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Identification of traits associated with stuttering

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Abstract

Stuttering has been considered a heritable disorder since the 1930s. There have been different models of transmission that have been proposed most involving a polygenic component with or without a major locus. In spite of these models, the characteristics being transmitted are not known. This study used two different tasks—a tapping task that is thought to probe hemispheric differences and a Stroop task, which appears to create interferences in speech motor programming and/or execution. The 48 participants in this study included individuals who stutter, high risk family members and controls for each group. Results indicated that for tapping at a comfortable rate, the experimental groups were significantly different from their control groups and for tapping at a fast rate, the stuttering and high risk groups were different from each other. The results of the Stroop test were not statistically significant.

Learning outcomes: Readers will learn about: (1) genetic aspects of stuttering; (2) hemispheric dominance in stuttering and high risk subjects; (3) understanding traits that may be associated with stuttering.

1. Introduction

Studies related to the familial nature of stuttering have been conducted since the 1930s (Berry, 1938; Bryngelson, 1935, 1939). There have been many reports indicating that stuttering clusters in families (see an early review by Sheehan & Costley, 1977).

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All studies, past and contemporary, suggest that individuals who stutter are more likely to have family members who stutter when compared with non-stuttering individuals. In a more recent review of the literature, Yairi, Ambrose, and Cox (1996) examined at least 26 studies related to familial history in stuttering. They too concluded that in spite of different findings in terms of the percentages, familial incidence of stuttering is consistently greater for individuals who exhibit stuttering than for the general population. Specifically, familial incidence of stuttering in very young children who stutter was found to be as high as 65%. In addition, twin studies examined by these authors indicated a high stuttering concordance rate of 63–83% among monozygotic twins.

Andrews and Harris (1964) were among the first to put forth a genetic model that explains the transmission of stuttering within families. They postulated either a purely polygenic model or a polygenic model with a possible component of major locus or loci. Using segregation analysis Kidd (1980), and later Cox, Kramer, and Kidd (1984), offered a polygenic multifactorial transmission model wherein genetic susceptibility is believed to be transmitted through the cumulative contribution of multiple unspecified genes, with some role assigned to multiple environmental factors. Later, Ambrose, Yairi, and Cox (1993) also used segregation analysis and were the first to report statistically significant evidence for both single major locus and polygenic components for stuttering. Recently, Viswanath, Lee, and Chakraborty (2004) conducted a segregation analysis on 56 pedigrees of adults with persistent developmental stuttering. Using a regression model, they found evidence for an autosomal dominant major gene effect. Two co-variates – sex and affection status of parents – influenced the major gene.

Although these findings and the emerging models have strengthened the conclusion that there is a strong genetic component to the familiality of stuttering, what exactly is being transmitted is not known. It could include abnormalities in biochemical pathways, structural differences, brain processing for speech and language, motor skills, and/or others (Cox, 1993; Yairi, 1998). Quite likely, the postulated major gene(s) and other genetic components transmit a number of traits, some of which contribute to the expression of stuttering. This constellation of genes may vary from one family to another. Furthermore, a particular characteristic that increases the susceptibility for stuttering may not, by itself, cause the stuttering. But, when it co-occurs with certain other characteristics, stuttering may be expressed. As stated, however, the nature of these genes and their expression is not currently known.

Some of the traits carried by genes underlying susceptibility to stuttering might also be expressed in non-stuttering family members. Susceptibility to stuttering is frequently transmitted by a non-stuttering parent having a family history of stuttering. Because the parents, as well as non-stuttering siblings, share some of the genes with the stuttering child, they may also share some of the fluency-related traits possessed by their child who stutters.

Many studies that have as their focus the identification of traits that are related to the primary liability to a complex disorder. For example, offspring in families in which both parents have type 2 diabetes have been studied longitudinally to identify traits that precede (and may predict) the onset of the disease in these individual who are clearly at high risk of developing diabetes (Warram, Sigal, Martin, Krolewski, & Soeldner, 1996). In general,