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# Ethics in personalized genomics; an overview of the issues

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# Outline

- Focus on issues related to direct to consumer genetic tests.
- What are the main ethical arguments?
- Where do they lead us in relation to policy?



## The context

- The ethical debate is heavily influenced by the fact that there is *uncertainty about the clinical validity and utility* of the information that such tests generate.
- “Direct to consumer genetic testing: ***Avoiding a culture war***”



# The context

- i.e. ESHG: communicated to the public that DTC genetic testing is neither accurate nor beneficial to consumers. (ESHG, press release 2011  
<https://www.eshg.org/13.0.html>)
- ESHG statement
- ASHG statement
- FDA recent review of DTC companies and hype in the media



## Principles and values

- Respect for autonomy
- Beneficence
- Non-maleficence
- Distributive justice /equity
- The state's responsibility to protect
- Right to know/ right not to know



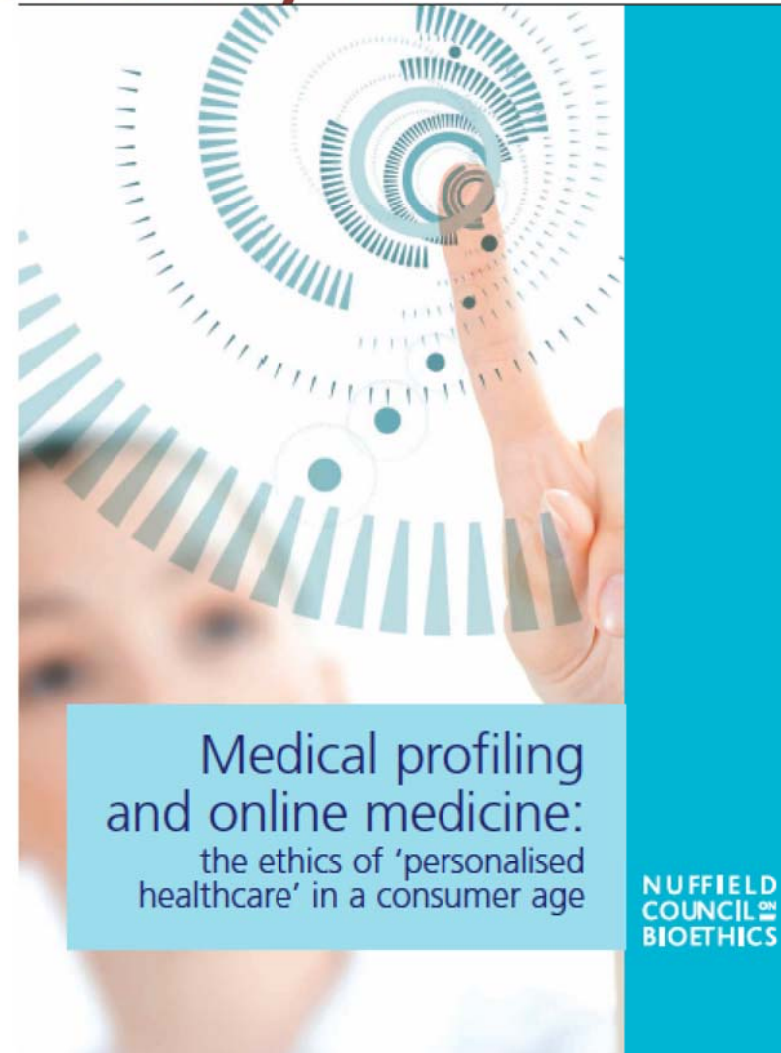
## Respect for autonomy I

- Self-determination.
- Access to direct to consumer genetic tests provides an opportunity for action to improve health (or inaction).
- Lack of access deprives one of the opportunity to act.



## Respect for autonomy II

- “consumerism”
- “responsabilization”
- Purchasable health goods
- Increased responsibility in one’s own health
- Empowerment of patients



# Respect for autonomy III

- Action or inaction related to the quality of information:
  - accurate
  - impartial
  - balanced
  - evidence-based
  - accessible
  - well written





# Respect for autonomy IV

- Safeguard: informed consent
  - Comprehensive and comprehensible information
  - Valid consent
  - Protect the vulnerable
- Safeguard: Gatekeepers, counseling
  - Point of contest

# Respect for autonomy V

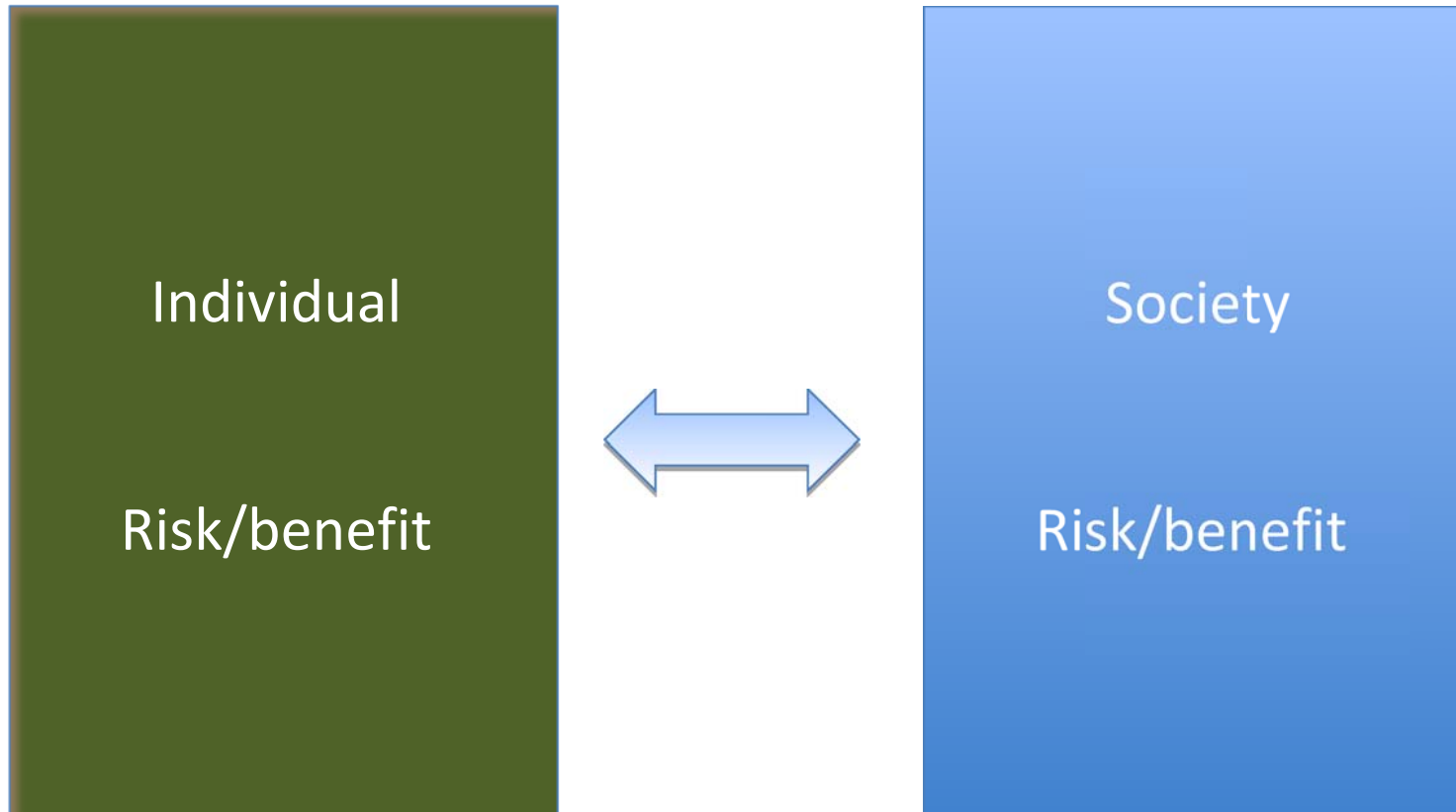
- Genetic test without one's permission

- Genetic tests of minors
  - Diminished autonomy
  - Evolving capacities of minors
  - Risk/benefit (especially if lack of counseling)





# Beneficence/non maleficence I



## Beneficence/non maleficence II

### Benefits\*

- Impact: life style change (personal control)
- Alert family members
- Increase genetic literacy
- Health care cost reduction if positive life-style changes and/or prevention leads to healthier individuals
- Reduce health care costs through better medicines
- Contribution to research

## Beneficence non/maleficence III

### Harms

- Incurred costs to individuals
- False reassurance or misplaced anxiety (fatalism)
- Potential for genetic discrimination
- Privacy, confidentiality
- Increase health care costs if information leads to additional interventions (unnecessary tests, physician time)
- Genetic determinism

## Beneficence/maleficence IV

- Risk/benefit?
- Do we have a good answer?
- Do we have data?
- Why is this relevant?
  - Moral issue of do no harm (or good than harm)
  - Policy implications (if decisions based on falsely derived ratio)



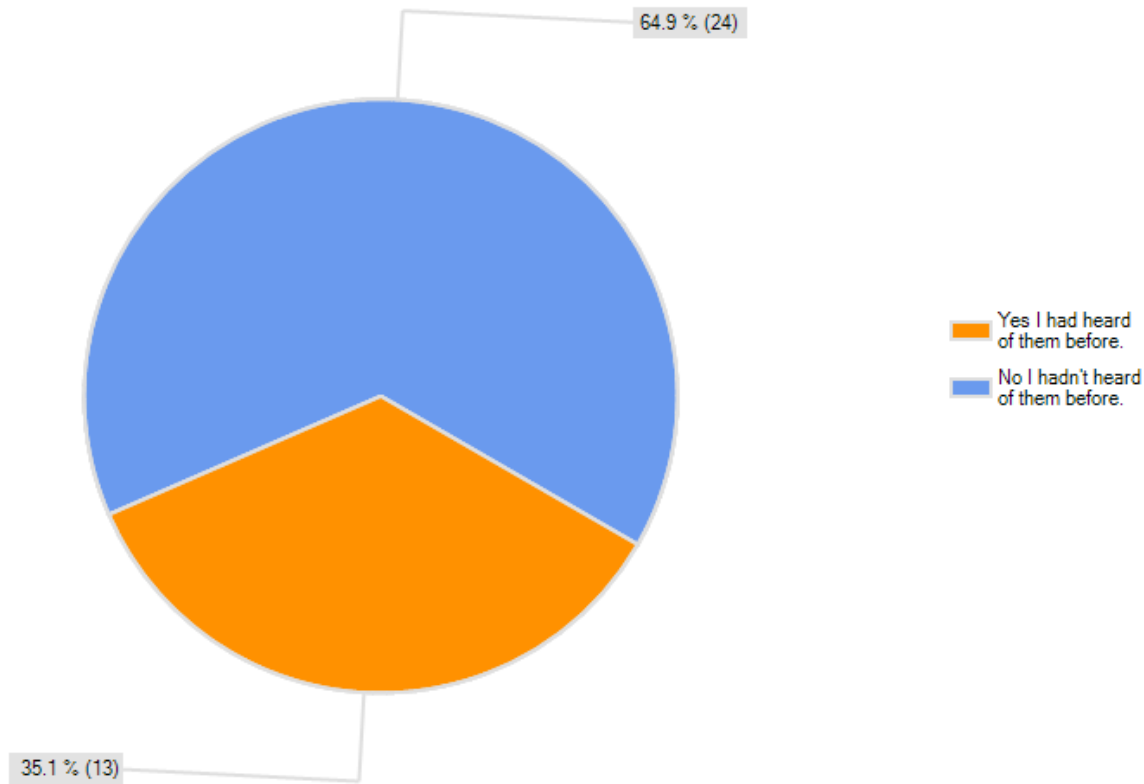
# Evidence

- Evidence about potential harm, or impact on behavior
- Probably leaning towards null hypothesis
- Caution regarding how the available evidence can be used
- Need for more evidence



# Preliminary results/Swiss study

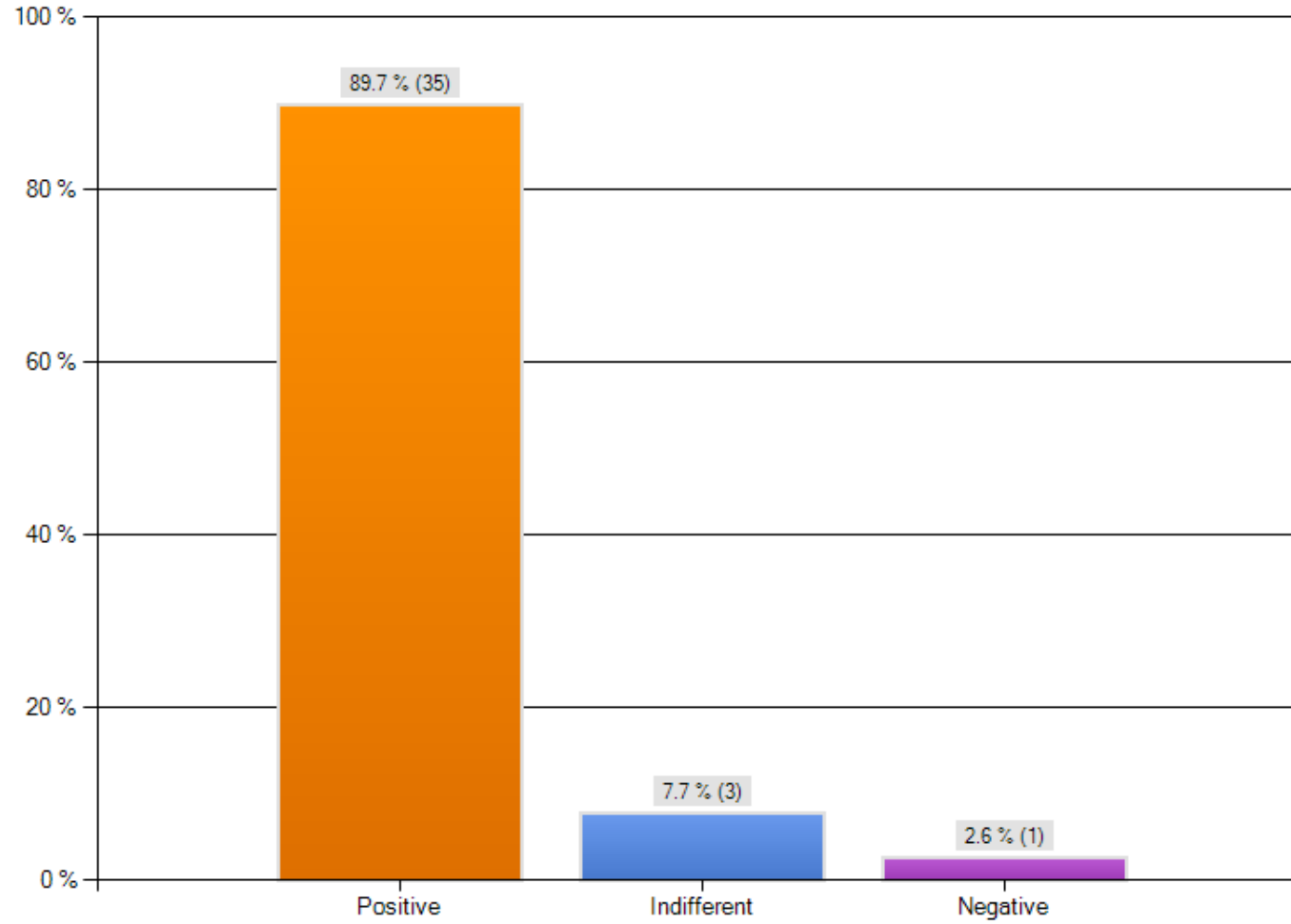
Had you heard of DTC genetic testing before the seminar/talk?





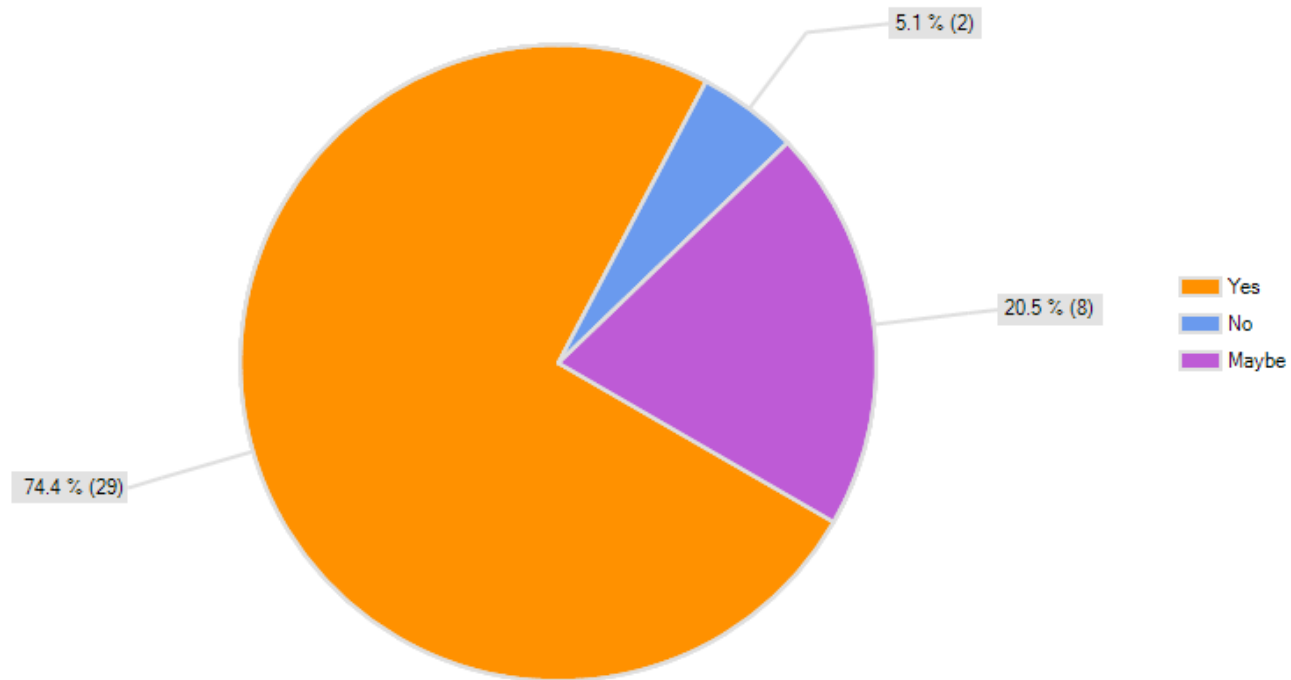


How would you rate your overall experience with doing this test?



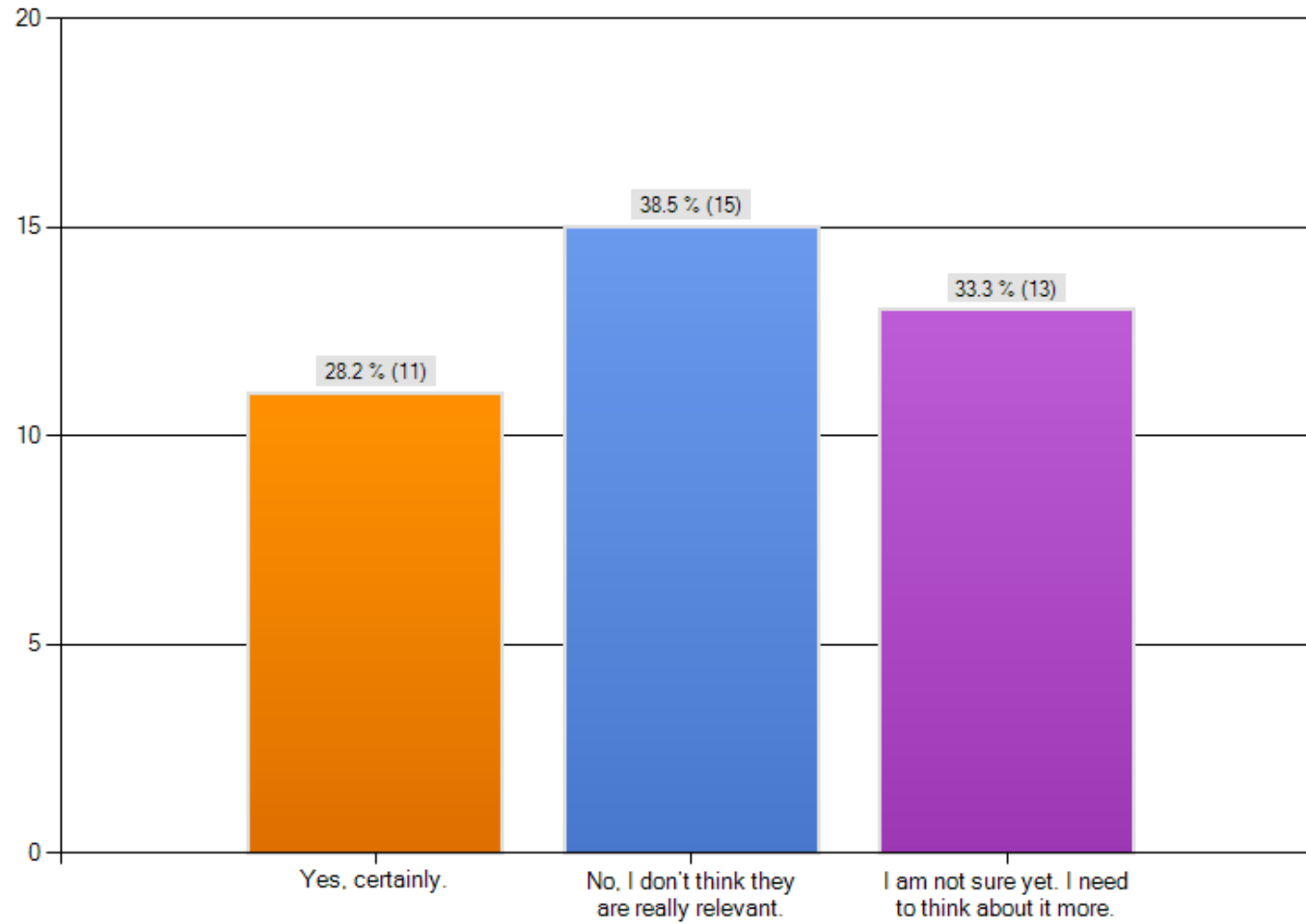


Would you recommend this test to a family member or friend?



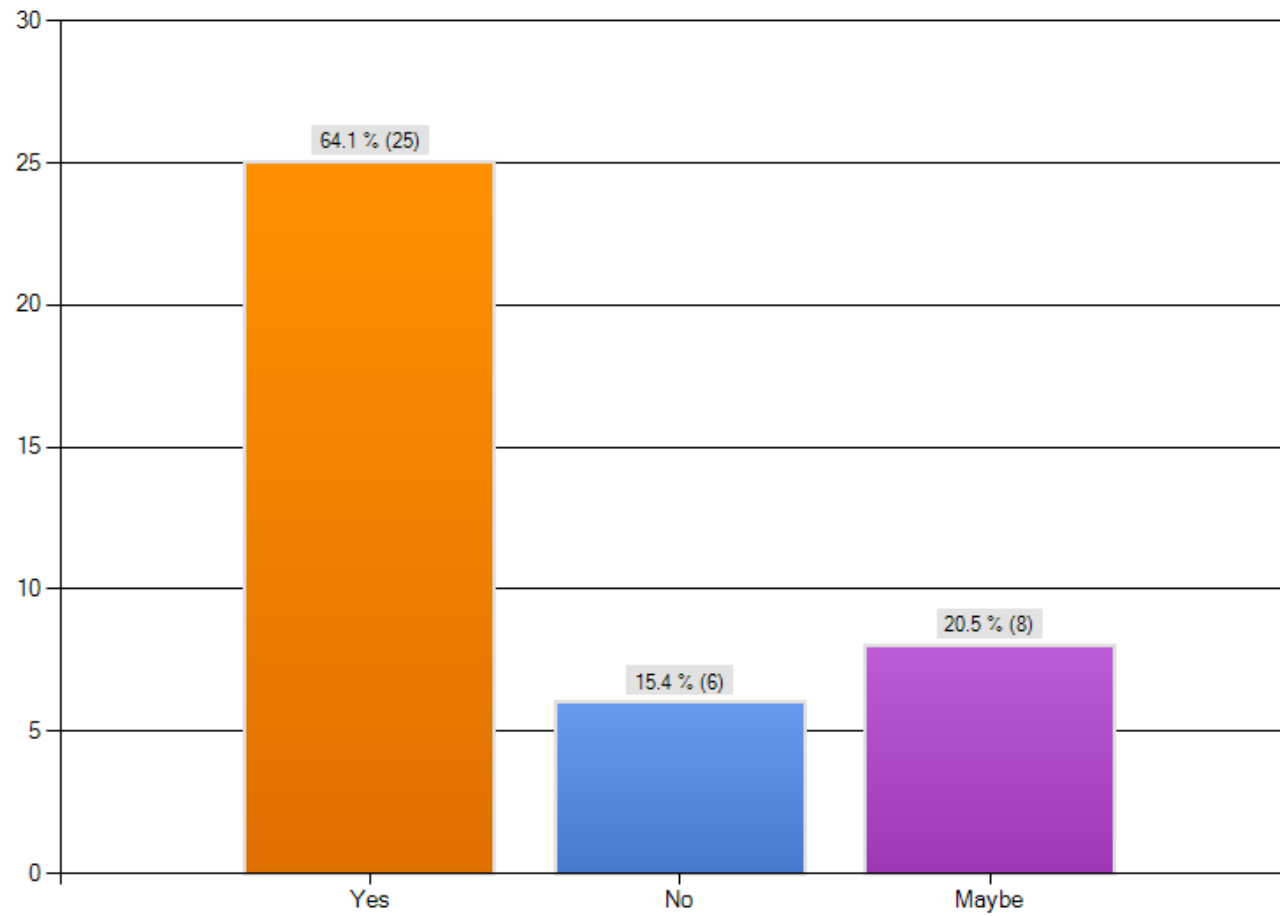


Do you intend to show your test results to your doctor?



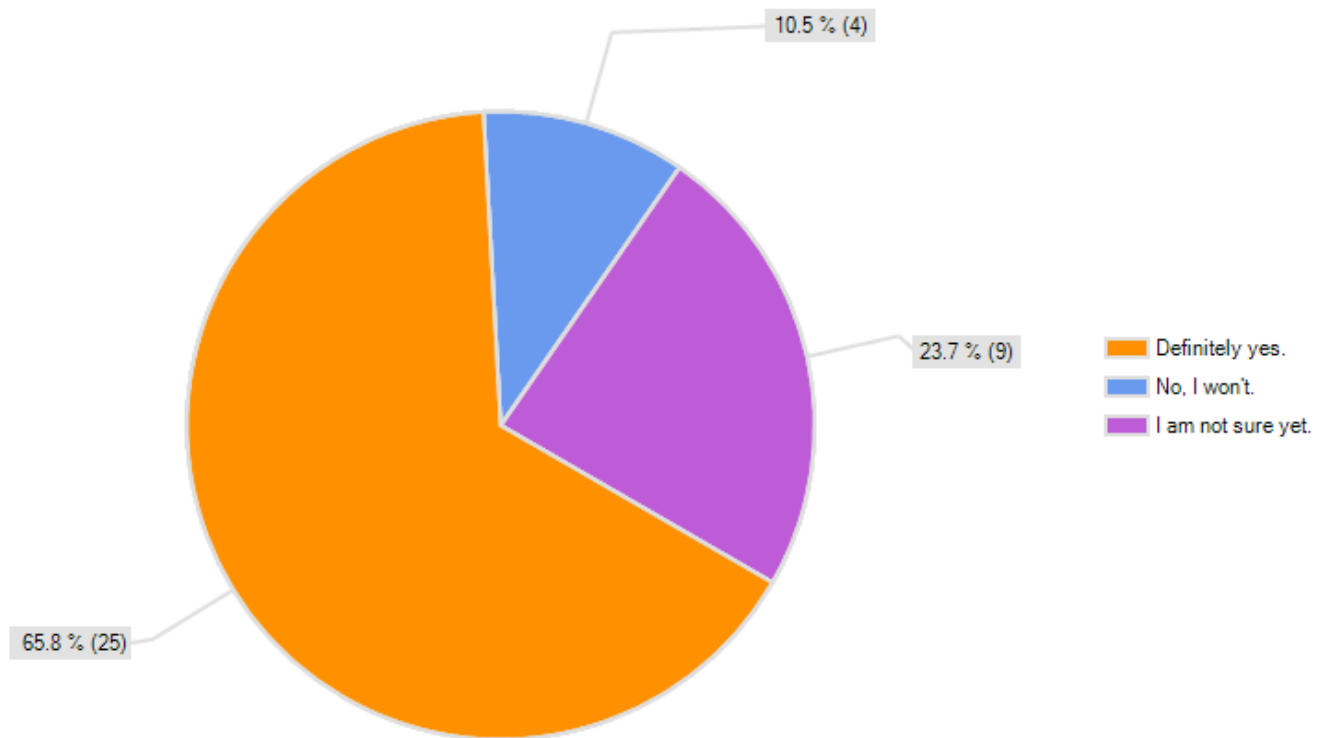


Are you planning to discuss your results with your family?





Do you intend to participate in the surveys offered by 23&We?





- Privacy concerns
- How well are genetic data protected by the companies?
- Is the legal protection adequate?
- Is our notion of privacy changing?



# Research

- Participation in research that DTC companies conduct.
- Is the research participant protected?
- Do mechanisms we already have in place for protecting human subjects suffice?



# Web-Based, Participant-Driven Studies Yield Novel Genetic Associations for Common Traits

Nicholas Eriksson<sup>1\*</sup>, J. Michael Macpherson<sup>1</sup>, Joyce Y. Tung<sup>1</sup>, Lawrence S. Hon<sup>1</sup>, Brian Naughton<sup>1</sup>, Serge Saxonov<sup>1</sup>, Linda Avey<sup>1</sup>, Anne Wojcicki<sup>1</sup>, Itzik Pe'er<sup>2</sup>, Joanna Mountain<sup>1,3\*</sup>

## Editorial

# Consent and Internet-Enabled Human Genomics

Greg Gibson<sup>1\*</sup>, Gregory P. Copenhaver<sup>2,3</sup>





“The editors of PLoS Genetics recognize that the decision to publish this study, without IRB review as human subjects research and with some concerns over the consent document, and the fact that there is limited access to the raw data, will not sit well with some, perhaps many, readers...

In so doing, we call for community input to spur efforts to standardize the IRB consent process for GWAS research.”



# Research

Research participation as social responsibility

Moral duty?



*H1N1 Vaccination trials /Photos from CNN on line edition*



## Distributive justice

- Equitable access
  - Within a country and at global level
  - Resource allocation issues
- Research participation
- Focus on certain populations, certain diseases
- The 10/90 health research gap



Most GWAS done one population of European descent (96%).

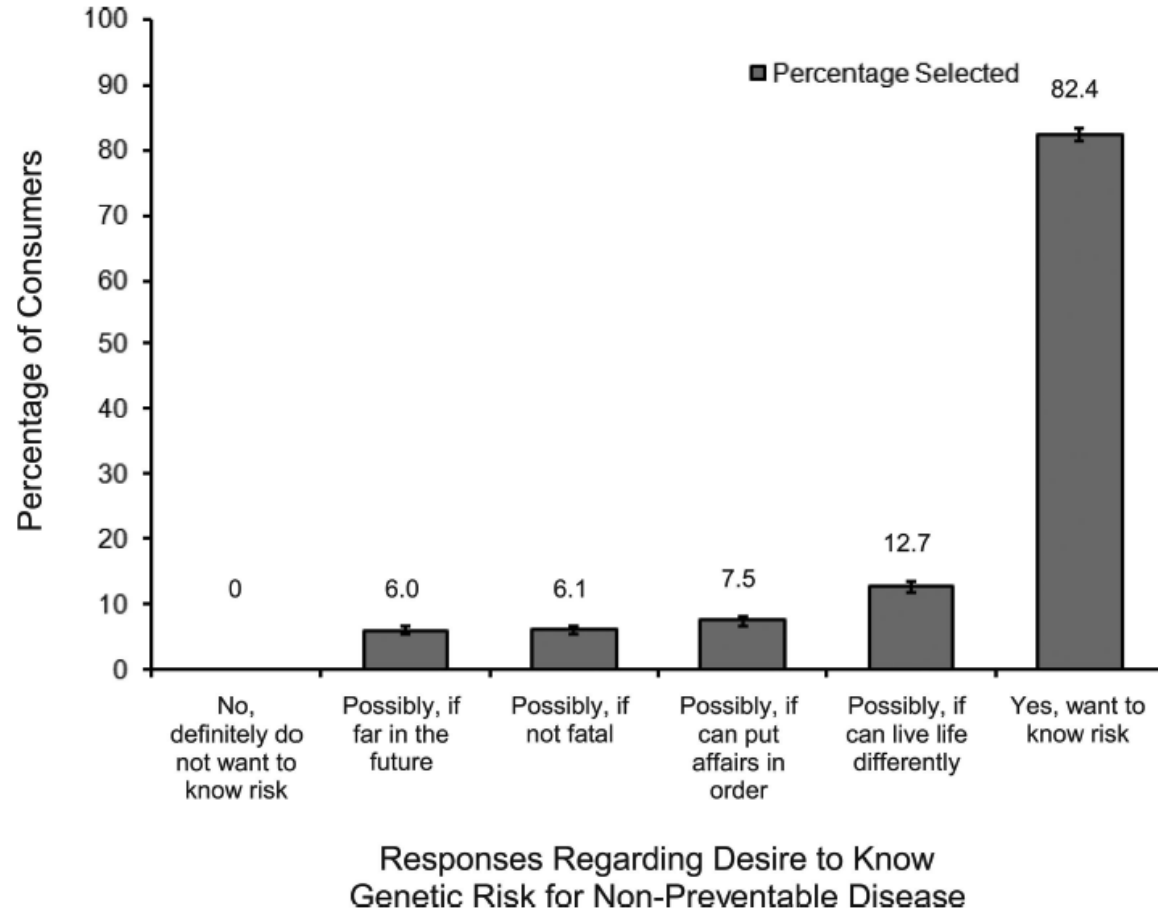
“It is tempting to focus on populations that are motivated, organized, medically compliant and otherwise easy to study. But by failing to develop resources, methodologies and incentives for underserved people, we risk perpetuating the health disparities that plague the medical system. Those most in need must not be the last to receive the benefits of genetic research.”

Bustamante et al., Genomes of the world. Nature 475, 163–165 (14 July 2011)



# Rights?

- Right to know genetic information
  - Medical information
  - Right to know enhances autonomy
- Right not know
  - Especially when lack of treatment is available



# Paternalistic approach?

- Is there a state responsibility for protection of harms from DTC GT?
- State protection versus personal liberties
- Genetic exceptionalism



Extreme libertarianism  
(all GT allowed)

Extreme conservatism  
(none allowed)

A diagram consisting of a horizontal blue line with a light blue circle centered on it. The circle is divided into two halves by the line. The left half of the circle is labeled 'Extreme libertarianism (all GT allowed)'. The right half is labeled 'Extreme conservatism (none allowed)'. The bottom half of the circle contains the text 'Intermediate position (treat as other medical tests Regulate aspects of the process)'.

Intermediate position  
(treat as other medical tests  
Regulate aspects of the process)





## Summary

- Developments in personal genomics challenge ethical and social norms
- There are conflicts of principles
- Need for caution in how arguments are used
- Need for identifying and evaluating the real ethical concerns early and iteratively in the process.