WEBVTT Kind: captions Language: en

00:00:04.220 --> 00:00:06.580 - The thing that's really remarkable about the skin

00:00:06.580 --> 00:00:08.250 is that it's a single cell

00:00:08.250 --> 00:00:10.150 that makes up the entire skin.

00:00:10.150 --> 00:00:12.630 But in fact, that skin cell requires

00:00:12.630 --> 00:00:14.950 an orchestrated expression of hundreds

00:00:14.950 --> 00:00:16.720 if not thousands of different genes

00:00:16.720 --> 00:00:19.300 in order to fulfill those different roles.

00:00:19.300 --> 00:00:23.190 And so when one of those genes goes wrong,

00:00:23.190 --> 00:00:24.893 the whole skin changes.

00:00:26.340 --> 00:00:28.040 - I have a condition called

00:00:28.040 --> 00:00:31.800 disseminated superficial actinic porokeratosis

00:00:31.800 --> 00:00:34.180 also commonly known as DSAP.

00:00:35.260 --> 00:00:37.080 The texture is very rough.

00:00:37.080 --> 00:00:39.180 It's a scaly feeling.

00:00:39.180 --> 00:00:40.270 If you've been in the sun,

00:00:40.270 --> 00:00:41.783 it can turn red.

00:00:43.470 --> 00:00:45.790 - Most disorders have a genetic basis.

00:00:45.790 --> 00:00:47.340 And in the case of skin disease,

00:00:47.340 --> 00:00:50.660 it's incredibly easy to see those genetic events.

00:00:50.660 --> 00:00:52.520 They are the basis of

00:00:52.520 --> 00:00:54.280 the moles that we have on our arm,

00:00:54.280 --> 00:00:57.760 they're the basis of even the acne or psoriasis

00:00:57.760 --> 00:00:58.933 that we might develop.

00:01:00.000 --> 00:01:03.130 - It's frustrating when you see

00:01:03.130 --> 00:01:05.330 the condition yourself getting worse

00:01:05.330 --> 00:01:08.183 and there's just absolutely nothing.

00:01:20.010 --> 00:01:22.870 - We were able to take a piece of your tissue

00:01:22.870 --> 00:01:25.310 or a swab from the inside of your cheek

00:01:25.310 --> 00:01:28.590 or a little bit of blood, take it back to the laboratory,

00:01:28.590 --> 00:01:30.640 analyze the entire genome

00:01:30.640 --> 00:01:33.210 and look for what mutations underlie that disease

00:01:33.210 --> 00:01:34.253 in the laboratory.

00:01:35.270 --> 00:01:38.500 - The opportunity to work with Yale,

00:01:38.500 --> 00:01:41.750 it was really what I saw as my best chance

00:01:41.750 --> 00:01:45.590 of being able to actually figure this condition out.

00:01:45.590 --> 00:01:49.140 - By studying genetic disease, we can discover pathways

00:01:49.140 --> 00:01:53.480 that are relevant to both the cause of that disease

00:01:53.480 --> 00:01:55.780 but also potentially to the cure of that disease.

00:02:02.200 --> 00:02:04.840 And so when we find a mutation in a new gene,

00:02:04.840 --> 00:02:06.917 the first thing that we do is you say,

00:02:06.917 --> 00:02:08.797 "Well, what is the normal function of this gene

00:02:08.797 --> 00:02:09.630 "within the skin?

00:02:09.630 --> 00:02:10.630 "What does it do?"

00:02:10.630 --> 00:02:12.260 We can then think about

00:02:12.260 --> 00:02:14.963 what this pathogenesis of the disease might be.

00:02:16.070 --> 00:02:20.890 - They found something similar in our case to another case

00:02:20.890 --> 00:02:22.600 and they propose the treatment.

00:02:43.540 --> 00:02:45.000 - Some of the mutations that we see

00:02:45.000 --> 00:02:46.900 in these rare scaly skin disorders

00:02:46.900 --> 00:02:50.320 actually also very relevant to garden variety eczema

00:02:50.320 --> 00:02:53.460 and so by identifying new parts of that genetic puzzle,

00:02:53.460 --> 00:02:55.540 these rare diseases unlock insights

00:02:55.550 --> 00:02:58.330 that become relevant to common diseases as well.

00:02:58.330 --> 00:03:00.500 - Typically you to a dermatologist

00:03:00.500 --> 00:03:02.720 and they tell you what they know

00:03:02.720 --> 00:03:03.880 and that's it.

00:03:03.880 --> 00:03:05.550 Working with Dr. Choate's team

00:03:05.550 --> 00:03:08.990 is you get access to not just the clinical side,

00:03:08.990 --> 00:03:11.020 but also the research.

00:03:11.020 --> 00:03:13.310 That is something that really gets to the root

00:03:13.310 --> 00:03:15.183 of the science behind all of this.

00:03:16.390 --> 00:03:19.260 The condition has become much more manageable.

00:03:19.260 --> 00:03:23.220 I no longer think twice about going outside

00:03:23.220 --> 00:03:25.820 and having to be completely covered up.

00:03:25.820 --> 00:03:27.320 It's made my life much better.

00:03:28.220 --> 00:03:30.520 - [Dr. Choate] We've really had the opportunity here at Yale

00:03:30.520 --> 00:03:34.050 to define over a dozen new genetic skin disorders

00:03:34.050 --> 00:03:35.860 to understand their genetic basis

00:03:35.860 --> 00:03:38.220 and in fact, in the case of a couple of them

00:03:38.220 --> 00:03:39.760 come up with cures.

00:03:39.760 --> 00:03:42.280 It's in studying these fundamental pathways

00:03:42.280 --> 00:03:44.423 within the skin and within human disease

00:03:44.423 --> 00:03:46.840 that we ultimately develop things

00:03:46.840 --> 00:03:49.413 that are relevant to treating people.