

# Association mapping of the xMHC in the risk of childhood acute lymphoblastic leukemia in Japanese

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## VI. References

1. The Leukemia & Lymphoma Society 2009
2. NCCN Guideline <<https://www2.tri-kobe.org/nccn/guideline/hematologic/english/all.pdf>>
3. Greaves M. (2018). A causal mechanism for childhood acute lymphoblastic leukaemia. *Nature reviews. Cancer*, 18(8), 471–484. <https://doi.org/10.1038/s41568-018-0015-6>
4. Urayama, K. Y., Chokkalingam, A. P., Metayer, C., Hansen, H., May, S., Ramsay, P., Wiemels, J. L., Wiencke, J. K., Trachtenberg, E., Thompson, P., Ishida, Y., Brennan, P., Jolly, K. W., Termuhlen, A. M., Taylor, M., Barcellos, L. F., & Buffler, P. A. (2013). SNP association mapping across the extended major histocompatibility complex and risk of B-cell precursor acute lymphoblastic leukemia in children. *PloS one*, 8(8), e72557. <https://doi.org/10.1371/journal.pone.0072557>.
5. Horton, R., Wilming, L., Rand, V., Lovering, R. C., Bruford, E. A., Khodiyar, V. K., Lush, M. J., Povey, S., Talbot, C. C., Jr, Wright, M. W., Wain, H. M., Trowsdale, J., Ziegler, A., & Beck, S. (2004). Gene map of the extended human MHC. *Nature reviews. Genetics*, 5(12), 889–899. <https://doi.org/10.1038/nrg1489>.
6. Barcellos, L. F., May, S. L., Ramsay, P. P., Quach, H. L., Lane, J. A., Nititham, J., Noble, J. A., Taylor, K. E., Quach, D. L., Chung, S. A., Kelly, J. A., Moser, K. L., Behrens, T. W., Seldin, M. F., Thomson, G., Harley, J. B., Gaffney, P. M., & Criswell, L. A. (2009). High-density SNP screening of the major histocompatibility complex in systemic lupus erythematosus demonstrates strong evidence for independent susceptibility regions. *PLoS genetics*, 5(10), e1000696. <https://doi.org/10.1371/journal.pgen.1000696>.
7. Hirota, T., Takahashi, A., Kubo, M., Tsunoda, T., Tomita, K., Doi, S., Fujita, K., Miyatake, A., Enomoto, T., Miyagawa, T., Adachi, M., Tanaka, H., Niimi, A., Matsumoto, H., Ito, I., Masuko, H., Sakamoto, T., Hizawa, N., Taniguchi, M., Lima, J. J., ... Tamari, M. (2011). Genome-wide association study identifies three new susceptibility loci for adult asthma in the Japanese population. *Nature genetics*, 43(9), 893–896. <https://doi.org/10.1038/ng.887>.
8. Kamatani, Y., Matsuda, K., Okada, Y., Kubo, M., Hosono, N., Daigo, Y., Nakamura, Y., & Kamatani, N. (2010). Genome-wide association study of hematological and biochemical traits in a Japanese population. *Nature genetics*, 42(3), 210–215. <https://doi.org/10.1038/ng.531>
9. Urayama, K. Y., Jarrett, R. F., Hjalgrim, H., Diepstra, A., Kamatani, Y., Chabrier, A., Gaborieau, V., Boland, A., Nieters, A., Becker, N., Foretova, L., Benavente, Y., Maynadié, M., Staines, A., Shield, L., Lake, A., Montgomery, D., Taylor, M., Smedby, K. E., Amini, R. M., ... McKay, J. D. (2012). Genome-wide association study of classical Hodgkin lymphoma and Epstein-Barr virus status-defined subgroups. *Journal of the National Cancer Institute*, 104(3), 240–253. <https://doi.org/10.1093/jnci/djr516>.
10. Klitz W, Gragert L, Trachtenberg E. Spectrum of HLA associations: the case of medically refractory pediatric acute lymphoblastic leukemia. *Immunogenetics*. 2012 Jun;64(6):409-419. DOI: 10.1007/s00251-012-0605-5.
11. Taylor GM, Dearden S, Ravetto P, Ayres M, Watson P et al. (2002) Genetic susceptibility to childhood common acute lymphoblastic leukaemia is associated with polymorphic peptide-binding pocket profiles in HLA-DPB1\*0201. *Hum Mol Genet* 11: 1585-1597. doi:10.1093/hmg/11.14.1585. PubMed: 12075003.
12. Taylor GM, Richards S, Wade R, Hussain A, Simpson J et al. (2009) Relationship between HLA-DP supertype and survival in childhood acute lymphoblastic leukaemia: evidence for selective loss of immunological control of residual disease? *Br J Haematol* 145: 87-95. doi:10.1111/j.1365-2141.2008.07571.x. PubMed: 19183185.

13. Urayama KY, Chokkalingam AP, Metayer C, Ma X, Selvin S et al. (2012) HLA-DP genetic variation, proxies for early life immune modulation, and childhood acute lymphoblastic leukemia risk. *Blood* 120: 3039-3047. doi:10.1182/blood-2012-01-404723. PubMed:22923493.
14. Hosking, F. J., Leslie, S., Dilthey, A., Moutsianas, L., Wang, Y., Dobbins, S. E., Papaemmanuil, E., Sheridan, E., Kinsey, S. E., Lightfoot, T., Roman, E., Irving, J. A., Allan, J. M., Taylor, M., Greaves, M., McVean, G., & Houlston, R. S. (2011). MHC variation and risk of childhood B-cell precursor acute lymphoblastic leukemia. *Blood*, 117(5), 1633–1640. <https://doi.org/10.1182/blood-2010-08-301598>
15. Klitz W, Gragert L, Trachtenberg E (2012) Spectrum of HLA associations: the case of medically refractory pediatric acute lymphoblastic leukemia. *Immunogenetics* 64: 409-419. doi:10.1007/s00251-012-0605-5. PubMed: 22350167.
16. Urayama, K. Y., Takagi, M., Kawaguchi, T., Matsuo, K., Tanaka, Y., Ayukawa, Y., Arakawa, Y., Hasegawa, D., Yuza, Y., Kaneko, T., Noguchi, Y., Taneyama, Y., Ota, S., Inukai, T., Yanagimachi, M., Keino, D., Koike, K., Toyama, D., Nakazawa, Y., Kurosawa, H., ... Manabe, A. (2018). Regional evaluation of childhood acute lymphoblastic leukemia genetic susceptibility loci among Japanese. *Scientific reports*, 8(1), 789. <https://doi.org/10.1038/s41598-017-19127-7>
17. Ishida, Y., Maeda, M., Urayama, K. Y., Kiyotani, C., Aoki, Y., Kato, Y., Goto, S., Sakaguchi, S., Sugita, K., Tokuyama, M., Nakadate, N., Ishii, E., Tsuchida, M., Ohara, A., & QOL committee of Tokyo Children's Cancer Study Group (TCCSG) (2014). Secondary cancers among children with acute lymphoblastic leukaemia treated by the Tokyo Children's Cancer Study Group protocols: a retrospective cohort study. *British journal of haematology*, 164(1), 101–112. <https://doi.org/10.1111/bjh.12602>
18. Muro, S. et al. Relationship Among Chlamydia and Mycoplasma Pneumoniae Seropositivity, IKZF1 Genotype and Chronic Obstructive Pulmonary Disease in A General Japanese Population: The Nagahama Study. *Medicine* 95, e3371, <https://doi.org/10.1097/MD.0000000000003371> (2016).
19. Seow, W. J. et al. Association between GWAS-identified lung adenocarcinoma susceptibility loci and EGFR mutations in never-smoking Asian women, and comparison with findings from Western populations. *Hum Mol Genet* 26, 454-465, <https://doi.org/10.1093/hmg/ddw414> (2017).
20. Izuhara, Y. et al. Mouth breathing, another risk factor for asthma: the Nagahama Study. *Allergy* 71, 1031-1036, <https://doi.org/10.1111/all.12885> (2016).
21. Barrett, J. C., Fry, B., Maller, J., & Daly, M. J. (2005). Haploview: analysis and visualization of LD and haplotype maps. *Bioinformatics (Oxford, England)*, 21(2), 263–265. <https://doi.org/10.1093/bioinformatics/bth457>
22. Pruim, R. J., Welch, R. P., Sanna, S., Teslovich, T. M., Chines, P. S., Gliedt, T. P., Boehnke, M., Abecasis, G. R., & Willer, C. J. (2010). LocusZoom: regional visualization of genome-wide association scan results. *Bioinformatics (Oxford, England)*, 26(18), 2336–2337. <https://doi.org/10.1093/bioinformatics/btq419>
23. Purcell, S., Neale, B., Todd-Brown, K., Thomas, L., Ferreira, M. A., Bender, D., Maller, J., Sklar, P., de Bakker, P. I., Daly, M. J., & Sham, P. C. (2007). PLINK: a tool set for whole-genome association and population-based linkage analyses. *American journal of human genetics*, 81(3), 559–575. <https://doi.org/10.1086/519795>
24. Krishnan, R., Boddapati, N., & Mahalingam, S. (2018). Interplay between human nucleolar GNL1 and RPS20 is critical to modulate cell proliferation. *Scientific reports*, 8(1), 11421. <https://doi.org/10.1038/s41598-018-29802-y>
25. Krishnan, R., Murugiah, M., Lakshmi, N. P., & Mahalingam, S. (2020). Guanine nucleotide binding protein like-1 (GNL1) promotes cancer cell proliferation and survival through

- AKT/p21 CIP1 signaling cascade. *Molecular biology of the cell*, 31(26), 2904–2919.  
<https://doi.org/10.1091/mbc.E20-04-0267>
26. Yusuf, N., Hidalgo, B., Irvin, M. R., Sha, J., Zhi, D., Tiwari, H. K., Absher, D., Arnett, D. K., & Aslibekyan, S. W. (2017). An epigenome-wide association study of inflammatory response to fenofibrate in the Genetics of Lipid Lowering Drugs and Diet Network. *Pharmacogenomics*, 18(14), 1333–1341. <https://doi.org/10.2217/pgs-2017-0037>
  27. T., Liu, T., Hu, X., Song, J., & Ying, B. (2020). A Notch4 missense mutation is associated with susceptibility to tuberculosis in Chinese population. *Infection, genetics and evolution: journal of molecular epidemiology and evolutionary genetics in infectious diseases*, 78, 104145. <https://doi.org/10.1016/j.meegid.2019.104145>
  28. Takam Kanga, P., Dal Collo, G., Midolo, M., Adamo, A., Delfino, P., Mercuri, A., Cesaro, S., Mimiola, E., Bonifacio, M., Andreini, A., Chilosi, M., & Krampera, M. (2019). Inhibition of Notch Signaling Enhances Chemosensitivity in B-cell Precursor Acute Lymphoblastic Leukemia. *Cancer research*, 79(3), 639–649. <https://doi.org/10.1158/0008-5472.CAN-18-1617>
  29. Takam Kanga, P., Dal Collo, G., Resci, F., Bazzoni, R., Mercuri, A., Quaglia, F. M., Tanasi, I., Delfino, P., Visco, C., Bonifacio, M., & Krampera, M. (2019). Notch Signaling Molecules as Prognostic Biomarkers for Acute Myeloid Leukemia. *Cancers*, 11(12), 1958. <https://doi.org/10.3390/cancers11121958>
  30. Ucisik-Akkaya, E., Davis, C. F., Gorodezky, C., Alaez, C., & Dorak, M. T. (2010). HLA complex-linked heat shock protein genes and childhood acute lymphoblastic leukemia susceptibility. *Cell stress & chaperones*, 15(5), 475–485. <https://doi.org/10.1007/s12192-009-0161-6>
  31. Zong, J., Lin, Y., Tian, Q., Zhao, X., Chu, K., Jiang, B., Li, S., Sun, G., & Liu, S. (2020). HSPA1L rs1061581 polymorphism is associated with the risk of preeclampsia in Han Chinese women. *Bioscience reports*, 40(2), BSR20194307. <https://doi.org/10.1042/BSR20194307>
  32. De Andrade, W. P., Da Conceição Braga, L., Gonçalves, N. G., Silva, L. M., & Da Silva Filho, A. L. (2020). HSPA1A, HSPA1L and TRAP1 heat shock genes may be associated with prognosis in ovarian epithelial cancer. *Oncology letters*, 19(1), 359–367. <https://doi.org/10.3892/ol.2019.11095>
  33. Weng, A. P., Ferrando, A. A., Lee, W., Morris, J. P., 4th, Silverman, L. B., Sanchez-Irizarry, C., Blacklow, S. C., Look, A. T., & Aster, J. C. (2004). Activating mutations of NOTCH1 in human T cell acute lymphoblastic leukemia. *Science (New York, N.Y.)*, 306(5694), 269–271. <https://doi.org/10.1126/science.1102160>