

**BIOMEDICAL DATA SCIENCE  
PRESENTS:  
BIODS 260  
10/13/22 1:30PM-2:30PM  
MSOB X303 (SEE ZOOM DETAILS BELOW)**

Lynn Petukhova  
Assistant Professor,  
Epidemiology and Dermatology  
at the Columbia University  
Medical Center

## **TITLE:**

Leveraging Information in the Human Genome to Improve Skin Health  
and to Advance the Practice of Dermatology

## **ABSTRACT:**

Title: Leveraging Information in the Human Genome to Improve Skin Health and to Advance the Practice of Dermatology

Abstract: The process of diagnosing a patient historically has largely relied on clinical observations of symptoms by physicians. Limitations of a clinical diagnosis have been identified with the use of genetic and genomic technologies, which demonstrate that a molecular diagnosis derived from biomedical data can provide greater diagnostic accuracy and inform subsequent management. I conduct human genetic studies as a starting point for leveraging information in the human genome to improve the accuracy and utility of a skin disease diagnosis. Statistical evidence for an association between an inherited genetic variant and a disease outcome is a definitive marker for a disease mechanism, but does not provide adequate resolution of the mechanism for clinical translation. The scale and complexity of biomedical data that is available to define disease mechanisms requires data-driven approaches to identify salient features and to detect patterns among them that link disease mechanisms to interventions and outcomes. Using the hair follicle as a model organ to understand mappings between disease mechanisms and clinical diagnoses, our group is using clustering, network, and tensor factorization methods to discover clinically relevant relationships among genetically-derived disease entities. I will present results from three studies that our group is conducting that leverages knowledge about inherited genetic variants, disease genes, pathways, and/or comorbidities to define an underlying causal structure of skin disease pathogenesis and to identify key genetic regulators of hair follicle health.

### **Suggested Readings:**

<https://www.nature.com/articles/s41598-017-16050-9>

<https://dl.acm.org/doi/abs/10.1145/3368555.3384464>

**Zoom link: [https://stanford.zoom.us/j/92865685887?  
pwd=YjIUM1cxOHZ4UnBZMkhqcG1JYzFNdz09](https://stanford.zoom.us/j/92865685887?pwd=YjIUM1cxOHZ4UnBZMkhqcG1JYzFNdz09)**

Meeting ID: 928 6568 5887

Password: 219826

