YAO LEI

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EDUCATION

Dec 2022 – Now	Ph.D.	The University of Hong Kong Systemic genomic study on systemic lupus erythematosus
Oct 2020 – Dec 2022	M.Phil.	The University of Hong Kong Casual mutation diagnosis for Neuromuscular disorders
Sep 2015 – Jul 2019	B.S.	Nankai University Major in Biotechnology

PUBLICATIONS

1. Cw Chan, S.#, **Lei, Y.#**, Yh Yap, D., Pw Lee, P., Lai, W. M., Ky Ying, S., Mh Leung, A., Mok, C. C., Lee, K. L., Lau, C. S., Yang, W., & Li, P. H. (2025). Distinct genetic risk loci between biopsy-proven renal and non-renal lupus: a 10-year longitudinal cohort. ***Rheumatology (Oxford, England)***, keaf027. Advance online publication. https://doi.org/10.1093/rheumatology/keaf027

2. Wang, F. Q.#, Dang, X., Su, H., **Lei, Y.**, She, C. H., Zhang, C., Chen, X., Yang, X., Yang, J., Feng, H., & Yang, W. (2024). Association of hyperactivated transposon expression with exacerbated immune activation in systemic lupus erythematosus. ***Mobile DNA***, 15(1), 23. https://doi.org/10.1186/s13100-024-00335-8

3. Liu, Z.#, Shao, L.#, Hou, F.#, Li, W.#, Wang, Y. F., Feng, H., Wang, F. Q., **Lei, Y.**, Zheng, L., Liang, R., Li, J., Guo, X., Zhang, L., Zhang, Y., Yang, J., Qin, X., Wei, W., Yang, X., Dang, X., Ma, W., … Yang, W. (2024). Transcriptomic features of systemic lupus erythematosus patients in flare and changes during acute in-hospital treatment. ***Rheumatology (Oxford, England)***, 63(10), 2810–2818. https://doi.org/10.1093/rheumatology/kead704

4. Tangtanatakul, P#., **Lei, Y#.**, Jaiwan, K., Yang, W., Boonbangyang, M., Kunhapan, P., Sodsai, P., Mahasirimongkol, S., Pisitkun, P., Yang, Y., Eu-Ahsunthornwattana, J., Aekplakorn, W., Jinawath, N., Neelapaichit, N., Hirankarn, N., & Wang, Y. F. (2024). Association of genetic variation on X chromosome with

systemic lupus erythematosus in both Thai and Chinese populations. ***Lupus science & medicine***, 11(1), e001061. https://doi.org/10.1136/lupus-2023-001061

5. Zhang, Y.#, Xu, L.#, **Lei, Y.#**, Chan, S. H. S., & Javed, A. (2022). VP.03 Diagnosing Titinopathy: Lessons from a multi-omics pilot study. ***Neuromuscular Disorders***, 32, S47–S48. https://doi.org/10.1016/j.nmd.2022.07.028

6. Wang, Y. F., Wei, W., Tangtanatakul, P., Zheng, L., **Lei, Y.**, Lin, Z., Qian, C., Qin, X., Hou, F., Zhang, X., Shao, L., Satproedprai, N., Mahasirimongkol, S., Pisitkun, P., Song, Q., Lau, Y. L., Zhang, Y., Hirankarn, N., & Yang, W. (2022). Identification of Shared and Asian-Specific Loci for Systemic Lupus Erythematosus and Evidence for Roles of Type III Interferon Signaling and Lysosomal Function in the Disease: A Multi-Ancestral Genome- Wide Association Study. ***Arthritis & rheumatology (Hoboken, N.J.)***, 74(5), 840–848. https://doi.org/10.1002/art.42021

7. Song, Q#., **Lei, Y#.**, Shao, L., Li, W., Kong, Q., Lin, Z., Qin, X., Wei, W., Hou, F., Li, J., Guo, X., Mao, Y., Cao, Y., Liu, Z., Zheng, L., Liang, R., Jiang, Y., Liu, Y., Zhang, L., Yang, J., ... Yang, W. (2021). Genome-wide association study on Northern Chinese identifies KLF2, DOT1L and STAB2 associated with systemic lupus erythematosus. ***Rheumatology (Oxford, England)***, 60(9), 4407–4417. https://doi.org/10.1093/rheumatology/keab016

8. Wang, Y. F#., Zhang, Y., Lin, Z., Zhang, H., Wang, T. Y., Cao, Y., Morris, D. L., Sheng, Y., Yin, X., Zhong, S. L., Gu, X., **Lei, Y.**, He, J., Wu, Q., Shen, J. J., Yang, J., Lam, T. H., Lin, J. H., Mai, Z. M., Guo, M., ... Yang, W. (2021). Identification of 38 novel loci for systemic lupus erythematosus and genetic heterogeneity between ancestral groups. ***Nature communications***, 12(1), 772. https://doi.org/10.1038/s41467-021-21049-y

SKILLS

Data Analysis	(1) Genome-wide association study
	(2) Whole-genome sequencing variant alignment, calling, and analysis
	(3) Single-cell RNA & single-cell ATAC sequencing data analysis
Coding	(1) R, Python, and C++ programming
	(2) Linux system construction and maintenance
	(3) Experience in high-performance computing environments & cloud-based servers

Theory	(1) Human genetics, transcriptomics, epigenomics
	(2) Statistics, Probability
	(3) Machine Learning, deep learning

POSTER PRESENTATIONS

2024 ASHG conference	Whole-genome sequencing identifies *SIKE1*, *CD36*, and *IFFO1* as candidate susceptible genes for systemic lupus erythematosus (SLE)
2023 ASHG conference	Phenotype-based prioritization strategy accelerates the identification of real causal variants in patients with Titinopathy
2022 AOMC conference	Ranking variant pathogenicity using Exomiser facilitated the identification of the missing second mutation in three recessive cases of congenital myopathy
2021 AOMC conference	*TTN* mutations in Limb-girdle muscular dystrophy with calpainopathy-like presentation in 2 siblings

RESEARCH EXPERIENCE

Ph.D. student

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong

- SNP array-based genome-wide association study on SLE:
  - Distinct genetic risk loci between biopsy-proven renal and non-renal lupus: a 10-year longitudinal cohort. (Oxford Rheumatology)
  - Association of genetic variation on X chromosome with systemic lupus erythematosus in both Thai and Chinese populations. (BMJ lupus science & Medicine)

- Whole-genome sequencing on SLE (on-going project):
  - Multiple GWAS studies have been performed and published on SLE, while these studies are focused on common variants only. In this study, we conducted large-scale whole-genome sequencing on our 1,500

SLE samples and collected WGS for 12,000 healthy control samples, to try to identify rare variants that enriched in genes, regulatory regions, or conservative regions in SLE.

M.Phil. student

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong

Due to the clinical and genetic heterogeneity of muscle diseases, mutations in *TTN* are often ignored. To improve the diagnostic rate, we collected patients who had been clinically diagnosed with muscle disease but no confirmed causal mutations identified, and performed whole-genome sequencing (WGS). Using the latest phenotype-based prioritization strategies, we identified candidate causal variants in the *TTN* gene for approximately 40% of these patients.

Research Intern

Department of Paediatrics and Adolescent Medicine, The University of Hong Kong

In this study, we collected SNP array data form 522 SLE and 1,017 controls samples from Jining, Shandong Province, China; and performed genome-wide association study (GWAS) followed-by a meta-analysis that integrated the in-house Chinese and European SLE cohorts. Thought this analysis, we identified *KLF2*, *DOT1L*, and *STAB2* as novel SLE associated loci.

TEACHING EXPERIENCE

Dec 2023 Teaching Fellow: Bioinformatics and PhD Experience Workshop for Undergraduates

Jun 2023 Lecturer: Undergraduate Introduction to Bioinformatics and Doctoral Research

AWARDS AND HONORS

Jun 2022 1st Place of Best e-Poster: Ranking variant pathogenicity using Exomiser facilitated the identification of the missing second mutation in three recessive cases of congenital myopathy (2022). The 20th Annual Scientific Meeting of Asian Oceanian Myology Center

Jun 2021

Best Poster Presentation: TTN mutations in limb girdle muscular dystrophy with calpainopathy-like presentation in 2 siblings (2021). The 19th Annual Scientific Meeting of Asian Oceanian Myology Center

May 2016 Third Prize in Basic Experiment Skills Competition of Biochemistry

Oct 2016 Third Prize in Botanical Basic Experiment Skills Competition