SARAH KIM-HELLMUTH – CURRICULUM VITAE

PERSONAL INFORMATION

Name	Sarah Kim-Hellmuth
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EDUCATION

2014	MD thesis at the Division of Clinical Pharmacology, Department of Internal Medicine, Ludwig-Maximilians-University, Munich, Laboratory of Prof. Dr. Gunther Hartmann Thesis project: "Recognition of microbial DNA by the innate immune system" Grade: summa cum laude
2002 - 2009	Medical training at Ludwig-Maximilians-University Munich (preclinical phase) and Technical University of Munich (clinical phase)

CLINICAL EXPERIENCE

Since 11/2021	Specialist in Human Genetics (Facharzt für Humangenetik) at the Department of Pediatrics, Dr. von Hauner Children's Hospital, University Hospital LMU Munich
2020 – 2021	Resident at the Department of Pediatrics, Dr. von Hauner Children's Hospital, University Hospital LMU Munich
2011 – 2015	Resident at the Institute of Human Genetics, University Hospital, University of Bonn

RESEARCH EXPERIENCE

Since 01/2022	Emmy Noether Group Leader with joint appointment at the Institute of Translational Genomics, Helmholtz Munich, and the Dr. von Hauner Children's Hospital, University Hospital LMU Munich, Germany
04-12/2021	Helmholtz Young Investigator Group Leader at the Institute of Translational Genomics, Helmholtz Munich, Germany (This competitive award funded the first year of my group and was replaced by the more generous Emmy Noether award)
2015 - 2019	Postdoctoral scientist at the New York Genome Center and Columbia University, Laboratory of Dr. Tuuli Lappalainen: Lead analyst of Genotype-Tissue Expression (GTEx) consortium and subgroup leader of the cell type composition analysis team
2011 - 2015	Research fellow at the Institute of Human Genetics, University of Bonn, Laboratory of Prof. Markus M. Nöthen and Institute for Clinical Chemistry and Clinical Pharmacology, University of Bonn, Laboratory of Prof. Veit Hornung
2011 - 2015	Guest researcher at Statistical Genetics, Max-Planck-Institute of Psychiatry, Munich, Laboratory of Prof. Bertram Müller-Myhsok
2009 - 2011	MD thesis research fellow at the Institute for Clinical Chemistry and Clinical Pharmacology, University of Bonn, Laboratory of Prof. Veit Hornung
2005 - 2009	Medical student at the Division of Clinical Pharmacology, Department of Internal Medicine, University of Munich, Laboratory of Prof. Gunther Hartmann and Prof. Stefan Endres

SCIENTIFIC FELLOWSHIPS, HONORS AND AWARDS

Since 2022	DFG Emmy Noether Programme
2022 - 2027	Elected member of Die Junge Akademie of the Berlin-Brandenburg Academy of Sciences and Humanities and the German National Academy of Sciences Leopoldina
2022	Friedmund Neumann Prize 2022
2022	Science Award for Basic Medical Research 2022 of the GlaxoSmithKline Foundation
2021	Adalbert Czerny Award 2021 of the German Society of Pediatrics and Adolescent Medicine
2021	Helmholtz Young Investigator Group Award
2017 - 2019	Marie Skłodowska-Curie Individual Global Fellowship
2016 - 2017	DFG Research Fellowship
2016	Dissertation award of the Münchener Universtitätsgesellschaft

SUPERVISION

Since 2020	Supervision of 3 PhD students and 4 MD students, Department of Pediatrics, Dr. von Hauner Children's Hospital, LMU Munich and Institute of Translational Genomics, Helmholtz Munich, Germany

TEACHING

Since 2020	Lecturer – 1. Genes vs. Environment – the delicate balance between health and disease, PhD program Genomic and Molecular Medicine; 2. Genomic Medicine, interdisciplinary CT seminar, Medical Faculty, University Hospital LMU Munich, Germany
2019	Instructor at *Functional Genetics Boot Camp*, Columbia University, USA
2016 - 2018	Teaching Assistant at Cold Spring Harbor Laboratory Course Statistical Methods for Functional Genomics, Cold Spring Harbor Laboratory, USA
2009 - 2015	Lecturer – 1. Introduction to Human Genetics, 1st clinical semester; 2. Practical Course Genetics, 2nd clinical semester; 3. Antibiotics, Clinical Pharmacology, 4th clinical semester, Medical Faculty, University of Bonn, Germany

PUBLICATIONS (citations = 9801, h-index = 24; i10-index = 29 – Google Scholar August 2022)

1. Flynn, E., Tsu, A., Kasela, S., **Kim-Hellmuth, S.,** Aguet, F., Ardlie, K. G., Bussemaker, H. J., Mohammadi, P. & Lappalainen, T. Transcription factor regulation of eQTL activity across individuals and tissues. *Plos Genet* 18, e1009719 (2022).
2. Brandt, M. K., **Kim-Hellmuth, S.**, Ziosi, M., Gokden, A., Wolman, A., Lam, N., Recinos, Y., Hornung, V. K., Schumacher, J. & Lappalainen, T. An autoimmune disease risk variant: A trans master regulatory effect mediated by IRF1 under immune stimulation? *PLoS Genet* 17(7), (2021). doi.org/10.1371/journal.pgen.1009684
3. Nadel, B. B., Oliva, M., Mitchell, K., Montoya, D. J., **Kim-Hellmuth, S.**, Stranger, B. E., Pellegrini, M. & Mangul, S. Systematic evaluation of transcriptomics-based deconvolution methods and references using thousands of clinical samples. *Briefings in Bioinformatics* bbab265, 2021. doi.org/10.1093/bib/bbab265
4. Warnat-Herresthal, S., Schultze, H., Shastry, K. L., Manamohan, S., Mukherjee, S., Garg, V., Sarveswara, R., Händler, K., Pickkers, P., Aziz, N. A., Ktena, S., Tran, F., Bitzer, M., Ossowski, S., Casadei, N., Herr, C., Petersheim, D., Behrends, U., Kern, F., Fehlmann, T., Schommers, P., Lehmann, C., Augustin, M., Rybniker, J., Altmüller, J., Mishra, N., Bernardes, J. P., Krämer, B., Bonaguro, L., Schulte-Schrepping, J., Domenico, E. D., Siever, C., Kraut, M., Desai, M., Monnet, B., Saridaki, M., Siegel, C. M., Drews, A., Nuesch-Germano, M., Theis, H., Heyckendorf, J., Schreiber, S., **Kim-Hellmuth, S.**, […], Deutsche COVID-19 Omics Initiative (DeCOI), Giamarellos-Bourboulis, E. J., Kox, M., Becker, M., Cheran, S., Woodacre, M. S., Goh, E. L. & Schultze, J. L. Swarm Learning for decentralized and confidential clinical machine learning. *Nature* 1–7 (2021).
5. de Goede, O. M., Nachun, D. C., Ferraro, N. M., Gloudemans, M. J., Rao, A. S., Smail, C., Eulalio, T. Y., Aguet, F., Ng, B., Xu, J., Barbeira, A. N., Castel, S. E., **Kim-Hellmuth, S**., Park, Y., Scott, A. J., Strober, B. J., GTEx Consortium, Brown, C. D., Wen, X., Hall, I. M., Battle, A., Lappalainen, T., Im, H. K., Ardlie, K. G., Mostafavi, S., Quertermous, T., Kirkegaard, K. & Montgomery, S. B. Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. *Cell* (2021).
6. Barbeira, A. N.*, Bonazzola, R.*, Gamazon, E. R.*, Liang, Y.*, Park, Y.*, **Kim-Hellmuth, S.**, Wang, G., Jiang, Z., Zhou, D., Hormozdiari, F., Liu, B., Rao, A., Hamel, A. R., Pividori, M. D., Aguet, F., GTEx GWAS working group, Bastarache, L., Jordan, D. M., Verbanck, M., Do, R., GTEx Consortium, Stephens, M., Ardlie, K., McCarthy, M., Montgomery, S. B., Segre, A. V., Brown, C. D., Lappalainen, T., Wen, X. & Im, H. K. Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. *Genome Biol.* 22, 49–24 (2021).
7. **Kim-Hellmuth, S.***, Hermann, M.*, Eilenberger, J., Ley-Zaporozhan, J., Fischer, M., Hauck, F., Klein, C., Haas, N., Kappler, M., Huebner, J., Jakob, A. & Both, von, U. SARS-CoV-2 Triggering Severe Acute Respiratory Distress Syndrome and Secondary Hemophagocytic Lymphohistiocytosis in a 3-Year-Old Child With Down Syndrome. *Journal of the Pediatric Infectious Diseases Society* 146, e2020009399–4 (2020).
8. **Kim-Hellmuth, S.†***, Aguet, F.*, Oliva, M., Muñoz-Aguirre, M., Kasela, S., Wucher, V., Castel, S. E., Hamel, A. R., Viñuela, A., Roberts, A. L., Mangul, S., Wen, X., Wang, G., Barbeira, A. N., Garrido-Martin, D., Nadel, B., Zou, Y., Bonazzola, R., Quan, J., Brown, A., Martinez-Perez, A., Soria, J. M., GTEx Consortium, Getz, G., Dermitzakis, E., Small, K. S., Stephens, M., Xi, H. S., Im, H. K., Guigó, R., Segre, A. V., Stranger, B. E., Ardlie, K. G. & Lappalainen, T. Cell type specific genetic regulation of gene expression across human tissues. *Science* 369, (2020).

9. Oliva, M.*****, Muñoz-Aguirre, M.*****, **Kim-Hellmuth, S.*,** Wucher, V., Gewirtz, A., Cotter, D., Parsana, P., Kasela, S., Balliu, B., Viñuela, A., Castel, S. E., Mohammadi, P., Aguet, F., Zou, Y., Khramtsova, E., Skol, A., Garrido-Martin, D., Reverter, F., Brown, A., Evans, P., Gamazon, E., Payne, A., Bonazzola, R., Barbeira, A. N., Hamel, A. R., Martinez-Perez, A., Soria, J. M., Consortium, G., Pierce, B., Stephens, M., Eskin, E., Dermitzakis, E., Segre, A. V., Im, H. K., Engelhardt, B., Ardlie, K. G., Montegomery, S., Battle, A., Lappalainen, T., Guigó, R. & Stranger, B. E. The impact of sex on gene expression and its genetic regulation across human tissues. *Science* 369, (2020).
10. GTEx Consortium[#]. The GTEx Consortium atlas of genetic regulatory effects across human tissues. *Science* 369, 1318–1330 (2020). [#]**Lead analyst**, see Fig. 7 and author contribution on page 72 in supplementary materials.
11. Demanelis, K., Jasmine, F., Chen, L. S., Chernoff, M., Tong, L., Delgado, D., Zhang, C., Shinkle, J., Sabarinathan, M., Lin, H., Ramirez, E., Oliva, M., **Kim-Hellmuth, S.**, Stranger, B. E., Lai, T.-P., Aviv, A., Ardlie, K. G., Aguet, F., Ahsan, H., GTEx Consortium, Doherty, J. A., Kibriya, M. G. & Pierce, B. L. Determinants of telomere length across human tissues. *Science* 369, (2020).
12. GTEx Consortium. Genetic effects on gene expression across human tissues. *Nature* 550, 204–213 (2017).
13. **Kim-Hellmuth, S.†,** Bechheim, M., Pütz, B., Mohammadi, P., Nédélec, Y., Giangreco, N., Becker, J., Kaiser, V., Fricker, N., Beier, E., Boor, P., Castel, S. E., Nöthen, M. M., Barreiro, L. B., Pickrell, J. K., Müller-Myhsok, B., Lappalainen, T., Schumacher, J. & Hornung, V. Genetic regulatory effects modified by immune activation contribute to autoimmune disease associations. *Nat Commun* 8, 266 (2017).
14. **Kim-Hellmuth, S.** & Lappalainen, T. Concerted Genetic Function in Blood Traits. *Cell* 167, 1167–1169 (2016).
15. Kehrer, C., Hoischen, A., Menkhaus, R., Schwab, E., Müller, A., **Kim, S.** et al. Whole exome sequencing and array-based molecular karyotyping as aids to prenatal diagnosis in fetuses with suspected Simpson-Golabi-Behmel syndrome. *Prenat Diagn* 36, 961–965 (2016).
16. Schäfgen, J., Cremer, K., Becker, J., **Kim, S.,** Aretz, S., Strom, T. M., Wieczorek, D. & Engels, H. De novo nonsense and frameshift variants of TCF20 in individuals with intellectual disability and postnatal overgrowth. *European Journal of Human Genetics* 24, 1739–1745 (2016).
17. **Kim, S.,** Becker, J., Bechheim, M., Kaiser, V., Noursadeghi, M., Fricker, N., Beier, E., Klaschik, S., Boor, P., Hess, T., Hofmann, A., Holdenrieder, S., Wendland, J. R., Fröhlich, H., Hartmann, G., Nöthen, M. M., Müller-Myhsok, B., Pütz, B., Hornung, V. & Schumacher, J. Characterizing the genetic basis of innate immune response in TLR4-activated human monocytes. *Nat Commun* 5, 5236 (2014).
18. **Kim, S.,** Kaiser, V., Beier, E., Bechheim, M., Guenthner-Biller, M., Ablasser, A., Berger, M., Endres, S., Hartmann, G. & Hornung, V. Self-priming determines high type I IFN production by plasmacytoid dendritic cells. *Eur. J. Immunol.* ***44,*** *807–818 (2014).*
19. Bauernfeind, F., Ablasser, A., Bartok, E., **Kim, S.,** Schmid-Burgk, J., Cavlar, T. & Hornung, V. Inflammasomes: current understanding and open questions. *Cell. Mol. Life Sci.* 68, 765–783 (2011).
20. **Kim, S.,** Bauernfeind, F., Ablasser, A., Hartmann, G., Fitzgerald, K. A., Latz, E. & Hornung, V. Listeria monocytogenes is sensed by the NLRP3 and AIM2 inflammasome. *Eur. J. Immunol.* 40, 1545–1551 (2010).
21. Bauernfeind, F., Ablasser, A., **Kim, S.,** Bartok, E. & Hornung, V. An unexpected role for RNA in the recognition of DNA by the innate immune system. *RNA Biology* 7, 151–157 (2010).
22. Berger, M., Ablasser, A., **Kim, S.,** Bekeredjian-Ding, I., Giese, T., Endres, S., Hornung, V. & Hartmann, G. TLR8-driven IL-12-dependent reciprocal and synergistic activation of NK cells and monocytes by immunostimulatory RNA. *J. Immunother.* 32, 262–271 (2009).
23. Ablasser, A., Poeck, H., Anz, D., Berger, M., Schlee, M., **Kim, S.** et al. Selection of molecular structure and delivery of RNA oligonucleotides to activate TLR7 versus TLR8 and to induce high amounts of IL-12p70 in primary human monocytes. *J. Immunol.* 182, 6824–6833 (2009).
24. Hornung, V., Ellegast, J., **Kim, S.,** Brzózka, K., Jung, A., Kato, H., Poeck, H., Akira, S., Conzelmann, K.-K., Schlee, M., Endres, S. & Hartmann, G. 5'-Triphosphate RNA is the ligand for RIG-I. *Science* 314, 994–997 (2006).

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