

# CURRICULUM VITAE

July 12, 2019



## Jinmeng Jia

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## Objective

A highly motivated individual, trained in both mathematics and biology, with good programming skills and problem solving abilities. Focusing on the causative gene identification and genotype-phenotype correlation analysis for rare diseases with statistical methods and bioinformatics algorithms, clinical data standardization and multi-omics data integration based on text mining.

## Education

**PhD, Tielu Shi's Lab, East China Normal University** **2016.09 – 2019.06**

**Major:** Biochemistry and Molecular Biology

**Main Courses:** Biochemistry and Molecular Biology, Statistics, Molecular Genetics, Population Ecology

**M.S, Tielu Shi's Lab, East China Normal University** **2014.09 – 2016.07**

**Major:** Biomedical Engineering

**Main Courses:** Machine Learning, Bioinformatics, Cellular Signal Transduction, Biostatistics, Mathematical Modeling

## Research Experience

### Investigation of Rare Disease Mechanism based on Multi-omics data integration and Network Analysis

- Established a standardized knowledge base of rare diseases by integrating multi-omics data.
- Supplemented a large number of rare disease-phenotype associations by text-mining nearly 10 million articles in the MEDLINE database.
- Built up gene-based disease network and phenotype based disease network for rare diseases and elaborated the role of disease networks in the study of disease molecular mechanisms, disease gene prediction, and clinical diagnostic assistance.
- In charge of the study design, text-mining, disease network building and manuscript writing.

### Infer protein-protein interaction based on a hierarchical vector space model

- Proposed a Hierarchical Vector Space Model (HVSM) for computing semantic similarity between different genes or their products, which enhances the basic vector space model by introducing the relation between GO terms.
- Introduced the concept of a Certainty Factor to calibrate the semantic similarity based on the number of terms annotated to genes.
- In charge of the study design, data analysis and manuscript writing.

#### **Machine learning system to support phenotype-based rare disease diagnosis**

- Adopted both phenotypic similarity method and machine learning method to build up four diagnostic models to support rare disease diagnosis.
- In charge of the study design, model construction and manuscript writing.

#### **(On-going Project) Guideline for the Minimal Information when reporting a proteomics/metabolomics quality control experiment. (Cooperation Project with EMBL-EBI)**

- Delivered a set of technical guidelines representing the minimal information required to report and sufficiently support assessment and interpretation of a proteomics/metabolomics experiment.
- Built up CV for qcML.
- Leading construction of MIAPE-QC (Manuscript is in preparation now).

#### **(On-going Project) Calculating phenotype similarity based on molecular function.**

- Quantified the similarity of disease phenotype and molecular function based on gene ontology.
- Established a phenotypic network based on molecular function.

### **Skills**

- **Programming languages:** Proficient in R, Python, Shell.
- **Bioinformatics/ Data science:** Expertise in text mining and nature language processing skills. Expertise in the usage of machine learning and deep learning algorithms. Experienced in processing and analysis of array and NGS-based transcriptomic data using standard statistical analysis.

### **Academic Conference Attended**

#### **Poster Presentation**

- April 2015, Beijing, China - The 8th International Biocuration Conference  
**Poster Presentation:** *Standardized metadata for LC-MS/MS Proteomics.*
- April 2016, Geneva, Switzerland - The 9th International Biocuration Conference  
**Poster Presentation:** *Rare disease annotation and Medicine.*

#### **Oral Presentation**

- April 2016, Geneva, Switzerland - The 9th International Biocuration Conference  
**Oral Presentation:** Standardized Multi-omics Annotation for Pediatric disease.
- April 2017, Stanford University, USA - The 10th International Biocuration Conference  
**Oral Presentation:** *eRAM: encyclopedia of rare disease annotations for precision medicine.*
- May 2018, Heidelberg, Germany: The HUPO-PSI Spring Meeting  
**Oral Presentation & Holding Workshop:** *MIAPE-QC: Minimum information guidelines for a*

## Host Workshop

- September **2016**, Taipei, **China** - The 15th Human Proteome Organization World Congress (HUPO 2016)

**Host Workshop (Co-host with Mathias Walzer):** *QCML: an exchange format for quality control metrics from mass spectrometry experiments.*

- September **2017**, Dublin, **Ireland** - The 16th Human Proteome Organization World Congress (HUPO 2017)

**Host Workshop (Co-host with PSI-QC group):** *The Human Proteome Organization-Proteomics Standards Initiative Quality Control Working Group: Making Quality Control More Accessible for Biological Mass Spectrometry*

## Publications

### Published

- Jia, J.**, An, Z., Ming, Y., Guo, Y., Li, W., Liang, Y., Guo, D., Li, X., Tai, J., Chen, G. et al. (2018) eRAM: encyclopedia of rare disease annotations for precision medicine. *Nucleic acids research*, **46**, D937-D943. (PMID: [29106618](#))
- Jia, J.**, An, Z., Ming, Y., Guo, Y., Li, W., Li, X., Liang, Y., Guo, D., Tai, J., Chen, G. et al. (2018) PedAM: a database for Pediatric Disease Annotation and Medicine. *Nucleic acids research*, **46**, D977-D983. (PMID: [29126123](#))
- Jia, J.**, Wang R., An, Z. et al. RDAD: A machine learning system to support phenotype-based rare disease diagnosis. *Frontiers in genetics*, **9**:587 (PMID: [30564269](#))
- Jia, J.** and Shi, T. (2017) Towards efficiency in rare disease research: what is distinctive and important? *Science China. Life sciences*, **60**, 686-691. (PMID: [28639105](#))
- Zhang, J., Jia, K., **Jia, J.** and Qian, Y. (2018) An improved approach to infer protein-protein interaction based on a hierarchical vector space model. *BMC bioinformatics*, **19**, 161. (PMID: [29699476](#))
- (Accept) Fu, Y<sup>#</sup>, **Jia, J<sup>#</sup>**, Yue, L., Yang R., Guo Y., Ni X. and Shi, T. (2018) Systematically analyze the pathogenic variations for Acute Intermittent Porphyria. *Frontiers in pharmacology*. (Co-first author)
- (Accept) Ren, M., Shi, J., **Jia, J.**, Guo Y., Ni X. and Shi, T. (2018) Genotype-phenotype correlations of Berardinelli-Seip congenital lipodystrophy and novel candidate genes prediction. *Frontiers in pharmacology*. (Third author)

### On-going

- (Second round review) Li, S<sup>#</sup>, **Jia, J<sup>#</sup>**, Guo Y., Ni X. and Shi, T. (2018) Systematically analyze the associations between Cystinuria and different pathogenic variations. *Frontiers in pharmacology*. (Co-first author)
- (Peer review) Cheng, G<sup>#</sup>, **Jia, J<sup>#</sup>**, Guo Y., Ni X. and Shi, T. (2018) Multi-level data integration analysis associates potential new pathogenic genes with Treacher Collins syndrome. *Frontiers in Genetics*. (Co-first author)

- (Peer review) Shi, J., Ren, M., **Jia, J.**, Tang, M., Guo Y., Ni X. and Shi, T. (2018) Genotype-Phenotype association analysis reveals new pathogenic factors for Osteogenesis imperfecta disease. *Frontiers in pharmacology*. (Third author)

## **Honors and Awards (*Recent 5 Years*)**

- **Outstanding graduates of Shanghai** - June **2019**, Shanghai, **China**.
- **Outstanding graduates of East China Normal University** - May **2019**, Shanghai, **China**
- **Evaluation of scientific research achievements in research group, First Prize (No. 1)** – February **2019**, Shanghai, **China**
- **National Scholarship Award** - October **2018**, Shanghai, **China**.
- **Excellent-Youngster Researcher** - May **2018**, Heidelberg, **Germany**.  
**The HUPO-PSI Spring Meeting.**  
**Presentation Title:** *MLAPE-QC: Minimum information guidelines for a Quality Control experiment in LC-MS/MS.*
- **The Thirteenth National Graduate Student Mathematical Contest in modeling, First Prize** – October **2017**, Chongqing, **China**.
- **Evaluation of scientific research achievements in research group, First Prize (No. 1)** – February **2017**, Shanghai, **China**
- **Travel Fellowship Award** - April **2016**, Geneva, **Switzerland**.  
**The 9th International Biocuration Conference.**  
**Presentation Title:** *Standardized Multi-omics Annotation for Pediatric disease.*
- **Excellent Student** – December **2015**, Shanghai, **China**.