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One Little Mutation Can Make All the Difference

An interview with **Carl de Boer**, Assistant Professor, University of British Columbia
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Our goal is to figure out how the genome is regulated. Well, we take a slightly different approach than many groups. Oftentimes people are trying to figure out how the genome works by studying the genome itself, but that's not our approach. We're trying to make new DNA, and then translating what we learned from the new DNA to the genome.

There's a couple different applications to our work. One of the biggest ones is in understanding heart disease or autoimmunity or mental illness. So we've studied these in great detail now, and we understand the genetics of them quite a bit. And what we've learned from the genetics is that much of the genetic cause of these diseases is actually in when and where these genes are turned on, not so much in the genes themselves.

So if we can understand the code that the cells are using to interpret regulatory DNA sequences, we can figure out, okay, this person has a mutation in this particular sequence. What is it actually going to do in that person? And figure out how it actually links to disease that way.

So in the long term, now that we understand how this one is contributing to disease, we can potentially reverse those changes by targeting either the mutation itself, something upstream of the mutation, or something downstream of the mutation.

In 15 years it would be amazing if we had computer models of the cell that could predict essentially everything that goes on in the cell. We could plug in a genome sequence and run it through this cellular program, and it will tell you exactly what that genome sequence is going to produce, potentially going all the way from an oocyte — a fertilized egg — all the way to an organism.