

Ethical and Medicolegal Issues in the Age of Genomics



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New challenges



The era of “Information-Based Medicine”

The IBM era

As family doctors we need to be prepared

- Our patients will expect us to have the answers and to know what to do next
- Wonca is leading the world in addressing ethical issues in the age of genomics through the work of our Special Interest Group on Ethical Issues chaired by Professor Manfred Maier

The 3 big challenges for family doctors

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

**So how will the Age of Genomics
impact on our consultations?**

Welcome to 2010

The 3 'P's of the era of IBM

Preventive medicine

Predictive medicine

Precision medicine

Predictive medicine will become a core part of family 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 Family 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 family medicine

- 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.

Ethical and Medicolegal issues

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

What do our colleagues have to say?

- “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.”

- “Will knowledge of their genetic profile have a positive or negative influence on our patients?”
- “I am worried about the impact.
- “And especially the possible negative effect on adolescents.”

- “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?”

- “Will this really add further information to what we already know?”
- “Or will it end up with people have lots of tests – like the Battery of Test Model used by junior doctors in emergency department?”

- “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 family doctors we need to stake a claim in this area.
- “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.”

- “I imagine there will be different use in different nations.
- “Will genetic testing be used by some countries to determine whether someone can migrate or not based on the potential to develop some disease in the future?”

- “New genetics can learn from old - pre Human Genome Project genetics.
- We need to get back to basics.
- As family 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?”

- “Our governments need to educate the public about genetics.
- “Otherwise we risk overburdening family doctors with responsibility for all of the education of all of our patients.”

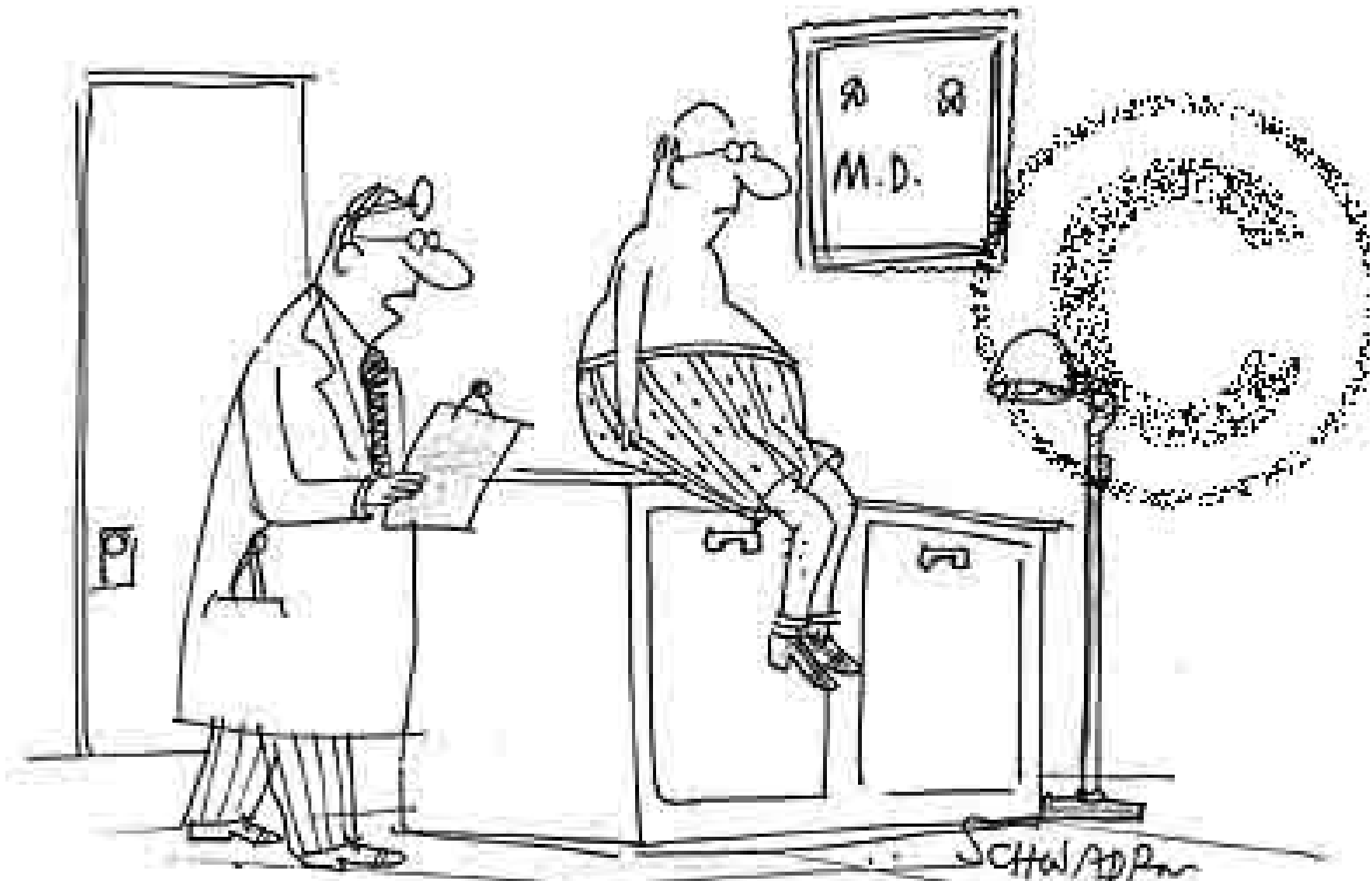
- “We need to remember that genes are not always “bad“.
- “Some may express good health characteristics.
- “For example the presence of a schizophrenia gene may be found to exist in all highly creative people.
- “There is a risk we could terminate the next Mozart or the next Einstein.”

- “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.
- “Tertiary hospital genetic counselling clinics will try to manage it all and they will fail.
- “Even worse gene testing quacks may appear.”

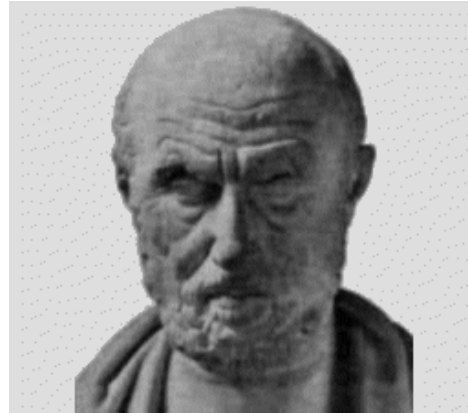
- “Genetics is the sexiest subject available to medics.”

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



"YOUR DNA TEST SHOWS YOU'RE PREDISPOSED TO SUE DOCTORS."

- My prediction back in the 2001:
- “In the trade display at the WONCA conference in 2007, 50% of all exhibitors will be gene testing companies - buy some shares now!”
- Was I right?



We have a duty to ensure privacy and confidentiality as part of our use of any new technology

“Whatever I see or hear, professionally or privately, which ought not to be divulged, I will keep secret and tell no one.”

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?

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?

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.

What can our patients do today at home, without us?





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



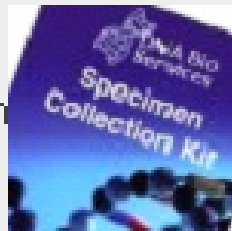
www.dnabioservices.com.au

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.



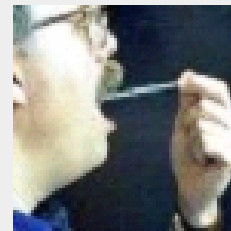
1. Order Test Online

You will receive your kit overnight in the mail



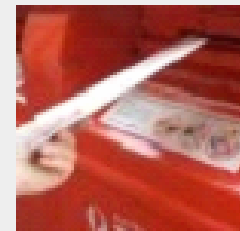
2. Swab Cheek

Swab the cheek of all the individuals to be tested



3. Return Kit

fill out the forms on the envelopes & return all together by mail to our office



Results in 5 - 10 days

The Results will be with you in 5-10 business days by mail, email or phone



Testimonials

“A girl whom I'd dated off and on came to me with the news that she was pregnant. Before any decision could be made I had to KNOW for sure if the child was actually mine. I called DNA Bioservices, I told them that I wanted to know if this child was mine without the mother knowing about it. The report came back telling me that the child was in fact 99.998% mine. I can finally look into her eyes and feel at ease.”



“No more living in doubt”

“I'm married with two children, but am separated from my spouse. I've been paying child support for more than 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.”

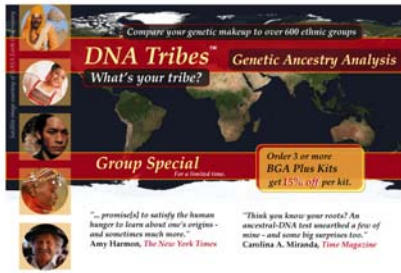


Cost

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

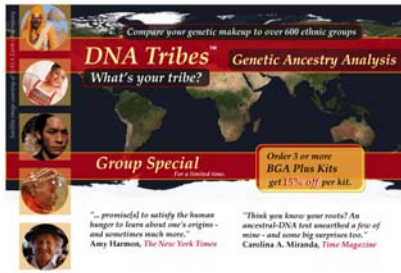
www.dnatribes.com





What is it?

- DNA Tribes Genetic Ancestry Analysis is a service that uses genetic material inherited from both maternal and paternal ancestors to measure your genetic connections to individual ethnic groups and major world regions.
- DNA Tribes identifies the peoples and places where your geographical genetic ancestry is most strongly represented. DNA Tribes is the only personal genetic analysis that compares your genetic profile to a population database that includes **over 193,000 individuals** from **655 populations** around the world, including **467 indigenous populations**.



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, as described by Spencer Wells, 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?

Get The Right Help From DNA Direct

- **DNA Direct** wants to make sure that you are doing the right thing for your health. We give you the impartial, professional medical advice you need.
- **There is never an obligation to buy a test.** You can use our consulting services without obligation and we will not try to sell you a test. Testing may not be right for you.
- **We offer advice before you take a test.** Getting the right test, for the right reasons, is very important. You can get qualified medical advice for just \$75 to \$150 depending on the condition. And most questions can be answered in a 30-minute session.



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

Drug Metabolism (CYP2D6 Gene)

- Approximately 25% of all prescribed drugs are metabolized by 2D6 enzymes, including some antidepressants, beta blockers, and pain relievers. These enzymes are produced by the 2D6 gene, which comes in many different forms.
- Some forms of the 2D6 gene metabolize drugs very quickly, while others don't process drugs as well. Some forms of the 2D6 gene don't metabolize drugs at all.
- **Knowing your genetic "profile" can help you and your doctor make sure you receive the right drug at the right dose for you.**

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](#)

Ethnic risks - recommendations

- European Caucasian (white)
 - [Blood Clotting Disorders](#)
 - [Cystic Fibrosis](#)
 - [Alpha-1 Antitrypsin Deficiency](#)
- Celtic, or Irish
 - [Hemochromatosis](#)

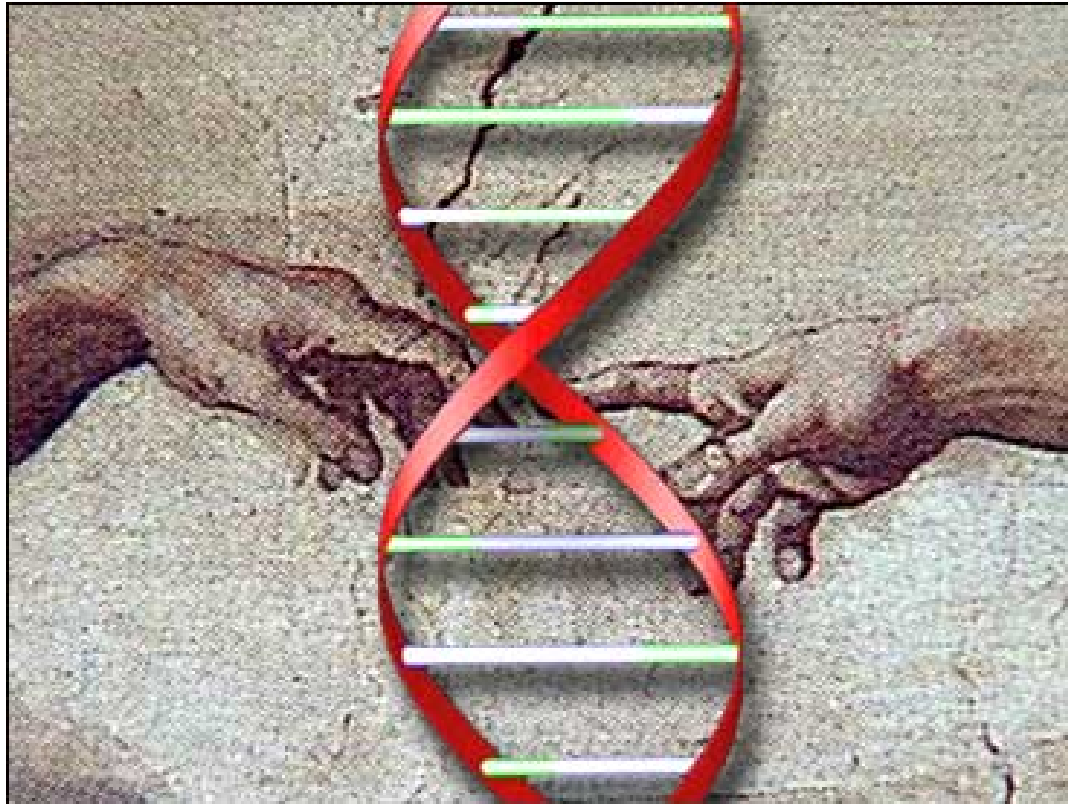
 - [Tay-Sachs Disease](#)
- Any ethnicity, and age 50 or older
 - [PreGen-Plus™ Colon Cancer Screening](#)

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 National Institutes of Health



At This Site

[GeneReviews](#)

Online publication of expert-authored disease reviews

[Laboratory Directory](#)

International directory of genetic testing laboratories

[Clinic Directory](#)

International directory of genetics and prenatal diagnosis clinics

[Educational Materials](#)

Including a link to the Genetic Tools website which provides materials for teaching genetics in primary care settings

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

How much have I spent?

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

- **TOTAL US \$5664.00**

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

So what do I do now?

- I now need to see my family doctor
- 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 family doctor have the answers?

Some initiatives to support family doctors



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

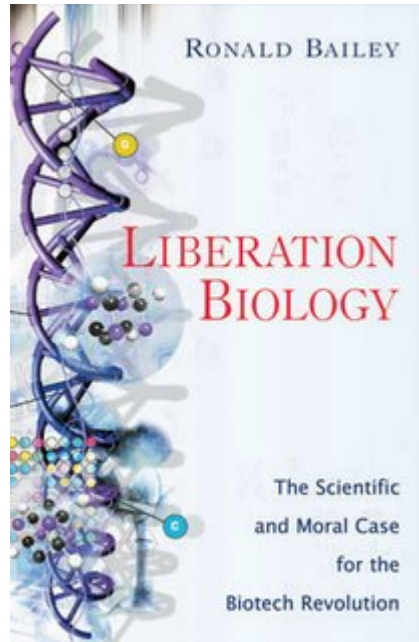
- There are many broader moral and philosophical issues which I have not had the opportunity to address in detail.
- Some commentators believe that for the decades to come we shall probably only have the most rudimentary understanding of the genetic contribution to complex characteristics, knowing already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

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

- We need to ensure our family doctors 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.

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 at the Wonca conference back here in Singapore in 93 years in the year 2100.
- But just in case he is wrong ...

I invite you to attend the 2008 Wonca Asia
Pacific Regional Conference in Melbourne
next October

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.

- The only thing we can say about the future for certain is that it is certain to surprise us.
- I hope all your surprises are pleasant ones.

