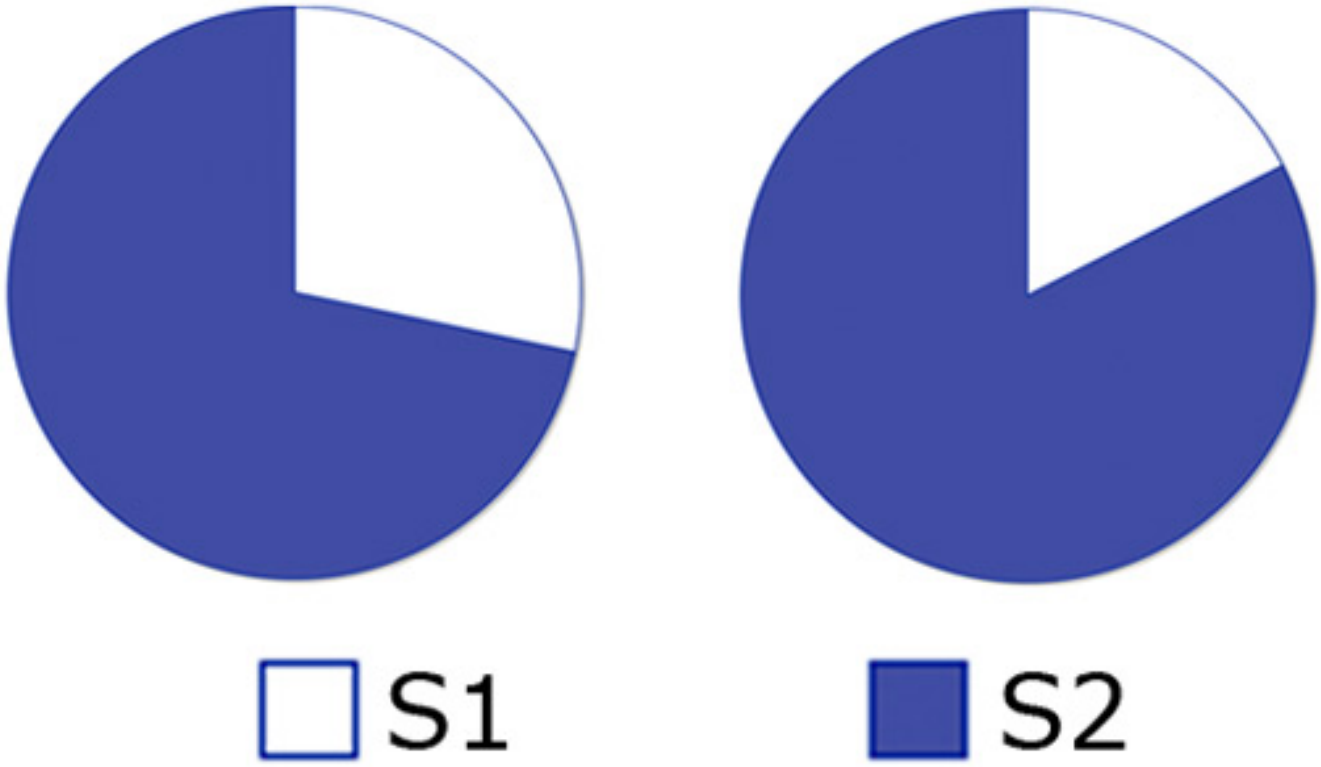
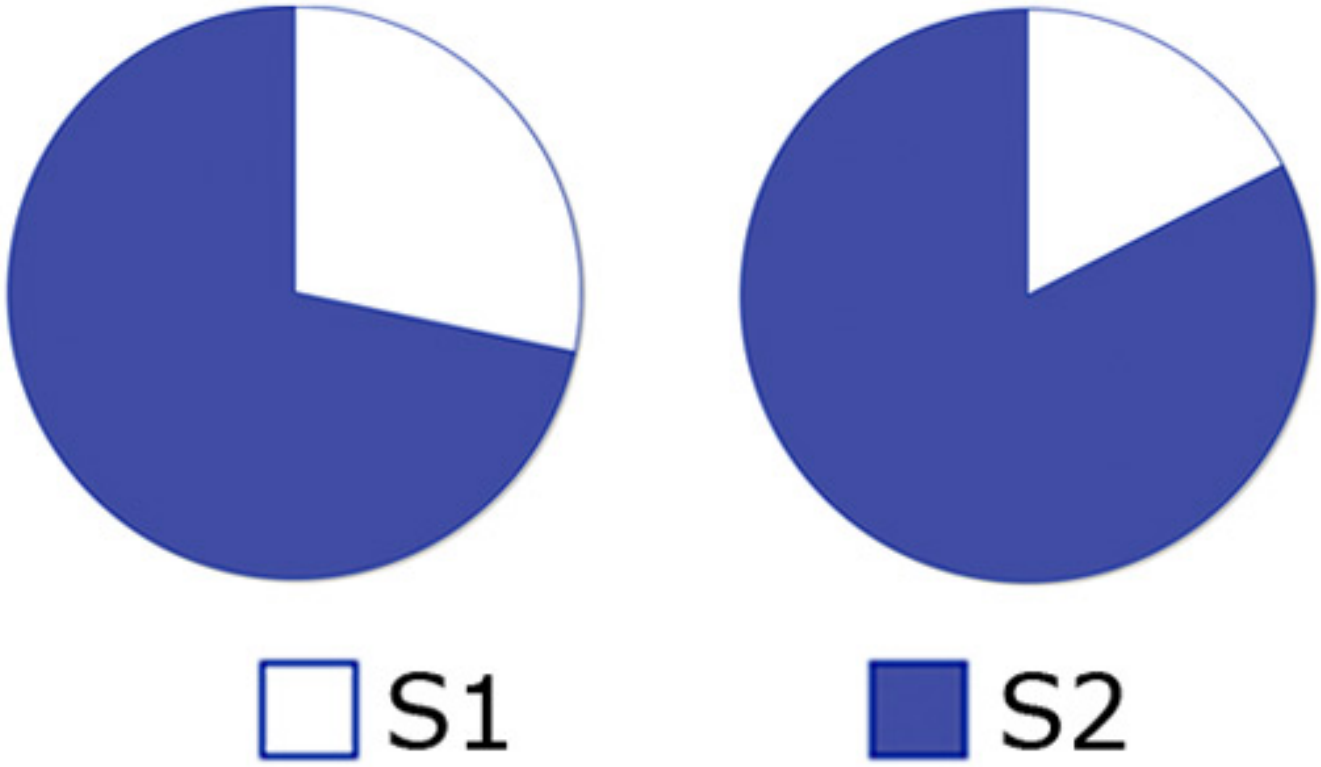
NEWS

# Genetics: Common mutation linked to risk of mental disorders

BY **JESSICA WRIGHT**

27 SEPTEMBER 2010






Relative risk: About 82 percent of the general population (right) and 72 percent among Asians (left) carry the S2 configuration of 16p12.1, a direct risk factor for the chromosomal region's deletion.

A common variation within a region on chromosome 16 **puts a large proportion of the general population at risk for intellectual disability**, according to a study published in September in *Nature Genetics.*

**Evan Eichler** and his colleagues reported in 2009 that **a small deletion in the 16p12.1 chromosomal region**, in combination with other mutations, can lead to inherited mental disorders.

The new study reports that this genetic region exists in two different structural forms, S1 and S2, both of which differ from the reference human genome.

The S2 configuration, seen in about 82.4 percent of the population, is much larger, containing several repeated regions. The duplications, and their specific orientation on either side of the region, make S2 prone to the type of rearrangement that would lead to its deletion.

The presence of the S2 configuration is a direct risk factor for the 16p12.1 deletion. The

researchers report that 34 of the 35 tested individuals with this mutation carry two copies of the S2 structure. None had two copies of S1.

When the researchers compared the chromosomal region among several species of primates, they found that it seems to have expanded during evolution to form the S1 and S2 configurations. Chimpanzees, gorillas and humans all have several duplications in this region compared with orangutans, gibbons and macaques, suggesting that the expansion took place in a relatively short evolutionary time.