Programme

8TH ANNUAL MEETING
Online · 2024

8th Meeting of the European Hereditary Tumour Group (EHTG)

Friday, Sept 27th – Saturday, Sept 28th, 2024

Sponsored by:

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The Newcastle upon Tyne Hospitals NHS Foundation Trust

Invitation 8th Meeting - Online

Dear Members, Friends, Affiliates and Sponsors,

We are delighted to invite you to the 8th annual EHTG meeting which will take place on the **27th and 28th of September 2024 – online!** Our lively programme includes selected hot and controversial topics, in depth debates, audience discussion including the opportunity to discuss challenging clinical cases. We also aim to utilise the event to further explore collaboration across our members.

Day 1 (Friday) will include plenary sessions, a debate on an area of current clinical equipoise and a keynote lecture on an emerging hot topic. On Day 2 (Saturday), there will be interactive working groups (WG), a Dragon's Den session where young scientists pitch their research ideas – organized by the yEHTG, and an update on chemoprevention in hereditary cancer. Furthermore, you will be able to present your most challenging cases and discuss them with an expert panel.

Please visit **www.ehtg.org** for more information and contact **gs007@ehtg.org** for all organisational queries. If you have any suggestions, please get in touch with any of the programme members.

We are excited to see you in September!

Sincerely yours
EHTG programme committee and board members

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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 Kiddom)** **Gabriela Möslein** (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) **Neil Ryan** (United Kingdom) **Laura Thomas** (United Kingdom)
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Registration Fee:	See online via www.ehtg.org
Certification:	We have applied for certification by EACCME
Technical organisation online part:	Miaglossa GmbH, Köln support@miaglossa.com

Notes

Scan the QR code or click the button to sign up today

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Scientific Programme

Day 1 – Friday, September 27th

12:55	**Welcome** *John Burn (United Kingdom)*
13:00-14:15	**Lynch syndrome** *Chairs: Ingrid Winship (Australia)* *Kevin Monahan (United Kingdom)*
13:00-13:15	**Lynch syndrome - beyond the colon and the endometrium** *John Burn (United Kingdom)*
13:15-13:30	**Dominantly inherited MSI cancers - an update of the PLSD** *Mev Dominguez-Valentin (Norway)*
13:30-13:45	**Update on the English National Lynch syndrome Registry (National Disease Registration Service)** *Steven Hardy (United Kingdom)*
13:45-14:00	**Lynch syndrome guideline harmonization - report on the work in progress** *Giulia Martina Cavestro (Italy)*
14:00-14:15	**Immunotherapeutic options for patients with Lynch syndrome** *Andrea Cercek (USA)*
14:15-15:15	**Fertility / Pregnancy** *Chairs: Denise Nebgen (USA)*
14:15-14:30	**Management of colorectal cancer during pregnancy** *Jonas Ludvigsson (Sweden)*
14:30-14:45	**Fertility preservation in gynaecological cancer** *Richard Anderson (United Kingdom)*
14:45-15:00	**Fertility preservation in patients with gastrointestinal cancer syndromes** *Valeria Vanni (Italy)*
15:00-15:15	**Surgery and quality of life** *José Gaston Guillem (USA)*

Scientific Programme

Day 1 – Friday, September 27th

15:15-15:30	**Coffee break**
15:30-16:30	**Guidance from patient organizations** *Chairs: Nicola Reents (Germany)* *Laura Monje-Garcia (United Kingdom)*
15:30-15:40	**Living with Lynch: patient perspective** *Georgina Hoffmann (Germany/Canada)*
15:40.15:50	**Examining Lynch syndrome Carriers' Perceptions of Care in the UK and Ireland** *BJ Olsen (Northern Ireland)*
15:50-16:00	**Qualitative interview study: Exploring the opinions and attitudes towards nutritional advice from individuals with Lynch syndrome** *Isabelle Rennocks (United Kingdom)*
16:00-16:10	**Who has the responsibility to inform relatives at risk of hereditary cancer? A population-based survey in Sweden** *Anna Rosén (Sweden)*
16:10-16:20	**CMMRD – A Family Perspective** *Marisa I. Lopez-Messecar (USA)*
16:20-16:30	**Enhancing Lynch Syndrome management and services in the UK: The dual role of Lynch Syndrome UK in supporting patients and healthcare professionals** *Tracy Smith (United Kingdom)*

The time scheduled in the programme is referring to CET (Central European Time)

Scientific Programme

Day 1 – Friday, September 27th

16:30-17:15	**Immunotherapy for colon and rectal cancer. Organ preservation for everyone?** *Chair: David Ljungman (Sweden)* Pro: *Julio Garcia-Aguilar (USA)* Contra: *Dieter Hahnloser (Switzerland)* Discussion with panel and auditorium
17:15-18:00	**Desmoid disease** *Chairs: Elizabeth Half (Israel)* *David Liska (USA)*
17:15-17:30	**Biological background and screening: Who and how should we screen for desmoids?** *Alona Zer (Israel)*
17:30-17:45	**How and when to treat desmoid tumors: Medical treatment options for desmoid disease** *Silvia Stacchiotti (Italy)*
17:45-18:00	**Surgical options for treatment of desmoid tumors** *Yann Parc (France)*
18:00-18:30	**Keynote lecture I** *Chair: Julian Sampson (United Kingdom)* Targeting the Gut Microbiome for Cancer prevention and treatment *Phil Quirke (United Kingdom)*

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1 Schmidt A, Beyna T, Schumacher B, Meining A, Richter-Schrag HJ, Messmann H, et al. Colonoscopic full-thickness resection using an over-the-scope device: a prospective multicentre study in various indications. Gut 2018 Jul;67(7):1280-1289.

2 Meier B, Stritzke B, Kuellmer A, et al. Efficacy and safety of endoscopic full-thickness resection in the colorectum: Results from the German colonic FTRD registry. Am J Gastroenterol 2020; 115(12):1998-2006.

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Scientific Programme

Day 2 – Saturday, September 28th

09:00-10:00	**Y-EHTG**
09:00-09:10	**Y-EHTG – who we are and what we do** *Saskia Haupt (Germany)* *Alethea Tang (United Kingdom)*
09:10-09:40	**Dragons' Den – update on ongoing projects** *Chairs:* *Núria Dueñas Cid (Spain)* *Sanne Bajwa-ten Broeke (The Netherlands)* *Naim Abu-Freha (Israel)* *Panelists:* *Julian Sampson (United Kingdom)* *Svetlana Bajalica-Lagercrantz (Sweden)* *Emma Crosbie (United Kingdom)* *Christina Therkildsen (Denmark)* *Speakers:* *Sanne Bajwa-ten Broeke (The Netherlands)* *Neil Ryan (United Kingdom)* *Caroline Kahn (Denmark)*
09:40-09:50	**Pitch new studies and projects of Y-EHTG members: Predicting the unpredictable: using machine learning models to classify non-coding variants in the MMR genes** *Carlos Urzua Traslavina (The Netherlands)*
09:50-10:00	**Discuss your ideas!** *Chairs:* *Maria Rasmussen (Denmark)* *Laura Roht (Estonia)*
10:00-12:00	**4 parallel Working Groups**
WG 1	**Polyposis**
WG 2	**Carcinogenic mechanisms and pathways / early onset cancers / novel associations**
WG 3	**Genetics Counselling & Management (incl. Lab diagnosis, RNA seq, MMR variant interpretation)**
WG 4	**Extra-colonic cancers in hereditary tumour syndroms**

Scientific Programme

Day 2 – Saturday, September 28th

10:00-12:00	**WG 1**	**Polyposis**
	Chairs:	*Dan Buchanan (Australia)* *Stefan Aretz (Germany)* *Gloria Zaffaroni (Germany/Italy)*
10:00-10:12	**Different orders of events lead to colorectal cancer in familial adenomatous polyposis and inherited MSI cancers: implications for screening.** *Pål Møller (Norway)*	
10:12-10:24	**Immune characterisation of adenomas in Familial Polyposis Syndrome patients** *Sion Roberts (United Kingdom)*	
10:24-10:36	**Molecular and clinical differences between constitutional and somatic POLE and POLD1 exonuclease domain pathogenic variants in cancer** *Julen Viana-Errasti (Spain)*	
10:36-10:48	**The Role of Ferroptosis in Sporadic and Familial Intestinal Cancers** *Imogen Bennett (United Kingdom)*	
10:48-11:00	**Clinical phenotype, diagnostic route and unique presentations of PTEN Hamartoma Tumor Syndrome** *Einat Ritter (Israel)*	
11:00-11:12	**Identifying APC mosaicism in unexplained adenomatous polyposis: paving the way for the new susceptibility gene discovery** *Eric Joo (Australia)*	
11:12-11:24	**PIGA mutations and glycosylphosphatidylinositol anchor dysregulation in polyposis-associated duodenal tumorigenesis** *Laura Thomas (United Kingdom)*	
11:24-11:36	**Extended family with an inherited pathogenic variant in POLD1 provides strong evidence for recessive effect of polymerase delta proofreading deficiency in human cells** *Laura Valle (Spain)*	

Scientific Programme

Day 2 – Saturday, September 28th

10:00-12:00	WG 2	**Carcinogenic mechanisms and pathways / early onset cancers / novel associations**
	Chairs:	*Aysel Ahadova (Germany)* *Julian Sampson (United Kingdom)*
10:00-10:12		**Systemic FSP-specific T cell responses as predictive biomarker for immune checkpoint blockade therapy response in patients with microsatellite-unstable cancers?** *Lena Bohaumilitzky (Germany)*
10:12-10:24		**Risk stratification of colorectal cancer using a combined effect of polygenic risk score and mismatch repair genes for Norwegian cohort** *Bayran Akdeniz (Norway)*
10:24-10:36		**Prevalence of unexplained mismatch repair deficiency in colorectal cancer diagnosed at a tertiary cancer referral centre** *Lauren O'Connell (Ireland)*
10:36-10:48		**Microsatellite instability at U2AF-binding polypyrimidic tract sites perturbs alternative splicing during colorectal cancer initiation (Lynch syndrome and sporadic)** *Hugo Montemont (France/Germany)*
10:48-11:00		**HLA genotype as a colorectal cancer risk modifier in Lynch syndrome** *Aaron Meyers (Australia)*
11:00-11:12		**Tumor occurrence during follow up of 93 patients with Lynch syndrome treated with immune-check point inhnibitor for metastatic digestive cancer between 2015 and 2024.** *Antoine Dardenne (France)*
11:12-11:24		**Artificial intelligence-based tools for molecular subtype classification of colorectal carcinomas** *Julius Krauch (Germany)*

Scientific Programme

Day 2 – Saturday, September 28th

11:24-11:36		**Aetiological insights into early-onset colorectal cancer and adenoma tumourigenesis through genomic tumour mutational signature profiling** *Daniel Buchanan (Australia)*
11:36-11:48		**Diet and early-onset colorectal cancer: Validation of a novel ad hoc designed food frequency questionnaire** *Marta Puzzono (Italy)*
11:48-12:00		**Constitutional epimutations in LTBP4 and in BRCA1 as drivers of early-onset colorectal cancer** *Mariona Terradas (Spain)*
10:00-12:00	WG 3 *Chairs:*	**Genetics Counselling & Management (incl. Lab diagnosis, RNA seq, MMR variant interpretation)** *Kelly Kohut (United Kingdom)* *Fiona Lalloo (United Kingdom)*
10:00-10:12		**A novel low cost amplicon based multiplex microsatellite instability assay with 22 driver BRAF/RAS mutations deployed across 3 million population allows discontinuation of routine colorectal cancer immunohistochemistry and enhanced Lynch syndrome detection.** *Richard Gallon (United Kingdom)*
10:12-10:24		**Interim outcomes from the NHS England Lynch syndrome Transformation Project: Finding the Missing 95%** *Kevin Monahan (United Kingdom)*
10:24-10:36		**The new English National Bowel Cancer Screening Programme (BCSP) for Lynch syndrome** *Kevin Monahan (United Kingdom)*
10:36-10:48		**Is less really enough? Extent of colorectal cancer resection in Lynch syndrome** *Robert Hüneburg (Germany)*

Scientific Programme

Day 2 – Saturday, September 28th

10:48-11:00	**Unmasking hidden genetic risks by comprehensive genetic testing of cancer patients** *Henriett Butz (Hungary)*
11:00-11:12	**Mutational spectrum of Lynch syndrome in the Mid-South of Israel** *Rakefet Chen-Shtoyerman (Israel)*
11:12-11:24	**ConEpiMut-DB: an international database to improve constitutional MLH1 epimutation diagnosis and research** *Paula Climent-Cantó (Spain)*
11:24-11:36	**The Family Gene Kit: A tool to help families communicate and understand hereditary cancer** *Caroline Leek (United Kingdom)*
11:36-11:48	**Introduction of a Mismatch Repair Deficiency Investigative Pathway to standardise the extent of testing in a tertiary cancer referral centre** *Rachel Clarke (Ireland)*

10:00-12:00	WG 4	**Extra-colonic cancers in hereditary tumour syndroms**
	Chairs:	*Christina Therkildsen (Denmark)*

10:00-10:12	**An audit of current endometrial cancer mismatch repair testing and onward genetic referral in the United Kingdom and Republic of Ireland between March 2022 to March 2023.** *Neil Ryan (United Kingdom)*
10:12-10:24	**Evaluation of upper gastrointestinal tract surveillance in individuals with Lynch syndrome (EARLY) – an international, multicenter registry** *Katrin van Beekum (Germany)*
10:24-10:36	**Germline pathogenic variants in patients with pancreatic cancers according to family and personal history of cancer** *Elena Vida (Spain)*

Scientific Programme

Day 2 – Saturday, September 28th

10:36-10:48	**Mutations in the ATRIP gene predispose to breast cancer** *Cezary Cybulski (Poland)*
10:48-11:00	**A new gangster in town – A POT1 Founder Variant Associated with Early Onset Recurrent Melanoma, Various Solid Malignancies and High Tumor Burden** *Yael Goldberg (Israel)*
11:00-11:12	**SELINA – all-cause mortality and cancer risk after optimization of selenium and arsenic blood levels with inherited predispositions to breast cancer** *Jan Lubinski (Poland)*
11:12-11:24	**Blood lead, molybdenum, zinc and iodine levels as a marker of cancers risk in BRCA1 carriers.** *Milena Matuszczak (Poland)*
11:24-11:36	**MRI Surveillance and Breast Cancer Mortality in Women With BRCA1 and BRCA2 Sequence Variations** *Jacek Gronwald (Poland)*
12:00- 12:30	Lunch break
12:30-13:15	**My most challenging/interesting case** *Chairs: Giulia Martina Cavestro (Italy)* *Laura Valle (Spain)*
12:30-12:45	**Case 1** **Catching the missing heritability in Lynch syndrome: the role of whole genome sequencing** *Vince Kornél Grolmusz (Hungary)*
12:45-13:00	**Case 2** **Early-onset hyperplastic polyposis and colon cancer with unexplained cause and clonal spread** *Esther Schamschula (Austria)*
13:00-13:15	**Case 3** **Metastatic Colorectal Cancer in Pregnancy: A Case Report** *Florian Scholz (Germany)*

Scientific Programme

Day 2 – Saturday, September 28th

13:15-14:15	**Prevention Update** *Chairs: Elke Holinski-Feder (Germany)* *Finlay Macrae (Australia)*
13:15-13:35	**The MESACapp Study – an update on chemo-prevention for colorectal cancer with mesalamine** *Ann-Sofie Backman (Sweden)*
13:35-13:55	**Risks and benefit of aspirin in prevention of hereditary colorectal cancer** *Andrew T. Chan (USA)*
13:55-14:15	**Immune prevention in Lynch syndrome** *Matthias Kloor (Germany)*
14:15-14:45	**Keynote lecture II** *Chair: John Burn (United Kingdom)*
	The promise of liquid biopsy to reduce the burden of cancer *Harpal Kumar (United Kingdom)*
14:45-15:00	**Coffee break**
15:00-16:30	**EHTG-N Nursing Group** *Chairs: Laura Monje-Garcia (United Kingdom)* *Mechelle Loughrey (Ireland)* *Celia Diez de los Rios (Spain)*
15:00-15:15	**A global collaboration to integrate genomics into nursing practice: The Global Genomics Nursing Alliance (G2NA)** *Kathleen Calzone (USA)*

Scientific Programme

Day 2 – Saturday, September 28th

15:15-15:30	**Innovating Research to Demonstrate Nursing Sensitive Outcome Indicators** *Mary Ryder, Ireland*
15:30-15:40	**Cancer nurses' role in risk reduction strategies with people with hereditary cancer syndromes: the importance of addressing health beliefs and lifestyle behaviors** *Celia Diez de los Rios (Spain)*
15:40-15:50	**From diagnosis of colorectal cancer to diagnosis of Lynch syndrome at a tertiary cancer referral center – an Advanced Nurse Practitioner led mainstreaming model** *Mechelle Loughrey (Ireland)*
15:50-16:00	**"It's a lifetime of worry" – making sense of uncertainty in Lynch syndrome: an interpretative phenomenological analysis** *Zanna Karsan (United Kingdom)*
16:00-16:10	**Lynch syndrome surveillance: outcomes from a new regional tertiary center hub** *Helen Francis (United Kingdom)*
16:10-16:20	**Nurse education to support Lynch syndrome genetic testing aligned with a national competency framework** *Karen Westaway (United Kingdom)*
16:20:16:30	**Advancing inherited colorectal cancer research and services through comprehensive patient and public involvement: a real-life application from the PSYLIVED project** *Laura Monje-Garcia (United Kingdom)*
16:30	**Meeting summary and closing remarks** *Gabriela Möslein (Germany)* *Toni Seppälä (Finland)* **Invitation to 9th EHTG Meeting Heidelberg, 19.-21.09.2025** *Aysel Ahadova (Germany)*

Faculty

Abu-Freha, Naim p 10	Soroka University Medical Center, Beer Sheva, Israel
Ahadova, Aysel p 12, 17	Heidelberg University Hospital, Heidelberg, Germany
Akdeniz, Bayram p 12	Center for Bioinformatics, Department of Informatics, University of Oslo, Oslo, Norway
Anderson, Richard p 6	University of Edinburgh, Edinburgh, United Kingdom
Aretz, Stefan p 11	Bonn University Hospital, Bonn, Germany
Backman, Ann-Sofie p 16	Karolinska Institute, Stockholm, Sweden
Bajalica-Lagercrantz, Svetlana p 10	Karolinska Institutet, Dpt. Of Oncololgy - Pathology Stockholm, Sweden
Bajwa-ten Broeke, Sanne p 10	University Medical Center Groningen, Groningen, The Netherlands
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Bohaumilitzky, Lena p 12	Heidelberg University Hospital, Department of Applied Tumor Biology Heidelberg, Germany
Buchanan, Dan p 11, 13	University of Melbourne, Melbourne, Australia
Burn, Sir John p 6, 16	Newscastle University, Newcastle upon Tyne, United Kingdom
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Calzone, Kathleen p 16	National Cancer Institute Bethesda, MD, USA

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Dardenne, Antoine p 12	Cochin. AP-HP, Service de gastroentérologie et d'oncologie digestive Paris, France
Diez de los Rios, Celia p 16, 17	Barcelona University, Faculty of Nursing, Bellvitge Campus Barcelona, Spain
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Faculty

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Möslein, Gabriela p 17	Bethesda University Hospital Duisburg, Germany
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Olsen, BJ p 7	Royal Victoria Hospital Belfast health and social care Trust Belfast, Northern Ireland
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Rosén, Anna p 7	Umeå University, Department of Diagnostics and Intervention, Oncology Umeå, Sweden

Faculty

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Ryder, Mary p 17	University College Dublin Dublin, Ireland
Sampson, Julian p 8, 10, 12	Cardiff University, Cardiff, United Kingdom
Schamschula, Esther p 15	Medical University of Innsbruck Department of Human Genetics Innsbruck, Austria
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Terradas, Mariona p 13	Catalan Institute of Oncology IDIBELL Hereditary Cancer Programme L'Hospitalet de Llobregat, Spain
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Thomas, Laura p 11	Cardiff University School of Medicine Cardiff, United Kingdom

Faculty

Urzua Traslavina, Carlos p.10	University Medical Center Groningen Groningen, The Netherlands
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Winship, Ingrid p 6	University of Melbourne Melbourne, Australia
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9TH ANNUAL MEETING
Heidelberg • 2025

SAVE THE DATE

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

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

Heidelberg, Germany

We would like to announce the 2025 Meeting which will take place in Heidelberg, Germany. Details to follow soon on our website: www.ehtg.org

10th Meeting of the European Hereditary Tumour Group (EHTG)

September 2026

To be announced

11th Meeting 2027:

Call for venue application 2027 will be announced early 2025.