A strong genetic disorder of autism

Health & Medicine, Healthcare

Over the recent years, autism has greatly become people’s concern. Autism has been defined as a severe neuropsychiatric disorder characterized by impairments in social and language abnormalities (American Psychiatric Association, 2013). Various definitions of autism have now been known to be universal as it is recognized as a spectral condition (Baird et al., 2016). Bailey et al’s. (1995) research is an ambitious attempt to resolve the contradictory argument whether autism is often or a consequence of environmentally determined brain damage or whether autism is a genetically determined disorder. The researchers reassessed the original sample from the twin study of Folstein and Rutter (1977a, b). A new British twin sample of autistic twins was recruited in a manner similar to the original study. Details on same-sex pairs, identical and non-identical, wherein either one or both twins had autism were requested from child psychiatrists and paediatricians. A standardized investigator- based interview gathered information on all the behavioural traits considered central to autism according to the ICD-10 criteria (World Health Organization, 1992). Twin participants were also assessed using the Autism Diagnostic Observational Schedule (Lord et al. 1989).

A neurodevelopmental examination was included in search for the stigmata of tuberous sclerosis, neurofibromatosis and the Fragile X syndrome; and the minor congenital anomalies examination of Waldrop & Halverson (1971). Zygosity was determined by typing of nine blood groups. Cytogenetic examination (including Fragile X) and plasma and urine amino acid chromatography were also performed. Results indicated that autism is strongly influenced through genetic control as multiple genetic loci were

involved in the mutation. 69% of the monozygotic (MZ) twins and 0% of the dizygotic (DZ) twins were concordant for autism from the new sample, concordance rates in the original sample were 36% in MZ pairs and 0% in DZ pairs. Concordance rates for cognitive and social abnormalities were 88% in MZ pairs and 9% in DZ pairs from the new sample, while 82% in MZ pairs and 10% in DZ pairs from the original sample. These findings support the assumption that autism is under a high degree of genetic component.

This study presented a few limitations. To begin with, the researchers did not acknowledge how environmentally determined aetiological factors also have a great contribution towards autism. The previous twin study by Steffenburg et al. (1989) suggested that severe perinatal hazards may be environmental factors that convert an inherited broader phenotype into autism. Environmental toxins such as mercury quite possibly from the sea food consumption by the expectant mother have been found to be amongst the few contributors towards the cause of autism. The chemical harms the child’s neuron development, causing a disturbance in the brain activities hence blocking the development of the brain (Raymond, 2011).

This epidemiological study has provided crucial information on the heritability of ASD. However, they have failed to inform us on the specific genes involved or on the number and the frequencies of the variants. Comparatively Pinto et al. (2014) performed a genetic study that associated genetic variants with autism as they used observations from their cytogenetic studies to detect genomic imbalances and reduce the prevalence of large chromosomal abnormalities. It was estimated that likely

gene disrupting mutations (present on both paternal and maternal chromosomes) could account for 3% of the patients with ASD. For the X chromosome, there was a significant 1. 5-fold increase in complete loss of function mutations in affected males compared to unaffected males, who could account for 2% of males with ASD.

However, this present article stood out for many reasons. This study confirmed previous findings on the genetic influences in the determination of autism. Because the latter study included several crucial methodological improvements over the original research (Folstein & Rutter, 1977a, b). This study had four significant design features: Firstly, clinics and special schools in the country were contacted, the twin registers were examined and there was a population screening for the cases. Secondly, systematic and standardized methods of diagnosis were employed such as parental interviews (Le Couteur et al., 1989) and diagnostic observations on the child were made on all behaviours that were considered central to autism in ICD-10, the twin pairs were also assessed using the Autism Diagnostic Observational Schedule (Lord et al., 1989). Thirdly, a systematic screening for medical conditions and chromosomal abnormalities was also employed, to mainly focus on the genetic influences in idiopathic autism. Fourth, blood groups were used to test for zygosity. This was the most important feature as parents and professionals thought the twins were fraternal based on their behavioral differences associated with autism, whereas in fact they were MZ.

The researchers in the present study did not include all participants from the original study in order to control for other variables that may predominantly

determine the causes of autism. However, the researchers have justified why the two probands from the original study were not included in their analysis and hey have also defined the scientific terms used in the justification. Bailey et al. (1995), state that the two probands did not meet the ICD-10 criteria for autism upon assessment. One discordant DZ pair was diagnosed with atypical autism: this is a disorder that is grouped within the pervasive developmental disorder (PDD) (Le Couteur et al. 1995, though it does not meet the ICD-10 criteria for autism. The other MZ pair was discordant for autism although they were concordant for cognitive and social impairments.

The findings from this study are concordant for autism, which suggest a consequence of genetic influences. The findings also follow the aim of this study logically and statistically. Since there are substantial differences between the concordance rates for autism in MZ and DZ twins, the increase in risk for the MZ twins for 10 times that of the DZ twins. The findings of this study have also been replicated in another study by Pickles et al. (1995), the findings in his study also effectively ruled out the possibility of just one gene to influence autism, he suggested that the three or four genes were the most probable, although Dykens and Volkmar (1997) hypothesized that a possibility between 2 and 10 genes was much more probable, this was backed by the falloff rate from the MZ and DZ twins, they also measured the genetic makeup of first-degree and second- degree relatives.

The findings from the Scandinavian study (Steffenburg et al. 1989) suggested that the genetic liability was usually manifest as autism; whereas variable expression was observed among the MZ cotwins in the British study

(Folstein & Rutter 1977 a, b). The findings from this study confirm that phenotypic expression is not confined to autism. At follow-up the MZ cotwins from the original sample had considerable social difficulties, and both cognitive and social difficulties were identified in the new non-autistic cotwins. The finding that over 90% of MZ pairs were concordant for a broader phenotype of social and/or cognitive difficulties supports the conclusion of substantial genetic influences and suggests that these apply to most cases.

In conclusion, the present study illustrates their main aim to prove that autism is a strongly genetic disorder. The authors further develop previous twin studies on genetics and autism, which contributes to the field with more solid findings for the causes of autism. The literature that has been reviewed in the research study has been very relevant and comprehensive. The design of the study was consistent with its aims. Personally, I found the article quite interesting as it has extended my knowledge on the causes of autism. This is the first epidemiological study of autistic individuals to report an association with increased head circumference.