

SARAH KIM-HELLMUTH – Publications

Current list of publications can be found on my [Google Scholar profile](#)

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., Gludemans, 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., Montgomery, 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. Schäfergen, J., Cremer, K., Becker, J., **Kim, S.**, Aretz, S., Strom, T. M., Wiczorek, 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., Guenther-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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