SPOTTED

Autism unsurprised; diagnostic camouflage; Neanderthal legacy and more

BY **EMILY WILLINGHAM**

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Autism unsurprised

Could it be that people with autism are fatalistic about change because the environment so consistently violates their desire for sameness? That's **one possible implication** of a small study published 31 July in *Nature Neuroscience*. Study participants with autism showed less surprise than neurotypical participants when their expectations were not met. In this case, the expectation was that a particular image would follow a specific sound.

The more pronounced participants' autism features, the less likely they were to find an unexpected image surprising. Paradoxically, this low reactivity could mask a constant hum of **sensory hyperarousal** for people with autism. Researcher **Rebecca Lawson** of the Wellcome Trust Centre for Neuroimaging at University College London **speculated in a statement** that if "people with autism are more often expecting volatility, that could help explain their propensity to sensory overload, enhanced perceptual functioning and context insensitivity."

SOURCES:

Nature Neuroscience / 31 Jul 2017
Adults with autism overestimate the volatility of the sensory environment
http://www.nature.com/neuro/journal/vaop/ncurrent/full/nn.4615.html

High-throughput potential

A pair of studies underscores the possibility that sequencing may become the first stop for diagnosing certain childhood conditions. Improvements in techniques for screening samples in bulk have greatly enhanced the diagnostic yield of these tests. These improvements may make them more feasible and cost-effective as first-tier tests, as the two studies, published 31 July in *JAMA Pediatrics,* suggest.

The first study compared how well different genetic testing approaches identify sequence changes **associated with early-life epilepsy**. About one-third of people with autism will **also have an epilepsy diagnosis**. These investigators found the best performance with approaches such as whole-exome sequencing, which spells out the coding regions of DNA. Whole-exome sequencing yielded genetic diagnoses in 27.5 percent of the cases, compared with 17 percent with microarray tests, which identify large duplications or deletions of DNA. Microarray tests **are a common choice** for genetic testing in autism.

The second study used whole-exome sequencing to try to pin down diagnoses in children thought to have various conditions traceable to single genes. The researchers **identified the causative gene variants in 52 percent** of cases. In one-third of these cases, the variant had not been at the top of the list of diagnostic possibilities.

SOURCES:

JAMA Pediatrics / 31 Jul 2017
Diagnostic impact and cost-effectiveness of whole-exome sequencing for ambulant children with suspected monogenic conditions
http://jamanetwork.com/journals/jamapediatrics/fullarticle/2643959

Autism buzz

A subset of honeybees that react atypically in hive social situations share some genetic traits with people who have autism. Involved genes are linked to ion flow in neurons and to stress-related proteins. Researchers published their **buzzy evolutionary findings** 25 July in the *Proceedings of the National Academy of Sciences*.

SOURCES:

Proceedings of the National Academy of Sciences / 25 Jul 2017
Deep evolutionary conservation of autism-related genes
http://www.pnas.org/content/early/2017/07/25/1708127114

Diagnostic camouflage

Women with autism use ‘camouflage’ — mimicking neurotypical behaviors — more often than men do to **navigate social settings**. Evidence suggests that the apparent autism gender gap **may trace to this capacity of girls and women** on the spectrum to practice social camouflage. NPR dug into the phenomenon in a 31 July story.

SOURCES:

NPR / 31 Jul 2017
'Social camouflage' may lead to underdiagnosis of autism in girls
http://www.npr.org/sections/health-shots/2017/07/31/539123377/social-camouflage-may-lead-to-underdiagnosis-of-autism-in-girls

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Rett therapy

A **significant proportion** of girls and women with Rett syndrome also have autism. Rett traces to loss of **MECP2**, a protein with multiple roles, including production of a critical brain growth factor. A drug mimic of this growth factor **improves brain function** in a Rett mouse model, according to results published 1 July in *Disease Models & Mechanisms*.

SOURCES:

Disease Models & Mechanisms / 01 Jul 2017
A small-molecule TrkB ligand restores hippocampal synaptic plasticity and object location memory in Rett syndrome mice
http://dmm.biologists.org/content/10/7/837

Neanderthal legacy

How far back in the human lineage can we trace conditions like autism? New findings suggest that the more Neanderthal DNA a person’s genome contains, the more **the person’s brain is like that of Neanderthals**. Neanderthal-associated brain regions are linked to tool use and visual discrimination and may represent tradeoffs with the ‘social brain,’ according to findings published 24 July in *Scientific Reports*.

SOURCES:

Scientific Reports / 24 Jul 2017
Neanderthal-derived genetic variation shapes modern human cranium and brain
https://www.nature.com/articles/s41598-017-06587-0

Blaming parents

About **one-third of people with fragile X syndrome** also have an autism diagnosis. A test for the associated mutation, which is the most common genetic cause of intellectual disability, can detect it in utero. Doctors in Australia are **calling for guideline updates** to include this testing option for pregnant women, reported *The World Today* on 30 July.

SOURCES:

The World Today / 30 Jul 2017
Fragile X: experts say all women should be offered screening for genetic condition
http://www.abc.net.au/news/2017-07-31/all-women-should-be-offered-fragile-x-screening:-study/8759570

Evidence obstacles

The biggest obstacle public-health workers face in the autism context is giving people on the spectrum access to evidence-based care, says **Sally J. Rogers**, co-developer of the **Early Start Denver Model**. In a wide-ranging interview published 2 August in the *Bulletin of the World Health Organization*, Rogers talks about early-intervention practices and **developmental commonalities** between people with and without autism.

SOURCES:

Bulletin of the World Health Organization / 02 Aug 2017
Making evidence-based approaches to autism accessible
http://www.who.int/bulletin/volumes/95/8/17-030817/en/

Funding aspirations

The U.S. **Interagency Autism Coordinating Committee** is calling for a doubling of autism research funding by 2020. The advisory panel, which consists of representatives from the autism

community and the federal government, wants federal and private funding to reach $658 million by the target year, which **means a nearly 15 percent annual increase**, reported *Disability Scoop* on 1 August.

Joshua Gordon, committee chair and director of the National Institute of Mental Health, called the request “a reasonable and aggressive start … that we can spend wisely and efficiently,” *Disability Scoop* reported.

SOURCES:

Disability Scoop / 01 Aug 2017
Federal panel wants autism funding doubled
https://www.disabilityscoop.com/2017/08/01/federal-panel-autism-doubled/24000/

News tips

Do you have a new paper coming out? Are you making a career move? Did you see a study or news story that you want to share? Send your news tips to **news@spectrumnews.org**.