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Genetic and environmental effects on stuttering: A twin study from Finland

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ABSTRACT

The present study explored the prevalence of self-reported stuttering in a Finnish twin population and examined the extent to which the variance in liability to stuttering was attributable to genetic and environmental effects. We analyzed data of 1728 Finnish twins, born between 1961 and 1989. The participants were asked to complete a questionnaire on speech, language, and voice. In two of the questions they were asked to report the occurrence of childhood and present stuttering of their own and that of their sibling. According to the results, 2.3% (52) of the participants were reported to have stuttered as children and 28.8% of them (15) were reported to continue to stutter in adulthood. There was no significant gender difference in the prevalence of stuttering in either childhood or adulthood. For childhood stuttering, the tetrachoric correlation was higher for monozygotic pairs ($r = .74$) than for dizygotic pairs ($r = .27$). By means of structural equation modeling it was found that 82% of the variance in liability to childhood stuttering was attributable to additive genetic effects, with the remaining 18% due to non-shared environmental effects. In conclusion, the results of the present study confirm findings from prior studies and support a strong genetic and only a moderate non-shared environmental effect on stuttering. Potential small differences in the prevalence of stuttering in different populations are suggested by our data.

Educational objectives: The reader will be able to recognize the contribution of genetic and environmental effects on stuttering.

1. Incidence and prevalence of stuttering

According to a review by Guitar (2006), the incidence of stuttering is about 5%. However, there is some variation in the incidence of stuttering in different studies depending on the population and the method used. The incidence of stuttering in three year old children, investigated by speech screening, was 5.2% in Månsson's (2000) study, while the overall incidence of stuttering was 2.2% in a population study carried out by telephone interviews by Craig, Hancock, Tran, Craig, and Peters (2002).

In twin studies, the reported incidence and prevalence rates vary to some extent. When the population was of adults and self-reports were used, the incidence was 3.2% for males and 1.2% for females in a study by Andrews, Morris-Yates, Howie, and Martin (1991), 8.8% in a study by Felsenfeld et al. (2000) and 5.6–5.8% in younger groups and 4.5% in the oldest group in the study by Fagnani, Fibiger, Skytthe, and Hjelmborg (2010).

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When parental reports for childhood stuttering have been used in twin studies, prevalence rates of 7% (Dworzynski, Remington, Rijsdijk, Howell, & Plomin, 2007), 6.7% for males and 3.6% for females (Ooki, 2005) and 4% (van Beijsterveldt, Felsenfeld, and Boomsma, 2010) have been reported. According to Guitar (2006) the overall prevalence of stuttering in kindergarten age is about 2.4%.

1.1. The etiology of stuttering

Historically there have been a variety of etiological explanations of stuttering. In spite of various approaches, including physiological, behavioral and psychological aspects, no single theory sufficiently explains the origin of stuttering. Several theories have focused on physiological factors (see e.g., Guitar, 2006). Because various brain areas are involved in the planning and production of speech, there is naturally a risk of disruptions in these processes. According to a review by Yairi (2007), deficient coordination between hemispheres due to deviant brain dominance might be a contributory cause of stuttering. Disturbances in auditory feedback have also been suggested as a possible cause. For example, Foundas et al. (2004) have reported an association between planum temporale asymmetry and auditory feedback disturbance.

There is also strong evidence to support the notion that disturbances in motor control could cause stuttering; that is, stuttering might be a result of deficiencies in neural connections controlling speech (Kent, 2000). When Fox et al. (1996) examined individuals who stutter using positron emission tomography they found evidence that stuttering could be a disorder where multiple neural systems involved in the speech production can be affected. In a review, Alm (2004) concluded that stuttering might be the result of an impaired ability of the basal ganglia to give motor timing cues which initiate speech. There is also evidence as how a brain injury in the basal ganglia can cause stuttering for adults (Tani & Sakai, 2011). According to the review by Smits-Bandstra and De Nil (2007) dysfunction in cortico-striato-thalamo-cortical connections can cause difficulties in sequence learning and automatization and be one of the etiological components in the development of stuttering.

Possible explanations of the physiological and psychological factors behind the stuttering may include both environmental and genetic effects (Alm, 2004; Guitar, 2006). Environmental factors, such as growing up in the same family, are considered to be shared environmental factors. On the other hand, factors that influence people on an individual level are considered to be non-shared environmental factors. They can be, for example, illness, injury, or harmful events during prenatal or perinatal development. Premature birth is known as one perinatal risk factor for stuttering (Ajdacic-Gross et al., 2010; Stromswold, 2006).

1.2. The role of genetics

According to a review by Yairi, Ambrose, and Cox (1996) stuttering is far more frequent among relatives of stuttering probands than in the general population. In the reviewed studies the percentages of stutterers with a positive family history of stuttering has ranged from 20% to 74%. In the non-stuttering control groups, the corresponding percentages have ranged from 1.3% to 42%. The variation in the results is likely to depend on differences in definitions of stuttering, the effect of spontaneous recovery, and the imbalanced gender ratio (Yairi et al., 1996).

One way to study genetic predisposition of speech disorders is a twin study. In contrast to family studies, twin studies can help in determining whether familial aggregation is genetic or environmental in nature. Monozygotic (MZ) twins have the same gene collection while dizygotic (DZ) twins share about 50% of their segregating genes (Carey, 2003). Twins often grow up in a shared environment and by making the equal environment assumption (EEA), that is, that the environmental influences are shared to the same extent by MZ and DZ twin pairs, differences between the two twin groups are attributable to genetic variance (Rutter, 2006).

In many twin studies of stuttering the concordancerate – an estimate of the probability that both twins are affected by stuttering if it is known that one of the twins is affected- has been reported to be higher in MZ than in DZ twins (Andrews et al., 1991; Dworzynski et al., 2007; Felsenfeld et al., 2000; Howie, 1981). If significantly more MZ twins than DZ twins are concordant for a certain disorder, it is probable that the difference depends on genetic factors (Rutter, 2006).

In studies from different parts of the world, the results of genetic model fitting analyses of stuttering vary slightly (Table 1). Approximately 70–85% of the variance in liability to stuttering has been found to be attributable to genetic effects, with the remainder due to non-shared environmental effects. The recent results of van Beijsterveldt et al. (2010) indicate that shared environmental factors may be significant for stuttering-like behaviors and high levels of normal nonfluency in young children.

Recent studies in molecular genetics have focused on finding susceptibility genes that might contribute to stuttering. The results of these studies differ from each other, suggesting that stuttering is likely to be a polygenic disorder for which several genes may increase susceptibility (Kang et al., 2010; Lewis, Ricci, Lukong, & Drayna, 2004; Raza, Riazuddin, & Drayna, 2010; Riaz et al., 2005; Shugart et al., 2004). The results of the study of Suresh et al. (2006) and Wittke-Thompson et al. (2007) support linkage on multiple loci throughout the genome. The results of a recent study by Kang et al. (2010) found that a mutation in gene GNPTAB, GNPTG and NAGPA occurred more often in stuttering probands relative to non-stuttering controls. These structures are associated with, for example, emotion and motor function. Expression of these three genes in the human brain is not known and it has to be examined further, but there are data that they have high levels of expression in the hippocampus, hippocampal formation, and cerebellum in mice (Kang et al., 2010). As support to the multiple loci