Titles (In Order of Appearance):

Evan Eichler [Pronunciation: IKE-lur]
Professor of Genome Sciences, UW School of Medicine
Investigator, Howard Hughes Medical Institute

Mitchell Vollger [Pronunciation: VUL-jur]
Postdoctoral Fellow, UW Medicine Eichler Lab

Glennis Logsdon
Postdoctoral Fellow, UW Medicine Eichler Lab

– – –

Evan Eichler
Professor of Genome Sciences, UW School of Medicine
Investigator, Howard Hughes Medical Institute

:05-:35 [Audio only :01-:31]
I was part of the original Human Genome Project back in 2001, and I was very interested in regions of the genome that were really complex – and complex from the perspective that they were highly repetitive, but also encoded genes. And so, when the human genome was declared finished or finishing, a lot of my regions weren't done. And I say they're my regions because I was interested in roughly those 500 genes that are in those regions, because they're important for both diseases as well as human evolution.

:36-1:13 [Audio only :32-1:08]
It's been an intense desire to finish it. I've always kind of come back to that point that, in order to really understand genetic variation comprehensively, we need to have a reference that's complete. Otherwise, we're missing pieces of the puzzle. And, you know, it's probably this idea that, you know, 95% of the puzzles being solved is good enough for some people. But I guess for me, getting that last 5% was so important because I believe so much of what we don't understand about disease, or we don't understand about evolution is disproportionately represented in that 5% of the genome that we didn't sequence first off.

1:14-1:35 [Audio only 1:09-1:31]
I consider it a privilege to actually build the next generation of scientists. And so, it's so much fun to see them start as students that feel they have a small piece of the world they can understand, and to feel that they can contribute to a big project, and then not only contribute to that project, but carry it to the next level. To me, that's one of the most exciting things about being a mentor now.

1:36-2:05 [Audio only 1:32-2:00]
This is not the end. Even though people would say, ‘Well, we're done, we finished the genome.’ We finished a genome, and there will now be hundreds, probably thousands over the next few

years. And I think our view of how humans differ from each other is going to be transformed, and how that genetic variation is important – not only for making us human, but making us different and susceptible and protected from various diseases. I think that's going to transform.

Mitchell Vollger
Postdoctoral Fellow, UW Medicine Eichler Lab

2:10-2:20 [Audio only 2:01-2:10]
I'm Mitchell Vollger. I'm also a postdoctoral fellow in Evan Eichler's lab and I study segmental duplications in the human genome.

2:21-2:48 [Audio only 2:11-2:39]
So, the Telomere-to-Telomere Project has been a really great international collaboration between a huge number of labs. And it started as sort of like a summer workshop, actually. It was supposed to be this small collection of scientists trying to finish the remaining parts of the genome, and it rapidly evolved into this huge collaboration to finally sort of get the 8% of sequence that's been missing since 2001 from the human genome.

2:49-3:10 [Audio only 2:40-3:01]
We were all able to really complement each other's strengths to make this happen in a relatively short time period. We had been working on the human genome for 20 years, and then in about the span of a year or so, we were able to resolve almost all the sequence that was previously missing.

3:11-3:43 [Audio only 3:02-3:34]
I remember as a kid, seeing the magazine covers for a complete human genome, in like 2001. And I remember thinking that that was like the coolest project, and how I was kind of disappointed that I would never get to do something that cool. So, I've thought about that a lot during this project that I actually got to contribute sequence to the human genome, and really that excites me a lot that I had the opportunity to do that.

3:44-3:55 [Audio only 3:35-3:46]
We're going to have so many more opportunities to do that. We're going to sequence so many more individuals and try to assemble them from telomeric-to-telomere to understand the scope of human variation.

Glennis Logsdon
Postdoctoral Fellow, UW Medicine Eichler Lab

4:00-4:10 [Audio only 3:47-3:56]
My name is Glennis Logsdon, and I'm a postdoctoral research fellow in Evan Eichler's laboratory at the University of Washington. And I study the centromere, which is an essential region found on our chromosomes.

4:11-4:34 [Audio only 3:57-4:20]
So, we had to develop new ways to target these regions, so we took advantage of new technology that were on the horizon, such as ultra-long read sequencing, to really traverse these regions for the very first time. And we also spent a lot of time on polishing the human genome sequence so we could get it highly accurate. I believe the accuracy is over 99.99999%, which is about as accurate as you can get it with the technologies that exist today.

4:35-5:01 [Audio only 4:21-4:46]
I think that this will fundamentally change how we do genomics moving forward. Instead of looking at small regions of a genome sequence, we'll be looking at the complete genome of an individual. We'll also be looking at the variation between individuals to determine how this impacts genetic and epigenetic diseases. So, I think that this will be a real tour de force, and will really impact how we change medicine in the future – and how we can detect and treat diseases moving forward.

5:02-5:16 [Audio only 4:47-5:02]
This is such an exciting time, and it means we have a new baseline for personalized medicine, from which we can move forward. We can look at the variation between individuals and how this affects disease. So yeah, I'm really super excited about it, and I'm looking forward to the discoveries we can make in the future.