

Number of Participants – 40

Number of Presented Diagnostic Lab Cases – 20, 10 minute presentation + 10 minutes

Q&A/discussion

Sponsors – TBD

*** All Times are listed in Eastern Daylight Time (UTC -4)***

WEDNESDAY, AUGUST 2

- 12:00 – 1:00pm** Welcome and Introductory Videos
- 1:00 – 2:00pm** Faculty Lecture
Genetics and Genomics: Concepts, Nomenclature and Analytical Approaches
Craig Platt, MD
- 2:00 – 3:00pm** Faculty Lecture
Genetics: Analysis Tools
Janet Chou, MD
- 3:00 – 3:30pm** Break
- 3:30 – 4:30pm** Attendee Case Presentations Session #1
Moderator: Rebecca Marsh, MD (3 Cases)
- 4:30 – 5:10pm** Attendee Case Presentations Session #2
Moderator: Janet Chou, MD (2 cases)
- 5:10 – 5:30pm** Genetic Variant Flash Cards
Rebecca Marsh, MD

THURSDAY, AUGUST 3

- 9:00 – 10:00am** Breakfast Small Group Mentoring in Breakout Rooms
How to prepare a concise effective presentation and write an impactful case report.
- Take 2 assigned cases (from previous year) and discuss with faculty support how to condense 10 slides to 2 and how to write an effective case report. Focus on key elements that convey the point you wish to make. Focus on diagnostic aspects and how they contribute to making a diagnosis, selection of treatment and monitoring of therapy. Hone in on novel/meaningful aspects.

- 10:00 – 11:00am** **Attendee Case Presentations Session #3**
Moderator: Roshini Abraham, PhD **(3 Cases)**
- 11:00 – 11:40am** **Attendee Case Presentations Session #4**
Moderator: Vijaya Knight, MD, PhD **(2 Cases)**
- 11:40 – 12:30pm** **Break**
- 12:30 – 1:30pm** **Sponsor Presentations A**
- 1:30 – 2:10pm** **Attendee Case Presentations Moderated Session #5**
Moderator: Sergio Rosenzweig, MD, PhD **(2 Cases)**
- 2:10 – 2:30pm** **Flow Cytometry Flash Cards**
Rebecca Marsh, MD
- 2:30 – 3:00pm** **Break**
- 3:00 – 4:00pm** **Mystery Case in Small Groups**
The Mystery Case incorporates all aspects of diagnostic testing – flow/cellular analysis, biomarker/non-genetic, non-flow studies, genetic analysis. Small groups x 4. Some faculty will be assigned to supervise and support each group.
- 4:00 – 4:30pm** **Large Group Mystery Case**
The groups rejoin and present their solutions to the patient diagnosis to everyone.

FRIDAY, AUGUST 4

9:00 – 9:30am **Breakfast Small Group Mentoring in Break Out Rooms**

9:30 – 10:30am **Didactic Buffet Session One**
Attendees select one of the following breakout rooms:

Room 1:

HLH Diagnostics –Rebecca Marsh, MD

SCID/CID Diagnostics –James Verbsky, MD, PhD

Room 2:

DNA Repair Defects –Roshini Abraham, PhD

Complement Diagnostics –Vijaya Knight, MD, PhD

- 10:30 – 10:40am:** Break
- 10:40 – 11:20pm** Attendee Case Presentations Session #6
Moderator: Helene Decaluwe, MD, PhD (2 Cases)
- 10:40 – 11:20pm** Attendee Case Presentations Session #7
Moderator: James Verbsky, MD, PhD (2 Cases)
- 12:00 – 1:00pm** Break
- 1:00 – 1:30pm** Sponsor Presentations Group B
- 1:30 – 2:10pm** Attendee Case Presentations Session #8
Moderator: Craig Platt, MD, PhD (2 Cases)
- 2:10 – 2:50pm** Attendee Case Presentations Session #9
Moderator: Lisa Forbes Satter, MD (2 Cases)
- 2:50 – 3:15pm** Break
- 3:15 – 3:45pm** Didactic Buffet Session Two
Attendees select one of the following breakout rooms:
- Room 1:
Signaling Pathway Defects – Sergio Rosenzweig, MD, PhD
- Room 2:
STAT GOF and other PIRD Diagnostics – Lisa Forbes Satter, MD
- 3:45 – 4:15pm** Faculty Lecture
COVID-19 and the Immune System
Helene Decaluwe, MD, PhD
- 4:15 – 4:30pm** Best Abstract Award and Wrap-Up
Roshini Abraham, PhD & Rebecca Marsh, MD