Ethical Aspects of Direct-to-Consumer Genetic Testing

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Ethics...bioethics...ELSI
A little Thursday morning survey…

• Who would you go through a DTC GT company to get ONE genetic test?
• Who would like to get a whole genome scan done?
• Why? What would you do with the information?
• Would you have your kids tested?
1- DTC GT

2- Ethical and Social Aspects:
   A. Company claims/potential benefits
   B. Ethical and social concerns
1. DTC GT

Human genome research begins to pay off...

Normal DNA

Oil company executives DNA

That explains everything.
• Some basics: lions, tigers & genes Oh my!
• What is DTC GT?
• What are the companies?
• What tests are being offered?
• Dynamics of the market
More about DTC market existence…

- Offer & Advertising
- HUGE variation!
- Not bc there is necessarily a need!
- Potential for profit
- Riding the wave of hype, desire to use info generated by projects like the HGP
- Consumer/patient interest in self-mediated health care
- Health promotion not health care -- disclaimers
- USA residents being used to paying out of pocket
- Right to know
- Relatively few barriers to market entry
What about consumers & DTC GT?

• 2007 survey by Goddard et al.
  – 14% aware of nutrigenomics tests
  – 0.6% had used them
  – Young, educated, high income

• 2009 survey by McGuire et al.
  – 6% (63/1087) had used DTC personal genome testing
  – 63% would use DTC PGT
2. Ethical & Social Issues

ONCE YOU UNFOLD ONE OF THESE THINGS, IT'S NEVER THE SAME.

MAP OF THE HUMAN GENOME

Ethical Questions
Legal Tangles
Privacy Concerns
Moral Issues
Medical Dilemmas
Insurance Applications
A. Claims/Potential Benefits

1. Increased access
   – Is this true? Is there a REAL NEED?
   – No empirical data yet
   – There are few GCs though...

2. Increased autonomy & empowerment
   – True for good quality information & services
   – Just bc people want something should they have it?

Can 1 & 2 can be achieved without commercialization?!
3. Ability to alter life-style to reduce known disease susceptibilities (i.e.: better health)
   – WILL consumers change habits?
   – Is genetic risk different than risk due to obesity?

4. Increased confidentiality (since results are not part of “official” medical report.)
   – Technically this is false
B. Ethical and Social Concerns

1. Analytical, Clinical validity and utility
2. Adequate counselling
3. Research agenda
4. Testing in children (& Testing of third parties)
5. Privacy and confidentiality
6. Challenge to public health care system
7. Informed Consent
8. Lack of adequate regulatory framework…
9. Equity and equal access
1. Validity & Utility of DTC GTs

- **Analytical validity**
  - Lab techniques are very good = Least of our worries

- **Clinical validity**
  - How good/strong is the association? **VARIES A LOT!**
  - Monogenic vs. complex trait

- **Clinical utility**
  - Is it useful for individual to get this information? **NOT FOR full genome testing!** (Janssens et al.)
  - Does it provide concrete treatment/lifestyle change etc…? **EAT RIGHT, EXERCISE, REDUCE STRESS!!!**
  - Even with this info will consumers change habits?
2. Genetic Counselling

- GC is generally recommended both pre & post testing (clinical setting)
- Considered one of the most expensive aspects of DTC GT, not many GCs in USA and Canada
- Most companies **do not offer GC**
- Consumer misunderstanding may lead to **too drastic or too lax measures**
- Understanding for family members
2. Genetic Counselling cont...

- How can a GC hired by a company be impartial or non-directive? (think pre-test)
- Would GC even understand full genome scan results? How will consumers understand?
- How could GC explain changes in risk profile with time?
- Causing re-questioning of role of GC in GT… not a bad thing!
3. Research Activities cont…

- Companies performing full genome testing also use consumer data for research
- Not always transparent
- Gene Essence “We may use your Necessary Profile Information and any Optional Profile Information that you provide to advance the science of genetics and provide you with a better understanding who you are genetically.”

- SeqWright “Please volunteer any diseases from which you currently suffer (this can help us advance medical research by enabling us discover new SNP/Disease associations).”
Even for those that are open about it:

https://www.23andme.com/researchrevolution/compare/
3. Research Activities

- Are consumers aware?

- Information given by companies is barely existent, found in "small print", or simply confusing (ie: 23andme)

- Is this informed consent? Are consumers becoming research subjects "willingly"?

- Do companies have Ethics Review Boards? If they did have them, would the ERB have veto power over research??
4. Testing in Children

- Risk of non-consensual testing of a third party is always a risk… but what about in minors?
- Some companies do perform genetic testing in minors

- Predictive genetic test are recommended
  - when “established, effective, and important medical treatment” can be offered
  - should be delayed until the person is old enough to make an informed choice
DTC GT in minors

- ESHG statement on DTC GT
  "...the very context of DTC genetic testing does not allow for an adequate assessment of the competence of minor. Therefore, the ESHG considers that DTC genetic tests should not be offered to individuals who have not reached the age of legal majority."
Study I: Content Analysis

Nature Reviews Genetics | AOP, published online 25 November 2008; doi:10.1038/nrg2501

Ethics watch

DIRECT-TO-CONSUMER GENOME SCANNING SERVICES. ALSO FOR CHILDREN?

Familial Cancer
DOI 10.1007/s10689-009-9253-9

Health-related direct-to-consumer genetic testing: a review of companies’ policies with regard to genetic testing in minors

Pascal Borry · Heidi C. Howard · Karine Sénécal · Denise Avard
Study II: Company survey: results

- 9/13 have received requests from parents (or LG) to test minors
- 10/13 DO perform GT in minors under parental (or LG) request
- Most do not base policy on established recognized policies (ESHG, AMA etc.)
5. Privacy and Confidentiality

- Technical aspects: saliva & DNA sample, personal info, data.
  - Where, by whom, and for how long are they kept?
  - How? Anonymized, coded?
- Third party access?
- Sharing information with “friends” & family
- What happens if company is sold?
6. Challenge to public health care system

- No data yet
- Survey out of Japan more about MD awareness
- European Survey under way
7. Informed Consent

**Personal Genome Testing: Do You Know What You Are Buying?**

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- See genetic counseling issues
- How much can consumers be expected to understand? For service? For research?
9. Equity

• Is knowing “your DNA” a right?
• If so, shouldn’t everyone be allowed to have it?
• For now, only for those who can afford it…
Let’s just remove the ethics gene...
DTC Advertising

• “The truth is direct-to-consumer advertising is used to drive choice rather than inform it”

Dr. Dee Mangin, Christchurch School of Medicine, New Zealand
“recreational” vs medical genetics

• Which is worse DTC?

Some say DTC full genome testing is a clear example of immature translation of technology and DTC monogenic tests if properly regulated via a HC professional is OK.

Vs

Others say the opposite: who cares about « recreational genetics » let the consumer waste his money, but leave the serious diseases out of DTC GT market
Thank you!

For more information:

http://www.dtcgenetest.org/

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Guidelines, laws and policies

DTGen is one of the modules of the HumGen International database (www.humgen.org), and as such, is a document repository of international, national and regional materials regarding the ethical, legal, and social issues pertaining to direct-to-consumer (DTC) genetic testing services. The academic, professional, and governmental publications gathered in the DTGen module all pertain to the marketing and/or offer of genetic testing services directly to consumers.

Since the completion of the sequencing of the human genome, there has been a steady increase in the number of genetic tests developed and offered via the traditional health care system. Furthermore, private commercial companies outside of the health care system have recently been offering genetic tests directly to consumers. This means that consumers can obtain genetic tests without ever seeing or speaking with a doctor or genetic counselor (or any other certified health care professional). This new model of genetic test provision has generated a great deal of debate and concern within the health care field and has the potential to have a concrete impact on consumers. For these reasons, research on DTC genetic testing is an important priority.

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