Trending: Seahawks star Richard Sherman on Donald Trump, Microsoft Surface, power of grit, favorite apps (http://www.geekwire.com/2016/seahawks-star-richard-sherman-donald-trump-microsoft-surface-power-grit-favorite-apps/)

Space & Science

UW prof delves into why we got genes linked to autism, but Neanderthals didn't

BY ALAN BOYLE (HTTP://WWW.GEEKWIRE.COM/AUTHOR/ALANBOYLE/) on August 3, 2016 at 8:53 pm

(http://cdn.geekwire.com/wp-content/uploads/2016/08/160803-skulls2.jpg)

These skulls of a modern human and a Neanderthal are from the Cleveland Museum of Natural History. (Credit: DrMikeBaxter / HairyMmuseumMatt via Wikimedia)

One of the biggest genetic differences between humans and other members of the primate family tree, including Neanderthals, predisposes people to a type of autism. The stretch of DNA appears to be an important piece of the human genome, but why?

University of Washington genome scientist Evan Eichler and his colleagues on an international research team focus on that question in a study published today by the journal Nature (http://www.nature.com/nature/journal/vaop/ncurrent/full/nature19075.html).

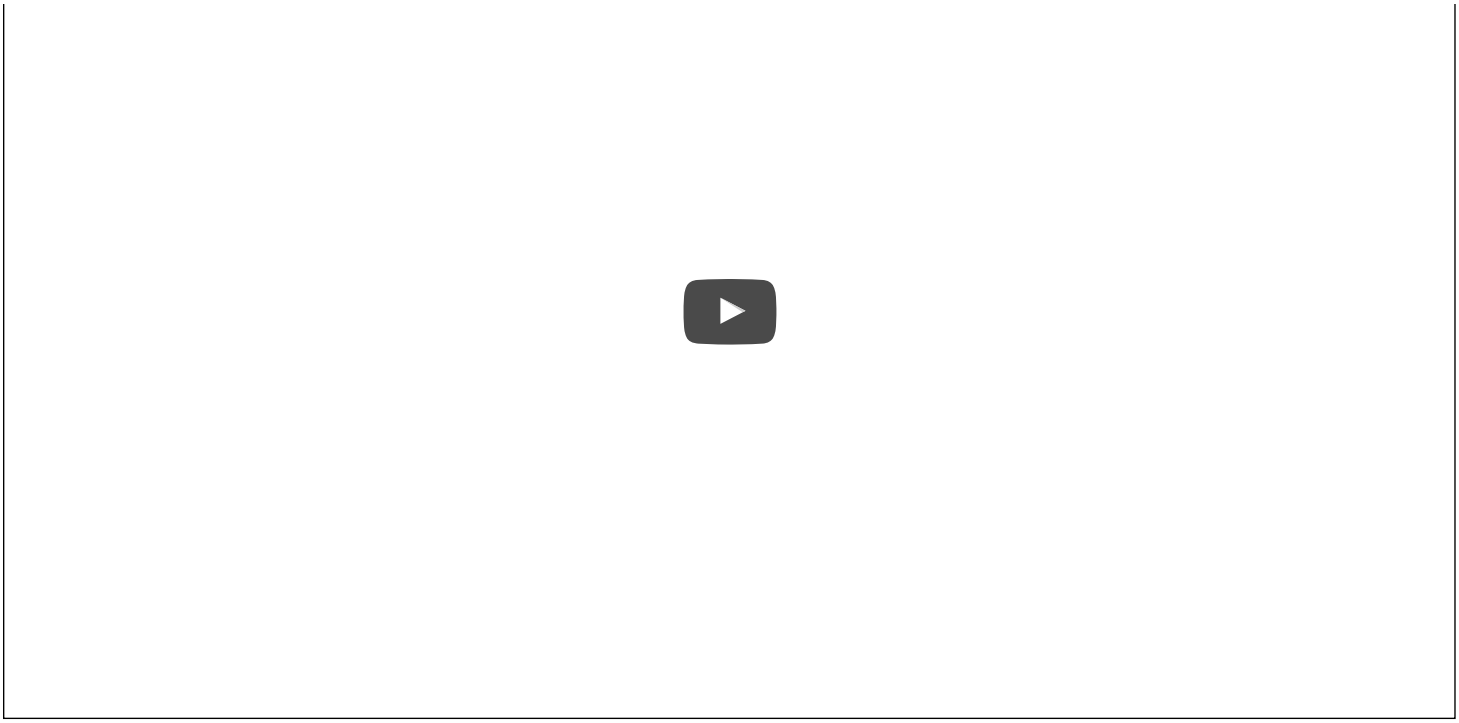
The key genetic structure consists of 95,000 molecular base pairs in a region on chromosome 16 that's known as 16p11.2. The structure includes 28 genes, flanked by blocks of DNA with duplicated sequences of genetic code known as copy-number variants.

Eichler's team compared the genomes of modern humans with the genetic code for chimps, gorillas and orangutans, as well as the code for Neanderthals and another strain of extinct pre-humans known as Denisovans. Humans were the only ones to have the structure in the 16p11.2 region.

The researchers' analysis indicates that the structure appeared in our ancestors' genome relatively suddenly, about 280,000 years ago. That time frame is about 80,000 years before anatomically modern humans – that is, Homo sapiens – show up in the fossil record.

"Most duplications in our genome are millions of years old, and the speed at which this structure transformed our genome is unprecedented," Eichler said in a news release (http://hsnewsbeat.uw.edu/story/human-neanderthal-gene-variance-involved-autism).

UW Medicine's influential scientists: Evan Eichler



Researchers have previously taken note of the structure in 16p11.2 because a mutation that results in the deletion of genetic code is correlated with about 1 percent of autism cases (http://www.nejm.org/doi/full/10.1056/NEJMoa075974). But the structure also contains a beneficial gene called BOLA2. That gene seems to plays a part in helping cells capture iron more efficiently and making iron available to proteins that need it.

The iron-building benefit is most pronounced early in cell development. “This ability to help humans to acquire and use this essential element early in life might confer a significant enough benefit to outweigh the risk of having some offspring with autism,” Eichler said.

There are a couple of additional twists to the tale: One is that the structure in the 16p11.2 appears to include a new gene that was created by fusing two regions of a BOLA2 gene with three regions from another gene. “We’re going to work with other research teams to find out what it does, but so far we haven’t a clue,” Eichler said.

The other twist is that Eichler has an autistic niece who’s now in her 20s. He’s always wondered why some children are born with autism while others are not.

“With genome technologies and our understanding of genetics and genomes, we’re now just beginning to crack the case,” Eichler said. “My hope is that in five to 10 years, you would treat children with different genetic causes of autism with different therapies that would be tailored really for those specific kids.”

In addition to Eichler, 29 other researchers are co-authors of the Nature paper, titled “Emergence of a Homo Sapiens-Specific Gene Family and Chromosome 16p11.2 CNV Susceptibility.” (http://www.nature.com/nature/journal/vaop/ncurrent/full/nature19075.html) Eichler is an investigator of the Howard Hughes Medical Institute as well as a UW professor of genome sciences.

Love space and science? *Sign up for our GeekWire Space & Science email newsletter (https://www.geekwire.com/space-newsletter/) for top headlines from Alan Boyle, GeekWire’s aerospace and science editor.*