

Together, we are stronger



Workshop on “Advanced Analysis for Understanding Rare Diseases”

Organizer: East China Normal University, China
Time: February 27th - March 1st, 2018
Venue: Room 534, Life Science Building
East China Normal University Minhang Campus,
500 Dongchuan Road., Shanghai, China

Organizing Committee

Dr, Tielu Shi, Professor - East China Normal University, China
Dr, Leming Shi, Professor - Fudan University, China
Dr, Duan Ma, Professor - Fudan University, China
Dr, Pawel Labaj, Professor - Boku University, Austria
Dr, David Kreil, Professor - Boku University, Austria
Dr Weimin Zhu, Professor - Beijing Proteome Research Center, China

QR Code



PARTNERS & SPONSORS



When there is love, there is hope.

AGENDA

Day 1

Date: 15:30-18:00 p.m. on Tuesday, February 27th, 2018

Time	Event	Speakers
15:30-15:45	Welcome Remark	Dr. Rongming Wang, Vice President of East China Normal University
15:45-16:00	Group Photo for All Participants	
16:00-17:00	Plenary Speech: Rare biology and rare cancers in astronauts	Dr. Christopher Mason, Weill Cornell Medicine
17:00-18:00	Plenary Speech: The personalized medicine plan of Andalusia: the first experience of use of genomic data within a public health system	Dr. Joaquin Dopazo, Foundation Progress and Health, Andalusian Public Health System
18:00-Midnight	Dinner & Fun	

Day 2

Session1: Data Analysis & Interpretation

Session Chair: Dr. David Kreil

Date: 09:00-12:00 on Wednesday, February 28th, 2018

Time	Title/Event	Speakers
09:00-09:45	Methylation profiling in therapy related AML	Dr. Polanska, Silesian University of Technology
09:45-10:15	Application of FDAlabel Database to Advance Rare Disease Study	Dr. Fang Hong, FDA/NCTR
10:15-10:45	Machine Learning for Rare Diseases	Dr. Cesare Furlanello, The Kessler Foundation
10:45-11:15	Determination of local chromatic interactions using a combined CRISPR and peroxidase APEX2 system	Dr. Yun Liu, Fudan University
11:15-11:45	Data interpretation with interactive visualization	Dr. William Zhou, SAS Inc.
11:45-12:00	Clustering of heterogeneous rare disease networks and Predicting disease-gene associations based on low-rank matrix Tri-factorization	Ms. Xia Pan, East China Normal University
12:00-13:30	Lunch	

Session2: Data Sharing & Standardization**Session Chair:** Dr. Leming Shi**Date:** 13:30-17:25 on Wednesday, February 28th, 2018

Time	Title/Event	Speakers
13:30-14:15	Deep study of chronic fatigue syndrome	Dr. Wenzhong Xiao, Harvard Medical School
14:15-14:45	Rare epigenetic diseases	Dr. Duan Ma, Fudan University
14:45-15:15	Biobank as the resource for rare disease study at Beijing Children's Hospital	Dr. Yongli Guo, Beijing Children's Hospital
15:15-15:45	Application of Genome Analysis Strategies in the Clinical Testing for Pediatric Diseases	Dr. Yaqiong Jin, Beijing Children Hospital.
15:45-15:55	Coffee break	
15:55-16:25	Crowd sourcing science to advance integrated analysis of Neuroblastoma	Dr. David Kreil, Mr. Maciej Kandula, Boku University
16:25-16:55	Single amino acid repeats in rare diseases and beyond	Dr. Pawel Labaj, Boku University
16:55-17:10	Phenome-based rare disease annotation and functional analysis	Ms. Jinmeng Jia, East China Normal University
17:10-17:25	Disease standardization platform	Ms. Zhihan Li, East China Normal University
17:25- Midnight	Dinner & Fun	

Day 3**Session3: Technology Advances in Rare Disease Diagnosis****Session Chair:** Dr. Wenzhong Xiao**Date:** 09:00-12:15 on Thursday, March 1st, 2018

Time	Title/Event	Speakers
09:00-09:45	Genetic tests for inherited diseases with high sensitivity	Dr. Yongyong Shi, Shanghai Jiaotong University
09:45-10:15	Global Genomic Data Platform Empowering Rare Disease Diagnosis and Clinical Research	Dr. Hongye Sun, WuXi NextCODE Genomics
10:15-10:45	Application of gene capture technologies in rare disease diagnosis	Dr. Jian Wu, MyGenostics Inc,
10:45-11:15	Comprehensive application of multiple technologies to hunt the genetic culprit.	Dr. Qian Zhao, SinoPath Diagnostics
11:15-11:45	The application of third generation sequencing technologies in rare disease diagnosis.	Dr. Depeng Wang, Grand Omics
11:45-12:00	RDAD: A Phenotype Based Rare Disease Auxiliary Diagnosis System	Mr. Zhongxin An, East China Normal University
12:00-12:15	Rdpa: A Phenotype-Disease Prediction Algorithm Based On Text	Mr. Xin Li, East China Normal University
12:15-13:30	Lunch	

Session 4: Clinical Applications in Rare Disease Diagnosis

Session Chair: Dr. Duan Ma

Date: 13:30-17:00 on Thursday, March 1st, 2018

Time	Title/Event	Speakers
13:30-14:15	TBX6 gene dosage-dependent phenotypic variability in both congenital renal and vertebral anomalies	Dr. Feng Zhang, Fudan University
14:15-14:45	In silico Drug Repositioning for Rare Disease at FDA/NCTR	Dr. Zhichao Liu, FDA/NCTR
14:45-15:15	Case Discussion – what we learned from a prenatal case	Dr. Luming Sun, Shanghai First Maternity and Infant Hospital
15:15-15:25	Coffee break	
15:25-15:55	Clinical application of next generation sequencing for rare diseases	Dr. Jian Wang, Shanghai Children's Hospital
15:55-16:15	Application of Chinese Gene Mutation Database for disease causative site detection in pedigree	Dr. Changsheng Liu, Virgilbio Inc
16:15-16:30	A gene and phenotype guided drug repositioning through collaborative filtering model for rare diseases	Ms. Yue Ming, East China Normal University
16:30-16:45	Modular-based mapping between HPO and MPO provides new insights into phenotypic mechanisms	Mr. Yunxiang Liang, East China Normal University
16:45-17:00	Phenotypic insights on the relation between adverse drug reaction and disease phenotype	Mr. Dongming Guo, East China Normal University

Participants (in alphabetical order):

Dr. Geng Chen

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Dr. Joaquin Dopazo

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Dr. Duan Ma

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Dr. Jian Wang
Professor, Director of Molecular Diagnostic Laboratory; Vice Director of Department of
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会议须知/Notice

会议须知

一、 注意事项

1. 请各位参会嘉宾带好会议胸牌，凭胸牌进入会场。
2. 会议开始请将手机铃声调为静音，避免影响会议的正常进行。

二、 会议信息

1. 地址：华东师范大学闵行校区(上海市闵行区东川路 500 号)
生命科学学院 534 报告厅
2. 午餐：华东师范大学闵行校区夏雨厅一楼 12:00-13:30 （凭餐券入场）
3. 酒店地址：沪华国际大酒店（上海市闵行区剑川路 368 号）

NOTICE

● Attention

1. Please carry out your conference badge during the conference.
2. Please ring the phone to mute during the conference.

● Information

1. Venue: Room 534, Life Science Building, East China Normal University
Minhang Campus
2. Lunch: 12:00-13:30, First Floor, Xiayu Cafeteria, East China Normal
University Minhang Campus
3. Hotel : Huhua International Hotel, 368 Jianchuan Rd., Shanghai, China