Nature Encyclopaedia of the Human Genome

Nature Publishing Group, £675/$950 (prepublication price), pp 5000
ISBN 0 333 80386 8
www.ehgonline.net

Rating: ★★★

Five volumes, 5000 pages, three million words, 15.2 kg, £675—if it's nothing else, the *Nature Encyclopaedia of the Human Genome* is a heavyweight publishing phenomenon. How is it possible to write a sensible review of an encyclopaedia? According to at least one dictionary definition an encyclopaedia "provides a general overview on a topic … a good place to start research." To test the encyclopaedia against this definition the three of us each chose a single topic to look up in the encyclopaedia. Here is what we found.

Steve Abbs, a clinical molecular geneticist, chose "DNA mutation nomenclature," a subject that is dear to his heart

Clinical molecular geneticists spend their working lives finding, naming, and investigating genetic mutations. When I looked up "mutation" in the contents I was reassured to discover that most of the topics I was interested in (and more) were covered by individual chapters, all written by international experts in their field. I looked in detail at the article "Mutation nomenclature," because this topic can be confusing for some mutations, and it is the sort of thing a molecular geneticist might want to look up in an authoritative reference work such as this. The chapter, written by Johan den Dunnen, an international authority on mutation nomenclature, gives a succinct account of current recommendations, including relevant examples. Readers are referred to several sources for further information, including the Human Genome Variation Society's website, which gives more examples—particularly of the more complex types of mutation that are not covered in depth in this chapter. The chapter is a useful introduction and reference source on current recommendations and is sufficient for putting a name to most mutations.

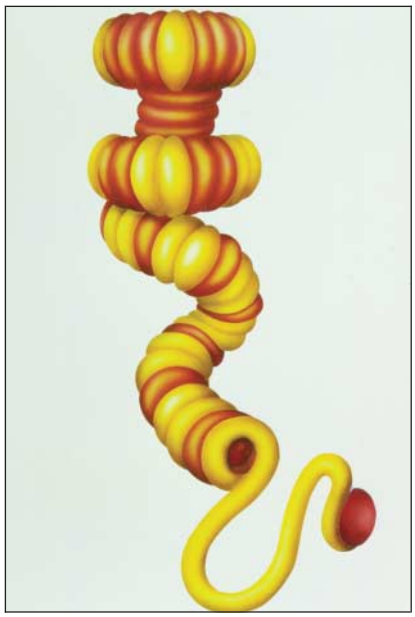
Unfortunately a recurring typesetting error causes confusion throughout this chapter. When I read the sentence "The terms Amutation and Apolymorphism are avoided" I began to wonder whether these were new terms that I hadn't come across before. I then realised that the letter A at the beginning of a word signifies that the word should have been printed in bold or italic. This needs correcting.

Tracy Bussoli, a genetic counsellor, looked up Duchenne's muscular dystrophy

I often see the young sisters of boys affected with Duchenne's muscular dystrophy, whose parents want to know whether the girls are carriers of this X linked recessive disorder. To prepare for such meetings I would want to remind myself about the disorder as well as to go over some of the ethical issues to do with testing children for being carriers of such genes. Kay Davies's chapter on the gene for Duchenne's muscular dystrophy and its mutations was easy to understand, but I could not find much information on the clinical details of the disease. It was also refreshing to see a chapter dedicated to genetic testing in children. This chapter's section on carrier testing raised many of the issues that would be pertinent if I were asked to do a Duchenne's muscular dystrophy carrier test on a girl of 13. In passing I noticed that the encyclopaedia had several excellent chapters dedicated to the subject of genetic counselling, including sections on consanguinity, the concept of non-directiveness, and the genetic counselling profession in Europe. These are all extremely useful topics for anyone working in my field.

Fred Kavalier, a primary care geneticist, tried to find out the answer to a question asked by a patient who carried a fragile X pre-mutation: "Will the fragile X gene have any effect on my health in later life?"

No article in the encyclopaedia is specifically devoted to the fragile X syndrome, although it is dealt with briefly in the articles "Fragile sites" and "Chromosome X." This seems odd for a relatively common genetic condition. The encyclopaedia's index contains references to 38 articles under the heading "Fragile X syndrome." None of these articles mentions the recent research that suggests that men who carry expansions in the fragile X gene may develop neurological symptoms later in life. Along the way I discovered a little discrepancy regarding the definition of a full mutation. On page 231 of volume 4 a full mutation is described as "more than 200 triplets." The table on the next page says a full mutation is "230-1000" repeats.



KAIROS/LATIN STOCK/SPL

Fragile X syndrome, a relatively common genetic condition, gets no separate entry

Searching through 38 articles in five volumes is heavy physical work. It is time consuming and requires a big desk. Online search facilities in the forthcoming electronic version of the encyclopaedia will be a big step forward.

Next door to the article on fragile sites I couldn't help noticing one on Rosalind Franklin. I was disappointed that the article's list of references did not include Brenda Maddox's *Rosalind Franklin: The Dark Lady of DNA*, which was published in 2002.

Summary

We were all pleasantly surprised by what we found lurking in the pages of this encyclopaedia. Like the human genome itself it provides an excellent starting point for anyone who is trying to find out about genetics. Like a good teacher it raises at least as many questions as it answers. Unlike the genome it is readable and full of reliable references and signposts. To save trees and remain up to date it needs to be published electronically.

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Items reviewed are rated on a 4 star scale (4=excellent)

Alarm call over reality gameshow

Doctors fear programme might have belittled dangers of sleep deprivation

A reality television gameshow that deprived contestants of sleep for a week and subjected them to tests including electric shocks has been criticised for exploitation.

Shattered—made by Endemol UK, which also produced *Big Brother*, and shown on Channel 4 last week (4 to 10 January inclusive)—featured 10 contestants, aged 19 to 33, living together in a purpose built studio in London's Docklands, where they underwent exercises designed to test memory, perception, reaction times, and mental agility. There was a "live elimination show" each evening to determine which contestant had lost the most function, and should therefore leave. The final remaining contestant would win the prize of a possible £100 000.

Contestants were allowed to sleep for up to two hours at a time, on the recommendation of the programme's medical advisers, but every "illegal sleep," when a participant shut their eyes for more than 10 seconds, meant £1000 was knocked off the prize money. In one challenge contestants had to work their way through a series of doors, some of which had handles that gave them an electric shock. This was despite advice from an independent ethics panel that electric shocks should not be used.

CHANNEL 4

Sleep challenge: the contestants of *Shattered*

Neil Douglas, professor of respiratory and sleep medicine at Edinburgh University, and president elect of the Royal College of Physicians of Edinburgh, said: "This is not a scientific experiment. It is voyeurism of people in distress to no benefit of anybody."

Professor Douglas feared that the programme—"an attempt to do something different for sensationalism"—might minimise the risks of sleep deprivation in the mind of the general public and make patients reluctant to seek help for sleep problems. "We have to stress that going about your life severely deprived of sleep does not work. You only have to think of the Selby rail crash."

Ten people were killed as a result of the crash in February 2001, when a driver (Gary Hart) fell asleep at the wheel of his car, which came off the M62 motorway on to a railway track. Hart, who was driving after a sleepless night, is now serving a five year sentence for manslaughter.

Professor Douglas said that sleep problems were prevalent in the general population but often went undiagnosed. The format of *Shattered*, "which would never have got past a hospital ethics committee," risked trivialising them, he said.

His concerns were echoed by Professor Jim Horne, director of the Sleep Research Centre at Loughborough University, who was a member of the independent ethics panel advising the makers of *Shattered*. He stressed that the programme should not be taken as a study of sleep loss.

Professor Horne was concerned about any impression that it was "somehow macho" to go without sleep, as many sleep related crashes in the United Kingdom involved men under 30. And he was worried that *Shattered* might pander to the idea of a 24 hour society. "This in no way reflects what would normally happen. Going without sleep in a group, under bright lights and television cameras with constant stimulus, is a long way from reality." If the contestants had been deprived of sleep completely they would not have been able to get beyond Wednesday, he added.

The Liberal Democrats' culture spokesman, Don Foster, said: "This is reality TV gone mad, reminiscent of the degrading American dance marathons of the Depression." He accused the programme of encouraging "voyeurism of pain." Viewers saw some contestants having hallucinations, slurring their speech, and causing muscle spasms in their feet to keep themselves awake.

Dr Trish McNair, another member of the ethics panel advising *Shattered*, said the programme makers had been responsive to many of the panel's suggestions, apart from ignoring its recommended ban on electric shocks. But, unlike a hospital ethics committee, this one had no power of veto, she said. And its role was different: "We had to examine how far the producers could be allowed to torture their participants in the name of entertainment."

Tim Hinks, creative director at Endemol UK, refuted criticism that *Shattered* exploited participants: "They were all adults and free to leave at any time. We carried out careful checks to make sure they had no medical problems and we do not accept that they were victimised in some way," he said.

Shattered might not improve public health, he said: "The aim was to make a show that would entertain and be watched." But at the same time he believed that television audiences were too sophisticated to take it as proof that going without sleep was safe.

Shattered was won by a 19 year old who is training to be a police officer. She was the youngest of the contestants and stayed awake 178 hours, beating a 30 year old psychiatrist to the prize of £97 000.

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Hit parade bmj.com

These articles scored the most hits on the *BMJ*'s website in the week of publication

NOVEMBER

1 **Clinical review: Managing sickle cell disease**
BMJ 2003;327:1151-5
4570 hits

2 **Editorial: What do we gain from the sixth coronary heart disease drug?**
BMJ 2003;327:1237-8
3983 hits

3 **Editorial: The criminalisation of fatal medical mistakes**
BMJ 2003;327:1118-9
3857 hits

4 **Editorial: BMJ Publishing Group to launch an international campaign to promote academic medicine**
BMJ 2003;327:1001-2
3677 hits

5 **News: *Lancet* accuses AstraZeneca of sponsoring biased research**
BMJ 2003;327:1005
3356 hits

6 **Career focus: How to write a case report**
BMJ 2003;327:s153-4
3156 hits

7 **10-minute consultation: Recurrent urinary tract infection in women**
BMJ 2003;327:1204
3107 hits

8 **Editorial: How many conditions can a GP screen for?**
BMJ 2003;327:1117
2820 hits

9 **Lesson of the week: Colchicine in acute gout**
BMJ 2003;327:1275-6
2795 hits

10 **Editorial: Diastolic heart failure**
BMJ 2003;327:1181-2
2763 hits

Fast and furious

Is media indignation over speed cameras undermining a valuable public health message?

It is not so unusual for a public health message to fail to make the impact that the medical profession feels that it deserves. To have a public health or safety message actively undermined by the media might, though, be felt to add insult to injury.

However, that has been the recent double whammy of a fate for the Speed Kills campaign. Newspaper coverage of the use of speed cameras in particular is so far off message as far as health and safety professionals are concerned that barely do the cameras' potential to save lives even get a mention.

Certain sections of the press have skewed the speed debate so much that it now seems to centre solely around the perceived infringement of individual rights, the "criminalisation" of the majority of drivers, and, in the most inflammatory aspect of the debate, the alleged siting of speed cameras purely for revenue-raising purposes.

The *Sunday Mirror* recently predicted that "Britain's roads will be spied on by an astonishing 20 000 speed cameras by the year 2013." The paper added that this apparent nightmare scenario heralded "a new age of misery for millions of motorists." Earlier this month a *Daily Mail* leader declared: "Seldom has a section of the British people been subjected to such a concerted and malevolent Government campaign."

Meanwhile, the *News of the World*, the country's best-selling paper, continues its Cameras . . . Action campaign—a "crusade to stop cops misusing speed cameras"—and the *Sun* offers readers free Stop the Highway Robbery car stickers.

The World Health Organization projects that by 2020 traffic fatalities could rank third among causes of death and disability—ahead of malaria, tuberculosis, and AIDS—and it is devoting this year's World Health Day to road safety under the slogan "Road Safety is no Accident." The United Kingdom Health Development Agency has also taken up the cause recently, calling for speed limits on residential roads to be cut to 20 mph to reduce children's deaths and injuries by 67%.

Inappropriate speed for road conditions is a factor in a third of crashes, and about 72 000 speed related incidents a year in Britain account for 1100 deaths and 12 600 serious injuries. The cost to the health service is vast. So why do some newspapers seem not to see speed as a health issue to be taken seriously?

The *News of the World*'s Jules Stenson insists that his paper does understand speeding as a health issue and does support the use of speed cameras in certain areas. "What we object to," he explains, "is the Big Brother excess of speed cameras, their overuse, and the thousands and thousands of otherwise law-abiding people being criminalised because of poorly and cynically placed cameras.

"We accept that speed kills but we don't think cameras are the way to slow people down. We'd like to see more measures such as police highway patrols and driver education."

Why then does the *News of the World* not campaign positively for more and better anti-speeding measures rather than focus on the alleged negative aspects of the cameras?

"But that's newspapers," says Stenson. "The most interesting facts go at the top."

He points out that his paper's campaign has had a massive resonance, with outraged readers inundating the paper with examples

Caught on camera: but are newspaper campaigns (above right) trivialising the dangers of speeding?

of badly placed cameras on which the paper duly reports.

Where, though, wonders Kevin Clinton, head of road safety at the Royal Society for the Prevention of Accidents, do the *News of the World* and other national newspapers—the majority of which have taken up the anti-camera campaign to some extent—carry the views of people who have been injured or lost family members in speed-related incidents? "Let's hear from them," he says. "The media do talk to us and they do publish our comments, so their stories have a little bit of counter-argument. But they don't like speed cameras and that comes across far more clearly."

He claims that many of the arguments that many journalists make cannot be substantiated. For example, he explains, it is little reported that speed cameras reduce death and serious injuries by 35% at the places where they are sited, and there is no evidence to support the wide claim that cameras are sited purely for revenue-raising purposes. Legislation, in fact, specifically prohibits it.

Zoe Stow, chairwoman of RoadPeace, the national charity for road crash victims, says that had speeding been a disease killing and injuring so many, there would long ago have been a public inquiry. Questions would have been asked in the House of Commons, and the full wrath of the media would have been unleashed.

"The public health message about speeding is failing to get through," she says. "It's illogical. Until we twist things around to show that if you speed then you're lucky if you don't hurt yourself or someone else, rather than unlucky if you do, nothing will change."

However, she acknowledges that it took around 20 years for the drink-driving message to be taken seriously. Just a decade into the Speed Kills campaign, maybe there is another decade to go before people see themselves as potential victims rather than unfairly persecuted. The media certainly seems less than keen to help push the message along.

Naomi Marks *freelance journalist, Brighton*

PERSONAL VIEW

Bioethics needs to rethink its agenda

What issues belong at the top of the agenda in bioethics? What important topics are commonly ignored? Does bioethics matter? As someone who writes about bioethics one of the lessons I have learnt is that the articles that typically attract the attention of editors and readers are the manuscripts addressing "sexy" topics. Ambitious researchers in bioethics know that if they want to obtain research funds and draw attention to their work they should focus on such topics as embryonic stem cell research, germ line gene therapy, and therapeutic and reproductive cloning. These topics practically sell themselves.

Not long ago researchers examining ethical issues in medicine and health care had a different focus. In the 1980s and 1990s the study of ethical issues in palliative care generated hundreds of articles, as doctors, philosophers, and lawyers addressed such topics as the withdrawal of fluids and nutrition, advance directives, surrogate decision making, defining death, medical futility, and physician assisted suicide. Scholars continue to address these subjects, but now the topics lack the air of novelty they once had. Researchers looking for the next big thing are shifting toward the study of "neuroethics" and genetic "enhancement technologies" supposedly on the verge of propelling us into a "post-human future." In contrast bioethicists rarely address urban poverty and inner city violence, even though poverty and violence raise important issues related to health. Although doctors, epidemiologists, and public health specialists treat firearm related violence, poverty, joblessness, and the breakdown of communities as important topics, such issues are largely off the radar screen of bioethicists. If clinicians recommend withdrawing treatment from a gunshot victim, and the family wants medical care to continue, then perhaps bioethicists become interested. However, bioethicists rarely engage the legal, economic, and social conditions underlying violence in poor communities.

Bioethicists commonly address ethical issues arising in wealthy developed countries. Discussions about priority setting and resource allocation typically occur within the context of a particular developed state. While analyses of globalisation and international inequity in access to basic goods such as food, clean water, and shelter are beginning to appear in bioethics scholarship, bioethicists have traditionally tended not to think at the transnational level of analysis or addressed issues relating to the developing world.

Perhaps one reason bioethicists are reluctant to address global ethical issues related to health, illness, and poverty is that bioethicists are deeply embedded in a global economic system that depends on the continued existence of impoverished societies. While there are many ways for corporations to generate profits, one effective means is to shift factories and jobs to places where employers are relatively free of government regulations and where labourers work for a pittance. In North America factories and jobs in industry migrated from north to south. Now factories, workshops, and labour intensive jobs in Japan, the United States, and Western Europe move to China and India. Many jobs are shifting from developed nations to poorer countries with low hourly wages, few workplace benefits, minimal health and safety standards, and patchy environmental regulations. Many of the goods enjoyed by citizens of wealthy nations are available for consumption because of the continued existence of massive economic disparities between wealthy and poor nations.

> Bioethics risks becoming a source of entertainment

Most of us would prefer not to confront the incredible disparity between living as a professor or clinician in a smart apartment in Manhattan and eking out a living in one of China's rapidly industrialising districts or a shantytown in South Africa. Scholars such as Solomon Benatar and Paul Farmer have drawn attention to such matters, but many bioethicists continue to see these questions as macrosocial economic issues falling outside the proper scope of bioethics. And yet questions of health and illness—and ethical issues related to health systems, social institutions, and economic policies—are connected to global markets and financial institutions.

Many of the questions that bioethicists address only make sense within the context of wealthy developed nations. Some of the favourite topics of bioethicists seem trivial compared with the important health issues facing people in the world's poor countries and in impoverished regions in rich countries. Greater consideration of global ethical issues related to health, illness, and suffering might generate a richer, more meaningful research agenda for bioethics. Otherwise bioethics risks becoming a source of entertainment and spectacle in wealthy societies whose inhabitants overlook the poverty and suffering found throughout most of the world.

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SOUNDINGS

Student selected caesareans

2004 is my 40th year in medical education, as student or teacher. Back in 1964 our class was special—the first to experience the New Curriculum. The idea had been to reduce factual learning and make the course more integrated.

I now know that reducing factual learning is a tradition stretching back to the 19th century. Each generation of educators discovers that designing a fully integrated and really challenging course is fun. It certainly beats teaching.

Our last new curriculum in Leeds had special study modules (SSMs). Our current one has student selected components (SSCs). When I asked about these damn fool initials, I was told they came from the General Medical Council (GMC). Ah yes. Back in Hallam Street it had seemed a good idea to let students choose subjects for in-depth study.

When the Leeds ethics theme team asked for topics for a third year SSC, I suggested: "Should women be allowed to choose caesarean section?" Later the emails started arriving, asking: "When can we meet?" That's when the magic started. After decades of force-feeding students the basics I had five lively undergraduates talking to me by choice.

Never having done obstetrics, they soaked up background information about indications and risks. I tried (but not too hard) to disguise my own opinion that a well-informed woman can choose how to have her baby. Then off they went to surf the literature.

Most of the stuff on the internet is written by people who believe that pushing a baby through your pelvic floor is a harmless and fulfilling experience. The students, rather pro-choice to begin with, were persuaded that caesarean section is risky, bad for bonding, and too expensive for the NHS. People who like it don't say so on the web.

The climax of the SSC was dramatised presentations. Emily wore a pregnancy belly with an umbilical ring. Hinaa was the NHS obstetrician and Imran the hospital manager. David played the suave Brazilian husband and Aysha provided the commentary.

They wrote the script but let me direct. "Now, luvvies, let's do the moves. Autonomy *here* . . . Beneficence *here* . . . Marvellous, darlings!" I got a text message when they were about to start. They were terrific. I went home a happy medical academic. Now that *is* new.

James Owen Drife *professor of obstetrics and gynaecology, Leeds*