**UWEC/PreventionGenetics Molecular Genetics Workshop**  
**November 2, 2017**

10:00-10:45 Helix Hall **Power of Genetics**

*Christina Zaleski, MS CGC* (UWEC alum)

- Introduction & overview for workshop

- About PreventionGenetics

- Power of Genetics & DNA Banking, dysmorphology exam pre-testing, ethical considerations & careers in genetics

10:45-11:15 Helix Hall **DNA Sequencing Technology at PreventionGenetics**

*Dr. Luke Drury, PhD Director of Sanger and Next Generation Sequencing*

-Overview of how NextGen sequencing works (short read technology)

-Variant calling (What is a vcf? What is a BAM?)

-Issues with homology  
-Examples of difficult cases (ambiguous reads, false positives, determining if variants are in cis/trans)

11:15-11:45 Helix Hall **Facility Tour  
 Leaders: Luke, Greg, Angie, Diane (4 groups of 4)**

11:45-12:15 Helix Hall **Lunch (Subway)/Break**

Discussion of Positions at PreventionGenetics/Laboratories and Work Environment

-(Q/A with Technician(s))  
-Renee will have table up in back. Discussion of open positions.

12:15-12:30 Helix Hall **Interpreting Sequence Variants: What Makes Something Pathogenic, Uncertain, or Benign?**

*Dr. Diane Allingham-Hawkins, PhD, FCCMG, FACMG, Dean of Geneticists*

-Discussion of Richards *et al.,* 2015 paper

-Tools used to interpret variants (gnomAD, ClinVar, literature, *in silico* programs)

-Example interpretations

12:30-1:00 Helix Hall **How Do Clinical Reports Impact Patients: Ramifications of Genetic Testing***Dr. Angela Gruber, PhD, Director of Accessioning* (UWEC Alum)  
-What’s next for patients after testing?  
-Secondary findings/incidental findings/emphasis on ethical concerns

1:00-1:15 **Break**

1:15-1:45 Helix Hall **Whole Exome Sequencing: Comprehensive Genetic Testing**

*Dr. Greg Fischer, PhD Human Molecular Geneticist* (UWEC Alum)

-What is exome sequencing?

-Clinically-relevant genes/variants  
-Programs, programming for sorting variants  
-Selecting clinically-relevant variant.

1:45-2:00 Helix Hall **Workshop Exercise: Interpretation and Reporting of Sequence Variants**

-Hand out mock cases with variants to take back to UWEC for interpretation assignment.

-Each student will have 1 clear pathogenic, 1 clear variant of uncertain significance, and 1 clear benign variant.

-Questions?