

PLOS Science Wednesday: Hi Reddit, this is Seth Weinberg. I am here with my colleagues today to discuss our paper in PLOS Genetics concerning the identification of genetic variants that influence human facial features.

PLOSScienceWednesday ¹ and r/Science AMAs¹

¹Affiliation not available

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Hi! Your research reminded me of [the Candela project](#).

My college was one of their research sites and in one presentation, the local coordinator argued that their results would allow them to predict risk profiles for certain diseases by using computer vision software that would link facial traits to mutations of clinical relevance.

I couldn't help but feel the professor was overselling the potential applications of such research, but he and I never got along so my assessment may have been biased.

How would you rate the feasibility of those claims?

[joevector](#)

Hi this is Seth: Yes there are a few companies working on things like this based on facial recognition methods developed in the computer science field. This kind of work is still in its early stages. As a side note: we work with some folks in the CANDELA consortium on a collaboration so stay tuned for that. Here's how I see the main applications of our work: studies like this can help us gain a better understanding of how particular genes/pathways influence facial development (more of a basic science application), improve our knowledge of how genetic variation relates to the types of features we see with certain birth defects and syndromes (like why are some individuals more or less severely affected), and possibly (in the future) make predictive models about how the face grows.

What might be one downstream application of figuring out most of the loci associated with facial structure? Would law enforcement or intelligence agencies be able to use these predictors?

[High Point Genetics](#)

Hi this is Seth: There are a number of groups trying go from DNA to face, with a great interest from agencies like the FBI and police. A critical thing to keep in mind is that this work is still in its infancy. Researchers looking at the genetic basis of human facial shape have collectively identified a very small fraction of the variants that influence these complex traits - thus, we can only explain a small amount of

facial variation at this point. Critically, though, even if ALL the genetic variants were known, we would still only have part of the story, because there are numerous non-genetic factors (diet, prenatal nutrition, mechanical forces) that influence the size and shape of our faces. So we are still a long way off in our view. This however has not stopped companies from making claims that they can identify faces from DNA. Right now, be suspicious of these claims.

Hi, thanks for the AMA. I have a couple questions, feel free to answer one or both!

1. Are there any related studies focused on skull structure, perhaps using x-ray images? It occurs to me that the soft tissue of the face might be more malleable by environmental factors than bone. (Maybe this is off - did the genes you identified explain most of the variance?) Also one might be able to more easily apply hard tissue findings to the fossil record to say something about human evolution.
2. Some evolutionary psychologists theorize that facial symmetry is attractive because it advertises "good genes". As someone who actually studies the genetics of facial structure, what do you make of such ideas?

[walkickswillwork](#)

Hi this is seth: There are some studies using x-ray images - mostly out of Japan. They are generally small samples. The big problem with this approach to imaging the face is the radiation exposure. We used 3d photography which is totally non-invasive. The downside, however, is that we cannot look directly at skeletal morphology. Both types of tissues are influenced by environmental factors.

Hi, my son was born with bilateral cleft lip and palate. When we met his craniofacial team before he was born (11yrs ago), we met with a geneticist. She told us that they weren't really sure what causes cleft and that there were only loose correlating factors like ethnicity and whatnot.

Simply put, do we know or at least are we any closer to knowing what causes cleft or maybe even how to prevent it?

[solofatty09](#)

Mary Marazita Thanks for the question. in the time since your son was born our research group and other groups around the world have been engaged in many genetics studies of cleft lip and palate. To date about 20 genetic regions and some specific genes have been identified as associated with cleft lip and palate but it is important to note that these results so far are at a population level, that is we do not yet know how to apply these results to individual families. So in summary, we are much closer to understanding the causes, developmental processes, and genes that contribute to cleft lip and palate but still have work to do.

Hi there! You mentioned that an application of this research might be to "*improve our ability to create forensic facial reconstructions from DNA*". How far off is this kind of technology, and how might it account for variations in facial structure attributable to environmental factors?

[sayeskay](#)

Hi this is Seth: Several people asked similar questions. See my response to High_Point_Genetics.

Hi! There . Thanks for the AMA.

I always think that people's faces can be classified . There may be thousands of really vague classes.

When somebody looks like someone , he/she must have something in common , Say Nelson Mandela and Morgan Freeman, Matt Damon - Mark Warlburg- John Cena etc .(some of my observations) Also include dopplegangers .

What is it that is common here?

[y2k2r2d2](#)

Hi this is seth: Such an interesting question. I would like to suggest that genes play a role, but a study like this has simply not been done yet. I found a website once where people could find their dopplegangers. My first thought was i would love to find these folks and enroll them in a study. We may be able to do this in reverse though, by looking at people with the closest DNA match in our sample and seeing how similar/different their faces are.

When playing a Role Playing Game (RPG) that allows character face customization (e.g. Fallout 3), how long does it take you to create your character?

[rabbiferret](#)

Hi this is seth: Unfortunately the issue has never come up. I wonder what factors go into how people choose make these faces? Do you have any insights.

When playing a Role Playing Game (RPG) that allows character face customization (e.g. Fallout 3), how long does it take you to create your character?

[rabbiferret](#)

John R. Shaffer: I recently spent way too long making my Bitmoji, and I recall doing the same making a Nintendo Wii avatar back in grad school. No genetic data was needed, though.

Have you found evidence of any genes controlling facial features that lie on the allosome (sex chromosome)? If so, what conclusions can you draw from that in terms of evolutionary morphology?

[bluealbino](#)

John R. Shaffer: Yes we find genes associated with facial features on the X chromosome. Across evolutionary time scales the sex chromosomes face different evolutionary consequences than the autosomes. We are not yet sure of the implications of this for facial morphology.

Are you familiar with Ehlers-Danlos Syndrome? We have a subreddit here, [/r/ehlersdanlos](#). EDS is known to affect facial morphology - narrow/high palette, small jaw, dental crowding, dark puffy circles around the eyes, downward-slanting eyes, etc. It has been said that we look like we could all be related. EDS is caused by several different mutations that affect the production of collagen, having wide-sweeping affects on the entire body, the most common expression being double-jointedness. No gene has been identified for the most common form of EDS, so-called "type 3". It sounds like there is a possibility that your research may be relevant to the search for this gene variant. And since there is also the possibility of an overlap between EDS and autism, plus a known correlation between head

size and autism, it may be relevant there, as well.

Can you provide any insight into these conditions, or detail how your research is/is not relevant?

[hosford42](#)

Katya: Hi, yes, we are familiar, but for our study we excluded people who had any genetic syndromes that affect the face, so we can't infer anything about EDS from this study, unfortunately.

If facial features depend on certain genes turned on or off (roughly speaking) would it possible to tell mathematically how many possible face feature combinations there could be?

[tomega](#)

John R. Shaffer: Interesting idea. I suppose that, in theory, we could determine all combinations of genetic variants related to facial morphology, assuming that we had cataloged them all (we haven't yet). It would take a lot of work to really understand how they all act together! And don't forget, genetics is only part of the story. Environmental and behavioral factors play a role, too.

Some novelists, and others, love to infer assignments of character from facial features. I wonder if an understanding of the genetics underlying said features offer any insight toward or away from that apprehension?

[clapter](#)

Hi this is Seth: I am fascinated by this stuff, especially from the historical perspective. The practice you are talking about is called physiognomy and stretches back as far as aristotle. what is really interesting is that there is all kinds of evidence that face and brain development are intimately linked. There are facial differences in individuals with schizophrenia...and brain differences in individuals with cleft lip and palate (i did my post-doc on this). Some recent studies have tried to connect facial morphology to aggression, which may make sense if both are influenced by hormones.

How come some people have asymmetrical faces?

[rogamore](#)

Hi this is Seth: such a complex phenomenon, with both genetic and non-genetic factors at play. Facial asymmetry can be exacerbated in certain known genetic conditions. We have not yet looked at this in our dataset, but will in the future.

I have several questions, they are maybe outside your scope of knowledge, and also they are not logical when seen together : 1. Do people more often inherit the fathers looks? If so, how would that work, genetically?

1. Some facial features seem dominant - do you know why? Like, all the relatives on my mothers side have a slight bump on the nose.
2. Other facial features seem to be able to become extra prominent if both the mother and the father has it. Does that mean that most features are usually a compromise between fathers genes and mothers genes?

3. And lastly: Do you know how nutrition affects looks? Will for example a lack of vitamin x while growing up give narrow eyes?

[Toove](#)

from Mary Marazita good questions--that for the most part we don't have answers to ! (1) there is a relatively new area of genetics studying imprinting which are situations in which some of one parent's genes are preferentially expressed in the offspring. There are now many examples of these in animal models, and a couple examples in humans. We don't yet know of any studies that have studied these mechanisms in human faces. (2) there are a variety of mechanisms that influence the expression of gene variants---one of those mechanisms is dominance of certain variants. (3) there are some nutritional factors that can influence facial features but these are usually quite severe cases of nutritional imbalances.

Are you related in any way to Wilhelm Weinberg, the man who came up with the Hardy-Weinberg principle (along with G.H Hardy)?

[smillerm](#)

Hi this is Seth: Not that i know of...but maybe i should go on Anscetry.com and find out!

In your study did you exclude participants in any way? For example environmental factors could influence development of facial features in utero or severe trauma to the face could alter facial features post-natal, these would both interfere with your statistical analysis correlating only genetic information to facial features

[D-Raj](#)

Katya: Yes, you're absolutely right that this is important for a study such as ours. We excluded subjects if they had a personal history of facial trauma, a personal history of facial reconstructive or plastic surgery, a personal history of orthognathic/jaw surgery or jaw advancement, a personal history of any facial prosthetics or implants, a personal history of any palsy, stroke or neurologic condition affecting the face, a personal or family history of any facial anomaly or birth defect, and/or a personal or family history of any syndrome or congenital condition known to affect the head or face.

I've always wondered, if someone is your doppelganger face-wise, does that make them more likely to share other genetically determined attributes (height, cancer likelihood, etc.)?

[mracidglee](#)

Hi this is Seth: someone else asked a similar question above. I tried to answer it.

I can imagine that depending on developmental environment, you might see varying degrees of influence on the expression of genetic variance. Is it possible come to any conclusions on the relationship between these gene variances and the environment - and if so, how do you control for environment differences?

Semi-related: how do you differentiate "noise" (small mutations from uv exposure, etc) from true "variants".

Other semi-related: Do you take genetic samples from multiple facial regions? Is mosaicism a consideration when it comes to the genes you are considering?

[WtheCore](#)

Mary Marazita Your last point about mosaicism is fascinating---for our study we took our genetic samples (DNA) from white blood cells, we did not take samples directly from the face. Mosaicism could potentially influence the develop of body parts including the face, but to our knowledge no study has investigaged mosaicism in tissues from the face.

How did you get into studying genetics?

What is the most exciting thing about studying genetics?

What is the proudest moment of your career so far?

[thatsconelover](#)

Hi this is Seth: The most interesting thing to me is the amount that we still don't know. Look at our study for example...people have been studying the human face for so long, yet we are only now finding some of the genes involved and we have only scratched the surface. Proudest moment: honestly, probably being notified that the grant that supported this work would be funded. This happened right at the start of my faculty position. That is a very rare thing to happen and it set me on a path to success.

What do you find most interesting about this type of research and what limitations does the field have, currently?

[AngryTable](#)

Hi this is Seth: I have always been interested in the face and human variation in general (my background is in anthropology). Most interesting to me is the connection to facial birth defects and syndromes, which is my other area of study. It is interesting that many of the regions contained genes implicated in these conditions. Maybe a mutation in these genes results in a syndrome, but common variation in these same genes can influence typical-range traits.

What do you find most interesting about this type of research and what limitations does the field have, currently?

[AngryTable](#)

John R. Shaffer: I have always been interested in genetics, but only recently began working on the genetics of facial variation. I think, as humans, we are very attuned to faces, which makes this sort of work very interesting to me.

Are the genes involved in facial features used for any thing else? For instance personality or immune system strength...

[brewmastermonk](#)

John R. Shaffer: Yes. A recurring theme in genetics is that many genes are like Swiss army knives – they have lots of different functions across tissues. As a field, we are still discovering all of these functions.

Hi! I work in cosmetics and something I've been trying to find data on is surface area of the human skin, in particular I would love to see some data on the average surface area of the face!

[kindofstephen](#)

Hi this is Seth: This is possible using the kind of data we collect. The difficulty would be defining where to define the borders of the face. Maybe we will try to do this at some point.

Is there are reason why the sequenced genomes come from unrelated individuals? Because it's possible to have visually similar facial structures derived from distinctly different gene variants (at least theoretically), wouldn't a pedigree be helpful to tracking particular variants as well as answering dominant/recessive questions?

[moleculewerks](#)

John R. Shaffer: Good question. In genetics we study both families and unrelated individuals, and some scientific questions can be better addressed in one study design or the other. In our study, unrelated individuals were used to help us understand population facial norms, and because it was helpful for the statistical analysis to have independent observations. You are right that family studies can be very helpful to investigate genetics of all kinds of traits, including facial variation.

Greetings from your friends and colleagues at Children's Hospital Colorado. Congrats on the publication and the AMA!

[gallen1119](#)

from Mary Marazita. Thanks for the kind words.

Greetings from your friends and colleagues at Children's Hospital Colorado. Congrats on the publication and the AMA!

[gallen1119](#)

Hi this is seth: Thanks! Hi from Pittsburgh.