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What Kind of Mutant Are You?

An interview with Professor Fritz Roth
Molecular Biologist, University of Toronto

How are we looking at mutations in genes?

The idea is to understand how, when something in our genome changes, when a letter in our dictionary of six billion letters changes, how is that propagated through the whole system? So each of us carries something like a hundred mutations or variants and so this is the real challenge is to figure out which of these mutations don't matter - and many of them don't - and which of them we should worry about.

How does your research work?

Some of what we can know about how our genes relate to disease - we can do statistics. We can just say: "people with that mutation, that variant, tended to get this disease and we can have this therapy and it works for those people". But what we see a lot in the clinic is rare variation, where we don't have a lot of statistics, we don't have a lot of opportunity to learn on a lot of people, so we actually have to have models of the disease and we actually have to understand how the system works to predict what's going to happen when we make that change.

What are you most excited about right now?

What I'm very excited about is a field called deep mutational scanning. In a model of a disease, we want to construct an assay for a particular gene that's involved in that disease. The idea is could we be efficient about this and actually test all possible mutations in a gene ahead of time? So that when somebody came into the clinic with a mutation, we said "hey, we already did the experiment. Even though we've never in humanity seen that mutation before, we did the experiment and we think that mutation's not a problem, or it is". For this, we need molecular biologists, we need geneticists, we need engineers, and we need computer scientists to put it all together to come up with one deep mutational scan that tells us which mutations are bad and which aren't.

TODAY'S RESEARCH. TOMORROW'S REALITY.