

9th ANNUAL MEETING
Heidelberg • 2025

9th Meeting of the European Hereditary Tumour Group (EHTG)

Friday, Sept 19th – Sunday, Sept 21st, 2025

Heidelberg, Germany

Final Programme





Invitation 9th Meeting - Heidelberg

Dear Members, Friends, Affiliates and Sponsors,

We are delighted to welcome you to the 9th Annual EHTG Meeting, taking place from September 19–21, 2025, in Heidelberg – the renowned City of Science.

Join us for a dynamic and thought-provoking programme featuring cutting-edge and controversial topics, in-depth expert debates, and valuable insights from the patient perspective.

Beyond the scientific sessions, this year's meeting offers a unique opportunity to strengthen collaboration within our growing community.

We look forward to inspiring discussions, new connections, and shaping the future of hereditary tumour research together.

Please visit www.ehtg.org for more information and contact gs007@ehtg.org for all organisational queries.

We are excited to welcome you to Heidelberg in September!

Sincerely yours

EHTG programme committee and board members



Organisation

Board Members: Aysel Ahadova (Germany)
 Peter Bauerfeind (Switzerland)
 John Burn (United Kingdom)
 Mev Dominguez-Valentin (Norway)
 Saskia Haupt (Germany)
 Kelly Kohut (United Kingdom)
 Florian Kühn (Germany)
 Fiona Lalloo (United Kingdom)
 Laura Monje-Garcia (United Kingdom)
 Gabriela Möslin (Germany)
 Neil Ryan (United Kingdom)
 Julian Sampson (United Kingdom)
 Toni Seppälä (Finland)

Programme Committee: Florian Kühn (Germany, Programme Director)
 Aysel Ahadova (Germany)
 Ann-Sofie Backman (Sweden)
 Richard Gallon (United Kingdom)
 Elizabeth Half (Israel)
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<https://www.frauenbad-heidelberg.de/>

Registration and Registration Fee: See online via www.ehtg.org

Certification: CME Accreditation (UEMS/EACCME):
 18 credit points



Landesärztekammer
 Baden-Württemberg

LÄK Baden- Württemberg:
 18 credit points Cat.B

Congress Organisation: Miaglossa GmbH,
 Pilgrimstraße 6, 50674 Köln



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Meeting Overview

Day 1 – Friday, September 19th

Time	Event
10:00-12:05	Hereditary Cancer Syndromes for Clinicians – Essentials you need to know!
12:05-13:15	Opening Ceremony and Welcome Lunch
13:15-14:30	Current Surgical Indications in Hereditary Cancer Syndromes
14:30-16:00	Artificial Intelligence - The Future Decision Maker in Hereditary Cancer Syndromes?
16:00-16:20	Coffee Break
16:20-16:50	Panel Discussion: Personalized Screening in Hereditary Cancer?
16:50-17:10	Abstracts: Gastroenterology I
17:10-18:10	Spotlight on Pancreas
18:10-18:30	Abstracts: Gastroenterology II
18:30-18:40	Award Ceremony – Travel Grants
18:40 +	Get Together at FRAUENBAD, Heidelberg
20:00 +	Y-EHTG evening at URBAN KITCHEN

Meeting Overview

Day 2 – Saturday, September 20th

Time	Event
08:00-08:45	PLSD Business Meeting
08:45-09:40	Update on Guidelines
09:40-10:00	Coffee Break
10:00-11:40	Rethinking Genetics
11:40-12:00	E-health – the Future of Digital Medicine
12:00-13:00	Y-EHTG
13:00-13:50	Lunch
13:50-14:00	AWARD Ceremony – Y-EHTG
14:00-14:30	Keynote: Tracing the Life History of Colorectal Cancer with Hypermutable DNA
14:30-15:45	Liquid Biopsy
15:45-16:05	Coffee Break
16:05-17:30	Translational Immunology and Immunoprevention
17:30-17:50	Big Trends in MSI Research
17:50-18:30	EHTG Nursing Session
20:00 +	Conference Dinner at MOLKENKUR



Meeting Overview




Day 3 – Sunday, September 21st

Time	Event
08:30-09:00	EHTG Business Meeting
09:05-10:40	Prevention of the Preventable
10:40-11:05	Coffee Break
11:05-12:15	Patient Perspectives
12:15-13:30	Gynecological Tumors - Implications for Hormonal Status and Fertility
13:30-14:00	Lunch Break
14:00-14:30	My most Challenging Case
14:30-15:45	CMMRD - what's new?
15:45-16:00	Meeting Summary and Closing Remarks



Scientific Programme

Day 1 – Friday, September 19th

- 10:00-12:05 Hereditary Cancer Syndromes for Clinicians – Essentials you need to know!**
Chairs: Neil Ryan (United Kingdom)
Sonia Kupfer (USA)
Naim Abu Freha (Israel)
- 10:00-10:15 Genetic Background of Hereditary Cancer Syndromes**
Laura Valle Velasco (Spain)
- 10:15-10:30 Diagnostics in MSI Tumors – IHC or PCR? Somatic or Germline?**
Matthias Kloor (Germany)
- 10:30-10:45 Gastroenterological Surveillance in Hereditary Cancer Syndromes**
  *Robert Hüneburg (Germany)*
- 10:45-11:00 Therapeutic Management of Lynch Syndrome: Immunotherapy for everyone?**
  *Romain Cohen (France)*
- 11:00-11:15 Basic Elements of Genetic Counselling for Hereditary Cancer**
Kelly Kohut (United Kingdom)
- 11:15-11:35 DISCUSSION**
- 11:35-12:05 ABSTRACTS**
- Outcomes from the English National Health Service Lynch Syndrome Transformation Project: Nationally Coordinated Delivery of Universal Testing for Lynch Syndrome**
and
The New English National Bowel Cancer Screening Programme (BCSP) for Lynch Syndrome
Kevin Monahan (United Kingdom)
- Expanding Lynch Syndrome Spectrum: the Role of Non-Colorectal/Endometrial Malignancies and Implications for Universal Tumor Screening**
and

Scientific Programme

Day 1 – Friday, September 19th

Analysis of the Performance of Universal Tumor Screening for Lynch Syndrome on Consecutive 1022 Patients Resected for Colorectal Cancer.

Emanuele Urso (Italy)

Upper Gastrointestinal Surveillance in Lynch Syndrome

Katrin van Beekum (Germany)

Distinct Genomic Landscape, Constrained Hypermutation and Absence of TERT Promoter Mutations in Lynch Syndrome-Associated Urothelial Cancer

Jussi Nikkola (Finland)

12:05-13:15 Opening Ceremony and Welcome Lunch

Welcome Address

Gabriela Möslin (Chair)

Florian Kühn (Programme Director)

Aysel Ahadova (Local Host)

Introduction of the IJC Special Issue

Christoph Plass (Editor-in-Chief, IJC)

13:15-14:30 Current Surgical Indications in Hereditary Cancer Syndromes

Chairs: Gabriela Möslin, (Germany)

Florian Kühn (Germany)

Benedito Rossi (Brazil)

13:15-13:30 Lynch Syndrome: do we still need Surgery?

David Liska (USA)

13:30-13:45 Surgical Intervention in Polyposis Syndromes - When is it Mandatory?

Ashish Sinha (United Kingdom)

13:45-14:00 Update on Technical Aspects of Surgery for FAP

Yann Parc (France)

14:00-14:05 DISCUSSION



Scientific Programme

Day 1 – Friday, September 19th

14:05-14:20 **Debate: Prophylactic Gastrectomy for CDH1 Carriers**

Uberto Fumagalli Romario (Italy)

Ophir Gilad (USA)

14:20-14:30 **ABSTRACTS**

Risk of Desmoid Formation in FAP after Minimally Invasive versus Open Restorative Proctocolectomy

Benjamin Zare (United Kingdom)

The Impact of Hysterectomy on Subsequent Colonoscopy in Women with Lynch Syndrome.

Hanne Hyldebrandt (Norway)

14:30-16:00 **Artificial Intelligence - The Future Decision Maker in Hereditary Cancer Syndromes?**

*Chairs: Magnus von Knebel Doeberitz (Germany)
Sanne Bajwa-ten Broeke
(The Netherlands)*

14:30-14:50 **Keynote: How to Employ AI to Fight Cancer?**

Julio Mayol (Spain)

14:50-15:05 **AI in Genetics**

Susanna Zucca (Italy)

15:05-15:20 **AI in Endoscopy**

Marco Spadaccini (Italy)

15:20-15:35 **AI in Histopathology - Helper or Killer?**

Peter Schirmacher (Germany)

15:35-15:45 **DISCUSSION**

15:45-16:00 **ABSTRACTS**

Evaluating the Potential of Artificial Intelligence-Based Algorithms to Predict Beta-2-Microglobulin Mutations in Mismatch Repair-Deficient Colorectal Cancer

Mozzam Motiwala (Germany)

Scientific Programme

Day 1 – Friday, September 19th

Evaluation of a RAG-based AI Chatbot for Neurofibromatosis Type 1 Using Simulated Patient Queries: a Genetic Counselor-Led Validation Study

Mashu Futagawa (Japan)

16:00-16:20 Coffee Break

16:20-16:50 Panel Discussion: Personalized Screening in Hereditary Cancer?



Sonia Kupfer (USA), Moderator

Kevin Monahan (United Kingdom)

Elizabeth Half (Israel)

Francesc Balaguer Prunés (Spain)

Elena Stoffel (USA)

16:50-17:10 Abstracts: Gastroenterology I

Chairs: Kevin Monahan (United Kingdom)

Elizabeth Half (Israel)

Adopting an Extended Interval Approach in Individuals with Lynch Syndrome Could Result in a High Rate of Interval Colorectal Cancer at Advanced Stage

Aharoni Golan (Israel)

Risk of Ileal Neoplasia in Lynch Syndrome Patients

Katrin van Beekum (Germany)

In Path_MMR Carriers, Diagnosis of Cancer in One Organ Does Not Modify the Probability for Cancer in Another Organ

and

Three Mechanisms May Explain Why Colonoscopy Does Not Reduce Colorectal Cancer Incidence in Three of the Four Lynch Syndromes

Pål Møller (Norway)



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Day 1 – Friday, September 19th

17:10-18:05 Spotlight on Pancreas

*Chairs: Ann-Sofie Backman (Sweden)
Imogen Bennett (United Kingdom)*

17:10-17:25 Who and How to Screen for Pancreatic Cancer?
Matthias Löhr (Sweden)

17:25-17:40 Long-term Yield of Pancreatic Cancer Surveillance in High-Risk Individuals
Kasper Overbeek (The Netherlands)

17:40-17:55 NGS-Based Treatment for Pancreatic Cancer - Indication and Success Rates
Christoph Springfeld (Germany)

17:55-18:05 DISCUSSION

18:05-18:30 Abstracts: Gastroenterology II

*Chairs: Elena Stoffel (USA)
Francesc Balaguer Prunés (Spain)
Stephanie Poo (United Kingdom)*

Excellent Survival for Lynch Syndrome Individuals with Screen Detected Pancreatic Cancer and MMRD Status: Data from the PLSD
Hadar Edelman-Klapper (Israel)

Survival of Lynch Syndrome Patients with Biliary Carcinoma: Data from the PLSD
Lior H. Katz (Israel)

Duodenal Involvement in MUTYH-Associated Polyposis: Findings from an International Prospective Study and Implications for Endoscopic Surveillance
Imogen Bennett (United Kingdom)

Follow-Up, Cancer Risk and Mortality in Peutz-Jeghers Syndrome: a Cohort Study from the PRED-IdF Network
Maxime Remond (France)

Scientific Programme

Day 1 – Friday, September 19th

18:30-18:40 Award Ceremony – Travel Grants

18:40 + Get-Together at Frauenbad, Heidelberg

20:00 + Y-EHTG evening at URBAN KITCHEN
(same building as Frauenbad)
at your own expense!



Scientific Programme

Day 2 – Saturday, September 20th

08:00-08:45 **PLSD Business Meeting**
Chair: Mev Dominguez-Valentin (Norway)

08:45-09:40 **Update on Guidelines**
Chairs: Mev Dominguez-Valentin (Norway)
Kai-Keen Shiu (United Kingdom)

08:45-09:00 **European Initiative on Harmonized
Lynch Syndrome Guidelines**
Giulia Martina Cavestro (Italy)

09:00-09:15 **EMQN Best Practice Guidelines for MSI**
Richard Gallon (United Kingdom)



09:15-09:30 **Updated BRCA Guidelines (Gastrointestinal)**
Elizabeth Half (Israel)
Zohar Levi (Israel)
Lior Katz (Israel)

09:30-09:40 **DISCUSSION**

09:40-10:00 **Coffee break**

10:00-11:40 **Rethinking Genetics**
Chairs: Maartje Nielsen (The Netherlands)
Vince Kornel Grolmusz (Hungary)
Romy Walker (Australia)

10:00-10:15 **What is Universal about Testing: How We Do It
and How We Should Do It?**
Heather Hampel (USA)

10:15-10:30 **Cascade Testing Barriers: How to Overcome?**
  *Joanne Ngeow (Singapore)*

10:30-10:45 **Improving Genetic Diagnosis of Hereditary
Tumor Diseases: From Expanded Gene Panels
to Functional Genomics**
  *Barbara Klink (Germany)*

Scientific Programme

Day 2 – Saturday, September 20th

10:45-10:55 DISCUSSION

10.55-11.40 ABSTRACTS

Decoding Lynch Syndrome: Resolving Difficult to Categorize Findings in the DNA Mismatch Repair Genes MLH1, PMS2 and MSH2

Rodney Scott (Australia)

Improving Interpretation of POLE and POLD1 Exonuclease Domain Missense Variants via Functional Studies and AI-Based Pathogenicity Predictors

Laura Valle (Spain)

Extra-Intestinal Cancer Risk, Including Breast, Ovarian, and Endometrial Cancer, in MUTYH-Associated Polyposis (MAP)

Maartje Nielsen (The Netherlands)

Cancer Risks for ATM Variant Carriers in Ataxia-Telangiectasia Families and in Families Identified in Family Cancer Clinics

Fabienne Lesueur (France)

Testing *in vitro* Efficacy of Werner Helicase Inhibitors on Patient-Derived Tumor Organoid Models of Lynch Syndrome-Associated Colorectal Cancer

Julia Kolikova (Finland)

Characterisation of Rare Germline Genetic Variation in the RNF43 Gene in People with Serrated Polyposis Syndrome

Daniel Buchanan (Australia)

Phenotypic and Genotypic Profile of Lynch Syndrome in Latin America: A Multicenter Study

Mev Dominguez Valentin (Norway)

Comprehensive Germline and Somatic Analyses Delineate Mutational Mechanisms in Two Independent Patients Carrying a Combination of Germline Pathogenic Variants in APC and PMS2

Vince Kornel Grolmusz (Hungary)



Scientific Programme

Day 2 – Saturday, September 20th

Direct Letters to Relatives at Risk of Hereditary Cancer – A Multi-Center Randomized Trial on Healthcare-Assisted versus Family-Mediated Risk Disclosure (The Swedish DIRECT-Study)

Anna Rosen (Sweden)

11:40-12:00

Keynote: E-health – the Future of Digital Medicine

Introduction: John Burn (United Kingdom)

Speaker: Magnus von Knebel Doeberitz (Germany)



12:00-13:00

Y-EHTG

Chairs: Alethea Tang (United Kingdom)

Maria Rasmussen (Denmark)

Saskia Haupt (Germany)

12:00-12:15

Introduction, Prizes, News

Saskia Haupt (Germany)

12:15-13:00

ABSTRACTS

Whole Exome Sequencing of MLH1 Lynch Syndrome Colorectal Cancer Reveals Frequent Copy Number Alterations in MLH1-CTNNB1 Locus

Kalle Hokkanen (Finland)

Cancer Risks in First-Degree Relatives of Individuals with Biallelic Somatic DNA Mismatch Repair Mutations

Romy Walker (Australia)

Epigenetic Therapy for Mismatch Repair-Deficient Tumors

Anna Scheßl (Germany)

Evaluating Cost-Effectiveness of Preventive Strategies and the Impact of Specialized Care Pathways for Peutz-Jeghers Syndrome in Europe

Amalia Nicole Nanciu (Germany)

Scientific Programme

Day 2 – Saturday, September 20th

Description of the Coding Mutational Landscape Involved in the MSI Lynch Syndrome Colorectal Carcinogenesis, from Initiation (MMR-Deficient Crypts) to Tumor Progression (Adenoma, Adenocarcinoma)

Hugo Montémont (Germany)

Diving into Sex-Based Disparities: Systemic Biomarker Associations to Physical Activity in Lynch Syndrome Carriers

Minta Kärkkäinen (Finland)

PCR-Based Microsatellite Instability Testing: Pitfalls and Strategies for Enhancing Accuracy

Maria Kessler (Germany)

Study Plan: The Interplay Between Aging, Physical Activity and Immune Cells in Cancer Risk Regulation

Henna-Riikka Littunen (Finland)

Urological Malignancies in Lynch Syndrome

Antonia Schuler (Germany)

13:00-13:50 **Lunch**

13:50-14:00 **AWARD Ceremony – Y-EHTG**

14:00-14:30 **Keynote: Tracing the Life History of Colorectal Cancer with Hypermutable DNA**

Introduction: Aysel Ahadova (Germany)

Speaker: Kamila Naxerova (USA)

14:30-15:45 **Liquid Biopsy**

Chairs: Alessandro Mannucci (Italy)

Alex Duval (France)

Maria Rasmussen (Denmark)

14:30-14:45 **New Horizons of Liquid Biopsy Analysis Using Blood and CSF for Patients with Cancer Predisposition Syndromes**



Uri Tabori (Canada)



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Day 2 – Saturday, September 20th

- 14:45-15:00 **PREDiLynch**
Mev Dominguez-Valentin (Norway)
Toni Seppälä (Finland)
- 15:00-15:15 **Urine-Based Surveillance in Lynch Syndrome**
Michael Jackson (United Kingdom)
- 15:15-15:25 **DISCUSSION**
- 15:25-15:45 **ABSTRACTS**
- Multi-Omics Approaches to Uncover Liquid-Biopsy-Based Cancer-Predicting Biomarkers in Lynch Syndrome**
Tiina Jokela (Finland)
- LIFE-CNA: A Whole-Genome Liquid Biopsy Approach Beyond Single Variant Detection in Diverse Tumour Entities**
and
Liquid Biopsy Analysis of Advanced Breast Cancer Patients – A Real-life Perspective of Actionable Variants
Thomas Keßler (Germany)
- Leveraging Ultra Low-Coverage Whole Genome Sequencing in Liquid Biopsy for Early Cancer Screening: Insights from the PreveLynch Cohort**
Tomas Szemes (Slovakia)
- 15:45-16:05 **Coffee break**
- 16:05-17:30 **Translational Immunology and Immunoprevention**
Chairs: Matthias Kloor (Germany)
Stefanie Bärthel (Germany)
Henna-Riikka Littunen (Finland)
- 16:05-16:20 **Cancer Immunoprevention Approaches: Current Status**
  *Steve Lipkin (USA)*

Scientific Programme

Day 2 – Saturday, September 20th

16:20-16:35 **Organ-Sparing Therapy Options for Patients with Inherited Cancers – Decision Making and Patient Outcomes**

Antoine Dardenne (France)

16:35-16:50 **Panel Discussion: Perfect Vaccine: How Does It Look? Why We Need Vaccines in the Era of Immunotherapy?**

Speakers and Chairs

16:50-17:30 **ABSTRACTS**

Immune Surveillance in Lynch Syndrome: T Cell Receptor Repertoire Mapping and Clonal Expansion in Colorectal Neoplasia

Penelope Edwards (United Kingdom)

Microsatellite Instability Degree and Immunological Profile of MSH6-mutated Colorectal Carcinomas compared to other Lynch Syndrome-Associated Colorectal Carcinomas

Noah Cornelis Helderman (The Netherlands)

Unveiling Immune Surveillance in Lynch Syndrome: Insights from T cell Population Characterization

Joaquin Andres Castillo (Spain)

HLA Fine-Mapping and Immuno-peptidome-Wide Association Studies Identify Functional Colorectal Cancer Risk Modifiers and Frameshift Neoantigen Targets in Lynch Syndrome

Aaron Meyers (Australia)

HLA Genotypes as a Potential Cancer Risk Modifiers in Women Living with Germline Pathogenic BRCA1 Variants

Marcell Kakonyi (Hungary)

Scientific Programme

Day 2 – Saturday, September 20th

17:30-17:50 Keynote: Big Trends in MSI Research

Introduction: Matthias Kloor (Germany)

Speaker: Alex Duval (France)

17:50-18:30 EHTG Nursing Session

Chairs: Laura Monje-Garcia (United Kingdom)

Mechelle Loughrey (Ireland)

**17:50-18:00 Nursing Innovation in Hereditary Cancer:
Advancing Scope, Genomics, and Research
Within the Multidisciplinary Team**

Mechelle Loughrey (Ireland)

18:00-18:30 ABSTRACTS

**Opening Pandora's Box: A Qualitative Study on
the Psychological Impacts of Living with
an Inherited Colorectal Cancer Predisposition
Syndrome (PSYLIVED)**

Laura Monje-Garcia (United Kingdom)

**Implementation of Mainstream Lynch
Syndrome Testing in a High-Volume Colorectal
Cancer Service: Outcomes from a UK Tertiary
Centre**

Helen Francis (United Kingdom)

**Declining Genetic Testing Uptake among
Individuals from Lynch Syndrome and
Hereditary Breast and Ovarian Cancer Families**

Elizabeth Half (Israel)

Evening Programme

Conference Dinner at MOLKENKUR

Saturday, September 20, 20:00

Arrival:

Individual arrival by foot walking through the Old Town, followed by a ride on the historic funicular railway from Kornmarkt to Molkenkur.



Departure:

Return to hotels via staggered bus transfers between 22:30 and 24:00.

The Schlosshotel Molkenkur is located above the Heidelberg Castle, surrounded by nature and offering a sensational panoramic view over the Rhine plain.

A Brief History of Molkenkur

The so-called "Upper Castle," located on the present-day Molkenkur parking area, was the original Heidelberg Castle, first mentioned in records in 1225. After the construction of the "New Castle," the old structure was struck by lightning and destroyed on April 25, 1537.

In 1851, Albrecht Wagner began construction of what is now known as the Swiss House, the central building of the Molkenkur complex. This structure is a listed heritage building and has remained nearly unchanged since its completion.

The name Molkenkur comes from the "whey cure" (Molkenkur in German) – a once-popular therapeutic treatment using dairy products that gave the site its name and historical significance as a health retreat.



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Day 3 – Sunday, September 21th

08:30-09:00 EHTG Business Meeting
Chairs: *Toni Seppälä (Finland)*
Gabriela Möslin (Germany)

09:00-10:40 Prevention of the preventable
Chairs: *C. Richard Boland (USA)*
Heather Hampel (USA)
Kelly Kohut (United Kingdom)

09:00-09:15 Addressing Lifestyle Factors to Prevent Cancer in High-Risk Individuals
John Mathers (United Kingdom)

09:15-09:30 Patients' Preference for Preventative Strategies / Uptake of Aspirin for Lynch Syndrome Chemoprevention
Michael Hall (USA)

09:30-09:45 Is Less More? CAPP3 Data on Aspirin for Cancer Prevention in Lynch Syndrome
 *John Burn (United Kingdom)*

09:45-10:00 Mesalamine-based Prevention of Inherited Cancer
Ann-Sofie Backman (Sweden)

10:00-10:15 Employing Microbiota to Prevent Cancer in Hereditary Tumor Syndromes
Phil Quirke (United Kingdom)

10:15-10:25 DISCUSSION

10:25-10:40 ABSTRACTS

Linking Lynch Syndrome Mutations to the Gut Microbiome Profile: A Predictive Approach for Colorectal Cancer Risk
Elizabeth Half (Israel)

Association between Blood Levels of Selenium, Zinc and Copper and Survival in Colorectal Cancer Patients with Lynch Syndrome
and

Scientific Programme

Day 3 – Sunday, September 21th

SELINA – Lowering of Cancer Risk and All-Cause Mortality by Optimization of Se and As Blood Levels in Females from Families with Hereditary Breast Cancer

Jan Lubinski (Poland)

10:40-11:05 **Coffee break**

11:05-12:15 **Patient Perspectives**

Chairs: Kelly Khout (United Kingdom)

Georgina Hoffmann (Germany)

Coping with Lynch syndrome – Patient Perspectives

11:05-11:15 **Modifiable Lifestyle Factors**

Aung Ko Win (Australia)

11:15-11:20 **Interview and Panel Discussion**

What is Helping Us Living with Lynch and Facing Our Lifelong Cancer Risk? Where Do We See Gaps in Care and Support?

Georgina Hoffmann (Germany)

11:20-11:25 **Lynch Choices:**

<https://canchoose.org.uk/> **Decision Aid Tool**

Kelly Kohut (United Kingdom)

11:25-11:30 **Lynch Syndrome UK**

Tracy Smith (United Kingdom)

11:30-11:45 **Raising Detection: Finding the Missing 95%**

ALL PATIENT ORGANIZATIONS

Lynch syndrome Awareness: Achievements and Future Aspiration

11:45-11:50 **Awareness Campaigns Alive and Kick'n**

Dave Dubin (USA)

11:50-11:55 **Awareness Campaign Lynch Syndrome Ireland**

Roberta Horgan (Ireland)

11:55-12:00 **Awareness Campaign SemiColon**

Georgina Hoffmann (Germany)

Nicola Reents (Germany)

12:00-12:05 **DISCUSSION**

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12:05-12:15 ABSTRACTS

Addressing the Psycho-Social Needs of Hereditary Cancer Risk Patients: “A Much Needed Specialist Service”

Louise O'Driscoll (Ireland)

Impact of Risk-Reducing Hysterectomy on Health-Related Quality of Life in Lynch Syndrome Women: PRESCORES Study

Ranjit Manchanda (United Kingdom)

12:15-13:30 Gynecological Tumors – Implications for Hormonal Status and Fertility

Chairs: Denise Nebgen (USA)

Neil Ryan (United Kingdom)

André Pfob (Germany)

12:15-12:30 Perspectives on Early Detection Strategies

Neil Ryan (United Kingdom)

12:30-12:45 Risk-Reducing Bilateral Salpingo- Oophorectomy for Ovarian Cancer and Opportunities for Novel Screening Approaches

Zsofia Stadler (USA)

12:45-13:00 Fertility Preservation in Patients with BRCA Mutations or Lynch Syndrome

Claudia Marchetti (Italy)

13:00-13:15 Keynote: Population Screening Strategies and the Implications of the PROTECTOR Study

Ranjit Manchanda (United Kingdom)

13:15-13:30 ABSTRACTS

What are the Lifetime Endometrial Cancer Risk Thresholds for Surgical and Medical Prevention of Endometrial Cancer and Estimating its Cost-Effectiveness

Ranjit Manchanda (United Kingdom)

A New Method of Screening Female Lynch Syndrome Carriers for Endometrial Cancer: The Ellele Sampling Device

and

Scientific Programme

Day 3 – Sunday, September 21th

Microsatellite Womb Surveillance (MEWS) Study

Neil Ryan (United Kingdom)

13:30-14:00 **Lunch**

14:00-14:30 **My most Challenging Case**

*Chairs: Giulia Martina Cavestro (Italy)
Toni Seppälä (Finland)*

Somatic PTEN Mosaicism in Colorectal Polyposis Patients

Diantha Terlouw (The Netherlands)

A Challenging Case of Gastric Cancer in an Elderly CDH1 Mutation Carrier Without a Family History

Hadar Edelman-Klapper (Israel)

Distal Cholangiocarcinoma Arising in Familial Adenomatous Polyposis: A Unique Case Presentation

Julius Hüneburg (Germany)

The Dark and the Light. The Difficult Journey of a Patient with a Huge Cancer of the Ascending Colon and Lynch Syndrome.

Emanuele Urso (Italy)

14:30-15:30 **CMMRD - what's new?**

*Chairs: Gabriel Capella (Spain)
Chrystelle Colas (France)
Steve Lipkin (USA)*

14:30-14:45 **ERN GENTURIS Guidelines**

*Chrystelle Colas (France)
Katharina Wimmer (Austria)*

14:45-15:00 **Care for CMMRD Database**

Pauline Hoarau (France)

15:00-15:15 **Updates from IRRDC: From Expanding Landscapes to Novel Therapies**

Anirban Das (Canada)



Scientific Programme

Day 3 – Sunday, September 21th

15:15-15:20 DISCUSSION

15:20-15:35 ABSTRACTS

Modelling the Origin of Glioblastoma in Constitutional Mismatch Repair Deficiency Syndrome

Manuel Torralba (Spain)

Screening Program to Improve Identification of Constitutional Mismatch Repair Deficiency in Spain

Oscar Mesia Carbonell (Spain)

First Report of POLD1 Homozygote Patient with Double Colon Cancer and Polyposis

Salwa Ben Yahia (The Netherlands)

15:35-15:45 DISCUSSION

15:45-16:00 **Meeting Summary and Closing Remarks**

Gabriela Moeslein (Germany)

Toni Seppälä (Finland)

Poster Exhibition

Abu Shtaya, Aasem
Israel

POT1 Variants: Drivers of
Telomeric Elongation, Diverse
Malignancies, and High Tumor
Burden

Akdeniz, Bayram
Norway

Understanding Cancer Risk
Variability in Lynch syndrome: The
Role of Polygenic Risk Scores

Aswolinskiy, Witali
Germany

Interpretable Deep Learning for
Microsatellite Instability Prediction
in Colorectal Cancer From H&E-
stained Whole Slide Images

Avsec, Eva
Slovenia

Mismatch Repair Deficient Breast
Cancer in Patients with Lynch
syndrome

Bulbaai, Marjolein
The Netherlands

Fish Intake and Colorectal
Neoplasm Risk in Individuals with
Lynch syndrome

Butz, Henriett
Hungary

When Two Is Not Enough:
Complexity of Cancer Patients with
Multiple Germline Mutations in
Tumour-Predisposing Genes

Campoy, Séphora
France

Systematic Review and Meta-
analysis of Lifetime Cancer Risk in
Lynch syndrome: Impact of
Methodology and Study Design

Colas, Crystelle
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Prevalence of Somatic Mosaicism
in the APC Gene in Patients with
Unexplained Colorectal
Adenomatous Polyposis or
Multiple Adenomatous Colorectal
Polyps.

Dardenne, Antoine
France

Cancer Incidence and Relevance
of Surveillance Recommendation
in Lynch syndrome Patients
followed in a Specialized Resource
Center

Dominguez Valentin, Mev
Norway

Microsatellite Instability and
Hereditary Risk in Ovarian
Cancer

Poster Exhibition

Erisen, Gizem Mehtap Germany	Spatial Heterogeneity of Primary Lynch syndrome-associated Cancers.
Erkan, Erdogan Pekcan Finland	Single Cell Profiling of Tissue and Tumor Microenvironments in Mismatch Repair Deficient Colorectal Cancers
Fritzell, Kaisa Sweden	Impact on Mental and Physical Health, and Well-being in Patients with Polyposis Syndromes: A Scoping Review
Fujita, Hiroko Japan	Genetic Counselling for three Japanese Families with Gastric Adenocarcinoma and Proximal Polyposis of the Stomach (GAPPS)
Guillen Ponce, Carmen Spain	Cascade Testing of Low and Moderate Risk Genes in Hereditary Breast and Ovarian Cancer Syndrome.
Guillen Ponce, Carmen Spain	Pathogenic Variants in Patients with Early-onset Gastrointestinal Cancer.
Hirasawa, Akira Japan	Prospective Cohort Study with a Biobank for Hereditary Tumor Syndromes in Japanese -Mid-West Japan Hereditary Tumor Cohort-
Kobrow, Christian Germany	Transformation Behavior of Images in Latent Space
Lautrup, Charlotte Kvist Denmark	New RPS20 Gene Variant Underpins – the Role of RPS20 as a Colorectal Cancer Predisposition Gene without Extracolonic Manifestations
Lubinski, Jan Poland	Elements Ratio as a Marker of Cancer Risk and Mortality in BRCA1 Mutation Carriers.

Poster Exhibition

Monahan, Kevin
United Kingdom

Diagnostic Yield of Genetic Testing and Risk Factors for Colorectal Cancer in Patients with Serrated Polyposis Syndrome

Montémont, Hugo
Germany

Immunoguided Laser-Assisted Microdissection for Whole Exome and RNA-Sequencing: A Precise Technology to Explore Genomics and Transcriptomics Alteration in Early Stage of Cancer

Olkinuora, Alisa
Switzerland

Exome Sequencing Study identifies DNA Glycosylase Gene Variants enriched in Swiss Colon Polyposis Patients

Poo, Stephanie
United Kingdom

Colonoscopy Yield in Lynch Syndrome at a UK Tertiary Centre with High Baseline Quality Metrics

Ryan, Neil
United Kingdom

A Mismatch in Testing: A Retrospective Analysis of Mismatch Repair Testing in Endometrial Cancer and Lynch syndrome Diagnosis in Multiple Specialist Centres in the UK and Ireland (March 2022–March 2023)

Silva, Sofia
Spain

NGS and all that it reveals

Tada, Haruka
Japan

A Parent-child case of Peutz-Jeghers Syndrome: Definitive Diagnosis using Microarray Chromosomal Analysis and Subsequent Surveillance

Urakawa, Yusaku
Japan

Late-Onset Ovarian Cancer in BRCA2 Carriers: Rethinking Age Thresholds for Risk Reducing Surgery.

Poster Exhibition

Urso, Emanuele
Italy

Analysis of a Consecutive Series of 402 Cases discussed at the Multi-disciplinary Oncology Group for Hereditary Gastrointestinal Tumors (GOM-TEGI) at the University Hospital of Padua - Veneto Institute of Oncology

Valle, Laura
Spain

APC I1307K and Clinical Management: Insights from UK Biobank on Colorectal and other Cancer Risks in Ashkenazi and Non-Ashkenazi Whites

van der Werf-'t Lam, Anne-Sophie
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Cancer Risks for MSH6 Pathogenic Variant Carriers

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