Autism Families Study

Children and families seen at the Social Communication Disorders Clinic are invited to take part in our primary research project.

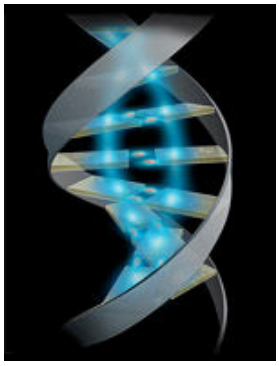
The child first and always

People with autism and autistic spectrum disorders usually have difficulties acquiring social skills that are important to get along with other people. As we understand more about the nature of these difficulties, the more effectively we will be able to research the genetic origins of conditions such as autism and Asperger's syndrome.

We want to understand more about the genes that influence the processing of social information, and hope eventually to discover new treatments for these conditions.

To understand how DNA can influence what we will be like in terms of our personality and abilities, it is useful to know more about what DNA is. Basically DNA is a molecule that is found in all cells of the body, and it contains instructions that provide almost all of the information necessary for us to grow and function.

Our brain is made up of cells and these cells receive their instructions from DNA, and from day to day experiences. If our cells are given the wrong instructions by our DNA, then our brains will not develop normally. We may then have learning difficulties or other problems in everyday life that affect the way in which we relate to other people. DNA is the chemical responsible for preserving, copying and transmitting information within cells and from generation to generation.



Each strand of DNA is made up of sugars, phosphates and bases. With the exception of identical twins, the sequence of the bases is different for everyone, which makes each of us unique. Segments of DNA form what are called chromosomes, and within the chromosomes genes are found. The complete set of genes is called a genome. Although we all look quite different from one another, we are surprisingly alike at the DNA level. The DNA of most people is 99.9 percent the same. We have approximately three billion base pairs of DNA in most of our cells, but only about 3 million base pairs are responsible for the differences among us. Yet these DNA base sequence variations influence most of our physical differences and many of our other characteristics, as well. Sequence variations occur in our genes, and the resulting different forms of the same gene are called alleles. People can have two identical or two different alleles for a particular gene.

The purpose of our research is to discover which alleles are important for increasing the risk of developing conditions on the autistic spectrum. Because DNA is inherited, this risk runs in families, although not everyone with the risk allele will be autistic. They might, however, have some very subtle differences in the way in which their brain works – another important aspect of our research is to discover whether these subtle differences (which we call endophenotypes) also run in families.

There are big differences in the frequency with which males and females develop disorders that affect emotional and social development, such as autism. The reason for this sex difference has never been explained. We believe it is linked in part to genes on the sex chromosomes. If we can identify which genes are responsible for the unusual pattern of information processing and brain structure seen in disorders of social and emotional development, we hope to be able to discover a cause for autistic spectrum disorders. In due course we may even be able to find more effective ways of treating those disorders.