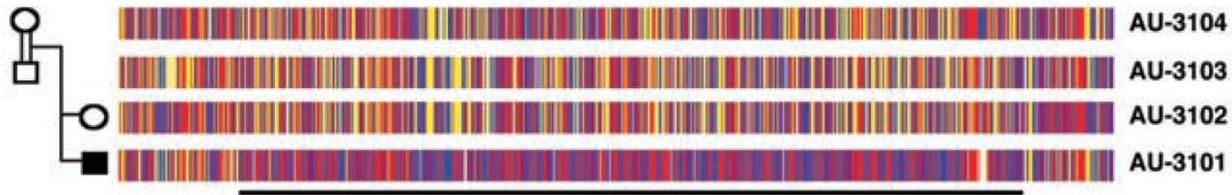




## Identifying Autism Genes by Tracking Gene Mutations



**Caption:** The diagram is a pedigree of a mother (AU-3104), father (AU-3103), daughter (AU-3102) and son with autism (AU-3101). Females are represented by circles and males by squares. Unaffected individuals are open symbols and the autistic son is a shaded symbol. 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 not found in the boy with autism's parents or sister.

### BACKGROUND INFORMATION

Autism is a disorder that affects brain development and is usually characterized by repetitive behaviors and interests, and by difficulties in communicating and interacting with others in social situations. Autism has a strong genetic component; however, it has been difficult to identify which specific genes contribute to autism. It's likely that many different genes are involved.

When parents share ancestry (for example, if cousins have a child together), their children are at increased risk of 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 disorder. The technique basically involves finding regions in the genome where the DNA of an individual with the recessive disorder 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 variations found throughout the human genome at known locations. 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 affected person's genome but not in the genomes of their unaffected relatives. Such regions are likely to hold the genes responsible for the individual's disorder.

Dr. Eric Morrow and colleagues mapped SNPs on chromosome 3 in a family in which the parents shared ancestry and had a son with autism and a daughter without the disorder.