

SPOTTED

Cautionary tale; lighter load; name game

BY KATIE MOISSE

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Cautionary tale

Doctors routinely perform prenatal screening for certain genetic disorders. But what about disorders **such as autism** where the evidence that any single genetic risk factor causes the condition is often slim?

The idea is not so far-fetched, as a number of genes **have ties to the condition**. A story in *The Atlantic* this week highlights the **hazards of this mentality**.

The piece, by journalist **Ed Yong**, explains that mutations classified as ‘pathogenic’ by one study may turn out to be harmless. Sadly, families have terminated pregnancies based on preliminary evidence.

“I think none of us really appreciated just how many rare, nasty-looking genetic variants exist in everyone’s genome,” **Daniel MacArthur**, associate director of medical and population genetics at the Broad Institute in Cambridge, Massachusetts, told *The Atlantic*.

New initiatives aim to tease out harmful mutations from normal variations in the genome by scouring sequences from thousands, and ultimately millions, of individuals. The goal is to be able to tell families, for instance, that a “variant has never been seen in 10 million healthy controls, but has been observed in 12 cases of the same disease,” MacArthur said. “Boom: There’s no ambiguity here.”

SOURCES:

The Atlantic / 16 Dec 2015

Clinical genetics has a big problem that's affecting people's lives

<http://www.theatlantic.com/science/archive/2015/12/why-human-genetics-research-is-full-of-costly-mistakes/420693/>

Planning ahead

The National Institutes of Health (NIH) unveiled their **strategic plan** for the next five years this week. The 48-page report kicks off by highlighting the sad state of federal research funding, which has dropped steadily since 2003.

“NIH currently funds about one in six grant applications, compared to its historical funding rate of one in three,” the report reads.

Beyond this plea for support, the report describes cutting-edge tools that could accelerate biomedical research, such as techniques for **analyzing gene expression in individual cells**. It also details plans to “catalyze innovative research” by **promoting high-risk, high-reward projects**.

SOURCES:

National Institutes of Health / 16 Dec 2015

NIH-wide strategic plan

<http://www.nih.gov/sites/default/files/about-nih/strategic-plan-fy2016-2020-508.pdf>

Lighter load

Many scientists spend a large chunk of their time writing grant proposals, applying for approval from ethics committees and editing manuscripts for publication. **C. Michael Stein**, professor of medicine at Vanderbilt University in Nashville, Tennessee, argues that all this administrative work **distracts from the ultimate goal** of advancing science.

In an editorial published Wednesday in *Science Translational Medicine*, Stein says that many of these clerical tasks are useful in theory. But in practice, they add up to a lot of wasted time. He outlines steps to reduce the administrative burden on researchers, such as implementing a system in which only grants deemed good enough to fund require stacks of paperwork.

Proposed **changes to the ethics approval process** in the U.S. could also lighten researchers' administrative load. The changes would allow scientists from different institutions to file joint paperwork for collaborative projects and exempt some projects from the ethics review process altogether.

SOURCES:

Science Translational Medicine / 16 Dec 2015

Academic clinical research: Death by a thousand clicks

<http://stm.sciencemag.org/content/7/318/318fs49>

Menacing myths

Some myths never die, and science has its share of persistent ones.

On Wednesday, *Nature* highlighted **five lingering legends** that “are hurting people and holding back science.”

What made the cut? The notion that screening for all types of cancer saves lives, the idea that antioxidants are good for you, and the fear that the human race is doomed because of overpopulation.

SOURCES:

Nature / 16 Dec 2015

The science myths that will not die

<http://www.nature.com/news/the-science-myths-that-will-not-die-1.19022>

Name game

Finally, a bit of humor for the holidays.

Slate published a list of, let's say, **'misguided' research paper titles** this week. The headlines range from clever (You Probably Think This Paper's About You: Narcissists' Perceptions of Their Personality and Reputation) to silly (Hung Jury: Testimonies of Genital Surgery by Transsexual Men) to unintentionally amusing (Role of Childhood Aerobic Fitness in Successful Street Crossing).

SOURCES:

Slate / 15 Dec 2015

Scientists' silly, dark, and sometimes inappropriate humor

http://www.slate.com/articles/health_and_science/science/2015/12/the_best_funny_clever_or_offensive_science_paper_titles.html
