

Trainee Excellence Spotlight: Elizabeth Plender, BS/BA

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Structural and genetic diversity in the secreted mucins MUC5AC and MUC5B

Position: PhD Candidate, Department of Genome Sciences, University of Washington & Fred Hutch Cancer Center

ASHG: Can you describe the type of research that has your primary focus?

Elizabeth Plender: I study genetic variation in the mucins, a set of heterogenous loci that encode large glycoproteins with defensive properties in mucosal



linings. Mucins are fascinating genes because they harbor coding variable number tandem repeats (VNTRs) that make them extensively polymorphic. Using long read sequencing, I characterize common mucin alleles, study their evolution across human populations, and try to contextualize this variation through epithelial disease associations.

ASHG: Throughout your life, what have been some of the biggest career goals that you have wanted to accomplish?

Plender: I hope to eventually run a lab that is focused on understanding the functional impacts of mucin genetic variability. These genes have been understudied because sequencing their VNTRs accurately has been a challenge. Long-read technologies, however, give us the haplotype-level sequence resolution that we need to begin understanding why the mucins are associated with so many common diseases, including cancer.

ASHG: What are some of the reasons you chose to study genetics instead of anything else?

Plender: Studying genetics has been a powerful way for me to understand my own humanity. I like to say that my job can be boiled down to looking at strings of A's, T's, G's, and C's all day, which sounds somewhat boring! Yet, I am privileged to spend immense amounts of time thinking about the origins of life's diversity. I am grateful to be in a community of people that have chosen genetics to be their life's vocation.

ASHG: What are three words that you would use to describe yourself?

Plender: Passionate, exuberant, and driven.

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