# WORKSHOP
# "Genetic Variant Interpretation for Common Diseases"

**"This activity is a Category 1 - Accredited Group Learning Activity as defined by the Ministry of Public Health's Department of Healthcare Professions – Accreditation Section and is approved for 3.0 hours"**

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| **Activity Title:** Genetic Variant Interpretation for Common Diseases |
| **Activity Code:** A318 |
| **Date:** October 9, 2021 |
| **Venue:** Lecture Hall C, Penrose House (LAS building), Education City |
| **Target Audience:** Physicians |

**Overall Learning Objectives**

At the end of the workshop, the participants will be able to:

- Recognize the basic bioinformatics tools and their exploitation to analyze genetic variation.
- Interpret genetic variations in a clinical context using available databases, resources, and software.

| Time | Speakers and talks | Session learning outcomes |
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| 8:30 – 8:55 | On-site Registration | |
| 8:55 – 9:00 | **Dr. Ayman Al Haj Zen**<br>Assistant Professor of Biological & Biomedical Sciences, College of Health & Life Sciences<br>Hamad Bin Khalifa University | • Welcome and introduction |
| | ***Theory session*** | |
| 9:00-9:45 | **Dr. Patricia Munroe**<br>Professor of Molecular Medicine<br>Queen Mary University of London<br><br>*Title: "**Genome-wide association studies – insights into biological mechanisms and disease risk stratification and prediction**"* | • Describe the genomic variation and the methods used to characterize genetic variants (common and rare variants).<br>• Explain how genetic variants are associated with specific traits and diseases.<br>• List polygenic risk scores and their utility.<br>• Demonstrate clinical applications of using polygenic risk scores. |
| 9:45 – 10:00 | Discussion - Q & A | |
| | ***Hands-on session*** | |
| 10:00-10:45 | **Dr. Borbala Mifsud**<br>Assistant Professor of Genomics and Precision Medicine<br>College of Health and Life Sciences<br>Hamad Bin Khalifa University<br><br>*Title: "**Understanding the effect of variants through genetic databases – Part I**"* | • Recognize the theoretical background to some common bioinformatics methods for variant classification.<br>• Apply Ensembl, a commonly used genome browser for retrieving information on variants.<br>• Define different disease or application specific databases to find information on genetic variants. |
| | 15 min ------ Coffee/Snack break --------- | |
| 11:00-11:45 | *Title: "**Understanding the effect of variants through genetic databases – Part II**"* | |
| 11:45 -12:15 | Discussion - Q & A | |
| 12:15-12:30 | Closing remarks | |

**The Scientific Planning Committee has reviewed all disclosed financial relationships of speakers and moderators, in advance of this CPD activity and has implemented procedures to manage any potential or real conflicts of interest.**