congressi, Firenze, Italia



7<sup>TH</sup> BIENNIAL *meeting*

July 5-8 2017

Palazzo dei Congressi

Piazza Adua, 1- Florence, Italy



FLORENCE 2017



International Society  
for Gastrointestinal Hereditary Tumours



# 7<sup>TH</sup> BIENNIAL *meeting*

Posters



FLORENCE 2017

**SESSION A - 6<sup>TH</sup> JULY 2017 FROM 08.00 TO 16.30****THE ASSOCIATION OF LOW PENETRANCE VARIANTS IN DNA REPAIR GENES WITH COLORECTAL CANCER: A SYSTEMATIC REVIEW AND META-ANALYSIS**

Kevin Monahan - Family History of Bowel Cancer Clinic, West Middlesex University Hospital, Chelsea and Westminster Hospitals NHS Trust, London, UK

**YOUR InSiGHT**

Jackie Hawkins - InSiGHT Secretariat, Harrow, UK

**RUPTURE OF SUPERIOR MESENTERIC ARTERY ANEURYSM IN DESMOID PATIENTS**

Xhileta Xhaja - Colorectal Surgery, Cleveland, Ohio, USA

**PREVALENCE OF LYNCH SYNDROME AND LYNCH-LIKE SYNDROME AMONG PATIENTS WITH COLORECTAL CANCER IN A JAPANESE HOSPITAL-BASED POPULATION**

Hidetaka Eguchi - Division of Translational Research, Research Center for Genomic Medicine, Saitama Medical University, Hidaka, JAPAN

**RISK FACTORS FOR ADVANCED DUODENAL AND AMPULLARY ADENOMATOSIS IN FAMILIAL ADENOMATOUS POLYPOSIS: A CLINICAL AND MOLECULAR PROSPECTIVE STUDY IN A BRAZILIAN POPULATION**

Fábio Guilherme - Campos Gastroenterology Department Hospital das Clínicas, University of São Paulo Medical School, Sao Paulo, BRAZIL

**ATTENUATED FAP - HOW SHOULD IT BE DEFINED AND WHAT ARE THE CLINICAL OUTCOMES?**

Chukwuemeka Anele - The Polyposis Registry, St Mark's Hospital, Harrow, UK  
Department of Cancer and Surgery, Imperial College, London, UK

**EPIGENETIC AND GENETIC CHANGES IN NONMALIGNANT TISSUES OF OVARIES AND ENDOMETRIUM AS POSSIBLE PRECURSORS IN OVARIAN MULTISTEP TUMORIGENESIS**

Anni Niskakoski - Medical and Clinical Genetics, Helsinki, FINLAND

**SEARCHING FOR A DRIVER MUTATION: LINKAGE ANALYSIS IN A SERRATED POLYPOSIS FAMILY**

Salman Rana - St. Mark's Hospital, London, UK

**THE QUESTION OF DESMOIDS AND POUCH SURGERY IN PATIENT WITH FAP: HOW MANY UPDATES DOES IT TAKE?**

Xhileta Xhaja - Colorectal Surgery, Cleveland, Ohio, USA

**NON-SYNDROMIC COLORECTAL CANCER (CRC) IN COLOMBIA -SOUTH AMERICA**

Mabel Bohorquez - University of Tolima, Ibagué, COLOMBIA

**THE INFLAMMATORY POTENTIAL OF THE DIET AND COLORECTAL TUMOR RISK IN PERSONS WITH LYNCH SYNDROME**

Jesca Brouwer - Department of Human Nutrition, Wageningen University & Research Wageningen, THE NETHERLANDS

**LYNCH SYNDROME-RELATED TUMORS IN THE PATIENTS WITH JAPANESE LYNCH SYNDROME**

Chiaki Saita - Tokyo Metropolitan Cancer and Infectious diseases Center Komagome Hospital, Tokyo, JAPAN

**COLORECTAL POLYPS AND CANCERS IN THE FOLLOW-UP OF ASYMPTOMATIC INDIVIDUALS WITH CONSTITUTIONAL MISMATCH REPAIR GENE MUTATIONS**

Luca Roncucci - Department of Diagnostic and Clinical Medicine, and Public Health, University of Modena and Reggio Emilia, Modena, ITALY

**IDENTIFICATION OF MISMATCH REPAIR DEFICIENT TUMOURS USING A MOLECULAR INVERSION PROBE BASED SEQUENCING ASSAY OF SHORT HOMOPOLYMER REPEATS**

Richard Gallon - Institute of Genetic Medicine, Newcastle University, Newcastle-upon-Tyne, UK

**A LARGE PROPORTION OF PATIENTS WITH LYNCH SYNDROME STILL UNDERGO GENETIC SCREENING IN CONNECTION TO THEIR DIAGNOSIS OF CANCER**

Sophie Walton - Bernstedt Center of Digestive diseases, Karolinska University Hospital, Solna, Stockholm, SWEDEN

**EUROPEAN REFERENCE NETWORK ON RARE GENETIC TUMOUR RISK SYNDROMES (ERN GENTURIS)**

Nicoline Hoogerbrugge - Human Genetics, Nijmegen, THE NETHERLANDS

**SURGICAL STRATEGY IN PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSIS**

Leonardo Duraes - Cleveland Clinic Foundation, Cleveland, Ohio, USA

**IS UNIVERSAL TUMOR TESTING FOR LYNCH SYNDROME TRULY UNIVERSAL?**

Sonia Kupfer - University of Chicago, Chicago, USA

**PRELIMINARY EXPERIENCE ON TESTING HEREDITARY GASTROINTESTINAL TUMOR RISK USING MULTI-GENE PANEL**

Jack Ji - Color Genomics Burlingame, CA, USA

**HEREDITARY COLORECTAL CANCER WITH DIAGNOSIS AFTER AGE 50 – CHARACTERISTICS OF THE “LATE ONSET” PHENOTYPE, MEF**

Mef Nilbert - HNPCC register, Copenhagen University, Copenhagen, THE NETHERLANDS

**LYNCH SYNDROME ASSOCIATED TO A RECURRENT CLASS 5 VARIANT IN THE 3'UTR OF THE MSH6 GENE**

Alessandra Viel - Functional onco-genomics and genetics, CRO Aviano, National Cancer Institute, Aviano, ITALY

**COMMON GENETIC VARIATION NEAR CDKN1A IS ASSOCIATED WITH COLORECTAL CANCER SUSCEPTIBILITY IN MALE PMS2 MUTATION CARRIERS**

Sanne W. ten Broeke - Clinical Genetics - Leiden University Medical Center, Leiden, DENMARK

**A POTENTIAL CHEMOPREVENTATIVE FOOD FOR FAMILIAL ADENOMATOUS POLYPOSIS: THE AUSFAP STUDY**

Finlay Macrae - Colorectal Medicine and Genetics, Royal Melbourne Hospital Parkville, Melbourne, AUSTRALIA



**PROGNOSTIC OR PREDICTIVE IMPACTS OF COLORECTAL CANCERS WITH LYNCH SYNDROME BY THE ADMINISTRATION OF 5-FU-BASED ADJUVANT CHEMOTHERAPY**

Nagahide Matsubara - Central Hospital, Amagasaki, JAPAN

**ENDOSCOPIC FOLLOW UP CAN SELECT PATIENTS FOR MULTI-GENE TESTING IN ATTENUATED ADENOMATOUS POLYPOSIS WITH NO APC OR MUTYH IDENTIFIED MUTATIONS**

Mara Fornasari - Oncological Gastroenterology, Aviano, ITALY

**CLINICAL AND MOLECULAR CHARACTERIZATION OF LATIN AMERICAN PATIENTS SUSPECTED TO HAVE LYNCH SYNDROME**

Mev Dominguez-Valentin - Department of Tumor Biology, Institute for Cancer Research, Oslo University Hospital, Oslo, Norway, Oslo, NORWAY

**ASSESSING HOW REDUCED EXPRESSION LEVELS OF THE MISMATCH REPAIR GENE PMS2 AFFECT REPAIR EFFICIENCY**

Mariann Kasela - Department of Biosciences, Helsinki, FINLAND

**GERMLINE MUTATIONS IN MICRORNAS IN HEREDITARY COLORECTAL CANCER**

Pilar Mur - Hereditary Cancer Program, Catalan Institute of Oncology, IDIBELL and CIBERONC, Barcelona, SPAIN

**A NOVEL TOOL FOR QUANTITATIVE ANALYSIS OF MICROSATELLITE MUTATIONS**

Alexej Ballhausen - Department of Applied Tumor Biology, Institute of Pathology, University Hospital Heidelberg, Heidelberg, GERMANY

**LYNCH SYNDROME FAMILY WITH CO-OCCURRENCE OF GERMLINE PATHOGENIC SPLICE SITE MUTATIONS OF MSH2 AND MSH6**

Ingrid Vogelaar - Department of Medicine (Oncology), Stanford Cancer Institute, Stanford University, Stanford, CA, USA

**IDENTIFICATION OF TYPE AND FREQUENCY OF TUMORS IN CHILEAN FAMILIES WITH LYNCH SYNDROME**

Francisco López-Köstner - Unidad De Coloproctología, Clínica Las Condes Santiago-Chile, CHILE

**ATTITUDE AND KNOWLEDGE TOWARDS GENETIC COUNSELING AND TESTING FOR THE MAIN COLORECTAL CANCER PREDISPOSITION SYNDROMES: A SURVEY AMONG PRIVATE PHYSICIANS IN SWITZERLAND**

Valeria Viassolo - Unit of Oncogenetics and Cancer Prevention, Division of Oncology, Geneva University Hospitals Geneva, SWITZERLAND

**UNIVERSAL MSI SCREENING FOR LYNCH SYNDROME IN JAPANESE COLORECTAL CANCER PATIENTS**

Tatsuro Yamaguchi - Tokyo Metropolitan Cancer and Infectious diseases Center Komagome Hospital, Tokyo, JAPAN

**POLYMERASE PROOFREADING ASSOCIATED POLYPOSIS: A PHENOTYPIC UPDATE**

Vicky Cuthill - The Polyposis Registry, St. Mark's Hospital, London, UK

**NATURAL HISTORY OF COLONIC POLYPOSIS IN CHILDREN AND YOUNG ADULTS WITH FAMILIAL ADENOMATOUS POLYPOSIS (FAP)**

Shashank Sarvepalli - Medicine Institute, Cleveland Clinic, Cleveland, Ohio, USA

**RAPID ASSESSMENT OF THE ACCEPTABILITY OF A WEB-BASED, PROVIDER-MEDIATED TOOL FOR COMMUNICATING CANCER GENETIC RISK FINDINGS TO AT-RISK RELATIVES**

Mala Pande - Gastroenterology, Hepatology & Nutrition, Houston, USA

**A MUCINOUS COMPONENT IN COLORECTAL CANCER WITH LYNCH SYNDROME**

Soichiro Natsume - Department of surgery, Bunkyo-ku, JAPAN

**COLORECTAL CANCER IN A 16-YEAR-OLD FEMALE WITH A DE NOVO MSH2 MUTATION**

Steven H. Erdman - Div of Gastroenterology, Nationwide Children's Hospital Columbus, Ohio, USA

**MOLECULAR FEATURES OF MICROSATELLITE INSTABILITY-HIGH COLORECTAL ISSUES WITH CLASSIFICATION OF AN MSH2 MISSENSE VARIANT ASSOCIATED WITH AN ATYPICAL MUIR-TORRE SYNDROME PHENOTYPE**

Alessandro Vaisfeld - Istituto di Medicina Genomica, Università Cattolica del Sacro Cuore; UOC Genetica Medica, Fondazione Policlinico Universitario A. Gemelli, Rome, ITALY

**ROUTINE MISMATCH REPAIR IMMUNOHISTOCHEMISTRY ANALYSIS AS A VALUABLE METHOD TO IMPROVE LYNCH SYNDROME'S DIAGNOSIS AMONG WOMEN WITH ENDOMETRIAL CANCER**

Alessandra Livi - Oncologic Gynecology Unit, S. Orsola-Malpighi Hospital, Bologna, Italy, Bologna, ITALY

**ROLE OF GENETIC TESTING IN SURGICAL DECISION MAKING FOR PATIENTS WITH HNPCC**

Leonardo Duraes - Cleveland Clinic Foundation, Cleveland, Ohio, USA

**BETA-2 MICROGLOBULIN AS A PROGNOSTIC BIOMARKER IN MISMATCH REPAIR DEFICIENT COLORECTAL CANCER**

Lyndsay Pearce - Central Manchester Foundation Trust Manchester, UK

**THE RISK OF METACHRONOUS COLORECTAL CANCER IN LYNCH SYNDROME: A COMPARISON OF RIGHT COLECTOMY AND LEFT COLECTOMY**

Misato Takao - Department of surgery, Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital Tokyo, JAPAN

**A 14-YEAR-OLD FEMALE WITH ADVANCED RECTAL CANCER, DIAGNOSED WITH LYNCH SYNDROME**

Mizuki Takatsu - Genetic Medicine and Services, National Cancer Center Hospital Tokyo, JAPAN

**ENDOSCOPIC RISK FACTORS FOR DUODENAL CANCER IN FAMILIAL ADENOMATOUS POLYPOSIS (FAP)**

Carol A. Burke - Sanford R. Weiss MD Center for Hereditary Colorectal Neoplasia, Cleveland Clinic, Cleveland, Ohio, USA

### THE INFLUENCE OF MUTATION CLASS ON THE SEVERITY OF LYNCH-RELATED CANCERS

Edenir Inêz Palmero - Molecular Oncology research Center, Barretos Cancer Hospital, Barretos, BRAZIL

### SOMATIC MLH1 MUTATION SCREENING IS SUPERIOR TO BRAFV600E MUTATION TESTING TO IDENTIFY SPORADIC MLH1-METHYLATED COLORECTAL CARCINOMAS

Christophe Rosty - Genetic Epidemiology Laboratory, University of Melbourne Centre for Cancer Research, Melbourne, AUSTRALIA

### TREATMENT OF DESMOID DISEASE IN A 51-YEAR-OLD MALE WITH FAMILIAL ADENOMATOUS POLYPOSIS (FAP)

Isabelle Danos - Cancer Genetics Unit, St Vincent's Hospital, Sydney, AUSTRALIA

### CLINICAL CHARACTERISTICS OF GASTRIC NEOPLASM IN PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSIS

Ryoko Shimizuguchi - Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital Gastroenterology, Bunkyo-ku, JAPAN

### NOVEL DUPLICATION OF SCG5/GREM1 AS A CAUSE FOR HEREDITARY MIXED POLYPOSIS SYNDROME (HMPS)

Louise Lynagh - Cancer Genetics Unit, St Vincent's Hospital Sydney, AUSTRALIA

### THE INSIGHT DATABASE – TRANSFORMATION FOR THE GENOMICS ERA

John-Paul Plazzer - Colorectal Medicine and Genetics, The Royal Melbourne Hospital Melbourne, AUSTRALIA

### CHARACTERISATION OF MISMATCH REPAIR VARIANTS SUBMITTED TO THE INTERNATIONAL MISMATCH REPAIR CONSORTIUM (IMRC)

Jeanette C. Reece - Centre for Epidemiology and Biostatistics, Melbourne, AUSTRALIA

### CDH1 PROMOTER REGION AS COFACTOR FOR A DIFFUSE GC OCCURRING AT YOUNG AGE

Valli De Re - Bio-Proteomics Facility, Aviano, ITALY

### COMPARISON OF LONG-TERM SURVIVAL BETWEEN TOTAL COLECTOMY WITH ILEO-RECTAL ANASTOMOSIS (IRA) AND PROCTOCOLECTOMY WITH ILEO-POUCH ANAL ANASTOMOSIS (IPAA) IN PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSIS (FAP): WITHIN AN OLD QUESTION. A REGISTRY-BASED

Ilaria Ardoino - Unit of Medical Statistics, Biometry and Bioinformatics - Fondazione IRCCS Istituto Nazionale dei Tumori Milan, ITALY

### USE OF TECHNOLOGY AND SOCIAL MEDIA TO FACILITATE FAMILY COMMUNICATION IN LYNCH SYNDROME

Susan Peterson - MD Anderson Cancer Center Houston, Texas, USA

### ENDOSCOPIC SURVEILLANCE OF THE UPPER GASTROINTESTINAL TRACT IN LYNCH SYNDROME PATIENTS

Robert Hüneburg - Department of Internal Medicine I, Center for hereditary tumor syndromes, University Hospital, Bonn, GERMANY

**PANEL TESTING FOR HEREDITARY COLORECTAL CANCER**

Emanuela Lucci-Cordisco - Institute of Genomics Medicine-Catholic University, Rome, ITALY

**FROM PHENOTYPE TO GENOTYPE: AN ATYPICAL LYNCH SYNDROME**

Daniela Barana - Oncology Unit, ULSS8 Berica, Arzignano, ITALY

**A NOVEL RISK SNP IN THE CRC RISK LOCUS ON CHROMOSOME 9q22**

Jessada Thutkawkorapin - Department of Molecular Medicine and Surgery, Karolinska Institute, Stockholm, SWEDEN

**PHENOTYPE IN A MALE INDIVIDUAL CARRYING BOTH MLH1 AND EPCAM GERM-LINE PATHOGENIC VARIANTS**

Marie Met-Domestici - Unit of Oncogenetics and Cancer Prevention, Division of Oncology, Geneva University Hospitals, Geneva, SWITZERLAND

**STANDARDIZATION OF THE IN VITRO MISMATCH REPAIR ASSAY USED FOR THE FUNCTIONAL CHARACTERIZATION OF VARIANTS IN MISMATCH REPAIR GENES**

Maribel González-Acosta - Hereditary Cancer Program, Catalan Institute of Oncology, ICO-IDIBELL and CIBERONC, Hospitalet de Llobregat, Barcelona, SPAIN

**A MUTATION IN CHECK2 CAUSING SIX DIFFERENT NEOPLASTIC TUMORS IN A SINGLE INDIVIDUAL**

Elizabeth Half - Gastroenterology, Haifa, ISRAEL

**DEEP INTRONIC SEQUENCING OF MUTATION-NEGATIVE LYNCH SYNDROM PATIENTS**

Anke Marie Nissen - MGZ - Medizinisch Genetisches Zentrum Munich, GERMANY

**THE MSH2 EXON 5 DELETION (C.792+8\_943-450DEL) IS A FOUNDER MUTATION IN PORTUGUESE LYNCH SYNDROME FAMILIES WITH A CENTER-SOUTH ANCESTRY**

Inês Francisco - Molecular Pathobiology Research Unit - Portuguese Institute of Oncology Francisco Gentil, EPE Lisbon, PORTUGAL

**RESULTS OF THE NGS ANALYSIS IN PATIENTS WITH SUSPECTED HEREDITARY COLORECTAL CANCER DUE TO FAMILY HISTORY**

Benedito Rossi - Sirio Libanes Hospital, São Paulo, BRAZIL

**TO NMD AND BEYOND: IDENTIFYING MUTATIONS IN THE 3' TERMINUS OF MISMATCH REPAIR GENES**

Brandon Smith - Ambry Genetics, Aliso Viejo, CA, USA

**MICROSATELLITE INSTABILITY-LOW AND LOSS OF HETEROZIGOSITY IN 2P MAY UNDERLIE INCREASED SUSCEPTIBILITY FOR FAMILIAL RECTAL CANCER**

Cristina Albuquerque - Molecular Pathobiology Research Unit- Portuguese Institute of Oncology Lisbon Francisco Gentil, Lisbon, PORTUGAL

**POLYMERASE PROOFREADING-ASSOCIATED POLYPOSIS (PPAP) AND LYNCH SYNDROME: NOVEL AND PREVIOUSLY REPORTED VARIANTS IN POLE AND POLD1 EXONUCLEASE DOMAIN AND COMBINED PMS2 AND POLE GERMLINE VARIANTS IN A PATIENT WITH EARLY ONSET COLORECTAL CANCERS**

Martina Calicchia - Istituto di Medicina Genomica, Università Cattolica del Sacro Cuore, Roma, ITALY

**MUTATIONAL SPECTRUM OF 35 LYNCH SYNDROME ARGENTINEAN FAMILIES:  
MORE TO SAY ABOUT LYNCH SYNDROME IN SOUTH AMERICA**

Marina Antelo - Oncology Section, Hospital of Gastroenterology "Dr.C.B. Udaondo"  
Buenos Aires, ARGENTINA

**DEFINING NEW COLORECTAL CANCER SYNDROMES IN A POPULATION BASED  
COHORT**

Anne Keränen - Laboratory Medicine, Karolinska Institute, Stockholm, SWEDEN

**ANALYSIS OF THE VARIANT MLH1 C.588+5G>C IN BRAZILIAN PATIENTS WITH  
LYNCH SYNDROME: IS THIS REALLY A VUS?**

Ivana Lucia de Oliveira Nascimento - Nucleo de Oncologia da Bahia, Salvador, BRAZIL

**INTERVAL AND SCREEN DETECTED GASTRO ESOPHAGEAL AND COLON CANCER  
AMONG LYNCH SYNDROME PATIENTS WITH A VERIFIED MUTATION**

Zohar Levi - Gastroenterology Petach Tikva, ISRAEL

**PREVALENCE OF ESOPHAGEAL REFLUX DISEASE AND INTERVAL ESOPHAGEAL  
SQUAMOUS CELL CANCER IN FANCONI ANEMIA PATIENTS PARTICIPATING IN AN  
ACTIVE SURVEILLANCE PROGRAM**

Zohar Levi - Gastroenterology Petach Tikva, ISRAEL

**WHAT IS THE BENEFIT OF EXTENDING IHC PRE SCREEN FOR ALL PATIENTS UP  
TO 60 YEARS OF AGE TO DETECT PATIENTS AT RISK OF LYNCH SYNDROME?**

Doug Speake - Department of Colorectal Surgery Western General Hospital Edinburgh

**CULTURAL PERSPECTIVE ON THE MANAGEMENT OF FAMILIAL ADENOMAS  
POLYPOSIS IN SAUDI ARABIA; A FAMILY CASE STUDY**

Sagal Ahmed Shire - RN, MPH Colorectal Registrar, King Faisal Specialist Hospital &  
Research Centre (KFSH&RC), SAUDI ARABIA

**IMPACT OF AN OPTIMIZED COLONOSCOPIC SCREENING PROGRAM FOR PATIENT  
WITH LYNCH SYNDROME. SIX YEARS' RESULTS OF A SPECIALIZED FRENCH  
NETWORK**

Elia Samaha - European Georges Pompidou Hospital, Paris, FRANCE

**ANALYSIS OF PATIENT MUTATIONS AND KNOCKOUT VARIANTS ON MUTYH  
EXPRESSION**

Guido Plotz - Zentrum der Inneren Medizin, Universitätsklinikum, Frankfurt,  
GERMANY

**STUDY OF RARE VARIANTS IN MISMATCH-REPAIR GENES IN SLOVAK POPULATION  
USING DATA FROM WHOLE GENOME TEST FOR PREGNANT WOMEN**

Tomas Szemes - Geneton s.r.o., Bratislava, SLOVAKIA

**SESSION B - 7<sup>TH</sup> JULY 2017 FROM 08.00 TO 18.00****MDM2 T309G POLYMORPHISM AND RISK OF COLORECTAL CANCER**

Kevin Monahan - Family History of Bowel Cancer Clinic, West Middlesex University Hospital, Chelsea and Westminster Hospitals NHS Trust, London, UK

**WORKING WITH CLINICIANS TO IDENTIFY BARRIERS TO FAMILY CANCER CLINIC REFERRALS FOR PATIENTS AT HIGH RISK OF LYNCH SYNDROME**

Rachel Williams - Hereditary Cancer Clinic, Prince of Wales Hospital Sydney, AUSTRALIA

**BONE MARROW TRANSPLANT AND GENETIC TESTING**

Patricia McGinty - The Polyposis Registry, London, UK

**WHOLE TRANSCRIPTOME ANALYSIS IN EARLY-ONSET COLORECTAL CANCER AS AN EFFECTIVE METHOD TO IDENTIFY RESPONSIBLE GENE**

Kenji Fujiyoshi - Department of Surgery, Kurume University, Fukuoka, JAPAN

**MOLECULAR PROFILE OF FAMILIAL COLORECTAL CARCINOMA (CRC) IN COLOMBIA**

Mabel Bohorquez - University of Tolima, Ibague, COLOMBIA

**WHAT ARE THE ADVANTAGES TO PAEDIATRIC CARE OF HAVING A PAEDIATRIC POLYPOSIS SERVICE?**

Jackie Hawkins - The Polyposis Registry, Harrow, UK

**CHARACTERIZATION OF A NOVEL POLD1 MISSENSE FOUNDER MUTATION IN A SPANISH POPULATION**

Rosario Ferrer-Avargues - Genética Molecular. Hospital Universitario Elche, Elche. SPAIN

**BURDEN AND NEEDS OF PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSIS (FAP)**

Philip P. Becker - Tillotts Pharma AG, Medical Affairs, Rheinfelden, SWITZERLAND

**EVALUATION OF AN ONLINE FAMILY HISTORY TOOL FOR IDENTIFYING HEREDITARY AND FAMILIAL COLORECTAL CANCER**

F.G.J. Kallenberg - Department of gastroenterology and hepatology, Academic Medical Center Amsterdam, THE NETHERLANDS

**A CASE OF COLORECTAL CANCER IN NIJMEGEN BREAKAGE SYNDROME**

Tara Clinick - General Surgery, Capital & Coast District Health Board, Wellington, NEW ZELAND

**EXOME SEQUENCING APPROACH FOR IDENTIFICATION OF HIGH-RISK GENETIC VARIANTS IN FCC-X FAMILIES**

Trinidad Caldes - Molecular Oncology. IdISSC, Madrid, SPAIN

**DEEP SEQUENCING OF LYNCH SYNDROME TUMORS HIGHLIGHTS EPIGENETIC EVENTS**

Noora Porkka - Department of Medical and Clinical Genetics, Helsinki, FINLAND

**HIGH PREVALENCE OF MSH6 AND PMS2 PATHOGENIC VARIANTS IN UNSUSPECTED LYNCH SYNDROME**

Jessica Stoll - University of Chicago Medicine, Chicago, USA

**UNIVERSAL SCREENING FOR LYNCH SYNDROME IN GASTRIC CANCER PATIENTS**

Kentaro Yamashita - Department of Gastroenterology and Hepatology, Sapporo Medical University, Sapporo, JAPAN

**DNA METHYLATION CHANGES IN LYNCH SYNDROME-ASSOCIATED NORMAL COLONIC MUCOSA, ADENOMAS AND CARCINOMAS**

Satu Maki-Nevala - Department of Medical and Clinical Genetics, University of Helsinki, Helsinki, FINLAND

**IMMUNOHISTOCHEMISTRY AND MSI OF TUMORS IN NORWEGIAN PMS2 MUTATION CARRIERS**

Eli Marie Grindedal - Department of Medical Genetics, Oslo University Hospital, Oslo, NORWAY

**INCIDENCE OF COLORECTAL AND ENDOMETRIAL CANCER IN INDIVIDUALS FROM FAMILIES SUSPECTED OF HAVING LYNCH SYNDROME: A PROSPECTIVE STUDY OF THE GERMAN HNPCC CONSORTIUM**

Christoph Engel - Institute for Medical Informatics, Statistics and Epidemiology, University of Leipzig, Leipzig, GERMANY

**CLINICOPATHOLOGICAL CHARACTERISTICS OF INITIAL COLORECTAL CANCER AND METACHRONOUS COLORECTAL CANCER DEVELOPMENT IN JAPANESE MISMATCH REPAIR GENE MUTATION CARRIERS**

Masashi Miguchi - Department of Surgery, Hiroshima, JAPAN

**LYNCH SYNDROME NETWORK IN LOMBARDY REGION**

Maria Grazia Tibiletti - Dpt of Pathology, Ospedale di Circolo, ASST Settelaghi Varese, ITALY

**CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY: ON THE SPOT DIAGNOSIS?**

Manon Suerink - Clinical Genetics, Leiden, THE NETHERLANDS

**ANNUAL COLONOSCOPY COULD NOT PREVENT SURGERY FOR COLORECTAL CANCER IN A FAMILY WITH MLH1 AND MSH2 MUTATION CARRIER**

Kohji Tanakaya - Department of Surgery, National Hospital Organization, Iwakuni Clinical Center Iwakuni, JAPAN

**YIELD OF UNIVERSAL TESTING FOR MISMATCH REPAIR PROTEIN DEFICIENCY IN 2077 COLORECTAL CARCINOMAS**

Christophe Rosty - Envoi Pathology, Brisbane, AUSTRALIA

**IMPACT OF THE TYPE OF SURGERY AT THE TIME OF DIAGNOSIS OF COLORECTAL CANCER IN PATIENTS WITH LYNCH SYNDROME**

Matilde Navarro - Hereditary Cancer Program, Catalan Institute of Oncology L'Hospitalet, Barcelona, SPAIN

**EXOME SEQUENCING IDENTIFIED POTENTIAL CAUSATIVE CANDIDATE GENES FOR UNEXPLAINED COWDEN SYNDROME**

Sophia Peters - Institute of Human Genetics, University of Bonn, GERMANY

**IDENTIFICATION OF GENETIC BIOMARKERS FOR CLINICAL MANAGEMENT OF FAMILIAL COLORECTAL TUMORS**

Mev Dominguez-Valentin - Department of Tumor Biology, Institute for Cancer Research, Oslo University Hospital, Oslo, NORWAY

### CLINICOPATHOLOGICAL FEATURES OF APC MUTATION-NEGATIVE FAP PATIENTS

Hiroyuki Ishida - Department of Surgery, Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital, Tokyo, JAPAN

### IDENTIFICATION OF A NEW POLE GERMLINE VARIANT WHEN EVALUATING THE DIAGNOSTIC UTILITY OF POLE AND POLD1 HOTSPOTS SCREENING IN A 600 POLYPOSIS PATIENT COHORT

Daniel Rueda - Hereditary Cancer Genetics Laboratory, Biochemistry Dept., 12 de Octubre University Hospital. Colorectal Cancer Research Group. i+12 Research Institute. Madrid, SPAIN

### NEXT GENERATION SEQUENCING INCREASES THE SENSITIVITY IN FAMILIAL ADENOMATOUS POLYPOSIS AND MUTYH-ASSOCIATED POLYPOSIS TESTING: A LABORATORY AND CLINICAL PERSPECTIVE

Elizabeth Alexander - Clinical Genetics Service, Manchester Centre for Genomic Medicine, Manchester, UK

### THE OPTIMAL DOSE AND DURATION OF DAILY ASPIRIN AS A CANCER PREVENTIVE IN LYNCH SYNDROME

John Burn - Institute of Genetic Medicine, Newcastle University Newcastle upon Tyne, UK

### EXPERIENCE OF AN E-LEARNING COURSE ABOUT GENETIC COUNSELING IN HEREDITARY CANCER FOR LATIN AMERICA HEALTH PROFESSIONALS

Francisco López-Köstner - Unidad De Coloproctología, Clínica Las Condes Santiago-Chile, CHILE

### ELUCIDATING THE MOLECULAR BASIS OF MSH2-DEFICIENT TUMORS BY COMBINED GERMLINE AND SOMATIC ANALYSIS

Marta Pineda - Hereditary Cancer Program, Catalan Institute of Oncology-IDIBELL L'Hospitalet de Llobregat, Barcelona, SPAIN

### FINDING YOURSELF IN FRONT OF THE MIRROR: DEVELOPMENT OF A THEORY REGARDING PRESYMPTOMATIC GENETIC TESTING IN YOUNG ADULTS

Lea Godino - Department of Medical and Surgical Sciences, Bologna University Hospital Authority St.Orsola-Malpighi Polyclinic, Genetic Unit, Italy, Bologna, ITALY

### IDENTIFICATION AND FUNCTIONAL CHARACTERIZATION OF A CDH1 MISSENSE VARIANT IN THREE UNRELATED FAMILIES WITH GASTRIC CANCER

José Perea - 12 de Octubre University Hospital, Madrid, SPAIN

### RELATIONSHIP BETWEEN LYNCH SYNDROME AND INFLAMMATORY BOWEL DISEASE- ASSOCIATED COLORECTAL CANCER

José Perea - 12 de Octubre University Hospital, Madrid, SPAIN

### COWDEN SYNDROME CAUSED BY LOW LEVEL MOSAICISM

Inbal Barnes Keda - Genetics, ISRAEL

### CHROMOENDOSCOPY IN COMBINATION WITH RANDOM BIOPSIES DOES NOT IMPROVE DETECTION OF GASTRIC CANCER FOCI IN CDH1 MUTATION POSITIVE PATIENTS

Robert Hüneburg - Department of Internal Medicine, Center for hereditary tumor syndromes, University Hospital Bonn, Bonn, GERMANY



**CANCER AND MUTATION SPECTRUM OF LYNCH SYNDROME IDENTIFIED BY UNIVERSAL TUMOR SCREENING**

Kiwamu Akagi - Saitama Cancer Center Molecular Diagnosis and Cancer Prevention, Saitama, JAPAN

**ASSESSING THE CLINICAL VALIDITY OF GENES IMPLICATED IN HEREDITARY COLORECTAL CANCER AND POLYPOSIS USING THE CLINGEN FRAMEWORK**

Matthew Ferber - Mayo Clinic Rochester, MN, USA

**GERMLINE MUTATIONS IN INDIVIDUALS REFERRED FOR "MULTIPLE" POLYPS**

Elena Stoffel - University of Michigan Ann Arbor, MI, USA

**DIFFICULTY AND IMPORTANCE OF THE GENETIC COUNSELLING IN HEREDITARY DIFFUSE GASTRIC CANCER**

José Perea - 12 de Octubre University Hospital Madrid, SPAIN

**MSH2 3'UTR AS PREDICTIVE BINDING SITE FOR THE MIRNA HSA-MIR-137**

Francesca Duraturo - Dept of Molecular Medicine and Medical Biotechnologies, University of Naples "Federico II" Naples, ITALY

**THE INVESTIGATION OF MALIGNANT TUMORS ASSOCIATED WITH PEUTZ-JEGHERS SYNDROME IN JAPAN**

Hidetaka Kawamura - Department of Surgery, Southern TOHOKU General Hospital, Department of Surgery, Tokyo Metropolitan Cancer and Infectious Diseases Center Komagome Hospital - Fukushima, Tokyo, JAPAN

**CLINICOPATHOLOGICAL FEATURES OF SERRATED POLYPOSIS SYNDROME IN JAPAN: SINGLE CENTER EXPERIENCE**

Akinari Taka - Departments of Gastroenterology, Tokyo Metropolitan Komagome Hospital, Tokyo, JAPAN

**CLINICAL AND MOLECULAR CHARACTERIZATION OF PATIENTS WITH COLORECTAL CANCER DIAGNOSED AT YOUNG AGES: EXPERIENCE OF A REFERENCE CANCER CENTER IN BRAZIL**

Edenir Palmero - Molecular Oncology research Center, Barretos Cancer Hospital, Brazil, Barretos, BRAZIL

**CHARACTERISTICS OF PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSIS IN BRAZIL - FIRST RESULTS OF THE BARRETOS CANCER HOSPITAL REGISTRY**

Edenir Inêz Palmero - Molecular Oncology research Center, Barretos Cancer Hospital, Brazil, Barretos, BRAZIL

**HOW COST EFFECTIVE IS ENDOSCOPIC SUBMUCOSAL DISSECTION IN REMOVING LARGE COLORECTAL POLYPS? A COMPARISON WITH LAPAROSCOPIC COLECTOMY**

Emre Gorgun - Cleveland Clinic Colorectal Surgery Department, Cleveland, Ohio, USA

**CLINICAL DIAGNOSTIC PMS2 TESTING IN A SINGLE COMMERCIAL LABORATORY: CHALLENGES IN GROSS DELETION/DUPLICATION ANALYSIS DUE TO PMS2CL INTERFERENCE**

Selvi Palaniappan - Ambry Genetics Aliso Viejo, CA, USA

**SCREENING INDIVIDUALS AT INCREASED RISK FOR PANCREATIC CANCER USING BIENNIAL CONTRAST MRI**

Ayelet Borgida - Zane Cohen Centre for Digestive Diseases, Mount Sinai Hospital  
Toronto, CANADA

**UNRAVELING THE SERRATED NEOPLASIA PATHWAY: IMMUNOHISTOCHEMISTRY AND NEXT-GENERATION SEQUENCING REVEAL UNIQUE FEATURES OF MLH1-PROFICIENT EARLY DYSPLASTIC SESSILE SERRATED LESIONS**

Arne Bleijenberg - of Gastroenterology and Hepatology, Academic Medical Centre  
Amsterdam, THE NETHERLANDS

**A NOVEL MUTATIONAL SIGNATURE CHARACTERIZES COLORECTAL CANCER IN PATIENTS WITH MUTYH-ASSOCIATED POLYPOSI**

Alessandra Viel - Functional Onco-Genomics and Genetics, CRO Aviano National  
Cancer Institute, Aviano, ITALY

**INITIAL RESULTS OF MULTIGENE PANEL TESTING FOR HEREDITARY COLORECTAL CANCER**

Roberto Piva - Medical Genetics Unit, AOU Città della Salute e della Scienza Torino, ITALY

**DIAGNOSTIC YIELD OF A COMPREHENSIVE GENE PANEL FOR HEREDITARY TUMOR SYNDROMES**

Isabel Spier - Institute of Human Genetics and Center for Hereditary Tumor Syndromes,  
University of Bonn, Bonn, GERMANY

**RISK ASSESSMENT AND SURVEILLANCE OF GASTROINTESTINAL CANCER IN PATIENTS WITH LI-FRAUMENI SYNDROME**

Kazuo Tamura - Kindai University Higashiosak, JAPAN

**HIGH PREVALENCE OF BENIGN AND MALIGNANT THYROID DISEASE IN PATIENTS WITH FAMILIAL ADENOMATOUS POLYPOSI (FAP)**

Robert Hüneburg - Department of Internal Medicine I, Center for hereditary tumor  
syndromes, University Hospital Bonn, GERMANY

**MOLECULAR CHARACTERIZATION OF A FAMILY WITH AN UNUSUAL POLYPOSI PHENOTYPE CAUSED BY A NOVEL POLE MUTATION**

Ester Castellsagué - Hereditary Cancer Program, Catalan Institute of Oncology, IDIBELL  
and CIBERON, Barcelona, SPAIN

**WORLDWIDE STUDY OF CANCER RISKS FOR LYNCH SYNDROME: INTERNATIONAL MISMATCH REPAIR CONSORTIUM (IMRC)**

Mark Jenkins - Centre for Epidemiology and Biostatistics, The University of Melbourne,  
Melbourne, AUSTRALIA

**SYSTEMATIC SCREENING FOR LYNCH SYNDROME IN A COHORT OF COLORECTAL AND ENDOMETRIAL CANCER PATIENTS IN SWITZERLAND: THE SYSSYL STUDY**

Aurélie Ayme - Division of Clinical Pathology - Unit of Oncogenetics and Cancer,  
Geneva, SWITZERLAND

**CLASSIFICATION OF A NOVEL MLH1 VARIANT THROUGH FUNCTIONAL ANALYSIS, CONFIRMING A NOVEL SPLICE-SITE MUTATION MECHANISM**

Bianca Desouza - North West Thames Regional Genetics Service, London, UK

**ELUCIDATING THE CLINICAL SIGNIFICANCE OF TWO PMS2 MISSENSE VARIANTS COEXISTING IN A FAMILY FULFILLING HEREDITARY CANCER CRITERIA**

Maribe González - Acosta Hereditary Cancer Program, Catalan Institute of Oncology, ICO-IDIBELL and CIBERONC, Hospitalet de Llobregat, Barcelona, SPAIN

**GENETIC CHARACTERIZATION OF FAMILIES FROM SIRIO LIBANES HOSPITAL HEREDITARY CANCER REGISTRY, SAO PAULO, BRAZIL**

Benedito Rossi - Sirio-Libanés Hospital, São Paulo, BRAZIL

**PERFORMANCE OF IMMUNOHISTOCHEMICAL TEST ON COLORECTAL CANCERS TO IMPROVE LYNCH SYNDROME IDENTIFICATION**

Stefano Signoroni - Unit of Hereditary Digestive Tract Tumors, Preventive-Predictive Department - Fondazione IRCCS Istituto Nazionale dei Tumori Milan, ITALY

**CONCORDANCE OF MULTI-GENE PANEL TESTING WITH PRIOR MICROSATELLITE INSTABILITY AND IMMUNOHISTOCHEMISTRY ANALYSES**

Carin Espenschied - Ambry Genetics, Aliso Viejo, CA, USA

**CONTRIBUTION OF MUTYH GERMLINE MUTATIONS TO EARLY ONSET NON-POLYPOSIS COLORECTAL CANCER**

Cristina Albuquerque - Molecular Pathobiology Research Unit (UIPM), Portuguese Institute of Oncology Francisco Gentil, EPE Lisbon

**CANCER PREVALENCE IN LYNCH SYNDROME PATIENTS: PRELIMINARY RESULTS OF A MULTICENTER NATION-WIDE STUDY**

Ariadna Sánchez - Hospital Clinic Barcelona, Barcelona

**FINDING THE GENETIC CAUSE IN SUSPECTED LYNCH SYNDROME PATIENTS**

Anne Maria Lucia Jansen - Department of Pathology, Leiden University Medical Center Leiden, THE NETHERLANDS

**GENETIC ALTERATIONS OF WNT PATHWAYS IN FAMILIAL VERSUS SPORADIC POLYPS**

Maria Cristina Curia - Department of Medical, Oral and Biotechnological Sciences, G. d'Annunzio' University Chieti, ITALY

**EUROPEAN cDNA MISMATCH REPAIR WORKING GROUP: COMPARISON OF DISTINCT STRATEGIES AND RECOMMENDATIONS FOR BEST PRACTICE IN RNA SPLICING ANALYSIS**

Monika Morak - Medizinische Klinik und Poliklinik IV, Campus Innenstadt, Klinikum der Universität München, Munich, Germany and MGZ – Medizinisch Genetisches Zentrum, Munich, GERMANY

**AN INTERDISCIPLINARY MODEL FOR GASTROINTESTINAL HEREDITARY SYNDROMES SUPPORTED BY NEXT GENERATION SEQUENCING TECHNOLOGY**

Paola Carrera - Division of Genetics and Cell Biology, Unit of Genomics for Human Disease Diagnosis and Laboratory of Clinical Molecular Biology, IRCCS San Raffaele Scientific Institute, Milan, ITALY

**DESCRIPTIVE ANALYSES OF MMR GENE MUTATION CARRIERS AT A SINGLE INSTITUTION IN BRAZIL**

Henrique Galvao - Oncogenetics Department, Barretos, BRAZIL

**UNIVERSAL SCREENING FOR LYNCH SYNDROME IN PATIENTS WITH ENDOMETRIAL CANCER**

Charlotte Kvist - Lautrup Department of Clinical Genetics, Aalborg University Hospital Aalborg, DENMARK

**COMPREHENSIVE VARIANT ANALYSES INCLUDING WHOLE GENOME SEQUENCING IN HEREDITARY COLORECTAL CANCER SYNDROMES**

Anna Rohlin - Department of Clinical Pathology and Genetics, Göteborg, SWEDEN

**WHOLE-EXOME SEQUENCING OF SIX RELATIVES WITH SERRATED POLYPOSIS**

Yasmijn van Herwaarden - Department of Gastroenterology and Hepatology Nijmegen, THE NETHERLANDS

**ANTICIPATION IN SWEDISH LYNCH SYNDROME FAMILIES**

Jenny von Salomé - Department of Molecular Medicine and Surgery, Karolinska Institutet, Stockholm, SWEDEN

**IDENTIFICATION OF FUNCTIONALLY DELETERIOUS GERMLINE GALNT12 VARIANTS IN A POPULATION-BASED COHORT OF INCIDENT COLORECTAL CANCER CASES**

Daniel R. Evans - Discipline of Genetics, Faculty of Medicine, Memorial University of Newfoundland St. John's, CANADA

**EARLY ONSET COLORECTAL CANCERS: CLINICOPATHOLOGICAL, MOLECULAR AND ONCOLOGICAL FEATURES**

Luigi Eusebi - Università di Bologna, ITALY

**EARLY ONSET COLORECTAL CANCERS VS SPORADIC COLORECTAL CANCERS: A CLINICOPATHOLOGICAL AND MOLECULAR COMPARISON**

Luigi Eusebi - Università di Bologna, ITALY

**SCREENING PATHWAYS FOR LYNCH SYNDROME: A SYSTEMATIC REVIEW OF THE EXISTING PATHWAYS AND A COST-EFFECTIVENESS ANALYSIS IN ITALY**

Lucci Cordisco Emanuela - Università Cattolica del Sacro Cuore, Roma, ITALY

**SPONTANEOUS REMISSION OF METACHRONOUS NEOPLASTIC LESIONS IN A LYNCH SYNDROME PATIENT: EFFICIENT IMMUNE REACTION DECIPHERED BY MODERN MEDICINE?**

Luigi Laghi - Humanitas Research Hospital, Milano, ITALY

**HAPLOTYPE ASSOCIATION ANALYSIS OF CANCER RISK SUSCEPTIBILITY IN A SWEDISH POPULATION**

Wen Liu - Karolinska Institutet Solna, SWEDEN

**HEREDITARY CANCER SYNDROMES: FROM GENETICS TO ENDOSCOPY, THE MULTIDISCIPLINARY APPROACH**

Annalisa Tortora - Gastroenterology, Fondazione Policlinico Universitario "A. Gemelli", Rome, ITALY



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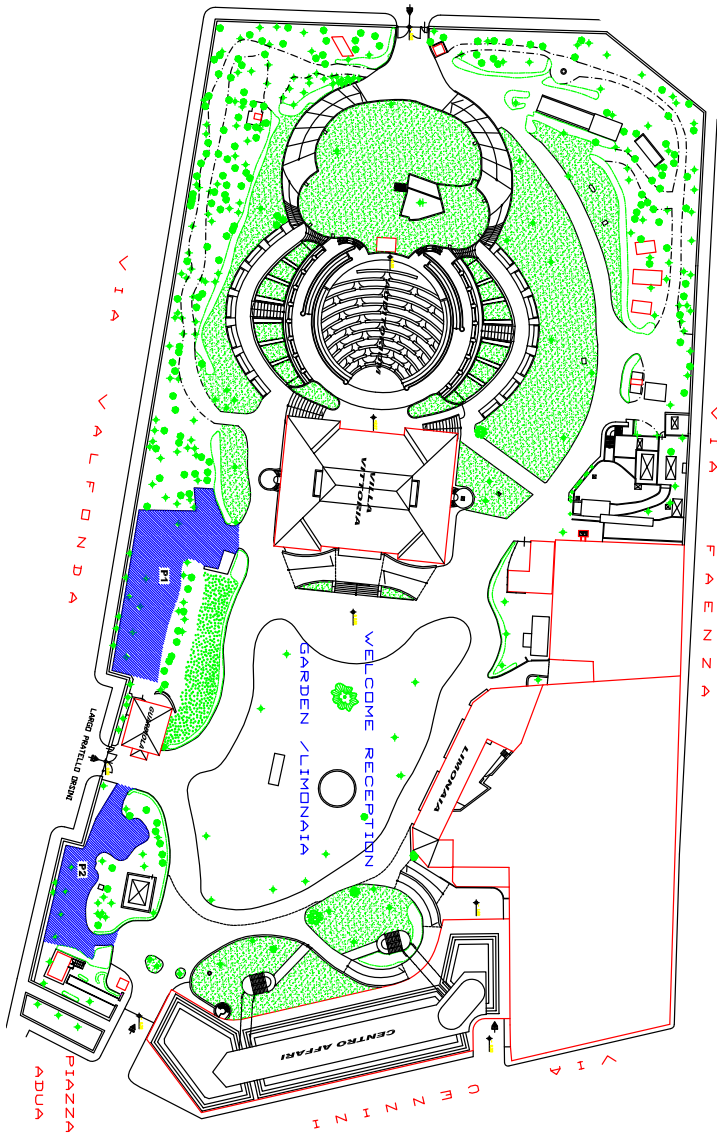


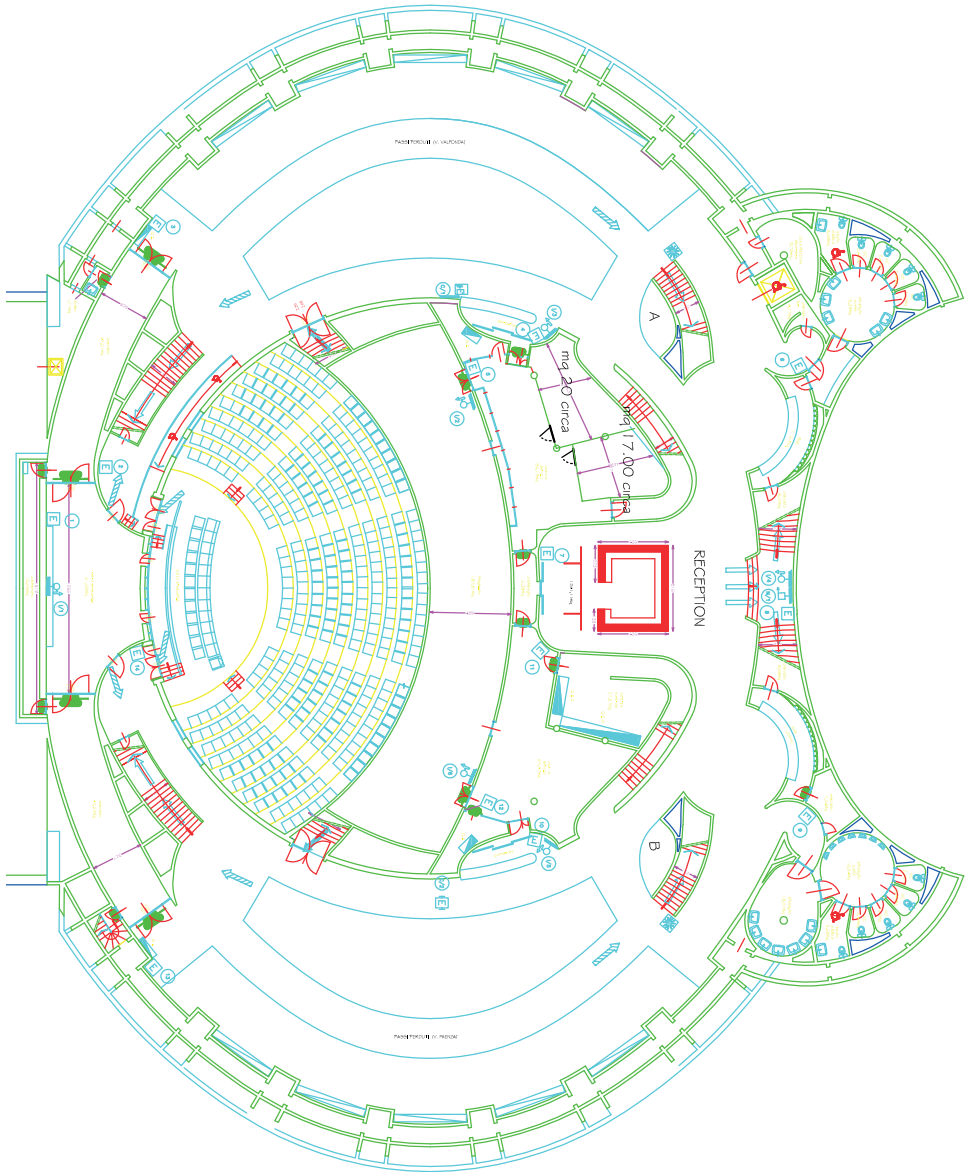
# 7<sup>TH</sup> BIENNIAL *meeting*

Maps

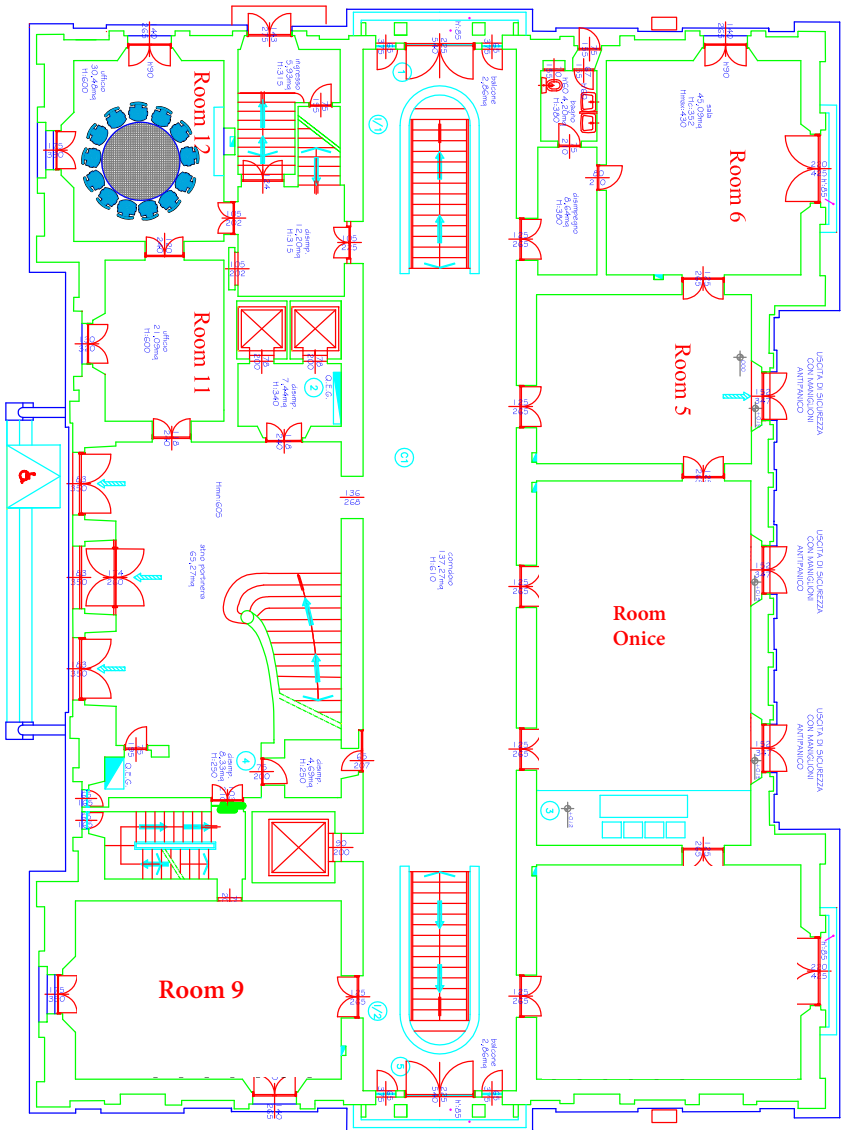


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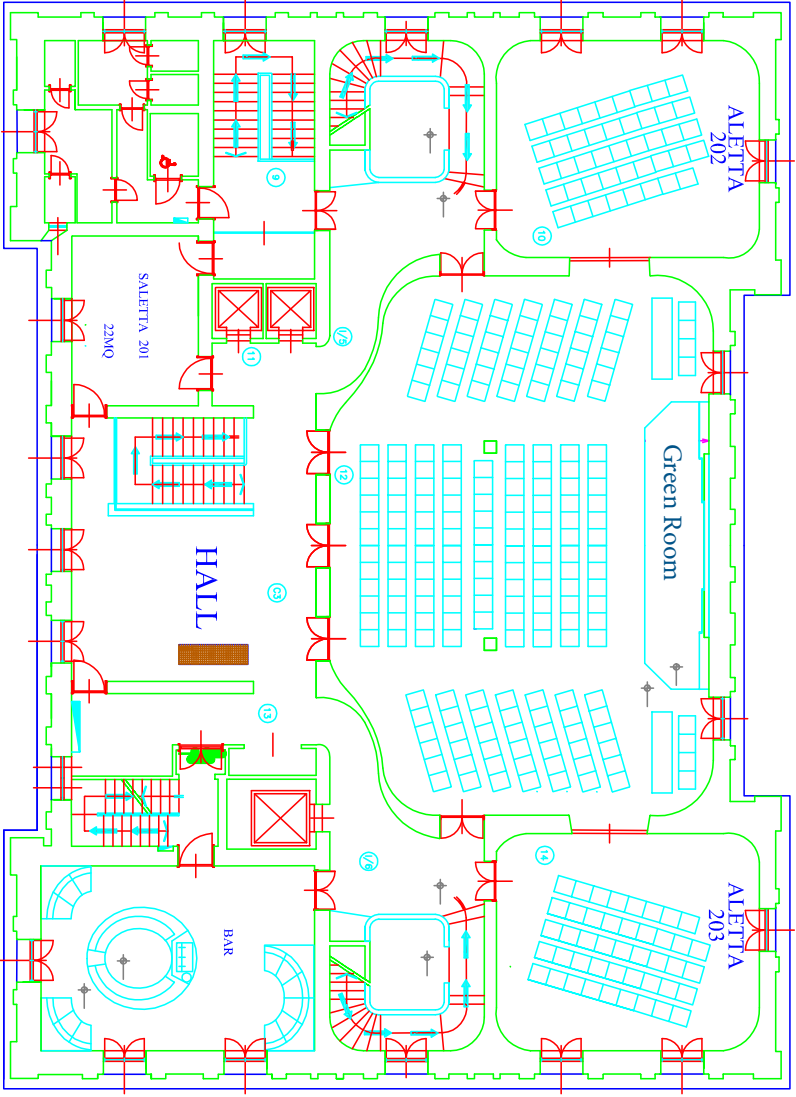




# Villa - Ground Floor



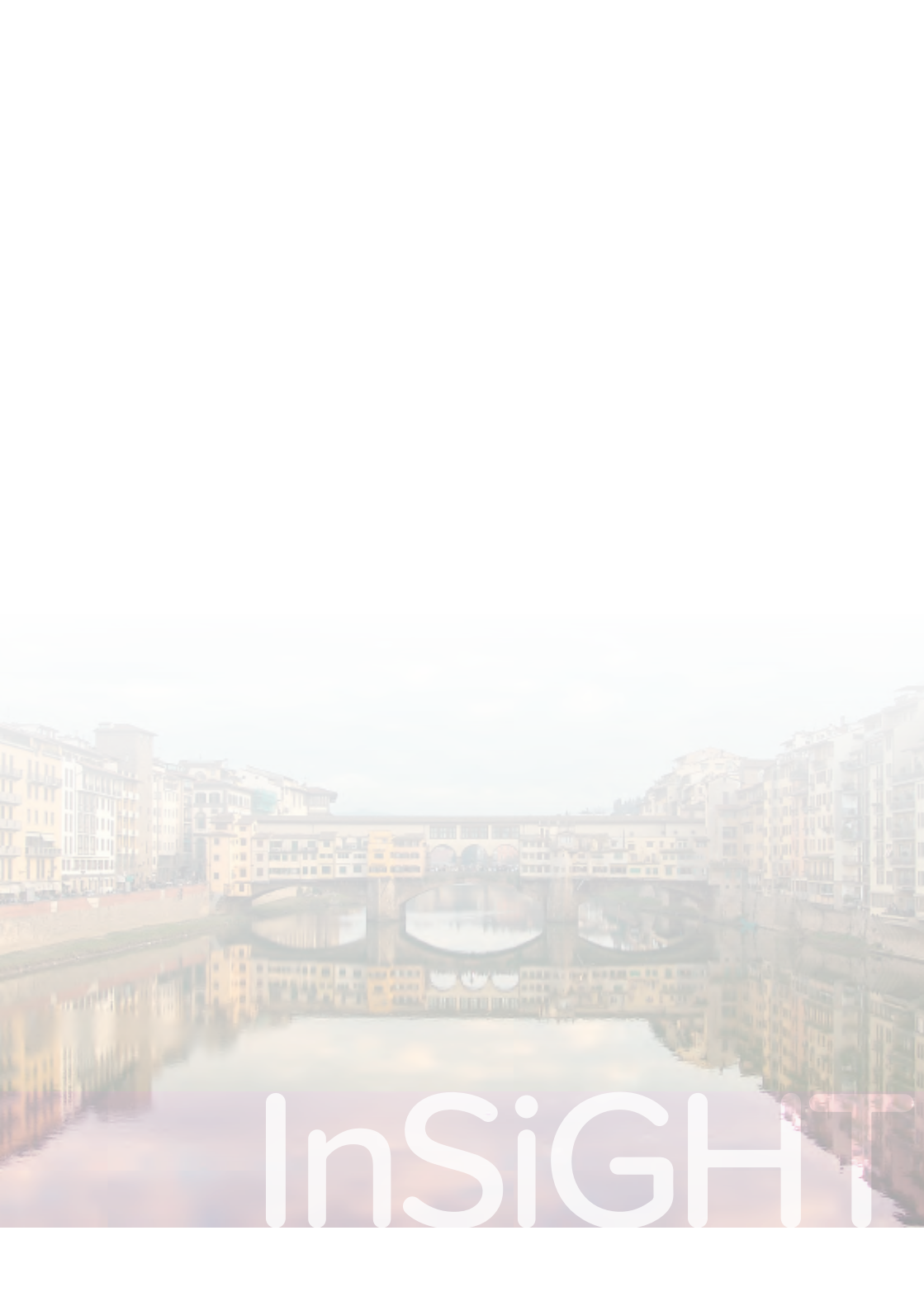




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