

Genetic testing and ethical dilemmas

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New challenges

- How might genetic testing change family medicine?
- What are some of the ethical challenges reported by family doctors from around the world?
- What are our patients up to?
- How do we support family doctors to include genetics in primary care?

As family doctors we will need to be prepared for the Age of Genomics

- Our patients will expect us to have the answers to their questions and to know what to do next
- As they always have

The 3 big challenges for family doctors in the Age of Genomics

1. Our knowledge of basic genetics
2. The lack of guidelines to assist us with screening and management of polygenetic conditions
3. The emerging ethical dilemmas

Ethical and Medicolegal issues

- It is clear that there are many ethical aspects to human genetic testing that we are going to face in medical practice.
- Many are encapsulated by the 3 'c's':
 - Consent
 - Counselling
 - Confidentiality

The 3 'P's of the era of IBM

Preventive medicine

Predictive medicine

Precision medicine

Preventive medicine is already
a core part of medical practice

Working with our patients in prevent ill
health or to diagnose serious
conditions early and prevent the
development of morbidity

Predictive medicine will become a core part of medical practice

- Advances in genetic testing will allow us to determine an individual's predisposition or susceptibility to numerous conditions, including cancers and infectious diseases.
- Comprehensive genetic testing is likely to become a standard part of medical practice.
- Our role will expand to include preventive management and genetic counselling of individuals and their families.

Precision medicine will become a core part of medical practice

- The ability to prescribe better preventive care before a patient begins to show symptoms.
- The ability to rule out drugs that are likely to have adverse or no effects on our patients and select those drugs designed to be most effective with minimum side effects.
- The ability to determine which of our patients will benefit most from which interventions.

- “Will this put medicine in a spin?”
- “If we have an idea of propensity for, say hypertension, then a family doctor can act early to prevent the condition or treat their patient.
- “We could see a revolution in prevention of long term morbidity and early mortality.
- “This seems comparable to the introduction of PAP smears.”

- “Is it irresponsible to screen for diseases that we can't do anything about?”
- “Should we have a new Golden Rule?”
- “Never screen in the absence of a remedy?”

- “What advice do I give my patients about privacy implications?”
- “Should we be worried about possible access to genetic information by employers or by insurance companies?”

- “What are the medicolegal consequences of giving explicit advice based on an as-yet imprecise science?”

- “This has serious potential for abuse.
- “I can envisage a patient seeking to terminate a foetus just because it has the propensity to develop an incurable disease at some time in the future.
- “There are considerable ethical consequences.”

- “As doctors our patients will expect us to have answers to their questions.
- “Commercial interests will develop in this area with the potential to prey on our unsuspecting patients.”

- “New genetics can learn from old - pre Human Genome Project genetics.
- We need to get back to basics.
- As doctors we are experts at taking and interpreting family histories. We know how to provide high quality counselling about tests for haemoglobinopathies and haemochromatosis.
- We need to build on our strengths.”

- “Will access to genetic screening at the age of 18 have a positive or a negative impact on young people.
- I can see people making choices about their future career based on possible genetic predictions.
- Will we see “bad back” genes which will prevent people training to be nurses, or “panic attack” genes which will prevent people training to be airline pilots?”

- “I am concerned about likelihood ratios.
- “For example, can I smoke or not?
- “Maybe we will find that some people can!
- “But what about the false positives and false negatives?
- “How accurate will these tests be?”

- “If family doctors don't accept responsibility for this, then others will.
- “ Gene testing quacks may start to appear.”

- “Genetics is the sexiest subject available to medics.”

- “This is all very interesting.
- “But if we don’t do something about climate change, this may all be irrelevant.”

Many emerging ethical issues

- Privacy of genetic samples
- The risk of discrimination
- Control of human genetic research
- Role of human research ethics committees
- Ownership of genetic materials
- Human genetic research databases

Many emerging ethical issues

- Responsibilities of health professionals with family genetic information
- Requests for parentage testing
- Use of genetics for immigration
- Use of genetics in identifying or selecting athletes for specific sports
- Is non-consensual genetic testing a criminal offence in your country?

Genomics may turn ethics upside down

- Genetic information cannot be de-identified
- It remains linked to an individual
- It is also linked to that individual's genetic relatives
- Should we destroy an individual's personal genetic information, when requested by the individual concerned, when such an action could adversely affect the health management of that individual's genetic relatives?

Informed consent and disclosure

- Informed consent needs to take into consideration that genetic samples can reveal information about other relatives, in addition to the individual being tested
- Should Privacy Laws allow a health professional to disclose genetic information about a patient to a genetic relative where the disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety, even where the threat is not imminent?

Possible uses of genetic information which could harm individuals

- Impact on families where one member starts to exhibit symptoms of an inherited condition
- Or where other members without the gene feel guilt
- Potential access to information by insurance companies
- Potential access to information by employers

Possible uses of genetic information which could harm individuals

- Will financial institutions wish to access genetic information to determine eligibility for a loan?
- Will potential adoptive parents wish to know a child's genetic status prior to adoption?
- Will genetic information be used by government to determine eligibility for health services?
- How to balance the risk of psychological harm?

The right not to know

- It cannot be assumed that everyone will wish to know that they might have inherited a disorder present in their family.



- Home gene testing
- 87,900,000 results
- Let's look at the top five
- Top 2 are highlighted “sponsored links”

www.dnabioservices.com.au

DNA Testing of all types, our simple DNA test offers you Peace of Mind concerning all **DNA Paternity Testing** issues, DNA tests can be carried in the privacy of your own home with our DNA test kit.

“No more living in doubt”



“I'm married with two children, but am separated from my spouse. I've been paying child support for five years. Thanks to DNA Bioservices I was able to do a private DNA test on both of my children.

I was so devastated when I got the Paternity Analysis Report that I thought I was going to die.

The Paternity Analysis Report read that the probability of paternity was 0% for both of my children.

I took this case to court and the Paternity Analysis Report served as evidence. The court dismissed the child support and freed me of all responsibilities.

Thanks again DNA Bioservices.”

Cost

- Kit is free
- US \$430 for paternity testing of father and one child

Cost

- \$US 199 for DNA testing and personal Genetic Ancestry Analysis including Genetic Profile, Native and Global Population Match and World Region Match analyses.
- The Genographic Project of National Geographic is only \$US 99.

- Testing for drug reactions
 - When it comes to medications, one size doesn't fit all. Your genes may affect how you respond to certain drugs.
- Testing for fertility and pregnancy
 - Looking for carrier screening? Having difficulty getting pregnant?
- Testing for lifestyle issues
 - If you carry certain genes, you can take steps to live longer and healthier.
- Testing for signs and symptoms
 - Do you have a chronic, undiagnosed condition? It could be genetic.
- Testing for ethnic risks
 - Are you more or less likely to carry certain disease-related genes?



TESTS & SERVICES WE OFFER

Alpha-1 Antitrypsin Deficiency
Ashkenazi Jewish Carrier Screening
Blood Clotting Disorders
Breast & Ovarian Cancer
Colon Cancer - PreGen Plus™
Cystic Fibrosis
Diabetes - deCODE T2™
Drug Metabolism (CYP2D6 Gene)
Hemochromatosis (Iron Overload)
Infertility
Recurrent Pregnancy Loss
Tamoxifen

Fertility and pregnancy

- Are you planning a family?
 - [Cystic Fibrosis](#)
- Are you trying to get pregnant, but have not had success?
 - [Infertility](#)
- Have you had two or more miscarriages?
 - [Recurrent Pregnancy Loss](#)
- Have you had pregnancy complications (preeclampsia, placental abruption, IUGR)?
 - [Blood Clotting Disorders](#)

Lifestyle risks - recommendations

- Are you age 50 or older?
 - [PreGen-Plus™ Colon Cancer Screening](#)
- Are you physically inactive or unable to regularly exercise?
 - [Diabetes Risk \(deCODE T2™\)](#)
- Are you planning travel or a surgery?
 - [Blood Clotting Disorders](#)

Cost

- US \$250 for 2D6 Drug Metabolism test
- US \$260 for cystic fibrosis test
- US \$995 for male infertility test
- US \$575 for colon cancer test
- US \$500 for type 2 diabetes test
- US \$380 for blood clotting (V Leiden) test
- US \$330 for Alpha-1 Antitrypsin deficiency test
- US \$199 for haemochromatosis
- US \$1266 for Tay Sachs Disease

- **TOTAL US \$4755**

CBS news report

- **Home Genetic Tests For Disease**
- **Send In Cheek Swab, Learn Your Disposition For Disease Online**



www.genetests.org

- a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons
- Funded by the U.S. National Institutes of Health

What else?

- Next five sites are all for companies offering paternity testing
- Sponsored links down the right side of the page include:
 - 6 ads for paternity testing
 - 1 for DNA bird sexing
 - 1 for testing for Alzheimer's Disease

DNA bird sexing

- **Pull 2 feathers** from the bird with one swift motion and place them in the bag or envelope labeled for that bird.
- **You cannot use feathers that have been picked up off the floor.**
- Send in:
 - Feathers
 - Payment US\$25 per bird
 - Sample Form
- Each bird sexed receives its own DNA sexing certificate.

- **Alzheimer's Genome Test NOW AVAILABLE!**
- Check your future susceptibility **BEFORE** symptoms occur.
- Pre-emptive insight into one's genetic predisposition can empower and allow for proactive prevention.
- No blood sample required.
- Results available in 3-4 weeks by mail or fax.
- This test for Apolipoprotein (E) Genome needs to be done **ONLY ONCE IN A LIFETIME.**

Cost

- US \$280 for the Alzheimer's Disease test

Human Sports Performance Gene ACTN3

- A fast, simple and painless genetic test can identify whether you may be naturally geared toward sprint/power events, or towards endurance sporting ability.
- Regardless of whether you are an accomplished athlete, or a beginner, your **ACTN3 Sports Gene Test**® result could assist you in optimising your training to make the most of your natural ability within a wide range of sports.
- If you are an Australian resident, you can also pick up a test kit from a [participating healthclub](#).
- Cost \$110.00

How much have I spent?

- \$430 paternity test
- \$199 ethnicity test
- \$4755 medical tests
- \$280 Alzheimer's test
- \$110 Sports Gene test

- **TOTAL US \$5774.00**

- Nothing I've done has relieved the growing pain in my epigastrium

So what do I do now?

- I now need to see my own GP
- We need to sit down together so I can receive help on how to interpret my test results
- Are these tests scientifically sound?
- Do I have to change my lifestyle?
- Do I need more tests?
- Will my GP have the answers?

Some initiatives to support doctors

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www.genome.gov

National coalition of health professional education in genetics

www.nchpeg.org

- Core principles and competencies
- Links to specific training offered through the American Academy of Family Physicians and other organisations

We need the capacity to harness the benefits of this new technology

- We need to ensure clinicians have ready access to the best available evidence about the clinical implications of genomics to support clinical decision making.
- This includes support for access to sources of clinical evidence and IT tools to enable us to assist our patients to interpret what they find and are told.

- There are many broader moral and philosophical issues which I have not had the opportunity to address in detail.
- Some commentators believe that for the decades to come we shall probably only have the most rudimentary understanding of the genetic contribution to complex characteristics, knowing already that awareness of an individual's genetic code is likely to allow only the crudest of predictions about those characteristics in a person.

And what special impact will the Age of Genomics have on you and me?

And what special impact will the Age of Genomics have on you and me?

- Ronald Bailey is the author of *Liberation Biology*
- He believes that those of us who are 50 years old today have a good chance of living another 100 years, thanks to the emerging ability to splice improved artificial human chromosomes into our genes.
- I look forward to seeing you all in 92 years time at the 99th Annual Conference of the Lebanese Society of Family Medicine in the year 2100.

Whatever happens ...

- Our important work as family doctors will continue.
- Never forget that we are privileged to work as family doctors.
- Each of us makes a positive difference to the lives of our patients every single day.