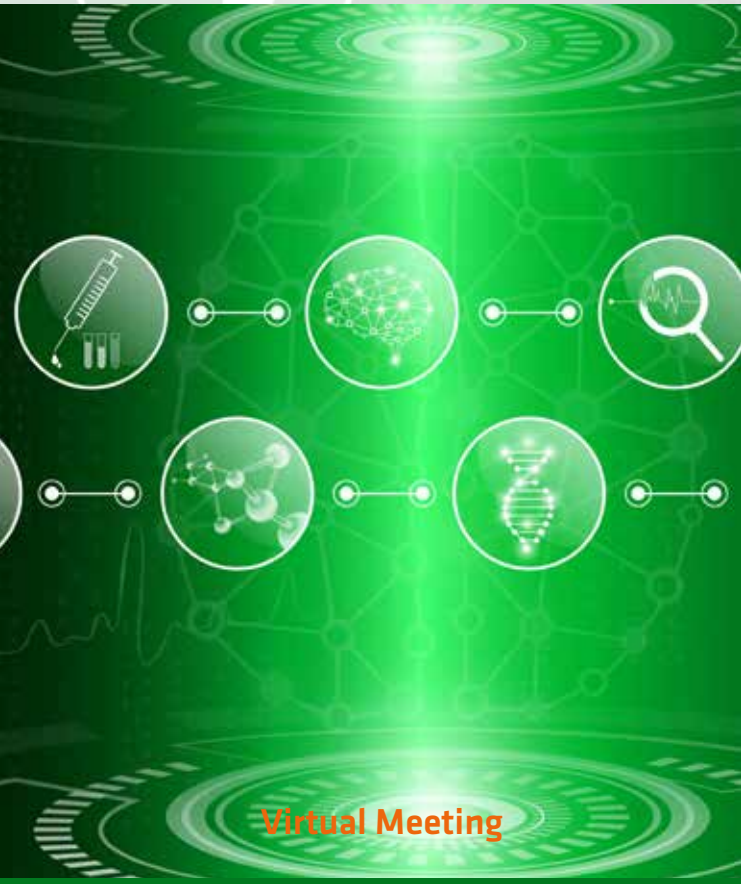
European Hereditary Tumour Group

Final Program
EHTG 5th Meeting

Friday 8 - Saturday 9 October 2021



PLSD

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 pandemic is still present worldwide 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:
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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

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 gyneacological 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

13:40-13:50 13 Development of a CE-Marked IVD Microsatellite Instability Test Utilizing Gold Standard Markers
Samantha Lewis, USA

13:50-14:50 **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**

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-Asso ciated 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 characte rizing 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 syndroms
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!

15:30-17:30 **WG 3**
Polyposis syndromes
Chairs: Daniel Buchanan, Australia / Anna Lepistö, Finland

15:30-16:00 29 Duodenal Polyposis: a significant complication in familial polyposis syndromes
Laura Thomas, United Kingdom

16:00-16:10 30 Diagnostic Yield of Constitutional Gene tic Testing in Patients with Multiple Colo rectal Adenomas (MCRA)
Sau Mak, United Kingdom

16:10-16:20 31 Germline chromothripsis of the APC locus in a patient with adenomatous polyposis
Florentine Scharf, Germany

16:20-16:30 32 An international study of duodenal disease in MAP: incidence of polyposis, cancer, and next steps
Becky Truscott, United Kingdom

16:30-16:40 33 Investigating the molecular mechanisms of adenomatous polyposis syndromes using 3D organoid models.
Angharad Walters, United Kingdom

16:40-16:45 34 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
Eric Joo, Australia

16:45-16:50 35 Differences between inherited and acquired polymerase proofreading deficiencies in cancer
Mur Pilar, Spain

16:50-16:55 36 Paired somatic-germline testing of 15 polyposis and colorectal cancer-predisposing genes highlights the role of APC mosaicism in de novo familial adenomatous polyposis
Paula Rofes, Spain

16:55-17:00 37 3D patient-derived intestinal organoid models for familial polyposis
Sara Seifan, United Kingdom

17:00-17:05 38 Phenotypic analysis of 106 serrated polyposis patients
Verena Steinke-Lange, Germany

17:05-17:10 39 APC mosaicism testing in milder polyposis phenotypes reveals pks+ E.coli bacteria as a possible additional explanation for the development of colorectal adenomas
Diantha Terlouw, The Netherlands

17:10-17:30 Discussion WG 3

17:30 **End of meeting day 1**
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 39 A **PLSD business meeting introduction**
Pål Møller, Norway

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

09:25-09:35 39 B **The Survival Inherited Cancer (SIC) initative**
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

10:50-10:55 45 Inherited Variants in BLM and the Risk and Clinical Characteristics of Breast Cancer
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**

10:00-11:50 **WG 6**
Surveillance & Clinical Management

Chairs: Maria Pellise, Spain /
Jukka-Pekka Mecklin, Finland

10:00-10:30 56 Using Family History and Other Clinical Factors to Personalize Surveillance and Management in Lynch Syndrome
Matthew Yurgelun, United Kingdom

10:30-10:40 57 Risk-stratified FIT for urgent colonoscopy in Lynch Syndrome: A clinical service throughout the COVID-19 pandemic
Anne Lincoln, United Kingdom

10:40-10:50 58 Revisiting the role of immunotherapy for Constitutional Mismatch Repair Deficiency related colorectal cancer treatment
Ellis Eikenboom, Netherlands

10:50-11:00 59 Age of onset of surveillance colonoscopy for MSH6 mutation carriers
Robert Hüneburg, Germany

11:00-11:10 60 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
Yoon-Jung Kang, Australia

11:10-11:20 61 Gastrointestinal Stromal Tumours: Five Years of Molecular Analysis and Referral to Cancer Genetics
Hazel O'Sullivan, United Kingdom

11:20-11:30 62 Molecular screening of urine for Mismatch Repair deficient urothelial tumours; an under-appreciated cancer in Lynch syndrome
John Burn, United Kingdom

Program Saturday, October 9, 2021

11:30-11:40 63 Broadening Risk Profile in Familial Colorectal Cancer Type X; increased risk for five cancer types in the national Danish cohort
Christina Therkildsen, Danmark

11:40-11:45 64 Interval Cancers in Patients with Hereditary Gastrointestinal Syndromes After One Year of the SARS-CoV-2 Pandemic
Giulia Martina Cavestro, Italy

11:45-11:50 65 Preliminary evaluation of highly sensitive assessment of microsatellite instability in endometrial biopsies as a tool for cancer risk individualization in Lynch syndrome
Julia Canet Hermida, Spain

11:50-12:20 **Lunch break**

10:00-11:50 **WG 7**
Immunology & Pathology

Chairs: Ari Ristimäki, Finland / Magnus von Knebel-Doeberitz, Germany

10:00-10:30 66 Cancer immune prevention in a Lynch syndrome mouse model
Steven Lipkin, USA

10:30-10:40 67 Collaborative study: INDICATE initiative – HLA Type as a modulator of tumor risk in Lynch syndrome?
Matthias Kloor, Germany

10:40-10:50 68 Mismatch repair deficiency and Lynch syndrome in a large series of patients with glioma.
Patrick Benusiglio, France

10:50-11:00 69 The immune profile of normal colonic mucosa as a possible tumor risk modifier in Lynch syndrome?
Lena Bohaumilitzky, Deutschland

11:00-11:10 70 B2M mutation status in stage IV gastrointes tinal microsatellite-unstable cancer - Influence onmetastatic patterns and res ponse to immune checkpoint blockade
Elena Busch, Germany

11:10-11:20 71 Immunogenicity and HLA binding affinity of MSI-associated frameshift peptide neoantigens
Alejandro Hernandez Sanchez, Germany

11:20-11:30 72 The germline and somatic landscape of mismatch repair proficient early-onset colorectal cancer
Khalid Mahmood, Australia

11:30-11:40 73 Lynch syndrome-associated epithelial ovari an cancer and its immunological profile
Maria Rasmussen, Danmark

11:40-11:50 74 Determining DNA mismatch repair deficiency from tumour features derived from next-generation sequencing for cancer types with a high prevalence of microsatellite instability
Romy Walker, Australia

11:50-12:20 **Lunch break**

Program Saturday, October 9, 2021

10:00-11:50 **WG 8**
Epidemiology of hereditary cancer

Chairs: Lone Sunde, Denmark / Gabriel Capella, Spain

10:00-10:30 75 Lynch syndrome colorectal cancer risk based on family history and type of DNA MMR gene mutation
Mark Jenkins, Australia

10:30-10:40 76 Gene-, age-, and gender-specific cancer risk of carriers of pathogenic mismatch repair variants from 24 countries worldwide: findings from the Prospective Lynch Syndrome Database
Mev Dominguez-Valentin, Norway

10:40-10:50 77 CNV analysis in a familial cancer cohort
Eivind Hovig, Norway

10:50-11:00 78 Genetic Testing for Assessment of Lynch Syndrome in Young Patients with Polyps
Ido Laish, Israel

11:00-11:10 79 Cancer predisposition and germline CTNNA1 variants
Silvana Lobo, Portugal

11:10-11:20 80 Colorectal cancer incidence in Lynch syndrome reported by IMRC and PLSD
Pal Möller, Norway

11:20-11:30 81 High consanguinity rate and Lynch Syndrome among Bedouin Population in southern Israel
Naim Abu-Freha, Israel

11:30-11:40 82 Upper tract urothelial carcinoma in Lynch syndrome: classification, clinical characterization and course of disease
Jussi Nikkola, Suomi

11:40-11:45 83 Genetic features of Lynch syndrome in the Israeli Arab population, preliminary results
Naim Abu-Freha, Israel

11:45-11:50 84 Computation of confidence intervals in PLSD
Saskia Haupt, Germany

11:50-12:20 **Lunch break**

12:20-13:20 **State of the Art 6-8**
Chairs: Luigi Ricciardiello, Italy / Giulia Martina Cavestro, Italy

12:20-12:40 85 **State of the Art 6**
Immune prevention in Lynch syndrome – Is vaccination feasible?
Matthias Kloor, Germany

12:40-13:00 86 **State of the Art 7**
Chemoprevention for hereditary syndromes
John Burn, United Kingdom

13:00-13:20 87 **State of the Art 8**
Novel technologies to improve performance of endoscopy
Robert Hüneburg, Germany

13:20-13:50 **Plenary Discussion State of the Art 6-8**

13:50-15:00 **Summary of all WG's and Plenary discussion**
Chairs: Aysel Adahova, Germany / Sanne ten Broeke, Netherlands

15:00-15:10 **Closing remarks**
Gabriela Moeslein, Germany

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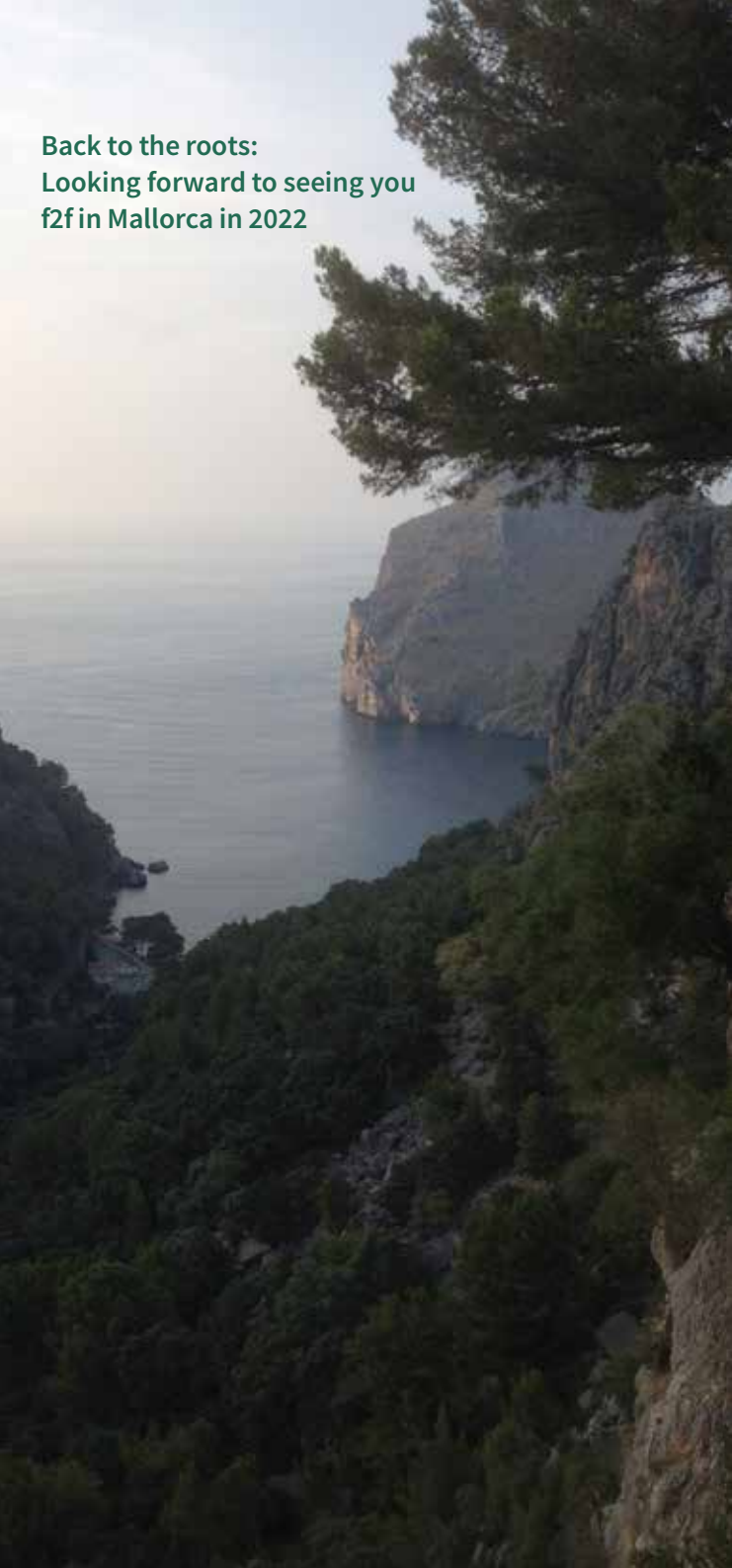
Notes



Back to the roots:
Looking forward to seeing you f2f in Mallorca in 2022