

Fanconi Anemia Cancer Screening Study Newsletter

Division of Cancer Epidemiology and Genetics • Clinical Genetics Branch

RECRUITMENT

We are excited that so many individuals have joined our FA Cancer Screening Study in the last year. A large amount of recruitment took place at the Fanconi Anemia Research Fund (FARF) FA Adult Meeting in Vancouver, British Columbia, Canada, this past September and at Fanconi Anemia Family Retreat at Camp Sunshine in Casco, Maine, USA, this past June. We thank FARF (now Fanconi Cancer Foundation) for their support in enrollment efforts through their quarterly newsletters delivered to families.

As of April 2024, the Study has enrolled 55 participants and so far, 14 have been seen in the NIH Clinical Center in Bethesda, Maryland, USA. We are looking forward to welcoming more than a dozen individuals for their first visit in 2024, and others returning for follow-up visits. Individuals who come to the NIH Clinical Center undergo screening and a battery of tests for early changes for oral, head and neck, esophageal, skin, anal and genital carcinomas, and myelodysplastic syndrome or leukemia for those who have not had a bone marrow transplant.

If you know someone who might also be interested in joining the Fanconi Anemia study, please scan the above QR code to the registration page to make an account and submit a screening for to help us determine if they are eligible, or visit service.cancer.gov/fanconi.

If you have difficulty with your account or online enrollment, please email NCIFamilyStudiesHub@nih.gov for technical assistance. To enroll by phone, please call 240-276-5047 or email NCIFamilyStudyReferrals@nih.gov.



Scan the QR code to navigate to the study registration page.



Photos were included upon approval from the individuals depicted.

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STUDY TEAM MEMBERS

Principal Investigator(s)



Neelam Giri, M.D., M.B.B.S., a pediatric hematologist/oncologist, is the principal investigator of the Fanconi Anemia study. She is in charge of all clinical activities for the Study.



Lisa J. McReynolds, M.D., Ph.D., is a pediatric hematologist/oncologist and co-investigator on the Study. She is responsible for the collaborative scientific research efforts through the Study.

Lead Medical Advisor



Sharon Savage, M.D., is a pediatric hematologist/oncologist and co-investigator. She serves as the lead medical advisor for the Study.

Clinical Team



Margarita Aryavand, CAPT., U.S.P.H.S., M.S.N., C.R.N.P., is a board-certified nurse practitioner and a Commissioned Officer and Captain in the U.S. Public Health Service. She is the lead clinician on the Study. She enrolls and consents patients to the study, conducts telehealth pre-visit, obtains medical records, and sees patients at the NIH Clinical Center.

Michele Nehrebecky, CAPT., U.S.P.H.S.,



F.N.P-BC., is a Family Nurse Practitioner and a Commissioned Officer and a retired Captain in the U.S. Public Health Service. She is a clinician on the Study. She enrolls and consents patients to the study, conducts telehealth pre-visit, obtains medical records, and sees patients at the NIH Clinical Center.

Study Managers



Cecilia Higgs, M.H.S., is a Program Manager. She leads the study management aspects of the Study and manages submissions to the Institutional Review Board.



Soundarya Avantsa, M.P.H., is a Program Manager. She provides scientific support and is involved in streamlining data collection and management.

Study Support



Lisa Leathwood, R.N., B.S.N., is the lead research nurse for the Study.



Kathryn Nichols, R.N., B.S.N., was the lead research nurse for the first year of the study coordinating clinic visits and obtaining medical records.



Jessica Hatton, M.S., C.G.C., is a genetic counselor for the Fanconi Anemia study.



Deborah Flamish, M.A., is a research assistant. She coordinates sample acquisition and sending of sample to collaborators.



Top row (left to right): Lisa Leathwood, Kathryn Nichols
Middle row (left to right): Cecilia Higgs, Deborah Flamish, Soundarya Avantsa
Second row (left to right): Lisa McReynolds, Margarita Aryavand
First row (left to right): Sharon Savage, Neelam Giri, Michele Nehrebecky

Please visit the NCI Fanconi Anemia website at <https://marrowfailure.cancer.gov/fanconi-anemia/study-team.html> to find out more about the key staff members and their roles on the study.

Additional NIH Consultants

Zohreh Khavandgar, D.D.S., Ph.D., is a dental physician and an oral and maxillofacial pathologist. She is in charge of the oral screening (including inspection, photographs, documentation and oral brush biopsies) on all FA individuals in the study.

Clint Allen, M.D., is the chief of the Head and Neck Surgery Oncology program with interest in early detection, diagnosis and treatment of head and neck cancer. He is in charge of oral and head and neck cancer screening on all FA individuals in this study.

Armando Filie, M.D., is the head of the cytopathology section in the Laboratory of Pathology at CCR, NCI. He provides cytopathology diagnosis on oral brush biopsy samples. Pathology lab provides histopathology diagnosis on tissue biopsies.

Theo Heller, M.D., is the section chief and senior investigator in the Clinical Research Section of Liver Disease Branch, NIDDK. He coordinates the

clinical screening and investigational studies for esophageal cancer.

Gracia Rodriguez, M.D., is a gastroenterology fellow with Dr. Theo Heller. She is responsible for seeing all FA individuals on this study and conduct gastrointestinal and hepatology (liver) screening.

Andrea Lisco, M.D., Ph.D., is an Assistant Clinical Investigator, NIAID. He specializes in evaluation of anal malignancy in immunocompromised population (HIV/AIDS). He is in charge of anogenital examination, diagnostic and research studies and coordinate management of dysplastic and anogenital SCC in males.

Sarah Temkin, M.D., a gynecologic oncologist, is an associate director for clinical research on women's health at NIH. She conducts gynecological evaluations in women with FA.

Edward Cowen, M.D., M.H.Sc., is the head of Dermatology consultation service at NIH. He is in charge for the dermatology screening on all FA individuals in the study.

SCREENING ACTIVITY

Oral screening including mouth pictures and brush biopsy for detection or early changes in the mouth area took place at the Fanconi Anemia Camp and the Fanconi Anemia Research Fund Adult Meeting. Screening also takes place at the NIH Clinical Center in Bethesda, Maryland.

We sincerely thank and appreciate Dr. Eunike Velleuer-Carlberg and Christine Krieg from The German Fanconi-Anemia team (<https://fanconi.de/>) for their involvement in the oral screening processes and for sharing their study materials for early detection of oral mucosal changes in FA, with us (icare@fanconi.de).

PRESENTATIONS

DiscovEHRing cancer risk in Fanconi anemia heterozygotes: a genome-first population study. 2023 Fanconi Anemia Research Fund (FARF) Scientific Symposium. McReynolds LJ.

Most Fanconi anemia heterozygotes are not at increased cancer risk: a genome-first DiscovEHR cohort population study. 65th Annual American Society of Hematology Meeting. McReynolds LJ.

Screening for Early Changes of Cancer in Fanconi anemia. 2023 FARF Scientific Symposium. Giri N.

Health Care Screening in Fanconi Anemia: Strengths and Limitations. 2023 FARF Scientific Symposium. Savage S.

Building a psychosocial research program to support individuals and families with rare disease: Lessons learned from Li-Fraumeni Syndrome. 2023 FARF Scientific Symposium. Werner-Lin A.

Non-invasive Technique of Detection of Squamous Cell Carcinoma in Fanconi anemia. 2023 The American Academy of Oral and Maxillofacial Pathology (AAOMP) Annual Meeting. Khavandgar Z.

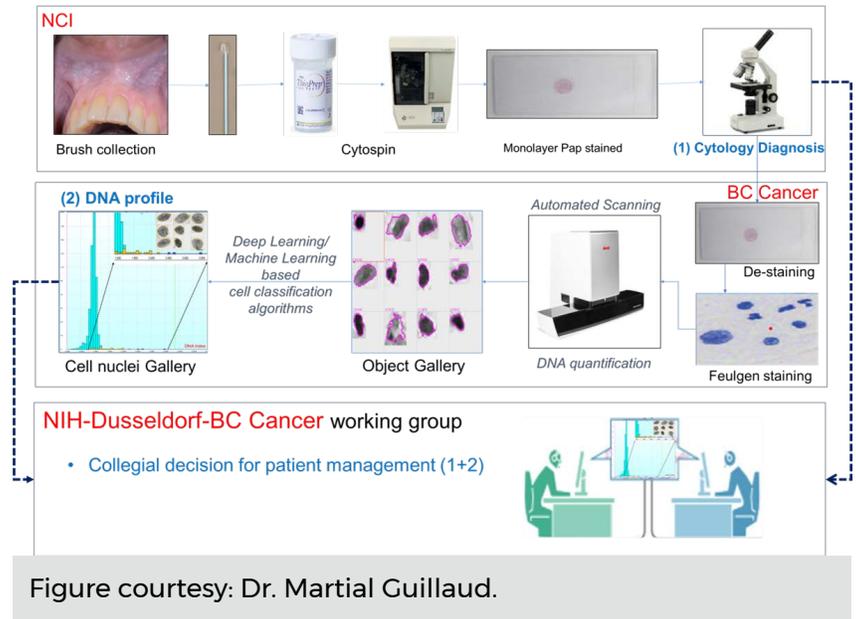
Quantification of large-scale DNA changes in oral brushes using image cytometry: towards integration in oral cancer screening and clinical management of FA patients - BCCRI collaboration. 2023 FARF Scientific Symposium. Guillaud M.

OUR COLLABORATORS – WHERE ARE YOUR SAMPLES GOING?

Martial Guillaud, Ph.D., and Denise Laronde, Ph.D., from the British Columbia Cancer Research Institute (BCCRI), BC Oral Cancer Prevention program at the University of British Columbia, conduct DNA cytometry for interpretation and quantification of aneuploidy on oral brush biopsy cytology slides.

Agata Smogorzewska, M.D., Ph.D., and Tamar Berger, M.D., M.H.A., from the Rockefeller University, NY perform tumor sequencing research to diagnose molecular changes that may help to categorize concerning changes from less urgent findings.

Our team at the NCI-NIH works collaboratively with Dr. Martial Guillaud's team at British Columbia, Dr. Eunike Velleuer and Christine Kreig from Dusseldorf, Germany, and the Rockefeller team Drs. Smogorzewska and Berger in oral screening and interpreting the brush biopsy cytology and DNA ploidy results. The graphic to the right depicts the collaborative process.



Jordi Surralles, Ph.D., and Maria Ramirez, Ph.D., University of Barcelona, Spain. They will perform micronuclei assay on oral brush biopsy samples to determine whether micronuclei frequency in exfoliated tissue can predict tumor evolution in FA.

Emily Vogtmann, Ph.D., M.P.H., from the NCI Metabolic Epidemiology Branch is an expert in the human microbiome. She oversees the oral microbiome analysis of this project.

Coming Soon!

Psychosocial Team

Dr. Emily Pearce, Ph.D., a CGB postdoctoral fellow, will be working alongside Dr. Neelam Giri and Dr. Sadie Hutson to design and implement a study of psychosocial experiences and needs of individuals with FA and their caregivers within the FA screening study currently underway. Dr. Pearce's study will include observation of FA screening visits as well as a brief survey about the screening experience, and follow-up telephone interviews.

We continue to develop partnerships with additional collaborators to study markers for early cancer detection and cancer development.

ADDITIONAL RESOURCES

The Fanconi Cancer Foundation (formerly the Fanconi Anemia Research Fund) has several educational resources published on their website to help people with FA and their families.

Find them here: <https://www.fanconi.org/explore/fa-educational-resources>.

The German Fanconi Anemia Hilfe eV Group has developed an app called “FAExam,” which offers a comprehensive guide for conducting oral self-examinations. You can find the app available for

download on both the iTunes App Store and the Google Play Store.

We would like to thank the Fanconi Cancer Foundation for funding our research efforts. Learn more about FCF here: <https://www.fanconi.org/>



*Thank you for
participating in our
Fanconi Anemia
Cancer Screening study!*