

# Masahiro Kanai

## Curriculum Vitae

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## Education

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**Harvard University** **Boston, USA**  
Bioinformatics and Integrative Genomics PhD Program, Harvard Medical School Aug. 2017–present

**Keio University** **Yokohama, Japan**  
B.S. in Bioinformatics, Department of Biosciences and Informatics Apr. 2013–Mar. 2017

## Research Experience

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**Massachusetts General Hospital & Broad Institute of MIT and Harvard** **Boston, USA**  
Graduate Student, Analytic and Translational Genetics Unit Jan. 2018–present

**Advisors:** Drs. Mark Daly & Hilary Finucane

**Research theme:** Cross-population analysis of complex human diseases and traits.

**Osaka University Graduate School of Medicine** **Osaka, Japan**  
Research Student, Department of Statistical Genetics Apr. 2017–present

**Advisor:** Dr. Yukinori Okada

**Research themes:**

- Genetic analysis of complex human diseases and traits in the Japanese population.
- Efficient visualization of high-dimensional multi-omics data.

**RIKEN Center for Integrative Medical Sciences** **Yokohama, Japan**  
Research Assistant, Laboratory for Statistical Analysis May 2015–Jul. 2019

**Advisors:** Drs. Yoichiro Kamatani & Yukinori Okada

**Research themes:**

- Genome-wide association study (GWAS) of ~200,000 individuals in the BioBank Japan Project.
- Interpretation of GWAS polygenic signals using epigenomic data.
- Population genetics analysis of 1,037 Japanese whole-genome sequences.

**Keio University** **Yokohama, Japan**  
Undergraduate Researcher, Laboratory for Bioinformatics Apr. 2016–Mar. 2017

**Advisor:** Dr. Yasubumi Sakakibara

**Bachelor thesis:** Integrative multi-omics analysis of renal cell carcinoma.

**Tokyo Medical and Dental University** **Tokyo, Japan**  
Technical Assistant, Department of Human Genetics and Disease Diversity Apr. 2014–Mar. 2016

**Advisors:** Drs. Yukinori Okada & Toshihiro Tanaka

**Research themes:**

- Empirical estimation of genome-wide significance thresholds based on GWAS simulations.
- HLA imputation analysis using the Japanese-specific reference panel.

## Certification

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**The Certification for Bioinformatics Engineers**  
certificated by the Japanese Society of Bioinformatics

2015

## Awards and Fellowships

<b>Reviewers' Choice Abstract Award</b>	<b>USA</b>
<i>The American Society of Human Genetics 2021 Virtual Meeting</i>	2021
<b>Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research</b>	<b>USA</b>
Finalist (Predoctoral), <i>The American Society of Human Genetics 2020 Virtual Meeting</i>	2020
<b>Reviewers' Choice Abstract Award</b>	<b>Houston, USA</b>
<i>The American Society of Human Genetics 2019 Annual Meeting</i>	2019
<b>Masason Foundation Fellowship</b>	<b>Tokyo, Japan</b>
Research grant for cross-population analysis of complex human diseases and traits.	2018–2022+
<b>Nakajima Foundation Fellowship</b>	<b>Tokyo, Japan</b>
Predocotrual fellowship which covers up to ~\$50,000/year for tuition and stipend.	2017–2022
<b>The 3rd place, Worldwide Finals</b>	<b>New York, USA</b>
<i>Microsoft Imagine Cup 2011 Windows 7 Touch Challenge</i>	2011
<b>The 1st place, National Finals &amp; Worldwide Finalist</b>	<b>Japan &amp; Poland</b>
<i>Microsoft Imagine Cup 2010 Software Design Competition</i>	2010

## Publications

### Selected Publications and Preprints

(\* = co-first)

1. **Kanai, M.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Karczewski, K. J., Fauman, E. B., Wang, Q. S., Jacobs, H., Aguet, F., Ardlie, K. G., Kerimov, N., Alasoo, K., Benner, C., Ishigaki, K., Sakaue, S., Reilly, S., The BioBank Japan Project, FinnGen, Kamatani, Y., Matsuda, K., Palotie, A., Neale, B. M., Tewhey, R., Sabeti, P. C., Okada, Y., Daly, M. J. & Finucane, H. K. Insights from complex trait fine-mapping across diverse populations. *medRxiv* (2021).
2. \*Sakaue, S., \***Kanai, M.**, Tanigawa, Y., Karjalainen, J., Kurki, M., Koshiba, S., Narita, A., Konuma, T., Yamamoto, K., Akiyama, M., Ishigaki, K., Suzuki, A., Suzuki, K., Obara, W., Yamaji, K., Takahashi, K., Asai, S., Takahashi, Y., Suzuki, T., Shinozaki, N., Yamaguchi, H., Minami, S., Murayama, S., Yoshimori, K., Nagayama, S., Obata, D., Higashiyama, M., Masumoto, A., Koretsune, Y., Ito, K., Terao, C., Yamauchi, T., Komuro, I., Kadowaki, T., Tamiya, G., Yamamoto, M., Nakamura, Y., Kubo, M., Murakami, Y., Yamamoto, K., Kamatani, Y., Palotie, A., Rivas, M. A., Daly, M. J., Matsuda, K. & Okada, Y. A cross-population atlas of genetic associations for 220 human phenotypes. *Nature Genetics* **53**, 1415–1424 (2021).
3. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* (2021).
4. \*Weissbrod, O., \***Kanai, M.**, \*Shi, H., Gazal, S., Peyrot, W., Khera, A., Okada, Y., The Biobank Japan Project, Martin, A., Finucane, H. & Price, A. L. Leveraging fine-mapping and non-European training data to improve trans-ethnic polygenic risk scores. *medRxiv* (2021).
5. \*Sakaue, S., \***Kanai, M.**, Karjalainen, J., Akiyama, M., Kurki, M., Matoba, N., Takahashi, A., Hirata, M., Kubo, M., Matsuda, K., Murakami, Y., Daly, M. J., Kamatani, Y. & Okada, Y. Trans-biobank analysis with 676,000 individuals elucidates the association of polygenic risk scores of complex traits with human lifespan. *Nature Medicine* **26**, 542–548 (2020).
6. Martin, A. R., **Kanai, M.**, Kamatani, Y., Okada, Y., Neale, B. M. & Daly, M. J. Clinical use of current polygenic risk scores may exacerbate health disparities. *Nature Genetics* **51**, 584–591 (2019).
7. **Kanai, M.**, Akiyama, M., Takahashi, A., Matoba, N., Momozawa, Y., Ikeda, M., Iwata, N., Ikegawa, S., Hirata, M., Matsuda, K., Kubo, M., Okada, Y. & Kamatani, Y. Genetic analysis of quantitative traits in the Japanese population links cell types to complex human diseases. *Nature Genetics* **50**, 390–400 (2018).

## Other Publications and Preprints.....

8. Tsuo, K., Zhou, W., Wang, Y., **Kanai, M.**, Namba, S., Gupta, R., Majara, L., Nkambule, L. L., Okada, Y., Morisaki, T., Neale, B., Daly, M. J., Martin, A. & Global Biobank Meta-analysis Initiative. Multi-ancestry meta-analysis of asthma identifies novel associations and highlights the value of increased power and diversity. *medRxiv* (2021).
9. Bhattacharya, A., Hirbo, J. B., Zhou, D., Zhou, W., Zheng, J., **Kanai, M.**, The Global Biobank Meta-analysis Initiative, Daly, M. J., Pasaniuc, B., Gamazon, E. R. & Cox, N. J. Best practices of multi-ancestry, meta-analytic transcriptome-wide association studies: lessons from the Global Biobank Meta-analysis Initiative. *medRxiv* (2021).
10. Wang, Y., Namba, S., Lopera-Maya, E. A., Kerminen, S., Tsuo, K., Lall, K., **Kanai, M.**, Zhou, W., Wu, K.-H. H., Fave, M.-J., Bhatta, L., Awadalla, P., Deelen, P., Lo Faro, V., Magi, R., Murakami, Y., Brumpton, B. M., Sanna, S., Uzunovic, J., Gamazon, E. R., Cox, N. J., Surakka, I., Okada, Y., Martin, A. R. & Hirbo, J. Global biobank analyses provide lessons for computing polygenic risk scores across diverse cohorts. *medRxiv* (2021).
11. Global Biobank Meta-analysis Initiative. Global Biobank Meta-analysis Initiative: powering genetic discovery across human diseases. *medRxiv* (2021).
12. Polygenic Risk Score Task Force of the International Common Disease Alliance. Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps. *Nature Medicine* (2021).
13. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19: an update. *medRxiv* (2021).
14. Luo, Y., **Kanai, M.**, Choi, W., Li, X., Sakaue, S., Yamamoto, K., Ogawa, K., Gutierrez-Arcelus, M., Gregersen, P. K., Stuart, P. E., Elder, J. T., Forer, L., Schönherr, S., Fuchsberger, C., Smith, A. V., Fellay, J., Carrington, M., Haas, D. W., Guo, X., Palmer, N. D., Chen, Y.-D. I., Rotter, J. I., Taylor, K. D., Rich, S. S., Correa, A., Wilson, J. G., Kathiresan, S., Cho, M. H., Metspalu, A., Esko, T., Okada, Y., Han, B., McLaren, P. J. & Raychaudhuri, S. A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nature Genetics* **53**, 1504–1516 (2021).
15. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., Davis, J. R., **Kanai, M.**, Yang, D. K., Butts, J. C., Guney, M. H., Luban, J., Montgomery, S. B., Finucane, H. K., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. *Cell* **184**, 5247–5260.e19 (2021).
16. Palmer, D. S., Zhou, W., Abbott, L., Baya, N., Churchhouse, C., Seed, C., Poterba, T., King, D., **Kanai, M.**, Bloemendal, A. & Neale, B. M. Analysis of genetic dominance in the UK Biobank. *bioRxiv* (2021).
17. Reilly, S. K., Gosai, S. J., Guterres, A., Ulirsch, J. C., **Kanai, M.**, Berenzy, D., Kales, S., Butler, G. B., Gladden-Young, A., Finucane, H. K., Sabeti, P. C. & Tewhey, R. Direct characterization of cis-regulatory elements and functional dissection of complex genetic associations using HCR-FlowFISH. *Nature Genetics* **53**, 1166–1176 (2021).
18. Ruotsalainen, S. E., Surakka, I., Mars, N., Karjalainen, J., Kurki, M., **Kanai, M.**, Mishra, P. P., Mishra, B. H., Sinisalo, J., Palta, P., Lehtimäki, T., Raitakari, O., Estonian Biobank research team, Milani, L., The Biobank Japan Project, Okada, Y., FinnGen, Palotie, A., Widen, E., Daly, M. J. & Ripatti, S. Loss-of-function of MFG8 and protection against coronary atherosclerosis. *medRxiv* (2021).
19. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., Cui, R., Albors, C., Cheng, N., Okada, Y., Aguet, F., Ardlie, K. G., MacArthur, D. G. & Finucane, H. K. Leveraging supervised learning for functionally informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. *Nature Communications* **12**, 1–11 (2021).

20. Namkoong, H., Edahiro, R., Fukunaga, K., Shirai, Y., Sonehara, K., Tanaka, H., Lee, H., Hasegawa, T., **Kanai, M.**, Naito, T., Yamamoto, K., Saiki, R., Hyugaji, T., Shimizu, E., Katayama, K., Takahashi, K., Harada, N., Naito, T., Hiki, M., Matsushita, Y., Takagi, H., Aoki, R., Nakamura, A., Harada, S., Sasano, H., Kabata, H., Masaki, K., Kamata, H., Ikemura, S., Chubachi, S., Okamori, S., Terai, H., Morita, A., Asakura, T., Sasaki, J., Morisaki, H., Uwamino, Y., Nanki, K., Mikami, Y., Uchida, S., Uno, S., Ishihara, R., Matsubara, Y., Nishimura, T., Ogawa, T., Ishiguro, T., Isono, T., Shibata, S., Matsui, Y., Hosoda, C., Takano, K., Nishida, T., Kobayashi, Y., Takaku, Y., Takayanagi, N., Ueda, S., Tada, A., Miyawaki, M., Yamamoto, M., Yoshida, E., Hayashi, R., Nagasaka, T., Arai, S., Kaneko, Y., Sasaki, K., Tagaya, E., Kawana, M., Arimura, K., Takahashi, K., Anzai, T., Ito, S., Endo, A., Uchimura, Y., Miyazaki, Y., Honda, T., Tateishi, T., Tohda, S., Ichimura, N., Sonobe, K., Sassa, C., Nakajima, J., Nakano, Y., Nakajima, Y., Anan, R., Arai, R., Kurihara, Y., Harada, Y., Nishio, K., Ueda, T., Azuma, M., Saito, R., Sado, T., Miyazaki, Y., Sato, R., Haruta, Y., Nagasaki, T., Yasui, Y., Hasegawa, Y., Mutoh, Y., Sato, T., Takei, R., Hagimoto, S., Noguchi, Y., Yamano, Y., Sasano, H., Ota, S., Nakamori, Y., Yoshiya, K., Saito, F., Yoshihara, T., Wada, D., Iwamura, H., Kanayama, S., Maruyama, S., Yoshiyama, T., Ohta, K., Kokuto, H., Ogata, H., Tanaka, Y., Arakawa, K., Shimoda, M., Osawa, T., Tateno, H., Hase, I., Yoshida, S., Suzuki, S., Kawada, M., Horinouchi, H., Saito, F., Mitamura, K., Hagihara, M., Ochi, J., Uchida, T., Baba, R., Arai, D., Ogura, T., Takahashi, H., Hagiwara, S., Nagao, G., Konishi, S., Nakachi, I., Murakami, K., Yamada, M., Sugiura, H., Sano, H., Matsumoto, S., Kimura, N., Ono, Y., Baba, H., Suzuki, Y., Nakayama, S., Masuzawa, K., Namba, S., Suzuki, K., Hizawa, N., Shiroyama, T., Miyawaki, S., Kawamura, Y., Nakayama, A., Matsuo, H., Maeda, Y., Nii, T., Noda, Y., Niitsu, T., Adachi, Y., Enomoto, T., Amiya, S., Hara, R., Kishikawa, T., Yamada, S., Kawabata, S., Kijima, N., Takagaki, M., Sasa, N., Ueno, Y., Suzuki, M., Takemoto, N., Eguchi, H., Fukusumi, T., Imai, T., Fukushima, M., Kishima, H., Inohara, H., Tomono, K., Kato, K., Takahashi, M., Matsuda, F., Hirata, H., Takeda, Y., Koh, H., Manabe, T., Funatsu, Y., Ito, F., Fukui, T., Shinozuka, K., Kohashi, S., Miyazaki, M., Shoko, T., Kojima, M., Adachi, T., Ishikawa, M., Takahashi, K., Inoue, T., Hirano, T., Kobayashi, K., Takaoka, H., Watanabe, K., Miyazawa, N., Kimura, Y., Sado, R., Sugimoto, H., Kamiya, A., Kuwahara, N., Fujiwara, A., Matsunaga, T., Sato, Y., Okada, T., Hirai, Y., Kawashima, H., Narita, A., Niwa, K., Sekikawa, Y., Nishi, K., Nishitsuji, M., Tani, M., Suzuki, J., Nakatsumi, H., Ogura, T., Kitamura, H., Hagiwara, E., Murohashi, K., Okabayashi, H., Mochimaru, T., Nukaga, S., Satomi, R., Oyamada, Y., Mori, N., Baba, T., Fukui, Y., Odate, M., Mashimo, S., Makino, Y., Yagi, K., Hashiguchi, M., Kagyo, J., Shiomi, T., Fuke, S., Saito, H., Tsuchida, T., Fujitani, S., Takita, M., Morikawa, D., Yoshida, T., Izumo, T., Inomata, M., Kuse, N., Awano, N., Tone, M., Ito, A., Nakamura, Y., Hoshino, K., Maruyama, J., Ishikura, H., Takata, T., Odani, T., Amishima, M., Hattori, T., Shichinohe, Y., Kagaya, T., Kita, T., Ohta, K., Sakagami, S., Koshida, K., Hayashi, K., Shimizu, T., Kozu, Y., Hiranuma, H., Gon, Y., Izumi, N., Nagata, K., Ueda, K., Taki, R., Hanada, S., Kawamura, K., Ichikado, K., Nishiyama, K., Muranaka, H., Nakamura, K., Hashimoto, N., Wakahara, K., Koji, S., Omote, N., Ando, A., Kodama, N., Kaneyama, Y., Maeda, S., Kuraki, T., Matsumoto, T., Yokote, K., Nakada, T.-A., Abe, R., Oshima, T., Shimada, T., Harada, M., Takahashi, T., Ono, H., Sakurai, T., Shibusawa, T., Kimizuka, Y., Kawana, A., Sano, T., Watanabe, C., Suematsu, R., Sageshima, H., Yoshifuji, A., Ito, K., Takahashi, S., Ishioka, K., Nakamura, M., Masuda, M., Wakabayashi, A., Watanabe, H., Ueda, S., Nishikawa, M., Chihara, Y., Takeuchi, M., Onoi, K., Shinozuka, J., Sueyoshi, A., Nagasaki, Y., Okamoto, M., Ishihara, S., Shimo, M., Tokunaga, Y., Kusaka, Y., Ohba, T., Isogai, S., Ogawa, A., Inoue, T., Fukuyama, S., Eriguchi, Y., Yonekawa, A., Kan-o, K., Matsumoto, K., Kanaoka, K., Ihara, S., Komuta, K., Inoue, Y., Chiba, S., Yamagata, K., Hiramatsu, Y., Kai, H., Asano, K., Oguma, T., Ito, Y., Hashimoto, S., Yamasaki, M., Kasamatsu, Y., Komase, Y., Hida, N., Tsuburai, T., Oyama, B., Takada, M., Kanda, H., Kitagawa, Y., Fukuta, T., Miyake, T., Yoshida, S., Ogura, S., Abe, S., Kono, Y., Togashi, Y., Takoi, H., Kikuchi, R., Ogawa, S., Ogata, T., Ishihara, S., Kanehiro, A., Ozaki, S., Fuchimo, Y., Wada, S., Fujimoto, N., Nishiyama, K., Terashima, M., Beppu, S., Yoshida, K., Narumoto, O., Nagai, H., Ooshima, N., Motegi, M., Umeda, A., Miyagawa, K., Shimada, H., Endo, M., Ohira, Y., Watanabe, M., Inoue, S., Igarashi, A., Sato, M., Sagara, H., Tanaka, A., Ohta, S., Kimura, T., Shibata, Y., Tanino, Y., Nikaido, T., Minemura, H., Sato, Y., Yamada, Y., Hashino, T., Shinoki, M., Iwagoe, H., Takahashi, H., Fujii, K., Kishi, H., Kanai, M., Imamura, T., Yamashita, T., Yatomi, M., Maeno, T., Hayashi, S., Takahashi, M., Kuramochi, M., Kamimaki, I., Tominaga, Y., Ishii, T., Utsugi, M., Ono, A., Tanaka, T., Kashiwada, T., Fujita, K., Saito, Y.,

- Seike, M., Omae, Y., Nannya, Y., Ueno, T., Takano, T., Katayama, K., Ai, M., Kumanogoh, A., Sato, T., Hasegawa, N., Tokunaga, K., Ishii, M., Koike, R., Kitagawa, Y., Kimura, A., Imoto, S., Miyano, S., Ogawa, S., Kanai, T. & Okada, Y. Japan COVID-19 Task Force: a nation-wide consortium to elucidate host genetics of COVID-19 pandemic in Japan. *medRxiv* (2021).
21. Koskela, J. T., Happola, P., Liu, A., Partanen, J., Genovese, G., Artomov, M., Myllymaki, M. N. M., **Kanai, M.**, Zhou, W., Karjalainen, J., Palviainen, T., Ronkainen, J., Sebert, S., Tukiainen, T., Palta, P., Kaprio, J., Kurki, M., Ganna, A., Palotie, A., Laitinen, T., Myllarniemi, M., Daly, M. J. & FinnGen. Genetic variant in SPDL1 reveals novel mechanism linking pulmonary fibrosis risk and cancer protection. *medRxiv* (2021).
  22. Nakatochi, M., Toyoda, Y., **Kanai, M.**, Nakayama, A., Kawamura, Y., Hishida, A., Mikami, H., Matsuo, K., Takezaki, T., Momozawa, Y., Biobank Japan Project, Kamatani, Y., Ichihara, S., Shinomiya, N., Yokota, M., Wakai, K., Okada, Y., Matsuo, H. & Japan Uric Acid Genomics Consortium (Japan Urate). An X chromosome-wide meta-analysis based on Japanese cohorts revealed that non-autosomal variations are associated with serum urate. *Rheumatology* **60**, 4430–4432 (2021).
  23. Turley, P., Martin, A. R., Goldman, G., Li, H., **Kanai, M.**, Walters, R. K., Jala, J. B., Lin, K., Millwood, I. Y., Carey, C. E., Palmer, D. S., Zacher, M., Atkinson, E. G., Chen, Z., Li, L., Akiyama, M., Okada, Y., Kamatani, Y., Walters, R. G., Callier, S., Laibson, D., Meyer, M. N., Cesarini, D., Daly, M., Benjamin, D. J. & Neale, B. M. Multi-Ancestry Meta-Analysis yields novel genetic discoveries and ancestry-specific associations. *bioRxiv* (2021).
  24. Pirastu, N., Cordioli, M., Nandakumar, P., Mignogna, G., Abdellaoui, A., Hollis, B., **Kanai, M.**, Rajagopal, V. M., Della Briotta Parolo, P., Baya, N., Carey, C. E., Karjalainen, J., Als, T. D., Van der Zee, M. D., Day, F. R., Ong, K. K., Morisaki, T., de Geus, E., Bellocco, R., Okada, Y., Børghlum, A. D., Joshi, P., Auton, A., Hinds, D., Neale, B. M., Walters, R. K., Nivard, M. G., Perry, J. R. B. & Ganna, A. Genetic analyses identify widespread sex-differential participation bias. *Nature Genetics* **53**, 663–671 (2021).
  25. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., Siewert, K. M., Kim, S. S., Luo, Y., Amariuta, T., Huang, H., Okada, Y., Raychaudhuri, S., Sunyaev, S. R. & Price, A. L. Population-specific causal disease effect sizes in functionally important regions impacted by selection. *Nature Communications* **12**, 1098 (2021).
  26. Atkinson, E. G., Maihofer, A. X., **Kanai, M.**, Martin, A. R., Karczewski, K. J., Santoro, M. L., Ulirsch, J. C., Kamatani, Y., Okada, Y., Finucane, H. K., Koenen, K. C., Nievergelt, C. M., Daly, M. J. & Neale, B. M. Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. *Nature Genetics* **53**, 195–204 (2021).
  27. Rämö, J. T., Kiiskinen, T., Karjalainen, J., Krebs, K., Kurki, M., Havulinna, A. S., Hämäläinen, E., Häppölä, P., Hautakangas, H., FinnGen, Karczewski, K. J., **Kanai, M.**, Mägi, R., Palta, P., Esko, T., Metspalu, A., Pirinen, M., Ripatti, S., Milani, L., Mäkitie, A., Daly, M. J. & Palotie, A. Genome-wide Screen of Otosclerosis in Population Biobanks: 18 Loci and Shared Heritability with Skeletal Structure. *medRxiv* (2020).
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## Presentations

### International Conference.....

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