Do your ears hang low? The complex genetics behind earlobe attachment

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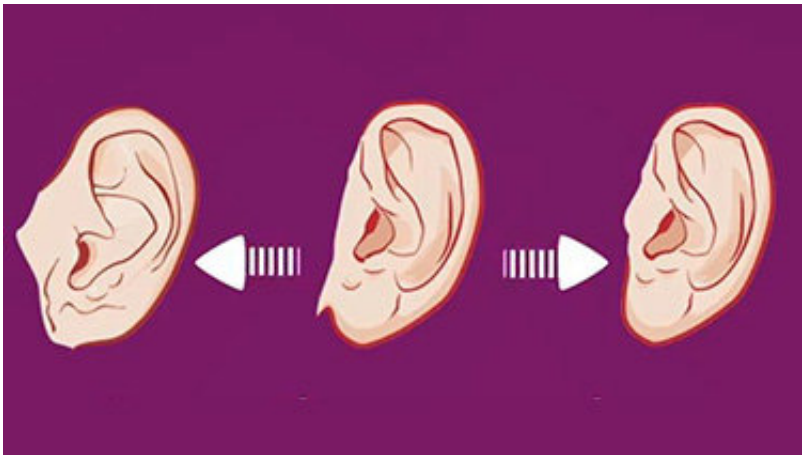

A common, hands-on method for teaching genetics in grade school encourages students to compare their earlobes with those of their parents: Are they attached and smoothly mesh with the jawline? Or are they detached and dangly? The answer is meant to teach students about dominant and recessive genes. Credit: UPMC

A common, hands-on method for teaching genetics in grade school encourages students to compare their earlobes with those of their parents: Are they attached and smoothly mesh with the jawline? Or are they detached and dangly? The answer is meant to teach students about dominant and recessive genes.

Simple, right? Not so fast.

New research led by the University of Pittsburgh Graduate School of Public Health and School of Dental Medicine, and published online today in the *American Journal of Human Genetics*, reveals that the lesson is much more complicated, with an interplay of at least 49 genes contributing to earlobe attachment.

"Sometimes the genetics of a fairly simple trait are actually quite complex," said lead author John R. Shaffer, Ph.D., assistant professor in Pitt Public Health's Department of Human Genetics and the Department of Oral Biology in Pitt's School of Dental Medicine. "By understanding that complexity, we can work toward treatments for genetic conditions, several of which have distinct facial features that involve the earlobe, including Mowat-Wilson Syndrome, which can cause cupped ears with protruding lobes."

The study was an international collaboration involving investigators in the United Kingdom and China, and included data from the U.S.-based personal genetics company 23andMe Inc.

"The great thing about these collaborations is that we not only had a large sample size, but we also had participants from different ethnicities, giving us a greater depth of genetic information," said senior author Seth M. Weinberg, Ph.D., associate professor in Pitt's departments of Oral Biology and Human Genetics.

The study - which is part of a broader investigation of the genetic basis of human facial features - also gives insight into a debate that has been raging in the genetics community: Which is better? Large-scale population studies that cast a wide net but, due to their size, cannot hone in on specifics, or more-focused studies that can obtain extensive participant information? For their earlobe study, the Pitt team did both.

First, the team investigated a sample of just under 10,000 participants

who, in addition to providing genetic data, were examined to determine if their lobes were attached, detached or partially attached. The participants provided some medical history and were screened for a family history of congenital malformations.

Then, the team partnered with 23andMe to include nearly 65,000 of its customers who consented to participate in research - dramatically increasing the study's statistical power. Unlike the more detailed assessment in the initial sample, the 23andMe participants provided self-reports about their own earlobe status as simply attached or detached.

"This is the unique power of 23andMe's research platform - to quickly and easily make genetic discoveries across a broad variety of traits, uncovering biology that might never otherwise see the light of day because it doesn't obviously meet the priorities for research funding," said Joyce Tung, Ph.D., 23andMe's vice president for research. "And yet, important insights can still be drawn even from seemingly insignificant traits."

Using just the information from the smaller group of 10,000 participants, the researchers were able to identify six genes that played roles in earlobe attachment. When the analysis was broadened to include the 23andMe participants, those six genes were again strongly implicated in earlobe attachment, but another 43 genes were added.

"So do we put our resources toward careful and rigorous study of a smaller set of individuals or is it better to get less detail, but recruit a lot more study participants?" said Shaffer. "In our study, we unite both of these approaches, and we see that using the bigger group gave us all the results of the smaller study, plus a whole lot more."

The results don't mean that grade school science teachers should stop using traits like earlobes to teach genetics. But the lesson needs to be

updated to show that even a seemingly simple inherited trait, such as earlobe attachment, involves a complex and fascinating interplay of genes that geneticists are only beginning to understand, said co-author Eleanor Feingold, Ph.D., professor of human genetics and biostatistics at Pitt Public Health.

"We've got these 49 genes that we know affect earlobe attachment, but we don't know how they work together or interact with one another," said Feingold, also senior associate dean at Pitt Public Health. "Figuring that out is the next step."

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