SPOTTED

Diagnostic odyssey; breaking through; leading role

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Diagnostic odyssey

Genetic disorders can be difficult to diagnose, as two parents in Houston, Texas, discovered firsthand.

Their son was born with **a confusing combination of features**: developmental delays, a disproportionately large head, extreme flexibility and weak muscles. After nearly four years of seeing specialists, they were no closer to knowing the cause of their son's condition. Then they took him to see **Daryl Scott**, a genetics specialist at Baylor College of Medicine in Houston.

Scott used a database of genetic abnormalities in mice and a database of human genetic sequences to trace the boy's features to a missing piece of the X chromosome. Scott combed a database of people with unidentified genetic disorders for other cases and found two other boys in the world with the same chromosomal abnormality and characteristics. He later found a third case through a colleague.

"That question mark dissolved," the boy's mother told *STAT* in a story this week. "I felt like I wasn't alone anymore."

Last month, *Spectrum* wrote about a mother's six-year quest for answers about her own son's genetic disorder, **a rare form of autism** that has been linked to mutations in the gene ADNP. She discovered a key sign of the condition: early appearance of teeth in babies.

SOURCES:

STAT / 16 May 2017
After four years, it took a geneticist a few hours to unravel a boy's puzzling illness
https://www.statnews.com/2017/05/16/medical-mystery-genetics/

Breaking through

Burnout is a prevalent problem among researchers, particularly graduate students and postdoctoral researchers, according to a story in *Nature* this week.

The term refers to a feeling of overwhelming fatigue and loss of motivation. It often results from working long hours for months on end. Although burnout is not a medical diagnosis, it can cause a drop in productivity and lead to mental health issues such as depression.

Perhaps the best way to avoid burnout is to take breaks.

“Knit, play computer games, build chainmail — anything that is completely different from thinking about research questions,” **Kay Guccione**, manager of researcher mentoring and coaching at the University of Sheffield in the United Kingdom, told *Nature*.

Guccione started the Twitter hashtag #takebreaksmakebreakthroughs to remind herself to limit her work hours and cultivate non-research interests.

SOURCES:

Nature / 17 May 2017
Work-life balance: Break or burn out
http://www.nature.com/nature/journal/v545/n7654/full/nj7654-375a.html

Leading role

An actor with autism will **play the lead role** in the play “**The Curious Incident of the Dog in the Night-Time**” in upcoming off-Broadway productions in **Indianapolis**, Indiana, and **Syracuse**, New York, *Salon* reported this week.

The play is based on a best-selling mystery novel by Mark Haddon that follows 15-year-old Christopher John Francis Boone, who has a number of autism features, as he investigates a crime.

“Playing this role is such an honor because there’s a lot of misinformation and stereotypes around autism,” actor Mickey Rowe, who will play the role of Christopher, told *Salon*. “All too often we learn about autism from non-autistic people instead of going straight to the source and learning about autism from autistic adults.”

Neurotypical actors have long been cast as characters with autism, from Dustin Hoffman in the film “Rain Man” to Jim Parsons in the television show “The Big Bang Theory.” Journalist **Matthew Rozsa**, who wrote the *Salon* story and has autism himself, praised the play’s casting decision.

“As an autistic person, I have thirsted to see individuals from our community represent us in popular culture,” he writes.

SOURCES:

Salon / 15 May 2017
For once, an autistic role will be played by an autistic actor
http://www.salon.com/2017/05/15/mickey-rowe-the-curious-incident-of-the-dog-at-night-time-autistic/

Life’s work

For almost 70 years, 98-year-old **Brenda Milner** has been breaking new ground in the field of neuroscience. And she is still going strong, according to a profile in *The New York Times* on Monday.

“People think because I’m 98 years old I must be emerita,” Milner, professor of psychology at McGill University in Montreal, Canada, told the newspaper. “Well, not at all. I’m still nosy, you know, curious.”

Milner has been involved in **some of the most influential neuroscience research** of the past century. Her early work on **Henry Molaison**, or H.M., a man who had memory loss after surgery to treat his **epilepsy**, identified the hippocampus as a brain area essential for memory formation. Continued research on H.M. led Milner to conclude that there are two memory systems in the brain: one for names, faces and experiences, and the other primarily for motor skills such as riding a bike.

A story in *Spectrum* last year explored the role of **the hippocampus** in autism. Some people with autism have difficulty remembering social information and describing autobiographical events, yet excel at tasks such as memorizing names, dates, definitions and facts.

SOURCES:

The New York Times / 15 May 2017
Brenda Milner, eminent brain scientist, is ‘still nosy’ at 98
https://www.nytimes.com/2017/05/15/science/brenda-milner-brain-cognitive-neuroscience.html

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Speaking out

An editorial in *Neuron* this week explains how neuroscientists can **communicate more effectively** with policymakers, service providers and the public.

Authors **Nathaniel Kendall-Taylor** and **Pat Levitt** draw on lessons they have learned from two decades of communicating their own research on early child development. Levitt's clinical work investigates children with autism who **also have gastrointestinal and other conditions**.

"There is an enormous gap between what we *know* about the power of neuroscience to spark substantive societal change and what we *do* with neuroscience research to inform policymakers, practitioners and members of the public," they write.

One of their recommendations is to use narratives and storytelling techniques to convey scientific information. They also suggest framing scientific issues in ways that allow nonscientists to grasp the broader importance of these issues.

"At a time when ideology divides the country and politics polarizes the population, there is an even greater need to tell science stories," they write. "Science transcends ideology and identity politics and focuses people's attention on issues, their causes and consequences, and solutions."

SOURCES:

Neuron / 17 May 2017
Beyond hat in hand: Science advocacy is foundational for policy decisions
http://www.cell.com/neuron/abstract/S0896-6273(17)30396-3

Job news

Making a career move? Send your news to **jobmoves@spectrumnews.org**.
