

## Precision medicine inches along

More than a decade ago, the US National Institutes of Health launched the Human Genome Project. Genetics, we thought, would lead us to discover why we get sick and how to cure disease. We thought there would be a gene for everything, or maybe, at most, a handful of genes that control this behavior or that. It turns out the entire picture was a lot more complicated than we thought. David Dobbs, writing at BuzzFeed explains part of [the great disappointment](#):

Our DNA held far fewer genes than expected, almost 20,000, which was confusing. Few held obvious function. Some seemed to do nothing. Some seemed to work fine one day but not the next, or to do one thing in one situation and another in another. And these genes were surrounded by vast stretches of DNA material that aren't really genes, and which some geneticists called junk, starting a big fight.

[Junk DNA](#), for example, does a lot more than we anticipated by turning genes on and off, ramping up or dampening the amount of protein genes make and completely silencing others, rendering them inert. Many researchers now eschew the word 'junk' accordingly.

As part of the 2015 State of the Union speech, President Obama announced that the government would spend [\\$215 million on precision medicine](#) initiatives. Although some commented that the program would help build the bridge from basic research, like that of the Human Genome Project, to relevant applications for patient care, others were not so keen. Dr. Michael Joyner at the Mayo Clinic called the initiative '[Moonshot Medicine](#)' in an op-ed for the *New York Times*:

Like most "moonshot" medical research initiatives, precision medicine is likely to fall short of expectations. Medical problems and their underlying biology are not linear engineering exercises, and solving them is more than a matter of vision, money and will.

I'm not sure what else Joyner thinks goes in the recipe for medical breakthroughs besides funding, insight and effort.

Joyner is right in part, for a healthy person the idea of precision medicine doesn't have a lot to offer right now considering the hype and promise. For someone contemplating a pregnancy, a genetic test can tell you and your mate the carrier status 38 genetically-linked diseases and quantify the potential of passing on disease to your children.

Reporter Alexandra Ossola [wrote about her experiences](#) with this kind of testing at *Popular Science*. But because she would donate only one half of her own genetics to her hypothetical children, she said the test didn't really do much for her. She would need to come back with a partner or do a fetal test after conception in order to glean any really useful information about health impacts.

The other side of the coin are direct-to-consumer genetic testing companies like 23andMe and larger,

private companies in the genetic data collection business. These groups are working on association studies using large amount of data to link mutations to traits using some hard core mathematical analysis [Ossola writes](#):

Each individual has a small combination of mutations, and each of them affects which others are expressed. That could have big effects on where the environmental factors fit in and, eventually, whether or not a person develops a disease. “You need a lot of data to figure out how those mutations fit together. Right now, that’s still too complex to interpret,” Mittelman says.

The pursuit of complex genes to traits relationships are also a [highly lucrative business](#), Dobbs writes. Genetic information is bought and sold all the time as companies try to amass the raw genetic data needed to find those connections. Consumers interested in their own genetics provide the raw material that companies then spend millions swapping back and forth. The hype of precision medicine is an important part of [drawing more and more consumers](#) in Dobbs writes:

Big Genomics is converting hype to cash at unsettling speed. After the FDA told consumer genomics company 23andMe it could no longer sell people health data, the company began selling that data to drug and biotech companies. An entire industry, potentially fed by almost anyone who draws blood, spit, or biopsies from you, is emerging to do likewise. Its growth, along with the increasingly routine collection of genetic data by hospitals, will feed the genomics bubble while putting private genetic and health information at increased risk.

Although genetics hasn’t yet cured diabetes, ended the obesity crisis or eradicated malaria, it’s important to note that precision medicine has had some success stories. They don’t always make the front page, but they do make news. Last week, the FDA approved Orkambi, a combination of two genetically-derived drugs that repair faulty proteins in cystic fibrosis patients caused by genetic mutations.

And later this month, the FDA will consider approval of a new class of cholesterol-lowering medications called [PCSK-9 inhibitors](#). These drugs were developed through genetic analysis of people with genetically-linked extremely high cholesterol called familial hypercholesterolemia and people with genetically-linked extremely low cholesterol. People with extremely high cholesterol make too much of the protein while people with extremely low cholesterol make too little.

The impact genetics has had on cancers is even more pronounced. While genetic testing for healthy people may have a very limited impact on people’s life, for cancer patients genetics are vital. Nearly every tumor is sequenced to understand the genetic malfunctions driving the cancer’s growth and to pinpoint which treatments are good targets in each patient. The promising new field of [immunotherapies](#), for example, almost always rely on matching a treatment to the faulty genetics of a cancer. Development of those drugs is expanding rapidly.

Dobbs, however, thinks it's [too little too late](#) to justify the time, effort and money involved, and more importantly, the hype precision medicine has generated:

But when it comes to how genes shape the traits and diseases that matter most to us — from intelligence and temperament to cancer and depression — genetic research overpromises and underdelivers on actionable knowledge. After 110 years of genetics, and 15 years after the \$3.8 billion Human Genome Project promised fast cures, after more billions spent and endless hype about results just around the corner, we have few cures. And we basically know diddly-squat.

While neither Dobbs nor Joyner call for a referendum on precision medicine, or even a halt to funding these initiatives, they both suggest that perhaps some of these efforts should be diverted to support of less sexy clinical measures we know that work. Lifestyle changes are as effective in preventing disease and treating some conditions as any drugs on the market. Put the money where we know it works [says Joyner](#):

We would be better off directing more resources to understanding what it takes to solve messy problems about how humans behave as individuals and in groups. Ultimately, we almost certainly have more control over how much we exercise, eat, drink and smoke than we do over our genomes.

**Meredith Knight is a contributor to the human genetics section for Genetic Literacy Project and a freelance science and health writer in Austin, Texas. Follow her [@meremereknight](#).**

#### **Additional Resources:**

- [Arm chair geneticists looking for enlightenment could provide useful data](#), Genetic Literacy Project
- [Precision medicine revolution hits cost and tech barriers](#), Genetic Literacy Project
- [That 'Precision Medicine' initiative? A Reality Check](#), Genetic Literacy Project