CH_Dr. Bupp & Dr. Rossetti_BTS_Full_Final

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Genetics and genomics is a wild world.

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We're all born with a genetic code.

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It's our blueprint.

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So we have cancer risk genes, we have genes that impact our cholesterol, we have genes that impact our brain function, whether we'll have Alzheimer's and more and more.

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There are opportunities where before those symptoms start, you now can start to anticipate or treat ahead of time.

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You can genetically test somebody to know that their heart is predisposed to have that sort of arrhythmia, which can be fatal, and you can go ahead and put a defibrillator or a pacemaker in.

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So that's if and when that does happen, that person will not die.

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And it truly is revolutionary.

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As the field has advanced and we're starting to talk about things like gene therapy or gene editing, the typical function of a gene, which is essentially just to make a protein, is restored.

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Spinal muscular atrophy is an example.

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Duchenne muscular dystrophy, sickle cell anemia, thalassemia.

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You're catching those conditions before they manifest.

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So before babies are having any symptoms of them, you're replacing all of those genes that the baby was born with that might not be working correctly with donor genes that do have all of the correct pathway parts in place.

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And those kids are essentially cured of that genetic condition.

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Yeah, newborn screens a really good example.

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Kids shouldn't be sick and in the hospital.

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Adults shouldn't have tons of medical things going on.

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I think one of the hardest things in genetics is seeing patients and families that they've had a diagnostic odyssey.

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They have gone years, sometimes decades, without an answer.

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What's going on?

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We have the opportunity now to look through people's genetic code and say This is why we can find diseases now that we just didn't know existed.

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It brings us a lot of hope and brings families a lot of hope that 10 years ago didn't exist.

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10 years ago we were having very different conversations.