SESSION LECTURE

No.29

Diagnosis and Therapy of Rare Diseases

Room: 405

Co-Chairs: Xue Zhang

Bill Newman

Day 2 October 28th (Sunday) 13:30 – 17:00		
Time	**Speaker**	**Title**
13:30-14:00	**Xue Zhang** *McKusick-Zhang Center for Genetic Medicine, Peking Union Medical College, China*	TBD
14:00-14:30	**Bill Newman** *Manchester Genomic Medicine Centre, UK*	Determining the contribution of noncoding variation to monogenic disorders
14:30-15:00	**Yiping Shen** *Harvard Medical School, USA*	The genetic etiologies of short staturein Chinese patients and their roles in determining treatment options
15:00-15:30	**Tea Break**	
15:30-16:00	**Muhammad Ansar** *Quaid-I-Azam University, Pakistan*	Diagnosis of Inherited Rare Disorders by Exome Sequencing in the Consanguineous Families: Lessons and Challenges
16:00-16:30	**Desheng Liang** *Central South University, China*	Targeted Gene and Cell Therapy for Monogenic diseases
16:30-17:00	**Shihui Yu** *KingMed Diagnostics, China*	An Overview of 121 Kinds of Rare Diseases with 30,000 Positive Cases Identified in a Diagnostic Laboratory in China

Xue Zhang

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Prof.Zhang 's research focuses on the molecular basis of rare Mendelian disorders. He has discovered the disease genes underlying several monogenic disorders, including familial acne inversa, Marie Unna hereditary hypotrichosis, familial idiopathic basal ganglia calcification and syndactyly type V, and identified the pathogenic genomic rearrange ments for congenital generalized hypertrichosis terminalis, X-linked congenital hypertrichosis syndrome and syndactyly type IV.

Yiping Shen

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Prof.Shen 's research interests include understanding the genetic basis of human development which involves physical development (short stature as a disease model) and behavioral and cognitive development (autism and intellectual disability as disease models). He is passionate in helping setting up principles and standards for proper use of genomic technologies for the purposes of effective prevention of severe genetic diseases and birth defects in China.

Desheng Liang

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professor and medical geneticist at the Center for Medical Genetics Central South University. He identified the rDNA locus as a “safe harbor” and developed a universal vector for targeted gene therapy using autologous stem cells. His research also involves in genome editing therapy for monogenic diseases. Dr. Liang serves as vice president of the Chinese Medical Geneticist Association and executive board member the East Asia Union of Human Genetics Societies.

Bill Newman

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He studied medicine at Manchester University and trained in internal medicine and clinical genetics in Manchester and Toronto, Canada. His main research has been in the application on novel genomic technologies in clinical practice. He has used next generation sequencing to define the basis of 20 rare disorders and has more recently defined non-coding variation underpinning rare disease. He is the Director of the Greater Manchester Genomic Medicine Centre and chief investigator on a pharmacogenetics study using point of care genetic testing in neonates.

Shihui Yu

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Chief scientist of KingMed Diagnostics, China. His research focuses on establishing clinical tests for genetic/genomic abnormalities and for early diagnoses of cancers. His group is also working on a project related to rare disease in China and has accumulated more than 60,000 positive cases of rare diseases.

Muhammad Ansar

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Professor of Human Genetics in the Biochemistry Department, Quaid-IAzam University, Pakistan. The aim of his research is to understand the genetic basis of various types of rare inherited disorders but currently his lab is focusing on intellectual disability and retinal dystrophies.