



10th Biennial Meeting
of the International Society for
Gastrointestinal Hereditary Tumours

InSiGHT
June 19 – 22, 2024
Barcelona



Venue:
Auditorium & Convention Centre – AXA
Avinguda Diagonal, 547 Barcelona

Organized by **INSIGHT**

 @Insightmeeting
#InSiGHT2024

Welcome message

Dear Colleagues, Dear Friends,

On behalf of the Board of Directors and the Program Committee it is a great pleasure to invite you to the 2024 Insight Biennial Conference to be held in Barcelona at 19th – 22nd June 2024.

This conference, which focusses on the world-wide improvement of care of patients and families with any hereditary condition resulting in gastrointestinal tumors, is a great opportunity and challenge. We aim to make this a great scientific meeting that attracts clinical and research experts in the field, including PhD students and other early career scientists and physicians.

The Program Committee has been working hard to put together a vibrant and comprehensive program for the meeting securing the participation of internationally acknowledged invited experts to address hot topics on key areas in Fundamental, Translational and Clinical Science in sessions with invited speakers and the best peer-reviewed abstracts as oral communications. Workshops will allow lively debating. We will also have the opportunity to listen the voice of patients critical to adequately design our care and our research.

Selected topics include among others: Mutational signatures and somatic evolution in normal tissue; the use of artificial intelligence (AI) in Digital pathology and Endoscopies; and monogenetic defects and Polygenic Risk Scores. Regarding clinical science we will be addressing surgical challenges as well as controversies in

surveillance and testing in pediatric and adult settings. Issues on colorectal, gynecological, gastric and biliopancreatic neoplasms will also be discussed. Also chemoprevention, vaccines and lifestyle modification will be tackled. Finally, we have joined forces with the European Society of Gastrointestinal Endoscopy to organize a pre-meeting educational event.

A scientific congress is not just a series of lectures or debates delivered from a podium. There is the social interaction, meeting old friends and making new acquaintances which may turn to become lifelong, discussing a project over a coffee, inviting colleagues to spend time in our unit. Especially for trainees, a congress represents the unique opportunity to meet renowned professionals and establish connections that can lead to new professional opportunities. In this regard, we have done our best to promote the active participation of the young people hoping to help providing effective enduring mentorship to the next generation of physicians, health professionals and scientists.

As Chairpersons of Insight we are very happy and honored to welcome all of you in Barcelona next June to share and learn the most up to date evidence in our specialty and to enjoy networking with your friends and colleagues.

Kind regards, also on behalf of the InSiGHT Scientific Committee



Conference directors

Meeting directors & core scientific committee



Prof. Dr. Nicoline Hoogerbrugge

MD, PhD, Internist-cancer
geneticist. Radboud university
medical center
Nijmegen, Netherlands



Dr. Gabriel Capellá

MD, PhD, Catalan Institute of
Oncology (IDIBELL),
Cancer geneticist
L'Hospitalet de Llobregat, Spain



Dr. Francesc Balaguer

MD, PhD, Gastroenterologist.
Hospital Clinic
Barcelona, Spain



Prof. dr. Marjolijn Ligtenberg

PhD, Molecular geneticist.
Radboud university
medical center
Nijmegen, Netherlands

Scientific committee

Dr. Joanne de Hullu

MD, PhD, Gynecologist. Radboud university medical center
Nijmegen, Netherlands

Dr. Marta Pineda

PhD, Biochemist. Catalan Institute of Oncology (ICO) & -
Bellvitge Biomedical Research Institute (IDIBELL)
L'Hospitalet de Llobregat, Spain

Dr. Leticia Moreira

MD, PhD, Gastroenterologist. Hospital Clinic
Barcelona, Spain

Dr. Joan Brunet

MD, PhD, Medical Oncologist. Catalan Institute
of Oncology - IDIBGI
Girona, Spain

Dr. Chella van der Post

MD, PhD, Pathologist. Radboud university medical center
Nijmegen, Netherlands

Dr. Laura Valle

PhD, Biochemist. Bellvitge Biomedical Research
Institute (IDIBELL)
L'Hospitalet de Llobregat, Spain

Dr. Maria Pellisé

MD, PhD, Gastroenterologist. Hospital Clinic
Barcelona, Spain

Dr. Miriam Cuatrecasas

MD, PhD, Pathologist. Hospital Clinic
Barcelona, Spain

Dr. Judith Balmaña

MD, PhD, Medical Oncologist. Vall d'Hebron Institute
of Oncology (VHIO)
Barcelona, Spain

Dr. Tanya Bisseling

MD, PhD, Gastroenterologist. Radboud university
medical center
Nijmegen, Netherlands



Pre-InSiGHT Programme for Patients

TUESDAY, JUNE 18TH

Virtual session

Organised in collaboration with ERN GENTURIS



European
Reference
Networks



ERN
GENTURIS



Pre-InSiGHT Programme for Patients

TUESDAY, JUNE 18TH

Virtual session

19:00 - 21:00 **Pre-InSiGHT meeting for patients and patient representatives with Lynch syndrome or polyposis**

Chairs:

Nicoline Hoogerbrugge, Radboud university medical center, Nijmegen, The Netherlands

Gabriel Capellá, Catalan Institute of Oncology (ICO) & Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

Organised in collaboration with ERN GENTURIS



European
Reference
Networks



ERN
GENTURIS

19:00 - 19:20 **What do you need to know about colonoscopy?**

Francesc Balaguer, Hospital Clínic Barcelona, Spain

19:20 - 19:40 **Different Lynch genes have different hereditary risks: PMS2 is the odd out**

Brandie Leach, Hereditary Cancer Medical Affairs, Exact Sciences, Cleveland, Ohio, USA

19:40 - 20:00 **Life after colectomy**

Toni T. Seppälä, Tampere University Hospital, Tampere University and TAYS Cancer Centre, Finland

20:00 - 20:30 **Promising news: Immune therapy in cancer treatment of patients with Lynch syndrome**

Dr. Judith Balmaña, Vall d'Hebron, Institute of Oncology (VHIO), Barcelona, Spain

20:30 - 21:00 **Questions from the audience**

21:00 **Closing remarks**



InSiGHT-ClinGen Polyposis-CRC VCEP Meeting

WEDNESDAY, JUNE 19TH



InSiGHT-ClinGen Polyposis-CRC VCEP Meeting

WEDNESDAY, JUNE 19TH

07:45 - 08:00 **Check-in, Coffee and Light Breakfast**

08:00 - 08:15 **Introduction, Overview**

08:15 - 08:45 **MMR subVCEP**

- History in InSiGHT, transition to InSiGHT ClinGen Expert Panel
- New MMR-Specific ACMG rules
- Future

9:25 - 10:05 **MUTYH subVCEP**

- Process
- New MUTYH-Specific Classification Rules
- Pilot Study
- Future

10:05 - 10:30  **Break, Coffee and Light Breakfast**

10:30 - 10:55 **Hamartomatous Polyp subVCEP SMAD4/BMP1A/STK11**

- Process
- HHT-Polyposis Joint effort
- Future

10:55 - 11:20 **POLD1/POLE subVCEP**

11:20 - 11:50 **U24 Grant Renewal Aims**

11:50 - 01:00  **Lunch**

This session is directed toward participants in the InSiGHT-ClinGen Hereditary CRC-Polyposis Variant Curation Expert Panel. If you would like to attend, kindly send an email to congresos@tacticsmd.net.



Pre-InSiGHT Programme

WEDNESDAY, JUNE 19TH



Pre-InSiGHT Programme

WEDNESDAY, JUNE 19TH

08:25 - 12:20

ESGE/ESDO postgraduate course:

"Advances in GI endoscopy and immunotherapy
in the management of hereditary GI syndromes"

Chairs:

Ian Gralnek, Institute of Gastroenterology and Hepatology,
Emek Medical Center, Afula, Israel

Thomas Seufferlein, Ulm University, Germany



08:00 - 08:30

Registration

08:25 - 08:30

Welcome message

08:30 - 09:30

SESSION 1:

Tips and Tricks for colonoscopy in high-risk conditions

Chairs:

Ian Gralnek, Institute of Gastroenterology and Hepatology,
Emek Medical Center, Afula, Israel

Francesc Balaguer, Hospital Clinic, Barcelona, Spain

- **Surveillance of the colon in Lynch Syndrome:** Tips and tricks
Maria Pellisé, Hospital Clinic, Barcelona, Spain
- **Surveillance of the pouch/rectum in FAP:** Tips and tricks
Andrew Latchford, St Mark's Hospital, London North West University
Healthcare NHS Trust, London, United Kingdom
- **Management of SPS:** How to diagnose and treat SSLs
Evelien Dekker, Amsterdam University Medical Centers, The Netherlands



Pre-InSiGHT Programme

09:30 - 10:30

SESSION 2:

Tips and tricks for upper endoscopy in high-risk conditions

Chairs:

Maria Pellisé, Hospital Clinic, Barcelona, Spain

Andrew Latchford, St Mark's Hospital, London North West University Healthcare NHS Trust, London, United Kingdom

- **Prevention of gastric cancer:** Tips and tricks for a high-quality upper GI endoscopy
Mário Dinis-Ribeiro, Instituto Português de Oncologia, Porto, Portugal
- **Prevention of gastric cancer in FAP**
Arthur Aelvoet, ANIOS MDL, Amsterdam, The Netherlands
- **Managing CDH1/CTNNA1 patients**
Tanya Bisseling, Radboud university medical center, Nijmegen, The Netherlands

10:30 - 11:00

Coffee Break

11:00 - 11:40

SESSION 3:

Surveillance of the duodenum in high-risk conditions

Chairs:

Evelien Dekker, Amsterdam University Medical Centers, The Netherlands

Tanya Bisseling, Radboud university medical center, Nijmegen, The Netherlands

- **How to deal with intestinal polyps in Peutz Jeghers?**
Begoña González, Teknon Medicaal Center, Barcelona, Spain
- **Management of duodenal polyposis in FAP**
John Gásdal Karstensen, University of Copenhagen, Denmark

11:40 - 12:20

SESSION 4:

New advances in immunotherapy in GI cancers

Chairs:

Thomas Seufferlein, Ulm University, Germany

- **Immunotherapy in colorectal cancer**
Eric Van Cutsem, University Hospital Gasthuisberg/Leuven, Belgium
- **Immunotherapy in upper GI cancer**
Thomas Seufferlein, Ulm University, Germany



Scientific programme of InSiGHT Conference

WEDNESDAY, JUNE 19TH

THURSDAY, JUNE 20TH

FRIDAY, JUNE 21ST

SATURDAY, JUNE 22ND



Scientific programme of InSiGHT Conference

WEDNESDAY, JUNE 19TH

12:00 - 13:00 **Registration**

13:00 - 13:20 **Welcome and announcements**

Nicoline Hoogerbrugge, Radboud university medical center,
Nijmegen, The Netherlands

Gabriel Capellá, Catalan Institute of Oncology (ICO) & Bellvitge Biomedical Research
Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

13:20 - 15:25 **SESSION 1 – TRANSLATIONAL SCIENCE**

Chairs:

Chella van der Post, Radboud university medical center, Nijmegen, Netherlands

Maria Pellisé, Hospital Clinic Barcelona, Spain

13:20 - 13:45

Use of AI in Endoscopies

Robert Hüneburg, Bonn University Hospital, Bonn, Germany

13:45 - 14:10

Use of AI in colorectal cancer: its application to GI hereditary tumors

Francesco Ciampi, Radboud university medical center, Nijmegen,
The Netherlands

14:10 - 14:55

Debate: Strategies to find all patients with hereditary cancer

Stefan Aretz, University Hospital Bonn, Bonn, Germany

Judith Balmaña, Vall d'Hebron Institute of Oncology (VHIO), Barcelona, Spain



Scientific programme of InSiGHT Conference

14:55 - 15:25

Abstract Presentations 1

14:55 - 15:05

Predictors of invasive signet ring cell carcinoma in CDH1 carriers – results from the multicenter Study of CDH1 Outcomes & Surveillance**Ophir Gilad**, University of Chicago, Illinois, United States

15:05 - 15:15

Familial serrated polyposis syndrome in the genetics of colonic polyposis study**Daniel Buckmann**, University of Pittsburgh, Pennsylvania, United States

15:15 - 15:25

Machine learning-based endoscopic classification for superficial mucosal lesions in hereditary diffuse gastric cancer**Leanne Wu**, Cambridge University, United Kingdom

15:25 - 15:35

Testing colonic ganglioneuromas as a clue to the diagnosis of PTEN hamartoma tumor syndrome**Chella Van der Post**, Radboud university medical center, Nijmegen, The Netherlands

15:35 - 15:45

Can genomic features of pre-malignant adenomas be used to identify biallelic MUTYH and NTHL1 carriers?**Romy Walker**, University of Melbourne Centre for Cancer Research, Victorian Comprehensive Cancer Centre, Melbourne, Australia

15:45 - 15:55

Multicentre approach to improve the identification and management of CMMRD patients in Spain**Fátima Marín**, Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

15:25 - 15:55

 **Coffee Break**

Scientific programme of InSiGHT Conference

15:55 - 17:50 **SESSION 2 – TRANSLATIONAL SCIENCE**

Chairs:

Marta Pineda, Catalan Institute of Oncology (ICO) & - Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

Elke Holinski-Feder, München Ludwig-Maximilians-Universität, Germany

15:55 - 16:20

Microbiome and (risk of) hereditary GE cancer

Annemarie Boleij, Radboud university medical center, Nijmegen, The Netherlands

16:20 - 17:50

● **Abstract Presentations 2**

16:20 - 16:30

HLA Type as a possible modulator of cancer risk in Lynch syndrome: first data from the INDICATE Network

Lena Bohaumilitzky, University Hospital Heidelberg, Germany

16:30 - 16:40

A faecal microbial signature to optimise colorectal cancer surveillance in Lynch syndrome

Sandra Taboada-López, GoodGut SL, Girona, Spain

16:40 - 16:50

Germline results following comprehensive genomic tumour sequencing of 1070 gynaecological cancers

Arianna Panfili, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy

16:50 - 17:00

IL-17A-Producing NKp44(-)ILC3s may promote duodenal adenoma formation in familial adenomatous polyposis

Jacob Nattermann, University Hospital Bonn, Germany



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17:00 - 17:10

Characterization of the mutational landscape of colorectal tumors from individuals with adenomatous or serrated polyposis

Anna Sommer, University Hospital Bonn, Germany

17:10 - 17:20

Lynch syndrome-related neoantigens prediction and validation for a dendritic-cell based cancer prevention vaccine

Cristina Bayó Llorens, Hospital Clínic Barcelona, Spain

17:20 - 17:30

Profiling germline and somatic mutations in consecutive early onset colorectal cancer cases in a European population

Penelope Edwards, The Centre for Familial Intestinal Cancer, St Mark's Hospital, London Northwest Healthcare Trust, United Kingdom

17:30 - 17:40

Prevalence of DNA mismatch repair (MMR) gene mosaicism and specific intronic pathogenic variants in people with MMR-deficient cancers

Romy Walker, University of Melbourne Centre for Cancer Research, Victorian Comprehensive Cancer Centre, Melbourne, Australia

17:40 - 17:50

Gastric epithelium and patient-derived gastric organoids from BRCA_{1/2} carriers harbor increased proliferation and DNA damage

Bryson Katona, University of Pennsylvania Perelman School of Medicine, PA, United States

17:50 - 18:00 **Adjourning**

19:00  **Welcome Reception**

Location: Conference Venue



Scientific programme of InSiGHT Conference

THURSDAY, JUNE 20TH

08:30 - 10:05 SESSION 3 – FUNDAMENTAL SCIENCE

Chairs:

Brandie Leach, Hereditary Cancer Medical Affairs, Exact Sciences, Cleveland, Ohio, United States

Ian Frayling, St Mark's Hospital, London & St Vincent's Hospital, Dublin

08:30 - 08:55

New genetic aspects of Lynch syndrome

Marjolijn Ligtenberg, Radboud university medical center, Nijmegen, The Netherlands

08:55 - 09:20

New genes for polyposis

Laura Valle, Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

09:20 - 09:50

● **Abstract Presentations 3**

09:20 - 09:30

Establishing a deep mutational scan for MSH6 missense variant classification

Anthony Scott, University of Michigan, Ann Arbor, MI, United States

09:30 - 09:40

The immune profile of Lynch syndrome-associated colorectal adenomas pinpoints the main determinants of immune activation

Lena Bohaumilitzky, Institute of Pathology, University Hospital Heidelberg, Germany

09:40 - 09:50

Diagnostic challenges of MLH1 constitutional epimutations involved in Lynch syndrome

Julie Leclerc, Jeanne de Flandre Hospital, Lille University Hospital, France



Scientific programme of InSiGHT Conference

09:50 - 10:05

Flashtalks

09:50 - 09:53

The germline POLD1 c.1420C>A p.(Leu474Ile) variant segregates with endometrial cancer and colonic adenomas demonstrating hypermutation and defective POLD1 mutational signatures

Khalid Mahmood, The University of Melbourne, Parkville, Victoria, Australia

09:53 - 09:56

Adenomatous polyps of germline pathogenic PMS2 mutation carriers: preliminary epidemiological and histopathological report from the LYNCH-GPS study

Katarina Andini, University Medical Centre Groningen, The Netherlands

09:56 - 09:59

Diagnostic yield and characterization of endoscopic surveillance in CDH1 pathogenic variant carriers in Spain

Joaquin Castillo, Hospital Clínic Barcelona, Spain

09:59 - 10:02

Mesalamine for colorectal cancer prevention program in Lynch Syndrome (MesaCAPP)

Ann-Sofie Backman, Karolinska Institutet, Huddinge, Stockholm, Sweden

10:02 - 10:05

Birth cohort effect on age of colorectal cancer onset in Lynch syndrome

Ophir Gilad, University of Chicago, Illinois, United States

10:05 - 11:05



Coffee Break + Poster session A



Scientific programme of InSiGHT Conference

11:05 - 12:25 **SESSION 4 – FUNDAMENTAL SCIENCE**

Chairs:

Maurizio Genuardi, University of Florence, Italy

Beneditto Rossi, Hospital Sirio Libanes, São Paulo, Brazil

11:05 - 11:30

Mutational signatures (somatic evolution in normal tissue)

Gene Koh, University of Cambridge, Cambridge, United Kingdom

11:30 - 11:55

Updated international guidelines on hereditary diffuse gastric cancer, hot news from Porto

Carla Oliveira, Ipatimup, Porto, Portugal

11:55 - 12:25

● **Abstract Presentations 4**

11:55 - 12:05

Genomic features of post-colonoscopy colorectal cancers in people with Lynch syndrome

Daniel Buckanan, Victorian Comprehensive Cancer Centre, The University of Melbourne, Parkville, Victoria, Australia

12:05 - 12:15

CaptureRNA-seq as a supplement to DNA germline testing to increase the diagnostic yield of hereditary tumor syndromes

Megan Hitchins, Cedars-Sinai Medical Center, Los Angeles, CA

12:15 - 12:25

The exome-wide genomic profile of Lynch syndrome-related colorectal cancers differs by affected gene: implications for determining pathways of tumourigenesis

Peter Georgeson, Victorian Comprehensive Cancer Centre, The University of Melbourne, Parkville, Victoria, Australia



Scientific programme of InSiGHT Conference

12:25 - 13:10

Debate: Surveillance in moderate risk genes

Sonia Kupfer, University of Chicago, Chicago, United States

Andrew Latchford, St Mark's Hospital, London North West University Healthcare

NHS Trust, London, United Kingdom

13:10 - 14:10



Lunch break

14:10 - 16:00

SESSION 5 – CLINICAL SCIENCE

Chairs:

Evelien Dekker, Amsterdam University Medical Centers, The Netherlands

Peter P. Stanich, The Ohio State University, Wexner Medical Center Columbus, United States

14:10 - 14:35

Endoscopic surveillance in hereditary GI syndromes

Francesc Balaguer, Hospital Clínic Barcelona, Spain

14:35 - 15:00

Surveillance in children with FAP

Warren Hyer, St Marks Hospital & Chelsea and Westminster Hospital, London, United Kingdom

15:00 - 16:00

Abstract Presentations 5

15:00 - 15:10

Pancreatic cancer is more common than melanoma in non-white CDKN2A positive individuals

Colin Young, Ambry Genetics, Aliso Viejo, California, United States

15:10 - 15:20

Lynch Syndrome (LS) patients with inflammatory bowel disease (IBD) have significantly higher intestinal neoplasia risk than LS patients without IBD

Swati G. Patel, University of Colorado – Anschutz School of Medicine, Rocky Mountain Regional Veterans Affairs Medical Center, Aurora, Colorado, United States



Scientific programme of InSiGHT Conference

15:20 - 15:30

Early Onset colorectal mortality trends in Argentina, 1997-2020. Is it time to reduce the age of screening in average-risk adults? First report in Latin America

Marina Antelo, SCo, UNLa, CONICET, Buenos Aires, Argentina

15:30 - 15:40

Detection of early gastric cancer during endoscopic surveillance in CDH1 and CTNNA1 pathogenic variant carriers: higher yield with targeted than with random biopsies

Tanya Bisseling, Radboud university medical centre, Nijmegen, the Netherlands

15:40 - 15:50

Psychosocial distress and quality of life in families with a germline CDKN2A pathogenic variant

Monique van Leerdam, Netherlands Cancer Institute, Amsterdam, The Netherlands

15:50 - 16:00

Variable cancer penetrance among CTNNA1 loss-of-function carriers: Initial results from the CTNNA1 Familial Expansion (CAFÉ) Study

Bryson W. Katona, Perelman Center for Advanced Medicine, Philadelphia, PA, United States

16:00 - 16:30

 **Coffee Break**

16:30 - 17:15

Debate: New-born testing on hereditary cancer genes

Héctor Salvador, Hospital Sant Joan de Déu, Barcelona, Spain

Ingrid Winship, University of Melbourne & Royal Melbourne Hospital, Melbourne, Australia



Scientific programme of InSiGHT Conference

17:15 - 18:15

● **Abstract Presentations 6**

17:15 - 17:25

Digestive burden of patients with constitutional mismatch repair deficiency syndrome, diagnostic pitfalls, and experience of immunotherapy: a report from the C4CMMRD database

Chrystelle Colas, Curie Institute, Paris, France

17:25 - 17:35

Gastric and duodenal cancer in individuals with Lynch syndrome: a nationwide cohort study

TBC

17:35 - 17:45

Familial uveal melanoma and other tumours in 25 families with monoallelic germline MBD4 variants.

Marie-Charlotte Villy, Curie Institute, Paris, France

17:45 - 17:55

An evaluation of user experiences, perceptions and attitudes towards Faecal Immunochemical Testing (FIT) for risk-stratified colonoscopy in Lynch syndrome patients

Annie Lincoln, Comprehensive Cancer Centre, King's College London, London, United Kingdom

17:55 - 18:05

Systematic review of the phenotypic cancer spectrum in NTHL1-associated tumour syndrome emphasizes the need to identify more families to assess cancer risks

Weilun Gao, Royal Melbourne Hospital & University of Melbourne, Australia

18:05 - 18:15

Differences in adenoma and post-colonoscopy colorectal cancer detection between Lynch syndrome carriers with and without a previous colorectal cancer

Ariadna Sánchez, Hospital Clínic Barcelona, Spain



Scientific programme of InSiGHT Conference

18:15 - 18:20 **Adjourning**

20:00 **Get Together**

Venue: Casa de Les Punxes

Address: Av. Diagonal, 420, L'Eixample, 08037 Barcelona

(Limited Places - Registration Required)

FRIDAY, JUNE 21ST

08:30 - 10:05 **SESSION 6 – CLINICAL SCIENCE**

Chairs:

Bryson W. Katona, Perelman Center for Advanced Medicine, Philadelphia, PA,
United States

Mathew Kalady, The Ohio State University, Comprehensive Cancer Center,
Columbus, OH, United States

08:30 - 08:55

Surgery in Lynch syndrome and polyposis

Gabriela Möslein, Academic Hospital of the University of Düsseldorf,
Duisburg, Germany

08:55 - 09:20

Recent advances in Immunotherapy for GI and gynecological tumors

Iosune Baraibar, Vall d'Hebron Institute of Oncology (VHIO), Barcelona, Spain



Scientific programme of InSiGHT Conference

09:20 - 09:50

● Abstract Presentations 7

09:20 - 09:30

Yield of multigene panel germline genetic testing among those with advanced colorectal adenomas

Swati G. Patel, University of Colorado Anschutz School of Medicine, Rocky Mountain Regional Veterans Affairs Medical Center, Aurora, Colorado, United States

09:30 - 09:40

Can butyrate prevent colon cancer? The AUSFAP study: A randomised, crossover clinical trial

Finlay Macrae, The Royal Melbourne Hospital, Melbourne, Australia

09:40 - 09:50

Cancer risks associated with germline pathogenic variants in MLH1, MSH2, MSH6, PMS2, and EPCAM genes

Christine Drogan, University of Chicago, IL, United States

09:50 - 10:05

| **Flashtalks**

09:50 - 09:53

Long read sequencing elucidates complex germline variants in individuals undergoing hereditary gastrointestinal cancer testing

Sami Belhadj, Enterprise, Aliso Viejo, California, CA, United States

09:53 - 09:56

Dietary habits of patients with or without hereditary cancer syndromes- A cross sectional cohort study

Hitasha Mittal, Università Vita Salute San Raffaele, Milano, Italy

09:56 - 09:59

A prospective analysis identified mismatch repair genes as candidate predisposing genes for uveal melanoma

Anaïs Le Ven, PSL Research University, Paris, France



Scientific programme of InSiGHT Conference

09:59 - 10:02

Evaluation of upper gastrointestinal tract surveillance in individuals with Lynch Syndrome (EARLY) - An international registry

Tim Marwitz, University Hospital Bonn, National Center for Hereditary Tumor Syndromes, Bonn, Germany.

10:02 - 10:05

Optimizing POLE and POLD1 variant interpretation: gene-specific classification guidelines and in vitro system for functional assessment

Julen Viana-Errasti, Catalan Institute of Oncology (ICO) & Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain

10:05 - 11:05

 **Coffee Break + Poster session B**

11:05 - 13:15

SESSION 7 – CLINICAL SCIENCE

Chairs:

Leticia Moreira, Hospital Clínic Barcelona, Spain

Rodrigo Jover, Hospital General Universitario Dr. Balmis, Alicante

11:05 - 11:30

Pancreas surveillance in high-risk individuals

Diane Simeone, NYU Pancreatic Cancer Center, New York, United States

11:30 - 12:30

● **Abstract Presentations 8**

11:30 - 11:40

Objective and subjective life expectancy in Lynch syndrome

Monique van Leerdam, Netherlands Cancer Institute, Amsterdam, The Netherlands

11:40 - 11:50

Racial and ethnic patterns of variants of uncertain significance (VUS) among patients with early-onset colorectal cancer

Andreana N. Holowatyj, Vanderbilt University Medical Center, Nashville, TN, United States



Scientific programme of InSiGHT Conference

11:50 - 12:00

Ileocecal intubation in Lynch syndrome patients – go all the way!

Katrin van Beekum, University Hospital Bonn, Germany

12:00 - 12:10

An Aspirin a day? The (still) secret weapon against Colorectal Cancer in Lynch Syndrome

Rebecca Tuckey, Te Whatu Ora – Health New Zealand Waitaha Canterbury, New Zealand

12:10 - 12:20

Colorectal adenoma and cancer incidences in path_MMR carriers undergoing surveillance colonoscopy

Pål Møller, Institute of Cancer Research, The Norwegian Radium Hospital, Oslo University Hospital, Norway

12:20 - 12:30

Early onset colon and rectal cancer mortality in Lynch syndromes carriers subjected to surveillance colonoscopy

Pål Møller, Institute of Cancer Research, The Norwegian Radium Hospital, Oslo University Hospital, Norway

12:30 - 13:15

Debate: Early onset pancreatic cancer

Toni T. Seppälä, Tampere University Hospital, Tampere University and TAYS Cancer Centre, Finland

Bryson Katona, University of Pennsylvania Perelman School of Medicine, PA, United States

13:15 - 14:15

 **Lunch break**



Scientific programme of InSiGHT Conference

14:15 - 16:05 **SESSION 8 – CLINICAL SCIENCE**

Chairs:

Joan Brunet, Catalan Institute of Oncology, Girona Biomedical Research Institute (IDIBGI), Girona, Spain

Stefan Aretz, University Hospital Bonn, Bonn, Germany

14:15 - 14:40

Endometrial cancer in hereditary cancer syndromes

Emma J. Crosbie, St Mary's Hospital, Manchester, United Kingdom

14:40 - 15:05

Ovarian cancer in hereditary cancer syndromes

Neil Ryan, St Mary's Hospital, Manchester, United Kingdom

15:05 - 16:05

● **Abstract Presentations 9**

15:05 - 15:15

Recruiting Lynch Syndrome Patients for clinical research via social media improves participant diversity

Swati G. Patel, University of Colorado Anschutz School of Medicine, Rocky Mountain Regional Veterans Affairs Medical Center, Aurora, Colorado, United States

15:15 - 15:25

Development of abdominal desmoid tumours after colectomy and ileorectal anastomosis versus proctocolectomy and ileal pouch-anal anastomosis in familial adenomatous polyposis

Arthur Aelvoet, Amsterdam UMC, The Netherlands

15:25 - 15:35

Outcomes of Lynch syndrome endometrial cancer surveillance in a nation-wide cohort

Lotte van Leeuwen, Erasmus MC Cancer Institute, Rotterdam, The Netherlands

15:35 - 15:45

Employing innovation to enhance the safety and reliability of restorative prophylactic surgical techniques for patients with familial adenomatous polyposis at a national referral centre

Bruno A. Alves Martins, St Mark's Hospital, London North West University Healthcare NHS Trust, London, United Kingdom



Scientific programme of InSiGHT Conference

15:45 - 15:55

Risk of cancer and secondary surgery following colectomy with ileorectal anastomosis and proctocolectomy with ileal pouch-anal anastomosis in familial adenomatous polyposis.

Hicham Bouchiba, Amsterdam UMC, University of Amsterdam Cancer Center, The Netherlands

15:55 - 16:05

The new National english bowel cancer screening programme for Lynch syndrome colonoscopic Surveillance

Kevin Monahan, St Mark's Hospital, London North West University Healthcare NHS Trust, London, United Kingdom

16:05 - 16:35

 **Coffee Break**

16:35 - 17:20

Debate: Strategies on endometrial and ovarian cancer in Lynch syndrome

Joanne A. de Hullu, Radboud university medical center, Nijmegen, The Netherlands

Anne van Altena, Radboud university medical center, Nijmegen, The Netherlands

17:20 - 18:20

Abstract Presentations 10

17:20 - 17:30

Optimizing mainstreaming of genetic testing in parallel with ovarian and endometrial cancer tumor testing: how do we maximize our impact?

Ying Liu, Memorial Sloan Kettering Cancer Center, New York, NY, United States

17:30 - 17:40

Cancer risk in hamatomatous polyposis syndromes: a focus on juvenile polyposis and Peutz-Jeghers

Emanuele Damiano Luca Urso, University of Padua, Italy



Scientific programme of InSiGHT Conference

17:40 - 17:50

The yield of artificial intelligence (GI genius) in Lynch syndrome – A randomized tandem-colonoscopy trial

Ido Laish, Sheba Medical Center, Israel

17:50 - 18:00

Specifications to the ACMG/AMP criteria enhance the classification of MMR variants

John Paul Plazzer, Royal Melbourne Hospital, Parkville, Australia,

18:00 - 18:10

The role of genotoxic gut bacteria in CRC tumourigenesis in Lynch syndrome

Yen Lin Chu, Victorian Comprehensive Cancer Centre, The University of Melbourne, Parkville, Victoria, Australia

18:10 - 18:20

Evaluating different testing approaches for identifying APC mosaicism in unexplained adenomatous polyposis: paving the way for the new susceptibility gene discovery

Eric Joo, The University of Melbourne, Parkville, Australia

18:20 - 18:25 **Adjourning**

18:30 **InSiGHT Business Meeting**

(Only for members)

20:00 **Congress Dinner**

Venue: Terraza Miramar

Address: Plaça de l'Armada, s/n, Montjuïc, Sants-Montjuïc, 08038 Barcelona
(Limited Seats - Registration Required)



Scientific programme of InSiGHT Conference

SATURDAY, JUNE 22ND

08:30 - 10:20 SESSION 9 – CLINICAL SCIENCE

Chairs:

Monique van Leerdam, Netherlands Cancer Institute, Amsterdam, The Netherlands
Sue Clark, St Mark's Hospital, Harrow, United Kingdom

08:30 - 08:55

Lifestyle and risk of hereditary cancer

Aung Ko Win, University of Melbourne Centre for Cancer Research,
Melbourne, Australia

08:55 - 09:20

Surveillance frequency in familial colorectal cancer

Veerle Coupe, VU Medical Centre, Amsterdam, The Netherlands

09:20 - 10:20

● **Abstract Presentations 11**

09:20 - 09:30

Prevalence of pathogenic genetic variants in gastric cancer patients ascertained through multigene panel testing

Ophir Gilad, University of Chicago, Chicago, Illinois, United States

09:30 - 09:40

Discovery of recessive effect of human polymerase delta proofreading deficiency through mutational analysis of POLD1-mutated normal and cancer cells

Laura Valle, Institute for Research in Biomedicine (IRB Barcelona), The Barcelona Institute of Science and Technology (BIST), Barcelona, Spain

09:40 - 09:50

Interrogating the landscape of immunogenic frameshift mutations in patients with Lynch syndrome and immune suppression programs supporting colorectal cancer development

Aimee Lucas, Icahn School of Medicine at Mount Sinai, New York, NY, United States



Scientific programme of InSiGHT Conference

09:50 - 10:00

Open-source bioinformatic pipeline to improve PMS2 genetic testing using short-read NGS data

Elisabeth Munté, Catalan Institute of Oncology (ICO), L'Hospitalet de Llobregat, Spain

10:00 - 10:10

Germline variants in DNA interstrand-cross link repair genes may contribute to increased susceptibility for serrated polyposis in particular with proximal/whole colon localization of polyps, with implications for clinical management and therapeutics

Patricia Silva, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal

10:10 - 10:20

KRAS-G 12C: a neglected biomarker for identifying MUTYH-associated polyposis patients

Giovana Torrezan, C.Camargo Cancer Center, São Paulo, Brazil

10:20 - 10:50 ☕ **Coffee Break**

10:50 - 12:25 **SESSION 10 – CLINICAL SCIENCE**

Chairs:

Allan Spigelman, University of New South Wales, Sydney

Tanya Bisseling, Radboud university medical center, Nijmegen, The Netherlands

10:50 - 11:15

Vaccines for Lynch syndrome

Eduardo Vilar, The University of Texas MD Anderson Cancer Center Houston, United States



Scientific programme of InSiGHT Conference

11:15 - 11:40

Chemoprevention of hereditary GI cancer

John Burn, Professor of Clinical Genetics, Cancer Prevention,
Newcastle University, Newcastle, United Kingdom

11:40 - 12:25

Debate: East versus West in colorectal cancer management

Joanne Ngeow, Cancer Genetics Service, National Cancer Centre Singapore

Finlay Macrae, The Royal Melbourne Hospital, Melbourne, Australia

12:25 - 13:25

● **Best Oral Communications**

12:25 - 12:35

Preventing Cancer in Lynch Syndrome: Vaccination with mutation-derived neoantigen-loaded dendritic cells eliminates precancerous cells

Asima Abidi, Radboud university medical center, Nijmegen, The Netherlands

12:35 - 12:45

Homopolymer switches mediate adaptive mutability in mismatch repair-deficient colorectal cancer

Marnix Jansen, CL Cancer Institute, University College London, London, United Kingdom

12:45 - 12:55

Development and validation of a MIRNA-Based signature, Powered by machine learning, for predicting 10-year recurrence-free and overall survival after curative-intent treatment in early-onset colorectal cancer

Alessandro Mannucci, Beckman Research Institute of City of Hope, Monrovia, CA, United States



Scientific programme of InSiGHT Conference

12:55 - 13:05

Colorectal carcinomas from path_MSH6 carriers display a lower degree and/or later onset of microsatellite instability and evoke weaker immune responses

Noah Cornelis Helderma, Leiden University Medical Center, Leiden, The Netherlands

13:05 - 13:15

Phase IIA trial of encapsulated rapamycin (ERAPA) in patients with familial adenomatous polyposis to reduce intestinal polyp burden: 6 month interim results

Carol Burke, Cleveland Clinic, Cleveland, OH, United States

13:15 - 13:25

Estimating cancer risk in carriers of Lynch syndrome variants in UK Biobank

Albert Tenesa, The University of Edinburgh

13:25 - 13:40

Concluding remarks

Nicoline Hoogerbrugge, Radboud university medical center, Nijmegen, The Netherlands

Gabriel Capellá, Catalan Institute of Oncology (ICO) & Bellvitge Biomedical Research Institute (IDIBELL), L'Hospitalet de Llobregat, Spain



Venue and main contact details

Venue

Auditorium & Convention Centre – AXA

Av. Diagonal, 547, Les Corts,
08029 Barcelona
+34 932 90 11 02

How to get there by public transport:

By underground:

L3, Maria Cristina station

By tram:

Line T3

By bus:

Lines 7, 33, 34, 63, 67, 68, 78, N12



Main contact details

Technical secretariat

General Information
Registration – Exhibition – Sponsorship



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Insight headquarters



The International Society for Gastrointestinal Hereditary Tumours (InSiGHT)

The St Mark's Centre for Familial Intestinal Cancer. St Mark's Hospital, Central Middlesex Hospital Site. Acton Lane, London NW10 7NS, UK.

admin@insight-group.org



General information

Registration fees

	Early Bird (Until April 1st)	Regular (From April 2nd to June 19th)	Onsite
InSiGHT Member Nurses, Genetic Counsellors, Students, Residents, Trainee*	325.00 €	500.00 €	600.00 €
InSiGHT Non Member - Nurses, Genetic Counsellors, Students/Residents, Trainee*	500.00 €	600.00 €	700.00 €
InSiGHT Member MD	600.00 €	700.00 €	800.00 €
InSiGHT Non Member MD	700.00 €	800.00 €	900.00 €

*** Students/Residents/Trainee:** Proof of status is mandatory - In order to benefit from the special fee, a submission of your status confirmation (approval letter signed by the Head of Department or copy of your status ID) must be uploaded during the Online registration.

The registration fee covers access to all scientific sessions, coffees and lunch breaks during the congress and the certificate of attendance.

Registration desk opening hours

Wednesday, from 07:45 to 20:00

Thursday, from 08:00 to 18:10

Friday, from 08:00 to 18:10

Saturday, from 08:00 to 13:00

Name badges

The name badges will be distributed from the Registration Area. All participants are kindly requested to wear their name badges throughout the conference.



General information

Certificate of attendance

Certificates of attendance will be sent directly right after the conference to the email address provided.

Exhibition opening hours

The exhibition will be open on 19 – 22 June 2024 during the congress hours:

Wednesday, from 09:30 to 17:45

Thursday, from 08:30 to 18:10

Friday, from 08:30 to 18:10

Saturday, from 08:30 to 13:00

Important dates

Deadline for early registration:

April 1st, 2024

Social programme

The social programme and activities for the InSiGHT 2024 Biennial Meeting are listed below:

Welcome Reception

Date | Time: Wednesday, June 19th 2024

Location: Auditori AXA Convention Center

** All INSIGHT2024 participants are cordially invited to the Welcome Reception.*

BCN Sunset InSiGHT Night - Dinner

Date | Time: Friday, June 21st | 20:00pm

Location: Terraza Miramar - Plaça de l'Armada,
s/n, Montjuïc, 08038 Barcelona

** Ticket must have been purchased in advance.*



General information

Wireless internet access

Username: INSIGHT2024

Password: HOLA

Social media

Conference website

In the conference website you will find all the information about the event, such as: scientific programme, speakers, committees, registration, venue and accommodation.

tactics/insight-2024



Follow us on @insightmeeting and tweet with the hashtag #InSiGHT2024.

[twitter/insightmeeting](https://twitter.com/insightmeeting)

EACCME accreditation

InSiGHT 2024 Biennial Meeting will request CME Accreditation.



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Lunch and refreshments

Lunch boxes are included in the participants' registration fee.

Coffee & tea will be available in the exhibition area during the breaks.

Safety & security

Please do not leave your bags or suitcases unattended at any time, whether inside or outside the session halls. Cloakroom will not be available in this occasion.



General information

Liability & insurance

By registering for the Congress and/or by participating in the exhibition joined to the Congress, participants and exhibitors agree that InSiGHT, the Organising Committee or the Congress Secretariat do not assume any responsibility for damage or injuries to persons or property during the Congress. Participants and exhibitors are advised to organize their own health, travel, and personal insurance.

Letter of invitation

Upon request, the Chairman of the Local Organising Committee will be pleased to send a personal letter of invitation to registered participants so as to enable participants to obtain visas for attending the Congress. These invitation letters cannot be considered as an offer of financial support by the organisers and will only be provided to registered participants once the payment has been received.

Eco-friendly & paperless

InSiGHT is a paperless, sustainable and environmentally friendly Conference encouraging the use of recyclable material.

Smoking

InSiGHT 2024 is a non-smoking congress.

Abstract book

Abstract book will be published online on the congress website.



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The 10th Biennial Meeting of the International Society for Gastrointestinal Hereditary Tumours, has been assessed as **COMPLIANT** with the MedTech Europe Code of Ethical Business Practice by EthicalMedTech.



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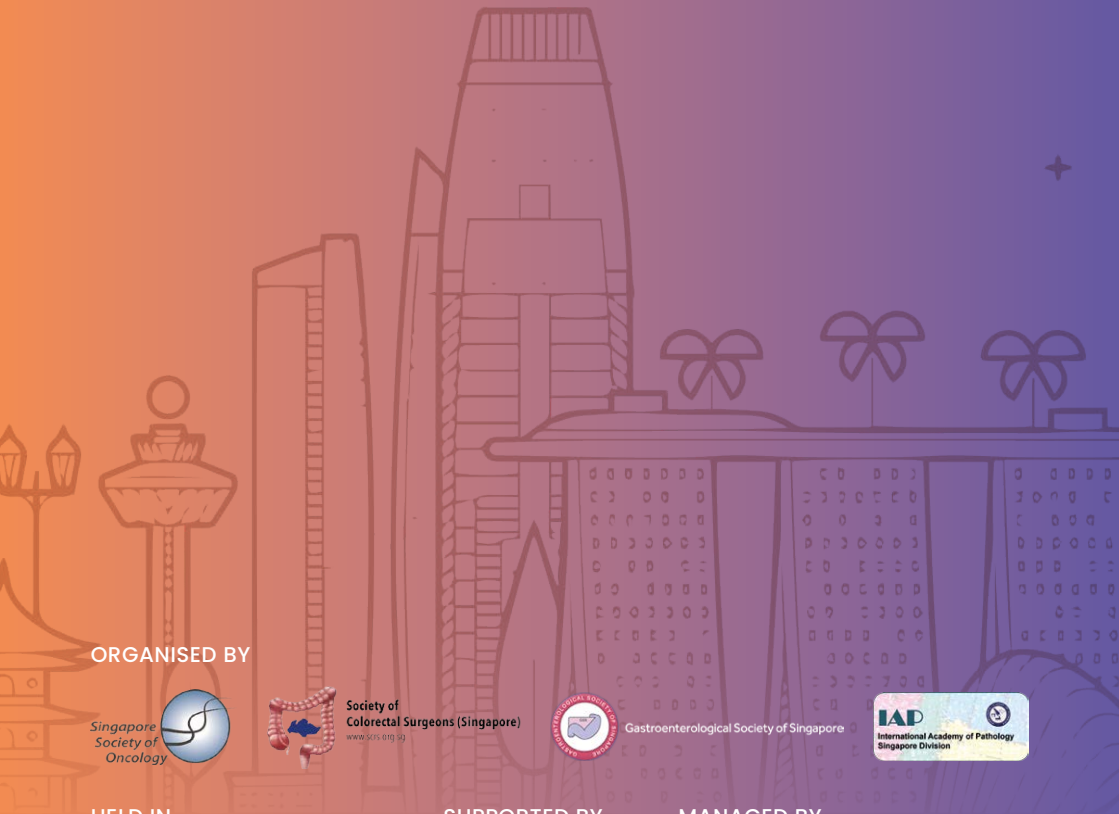
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11th Biennial Meeting of the International Society for Gastrointestinal Hereditary Tumours

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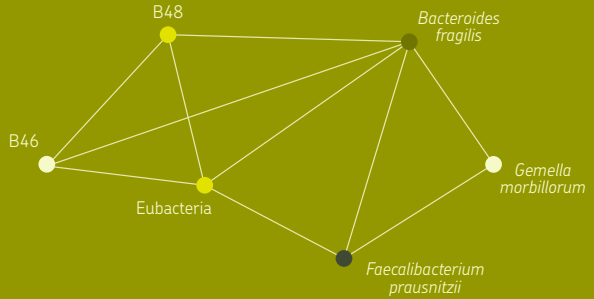


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Non-invasive faecal microbial test as a key tool to increase Colorectal Cancer screening efficiency by 12-15%¹

¹Malagón M, Oliver L, Ramió-Pujol S, Guardiola J, Balaguer F. Returning to endoscopy normality through the support of a new non-invasive faecal test based on microbial signatures. *Digestive and Liver Disease*. 2021; 53(12):1666–1668

