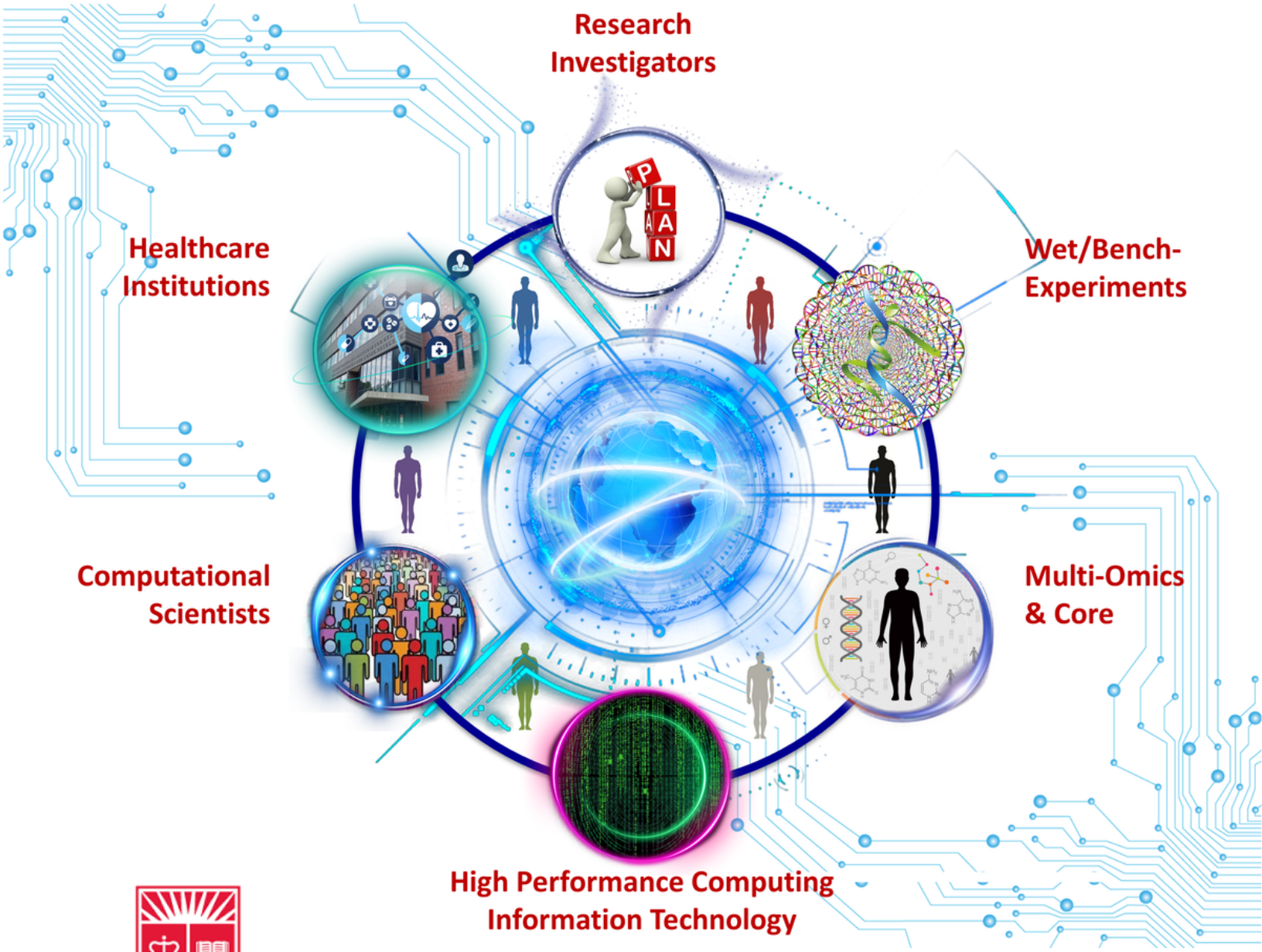
Practicing Precision Medicine with Data Analysis

Course designed and delivered by Zeeshan Ahmed, Ph.D.
Spring 2022 01:090:101 section 11

Editor: Nicole Swenarton



RUTGERS
THE STATE UNIVERSITY OF NEW JERSEY

BYRNE SEMINARS

Institute for Health, Health Care Policy and Aging Research.
Robert Wood Johnson Medical School.

Practicing Precision Medicine with Data Analysis

SPRING 2022 01:090:101 SECTION 11

Background

Precision medicine aims to empower clinicians to predict the most appropriate course of action for patients with complex diseases, and improve routine medical and public health practice. However, practicing precision medicine is not straightforward, as significant efforts are required from the experts in multidisciplinary sciences.

In this course, we have focused on discussing three important areas that heavily contribute to the development of precision medicine initiative: **1**) understanding complexities of Electronic Healthcare Records; **2**) bioinformatics applications for genomics data analysis; and **3**) intelligent and integrative data analysis with machine learning algorithms. Active participation has helped students in learning about operational and academic medical systems, intelligently linking curated clinical data with computationally processed genomic data to identify functional variants among expressed genes, and investigating genotype and phenotype associations.

This course has been grouped with theoretical discussions, basic and life science concepts, and computational skills. Case-studies discussed in this course include peer reviewed findings related to COVID-19, Cardiovascular, Cancer, and other diseases.

Rutgers Institute for Health, Health Care Policy and Aging Research.

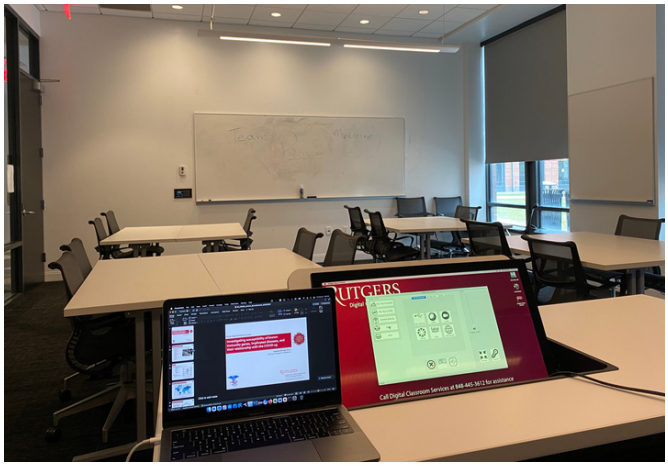
Topics

- Practicing Precision Medicine with Data Analysis.
- Practicing Precision Medicine & Healthcare Data Analysis.
- Practicing Precision Medicine & Genomics Data Analysis.
- Human Gene and Disease Associations for Clinical-Genomics & Precision Medicine.
- Whole genome/exome sequence data processing, management, and gene-variant discovery, annotation, prediction, and genotyping.
- Case Study: Investigating susceptibility of known immunity genes, implicated diseases, and their relationship with the COVID-19.
- Bioinformatics applications for variable gene-disease annotation, visualization, and expression analysis.
- Artificial intelligence for better healthcare, genomics, and precision medicine.
- Multi-omics strategies for personalized and predictive medicine: past, current, and future.
- Role of Clinical-Genomics, Pharmacogenomics, Artificial Intelligence, and Big Data in Precision Medicine Development.

Course overview:

This course was designed with a new research-based approach, where students were introduced to the concepts of modern, basic and medical sciences. This course delivered seminars/lectures based on the critical topics covering various aspects of practicing precision medicine. Students were encouraged to search and learn from the most relevant - peer reviewed scientific literature published in the last five years and available through PubMed Central (PMC) and National Center for Biotechnology Information (NCBI) mainly. **Students conducted their research into four groups: health, genome, data, and AI.**

During the first half of the course, each group focused on learning about healthcare and multi-omics data generation, processing, integration and analysis for precision medicine. Whereas in the second half of the course, students were introduced to complex scientific areas, including different case studies, bioinformatics applications for gene-disease data annotation, and implementing artificial intelligence (AI) and machine learning (ML) algorithms and their applications for better healthcare. Their goal was to understand the role of clinical-genomics, pharmacogenomics, AI/ML, and big data in precision medicine development.



Students were evaluated based on their class attendance, active participation, two major group assignments and final presentations. As a result of the excellent performance of all the participating students in both group assignments, a combined manuscript has been drafted for a future publication. Furthermore, many of the students showed great interest in perusing their academic research career in the field precision medicine at Rutgers.

Course designer and lecturer:

Zeeshan Ahmed, PhD is Assistant Professor of Medicine – Tenure Track and Core Member at the Institute for Health, Health Care Policy and Aging Research; and Department of Medicine – Division of General Internal Medicine, Robert Wood Johnson Medical School, Rutgers Biomedical and Health Sciences. Rutgers, The State University of New Jersey. Dr. Ahmed is focused on dealing with unprecedented challenges in data science and provide better understanding of biology to revolutionize the field of medicine. His research is underpinned with skills and resource development to build expertise in sequence-based genomic analysis, clinical variant interpretation, and evidence-based diagnostic and prediction model development and validation.

Group Health

Healthcare, Claims and Open Access Clinical Data in Precision Medicine. The Group Health weas introduced to different types of healthcare data such as claims data and open access clinical data models like OMAP etc; and their uses when it comes to supporting precision medicine. The students also learned the usage of Electronic Health Record and how physicians use this data to understand more of the patient's history and draw conclusions on future treatment plans. Case studies were presented showing implementation of this data in precision medicine to help train predictive models and increase early prognosis rates. Furthermore, they explored the importance of open access clinical data to advancing research by increasing accuracy rates in assessment and consequential prescription. The Group Health also identified advantages and limitations of claims data such as it needs to be normalized across multiple platforms as no unified pipeline exists to deal with a combination of these types of data.

Group AI

Artificial Intelligence / Machine Learning (AI/ML) in Precision Medicine. Members of Group AI explored the various applications of AI/ML algorithms in precision medicine and their role in multi-omics (genomics, transcriptomics, epigenomics and microbiome) data analysis. They were taught not only the applications of supervised and unsupervised methods of machine learning, but they also discussed the possibilities of AI/ML algorithms in risk assessments, therapy planning and early disease detection leading to better prognosis.

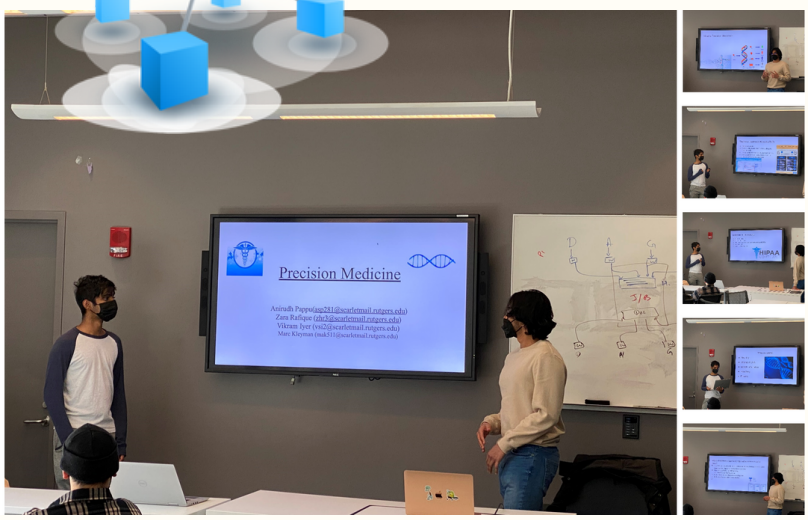
They learned that implementing prediction models can help physicians with not only data processing but also data interpretation and consolidation. Additionally, case studies helped this group identify the advantages and limitations of AI/ML models. This included lack of standardizing data formats and the unavailability of AI/ML ready datasets in the format of a user-friendly graphical interface making this technology hard to implement.

Group Genome

Genomics and Pharmacogenomics in Precision Medicine. The Group Genome learned pharmacogenomics in relation to precision medicine and its involvement in processes such as drug discovery and product development, preclinical and clinical drug development. They also analyzed the genomic and transcriptomic data and its ability to control the body's drug response. This allows physicians to gain a better understanding when prescribing certain medications and being mindful of the drug dosage and selection.

Members from this group also discussed that although pharmacogenomics in precision medicine is a relatively new field, it has aided physicians in better comprehending the drug response mechanism and increasing drug efficiency. Since this field is in its early stages, Group Genome concluded that in the future some limitations might be inadequacy related to pharmacogenomic testing when it comes to multiple medications.

Group Data



High-throughput, Big Data in Precision Medicine. In Group Data the students were responsible for learning multiple types of large, high-throughput genetic data. They focused on the applications of big data in precision medicine including its advantages and limitations. They also analyzed different types of computing environments such as High-Performance Computing (HPC) Environment and Cloud Computing Environment. They learned how big data can be used to predict different kinds of drug responses in patients who have the same diagnosis.

Students explored the idea of Next-Generation Sequencing and the ability to sequence millions of DNA strands at once in a cost-effective manner. Additionally, this group was able to conclude the HPC and cloud computing will outperform in analyzing big data. While big data can be extremely efficient in creating a personalized medical plan, members found that there is still a heightened need for collaborative networks that will lead to standardization of data content.

Students

ACHUTH SURESH NAIR

B.S. Computer Science, Rutgers School of Arts and Sciences

Interests: Research related to Computational Medicine and Human Computer Interaction. I would like to pursue higher studies in those fields. I would like to join the industry to pursue a career in the healthcare technology sector and contribute to the healthcare system. I have learnt about the essential details of Precision Medicine along with an in-depth understanding of its power to revolutionize the healthcare sector to treat, manage and prevent diseases.

ROTHELA SAMADI

B.S. Public Health, Rutgers School of Arts and Sciences

Interests: Research related to the biological sciences and medicine. I am open to both a research and industry career, and am not yet sure which one I would like to pursue. I've learned a lot about the healthcare field, genomics, the research process, and precision medicine/the future of medicine in general, all which I think will be very helpful in my future career development.

VIGNESH V VENKAT

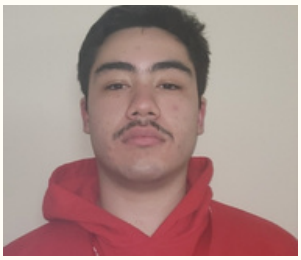
B.S. Computer Science, Rutgers School of Arts and Sciences

Interests: Data Analytics in genomic data. I would like to pursue research through industry. I have learned the magnitude of computation that is required for data analytics in genomics.

MATTHEW J DINATALE

SEBS, Rutgers School of Environmental and Biological Sciences

Interests: Experimental medicines and medical procedures as they pertain to larger mammals, mainly humans. My preference for research would be through that route: Academia. I have learned more about precise sampling techniques of the genome, as well as their importance to a precise diagnosis and prescription, and their applications.



DAVID A MENNA

B.S. Biomathematics, Rutgers School of Arts and Sciences

Interests: Field of Biology and data analytics. I would like to pursue a career either in research or a career in medicine. I have learned the multidisciplinary nature of biology and how computer science is revolutionizing many fields.

RAGHUNANDAN WABLE

B.S. Biomathematics, Rutgers School of Arts and Sciences

Interests: Machine Learning and Artificial Intelligence. I would like to pursue a Masters in Computer Science with a minor in Data Science and possibly conduct research if given the chance. I have learned about the role of Machine Learning and Artificial Intelligence in the field of Medicine, which will prove to be useful towards my future research prospects as well as industry prospects should I choose to work for a company.

Students

NISHI PATEL

B.S. Biological Sciences, Rutgers School of Arts and Sciences

Interests: Genomics and healthcare. I would like to pursue an M.D. and practice medicine in the future. I have learned about precision medicine and analyzing research, which will help me progress in the field of medicine as well as in my future research.

ALLYSON FU

B.S. Computer Science, Rutgers School of Arts and Sciences

Interests: Research related to the biological sciences and medicine. I am open to both a research and industry career, and am not yet sure which one I would like to pursue. I've learned a lot about the healthcare field, genomics, the research process, and precision medicine/the future of medicine in general, all which I think will be very helpful in my future career development.

MAYANK PALIWAL

B.S. Molecular Biology and Biochemistry, Rutgers School of Arts and Sciences

Interests: Biochemistry, genomics, and medicine. I would like to pursue an M.D and become a physician in the future. I have learned about the background and applications of precision medicine; I am sure I will be incorporating multi omics-based data when I practice medicine in the future, as personalized medicine utilizes many factors in order to diagnose and treat patients.

KUSH K PATEL

B.S. Biochemistry, Rutgers School of Environmental and Biological Sciences

Interests: Genomics, healthcare, and cardiovascular disease. I would like to pursue an M.D. to become a surgeon. I have learned about the role of precision medicine and genomics in healthcare as well as its research applications for developing more personalized medicine to advance treatments for complex diseases.

CYNTHIA VENKATESAN

B.S. Biochemistry, Rutgers School of Environmental and Biological Sciences

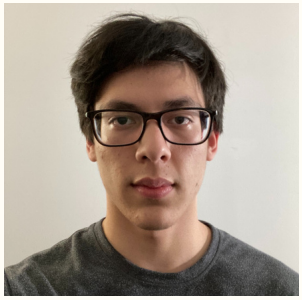
Interests: Medicine, genomics, and healthcare. I would like to pursue an M.D. and be involved in genetic research. I have learned about precision medicine and Gene-Disease databases, which is something I want to learn more about in the future.

ISHAN S KALOVE

B.A. Cell Biology and Neuroscience, Rutgers School of Arts and Sciences Honors Program

Interests: Genomics, medicine, and neuroscience. I would like to pursue an M.D. alongside neuroscience research. I have learned about precision medicine and its important implementation into patient care due to its increased personalization of care that the future of medicine revolves around.

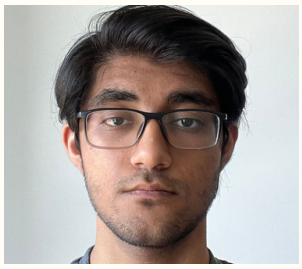
Students



JOSEPH S KOUTSOUTIS

·B.A. Genetics Student, Rutgers School of Arts and Sciences

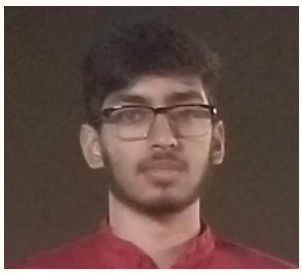
Interests: Genomics and programming. I would like to pursue a career in the bioinformatics industry. I have learned about precision medicine and the pipeline of genomics, and this will help me to get involved with research at Rutgers.



ROSHAN D VARADHAN

·B.S. Business Analytics & Information Technology Student, Rutgers Business School

Interests: Artificial Intelligence, Machine Learning, and Data Analytics. I would like to pursue a career in the software engineering industry. I have learned about the current issues with Big Data in Precision Medicine, which can be a potential career path for me in the future and something to learn more about.



THIRTH A PATEL

B.A. Genetics Student, Rutgers School of Arts and Sciences

Interests: Molecular modeling, medicine, and programming. I would like to pursue an M.D. while hopefully doing genetic research. I have learned about precision medicine and the untapped potential it has for the future of medicine, which I hope to research in the future.

ANIRUDH S PAPPU

B.S. Computer Science, Rutgers School of Arts and Sciences

Interests: Computer programming and healthcare. I am currently undecided between working in a company/industry and doing research. I would enjoy a career in either. I have learned how data science, artificial intelligence, and machine learning can be applied in ways I was unaware of. It also helped me realize that computer programming is very beneficial in healthcare.

ZARA RAFIQUE

B.S. Biological Sciences Student, Rutgers School of Arts and Sciences

Interests: Neurobiology, healthcare, and medicine. I would like to do a PhD in Medicine, and as well aim for a research career in academia. I have learned much about precision medicine and date research. It has definitely guided me in the planning of my future career in medicine.

VIKRAM S IYER

B.S. Finance, Rutgers School of Business

Interests: Heath conditions compared to financial situations (i.e. illness prevalence in each economic class). I would prefer to join industry, I want to work at a business firm. I learned how to analyze and assort data in this class as well as how to organize materials in an orderly manner which can assist my future career.

Students

MARC A KLEYMAN

B.S., Rutgers School of Arts and Sciences

Interests: Subjects of genetics, neuroscience, and exercise science. I would prefer to join an industry which includes research of exercise science. I have learned the possibilities of how programming can be used to create algorithms for genetics and solve many people's possible future health problems by knowing what to do in case of a possible incoming disease.

Teacher Assistants

Sreya Vadapalli

Research Assistant at Ahmed Lab.

Institute for Health, Health Care Policy and Aging Research.

Habiba Abdelhalim

Research Assistant at Ahmed Lab.

Institute for Health, Health Care Policy and Aging Research.

Magazine Editor

Nicole Swenarton

Communications Director,

Institute for Health, Health Care Policy and Aging Research.

Consulted Scientific Literature

- **Precision medicine with multi-omics strategies, deep phenotyping, and predictive analysis.** Progress in Molecular Biology and Translational Science (PMBTS): Precision Medicine. 190, 10. Ahmed Z. (2022). [Book Chapter]
- **Multi-omics strategies for personalized and predictive medicine: past, current, and future translational opportunities.** Emerging Topics in Life Sciences. PMID: 35234253. Ahmed Z. (2022).
- **Practicing precision medicine with intelligently integrative clinical and multi-omics data analysis.** Human Genomics. 14(1), 35. PMID: 33008459. Ahmed, Z. (2020).
- **Artificial intelligence with multi-functional machine learning platform development for better healthcare and precision medicine.** Database: the journal of biological databases and curation. baaa010. PMID: 32185396. Ahmed, Z., Mohamed, K., Zeeshan, S., & Dong, X. (2020).
- **100 Years of evolving gene-disease complexities and scientific debutants.** Briefings in Bioinformatics. 21(3), 885–905. PMID: 30972412. Zeeshan, S., Xiong, R., Liang, B. T., & Ahmed, Z. (2020).
- **Advancing clinical genomics and precision medicine with GVViZ: FAIR bioinformatics platform for variable gene-disease annotation, visualization, and expression analysis.** Human Genomics. 15(1), 37. PMID: 34174938. Ahmed, Z., Renart, E. G., Zeeshan, S., & Dong, X. (2021).
- **Genomics pipelines to investigate susceptibility in whole genome and exome sequenced data for variant discovery, annotation, prediction, and genotyping.** PeerJ. 9:e11724. PMID: 34395068. Ahmed, Z., Renart., E., & Zeeshan, S. (2021).
- **JWES: A new pipeline for whole genome/exome sequence data processing, management, and gene-variant discovery, annotation, prediction, and genotyping.** FEBS Open Bio. 11(9):2441-2452. PMID: 34370400. Ahmed, Z., Renart., E., Mishra, D., & Zeeshan, S. (2021).
- **RNA-seq driven expression and enrichment analysis to investigate CVD genes with associated phenotypes among high-risk Heart Failure patients.** Human Genomics. 15, 67. PMID: 34774109.Ahmed, Z., Zeeshan, S., & Liang, B. T. (2021).
- **Intelligent health system for investigation and consenting COVID-19 patients and precision medicine.** Personalized Medicine. 18(6), 573–582. PMID: 34619976. Ahmed, Z. (2021).
- **Human gene and disease associations for clinical-genomics and precision medicine research.** Clinical and Translational Medicine. 10(1), 297–318. PMID: 32508008. Ahmed, Z., Zeeshan, S., Mendhe, D., & Dong, X. (2020).
- **MAV-clic: management, analysis, and visualization of clinical data.** JAMIA open. 2(1), 23–28. PMID: 31984341. Ahmed, Z., Kim, M., & Liang, B. T. (2019).
- **Debutant iOS app and gene-disease complexities in clinical genomics and precision medicine.** Clinical and Translational medicine. 8(1), 26. PMID: 31586224. Ahmed, Z., Zeeshan, S., Xiong, R., & Liang, B. T. (2019).
- **MAV-seq: An interactive platform for the Management, Analysis, and Visualization of Sequence Data.** Human Genomics, 10. PMID: 27294413. Ahmed, Z., Bolisetty, M., Saman, Z., Anguiano, E., Ucar, D. (2016).
- **Lipid-Pro: A computational lipid identification solution for untargeted lipidomics on data-independent acquisition tandem mass spectrometry platforms.** Bioinformatics. 31, 1150-1153. PMID: 25433698. Ahmed, Z., Michel, M., Saman, Z., Dandekar, T., Mueller, M., & Fekete, A. (2015).
- **Software Applications toward Quantitative Metabolic Flux Analysis and Visualization.** Briefings in Bioinformatics. 15, 91-107. PMID: 23142828. Dandekar, T., Fieselmann, A., Saman, M., & Ahmed, Z. (2014).
- **Isotopo Database and Tool for Facile Analysis and Management of Mass Isotopomer Data.** Database: The Journal of Biological Databases and Curation. 2014. PMID: 25204646. Ahmed, Z., Saman, Z., Huber, C., Hensel, M., Schomburg, D., Münch, R., Eylert, E., Eisenreich. W, & Dandekar, T*. (2014).
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Acknowledgments

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Recognizing the contributions of Rutgers Information Technology for providing excellent media system and technology support. Great respect for the administration of *The Honors College of Rutgers - New Brunswick* for providing excellent class rooms and world class learning facility.

Byrne Seminars Program

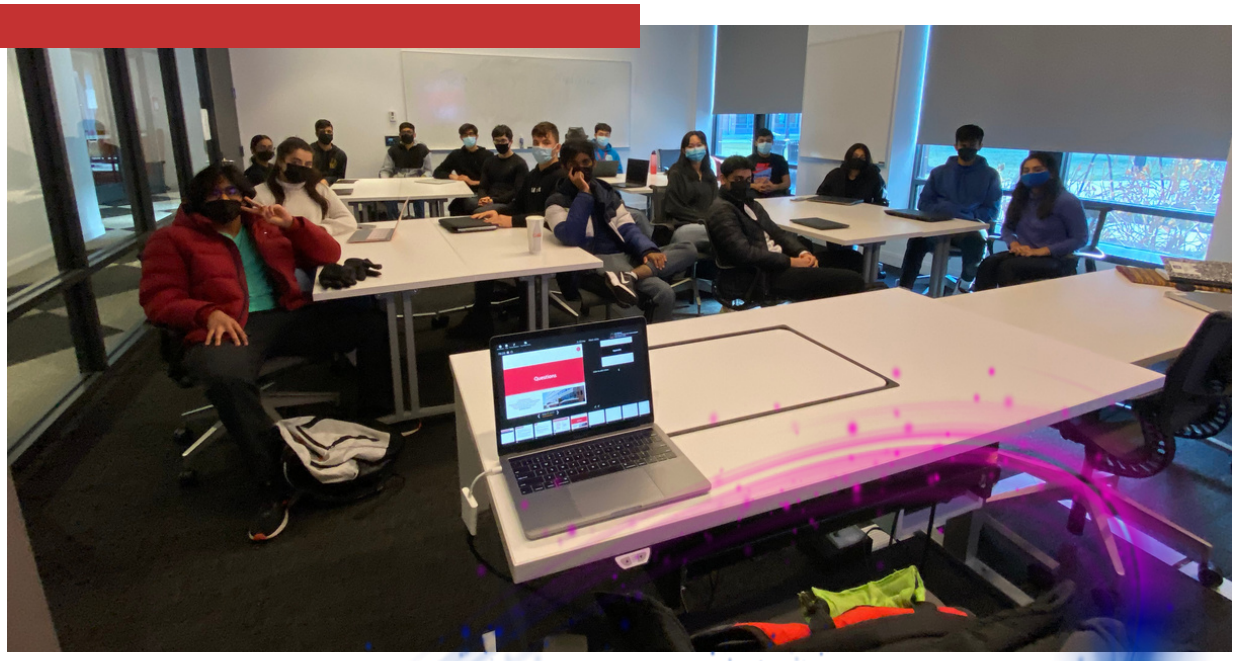
Byrne Seminars Program at the *Rutgers - New Brunswic* offers the opportunity to experience the excitement of original research, as faculty members share their curiosity, their intellectual passion, and their new ideas and fields of knowledge.
Details are available at: <https://nbprovost.rutgers.edu/byrne-seminars>

Ahmed Lab at Rutgers IFH

Ahmed lab is part of the Rutgers Institute for Health, Health Care Policy and Aging Research (IFH). This lab is directed by Dr. Zeeshan Ahmed, the designer and lecturer of this course. The Rutgers IFH is an excellent hub for multidisciplinary and translational research focused on improving population health.
Details are available at: <https://ifh.rutgers.edu/>

Ahmed lab is involved in research with focus on intelligent and multi-functional systems development for integrative healthcare and multi-omics data analysis to discover disease biomarkers and phenotypic characterization for advanced diagnostic and personalized treatment in precision medicine.
Details are available at: <https://promis.rutgers.edu/>

This course is part of the precision medicine project by the Ahmed lab.
Details are available at: <https://sites.rutgers.edu/precision-medicine/>



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URL - IFH: <https://ifh.rutgers.edu/>