VIDEO TRANSCRIPT

Difference, Not Deficit: Reframing the Conversation around Genetics, Deafness, and Disability

Link to video.

Introduction

The presentation in this video was provided in ASL (American Sign Language) by Professor Derek Braun from Gallaudet University. The transcript below is the spoken translation by the live ASL interpreter.

Transcript

[**ASL interpreter**]: Hello, everyone. I would like to introduce myself. My name is Derek Braun, and I am a microbiologist and geneticist at Gallaudet University. My doctoral degree is from the University of Maryland.

I have been teaching at Gallaudet first as an adjunct, then later as a tenure track for 26 years now. I am also on the editorial board of the journal CBE Life Sciences Education.

To better understand why things are the way they are today, I think it's important to go back and look at the historical roots of genetics research. The beginnings of the study of human genetics in this country was intertwined with eugenics movements at the turn of the 20th century.

This was a popular movement that has had a lasting impact. And it was prevalent. College textbooks of the time put a heavy emphasis on eugenics, authored by writers like Charles Benedict Davenport.

Among many things, Davenport studied the shapes of skulls with the intent of identifying inferior races. He founded the eugenics record office and directed Cold Spring Harbor laboratory in Massachusetts.

Alexander Graham Bell, I will refer to him as a G Bell, was a prominent eugenicist. As a patent holder for the telephone, he was wealthy and influential.

Just one second, I need to reset my, my different windows on my screen.

So going back to Alexander Graham Bell, now he didn't invent the telephone, but he does hold the patent for the telephone. He was wealthy and very influential and very passionate about his various causes.

I've had the privilege of reading many of his personal correspondence because I work with a historian at Gallaudet University and we've looked over his letters, and I can tell you that Bell was extremely stubborn to a fault.

He was so stubborn that he damaged many of his professional relationships during his lifetime because of his persistence, because he would not let things go.

One of Bell's biggest causes was the deaf community. Bell's mother and his wife were both deaf. His father taught deaf students and so did his brother. Bell himself worked as a teacher of the deaf for some time. He was very outspoken about deaf education throughout his life.

When Alexander Graham Bell was elected to the National Academy of Sciences in 1884, he was invited to give a presentation. The Academy probably thought that he would talk about the telephone, but he surprised them with a speech about the dangers of deaf intermarriage.

The title of his speech was “Upon the formation of a deaf variety of the human race”.

After his speech at the National Academy of Sciences, Bell founded an organization called the Volta Bureau with the purpose of researching deafness and deaf education.

In 1892 he partnered with Edward Allen Fay. Fay was the Vice President of Gallaudet College at the time. He wanted to do a study on whether deaf intermarriage was increasing the number of deaf people. This was a massive undertaking and funded at great expense by Bell.

Doctor Fay collected and collated pedigrees from 4471 deaf marriages from all across the country. This was probably the largest study of human genetics ever done to date. And we still use that data today. And I believe that study possibly marks the beginning of the formal study of human genetics.

Here I am showing a family portrait of John and Annie McNeill Sherman who participated in the Fay study. I found their photo in the database.

So, the results of the Fay study actually disproved Bell's hypothesis. Fay and Bell were unaware of Mendel's work and their simple analytic approach that they used with Fay's data failed to show clear inheritance patterns. But Fay showed that deaf parents almost always had hearing children. And that most deaf children typically are born to hearing parents.

As the decades passed after Fay's study, Mendel's theories were rediscovered and expanded upon. Scientists also developed theories in population genetics with the work of Sewall Wright and RA Fisher and many others.

We now know that deaf intermarriage can cause a small increase in phenotypic expression of deafness. But we know that the alleles will not change because deaf people have no advantage over hearing people, there are no reproductive advantages. And under present conditions, deaf people becoming parents simply cannot lead to the formation of a "deaf variety of the human race".

But this has not stopped geneticists from repeating this disproven idea in the discussion section of many papers. Even today, this idea is often repeated. I point to a 2023 paper from a group of Russian scientists that were built on that hypothesis.

I want to show how powerful AG Bell was. Alexander Graham Bell had a strong opinion that learning sign language interfered with learning spoken language. This theory wasn't based on any science. It was just Bell's intuition. And even though that was back in the 1890s, it is still a popular opinion today.

It currently causes a lot of problems because cochlear implants don't always work as advertised. When they don't work well, the deaf child who is not allowed to learn sign language is left with no language at all. Once the window of language acquisition closes in early childhood, this causes permanent cognitive impairment. We call this language deprivation.

So why is there a prevalence of connexin 26? Connexin deafness is surprisingly common. Connexin deafness alone causes half of all genetic deafness and about 1/4 of all congenital deafness. It is no surprise that geneticists are wondering why it's so common.

Connexin deafness is usually autosomal recessive. The three commonest alleles for it are found in between 1 to 5% of the populations where it's studied. They are associated with an ethnic group. 35delG is found in Caucasians and is thought to have originated in ancient Anatolia. 235delC is found in Asians. And 167delT is found in Ashkenazi Jewish ancestry.

There have been several hypotheses to try and explain why it's prevalent. You can guess that deaf intermarriage was one of those hypotheses. That's been disproven as I've mentioned Another hypothesis was the locus of hyper mutable. That has also been disproven. My laboratory has data showing that mutation rate of this locus on chromosome 13 is average for the human genome.

This leaves us with just two other hypotheses for the high prevalence of connexin deafness. One hypothesis is our demographic history that we are founders and we've migrated from Europe. A second and more interesting hypothesis is balancing selection. We are still investigating that question.

So, deafness remains a disproportionate target of genetics research. So, this brings us to the present day. Even today, deafness continues to be in the crosshairs of genetics research.

Recently, Denis Rebrikov, a Russian scientist announced that he would use CRISPR to edit deafness from human eggs. His first proposed genetic target was a polymorphism which confers HIV resistant. The second genetic target was connexin deafness.

Now, why deafness? The OMIM database, the Online Mendelian Inheritance in Man database – OMIM, catalogs more than 1000 human diseases. It includes metabolic diseases that cause suffering and death during infancy or early childhood.

Wouldn't those be less controversial targets for CRISPR? Why deafness?

As a society, we need to learn how to get consent from target populations. We should poll the population and see how they feel about the research. If there is consensus in the population, then there would be consent.

We should also involve them in the research as scientists or even encourage them to lead their own research.

Consent is important, particularly when looking across a cultural lens.

Even today, NIDC, they focus on deafness and “other communication disorders”. But that needs to be dismantled because it doesn't really need to focus on “other communication disorders”.

We have American Sign Language and that is a complete and whole language. Noam Chomsky has agreed that it is a full language.

Deaf people communicate in ASL they have no issues communicating in that ASL language. People that struggle with ASL are the people that don't know ASL. They don't have fluency, so of course, they will struggle.

There have been papers published on deafness and they talk about "pathological alleles" in terms of deafness. But it's very common. And based on that commonality, we should be calling it a "variant", not pathogenic.

This may sound like lots of little points, but they all add up. They add up to what we call microaggressions.

So, keep in mind that deaf people work, they pay taxes for the government. The government continues to remind us that we are broken. And that is how it feels to be a deaf person in America.

I don't know if you've seen the movie Gattaca, if you haven't, it's a great movie. I have my genetics class, watch it every year and write a short essay on it. It came out in 1997 but it's scary how much of the science fiction they show in that movie has become reality.

At the end of the movie, Ethan Hawke turns to Uma Thurman and says, "they've got you looking so hard for any flaw that after a while, that's all you can see". It's a great quote from that movie.

We know that diversity in scientists leads to better science.

Scientists draw upon their own personal observations and life experiences, informing research directions. Genetic diseases in Ashkenazi, Jewish ancestries are well understood because they were studied by Jewish scientists who were interested in the challenges of their own community.

Women's medical issues weren't well understood until recently, when a critical mass of women finally became medical researchers themselves.

Deaf scientists studying American Sign Language allowed us to identify commonalities in neurological activity between spoken and signed languages. And this has led to a deeper understanding of how language processes, how language is processed in the brain.

Research shows that diverse research groups consider more diverse solutions to problems and ultimately choose a more effective solution.

The pipeline for creating deaf scientists is long and has many leaks. We need more solutions.

At the beginning, some deaf children are lost due to language deprivation because of a cochlear implant or oral training that wasn't completely successful. And that child was never given access to ASL.

Educational opportunities for children in primary and secondary school are not equal.

Once a child reaches college age accommodations are required by law, but they are not always provided. Most importantly in college, science is notorious for having an unwelcome atmosphere.

The unanimous conclusion from a whole host of studies is that the difference between people who stay in science and those who switch to other careers is how well they can tolerate the culture of science. It is a culture that disproportionately weeds out women, racial and cultural minorities, and disabled students.

This is the topic of some exciting current research. Recent research shows that cultural competence by faculty and mentors and advisors goes a long way toward improving outcomes with cultural minorities. This includes deaf students as well.

Research shows that cultural competence can also probably be taught.

There's also research showing that science identity, when a student feels welcome and identifies them as a scientist, is also important for persistence.

There have been some tremendous success stories at Gallaudet and at Rochester Institute of Technology. If you're interested in the latest information, the latest research, or how to support a deaf student through science program or research mentoring, I can recommend two publications.

Another exciting development is the concept of “deaf space”. Deaf space recognizes that in a purely visual centric environment, things are to be designed a little differently. It's based on universal design principles and, unsurprisingly, it benefits everyone.

I led in the design of the first deaf space biology research lab on Gallaudet campus. It has been a wonderful space to work and teach in. My office has glass windows. sitting in my office, II can see across the lab and converse with students and staff. We've sacrificed a little shelf space for clean visual lines.

The benches are a little further apart than usual allowing deaf people to have conversations in the aisles without blocking traffic.

We've gone green by incorporating windows and using white shelving and bench tops which reflect the natural light. We typically do not use the overhead lights. Instead, we rely on the natural light.

We train deaf undergraduate students and summer interns in our laboratory. The last time we recorded statistics, 29% of our deaf interns had gone on to graduate in STEM fields and 21% were in a post baccalaureate research position. Most went to NIH or at the University of Rochester in New York.

I hope that today I have been able to open your eyes a little bit about the lens that deaf people are viewed through in genetics and genetics research.

It's clear this happened because human genetics research started in the era of eugenics. And the first major topic of study was hereditary deafness.

I hope I've helped you see how this has built a deficit lens. Even today, this still creates an unwelcoming atmosphere for deaf people and for deaf students who want to become scientists.

You can help us by coming, you can help us by becoming an ally, we can reframe discussions so that we emphasize ability, not disability. This includes revisiting the terms we use in our scientific research.

We can become more culturally aware.

We can support the idea of having diversity of scientists knowing that this will bring even more perspectives to the table and lead to a greater variety of research questions being asked and answered.

Thank you for listening.