



# Identifying Autism Genes by Tracking Gene Mutations



*Caption: The diagram is a pedigree of a mother (AU-3104), father (AU-3103), daughter (AU-3102), and son (AU-3101). Women are represented by circles and men by squares. Nonautistic individuals are shown as open symbols and autistic individuals as shaded symbols. The double line connecting the two parents indicates that they are related. Each horizontal bar represents a map of single nucleotide polymorphisms (SNPs) along one arm of chromosome 3 of the family members. Red and blue vertical stripes indicate homozygous SNPs for either one of two alleles, yellow stripes represent heterozygous SNPs, and white gaps represent genetic deletions. The horizontal black line demarcates a region with a pattern of homozygosity found in the autistic individual but not in the nonautistic individuals.*

OBSERVATIONS, NOTES & QUESTIONS	
BACKGROUND INFORMATION	BIG IDEAS, NOTES & QUESTIONS
<p>Autism is a disability that involves differences in development that can manifest in a variety of ways. Autistic individuals may engage in repetitive behaviors, have intensely focused interests, and have differences in communicating and interacting with others in social situations. The differences associated with autism can be strengths, neutral traits, or impairments depending on the social context in which they occur.</p> <p>Autism has a strong genetic component. Researchers who study autism genetics should work closely with the autistic community to make sure that their research will benefit autistic people. In general, the autistic community does <i>not</i></p>	

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want to prevent or “cure” autistic differences, since those are not inherently bad, just another way of being. Instead, they may want to identify genes related to autism to help treat co-occurring genetic conditions that decrease quality of life (for example, epilepsy, a condition that causes seizures). However, it has been difficult to identify specific genes related to autism, likely because many different genes are involved.

When parents share recent ancestry (for example, if cousins have a child together), their children are more likely to have conditions caused by recessive genetic mutations. This is because relatives are more likely to have the same recessive mutations, which may be rare in the population as a whole, and they can pass these mutations on to their children, who will then be homozygous for the mutations. In these families, scientists use a technique called “homozygosity mapping” to identify the locations of mutations associated with a recessive genetic condition. The technique basically involves finding regions in the genome where the DNA of an individual with the recessive condition is homozygous. Scientists can then further study those regions to determine which genes and mutations are there.

To identify regions of homozygosity, scientists map single nucleotide polymorphisms (SNPs), which are positions throughout the human genome where individuals are known to have different nucleotides. For each SNP, researchers determine whether an individual is heterozygous or homozygous at that location. If an individual is homozygous for two adjacent SNPs, which are typically mapped an average of 6,000 base pairs apart, then one can reasonably assume that the entire stretch of DNA between the two SNPs will also be homozygous. Scientists then determine which regions of homozygosity are in the genome of the person with the condition but not in the genomes of their relatives without the condition. Such regions are likely to hold genes related to the individual’s condition.

Eric Morrow and colleagues mapped SNPs on chromosome 3 in a family in which the parents shared recent ancestry and had an autistic son and a nonautistic daughter.