SECRETS OF MY DNA

By ADAM HIGGINBOTHAM
Photography: GREGG SEGAL

Raymond McCauley is betting on cheap gene tests plus some amateur science to minimise his chances of succumbing to a sight-threatening syndrome. Who needs doctors in the new era of health hacking?

LITTLE MORE THAN THREE YEARS AGO, SAN DIEGO-BASED biotech firm Illumina, a market leader in providing genetic-analysis services and equipment to labs and researchers worldwide, entered a landmark partnership: to develop the first genotyping kits for one of the first direct-to-consumer gene-testing services, 23andMe.

The deal, with the help of cocktails-and-testing-kits "spit parties" and a TV appearance on *Oprah*, would help to take the results of Illumina's work well beyond the research community and into thousands of homes across the United States. To celebrate, the company's employees were offered an idiosyncratic company perk: cut-rate gene sequencing.

At the time, the cost to have your entire genome sequenced by Illumina was $350,000 (£218,000). 23andMe was marketing a partial sequencing, covering only certain areas of the genome and producing reports for ten disease risks and four genetic traits, for $999. Under the terms of the deal, Illumina employees could get the partial sequence for $249. (Advances in technology and knowledge mean that a current 23andMe test starts at $199, providing 182 reports.)

In Illumina's offices in Hayward, California, senior bioinformatics scientist Raymond McCauley could hardly believe it. "My eyes got really big," he recalls. "I thought: 'That's fantastic. I'll know everything there is to know.'" McCauley, a softly spoken Texan who, at 44, describes himself as a "nuts-and-bolts guy" of computational biology, has worked in the commercial genetics industry almost since its inception. He had known that one day gene sequencing would become cheap enough for him to be able to afford it himself. But he hadn't imagined it would arrive so quickly. Just seven years earlier, when he was working for Rapigene Incorporated in Seattle, by his estimates the company would have charged at least $2.5 million for the same analysis he was now being offered through the post for the price of a PlayStation.

McCauley immediately ordered a test for himself and, soon afterwards, for his extended family: 11 tests in total, including ones for his partner, their twin sons, his mother, his sister, his mother-, father- and sister-in-law. Two months later, he received an email from 23andMe telling him that the analysis was complete, and logged on to its website. Today, the report that 23andMe provides its customers is clearly sorted in a neat and user-friendly profile, presenting the highest-risk diseases on a single page. But back in 2007 you had to sift through the information yourself. Fearful of what he might find – Alzheimer's, for example – it took McCauley three days to work through his risks, opening 30 pages, one link at a time. It was, he says, "like turning over a rock, where you're not sure you want to see what's under it".

There were some surprises: the results on his ethnicity revealed that the family lore that his grandmother was half Cherokee was groundless. Elsewhere, the analysis of disease risk showed that his chances of heart complaints and type II diabetes were slightly higher than average. Clicking deeper into the data, McCauley found that his DNA revealed something much more rare: that he was four or five times more likely than most people eventually to develop age-related macular degeneration (AMD).

McCauley had no idea what this was. At first he thought it sounded like something to do with his jaw, but he soon discovered that the condition, which occurs mostly in adults from their late sixties onwards, is a progressive blindness caused by a choking of the blood supply to the retina. AMD slowly destroys patients' sight from the centre of the retina outwards, until they're left with nothing but peripheral vision – a blurred halo of an image surrounding a ragged black hole. They are unable to read or recognise faces – legally blind. The disease advances very swiftly and is incurable. And of all the ailments detailed in the 23andMe profile, AMD has one of the strongest genetic associations yet established. "Well, I looked at that and I was like, 'Oh, that's not good news!'"

McCauley read that there were a few preventative measures he could take to reduce the chances of AMD one day rendering him blind: don't smoke and avoid ultraviolet light, for instance. Also, it seemed, he could try taking a special combination of vitamins, including B12 and lutein. But when he consulted the research, he could find little evidence to support the effectiveness of the regime, based on his genotype. He was convinced that there should be some way of finding out, a way to use his genetic data to create a customised preventative treatment devised just for Raymond McCauley – what he calls "actionable personal information". At the beginning of 2010, he decided to do so in the most direct way he could think of: he began experimenting on himself.

onsumer genomics expanded at prodigious speed after the announcement from the Human Genome Sequencing Center in Houston, Texas, in May 2007 that DNA pioneer James Watson had become the first person to have his entire genome sequenced. The first direct-to-consumer genome tests appeared on the market a few months later, and after that a steady trickle of individuals' genomic information began appearing online. Initially it was curated by a handful of geneticists – including George Church of Harvard Medical School, whose Personal Genome Project has so far made publicly available the complete genomes of 12 men and women, and Hugh Rienhoff, a doctor and biotech entrepreneur who in October 2007 founded mydaughtersdna.com as part of his attempts to discover more about the illness afflicting his daughter Beatrice. But by June 2010, an estimated 100,000 people had sent spit samples and cheek swabs to companies including 23andMe, Navigenics, Pathway Genomics and Decodeme for mail-order DNA analysis.

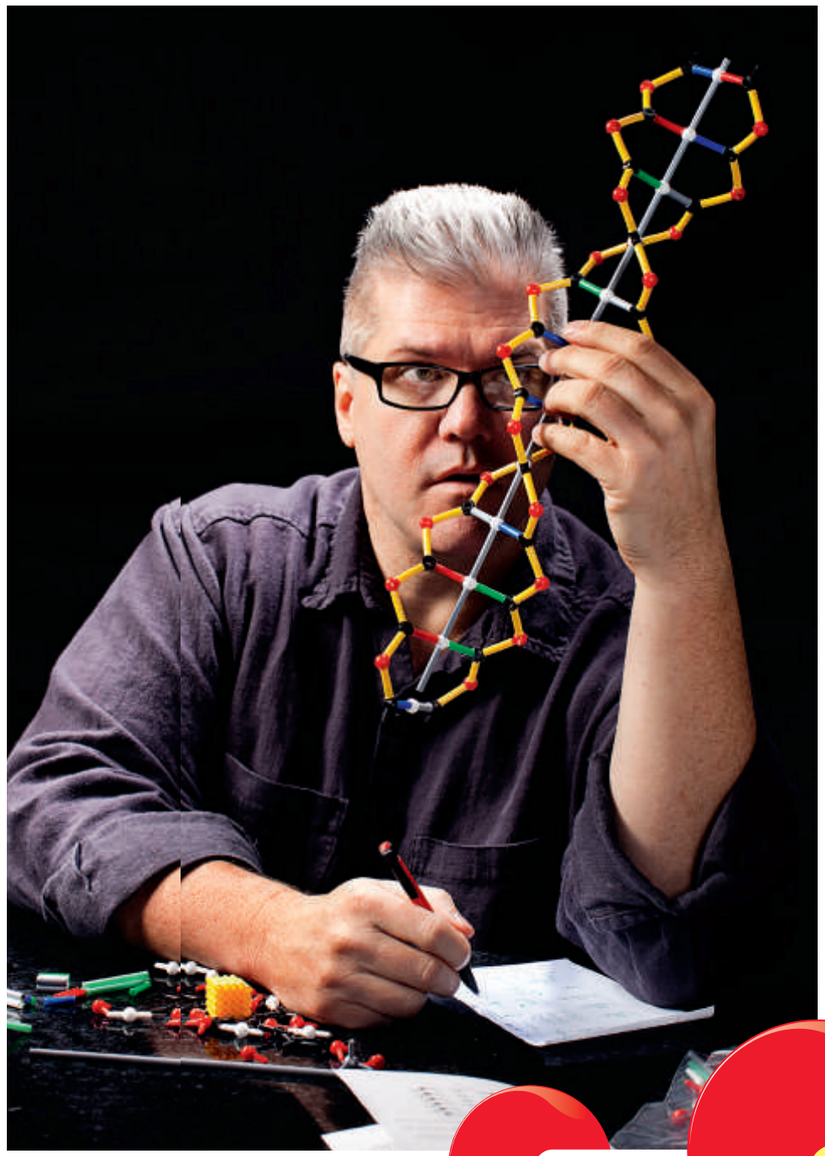
Although these tests offer only a selective analysis of the most useful chunks of DNA, a few dozen wealthy individuals have also gone directly to labs such as Knome or Illumina and paid to have sequenced their entire genomes – all six billion letters of genetic code, each representing a single nucleotide base, either an A (adenine), C (cytosine), T (thymine) or G (guanine). Among the tech elite and early adopters of Silicon Valley, having your DNA sequenced has become as socially valued as being early to the iPad or driving a Tesla. And, of course, the more of the genome you've had decoded, the more impressive it is: "It's, 'Have you had 23andMe done? Just a fraction of a per cent?'" Raymond McCauley explains one evening in San Francisco. "'Have you had the whole thing done?' That's *swank*."

But as the sophistication of sequencing increases and its price plummets, the process is becoming democratised. The decoding of Watson's DNA in 2007 took four months and reportedly cost the lab $1 million; to have the same thing done now would take less than a week, for a retail price as low as $20,000. Speed and price continue to fall, outstripping even the pace of advances in printed circuits. "It beats Moore's Law with a stick," says McCauley, who believes that the $100 genome is only three years away. "It's going to be so cheap that it will be a choice between, 'Do I want to buy a pizza tonight or should I go ahead and get my genome sequenced?'"

The reach and impact of personal genomics could revolutionise the way that we approach healthcare; almost anyone will be able to learn the contents of their thousands of lines of DNA code, without even making an appointment with a doctor. Yet the decoding alone is of little use without the means or knowledge to interpret it. "At that point you're going to have all the data but no information," McCauley says. "That's what got me started. How do I make all of those As, Cs, Gs and Ts useful? I know that I've got a C and a T here, but what does that matter? And what difference is that going to make to me in my life?"

The answers may come from a small group of individuals such as McCauley who, armed with their own genomic data, have become citizen scientists – amateur biologists who are part of a wider trend towards what Melanie Swan, a Silicon Valley futurist who recently founded DIYgenomics, an online start-up dedicated to crowdsourced clinical trials and personal-genome apps, calls "health hacking".

Health hackers, Swan says, are using online resources such as PubMed – the US Library of Medicine's colossal database of biomedical literature – to help to conduct experiments upon their own bodies with the same ease that anyone with access to the internet can now trade stocks and shares. "Twenty years ago, you had to have a stockbroker. Now there's enough information and tools available on the internet, you can dig into it yourself and experiment. The same is true for health," says Swan.



"Scientists," says Raymond McCauley (*left, with a DNA model*), "are people trying to find answers"

Increasingly, health hackers with access to their own DNA analysis are intervening more directly in their healthcare. Rather than having to rely on general information from books and clinical studies designed to draw median conclusions about the population in general, Swan and McCauley want to customise their own drug and diet regimens. They hope to devise preventative treatments for themselves based on their personal genetic predispositions towards everything from disease to drug response. "It's enabling people to know not just what's in their genome, but to figure out what that means – instead of waiting for some scientist to do a study that may relate to them," says McCauley.

Early last year, Swan and McCauley began contributing to the development of Genomera, a website founded by Silicon Valley entrepreneur Greg Biggers to enable health hackers to share their genetic and experimental data. Biggers had visited Hugh Reinhoff's attic laboratory but was appalled at how antiquated and laborious the analysis of DNA data remained. He recognised an opportunity: no longer were huge quantities of complex data restricted to scientists in the new era of consumer genomics. So consumers would pay for a service that helped them. "User experience is going to be huge in a way that wasn't necessary for the phase of biology in which it lived in laboratories," he says. As a result, Genomera has been designed with a simple interface and community features that Biggers hopes will soon turn healthcare into a social activity. "It's Facebook for genomes," Melanie Swan explains over coffee one afternoon in a café in Palo Alto.

And when mass sharing of DNA data does arrive, citizen geneticists will play a key role. "I think of all the things that citizen scientists can do – astronomy, weather, geology, fossil-hunting – this is probably the one with the biggest impact on peoples' lives," says George Church. "If you find another tyrannosaurus, that's kind of cool – though it's not a life-or-death matter. But if you find some family that has somehow escaped the ravages of Huntington's, even though they have the Huntington's allele, that would be a really big impact. If you found a family that avoided diabetes, despite having all the risk factors, that would be even bigger. It's something that scientists can't just do sitting in a laboratory. This is one of the few things where scientists really need citizen science."

At 6ft 2in and 113kg, Raymond McCauley is a big man. But he used to be much bigger, he tells WIRED one October afternoon in the small, cluttered upstairs room he uses as an office in his house in Mountain View, California. "I was a 300lb [136kg] guy," he says. "Really big." At the end of 2008, McCauley began making drastic changes to his diet, switching from burritos and hamburgers to fresh fruit and yoghurt, slashing the number of calories he consumed by two-thirds. In the first two weeks he lost 4kg through water loss alone; by March 2009 he had dropped four clothes sizes and lost tens of kilos.

The dramatic change in McCauley's weight is the most visible result of the citizen-science project he has been pursuing since the arrival of his 23andMe data at the end of 2007. Weight loss was not exactly a scientific leap in the dark: on his father's side, the family had endured a long history of heart trouble, and for years McCauley's doctor had been telling him to eat less and exercise more. But he had simply ignored the advice. "I was like, yeah, yeah, yeah, OK," he says. But when McCauley saw the elevated risks of atrial fibrillation and diabetes in his DNA profile, "I decided that the most actionable, important thing I could do was to lose weight – if I wanted to live longer."

He procrastinated about actually embarking on a diet ("I delayed it as long as I could," he admits). He connected a digital scale to his home computer and began poring over research journals that told him essentially what he already knew. ("Most of it basically came down to: eat less, exercise more".) Yet McCauley remained preoccupied with the one outstanding risk he had found in his genetic data: his high chance of developing the creeping blindness of age-related macular degeneration.

His first stop to find out more about AMD was the Mayo Clinic website – "They've got articles on everything from removing a splinter to the different forms of brain surgery," he says. There, he had found advice about avoiding smoking and UV light. He discovered a self-diagnostic tool, the Amsler Grid – a pattern of bold lines and a dot that a patient is required to print out on a piece of paper and then stare at. If you notice that the lines begin to distort and warp after a few seconds, then you've probably got AMD.

Seeking more information – "doing the scientist thing" – in mid-2008 he took a trip to Stanford University to visit a research ophthalmologist conducting clinical trials on AMD. McCauley wanted to hear news from the cutting edge of work on the disease and its links to genetics, and to put himself forward as a subject for long-term study. But he was told that although researchers were very interested in the genetic markers of the disease, nobody at Stanford had considered conducting genomic testing for AMD. McCauley was certainly the only person who had ever arrived bearing their own DNA information. His offer to become a subject for study was politely declined. "It was, 'Gosh – thank you. It's really interesting that you're proactive about this, but we don't really do that.'"

Back at home, McCauley considered taking vitamins as a preventative. He discovered a clinical study which suggested that the chances of contracting the disease were significantly reduced in patients who had taken a special mix of supplements, including B12, zinc and lutein. The results of the trials seemed conclusive. Even McCauley's doctor – "One of these guys who says, 'Vitamins? Vitamins are really good for giving you expensive urine,'" he explains – saw its value and advised him to begin taking the supplements.

Yet McCauley also remembered reading research into diabetes treatments, which suggested that large doses of vitamin B9 are important. The study mentioned that some patients with a specific genetic defect could not absorb the most widely available form of vitamin B9. To overcome this, diabetics were prescribed an active form of the vitamin, L-methylfolate. McCauley began to wonder if the same defect might affect him, and what the basis might be for the usefulness of taking L-methylfolate: "Is it snake oil or is it really helping people? How do you test it? How do you test if this is really working?"

He searched for information about the genetic defect, and for something that would tell him about his own potential response to vitamins, any study from which he could personalise his treatment, based on what he knew from his own DNA make-up. By cross-referencing what he found on PubMed with what he had been told by other subscribers to the 23andMe message boards, he was eventually led to a single gene, methylenetetrahydrofolate reductase (NAD(P)H), the status of which directly affects the body's ability to metabolise vitamins B9 and B12. The gene provides the body with instructions to manufacture an enzyme crucial to the chemical reaction by which one specific amino acid is processed into another. Its official name is usually abbreviated to MTHFR. Noting the opportune sequence of consonants in the initials, McCauley and a friend began referring to it as "Samuel L Jackson's favourite" – or, often, in a less oblique and more coarse way. And so, when they eventually found their focus, Raymond McCauley's citizen-science experiments would close in on the ten variants of what he had come to know as "the motherfucker gene".

The DNA marketplace

23andMe
An ancestry test costs $399, a health test $429: bundle both and you'll cover 175 health traits for $499. Founded in 2006, the Mountain View company raised $22 million in series C funding, including investment from Google Ventures, in November 2010.
23andme.com

Pathway Genomics
Founded in San Diego in 2008, Pathway sells four types of test – health conditions, drug response, carrier status, and fitness – through doctors for about $500 each. It screens for 27 complex conditions, 76 carrier statuses and 12 drug responses, and surveys 100 genes for metabolism.
pathway.com

Decode Me
A "complete scan" covers 47 medical conditions and costs $2,000; scans specific to cancer and cardio are $500 each. The Icelandic company was founded in 1996. It courted controversy by proposing an Icelandic Health Sector Database containing the genetic data of all 300,000 Icelanders.
decodeme.com

he journey Raymond McCauley has made from computer technician to amateur microbiologist has been long and frequently slow, but apparently inexorable. As a 13-year-old in Weslaco, in southern Texas, he made a plan to become an astronaut, and for the next ten years dedicated himself to its methodical execution. At Texas A&M University, he enrolled for a double major in electrical engineering and computer science, but that alone wasn't enough: "If you want to be an astronaut, you can't just be pretty good at something, you've got to be really good at a lot of things," he explains. So he considered a third degree in aeronautics before deciding that, in order to increase his chances of qualifying as a shuttle or space-station flight surgeon, he should take a medical subject instead. He chose genetics, at the time a discipline so little understood and apparently arcane that his decision frequently provoked a blunt response: "You're crazy. What use is that?"

McCauley eventually graduated with degrees in the first two subjects he chose, but never completed the one in genetics. He took a job just outside Houston with Boeing and interned with Nasa, working on a biosphere-like Closed Environment Life Support System project at Johnson Space Center. But the programme cancellations that followed the *Challenger* disaster in 1986 placed his dreams of one day going into space increasingly out of reach. He found himself sleeping on his sister's couch, looking for work and, eventually, his career goals were defined by very practical considerations: "Just having to eat," he says.

So McCauley became a computer programmer and systems analyst back on the university campus at Texas A&M. While there, he began attending classes in whatever took his fancy – from more science and engineering to philosophy, business and languages. He stayed for ten years and became, he says, a "professional student". But in 1997, he began a master's degree in biochemistry and biophysics. And as the world of commercial biotechnology began to expand, his once eccentric combination of computer skills and life-science qualifications suddenly seemed prescient. The unprecedented volume of data produced by the rapid advances in DNA sequencing overwhelmed the simple spreadsheets then used by biologists to process the information, creating a problem best solved by someone who understood both the technicalities of programming and the finer points of genetics.

McCauley was still working on the master's when, in 2000, he began work at Rapigene, bridging the gap between software engineers and biologists in a newly conceived field: bioinformatics. Yet, even after completing the master's in biochemistry and biophysics, McCauley was frustrated by his lack of direct experience in the laboratory; he longed to get his hands dirty. "I've always been an empiricist," he says. "I think you ought to be able to look at something and understand what's going on. I mean, I can design a computer, build it from spare parts, tell you which electrons are going where and build that up into seven layers of logic and an interpretive language. But in biological science, there's a lot of stuff that has been a real black box to me. I've never been a bench scientist, a hands-on guy. I understand a lot of the process. But if I had to go and do it, I would hurt myself with the glassware."

In 2008, McCauley stumbled upon the DIY bio movement (see WIRED 09.09). He was searching for cheap ways to perform PCR – the polymerase chain reaction, the DNA-copying process as fundamental to biotechnology as mixing flour and water is to breadmaking – without access to a research laboratory. "I found all these people who were playing with almost exactly the same things," he says.

The members of DIYbio, a network which now has groups in New York, London and Paris, are amateur scientists and biohackers who hope to kickstart a culture of low-cost commercial biotechnology in the same way that the Homebrew Computer Club did in personal computing in the 70s. The group was launched in May 2008 with a meeting of 25 people in an Irish pub just up the road from MIT. Members use second hand equipment bought on eBay and budget tools improvised at home – microscopes made by removing the lens from a webcam and reinserting it backwards, centrifuges built from Dremel miniature drills– to perform microbiology experiments in makeshift labs built in kitchens and garages.

McCauley started attending DIYbio meetings in Mountain View and soon became a regular on the homebrew microbiology scene. One weekend, his group learned how to build an algae bioreactor at home; on another, they genetically engineered a version of *E. coli* that glowed in the dark. They collected money in a hat, ordered some DNA over the internet and, a few weeks later, did some gene splicing right there in the garage. McCauley was amazed at how easy it all was. "Even being in the field," he says, "that was a real 'aha!' moment for me."

One Saturday morning, in autumn 2009, McCauley went to a meeting in a Mountain View garage where local DIYbio group BioCurious was experimenting with cancer-cell-design modelling using equipment bought on eBay. It was there that he met Melanie Swan.

Unlike McCauley, Swan has no scientific qualifications. Before moving to Silicon Valley in 1998 at the start of the dotcom boom to develop her own startup, Swan had taken an MBA in finance and accounting and then worked for two years as a banker at JP Morgan in New York. After selling the startup, she became a consultant on emerging technologies for clients such as Siemens and AT&T. She now describes herself as a hedge-fund manager and applied-genomics expert. "Three years ago, I decided there was no choice: I have to start learning about life sciences," says Swan, a pale, earnest 42-year-old. "I realised that this is going to be the next big area to hit, and I want to be a part of creating that."

She began educating herself from scratch – taking courses and attending conferences where, increasingly, she gave presentations about her findings. She added personal genomics to her portfolio of interests in 2008, when she took the 23andMe test. She had wanted to establish whether she had a personal genetic predisposition to colon cancer, which had killed her mother, but when the results revealed no susceptibility to the disease in her genes, she became curious about what else she could do with her data.

Sharing their frustrations about the limited "actionable information" that could be extracted from their 23andMe analysis, Swan and McCauley agreed that there should be some way in which anyone who had their genes sequenced should be able to use the

data to immediately change their day-to-day lives beyond "eat less, exercise more". They agreed to look into it and, at the start of 2010, began meeting every weekend at McCauley's home, spending hours in front of the computer and poring over research papers. They hoped to find material on what McCauley calls "lifestyle genetics", a study establishing a link between a person's make-up and their nutrition or metabolism. "Is there anything I do that I would want to do differently because of my genetics?" McCauley says. "How can I adjust what I eat, or how I'm activating my body? Is there something that tells me if I'm more of a morning person than a day person? Because those are probably not environmental differences. That really ought to be hidden in your genetic code."

Swan and McCauley covered every subject area they could think of, to see if anyone had ever executed the kind of study that could answer any one of his battery of questions: "If you want to lose weight, what's the best way? If you want to enhance your cognition, start faster in the morning and think at a higher level? Or have more energy? Or less joint pain? We were all over the place."

But wherever they looked, they were disappointed. They would simply have to conduct some practical research for themselves. Swan and McCauley decided to devise their own clinical study in lifestyle genomics. But first they had to pick a subject to investigate. "What's the simplest thing that is not controversial like some Alzheimer's thing?" McCauley says now. "Vitamins." So they turned their attention to the motherfucker gene.

enetic research was, in McCauley and Swan's first home-brew clinical trial, being conducted to answer a question that had become increasingly important to McCauley in the months since he first saw his 23andMe results: was there any link between individual genotype and vitamin metabolism? Specifically, could they prove a connection between variants in the MTHFR gene and the ability to metabolise B vitamins, one that might affect McCauley's efforts to head off AMD? They decided to find out with a series of simple experiments using the two forms of the vitamin, followed by blood tests. In the tradition of scientific pioneers of the past, they selected themselves as initial test subjects. They needed more – and discussed conducting the trial through a university lab where Swan had connections and could canvass a sample of student volunteers. But they quickly realised that wasn't the point: they didn't just want to conduct a single experiment that answered questions about B vitamins, they wanted to build a model trial that could be easily replicated by citizen scientists anywhere, with the minimum of effort. If the vitamin study were a success, they could post their findings online and invite anyone who was interested to help them repeat it on a larger scale, or to investigate any number of genetic questions about ageing, athletic performance, skincare products – anything at all.

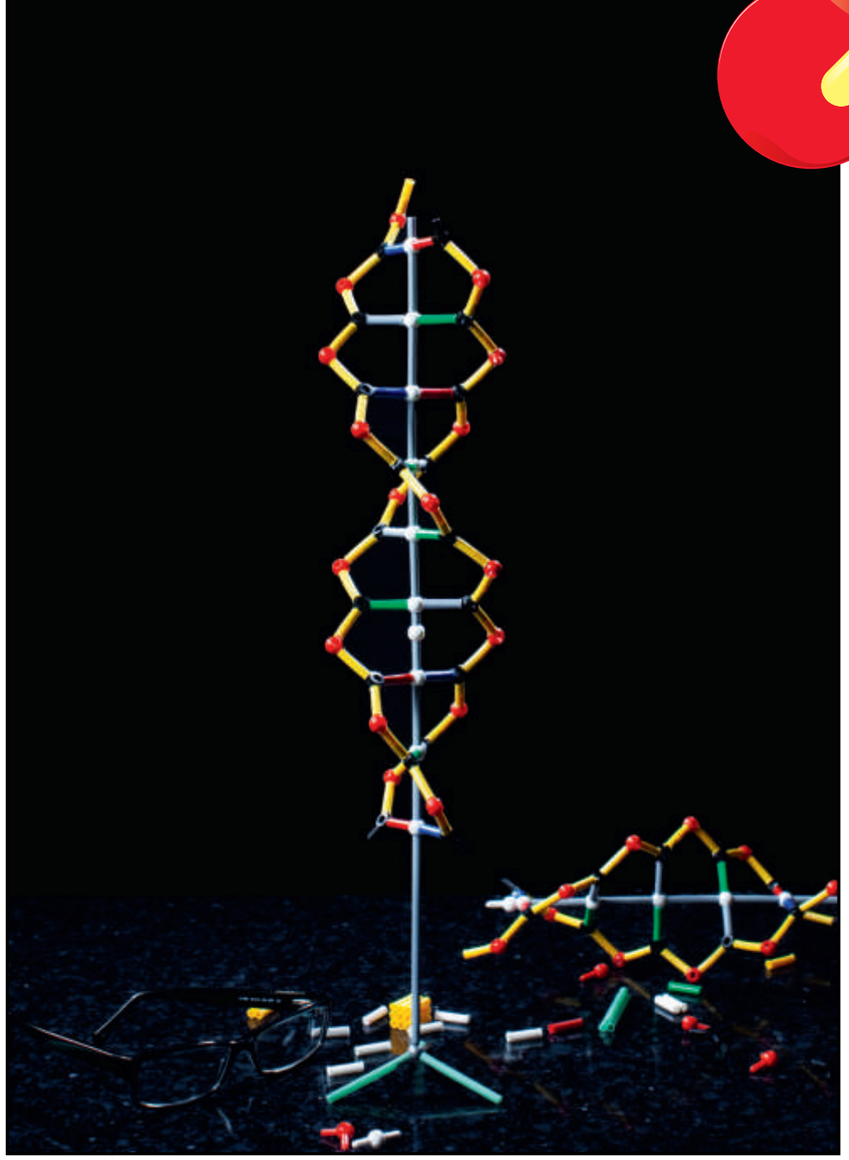
The experiment required five test subjects. In addition to themselves, McCauley and Swan solicited help from friends and acquaintances who had taken the 23andMe test. They signed up Chris Hogg, director of commercial strategy at a California biotech company, and Aaron Vollrath, a doctoral biology student living in the Bay Area. The fifth slot went to McCauley's partner, Kristina Hathaway. The experiment took place in three stages, starting with a two-week period during which the group took no vitamins at all. This was followed by a fortnight during which they all took Centrum multivitamins, then another fortnight taking the active form of vitamin B9, L-methylfolate. During the third period they took both supplements at once, followed by the "washout phase" when they took nothing. As each phase ended, they took a blood test to monitor each individual's level of homocysteine, the undesirable amino acid that is converted into a more benign substance if the body is efficiently absorbing B vitamins.

This proved the most potentially expensive part of the experiment: in California, drawing blood may be done only by a licensed technician, at a cost of around $100 for each test. So when, in April last year, Swan discovered a local lab running a three-day, two-for-one offer on blood tests, they decided to start the trials immediately. By the beginning of June, they had all uploaded their results, Wikipedia-style, to Swan's DIYgenomics.org website.

Relying on such a small sample, the findings of the experiment were hardly definitive. But they did confirm McCauley's suspicions about his genotype, which – "homozygous at both SNPs that coded for MTHFR", according to a recent report in the journal *Nature Medicine* – proved to be unique among the five people tested. The four other participants in the study responded well to taking both forms of B-vitamin supplement (Centrum and L-methylfolate), which reduced their levels of homocysteine by almost a third. But during the Centrum phase of the trial, the amount of the hormone in McCauley's blood actually rose; only when he began taking L-methylfolate did it fall. This suggested that if he wanted to have any benefit from taking B vitamins he would have to take the active form of it after all. For everyone else, the study demonstrated that taking vitamins certainly had at least one positive, quantifiable effect.

Most importantly, the MTHFR study was a perfect proof-of-concept trial for other citizen scientists interested in contributing to the experiment, or to guide them in devising their own. For the price of a handful of vitamins and some budget blood analysis, anyone could repeat the MTHFR test for themselves and add their results to the wiki at DIYgenomics.org. With each contribution, the overall study would become statistically more significant.

When Swan and McCauley mentioned to Greg Biggers, whom they both knew from conferences and Silicon Valley health-hacking meetings, that they wanted to build a new interface to cope with the volume of data necessary to scale up these contributions into a clinically valid study, he told them not to bother. They could use the one he was already building. The result, Genomera, is still in beta testing,



DNA's base pairs might hold the secret of keeping disease at bay

but promises to provide a place where individuals can upload their decoded genomic information to share and compare with other users, and gain easy access to medical databases where they can find information related to their DNA profiles. 23andMe has already shown the scientific benefits of consumers sharing their DNA data, with an analysis of its customers' information that revealed previously undiscovered connections between genetic variations and traits such as freckles. But Biggers plans to enable users to create and upload their own research data and provide fields for simple comparison, creating a truly interactive global platform for citizen science.

The second phase of the MTHFR study will be the first such open-participation experiment available on Genomera: anyone with their genomic data can participate for what Melanie Swan estimates will be around $500 each. McCauley has been surprised by the response. "I thought this was a narrow, geeky project," he says, adding that he's now heard from people who are planning to have a gene scan just so they can participate. The next stage Swan has planned is a long-term ageing study, which she hopes will involve 10,000 people.

Beyond that, McCauley sees citizen-science genetic experiments expanding as far as broadband will take them. At the discussion group in San Francisco where I first meet him, he's asked to introduce himself to the room in three words. "Genomics for grandmas," he replies.

Back in his office, McCauley says he understands that the small scale and modest aspirations of his investigation of the motherfucker gene make it easy to dismiss. "In some ways, what we're doing with this little experiment is not a big deal. It's high-school science. Instead of building a volcano or measuring the temperature in the garden, we've got genomic information and some blood-test info."

But he says that the power of citizen genomics lies, by definition, in how personal its appeal is. Someone need be motivated only by self-interest – "Does Centrum work for me?" – to contribute their valuable genetic data to a study that might have consequences for everyone, and could otherwise have cost millions.

"Scientists are not people with degrees," he says. "Scientists are people who have questions and they're trying to find answers. I guess my big thing here, beyond asking if vitamins work, is that if you want a democratic, technological society you have to have people who understand how science really works. To me, that's what this is. If somebody does this and doesn't get a good result, or they look at it and never participate but read up about it a little, they've asked a question. They understand, viscerally, how the scientific method works and how people really apply it with today's technology. I think that's a big deal. If we don't do more of it, I think we're doomed."

A few weeks later, McCauley writes to say that he has decided to leave his job at Illumina, to take up a full-time position with Genomera. "Helping people understand what's going on in their bodies, in their lives," he says in an email. "That's a good life's work."

Meanwhile, McCauley's home science has ensured that he's taken steps to offset his increased risks of developing AMD. He wears spectacles that reduce his exposure to ultraviolet light. And every afternoon he takes 1,000mcg of L-methylfolate and other vitamins.

The chances are he'll be fine, and it remains possible that the vast areas of the genome about which we still know nothing might contain a variation that counteracts the one linked to AMD. "It's a crap shoot," he says. "Most of it's unknown." But in case none of his preventative measures works, he's taking out one more piece of insurance. On his computer, he has a checklist of places he wasn't expecting to visit until after he'd retired, but now wants to make sure he sees while he can still fully appreciate them. "The Grand Canyon, from the river, or Mount Fuji... I don't want to look at it out of the corner of my eye. I'm gonna go ahead and do some [of that] stuff now."

Adam Higginbotham is a writer based in New York. He wrote about wireless electricity in 11.10