Xinyue Zhao

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University of Melbourne, master of bioinformatics

EDUCATION

University of Melbourne **Melbourne, Australia**
M. S. in bioinformatics (Science) Jul. 2021-Aug. 2023
- Overall score: 83.1%

Peking Union Medical College **Beijing, China**
M. S. in Medical Genetics Sep. 2018-Jul. 2021
- Overall score: 88.5%, Major score: 94%

School of the Gifted Young, University of Science and Technology of China **Hefei, China**
B. S. in Chemical Biology Sep. 2014-Jul. 2018
- Overall score: 84.9%, Major score: 86.2%
- Admitted to *Experimental Class for the Sciences*, an honors class for top students

CuriousU International Summer School, University of Twente **Enschede, Netherlands**
Summer Student Aug. 2016

RESEARCH EXPERIENCES

Heejung Shim lab, Melbourne Integrative Genomics (MIG), School of Mathematics and Statistics, University of Melbourne **Melbourne, Australia**
Supervisor: Associated Prof. Heejung Shim
Project: Downstream analysis of Nanopore sequencing data Feb. 2022-Jun. 2023

- Investigating the Sequin, SIRV, and biological cDNA sequencing data from Oxford Nanopore technology;
- Using the information on mapping quality and high-quality splice sites from other reads to correct unknown splicing sites in transcripts and identify full-length RNA isoforms;
- Programing via Python and Spartan (HPC), and downstream analysis of output from NanoSplicer, a software developed by Heejung Shim's Lab.

McKusick-Zhang Center for Genetic Medicine, Chinese Academy of Medical Science and Peking Union Medical College **Beijing, China**
Supervisor: Prof. Xue Zhang, Member of the Chinese Academy of Engineering (CAE)
Project 1: Investigating a Novel Pathogenic Mutation in a Chinese Cystic Fibrosis (CF) Patient Sep. 2018-May. 2019

- Detected the potential disease-causing mutations in a Chinese CF patient in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene;
- Identified a novel untypical splicing site variant and a known $TG_{12}T_5$ sequence polymorphism 3bp upstream in *cis*, both of which may affect splicing patterns of *CFTR*;
- Conducted minigene assay, site-directed mutagenesis technique, and T-clone assay *in vitro* and verified the pathogenicity of the novel splicing mutation alone or in combination with the polymorphism.

Project 2: A Clinical and Genetic study of 70 Primary Ciliary Dyskinesia (PCD) individuals Sep. 2018-Jan. 2020

- Performed whole-exome sequencing (WES) and/or whole-genome sequencing (WGS) on 70 suspected PCD individuals of Chinese origin.
- Reported clinical characteristics and genetic spectrum of 26 confirmed PCD patients
- Identified a total of 32 pathogenic mutations scattering among 8 disease-causing genes, this is **the first and largest** single-center study for Chinese adult PCD patients.
- Detected a copy number variant carried by a PCD patient and verified it by qPCR; underlined the potential ability of WES associated with low-pass WGS as a diagnostic tool.

Project 3: Investigation of a New PCD Causative Gene CFAP54 and verifying its pathogenicity May. 2019-Jul. 2021

- Detected a patient carrying compound heterozygous mutations in a suspected new PCD causative gene *CFAP54*.
- Validated the reduction of RNA expression caused by variants in *CFAP54* through qPCR and minigene assay
- Validated infertility, hydrocephalus, mucus accumulation in nasal sinuses, and other PCD-relevant phenotypes in a knock-in mouse model built by CRISPR/Cas9.
- Established an air-liquid-interface cell-culture platform for mouse tracheal epithelium to further detect the beat frequency of respiratory motile cilia.
- Performed high-speed video analysis (HSVA) and measured the beat frequency of mutant and wild-type respiratory cilia.
- Identified the position of Cfap54 protein in a single ciliary cell using confocal immunofluorescence microscopy.

PUBLICATIONS

- **Xinyue Zhao**, Keqiang Liu, Wenshuai Xu, et al. The novel mutation c.1210-3C>G in *cis* with a poly-T tract of 5T affects *CFTR* mRNA splicing in a Chinese patient with cystic fibrosis. *Frontiers of Medicine* (IF: 9.927); PMID: 34302615
- **Xinyue Zhao**, Chun Bian, Keqiang Liu, et. al. Clinical characteristics and genetic spectrum of 26 individuals of Chinese origin with primary ciliary dyskinesia. *Orphanet Journal of Rare Disease* (IF: 4.303); PMID: 34210339
- **Xinyue Zhao**, Haijun Ge, Wenshuai Xu, et. al. Lack of CFAP54 causes primary ciliary dyskinesia in both a mouse model and human patients. Accepted by *Frontiers of Medicine*

AWARDS

- Silver Award for Outstanding Student Scholarship 2014
- Bronze Award for Outstanding Student Scholarship 2016
- Second Award for Postgraduate Scholarship 2020

SKILLS

- **Computer skills**: Python, HPC, C programming language, SPSS, R studio, Photoshop, PyMOL, Origin, Codon Code, Prism, Galaxy, etc.
- **Molecular biology techniques:** DNA cloning, transformation and transfection, mouse sampling and histological analysis, primary cell culture, smear microscopy, (confocal) immunofluorescence microscopy, TEM, HSVA (high-speed video analysis), MLPA, DNA/RNA extraction, qPCR, etc.
- **Chemistry experimental skills**: organic molecules' synthesis, extraction, and purification, chromatography, HPLC, NMR, etc.