INTERNATIONAL GENETIC EPIDEMIOLOGY SOCIETY

Newsletter – December 2024

Denver City Hall

Letter from IGES President Elizabeth Gillanders

This was an outstanding year for the International Genetic Epidemiology Society. Thank you to the more than 200 people who attended our 33rd annual meeting in Denver November 3-5th 2024. The meeting was a tremendous success. A special thank you to Past-president John Witte, IGES Board member Stephanie Santorico and Scientific Program Committee chair Linda Kachuri for organizing an outstanding meeting. I also want to thank Marie-Pierre Dubé and the ELSI committee who organized a thought-provoking, interactive ELSI activity, which went on during the meeting. Finally, I would like to thank the 2024 Education Committee chaired by Chunyu Liu and Luke Grosvenor for organizing a joint education workshop with ASHG following our meeting focused on '*Development and Evaluation of Polygenic Risk Scores for Application in Diverse Populations*'. The workshop was fantastic, with nearly 70 attendees.

Congratulations to the winners of the 2024 meeting awards: Wenmin Zhang, from the Montreal Heart Institute, won the James V. Neel Young Investigator Award for the best IGES presentation by a young scientist;

Mykhaylo Malakhov from the University of Minnesota won the Roger Williams Memorial Award for the best presentation by a student;

Haoyu Zhang from the National Cancer Institute, Daniel Panyard from Standford University, and Eric Sanders from the University of Toronto were 1st, 2nd, and 3rd place winners, respectively, for the Best Poster Award; and

Gregory Zajac won the Robert C. Elston Award for Best Paper for his paper entitled: *A fast linkage method for population GWAS cohorts with related individuals*.

Congratulations again to Peter Kraft on receiving the IGES Leadership Award! This is the highest honor of the society and recognizes the awardee's contributions to both IGES and the field. David Conti did a wonderful job highlighting Pete's contributions to the society, to research in genetic epidemiology, and his record of training and mentoring. Starting with a quote from ChatGPT: "Pete Kraft's work has significantly advanced the field of genetic epidemiology, particularly in understanding the genetic architecture of cancer and developing methods to better analyze genetic data." But as David noted, what this doesn't capture is Pete's tremendous impact on the field and his colleagues. Peter has provided extraordinary service to IGES, including as President (2020), a member of the IGES Board of Directors (2009-2011), and as a member of the publications committee (2013-2015). Not to mention, even before receiving this award Pete had agreed to serve as a member of the program committee (2025-2027). This is a well-deserved honor recognizing Peter's leadership in IGES and the field.

IGES is a member-led organization, requiring substantial contributions by many people. In addition to those named above, thanks to all the people who helped with the meeting, including:

Chloé Sarnowski and Anthony Herzig of the Young Investigators Committee, which organized social events and a mentoring lunch, and judged the poster prizes;

Hae Kyung Im, chair of the Publications Committee, which selected the Robert C. Elston Award for Best Paper published in Genetic Epidemiology in 2023;

Elizabeth Blue and Lisa Strug, IGES Board members for spearheading fundraising activities, including our IGES t-shirt drive!

By the way, It's not too late, you can still order an IGES tshirt here.
https://www.bonfire.com/international-genetic-epidemiology-society/

Finally, last but not least, Vanessa Olmo and Zenaida Mendoza who helped organize and run the meeting. The Society would be lost without them.

I'm happy to report that our annual general business meeting at the end of IGES was well attended. We are always happy to have reports from the various IGES committees as well as hear suggestions from members about the society and future meetings. I am very happy to report that we expect to turn a sizable profit for IGES 2024.

The Young Investigators Committee (YIC) is calling for applications for early-career researchers to join IGES committees for two years. Applications are currently open **until December 31st.** Nominations will be announced by the YIC around January 27, 2025. For more information on the committees and to submit your application, visit this link.

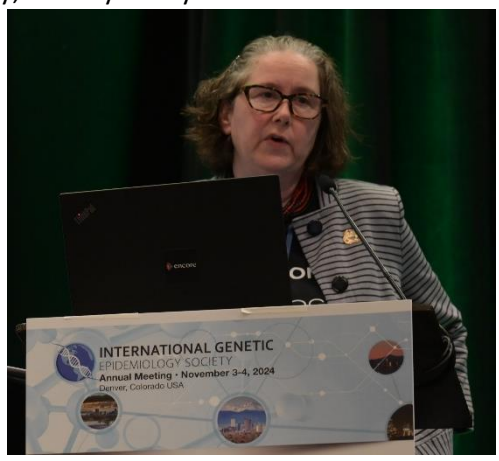
Please feel welcome to give me or other Board members suggestions to improve IGES, including ideas for the 34th Annual IGES Meeting which will take place in Cologne, Germany on August 31 - September 2, 2025. Early abstract submission will start Friday January 24th 2025.

Cologne is a beautiful city with sweeping views of the Rhine River in western Germany. You won't want to miss visiting there. Supplementary information on how to get to Cologne and what to do in and around the city can be found here: https://tinyurl.com/IGES-2025

Finally, don't forget to vote for the President-Elect and Board members. The election for a new President-elect and three members of the Board of Director is now underway. We have excellent candidates. Emmanuelle Génin is our candidate for President-elect. Board candidates are Silke Szymczak, Mohamad Saad and Anne-Louise Leutenegger. Short biographies of our candidates are given in the following pages. Please vote!

Wishing all of our members a happy holiday season and a happy, healthy new year.
See you in Cologne!

Elizabeth Gillanders



2025 IGES Elections
Candidate for IGES President – Emmanuelle Génin

Emmanuelle Génin is the Director of the Inserm Genetics, Genomics, and Bioinformatics Thematic Institute in Paris and the head of the UMR1078 research unit in Brest, France. She trained in genetic epidemiology under the supervision of Françoise Clerget-Darpoux at Josué Feingold's laboratory, initially focusing on Bayesian linkage analysis. Her PhD research then explored methods to map disease-associated genes in inbred populations, showing the importance of accounting for population structure in both linkage and association studies.

During her postdoctoral experience at UC Berkeley with Dr. Glenys Thomson, she developed her expertise into population genetics. Later, she joined Hexagen, a Cambridge-based startup, to lead statistical genetic data analyses. Returning to France, she investigated multifactorial diseases, particularly autoimmune conditions such as multiple sclerosis, while modeling the role of HLA components in disease susceptibility. In 2010, she collaborated with the Sanger Institute during a fellowship at Churchill College, Cambridge where she worked on rare variant association tests and the influence of population stratification. In 2012, she joined the team of Claude Férec at the University of Brest, focusing on dating various mutations associated with cystic fibrosis. Her collaborative research with the University of Wisconsin revisited the origin and dissemination of the p.Phe508del mutation involved cystic fibrosis, suggesting its spread through Bell-Beaker culture populations.

Emmanuelle's current work focuses on French population genetic diversity to optimize association studies for complex diseases and understand the molecular bases of rare diseases. She leads one of the pilot projects under the French Genomic Medicine Initiative, focusing on population references for sequence data interpretation.

An active member of IGES since 1994, she has contributed to the organization of its annual meetings in Nöördwijkerhout (NL) in 2004 and Paris in 2022. She served on the Board of Directors from 2004 to 2006 and has been a member of the editorial board of Genetic Epidemiology since 2011.

Five Selected Publications:

1. Genin E, et al. "APOE and Alzheimer disease: a major gene with semi-dominant inheritance." Molecular Psychiatry, 2011.

2. Farrell P, et al. "Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis." *European Journal of Human Genetics,* 2018.

3. Saint Pierre A, et al. "The genetic history of France." European Journal of Human Genetics, 2020.

4. Bocher O, et al. "Testing for association with rare variants in the coding and non-coding genome: RAVA-FIRST." PLOS Genetics, 2022.

5. Ogloblinsky et al. “PSAP-Genomic-Regions: A Method Leveraging Population Data to Prioritize Coding and Non-Coding Variants in Whole Genome Sequencing for Rare Disease Diagnosis.” Genetic Epidemiolog

2025 IGES Elections
Candidate for Board – Anne-Louise Leutenegger

Anne-Louise Leutenegger is a Senior Research Associate at INSERM, the French National Institute for Health and Medical Research, in Paris. After graduating in statistics, economics and finance from the University of Paris-Dauphine and ENSAE, she moved to the USA to study statistical genetics in Seattle. She obtained her PhD jointly at the University of Washington and the Université Paris-Sud. Her thesis on modelling genome sharing within and between individuals was awarded by the French Biometric Society. After a post-doc on Parkinson's disease genetics, she joined INSERM in 2006 as a full-time researcher in genetic epidemiology.

Her research interests focus on statistical developments for specific populations, such as population isolates, and how population genetics can contribute to disease gene mapping. She has contributed to many collaborative projects aimed at identifying the molecular basis of human traits, both for rare monogenic disorders and for common multifactorial traits.

At INSERM, she has participated in recruitment and evaluation committees and has been a member of the Scientific Advisory Board (2017-2021). She is currently co-leading the methodological developments of INSERM's transversal programme on genomic variability.
She has been a member of IGES since her PhD and is an IGES Fellow. She has been on the editorial board of *Genetic Epidemiology* since 2011. She was part of the organising committee for the IGES 2022 meeting in Paris.

In her spare time, she enjoys cinema, photography and discovering new places (near and far) with her husband and 2 young daughters.

Five selected publications:

1. Ruggiero D, Nutile T, Nappo S, Tirozzi A, Bellenguez C, **Leutenegger AL**, Ciullo M. Genetics of PlGF plasma levels highlights a role of its receptors and supports the link between angiogenesis and immunity. Sci Rep. 2021 Aug 19;11(1):16821.

2. Herzig AF, Nutile T, Babron MC, Ciullo M, Bellenguez C*, **Leutenegger AL***. Strategies for phasing and imputation in a population isolate. Genet Epidemiol. 2018 Mar;42(2):201-213

3. Gazal S, Sahbatou M, Babron MC, Genin E, **Leutenegger AL**. FSuite: exploiting inbreeding in dense SNP chip and exome data. Bioinformatics. 2014 Jul 1;30(13):1940-1

4. Edery P, Marcaillou C, Sahbatou M, Labalme A, Chastang J, Touraine R, Tubacher E, Senni F, Bober MB, Nampoothiri S, Jouk PS, Steichen E, Berland S, Toutain A, Wise CA, Sanlaville D, Rousseau F, Clerget- Darpoux F, **Leutenegger AL**. Association of TALS developmental disorder with defect in minor splicing component U4atac snRNA. Science. 2011; 8;332(6026):240-3

5. **Leutenegger AL**, Prum B, Genin E, Verny C, Lemainque A, Clerget-Darpoux F, Thompson EA. Estimation of the inbreeding coefficient through use of genomic data. Am J Hum Genet. 2003 Sep;73(3):516–23.

2025 IGES Elections
Candidate for Board – Mohamad Saad

My name is **Dr. Mohamad Saad**. I am a Senior Research Scientist and group leader at the Qatar Center of Artificial Intelligence and Qatar Computing Research Institute, part of Hamad Bin Khalifa University. I have a background in applied mathematics and statistics. My research interests are in statistical genetics and bioinformatics. I obtained my bachelor's degree in applied mathematics (majoring in statistics) at the Lebanese University in 2006, my master's and PhD degrees in Statistical Genetics in France at the University of Montpellier II (2007) and the University of Paul Sabatier III (2012), respectively. Between 2012 and 2016, I did my postdoctoral senior fellowship under the supervision of Dr. Ellen Wijsman at the Department of Biostatistics at the University of Washington, Seattle. In 2017, I joined Qatar Computing Research Institute as a research scientist.

I have been an IGES fellow and a member of the society for the past 10 years. I won the Neel Award for the best presentation by a young investigator at IGES Vienna, 2014. I have been serving on the IGES ELSI committee for the past 4 years, and I was a member of Scientific Program Committee of IGES 2024 in Denver. Unfortunately, I could not attend previous IGES conferences (2023-24) due to pending visa application to the US.

Over the past few years, I have been working on data from the Qatar Biobank and Qatar Genome Program. With my team, we are developing Middle Eastern polygenic risk scores, performing GWAS for common and rare variants for cardiometabolic traits, and using machine learning to integrate multi-omics data (metabolomics, proteomics, and genomics) to study cardiometabolic traits, especially type 2 diabetes. See you in Germany!

Five selected publications:

1. **Saad M**, Mokrab Y, Halabi N, Shan J, Razali R, Kunji K, Syed N, Temanni R, Subramanian M, Ceccarelli M, et al. Genetic predisposition to cancer across people of different ancestries in Qatar: a population-based, cohort study. ***Lancet Oncololgy***. 2022;23:341-352. doi: 10.1016/S1470-2045(21)00752-X

2. **Saad M**, El-Menyar A, Kunji K, Ullah E, Al Suwaidi J, Kullo IJ, Validation of Polygenic Risk Scores for Coronary Heart Disease in a Middle Eastern Cohort Using Whole Genome Sequencing. ***Circulation, Genomic and Precision Medicine***, 2022;0:e003712, https://doi.org/10.1161/CIRCGEN.122.003712

3. Caliebe A, Tekola-Ayele F, Darst BF, Wang X, Song YE, Gui J, Sebro RA, Balding DJ, **Saad M**, Dube MP. Including diverse and admixed populations in genetic epidemiology research. ***Genet Epidemiol***. 2022;46:347-371. doi: 10.1002/gepi.22492

4. Sayaman RW*, **Saad M***, Thorsson V, Hu D, Hendrickx W, Roelands J, Porta-Pardo E, Mokrab Y, Farshidfar F, Kirchhoff T, et al. Germline genetic contribution to the immune landscape of cancer. ***Immunity***. 2021;54:367-386 e368. doi: 10.1016/j.immuni.2021.01.011

5. Ullah E, Mall R, Abbas MM, Kunji K, Nato AQ, Jr., Bensmail H, Wijsman EM, **Saad M**. Comparison and assessment of family- and population-based genotype imputation methods in large pedigrees. ***Genome Res***. 2019;29:125-134. doi: 10.1101/gr.236315.118

Keywords: Inclusion, Open Science, Statistical Genetics, Artificial Intelligence, Precision Medicine

2025 IGES Elections
Candidate for Board – Silke Szymczak

Silke Szymczak is Professor of Genetic Epidemiology at the Institute of Medical Biometry and Statistics, University of Lübeck in Germany. During her Bioinformatics diploma thesis (equivalent to a Master's thesis) she generated some of the molecular data in the lab by herself. She earned a PhD in Statistical Genetics from the University of Lübeck, supervised by Andreas Ziegler. During this time she was involved in one of the first GWAS on myocardial infarction. She was a postdoctoral fellow in the Statistical Genetics Section, Inherited Disease Research Branch, National Human Genome Research Institute, National Institutes of Health, Baltimore, MD, USA under the supervision of Joan Bailey-Wilson where she performed her first linkage analysis. Back in Germany, she was head of a junior research group developing and evaluating machine learning methods for systems medicine before being appointed professor in 2020.

Silke's methodological research focuses on the development and evaluation of machine learning approaches for prediction modeling in precision medicine, with a special interest in random forests (RFs). Research topics include variable selection, the integration of pathway or network information into the RF algorithm, RF extensions for the analysis of longitudinal data, providing interpretable models using artificial representative trees and integrating multi modality, e.g. multi-omics, data. Her group is especially interested in performing systematic comparison studies using simulated and experimental omics data. Furthermore, she advocates open science by publishing preprints and open access papers, and making code and simulated data publicly available.

Furthermore, Silke has more than 15 years of experience in the statistical and computational analysis of different types of omics data (genome, transcriptome, proteome, metabolome, methylome, microbiome) in many collaborative research projects focusing on different diseases from cardiology, dermatology, gastroenterology, neurology and oncology.

Silke is a fellow member of IGES, attended her first IGES meeting in 2006 (St. Petersburg) and only skipped a few meetings since then. From 2015 to 2020 she was a member of the Education Committee

and supported the development of the Coursera Genetic Epidemiology Foundations Course. In addition to serving on the Publications Committee (2022-2024) she is organizer of the IGES Journal Club and will be member of the Program Committee from 2025 on. She would be honored to continue her support for IGES by serving on the IGES Board of Directors with a special focus on promoting internationality, junior researchers and open science.

Five selected publications:

1. Balck A*, Borsche M*, Campbell P, Luo X, Harvey J, Brückmann T, Ludwig C, Harms A, Lohmann K, Morris H, Schapira AHV, Hankemeier T, Fleming R, **Szymczak S***, Klein C*. The role of dopaminergic medication, lipid, and endocannabinoid pathway alterations in idiopathic and PRKN/PINK1-mediated Parkinson's disease - a large-scale targeted metabolomics study. *medRxiv* 2024 (under revision at Science Advances)

2. Hu J, **Szymczak S**. A review on longitudinal data analysis with random forest. *Brief Bioinform* 2023,19:bbad002

3. Degenhardt F, Seifert S, **Szymczak S**. Evaluation of variable selection methods for random forests and omics data sets. *Brief Bioinform* 2019;20:492-503

4. Sei Y, Zhao X, Forbes J, **Szymczak S**, Li Q, Trivedi A, Voellinger M, Joy G, Feng J, Whatley M, Jones MS, Harper UL, Marx SJ, Venkatesan AM, Chandrasekharappa SC, Raffeld M, Quezado MM, Louie A, Chen CC, Lim RM, Agarwala R, Schäffer AA, Hughes MS, Bailey-Wilson JE, Wank SA. A Hereditary Form of Small Intestinal Carcinoid Associated With a Germline Mutation in Inositol Polyphosphate Multikinase. *Gastroenterology* 2015;149:67-78

5. Samani NJ, Erdmann J, Hall AS, Hengstenberg C, Mangino M, Mayer B, Dixon RJ, Meitinger T, Braund P, Wichmann H, Barrett JH, König IR, Stevens SE, Szymczak S, Tregouet D, Iles MM, Pahlke F, Pollard H, Lieb W, Cambien F, Fischer M, Ouwehand W, Blankenberg S, Balmforth AJ, Baessler A, Ball SG, Strom TM, Braenne I, Gieger C, Deloukas P, Tobin MD, Ziegler A, Thompson JR, Schunkert H. Genomewide association analysis of coronary artery disease. *N Engl J Med* 2007;357:443-453

* contributed equally

Keywords: problem-solving, collaborative, interdisciplinary, approachable, reproducibility

Report from IGES Treasurer
Julia Bailey

Greeting from the Treasurer.

It was great seeing many of you at our conference in Denver and I'm happy to say that our meeting was successful from both a financial as well as an academic standing. I hope everyone is planning for our next meeting in Cologne, Germany and have noted it's early, at the beginning of September.

Thanks to everyone who renewed their membership. Membership also entitles you to reduced fees for meeting registration and eligibility to participate as a member on one of the various IGES committees or as a board member. Note that IGES is a charitable and educational organization, and any contribution provided by its members is tax-deductible as allowable by law. This can be done along with your membership renewal process – your continuous support of IGES is appreciated!

We are always looking for industry and institutional sponsors to support our meetings.

Our new T-shirt/hoodie fundraiser is being quite successful and there were many people wearing IGES clothes at the meeting. New designs are being developed. You can see and purchase them using the link in the President's Letter of this newsletter (page 4).

I am also in need of a successor to take over in 2026. If you have any interest in the position, please let me know via email (jbailey@mednet.ucla.edu) or in person in Cologne.

Best,

Julia Bailey

Treasurer