

REALITY CHECK Ray Moynihan

Beware the fortune tellers peddling genetic tests

We need urgently to evaluate and regulate the next wave of overmedicalisation

An enduring memory of my niece's third birthday party is the fortune telling session that took place under the dining room table. A creative parent had donned a headscarf and extravagant earrings, and soon a line of toddlers were waiting to hear about the magic of their future. Given the state of the science a decade later it's highly possible that this fortune telling was just as reliable as the high tech horoscopes arising from the marketing of genetic tests for common diseases.

When the US Government Accountability Office recently ran a covert operation on genetic tests for 15 common conditions, including Alzheimer's disease, breast cancer, and restless legs syndrome, it uncovered the most extraordinary mess.¹ It found that identical DNA samples produced wildly contradictory results. One donor was told by four different firms that he was at below average risk, average risk, and above average risk of having high blood pressure and prostate cancer.

Its report concluded that genetic tests marketed directly to the public were "misleading and of little or no practical use." Yet hundreds of thousands of people worldwide are sending off samples of their saliva in good faith and receiving predictions that can have life changing consequences on the basis of tests that remain poorly evaluated and grossly under-regulated.

For anyone concerned about the creeping medicalisation of life, the marketplace for genetic testing is surely one of the latest frontiers, where apparently harmless technology can help mutate healthy people into fearful patients, their personhood redefined by multiple genetic predispositions for disease and early death.

Certainly there's promise and hope among the hype, particularly when a rare single gene disorder may be involved. Researchers have used a genetic test to identify infants with a rare form of diabetes who benefited greatly from subsequent treatment.² Yet there are many examples of false hopes. Recent findings have provided no good evidence that genetic testing benefits people who have a history

of venous thromboembolism,³ those contemplating antidepressant treatment,⁴ or those wanting to identify their risk of developing cardiovascular disease.⁵

Again a tool that's proved useful in the laboratory has escaped like a virus into the marketplace, incubated by entrepreneurs, lazy reporters, and the power of our collective dreams of technological salvation, this time in the form of personalised medicine to treat us according to our individual genetic profiles.

Recent reports have welcomed the potential benefits of the different forms of genetic testing but warn governments to develop rational policy responses, to set enforceable rules for evaluating the technology, and to regulate its marketing.

A Nuffield Council on Bioethics report found that test results can be unreliable, be difficult to interpret, and cause further unnecessary testing.⁶ It concluded that many claims for individualised diagnosis and treatment "seem to be overstated and should be treated with caution." A technology review commissioned by the UK Conservative Party found that results can be "inaccurate and misleading" and cited calls for "proper regulation."⁷

Just before Christmas the Cambridge based Public Health Genetics Foundation decried the hype associated with "premature interventions" and argued that the biggest challenge now is to generate an evidence base so that we know when use of genetic information can improve global public health in a "safe, effective and cost-effective manner."⁸ Likewise the medical literature is full of cautionary notes alongside the technology's celebration, with one international group of researchers proclaiming that it could be decades before personalised medicine tailored to genetic profiles became a reality.⁹

The genetics researcher David Melzer and colleagues argued in the *BMJ* in 2008 that the science of genetic predispositions for common diseases was still so uncertain that onlookers may view it as "genetic astrology, producing entertaining horoscopes."¹⁰ The authors concluded that marketing poorly evaluated tests in this evidence based age was an "unwelcome



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anomaly" and that "preventing misleading claims should also be a priority." Others have made the valuable point that getting the regulation right might be as important as the science itself.

Evaluating genetic tests is a complex business, requiring assessment of how well the test measures what it claims to measure, how well the genetic variation predicts actual disease, how useful the results are in terms of treatment, and what the social and ethical issues might be. Clearly there's potential for exaggerating the value of a genetic test, which is one reason Germany has imposed severe restrictions on direct to consumer testing. In the United States they're talking of a new test registry on a government website, raising immediate concerns that it could lend legitimacy to unproved and potentially harmful products.

In Britain a government advisory body recently released a set of principles that it hopes will be taken up as a voluntary code of practice—a pusillanimous response already criticised as helping facilitate marketing rather than ensure proper regulation. Meanwhile the not for profit group GeneWatch UK warns that genetic tests may be used to sell unnecessary preventive drugs to healthy people and suggests that the tests be restricted to situations that produce health benefits and are ethically just.

Professor Melzer believes that there's a much wider problem of poor evaluation of diagnostic tests. Governments, he argues, should simply create a kind of compulsory Wikileaks for tests, with full disclosure of evidence, "so people know what junk they're buying." As for my niece, she's now a flourishing teenager, still confident that the camel ride predicted under the dining room table will one day come to pass.

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Competing interests: RM has published books on the medicalisation of life, including the 2005 *Selling Sickness*. His latest book is *Sex, Lies and Pharmaceuticals*. See www.raymoynihan.net.

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LOBBY WATCH Jane Cassidy

ResPublica



Who are they?

Described by the *Guardian* as the think tank “du jour,” this precocious new kid on the block is just a year old. It was founded by its director, Phillip Blond, a one time theology lecturer and author of *Red Tory*. He believes that “broken Britain” can be mended by strengthening civil society and local communities.

The *Guardian*’s John Harris described *Red Tory* as “a critique-cum-credo that harks back to the old paternalist Conservatism that was all but obliterated by Margaret Thatcher, but is also aimed at providing an answer to an array of very modern problems.”

David Cameron was at the think tank’s launch, and the prime minister’s “Big Society” concept grew out of the *Red Tory* thesis.

Last year ended on a high note for Blond as he was named by the *Evening Standard* as one of London’s 1000 most influential people for devising his brand of “muscular civic Toryism.”

Meanwhile ResPublica picked up the “One to watch” prize at *Prospect* magazine’s 2010 think tank of the year awards.

What agenda do they have?

ResPublica can mean “commonwealth.” The think tank’s motto is “Changing the terms of debate,” and its core belief is that mutualism and localism have a central role in shaping a more equitable society.

So, it must like the government’s plans for health reforms, such as putting commissioning in the hands of GPs? Surely this fits with its ethos of devolving power to communities?

Actually it’s not impressed. Its associate director and head of housing, health, and environment, Matt Leach, labelled the proposals “timid.” Many aspects of the health service reform agenda look too much like a closed dialogue between bureaucrats and professionals, rather than one in which communities are included, he said.

He criticised the new GP commissioning consortiums, which he said were likely to operate on too large a scale and not necessarily in a way that involved local people. He also questioned whether the proposed new structures would be capable of enabling discussion across communities



ResPublica director Phillip Blond was named one of London’s 1000 most influential people

about hard decisions on issues such as which services to prioritise.

What does the government think of them?

The think tank points to the government’s Decentralisation and Localism Bill, currently before parliament, as a key example of the major effect it has had at the heart of coalition policy. The legislation aims to devolve greater powers to councils and neighbourhoods and give local communities control over decisions on housing and planning.

How influential are they?

Anyone seeking to influence government policy or, in ResPublica speak, “gain traction,” is said to be beating a path to its Westminster office in the shadow of the Houses of Parliament.

Members of its advisory board include the communities and local government minister, Greg Clark, the further education, skills, and lifelong learning minister, John Hayes, and their fellow Conservative MP Zac Goldsmith.

Where do they get their money from?

Describing itself as non-party political, it gets funding from corporate donors and members. It carries out research for the National Endowment for Science, Technology and the Arts (NESTA), which made Blond a fellow in 2009.

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FROM BMJ.COM

The value of one minute

As we welcome in a new year, Aser García Rada writes about how expectant Spanish mothers have been rushing to deliver their babies before 1 January.

“It is a common Spanish tradition to play the ‘Christmas lottery’ on December 22—it is the most important draw of the year. Those who don’t win still keep some hope for the second lottery—the ‘kids’ lottery’ on January 6th. But this year, as the front page headline of the Spanish newspaper *La Razón* says, there is still one more chance to be a lucky winner if you happen to be pregnant. The ‘baby cheque’ is the other kids’ lottery. And it is indeed a lottery, the one showing, like rarely before, the true value of a single minute. To be precise, a value of €2500 (£2150; \$3350).

“In July 2007, the Spanish president Jose Luís Rodríguez Zapatero approved a bill to give €2500 to every mother having a new baby. Since then, more than 1.5 million mothers have received a so called ‘baby cheque.’ At first glance, it doesn’t seem a bad idea for a country that has for many years had the lowest birth rate in the world. But the distribution of the aid has been far from ideal. For example, someone overheard a very well dressed woman remark while doing her shopping: ‘I think I am going to spend the €2500 on a flat screen plasma TV.’

“Seems unfair? Yes, because it actually is. But now the cash benefit is set to be scrapped from 2011 as part of Spain’s austerity measures. And it is also unfair to cancel the measure, or at least for those people who really need it. Let’s remember that Spain has around 20% unemployment, the highest rate among developed countries. The government’s decision will make a great difference for some of those born between midnight and 0:01 tonight.

“Still, this would be merely something to fill the front pages on days of low political activity, if it didn’t unveil questionable practice in private Spanish clinics.

“This is what a midwife from a large hospital in Seville said to *El País* a few days ago: ‘In the public system it won’t work, but I have colleagues that are seeing this in private clinics in Andalucía.’ She is referring to pregnant women who are due to deliver in the first fortnight of January that are coming to visit the doctor early and suggesting they have gone into labour. ‘They don’t dare ask openly, but we know they wish to bring forward their delivery date,’ she said.”

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