Final Program
EHTG 5th Meeting
Friday 8 - Saturday 9 October 2021

Virtual Meeting
Dear members, colleagues, global participants and friends,
as chair of the EHTG and in the name of our board, it is
a great pleasure to invite you to our upcoming annual
meeting 2021, to be held on
October 8 and 9, 2021.
The uncertainty of the pan-
demic is still present world-
wide and therefore we have
planned our meeting as
a virtual event – and hope to be able to meet you again in
Mallorca in 2022.
The program committee and the entire board have spent
a lot of thought in planning the virtual format – especially
difficult for a group such as our which relies so heavily on interaction. The Working Group format has always been an asset of our society and keeping this identity in a virtual format has been challenging. Together with our fantastic new secretariat and the technical support by SynopticCon, we will have parallel working groups, maintaining the specialized expertise on one hand and sharing the most relevant aspects of each subgroup in combined plenary.
Although we will miss our always exciting social program this year, the virtual format does allow for a global attendance and this in itself poses an opportunity for such a specialized medical field as hereditary predisposition to cancer. We are delighted to have received so many excellent abstracts on hot topics and therefore have maintained the short abstract format – certainly a challenge – but one that allows for an exciting meeting. Thank you to all that are supporting this meeting and EHTG. After a difficult year 2020 we are absolutely enthusiastic to be back in 2021!
We sincerely hope that you will join us and look forward to future collaborations and networking
Gabriela Möslein
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Organisation

Board Members: Aysel Ahadova, Germany
John Burn, Great Britain
Mev Dominguez, Norway
Gabriela Moeslein, Germany
Pål Møller, Norway
Sanne ten Broeke, Netherlands
Julian Sampson, Great Britain
Toni Seppälä, Finland
Rolf Sijmons, Netherlands

Program Committee: Chair:
Mev Dominguez-Valentin
Toni Seppälä
Aysel Ahadova

Y-EHTG (Young EHTG): Chair:
Sanne ten Broeke

Secretariat: Gabriele Sponholz
Gs007@ehtg.org
+49-160-8459502

Location: only virtual in 2021
Registration: www.ehtg.org
Registration fee: options: see website
CME points: the EHTG congress has been granted 9 CME credits by the EACCME / UEMS
8.10.21: 5 credits
9.10.21: 4 credits
The online participation per day will be measured by the IT system and the certificates will be sent to each participant by mail.

Abstract submission: time for submission over
Important notice: the time information refers to CET – Central European Time

EHTG has received an educational grant from Promega Corporation
Program Friday, October 8, 2021

09:00-09:30  EHTG Business Meeting & Board elections

09:30-10:20  Proposals for Collaborative Studies & Guidelines
Chairs: Gabriela Moeslein, Germany / John Burn, United Kingdom

09:30-09:45  1a Survival inherited cancer (SIC)
Toni Seppälä, Finland

09:45-10:00  1 Evaluation of upper gastrointestinal tract surveillance in individuals with Lynch syndrome. An international, multicenter registry. EARLY-Study
Jacob Nattermann, Germany

10:00-10:20  2 Prenatal / pre-gestational diagnosis of adult onset conditions, in particular cancer predisposition syndromes
Yael Goldberg, Israel
Interactive with online voting!

10:20-12:00  State of the Art 1-5
Chairs: Julian Sampson, United Kingdom / Gareth Evans, United Kingdom

10:20-10:40  3 PLSD Update
Pål Møller, Norway

10:40-11:00  4 Preventive management of gynecological LS-associated tumors:
Annika Auranen, Finland

11:00-11:20  5 Immune checkpoint blockade in dMMR cancer (GI)
Georg Martin Haag, Germany

11:20-11:40  6 FIT for Lynch: A national COVID19 pandemic response service'
Kevin Monahan, United Kingdom

11:40-12:00  6a Splicing analyses for variants in MMR genes: best practice recommendations from the European Mismatch Repair Working Group
Marta Pineda Riu, Spain

12:00-12:30  Lunch break

12:30-13:50  Selected Abstracts
Chairs: Bryson Katona, USA / Maurizio Genuardi, Italy

12:40-12:50  7 Blood arsenic levels as a marker of breast cancer risk among BRCA1 carriers
Jan Lubinski, Poland

12:50-13:00  8 Defective DNA repair in polyposis susceptibility
Alisa Olkinuora, Finland

13:00-13:10  9 Development of an interactive, tailored decision aid for people with Lynch Syndrome to supplement genetic counselling
Kelly Kohut, United Kingdom

13:10-13:20  10 Precision medicine, germline genetic testing and pancreatic cancer: successful implementation of a mainstreaming pathway in French patients
Thomas Pudlatz, France

13:20-13:30  11 Delineating genotype and parent of origin effect on the phenotype in MSH6 associated Lynch syndrome
Anne-Sophie van der Werf- t Lam, The Netherlands

13:30-13:40  12 First estimations of diffuse gastric cancer risks in CTNNA1 pathogenic variants carriers
Marie Coudert, France

Samantha Lewis, USA

13:50-14:00  Podium Discussion
Explanations for limited colonoscopy effect on CRC prevention in Lynch syndrome”

13:50-14:00  13X Introduction: Julian Sampson, United Kingdom
Experts:

14:00-14:05  13A Toni Seppälä, Finland /
14:05-14:10  13B Pal Møller, Norway /
14:10-14:15  13C Zohar Levi, Israel /
14:15-14:20  13D Iris Nagtegaal, Netherlands /
14:20-14:25  13E Aysel Ahadova, Germany
14:20-14:50  Discussion
**Program Friday, October 8, 2021**

**14:50-15:30** | **Coffee break**
---|---
**15:30-17:20** | **WG 1**
| **Carcinogenic mechanisms and pathways**
| Chairs: Paivi Peltomaki, Finland / Maartje Nielsen, Netherlands
**15:30-16:00** | **14** Evolution of colon cancer and cancer phylogenetics
| Kamila Naxerova, USA
**16:00-16:10** | **15** Simultaneous inactivation of MLH1 and stabilization of beta-catenin in Lynch syndrome colorectal cancer: “one hit less” hypothesis
| Aysel Ahadova, Germany
**16:10-16:20** | **16** Bacteroides Fragilis toxin is associated with colonic right-sided precursor lesions in the follow-up of Lynch Syndrome patients
| Carlijn Bruggeling, Nederland
**16:20-16:30** | **17** A tumour-focused approach can successfully resolve a diagnosis of suspected Lynch syndrome
| Daniel Buchanan, Australia
**16:30-16:40** | **18** Mathematically modeling Lynch syndrome colorectal carcinogenesis at different scales
| Saskia Haupt, Germany
**16:40-16:50** | **19** The Molecular Profile of MSH6-Associated Colorectal Carcinomas From Patients With Lynch Syndrome
| Noah Helderman, Netherlands
**16:50-17:00** | **20** Blood Cadmium Level and the Risk of Cancer in Women with BRCA1 Mutations
| Jan Lubinski, Poland
**17:00-17:10** | **21** Highly-sensitive approach for characterizing microsatellite instability in normal tissue and tumors from biallelic germline mismatch repair mutation carriers
| Fátima Marín, Spain
**17:10-17:20** | **22** Systemic circulating microRNA profiles in Lynch syndrome
| Tero Sievänen, Finland

**15:30-17:20** | **WG 2**
| **Surgical management of hereditary tumor syndromes**
| Chairs: Rodrigo Guindalini, Brazil / Matt F. Kalady, USA
**15:30-15:50** | **23** Management of Hereditary Pancreatic Cancer: What do we actually know – an update
| Giulia Martina Cavestro, Italy
**15:50-16:10** | **24** Management of adrenal incidentalomas in hereditary syndromes
| Peter Goretzki / Martina Mogl, Germany
**16:10-16:30** | **25** Obesity and FAP requiring proctocolectomy in the elective situation – surgical considerations
| Antonio de Lacy, Spain
**16:30-16:40** | **26** Superior Rectal Artery sparing in total colectomies with ileorectal anastomosis to reduce anastomotic leakage in patients with familial polyposis.
| Gaia Colletti, Italy
**16:40-16:50** | **27** Long-term neoplastic outcomes of pouch surveillance in familial adenomatous polyposis (FAP): time to reduce surveillance intensity?
| Roshani Patel, United Kingdom
**16:50-17:00** | **28** Surgical desmoid management: special cases
| Gloria Zaffaroni, Italy/Germany
**17:00-17:30** | **Surgical Case Discussion & Voting**
| Interactive with online voting!
**17:30** | **End of meeting day 1**
| enjoy your evening and see you tomorrow!
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<tr>
<th>Time</th>
<th>Session</th>
<th>Presentation</th>
<th>Speaker</th>
<th>Location</th>
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<tr>
<td>15:30-17:30</td>
<td>WG 3</td>
<td><strong>Polyposis syndromes</strong></td>
<td>Chairs: Daniel Buchanan, Australia / Anna Lepistö, Finland</td>
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<tr>
<td>15:30-16:00</td>
<td>29</td>
<td>Duodenal Polyposis: a significant complication in familial polyposis syndromes</td>
<td>Laura Thomas, United Kingdom</td>
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<tr>
<td>16:00-16:10</td>
<td>30</td>
<td>Diagnostic Yield of Constitutional Genetic Testing in Patients with Multiple Colorectal Adenomas (MCRA)</td>
<td>Sau Mak, United Kingdom</td>
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<td>16:10-16:20</td>
<td>31</td>
<td>Germline chromothripsis of the APC locus in a patient with adenomatous polyposis</td>
<td>Florentine Scharf, Germany</td>
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<tr>
<td>16:20-16:30</td>
<td>32</td>
<td>An international study of duodenal disease in MAP: incidence of polyposis, cancer, and next steps</td>
<td>Becky Truscott, United Kingdom</td>
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<tr>
<td>16:30-16:40</td>
<td>33</td>
<td>Investigating the molecular mechanisms of adenomatous polyposis syndromes using 3D organoid models</td>
<td>Angharad Walters, United Kingdom</td>
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<tr>
<td>16:40-16:45</td>
<td>34</td>
<td>The sequential and dynamic changes in genome-wide DNA methylation and microbiome portray the neoplastic and tumourigenic transformations of colonic tissue in people with serrated polyposis syndrome</td>
<td>Eric Joo, Australia</td>
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<tr>
<td>16:45-16:50</td>
<td>35</td>
<td>Differences between inherited and acquired polymerase proofreading deficiencies in cancer</td>
<td>Mur Pilar, Spain</td>
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<td>16:50-16:55</td>
<td>36</td>
<td>Paired somatic-germline testing of 15 polyposis and colorectal cancer-predisposing genes highlights the role of APC mosaicism in de novo familial adenomatous polyposis</td>
<td>Paula Rofes, Spain</td>
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<tr>
<td>16:55-17:00</td>
<td>37</td>
<td>3D patient-derived intestinal organoid models for familial polyposis</td>
<td>Sara Seifan, United Kingdom</td>
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<tr>
<td>17:00-17:05</td>
<td>38</td>
<td>Phenotypic analysis of 106 serrated polyposis patients</td>
<td>Verena Steinke-Lange, Germany</td>
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<tr>
<td>17:05-17:10</td>
<td>39</td>
<td>APC mosaicism testing in milder polyposis phenotypes reveals pks+ E.coli bacteria as a possible additional explanation for the development of colorectal adenomas</td>
<td>Diantha Terlouw, The Netherlands</td>
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<tr>
<td>17:30</td>
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<td><strong>End of meeting day 1</strong></td>
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Enjoy your evening and see you tomorrow!
09:00-10:00  **PLSD Business Meeting**  
Chairs: Pål Møller, Norway  
Toni Seppälä, Finland

09:00-09:15  **39A PLSD business meeting introduction**  
Pål Møller, Norway

09:15-09:25  **Discussion**

09:25-09:35  **39 B The Survival Inherited Cancer (SIC) initiative**  
Toni Seppälä, Finland

09:35-09:45  **39 C Gastric cancer surveillance among LS carriers**  
Lior Katz, Israel

09:45-10:00  **Discussion**

10:00-11:50  **WG 5 Genetics & Counselling**  
Chairs: Demetra Georgiou, United Kingdom / Rolf Sijmons, Netherlands

10:00-10:30  **40 Genetics and genetic counselling - issues for hereditary cancer in 2021**  
Ingrid Winship, Australia

10:30-10:35  **41 The Spectrum of Mutations Predisposing to Familial Breast Cancer in Poland**  
Cezary Cybulski, Poland

10:35-10:40  **42 Mutations in ATM, NBN and BRCA2 predispose to aggressive prostate cancer in Poland**  
Dominika Wokołorczyk, Poland

10:40-10:45  **43 The utility of base-excision repair tumor mutational signatures for identifying biallelic MUTYH carriers and classifying germline variants of uncertain clinical significance using colorectal cancer panel-sequenced genomic data**  
Peter Georgeson, Australia

10:45-10:50  **44 Role of single nucleotide polymorphisms of PDCD1 and CD274 in Lynch syndrome**  
Vince Kornél Grolmusz, Hungary

Wojciech Kluźniak, Poland

10:55-11:00  **46 Germline MBD4 mutations and predisposition to uveal melanoma**  
Marine Le Mentec, France

11:00-11:05  **47 Developing a nurse led genetic clinic for lynch syndrome testing in colorectal cancer**  
Mauro Proserpio, United Kingdom  
Lora Fenton, United Kingdom

11:05-11:10  **48 WNT pathway components in the predisposition to serrated polyposis**  
Isabel Quintana, Spain

11:10-11:15  **49 Inherited Variants in XRCC2 and the Risk of Breast Cancer**  
Cezary Cybulski, Poland

11:15-11:20  **50 Assessment of expression and splicing of cancer-related transcripts by long-read mRNA sequencing**  
Vincent Schwenk, Germany

11:20-11:25  **51 Discordant IHC MMR staining and MSI results in tumors of MSH6 variant carriers**  
Anne-Sophie van der Werf-t Lam, The Netherlands

11:25-11:30  **52 Identification of two Lynch syndrome families harboring inherited MLH1 epimutations**  
Covadonga Vara, España

11:30-11:35  **53 MSH3 as a new predisposing gene for adenomatous polyposis, and beyond**  
Marie-Charlotte Villy, France

11:35-11:40  **54 PALB2 Mutations and Prostate Cancer Risk and Survival**  
Dominika Wokołorczyk, Poland

11:40-11:45  **55 Adherence to mismatch repair testing in early invasive colorectal cancer diagnosed before the age of 70 years**  
Berbel Ykema, Netherlands

11:50-12:20  **Lunch break**
**Program Saturday, October 9, 2021**

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<tr>
<th>Time</th>
<th>Session</th>
<th>Title</th>
<th>Speaker(s)</th>
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<tr>
<td>10:00-11:50</td>
<td>WG 6</td>
<td>Surveillance &amp; Clinical Management</td>
<td>Chairs: Maria Pellise, Spain / Jukka-Pekka Mecklin, Finland</td>
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<tr>
<td>10:00-10:30</td>
<td>56</td>
<td>Using Family History and Other Clinical Factors to Personalize Surveillance and Management in Lynch Syndrome</td>
<td>Matthew Yurgelun, United Kingdom</td>
</tr>
<tr>
<td>10:30-10:40</td>
<td>57</td>
<td>Risk-stratified FIT for urgent colonoscopy in Lynch Syndrome: A clinical service throughout the COVID-19 pandemic</td>
<td>Anne Lincoln, United Kingdom</td>
</tr>
<tr>
<td>10:40-10:50</td>
<td>58</td>
<td>Revisiting the role of immunotherapy for Constitutional Mismatch Repair Deficiency related colorectal cancer treatment</td>
<td>Ellis Eikenboom, Netherlands</td>
</tr>
<tr>
<td>10:50-11:00</td>
<td>59</td>
<td>Age of onset of surveillance colonoscopy for MSH6 mutation carriers</td>
<td>Robert Hüneburg, Germany</td>
</tr>
<tr>
<td>11:00-11:10</td>
<td>60</td>
<td>Should different surveillance options be offered to Lynch syndrome carriers with different mismatch repair gene mutations?: The predicted impact and cost-effectiveness of tailored colonoscopic surveillance strategies in individuals with Lynch syndrome</td>
<td>Yoon-Jung Kang, Australia</td>
</tr>
<tr>
<td>11:10-11:20</td>
<td>61</td>
<td>Gastrointestinal Stromal Tumours: Five Years of Molecular Analysis and Referral to Cancer Genetics</td>
<td>Hazel O’Sullivan, United Kingdom</td>
</tr>
<tr>
<td>11:20-11:30</td>
<td>62</td>
<td>Molecular screening of urine for Mismatch Repair deficient urothelial tumours; an under-appreciated cancer in Lynch syndrome</td>
<td>John Burn, United Kingdom</td>
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**Program Saturday, October 9, 2021**

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<tr>
<td>11:30-11:40</td>
<td>63</td>
<td>Broadening Risk Profile in Familial Colorectal Cancer Type X; increased risk for five cancer types in the national Danish cohort</td>
<td>Christina Therkildsen, Danmark</td>
</tr>
<tr>
<td>11:40-11:45</td>
<td>64</td>
<td>Interval Cancers in Patients with Hereditary Gastrointestinal Syndromes After One Year of the SARS-CoV-2 Pandemic</td>
<td>Giulia Martina Cavestro, Italy</td>
</tr>
<tr>
<td>11:45-11:50</td>
<td>65</td>
<td>Preliminary evaluation of highly sensitive assessment of microsatellite instability in endometrial biopsies as a tool for cancer risk individualization in Lynch syndrome</td>
<td>Julia Canet Hermida, Spain</td>
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<td>11:50-12:20</td>
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<td>Lunch break</td>
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<tr>
<td>Time</td>
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<td>10:00-11:50</td>
<td>WG 7</td>
<td>Immunology &amp; Pathology</td>
<td>Ari Ristimäki, Finland / Magnus von Knebel-Doeberitz, Germany</td>
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<tr>
<td>10:00-10:30</td>
<td>66</td>
<td>Cancer immune prevention in a Lynch syndrome mouse model</td>
<td>Steven Lipkin, USA</td>
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<tr>
<td>10:30-10:40</td>
<td>67</td>
<td>Collaborative study: INDICATE initiative – HLA Type as a modulator of tumor risk in Lynch syndrome?</td>
<td>Matthias Kloor, Germany</td>
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<tr>
<td>10:40-10:50</td>
<td>68</td>
<td>Mismatch repair deficiency and Lynch syndrome in a large series of patients with glioma.</td>
<td>Patrick Benusiglio, France</td>
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<tr>
<td>10:50-11:00</td>
<td>69</td>
<td>The immune profile of normal colonic mucosa as a possible tumor risk modifier in Lynch syndrome?</td>
<td>Lena Bohaumilitzky, Deutschland</td>
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<tr>
<td>11:00-11:10</td>
<td>70</td>
<td>B2M mutation status in stage IV gastrointestinal microsatellite-unstable cancer - Influence on metastatic patterns and response to immune checkpoint blockade</td>
<td>Elena Busch, Germany</td>
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<tr>
<td>11:10-11:20</td>
<td>71</td>
<td>Immunogenicity and HLA binding affinity of MSI-associated frameshift peptide neoantigens</td>
<td>Alejandro Hernandez Sanchez, Germany</td>
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<tr>
<td>11:20-11:30</td>
<td>72</td>
<td>The germline and somatic landscape of mismatch repair proficient early-onset colorectal cancer</td>
<td>Khalid Mahmood, Australia</td>
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<tr>
<td>11:30-11:40</td>
<td>73</td>
<td>Lynch syndrome-associated epithelial ovarian cancer and its immunological profile</td>
<td>Maria Rasmussen, Danmark</td>
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<tr>
<td>11:40-11:50</td>
<td>74</td>
<td>Determining DNA mismatch repair deficiency from tumour features derived from next-generation sequencing for cancer types with a high prevalence of microsatellite instability</td>
<td>Romy Walker, Australia</td>
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<td>11:50-12:20</td>
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<td>Lunch break</td>
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<td>12:20-13:20</td>
<td>State of the Art 6-8</td>
<td>Chairs: Luigi Ricciardiello, Italy / Giulia Martina Cavestro, Italy</td>
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<tr>
<td>12:20-12:40</td>
<td>State of the Art 6</td>
<td><strong>Immune prevention in Lynch syndrome – Is vaccination feasible?</strong></td>
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<td>Matthias Kloor, Germany</td>
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<td>12:40-13:00</td>
<td>State of the Art 7</td>
<td><strong>Chemoprevention for hereditary syndromes</strong></td>
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<td>John Burn, United Kingdom</td>
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<td>13:00-13:20</td>
<td>State of the Art 8</td>
<td><strong>Novel technologies to improve performance of endoscopy</strong></td>
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<td>Robert Hüneburg, Germany</td>
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<td>13:20-13:50</td>
<td>Plenary Discussion State of the Art 6-8</td>
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<td>13:50-15:00</td>
<td>Summary of all WG’s and Plenary discussion</td>
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<td></td>
<td>Chairs: Aysel Adahova, Germany / Sanne ten Broeke, Netherlands</td>
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<td>15:00-15:10</td>
<td>Closing remarks</td>
<td>Gabriela Moeslein, Germany</td>
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**Faculty**

- **Abu-Freha, Naim**
  Ben-Gurion University of the Negev, Omer, Israel
- **Ahadova, Aysel**
  University Hospital, Heidelberg, Germany
- **Auranen, Annika**
  University of Turku, Turku, Finland
- **Benusiglio, Patrick**
  Hôpitaux Pitié-Salpêtrière et Saint-Antoine, Paris, France
- **Bohaumilitzky, Lena**
  University Hospital, Heidelberg, Germany
- **Bruggeling, Carlijn**
  Radboud UMC, Nijmegen, The Netherlands
- **Buchanan, Daniel**
  University of Melbourne, Melbourne, Australia
- **Burn, John**
  Newcastle University, Newcastle upon Tyne, United Kingdom
- **Busch, Elena**
  University Hospital, Heidelberg, Germany
- **Canet Hermida, Julia**
  IDIBELL - L’Hospitalet de Llobregat, Barcelona, Spain
- **Capella, Gabriel**
  IDIBELL - L’Hospitalet de Llobregat, Barcelona, Spain
- **Cavestro, Giulia Martina**
  Università Vita-Salute San Raffaele, Milano, Italy
- **Colletti, Gaia**
  Università Statale di Milano, Milano, Italy
- **Coudert, Marie**
  Institut Curie, Paris, France
- **Cybulski, Cezary**
  Pomeranian Medical University, Szczecin, Poland
- **de Lacy, Antonio**
  Hospital Clínic de Barcelona, Barcelona, Spain
Faculty

Dominguez-Valentin, Mev
University Hospital, Oslo, Norway

Eikenboom, Ellis
Erasmus MC Cancer Institute, Rotterdam, The Netherlands

Evans, Gareth
Manchester University, Manchester, United Kingdom

Fenton, Lora
Imperial College Healthcare NHS Trust, London, United Kingdom

Genuardi, Maurizio
Policlinico Gemelli, Rome, Italy

Georgeson, Peter
University of Melbourne, Melbourne, Australia

Georgiou, Demetra
Cardiff University, Cardiff, United Kingdom

Goretzki, Peter
Charité - University Medicine, Berlin, Germany

Guindalini, Rodrigo,
University of São Paulo, Brasilia, Brazil

Haag, Georg Martin
University Hospital, Heidelberg, Germany

Haupt, Saskia
University Hospital, Heidelberg, Germany

Helderman, Noah
University Medical Centre, Leiden, The Netherlands

Hernandez Sanchez, Alejandro
German Cancer Research Center (DKFZ), Heidelberg, Germany

Hovig, Eivind
University Hospital, Oslo, Norway

Hüneburg, Robert
University Hospital, Bonn, Germany

Jenkins, Mark
University of Melbourne, Melbourne, Australia

Joo, Eric
University of Melbourne, Melbourne, Australia

Kalady, Matt F.
Ohio State University, Columbus, OH, USA

Kang, Yoon-Jung
The Daffodil Centre, Woolloomooloo, Australia

Katona, Bryson
Penn Medicine - University of Pennsylvnia, Philadelphia, USA

Kloor, Matthias
University Hospital, Heidelberg, Germany

Kluźniak, Wojciech
Pomeranian Medical University, Szczecin, Poland

Kohut, Kelly
University of Southampton, London, United Kingdom

Laish, Ido
Sheba medical center, Ramat Gan, Israel

Le Mentec, Marine
Institut Curie, Paris, France

Lepistö, Anna
University of Helsinki, Helsinki, Finland

Levi, Zohar
Rabin Medical Center, Tel Aviv, Israel

Lewis, Samantha
Promega Corporation, Palatine, IL, USA

Lincoln, Anne
King’s College, Guy’s Hospital, London, United Kingdom

Lipkin, Steven
Weill Cornell Medicine, New York, N.Y., USA
Faculty

Lobo, Silvana  
Instituto de Investigação e Inovação em Saúde, Porto, Portugal

Lubinski, Jan  
Pomeranian Medical University, Szczecin, Poland

Mahmood, Khalid  
University of Melbourne, Melbourne, Australia

Mak, Sau  
St. Mark’s Hospital, London, United Kingdom

Marín, Fátima  
IDIBELL - L'Hospitalet de Llobregat, Barcelona, Spain

Mecklin, Jukka-Pekka  
Central Hospital Central Finland, Jyväskylä, Finland

Moeslein, Gabriela  
Bethesda Hospital, Duisburg, Germany

Møller, Pål  
The Norwegian Radium Hospital, Oslo, Norway

Monahan, Kevin  
St. Mark’s Hospital, London, United Kingdom

Nagtegaal, Iris  
Radboud UMC, Nijmegen, The Netherlands

Nattermann, Jacob  
University Hospital, Bonn, Germany

Naxerova, Kamila  
Massachusetts General Hospital, USA

Nielsen, Maartje  
University Medical Center, Leiden, The Netherlands

Nikkola, Jussi  
University Hospital, Tampere, Finland

Olkinuora, Alisa  
University of Helsinki, Helsinki, Finland

O’Sullivan, Hazel  
The Royal Marsden NHS Trust, London, United Kingdom

Patel, Roshani  
The Polyposis Registry, St Mark’s Hospital, London, United Kingdom

Pellise, Maria  
Hospital Clínic Barcelona, Barcelona, Spain

Peltomaki, Paivi  
University of Helsinki, Helsinki, Finland

Phelps, Rachel  
Newcastle University, Newcastle upon Tyne, United Kingdom

Pilar, Mur  
IDIBELL - L’Hospitalet de Llobregat, Barcelona, Spain

Pineda Riu, Marta  
IDIBELL - L’Hospitalet de Llobregat, Barcelona, Spain

Proserpio, Mauro  
Imperial College Healthcare NHS Trust, London, United Kingdom

Pudlarz, Thomas  
Sorbonne Université, Paris, France

Quintana, Isabel  
IDIBELL - L’Hospitalet de Llobregat, Barcelona, Spain

Rasmussen, Maria  
Copenhagen University Hospital, Copenhagen, Danmark

Ricciardiello, Luigi  
University of Bologna, Bologna, Italy

Ristimäki, Ari  
University of Helsinki, Helsinki, Finland

Rofes, Paula  
IDIBELL - L'Hospitalet de Llobregat, Barcelona, Spain

Sampson, Julian  
Cardiff University, Cardiff, United Kingdom
Faculty

Scharf, Florentine  
MGZ – Medizinisch Genetisches Zentrum, München, Germany

Schwenk, Vincent  
MGZ – Medizinisch Genetisches Zentrum, München, Germany

Seifan, Sara  
Cardiff University, Cardiff, United Kingdom

Seppälä, Toni  
University Hospital, Helsinki, Finland

Sievänen, Tero  
University of Jyväskylä, Jyväskylä, Finland

Sijmons, Rolf  
UMCG - University Medical Center, Groningen, The Netherlands

Steinke-Lange, Verena  
MGZ – Medizinisch Genetisches Zentrum, München, Germany

Sunde, Lone  
Aarhus University, Aarhus, Finland

ten Broeke, Sanne  
UMCG - University Medical Center, Groningen, The Netherlands

Terlouw, Diantha  
University Medical Center, Leiden, The Netherlands

Therkildsen, Christina  
Copenhagen University Hospital, Copenhagen, Danmark

Thomas, Laura  
Swansea University Medical School, Swansea, United Kingdom

Truscott, Becky  
Cardiff University, Cardiff, United Kingdom

van der Werf-‘t Lam, Anne-Sophie  
University Medical Center, Leiden, The Netherlands

Faculty

Vara, Covadonga  
IDIBELL - L'Hospitalet de Llobregat, Barcelona, Spain

Villy, Marie-Charlotte  
Institut Curie, Paris, France

von Knebel Doeberitz, Magnus  
University Hospital, Heidelberg, Germany

Walker, Romy  
University of Melbourne, Melbourne, Australia

Walters, Angharad  
Cardiff University, Cardiff, United Kingdom

Winship, Ingrid  
The Royal Melbourne Hospita, Melbourne, Australia

Wokolortczyk, Dominika  
Pomeranian Medical University, Szczecin, Poland

Ykem, Berbel  
Netherlands Cancer Institute, Amsterdam, The Netherlands

Yurgelun, Matthew  
Dana-Farber Cancer Institute, Boston, MA, USA

Zaffaroni, Gloria  
University of Milan, Milano, Italy and Duisburg, Germany
Responsible in accordance with press law:
Prof. Dr. med. Gabriela Moeslein
Evangelisches Bethesda Krankenhaus
Zentrums für Hereditäre Tumorerkrankungen
g.moeslein@bethesda.de

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