

## INTRODUCTION TO PRECISION MEDICINE

Columbia University Medical Center, Institute for Genomic Medicine

Wednesday, October 24, 2018

4:30 PM–6:30 PM

### INTRODUCTION

► **Welcome and Precision Medicine Overview**

Dr. Vimla Aggarwal, Director of Diagnostic Genomics, Institute for Genomic Medicine

### WORKSHOPS

**1. Curation of Genetic Variants**

**Anya Revah-Politi, MS, Certified Genetic Counselor**

Learn about genetic variation and human disease. Review real clinical cases with genomic data, and complete research to determine the cause of disease in the cases.

**2. Family History and Inheritance**

**Jessica Giordano, MS, Certified Genetic Counselor**

Review how to draw pedigrees and discuss inheritance patterns. Using real clinical cases, draw a pedigree, and identify the inheritance pattern of disease in the family history.

**3. Ethics and Genetics**

**Amanda Bergner, MS, Certified Genetic Counselor**

Using information from an overview of ethical concepts, gather in small groups to discuss cases with a strong ethical component.

### CAREERS IN GENETICS

Meet genetics professionals from Columbia University who will give brief overviews of their path to working in genetics and their current roles.

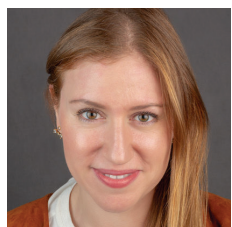
WORLD SCIENCE ACADEMY PARTNERS

## PARTICIPANTS



### **DR. VIMLA AGGARWAL | GENETICIST**

Dr. Vimla Aggarwal is the Director of Diagnostic Genomics at the Institute for Genomic Medicine. She received her medical degree from the University of Mumbai and then specialized in clinical molecular genetics and clinical cytogenetics. Her main interest is the identification of the molecular basis of genetically complex disorders (like autism) and the use of new technologies to diagnose and better take care of patients.



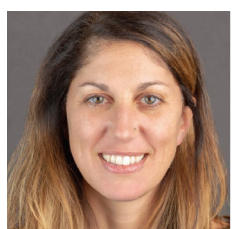
### **ANYA REVAH-POLITI | GENETIC COUNSELOR**

Anya Revah-Politi, MS, CGC, is a genetic counselor at the Institute for Genomic Medicine at Columbia University Medical Center. Her roles include enrolling patients into the precision medicine research studies, variant curation, and presentation and publication of results. She has over ten years of experience in the field of clinical and research genetic counseling.



### **JESSICA GIORDANO | GENETIC COUNSELOR**

Jessica Giordano, MS, CGC, has ten years of experience in clinical prenatal genetic counseling. She manages the fetal anomaly whole exome sequencing project and has experience working for a genetic testing start up. Originally from New York, she studied biology at Boston College and completed her MS in Human Genetics at Sarah Lawrence College.



### **AMANDA BERGNER | GENETIC COUNSELOR**

Amanda Bergner, MS, CGC, is the Program Director of the Genetic Counseling Graduate Program at Columbia. She has 19 years of experience in clinical care, education, industry work, and research. She has a biopsychosocial orientation to her practice, and believes strongly in the therapeutic potential of genetic counseling, and the capacity of the profession to support people engaging with questions about how genetic and genomic medicine impacts them as individuals, families, communities, and society.



### **ELAINE PEREIRA, MD | PEDIATRICIAN WITH A SPECIALTY IN GENETICS**

Dr. Pereira, a native New Yorker, completed her residency in pediatrics and medical genetics at the Children's Hospital at Montefiore. In her current role at NYP/CUMC, Dr. Pereira works with the clinical team as well as the DISCOVER program. Her areas of interest within genetics include collagenopathies (Marfan syndrome, osteogenesis imperfecta) and congenital hearing loss.



### **BRYNN LEVY | PATHOLOGIST, CELL BIOLOGIST**

Brynn Levy, MSc, PhD, is the Medical Director of the Genetics Laboratory at New York Presbyterian Hospital, Columbia University Medical Center, as well as a Professor of Pathology and Cell Biology. Dr. Levy is regarded internationally as an expert in the clinical utility of genomic technologies in reproductive medicine.

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## PARTICIPANTS



### **LOUISE BIER | GENETIC COUNSELOR**

Louise Bier, MS, CGC, is the director of Genetic Counseling and Clinical Engagement at the Institute for Genomic Medicine. She oversees all precision medicine projects at the IGM, and has extensive experience in the design and implementation of clinical genetic research studies.



### **KATE STANLEY | DATA ANALYST**

As a data analyst at the Institute for Genomic Medicine, Kate Stanley works closely with Exome sequencing data and bioinformatics pipelines to understand the genetic architecture of disease. She uses statistical approaches to identify gene-disease associations. She has a B.A. in Neuroscience from Columbia University.

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### **ADDITIONAL PARTICIPANTS IN THE CURATION WORKSHOP**

- Neha Raghavan, PhD
- Jun Liao, PhD
- Christie Buchovecky, PhD
- Michelle Ernst, MS, CGC
- Halie Holmes, MS
- Kate Stanley, BS

### **ADDITIONAL PARTICIPANTS IN THE PEDIGREE WORKSHOP**

- Priyanka Ahimaz, MS, CGC
- Anna Alkelai, PhD
- Meredith Ross, MS, CGC
- Brynn Levy, PhD

### **ADDITIONAL PARTICIPANTS IN THE ETHICS WORKSHOP**

- Elizabeth Rafikian, MS
- Hila Milo Rasouly, PhD
- Sara Berger, MS, CGC
- Volkan Okur, MD