Curriculum Vitae (Update: Apr 2025)

Yao LEI

Tel: (+852) 6367 0230 or (+86) 13207513180 | Email: leiyao1997@connect.hku.hk
Address: 1F, Jockey Club Building for Interdisciplinary Research, 5 Sassoon Rd, Hong Kong Island, Hong Kong, China, 999077

EDUCATION

Ph.D. in Bioinformatics	Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong, Hong Kong, China, Dec.2022-Now
M.Phil. in Bioinformatics	Department of Paediatrics and Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong, Hong Kong, China, Oct.2020-Dec.2022
B.S. in Biotechnology	College in Life Science, Nankai University, Tianjin, China, Sep.2015-Jul.2019

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

MAJOR RESEARCH EXPERIENCE

Identification of common and rare variants that associated with systemic lupus erythematosus using whole-genome sequencing. **Role: data analyzer**

Advisor: Dec.2023-Now

Prof. Wanling Yang	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Clara Tang	*LKS faculty of Medicine, The University of Hong Kong*
Prof. Brian Chung	*Hong Kong genome institute, Hong Kong.*
Prof. Bo Ban	*Affiliated Hospital of Jining Medical University, Jining, China.*
Prof. Huijun yuan	*West China Hospital of Sichuan University, Chengdu, China.*

- Systemic lupus erythematosus (SLE) is a complex autoimmune disease. Numerous genome-wide association studies (GWAS) have been conducted, and more than 300 common loci have been reported to be associated with SLE. However, these loci explain only a limited proportion of the disease's heritability. The goal of this study is to uncover the missing heritability of SLE. In collaboration with the Hong Kong Genome Institute, West China Hospital of Sichuan University, and Jining Medical University, we collected whole-genome sequencing (WGS) data from 2,000 SLE patients and 40,000 control individuals. Through this study, we aim to identify common and rare genetic variants and loci for SLE.

Identification of common and rare variants that associated with growth hormone deficiency using whole-genome sequencing. **Role: data analyzer & project initiator**

Advisor: Dec.2024-Now

Prof. Wanling Yang	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Bo Ban	*Affiliated Hospital of Jining Medical University, Jining, China.*

- Growth hormone deficiency (GHD) is a subtype of human short stature caused by insufficient growth hormone production. Although human height is a complex trait, GHD is often considered a Mendelian disease. In this study, using the our in-house WGS as control, we plan to collect whole-genome sequencing (WGS) data from 1,200 GHD patients and perform genome-wide association

studies (GWAS) and rare variant association studies (RVAS) to identify common and rare variants or genes associated with GHD.

Distinct genetic risk loci between biopsy-proven renal and non-renal lupus: a 10-year longitudinal cohort. (Oxford Rheumatology) **Role: data analyzer & algorithm dev**

Advisor: Dec.2022-Aug.2023

Prof. Wanling Yang	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Bo Ban	*Affiliated Hospital of Jining Medical University, Jining, China.*
Prof. Shirley Chan	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Philip Li	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Chak-sing Lau	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*

- Systemic lupus erythematosus (SLE) is a prototypical autoimmune disease that can affect multiple organ systems. Lupus nephritis (LN), characterized by inflammation of the kidneys, is a severe subtype of SLE. In this study, using 10 years of clinical follow-up data, we classified patients into LN and non-nephritic (NLN) groups. First, we replicated the finding that the LN group has a higher distribution of polygenic risk scores compared to the NLN group. We also identified that loci in *HLA* region, *TNFAIP3*, and *BLK* are significantly associated with the LN group but not in NLN group. Furthermore, using heterogeneity testing, we found that loci in *ELF1*, *OX40*, *DUSP22*, and *TPCN2* differ significantly between the LN and NLN groups.

Association of genetic variation on X chromosome with systemic lupus erythematosus in both Thai and Chinese populations. (BMJ lupus science & Medicine) **Role: data analyzer**

Advisor: Dec.2022-Aug.2023

Prof. Wanling Yang	*LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*
Prof. Yongfei Wang	*The Chinese University of Hong Kong, Shenzhen, China.*
Prof. Pattarin Tangtanatakul	*Chulalongkorn University, Thailand.*

- Genome-wide association study (GWAS) is a well-established method for identifying loci associated with human complex diseases. Several GWAS have been conducted on the X chromosome to discover candidate loci for systemic lupus erythematosus (SLE). However, studies in the Thai population are still lacking. In this study, we collected SNP array data from Thai individuals and performed X chromosome-specific GWAS, combining these data with our in-house Hong Kong cohort. We successfully replicated previously reported loci, including *TEME187-IRAK-MECP2*, *PRPS2*, *TLR7*, and *GPR173*, and identified two novel candidate loci associated with SLE: *LOC389895-SOX3* and *CT83-KLHL13*. Additionally, we confirmed a higher frequency of trisomy X among SLE patients compared to controls in both the Thai and Hong Kong populations.

Phenotype-based prioritization enhances diagnosis rate of patients with Titinopathy. (Manuscript preparing) **Role: data analyzer**

Advisor: Oct.2020-Dec.2023

Prof. Chan Hoi Shan Sophelia *LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*

Prof. Wanling Yang *LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*

- Titinopathy is a Mendelian muscle disease caused by mutations in the *TTN* gene. However, due to the clinical and genetic heterogeneity of muscle diseases, mutations in *TTN* are often overlooked. To improve the diagnostic rate, we collected patients who had been clinically diagnosed with muscle diseases but in whom no confirmed causal mutations had been previously 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.

Genome-wide association study on Northern Chinese identifies *KLF2*, *DOT1L* and *STAB2* associated with systemic lupus erythematosus. (Oxford Rheumatology) **Role: data analyzer**

Advisor: Aug.2018-May.2019

Prof. Wanling Yang *LKS faculty of Medicine, The University of Hong Kong, Hong Kong.*

Prof. Bo Ban *Affiliated Hospital of Jining Medical University, Jining, China.*

- Systemic lupus erythematosus is a complex autoimmune disease. We collected SNP array data form 522 SLE and 1017 controls samples from Jining, Shandong Province, China. Genome-wide association study was performed on these samples, followed-by a meta-analysis that integrated our in-house Chinese and European SLE cohorts. Thought this analysis, we identified *KLF2*, *DOT1L*, and *STAB2* as novel SLE associated loci.

AWARDS

1. 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.2022

2. 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 — Jun.2021

3. Third Prize in Basic Experiment Skills Competition of Biochemistry — May.2016

4. Third Prize in Botanical Basic Experiment Skills Competition — Oct.2016