Friday, August 30th @1:00-3:00pm

MOLECULAR PATHOLOGY CASE STUDIES
- CONNECTING THE DOTS BETWEEN GENETICS, MOLECULAR BIOLOGY & BIOCHEMISTRY IN REAL PATIENTS -

HOW YOU NEED TO PREPARE!
- Work with your group on your assigned case study.
- Figure out what is going on with your patient at the molecular level.
- Be prepared to share your patient and his/her molecular pathology with the class!

With recent advances in the integration of various disciplines of molecular science and technological developments in genetic analysis, it is now possible to implement truly “personalized” medicine. The growing adoption of “Precision Medicine” involves the full understanding of a patient, including their own specific molecular pathology and disease etiology, which can help to establish an accurate diagnosis and to select an effective therapy.

NCBI has long had online resources for biologists to explore what is known about a biological molecule including its structure and function, but has recently developed clinically-focused resources enabling scientists and clinicians to integrate known molecular biological information with clinically-relevant genetic variations.

In Wednesday’s Session:
- We will discuss the state of clinical practice with regard to the application of precision medicine principles.
- Together we will explore a real-world case study and follow a workflow to discover the patients’ molecular pathology for an undiagnosed/misdiagnosed problem.
- You will then be given a practice case study to solve, and we will go over the case and how to present your findings in preparation for…..

Before Friday’s Session:
- Your group will be assigned your own case study to explore and discover what is happening in your patient on the molecular level. You will be guided to prepare a summary that you can share with the class.

In Friday’s Session:
- Each groups’ case will be presented to the class so that you can see additional examples of molecular pathology in real patients - and see diversity of molecular pathology even in patients with the same disease or who have pathogenic genetic variants in the same gene.

Facilitator: Rana Morris, PhD - an NCBI Customer Experience team member and Team Lead for Educational Programs (Courses/Workshops, Webinars, Educational Materials). Since 2002, she has provided user support and training, as well as working with supervisors and development teams to improve NCBI resources based on user-centered design principles. Her doctoral, post-doctoral and research fellowship work integrated disciplines of computational and experimental biochemistry, molecular and cellular biology and genetics, and has included diagnostic development, drug design and coordination of genetics/genomics components of clinical trials.