



Preliminary PROGRAM 6th Meeting 2022

Thursday, Sept.29 to Saturday, Oct.1,
in person (+ live streaming) at the Hotel Victoria Gran Melia, Palma de Mallorca,
and virtual event online

THU, Sept.29, 2022

08:00-10:00 a.m. REGISTRATION on siteplace

10:00-11:00 a.m.

The 12 best visual posters (3* + 2' each)

Chairs: Ingrid Winship (Australia), Saskia Haupt(Germany)

- 1) HLA-specific immunogenicity of Lynch Syndrome associated frameshift peptide neoantigens – towards next-generation cancer vaccines
Hernandez Sanchez, Alejandro (Germany)
- 2) APC mosaicism is a relevant explanation in (mild) polyposis phenotypes
Nielsen, Maartje (Netherlands)
- 3) Clinical characteristics of pancreatic and biliary tract cancers in Lynch syndrome: a retrospective analysis from the Finnish national Lynch syndrome research registry
Zalevskaja, Kristina (Finland)
- 4) Combined somatic copy number alteration and fragmentation analysis for treatment monitoring in colorectal cancer patients using liquid biopsy
Suárez Sánchez, Aida (Spain)
- 5) Molecular carcinogenesis pathway of MLH1-associated Lynch syndrome colorectal cancer unraveled: “two-in-one hit” model
Ahadova, Aysel (Germany)
- 6) The prevalence of mismatch repair deficiency in ovarian cancer: systematic-review and metaanalysis
Ryan, Neil (UK)
- 7) Detection of mismatch repair deficiency in colonoscopic biopsies and urine using a simple PCR multiplex with potential for postal urinary tumour surveillance in Lynch syndrome patients
Jackson, Michael (UK)
- 8) Genetic testing in early onset col-rectal cancer: experience of an oncogenetic consultation in the era of NGS sequencing
Farely, Solenne (France)
- 9) Lynch Syndrome is associated with fecal and salivary dysbiosis
Mannucci, Alessandro (Italy)
- 10) Why the combination of modeling and machine learning could be the future direction in mathematical oncology
Haupt, Saskia (Germany)
- 11) The Person-Based Approach to optimising a personalised, interactive patient decision aid for people with Lynch syndrome
Kohut, Kelly (UK)
- 12) Mesalamine for Colorectal Cancer Prevention Program in Lynch Syndrome (MesaCAPP)
Backman, Ann-Sofie (Sweden)

11:00-11:30 a.m.

COFFEE BREAK



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11:30 a.m.-01:30 p.m.

Plenary Session 1

Chairs: Annika Auranen (Finland), Matt Yurgelun (USA), Michael Scharl (Switzerland)

1. Protecting fertility in patients with hereditary cancer syndromes
Somigliana, Edgardo (Italy)
2. What do the patients value in risk management – a patient association perspective
Nicola Reents (Germany)
3. What do the patients value in risk management – a cancer family perspective
Dubin, Dave (USA)
4. Cancer vaccines
Vilar, Eduardo (USA)
5. Immune surveillance and immune prevention in Lynch syndrome – new evidence for the feasibility of cancer-preventive vaccines
Kloor, Matthias (Germany)
6. Clinical description and first estimates of age-associated cancer risk and survival in the context of constitutional mismatch repair system deficiency (CMMRD syndrome) : report from the European database of the European consortium C4CMMRD
Colas, Chrystelle (France)

01:30-02:30 p.m. Industry Symposium (PAICON) / LUNCH BOX

02:30-03:30 p.m.

Big Debate 1

Chairs: Dan Buchanan (Australia)

Systematic tumor testing for genomic variants should be implemented (incl. costs-efficiency and capacity, RET pathogenic variants)

Speakers: Judith Balmana (Spain), Sigurdís Haraldsdóttir (Iceland)

Discussion

03:30-04:30 p.m.

Big Debate 2

Chair: Zohar Levi (Israel)

The term of oligo polyposis serves no purpose

Speakers: Elisabeth Half (Israel), Maartje Nielsen (Netherlands)

Discussion

04:30-05:00 p.m.

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05:00-06:00 p.m.

Evolving Technologies: applications of AI

Chairs: Ann-Sophie Backman (Sweden), Magnus von Knebel-Doeberitz (Germany)

1. AI-assisted Pathology
TBD
2. AI-assisted Endoscopy
Helmut Messmann (Germany)
3. AI assisted diagnosis
TBD

06:00-07:00 p.m. WELCOME RECEPTION on the MELIA Victoria hotel terrace

END OF DAY ONE

FRI, Sept.30, 2022

08:00-09:00 a.m.: PLSD Meeting

1. Møller, Pål: Lynch Syndrome: Which cancer comes first?
2. Møller, Pål: The four Lynch syndromes
3. Møller, Pål: An additional carcinogenetic mechanism for colon cancer in Lynch syndrome

OPEN PLSD Business Meeting

09:00-11:00 a.m.

Parallel Working Groups

WG 4: Genetics & Counselling (F2F)

Chairs: Rolf Sijmons (Netherlands) / Heather Hampel (USA)

Speaker: TBD and Topic

- 1) A national picture of molecular testing for Lynch syndrome in England highlights gaps in testing and opportunities for improved diagnosis
McRonald, Fiona (UK)
- 2) A NURSE LED GENETIC CLINIC FOR LYNCH SYNDROME TESTING IN COLORECTAL CANCER 1 YEAR LATER
Langford, Lora (UK), Mauro Proserpio (UK)
- 3) APC-specific ACMG/AMP variant classification guideline alleviates the burden of variants of uncertain significance in ClinVar and locus-specific databases
Aretz, Stefan (Germany)
- 4) Targeted short-read RNA sequencing as supplement to DNA germline testing to increase diagnostic yield
Romic-Pickl, Julia (Germany)



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- 5) Existing with a cancer ghost in the back of my mind- Written narratives of living with Lynch syndrome
Eriksen, Elin O. (Norway)
- 6) Mutational and transcriptomic landscape and somatic evolution in Li-Fraumeni Syndrome-associated tumorigenesis
Torra I Benach Maria (UK)
- 7) Psychological well-being of people living with a colorectal cancer predisposition syndrome: evidence from a systematic review
Monje-Garci, Laura (UK)

WG 1: Carcinogenic mechanism and pathways (virtual)

Chairs: Julian Sampson (UK), Sanne ten Broeke (Netherlands)

Speaker: Richarda de Voer (Netherlands) (talk about **MBD4/BER**, exact title follows)

- 1) Clinicopathological Features of Deficient Mismatch Repair (dMMR) Protein Expression Patterns in Colorectal Cancer in a Spanish Cohort
Suárez Sánchez, Aida (Spain)
- 2) Constitutional MLH1 methylation, frequency and mode of identification
Dardenne, Antoine (France)
- 3) Genome-wide methylome of Lynch syndrome and Familial adenomatous polyposis-associated colorectal tumors
Mäki-Nevala, Satu (Finland)
- 4) Germline MBD4 mutations in cancer predisposition
Valle Velasco, Laura (Spain)
- 5) HLA-specific immunogenicity of Lynch Syndrome associated frameshift peptide neoantigens – towards next-generation cancer vaccines
Hernandez Sanchez, Alejandro (Germany)
- 6) Molecular carcinogenesis pathway of MLH1-associated Lynch syndrome colorectal cancer unraveled: “two-in-one hit” model
Ahadova, Aysel (Germany)
- 7) APC mosaicism is a relevant explanation in (mild) polyposis phenotypes
Nielsen, Maartje (Netherlands)

WG 3: Epidemiology of hereditary cancer (virtual)

Chairs: Mark Jenkins (Australia) / Christoph Engel (Germany)

Speaker: Karl Heinimann (Switzerland): Prevalence of pathogenic variants in hereditary cancer by whole exome / genome analysis

- 1) Cancer mortality by organ, gene and gender in carriers of pathogenic mismatch repair gene variants receiving surveillance for early diagnosis and treatment: A report from the Prospective Lynch Syndrome Database
Dominguez Valentin, Mev (Norway)
- 2) Central Amalgamation of a nationally complete registry of Lynch Syndrome carriers in England
Huntley, Catherine (UK)
- 3) Germline Mismatch Repair (MMR) gene analyses from English NHS regional molecular genomics laboratories 1996-2020: development of a national resource of patient-level genomics laboratory records
Turnbull, Clare (UK)



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- 4) Implementation of population-wide effective CRC screening for Lynch syndrome using the MSI-Plus Assay
Herrero Belmonte, Patricia (UK)
- 5) The English National Lynch syndrome transformation project: An NHS Genomic Medicine Service Programme
Monahan, Kevin (UK)
- 6) Universal Testing of Asturian Population Diagnosed with Lynch Syndrome
Díaz Vico, Tamara (Spain)

11:00-11:30 a.m. COFFEE BREAK

11:30 a.m.-01:30 p.m.

Parallel Working Groups

WG 9: Systemic biomarkers (F2F)

Chairs: Mev Dominguez-Valentin (Norway) / Aysel Ahadova (Germany)

Speaker: TBD and Topic

- 1) A Demonstration of Microsatellite Instability Analysis with Circulating Tumor DNA in Endometrial Cancer
Lewis, Samantha (USA)
- 2) Analysis of plasma cell-free DNA genome-wide methylation in colorectal cancer and adenoma patients - towards a cell-free DNA-based surveillance
Eikenboom, Ellis (Netherlands)
- 3) Associations of circulating microRNAs with body mass index, waist circumference, and physical activity in Lynch syndrome
Sievänen, Tero (Finland)
- 4) Combined somatic copy number alteration and fragmentation analysis for treatment monitoring in colorectal cancer patients using liquid biopsy
Hallermayr, Ariane (Germany)
- 5) Highly sensitive Liquid Biopsy Duplex Sequencing complements tissue biopsy to enhance detection of clinically relevant genetic variants
Romic-Pickl, Julia (Germany)
- 6) Whole-exome sequencing of cell-free DNA reveals mutational signatures associated with Lynch syndrome
Kauma, Liro (Finland)

WG 7: Screening for gynecology, urology and beyond (virtual)

Chairs: Neil Ryan (UK) / Denise R. Nebgen (USA)

Speaker: TBD

- 1) Hypercalcemic Ovarian carcinoma and mutation of SMARCA4: we need a prematurely testing
Perez-Lopez, Maria-Eva (Spain)
- 2) The prevalence of mismatch repair deficiency in ovarian cancer: systematic-review and meta-analysis
Ryan, Neil (UK)



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- 3) The genomic and clinico-pathological characteristics of sebaceous skin lesions from people with Lynch syndrome
Buchanan, Dan (Australia)
- 4) Detection of mismatch repair deficiency in colonoscopic biopsies and urine using a simple PCR multiplex with potential for postal urinary tumour surveillance in Lynch syndrome patients
Jackson, Michael (UK)
- 5) Distinguishing the molecular profile of endometrial cancer by spectroscopy: A Diagnostic Cross-Sectional Study
Ryan, Neil (UK)
- 6) NON-BRCA MUTATION PREVALENCE IN HEREDITARY BREAST AND OVARIAN CANCER SYNDROME IN OUR CENTRE
Jimenez Ruiz, Francisco Javier (Spain)

WG 2: Early onset cancers (virtual)

Chairs: José Perea (Spain) / TBD

Speaker: Giulia Martina Cavestro, **DIRECT**: Delphi initiative for EoCRC. CGA-IGC, EHTG, AIFET
Guidelines for the Management of Early-Onset Colorectal Cancer

- 1) Genetic testing in early onset col-rectal cancer: experience of an oncogenetic consultation in the era of NGS sequencing
Farely, Solenne (France)
- 2) Young-onset gastric cancers are often early gastric cancers
Puzzono, Marta (Italy)
- 3) Comprehensive characterization of hereditary cancer cases from Peru: Bringing precision medicine to a low-resource setting
Dominguez Valentin, Mev (Norway)
- 4) Germline pathogenic variants detected in microsatellite-unstable cancers: a two-year prospective experience from a Hungarian center
Grolmusz, Vince Kornél (Hungary)

01:30-02:30 p.m. LUNCH

02:30-04:30 p.m.

Parallel Working Groups

WG 8: Surgery and Gastroenterology (F2F)

Chairs: Gabriela Moeslein (Germany) / Peter Bauerfeind (Switzerland)

Speaker: Alessandro Manucci, "Duodenal Polyposis in FAP and revision of the Spigelman Classification"

- 1) Is there a correlation between genotype and duodenal phenotype in Familial Adenomatous Polyposis?
Dhooge, Marion (Italy)
- 2) Duodenal disease in 579 individuals with MUTYH-associated polyposis: updated findings from an international prospective observational study
Truscott, Becky (UK)



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- 3) Small bowel cancer in patients with Familial Adenomatous Polyposis – report of two clinical cases and review of literature
Colletti, Gaia (Italy)
- 4) Ileoanal pouch cancers in Ulcerative Colitis and Familial Adenomatous Polyposis: A systematic review and meta-analysis
Pellino, Gianluca (Italy)
- 5) Metachronous CRC after surgery in Lynch Syndrome
Seppälä, Toni (Finland)
- 6) Functional Outcome Differences between Males and Females Who have Undergone Reconstructive Surgery with Ileorectal Anastomoses (IRA) or Ileal Pouch-Anal Anastomoses (IPAA) due to Familial Adenomatosis Polyposis - A Prospective Cohort Study
Mira, Daniel (Sweden)
- 7) Real-time use of artificial intelligence (CADEYE) in colorectal cancer surveillance of patients with Lynch syndrome – a randomized controlled pilot trial (CADLY)
Hüneburg, Robert (Germany)

WG 5: Novel associations (virtual)

Chairs: TBD/ TBD

Speaker: TBD

- 1) High Prevalence of MUTYH Associated Polyposis Among Minority Populations in Israel, due to Rare Founder Pathogenic Variants
Reznick Levi, Gili (Israel)
- 2) Lynch Syndrome is associated with fecal and salivary dysbiosis
Mannucci, Alessandro (Italy)
- 3) Oligodontia-colorectal cancer syndrome with cleft palate as a possible new feature in a cohort of 13 AXIN2 variant carriers
Roht, Laura (Estonia)
- 4) Risk of Second Primary Thyroid Cancer in Women with Breast Cancer
Lubinski, Jan (Poland)
- 5) Why the combination of modeling and machine learning could be the future direction in mathematical oncology
Haupt, Saskia (Germany)

WG 6: Risk and Clinical Management (virtual)

Chairs: Lubinski, Jan (Poland) / Helmut Messmann (Germany)

Speaker: TBD

- 1) The Person-Based Approach to optimising a personalised, interactive patient decision aid for people with Lynch syndrome
Kohut, Kelly (UK)
- 2) Uncertainty quantification in oncology - How do uncertain data influence model results and clinical decision making?
Zeilmann, Alexander (Germany)
- 3) From Diagnosis of Colorectal cancer to Diagnosis of Lynch Syndrome: The RM Partners Quality Improvement Project
Monje-Garcia, Laura (UK) / Ana Koziel (UK)



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- 4) Improvement of IAFLD under TEDUGLUTIDE in a patient with familial adenomatous polyposis (FAP) and short bowel syndrome
Bauerfeind, Peter (Switzerland)
- 5) Mesalamine for Colorectal Cancer Prevention Program in Lynch Syndrome (MesaCAPP)
Backman, Ann-Sofie (Sweden)
- 6) Clinical characteristics of pancreatic and biliary tract cancers in Lynch syndrome: a retrospective analysis from the Finnish national Lynch syndrome research registry
Zalevskaia, Kristina (Finland)

04:30- end of congress day for all non-participants of FAP Guidelines

04:30-06:30 p.m.

FAP Guidelines (by invitation only) **F2F and virtual**

07:30 p.m. – OPEN END: CONFERENCE DINNER

END OF DAY 2

SAT, Oct.1, 2022

09:00-11:00 a.m.

WG Reporting Back Session and Collaborative Studies

Chairs: Rolf Sijmons (Netherlands), Heather Hampel (USA)

11:00-11:30 a.m. COFFEE BREAK

11:30 a.m.-12:45 p.m.

Hot Topics

Chairs: Stefan Aretz (Germany), Brandie Leach (USA)

- 1) Challenges of organizing European patient cancer registries for research (online)
Speaker: Hoogerbrugge, Noline (Netherlands) (on behalf of ERN GENTURIS)
- 2) **ESCP/EHTG – Joint symposium; Topic: PD-1 blockade in mismatch repair-deficient cancer**
Introduction: Triantafillos Doulias (USA)
Speakers:
ESCP/EHTG – Joint symposium,
Shiu, Kai-Keen (UK)

Plenary discussion moderated by chairs

Discussant:

Burn, John (UK)



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12:45-01:30 p.m.

Networking with patients and their organisations

Chair & Organizer: **Nicola Reents**, 1-2 patient rep's (TBD)

Question: How would you change the health system? (3'+2')

1. Semi-Colon patient advocacy group (Germany)
2. Mutagens (Italy)
3. AFALynch (Spain)
4. Lynch Syndrome (UK)
5. Lynch syndrome (Ireland)
6. Colores (Finland)
7. Lynch+Polyposis syndromes (Netherlands)

01:30-02:30 p.m. EHTG Business Meeting & LUNCH BOX

02:30-03:15 p.m.

Big Debate 3

Chairs: Gabriel Capella (Spain)

Aspirin prevents more LS colorectal cancers than endoscopy

Speakers: Burn, John (UK), Bu Hayee (GB)

Discussion

03:15-03:45 p.m.

Best of abstracts

Chair: Matthias Kloor (Germany)

- 1) Assessment of the ability of the polygenic background to refine colorectal cancer risk in Lynch syndrome
Dueñas Cid, Nuria (Spain)
- 2) Mutation rate evolution drives immune escape in mismatch repair-deficient cancer
Jansen, Marnix (UK)

03:55-04:30 p.m.

Big Debate 4

Chairs: TBD

Molecular testing can direct management of colorectal cancer

Speakers: Pellino, Gianluca (Italy), Seppälä, Toni (Finland)

Discussion

04:30-05:00 p.m. COFFEE BREAK

04:30 p.m. Farewell
Gabriela Moeslein



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General remark:

The meeting will be a plenary F2F meeting (with live-streaming) in the conference rooms of the beautiful Hotel Gran Melia Victoria in Palma de Mallorca. On Thursday, Sept. 29, there will be a plenary sessions day, same as on Saturday, October 1. On Friday, we have planned the Working Group day with a total of 9 (3x3) parallel working groups, of which a total of three successive working groups will be F2F in the conference meeting room. 2 parallel working groups throughout the day (3 x 2) will be a virtual meeting, of course with live interaction.