Plan for this session

- Discuss three papers:

- Paper 1: *Genomic Justice for Native Americans: Impact of the Havasupai Case on Genetic Research*

- Paper 2: *Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior.*

- Paper 3: *The Ethics of Big Data in Genomics: The Instructive Icelandic Saga of the Incidentalome*

Some notes

> You will be divided into 3 groups
> We will spend 15 mins on each paper: 10 mins in small group discussions and 5 minutes as a large group.
> For paper 1, group 1 will report on their discussion to the large group
> For paper 2, group 2 will report on their discussion to the large group
> For paper 3, group 3 will report on their discussion to the large group
> Everyone will read and discuss all 3 papers

Ground rules

> Respect each other – we all have different lived experiences that impacts our thoughts

> If you are comfortable and feel that it is relevant to the discussion, state your positionality to the group. For example, I identify as a white able-bodied non-religious cis-gendered lesbian immigrant.

> Make sure that everyone has the opportunity to speak

> There are no right or wrong answers – try and see these questions from different points of view.

Paper 1: *Genomic Justice for Native Americans: Impact of the Havasupai Case on Genetic Research*

In 2004, the Havasupai Tribe filed a lawsuit against the Arizona Board of Regents and Arizona State University (ASU) researchers upon discovering their DNA samples, initially collected for genetic studies on type 2 diabetes, had been used in several other genetic studies. The lawsuit reached a settlement in April 2010 that included monetary compensation and return of DNA samples to the Havasupai but left no legal precedent for researchers.

Interviews of institutional review board (IRB) chairs and human genetics researchers at US research institutions revealed that the suit drew attention to indigenous concerns over genetic studies and increased their awareness of indigenous views. However, interviewees perceived no direct impact from the Havasupai case on their work; if they did, it was the perceived need to safeguard themselves by obtaining broad consent or shying away from research with indigenous communities altogether, raising important questions of justice for indigenous and minority participants.

If researchers and IRBs do not change their practices in light of this case, these populations will likely continue to be excluded from a majority of research studies and left with less access to resources and potential benefit from genetic research participation.

Paper 1: *Genomic Justice for Native Americans: Impact of the Havasupai Case on Genetic Research*

Q1: What are your thoughts on the Havasupai Case?

Q2: What are the implications of broad vs. narrow informed consent? How do you balance broad consent with responsible conduct of research?

Q3: What (if anything) should the researchers have done differently?

Paper 2: *Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior.*

We performed a genome-wide association study (GWAS) on 477,522 individuals, revealing five loci significantly associated with same-sex sexual behavior. In aggregate, all tested genetic variants accounted for 8 to 25% of variation in same-sex sexual behavior, only partially overlapped between males and females, and do not allow meaningful prediction of an individual's sexual behavior.

Overall, our findings provide insights into the genetics underlying same-sex sexual behavior and underscore the complexity of sexuality.

Paper 2: *Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior.*

Q1: What are your thoughts on this study?

Q2: What do you think the researchers wanted to achieve with this study?

Q3: At the end of the paper, the authors state: *"To communicate the results of the study to the broader audience, we engaged with different LGBTQIA+ (lesbian, gay, bisexual, transgender, queer, intersex, asexual, and other+) and science communication organizations and created multimedia materials for a lay audience."*

> What/who are the gatekeepers? What role do stakeholders play in this kind of research? Is it enough to consult them? How should we report stakeholder engagement?

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HOW IT WORKS PRIVACY BLOG

ACK TO THE GENETIC APPS STORE

122 SHADES OF GRAY

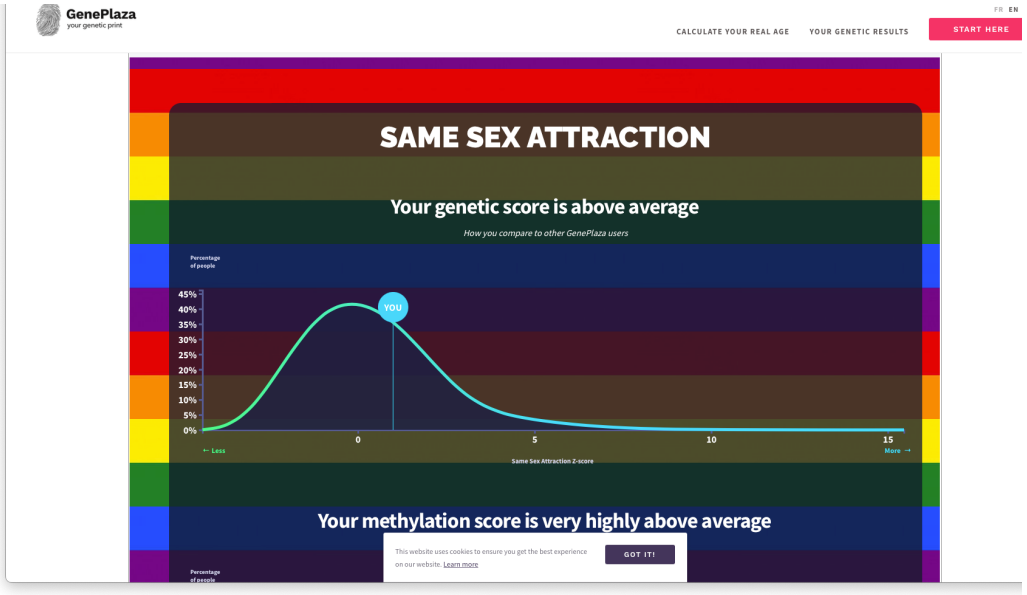
INSOLENT AI & JOEL BELLENSON

Languages:

DISCLAIMER The authors of the science paper and the authors who released the raw data are not associated with this application.

This App does **NOT** predict same sex attraction.
In the published study, evidence was found that many genetic variants contribute to same-sex sexual behavior, and each has a small influence. When added together, these variants explain only a minority of this behavior. This app attempts to quantify the sum of these small effects. This app does not account for any environmental factors that can possibly influence this trait

Your sexual preference phenotype exists in reality and independently of our biocomputational model. Which cannot be expected to match with your reality.
No longer available



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October 14, 2019

Dear Developers of GenePlaza,

We have recently been made aware of an app titled "How gay are you," which claims to show an individual's genetic score for "same sex attraction". As authors of the Science paper, which you cite, we are writing to **urge you to take down this application immediately**.

The "score" you are marketing through this app is a total misrepresentation of the conclusions of the work. Our study indicated that individual-level prediction is impossible for same-sex sexual behavior. The promotion of this app and, in particular, the claims it makes are a gross and dangerous mischaracterization of the work.

In our manuscript we explicitly state that "these scores could not be used to accurately predict sexual behavior in an individual." We also state in our public website (https://geneticsexbehavior.info/what-we-found/) that: "[t]he findings are based on statistical patterns in the data as a whole, and no conclusions can be drawn for any particular individual. It is not possible to predict or identify someone's sexual behavior or sexual orientation from their DNA, nor was doing so our intention."

At minimum, we request that you discontinue claiming that your app is drawn from our work in any way as it does not reflect the content or conclusions of the work.

The right thing to do is to remove the app from Geneplaza completely, which we hope you will do.

Sincerely,

Benjamin Neale

On behalf of co-authors:
Abdel Abdellaoui, Alexander Busch, Andrea Ganna, Rober Maier, Eden Martin, Michel Nivard, John Perry, Alan Sanders, Karin Verweij, Robbee Wedow, Brendan Zietsch

Paper 3: *The Ethics of Big Data in Genomics: The Instructive Icelandic Saga of the Incidentalome*

DeCODE Genetics, Inc. recently introduced a free website that permits Icelanders to learn whether they carry mutations in the *BRCA2* gene that are known to increase cancer risk, even if these citizens have never participated in genetic testing.

This site is made possible by the consanguinity of Icelandic citizens, who number fewer than 350,000, and their detailed genealogical records dating back centuries, a set of circumstances that presents a unique opportunity to study genetic mutations and the medical disorders associated with them. Using such information, deCODE has the ability to impute genetic information about individuals without any legal requirement to obtain their informed consent.

Paper 3: *The Ethics of Big Data in Genomics: The Instructive Icelandic Saga of the Incidentalome*

Q1: The researchers used existing genotype data and genealogy databases to impute the genomes of those who were not part of the study, an approach that was ultimately stopped by the Icelandic Data Protection Agency. As a compromise, the researchers only kept imputed data temporarily to conduct analyses and then discarded the imputed genomes, thus not saving any individual data. What are your thoughts about this approach? Note that using the extra data led to many new scientific discoveries

Q2: When the researchers were not allowed to contact those who were heterozygotes for the *BRCA2* variant, they launched a website where Icelandic individuals could login to learn their results. What are your thoughts about this solution?

Q3: Would you want to be contacted if you were part of a cancer research project and researchers found that you were heterozygote for a high-penetrant variant? What if you were not aware of the study but researchers had identified you because of your relation to family members who participated?

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