How genetically different are individuals diagnosed with autism compared with non-autistic people in the general population? According to a new study into the genetics of autistic spectrum disorder (ASD), there may be more similarities than previously thought.

ASD is a neurodevelopmental condition characterised by deficits in social behaviour and communication, along with restrictive and repetitive interests. They may include elements such as a lack of understanding of non-verbal communication, difficulties with social interaction, repetitive motor movements and speech, fixation with routines, particular foods and unusual objects, and extreme distress at small changes.

ASD is estimated to occur in around 1% of people worldwide. It’s thought to be caused by a combination of different genetic and environmental factors yet, despite extensive research over the past decade, its exact causes are not fully understood.

For thousands of rare diseases, genetic causes can be pinned down to a single genetic factor that has a large and detrimental biological impact. In Huntington’s disease, for example, a single gene mutation causes severe neurodegeneration and brain damage.

But this is not the case for nearly all common diseases, including asthma, heart disease, diabetes, arthritis and inflammatory bowel disease. For these common diseases the evidence points firmly to the existence of numerous genetic factors – often in the order of many thousands. In these cases, each genetic factor on its own has a tiny impact but, added together, they influence disease.

This is what has been found for ASD, where thousands of small genetic factors seem to be acting together in combination to produce a detectable difference in ASD-related traits.

Over the years, scientists discovered that almost all genetic risk factors that have been found for ASD can also be found in unaffected people from the general population. However, the effects of these genetic risk factors in non-autistic individuals were previously unknown. Until now.

In a recent study published in *Nature Genetics* (http://tinyurl.com/z2hg6qo) a team of international researchers, including Prof David Evans of the University of Queensland’s Diamantina Institute, studied thousands of autistic and non-autistic individuals from all around the world. The researchers wanted to find out what impact genetic factors for ASD have in the general population. To do so, they analysed genetic data from individuals diagnosed with autism, together with data from British children in the general population whose social and communication skills had been assessed.

They discovered that around one-quarter of the genetic factors
Constantino of Washington University referred to tensions between clinical judgements about where these thresholds should lie and the criteria for ASD described by the current Diagnostic and Statistical Manual of Mental Disorders (DSM-5). He argued that a compelling case can be made for adopting percentile-based thresholds for ASD criteria, much like IQ scales for intellectual disability. Certainly, the genetic findings of the Nature Genetics study add to the debate surrounding whether diagnostic procedures for ASD may be improved by focusing on continuum-based traits.

The study findings hold promising implications for the future of ASD research. First, it suggests that studying the behavioural traits of people in the general population could lead to new insights into what causes ASD. For example, determining the genetic factors and biological processes in normally-developing children may help to determine processes that are disturbed in children diagnosed with ASD. This has the huge advantage that any individual from the general population can be studied, and finding individuals diagnosed with ASD will not necessarily be required.

A second exciting prospect is that very large studies of the genetic causes of ASD-related traits could be undertaken much more easily by studying traits in the general population. While a handful of specific genetic mutations have already been linked to ASD, this number falls far short of the thousands believed to be implicated in the condition. The lack of known specific genetic factors for ASD is at least partly due to the fact that the very small individual genetic effects cannot be detected from studies of the sizes that are currently available.

This is because for genetic studies of common diseases, extremely large numbers of people are needed to uncover these small, specific genetic effects. In some cases, hundreds of thousands of people would be required. While this number would be difficult to achieve for a study of ASD individuals, this is largely feasible if scientists were able to study individuals in the general population.

Essentially, undertaking future ASD-related research in large numbers of the general population could potentially address some of the existing issues of study size currently limiting genetic studies of ASD. This could lead to the discovery of vastly more genetic factors for ASD-related traits.

Beyond ASD, studying continuum-based behavioural traits that affect the whole population also holds encouraging prospects for many other neurological and psychiatric diseases. Hence the new study could lead to improved genetic research for many complex and elusive neuropsychiatric conditions, such as schizophrenia.