Genetic susceptibility to stuttering

Essay
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By:
Rania Rafik Girgis
M.B.B.Ch.

Supervised by:
Prof. Dr. ***Mohamed Aly Saad Barakah***
Prof. of phoniatrics
Faculty of medicine
Ain Shams university

Dr. ***Hassan Hosni M.Ghandour***
Assistant Prof. of phoniatrics
Faculty of medicine
Ain Shams university

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Rania Rafik Girgis

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Introduction

Stuttering, also known as stammering, is a common speech disorder that has been recognized since antiquity and affects all populations and language groups ***(Bloodstein and Ratner, 2008).***

It is defined by the **World Health Organization (1977)** as a disorder in the rhythm of speech in which the individual knows precisely what he wishes to say, but at the time is unable to say it because of an involuntary repetition, prolongation or cessation of a sound ***(Johnson, 1995).***

The abnormal speech behaviors displayed by stutterers can be divided into blocks and prolongations/repetitions. Vocal blocks are the most disruptive to speed

because the stutterer is unable to talk at the time of the vocal block. Stutterers also often display abnormal movements of the lips, mouth and facial musculature including the eyes ***(Ludlow and Loucks, 2003)***.

Stuttering appears in all cultures and has been a problem for human kind for at least 40 centuries. It has been a controversial topic for decades concerning its definitions, different methods of assessment and treatment. Stuttering is a disorder of the neuromotor control of speech influenced by interactive process of language production and intensified by learning process ***(Peters and Guitar, 1991).***

Although the underlying causes of stuttering are unknown, results of twin studies, adoption studies, and family studies support a role for genetic contributions in the etiology of

this disorder. Genetic-linkage studies have provided suggestive or significant evidence of linkage with numerous loci across the genome ***(Bloodstein and Ratner, 2008).***

Susceptibility to nonsyndromic stuttering is associated with variations in genes governing lysosomal metabolism ***(Olson et al., 2010).***

Disorders that disrupt the development of speech, language, or reading have substantial effects on social function. Researchers have implicated specific genetic variants in common language impairments, and dyslexia ***(Fisher and Scharff, 2009).***

The incidence of stuttering is reported to be 1% worldwide with a greater incidence of males to females (3:1). Theories of causation

are briefly identified including a genetic predisposition and neurological factors. In the process of acquiring speech skills, preschool children normally pass through a transitional stage of speech dysfluency. This fact may impede recognition of early stuttering behavior, which can be emotionally painful for the child and may interfere with psychological development ***(Dowling, 1994).***

As regards family history and its role in stuttering, 30% of stutterers had relatives who stuttered ***(Andrews and Harris, 1964).***

For explanation of both genetic transmission of stuttering and the fact that evidence of genetic transmission is lacking in some cases, it could be said that genetic transmission of stuttering in many cases may be through two factors: anomalous neural

organization for speech and sensitive temperament ***(Guitar, 1998).***

There is a growing opinion among speech pathologists that most stuttering is a genetically inherited neurologic disorder ***(Kidd et al., 1986).*** It is now well established that stuttering is a heritable disorder ***(Watkins et al., 2002).***

Aim of the work

The aim of this work is to review the literature of genetics and heritability as suspected factors in the etiology of stuttering in order to be taken in consideration while investigating and treating a stutterer.

Stuttering

Speech is not a simple movement; it is the result of a large number of complex processes on various levels. These levels of processing include; level of conceptualization, level of formulation and level of articulation. Level of conceptualization is where the concepts one likes to express are specified and transformed into a preverbal message; then follows the level of formulation, during which preverbal message is transformed into verbal or linguistic form and this level includes process of grammatical encoding. Finally, the level of articulation or speech production complies preparation of muscle command and motor command execution leading to speech. In stuttering, difficulties of execution of the final product are what we hear ***(Postma and Kolk, 1990).***

There are many variables that determine fluency reflecting the temporal aspects of speech production; such variables as pauses, rhythm, intonation, stresses and rate of speech are controlled by when and how fast we move our speech structures. So, our temporal control of movements of these structures determines our fluency ***(Starkweather, 1997).***

Fluency is the production of a more or less continuous speech at a relatively rapid rate with optimum effort ***(Yairi and Ambrose, 1992).*** So; the speech of normal subject is perceived as continuous and fluent because pauses are linguistically appropriate and serving an important communicative function without any apparent muscular tension in the body or voice ***(Nicolosi et al., 1996).***

Certain variables suggested to be useful

in distinguishing between fluent and dysfluent speech ***(Dalton and Hardcastle, 1977):-***

1. *Presence of extra sounds*: Such as repetitions, prolongations, interjections and revisions. If a speaker says, " I-I-I nnnnneed to have uh my uh, well, I-I-I should get mmmmmy car fixed", he sounds dysfluent.
2. *Location and frequency of pauses:* If a speaker says, "Whenever I remember to bring my umbrella (pause), it never rains", he sounds fluent. But if he says, "Whenever (pause) I remember to bring (pause) my (pause) umbrella, it never (pause) rains", he sounds dysfluent.
3. *Rhythmical patterning in speech*: English is typically spoken with stressed syllables at relatively equal intervals; in general, stressed syllables are followed by several unstressed syllables. When this pattern is

deviated from markedly, as in cerebellar disease, when the speaker stresses all syllables equal, the speaker sounds dysfluent.

4. *Intonation and stress*: If a speaker does not vary intonation and stress and is therefore monotonous, he may be considered dysfluent. Abnormal intonation and stress patterns may also be considered dysfluent.
5. *Overall rate*: If a speaker has a very slow rate of speech or if he has bursts of fast rate interspersed with slower rate, he may be considered dysfluent.

Definition of fluency disorders:

Fluency disorders are defined as a deviation in continuity, smoothness, rhythm and/or effort with which phonology, lexical, morphologic and / or syntactic language are spoken ***(ASHA, 1999).***

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Curlee (1993) defined stuttering as an impairment, disability and handicap. Impairment is any loss or abnormality of psychological, physiological or anatomic structure or function; so, in stuttering it is present in the form of neuropsychological and neurophysiological events that immediately precede and accompany the audible and visible events of stuttering. Disability is defined as

restriction or lack of ability to perform an activity in the normal range; in stuttering, disability manifests as audible and visible events which are behavioral manifestations of stuttering. Handicap is defined as disadvantage for a given individual resulting from an impairment or disability preventing the fulfillment of a normal role; in stuttering, handicap presents in the form of disadvantages resulting from reaction to audible and visible events of stuttering.

American Speech-Language Hearing Association defined stuttering as a speech event that contains intraphonemic disruption, part-word repetitions, monosyllabic whole word repetitions, prolongations and silent fixations (blocks). This may be or may be not accompanied by secondary behaviors used to escape and / or avoid these speech situations

(ASHA, 1999).

The onset of stuttering may occur at any time during childhood between the beginning of two-word utterances (around 18 months) and puberty (11 or 12 years). It is most likely to occur between ages 2 and 5 years ***(Andrews et al., 1983 and Craig et al., 2003).***

Epidemiology of stuttering:

Andrews et al. (1983) found that the incidence of stuttering appears to be about 5%. The incidence of stuttering ranges from 2.1% in adults (21-50 years) to 2.8% in younger children (2-5 years) and 2.4% in older children (6-10 years) ***(Craig et al., 2003).***

Bloodstein (1995) found that the prevalence of stuttering appears to be 1%. The prevalence of stuttering over whole population