Press Information, August 30, 2022

A Matter of Genes

How the risk of disease differs from individual to individual

The Schering Stiftung awards the Friedmund Neumann Prize 2022 to Sarah Kim-Hellmuth for her outstanding work in studying the genetic influence on human gene activity variability. The research award includes a prize money of 10,000 euro.

Dr. Sarah Kim-Hellmuth
Photo: Judith Häusler

“Why do I get ill while others don’t – even though I live more healthily?” We all may have encountered this question at some point in our lives. Dr. Sarah Kim-Hellmuth’s research sheds light on this question. In her large-scale functional genetic analyses, she studies the genetic make-up of hundreds of people simultaneously. She then links the observed genome variation with the gene activity in different body tissues and the potential risk for certain diseases. Her work has highlighted that very different sections of the genome influence the activity of disease-associated genes. This knowledge can now help to develop improved and personalized therapies for the treatment and prevention of a wide variety of diseases including diabetes, cardiovascular diseases, autoimmune diseases or schizophrenia.

Sarah Kim-Hellmuth receives the prize in particular for the study results she delivered as lead analyst in the Genotype-Tissue Expression (GTEx) consortium. She was able to show that the genetic regulation of gene activity is strongly context-specific and that it can differ, for example, between cell types or between women and men. Her work is characterized by the fact that she studies a large number of different tissue types harvested from hundreds of donors. The resulting enormous dataset makes it possible to compare healthy and pathological gene activity in complex diseases. In the future, the knowledge thus gained will facilitate more targeted, disease-specific research and the identification of targets for drug development.

On September 29, clinical geneticist and research group leader Dr. Sarah Kim-Hellmuth will be awarded the **Friedmund Neumann Prize 2022** for her pathbreaking work in studying the genetic influence on human gene activity variability. “Through her work, Dr. Sarah Kim-Hellmuth has significantly expanded our understanding of disease-associated gene variants. The better we understand how our genetic make-up influences the risk of disease, the better we can take into account personal genetic endowment when it comes to prevention and treatment, and the development of personalized therapies,” said Prof. Dr. Dr. h.c. Stefan H. E. Kaufmann, Chair of the Foundation Council of the Schering Stiftung, to explain the jury’s decision.

The Schering Stiftung has awarded the 10,000-euro prize since 2011 to recognize young scientists for their outstanding basic research in human biology, organic chemistry or human medicine. The prize aims to make visible excellent scientific achievements, honor the early development of a distinctive scientific profile, and help the prize winners establish themselves in their field of science.

Sarah Kim-Hellmuth was nominated for the Friedmund Neumann Prize 2022 by Prof. Dr. Eleftheria Zeggini, director of the Institute of Translational Genomics at the Helmholtz Zentrum München. Prof. Dr. Dr. Christoph Klein, director of the Dr. von Hauner Children’s Hospital of the University of Munich (LMU), said about the prize recipient: “From a scientific and clinical perspective, Sarah Kim-Hellmuth and the researcher team of the GTEx consortium have contributed to a milestone that makes possible faster and improved discovery of disease mechanisms and that paves the way toward personalized medicine.”

Award Ceremony

September 29, 2022, from 5:00 p.m.
Berlin-Brandenburg Academy of Sciences and Humanities | Markgrafenstr. 38 | 10117 Berlin | Leibniz Hall

5:00 p.m. Ernst Schering Prize Lecture
Prof. Dr. Gisbert Schneider:
How artificial intelligence has revolutionized drug research

6:00 p.m. Award Ceremony of Friedmund Neumann Prize and Ernst Schering Prize
Award Presentation: Ulrike Gote, Senator for Higher Education and Research, Health, Long-Term Care and Gender Equality

Attendance by registration only. Please register with Maren Isabel Fritz, fritz@scheringstiftung.de .

Lectures by Dr. Sarah Kim-Hellmuth

September 30, 2022
Lecture to students: The fascinating world of genetic diversity and its influence on the human body
Oberstufenzentrum Lise Meitner – School of Science, Berlin-Neukölln (not open to the public)

October 4, 2022, 2:00 p.m.
Public scientific lecture (online): Understanding the diversity of genetic effects on gene expression in health and disease
Berlin Institute of Health at Charité (BIH)
In English. | Please check the website www.bihealth.org for updated registration information.

Background Information

The GTEx consortium has generated the most comprehensive atlas containing 15,201 RNA sequencing samples from 49 tissues from 838 post-mortem donors and data on the sequencing of the entire genome of each donor. With the aid of this enormous dataset, Dr. Sarah Kim-Hellmuth and other members of the consortium have mapped how genetic variants influence gene regulation and how these cellular changes contribute to the genetic risk of both frequent and rare diseases. Together with other scientists of the GTEx project, Dr. Kim-Hellmuth led two studies on cell-type specificity and sex differences in the genetic regulation of gene activity in up to 49 different body tissues from 838 donors. She showed, for example, how it is possible, by using new kinds of bioinformatic analyses, to study genetically modified disease mechanisms at the cell-type level not only through elaborate cell experiments but also through mixed tissue samples that are clinically more easily accessible. In its work on sex differences, the researcher team discovered that up to a third of all genes expressed in the body have different activity in women and men. These genes participate in many different biological processes, including the response to drugs, the control of blood sugar levels in pregnancy, and in cancers. Both studies thus contributed to the largest approach of the GTEx project yet: generating an atlas of genetic regulatory effects. Kim-Hellmuth says about her work as part of the GTEx project: “Worldwide, the data of the GTEx project are being used by numerous scientists for their own research: to compare healthy and pathological gene activity in complex diseases, to identify disease-associated variants and their genes in the relevant tissue, and to integrate this knowledge concretely into the development of drugs.”

Sarah Kim-Hellmuth studied medicine at the University of Munich (LMU) and the Technical University of Munich (TU) and did her specialist training at the Institute of Human Genetics of the University Hospital Bonn, followed by a multi-year postdoc at the New York Genome Center and at Columbia University in New York, where she was lead analyst of the Genotype-Tissue Expression (GTEx) consortium. Since 2021, she has been a clinical geneticist and since early 2022 has led an Emmy Noether Junior Research Group at the Institute of Translational Genomics at the Helmholtz Zentrum München and the Dr. von Hauner Children’s Hospital of the University of Munich (LMU).

Further Information

This press release as well as visual material can be found at https://scheringstiftung.de/presse/.

Press contact:
Maren Isabel Fritz, Project Manager Science
Schering Stiftung | Unter den Linden 32-34 | 10117 Berlin | Phone +49-(0)30-20 62 29-67 |
fritz@scheringstiftung.de