

NEWS

Community Newsletter: Completed human genome, chromosome 16 convergence, avoiding ableism

BY SPECTRUM

3 APRIL 2022

Welcome to April and **#AutismAcceptanceMonth**.

The cover of *Science* on Friday hailed a milestone years in the making: The **Telomere-to-Telomere Consortium** has at last **filled in nearly all of the gaps** from the original Human Genome Project, adding about 200 megabases of genetic information, more than 1,900 genes and a complete sequence for the Y chromosome.

To mark the occasion, **Evan Eichler**, professor of genome sciences at the University of Washington in Seattle, tweeted for the first time ever, noting that the project, which involved more than 100 researchers, had been a “**wonderful collaborative team effort**.”

It took 20+ years but we finally finished a human genome. What a wonderful collaborative team effort! **#T2T** (my first tweet)

— Evan Eichler (@EichlerEE) **March 31, 2022**

“Congratulations!” tweeted **Claudia Gonzaga-Jauregui**, assistant professor of genetics at the National Autonomous University of Mexico in Mexico City, calling the results an “**awesome resource**” — and noting that Eichler’s tweet comes nearly a decade after he created an account.

Congratulations! Great achievement and awesome resource for the human genomics community. (You've had Twitter since 2013?!)

— Claudia Gonzaga-Jauregui (@cgonzagaj) **April 1, 2022**

Simon Barnett, a genomics analyst at ARK Investment Management in St. Petersburg, Florida, joked that Eichler had “**set a new bar for first tweets.**”

You've set a new bar for first tweets! Congratulations!

— Simon Barnett (@sbarnettARK) **March 31, 2022**

“Welcome to **the dark side!**” **Jonathan Sebat**, professor of psychiatry and cellular and molecular medicine at the University of California, San Diego, chimed in.

Welcome to the dark side! <https://t.co/SN1UCTVX0w>

— Jonathan Sebat (@sebatlab) **March 31, 2022**

Over in the realm of autism-specific genetics, **Daniel Weiner**, a graduate student at Harvard University working in **Elise Robinson**'s laboratory, issued a 14-tweet thread about their new **preprint** that finds **functional convergence** of common and rare genetic variants linked to autism around the chromosomal region 16p11.2.

Excited to share our latest work!!

We uncover functional convergence of common+rare genetic risk factors for autism around 16p11.2, a region of longstanding mystery in neuropsych research ????

???? ???? (1/14)<https://t.co/W0vUbFFHYG>

— Dan Weiner (@danweiner92) **March 24, 2022**

Both common and rare risk factors “**converge on decreased gene expression**” across 16p, “facilitated by 3D genome architecture,” he writes.

In summary:

Hypothesized a model of ASD liability arising from 16p:

Distinct genetic risk factors – common 16p ASD PRS / rare 16p11.2 CNV – converge on decreased gene expression in region of increased brain relevance, facilitated by 3D genome architecture (10/14) pic.twitter.com/HXqqz2Zo8X

— Dan Weiner (@danweiner92) **March 24, 2022**

“This was the **most fun**,” tweeted Robinson, assistant professor of epidemiology at the Harvard University, asking for feedback.

This was the most fun. Check out the paper - we'd be excited to hear your ideas.
<https://t.co/XpNrmQ2AJx>

— Elise Robinson (@elisebrobinson) **March 24, 2022**

The **Policy Impact Project** — launched by **Lindsay Shea**, director of the Policy and Analytics Center at Drexel University in Philadelphia, Pennsylvania, to link autism research to policy change — tweeted about its upcoming “**power lunch**” for autism researchers. The webinar, scheduled for 13 April from noon to 1 p.m. EST, will discuss how to avoid ableist language.

Our next Policy Power Lunch will feature the authors of *Avoiding Ableist Language: Suggestions for Autism Researchers*. [@KristenBott](#), [@drstevenkapp](#), [@NinaLester](#),

@Noahsasson & @BN_Hand

Click here to register and to learn more about the panelists: <https://t.co/0fz6UMOddq>
<https://t.co/s3nltNgoFv>

— Policy Impact Project (@impact_policy) **March 28, 2022**

That's it for this week's Community Newsletter! If you have any suggestions for interesting social posts you saw in the autism research sphere, feel free to send an email to news@spectrumnews.org.

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