

Role of Genes and the Environment on Stuttering

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Abstract

This paper presents the role genes and the environment on stuttering. It covers an overview of stuttering, epidemiology, causes, genetic and environmental relation, treatment, and future research on treatment. The paper draws information from existing literature and previous research studies published on the pubmed website. Across the studies, stuttering is defined as a speech or communication disorder that involves disfluencies in words. The disruption in language can be partial or whole repetition of words. The severity of stuttering varies between people. From the studies reviewed, the stuttering among adults and children is about 2% and 5% respectively. The studies link stuttering to either genetics or environment, however the real cause of stuttering remains uncertain. On the genetics as a cause, most studies suggest that people that are related to a family member, who stutters, are more likely to stutter than people from a family where no members stutter. Further, most studies found that boys are more likely to stutter than men. On the environment causes, studies show that social anxiety is related to stuttering. Further, developmental stages of a child and experiences might aggravate stuttering. The common treatment for stuttering include therapist drugs and practices by the Speech-Language Pathologists. However, there is need to research further on the effectiveness of these methods to treat stuttering.

Introduction

Stuttering is a speech disorder that makes it difficult for those affected to communicate comfortably. It is associated with difficulty in pronouncing sounds of initial consonants of a word (Alm, 2014). Those who stutter feel uncomfortable having a smooth conversation with people and in most cases, they just keep things to themselves. According to Trichon & Tetnowski (2011), these are people who can make meaningful contribution to a conversation but end up keeping quiet. If the people associating with those who stutter do not understand the condition and how to deal with the affected person, the stuttering person becomes emotionally affected (Arenas, Zebrowski & Moon, 2012). The person might react angrily to the surrounding and feel that people do not want to listen. While some studies link stuttering to genetic variation, others associate stuttering with the environment from which a person grows.

The real cause of stuttering is still uncertain among scientists, but they agree that genetics and the environment play a significant role on stuttering. Most of the studies attribute stuttering to genetics. Beilby, Byrnes & Young (2012) found that a person from a lineage where a family member stutters is likely to stutter in some point in life. These findings are in line with Daniels, Gabel & Hughes (2012) who showed that there is a link between genetics and stuttering. The interesting thing about this studies is that there is no relationship between the severity of stuttering and being related to a family member who stutters. Numerous studies acknowledge that males are carriers of stuttering genes. Researchers show that the environment plays a significant role in stuttering (Eggers & Leahy, 2011). Therefore, the environment from which a person is born affects language and speech. Social interaction, especially in school plays a significant role in speech development. According to Bauerly & Nil (2011), knowledge develops

from social interaction and knowledge is a sum of social and cognitive process. This means that experiences of a person influence a child's cognitive development which affects speech.

Treatment of stuttering depends on the severity of the problem (Gerlach & Subramanian, 2016). It is therefore clear that diagnosing the cause of stuttering leads to the most effective method of treating stuttering. Treatment for people who stutter revolves around skills, strategies, and behaviors. These strategies aim at enhancing oral communication of the person affected (Zraick, Atcherson, & Brown, 2012). While these methods can work independently, in case of severe stuttering, a combination of two or more methods can be very effective. These methods; controlling speech rate and breathing aim at shaping fluency in speech. Knowing the role of genes and the environment on stuttering helps in designing new methods of treatment for problem (Hinckley, 2016). The purpose of this review paper therefore, is to present the role of genes and the environment on stuttering, and the treatment available for the problem.

Epidemiology

Studies link stuttering to childhood whereby about 5% of children are likely to be affected. In most cases, these children avoid speaking, which makes learning difficult (Holland, 2016). These children are often silent because they fear being laughed at by their peers. They often have speech associated anxiety and they will be seen trembling and out of breath during a conversation. In the worst scenarios, opts for social withdrawal becomes of the psychological effects associated with stuttering (Bauerly& Nil, 2011). Stuttering incidence rates range from 0.7% to 15.4%. However, the morbid risk, which is the average rate of stuttering among the literature reviewed is 5% (Beilby, et al., 2012). Most studies show that stuttering occur in children before the age of 10 years. Afterwards, the disorder disappears or can persist during the child's adolescent, adulthood and old age. For example, King George VI of England had the

stuttering problem and needed a speech therapist to train him and practice some words before giving a public talk (Lasalle & Wolk, 2011).

There are variations in phenotypic severity and symptom expression for those affected by stuttering. Most of the studies show mild severity of the stuttering population. Children can either recover from stuttering either partially or completely (Benditt, Hoit & Britton, 2016). Irani, Gabel, Daniels & Hughes (2012) estimated that recovery rate range from 36 to 89% and that boys have higher chances of stuttering than girls and take longer time to recover. Further, Logan & Willis (2011) show that men stuttered more than females and the distribution might be attributed to either the environment or genetic variation. The approximate gender ratio for stuttering among young children is 2:1 and among adults who continue stuttering is 4 or 5:1 (Coalson & Byrd, 2016). The distribution shows that females have the higher probability than men to recover from stuttering.

Causes

The etiology of stuttering remains uncertain despite centuries of research. While some relate the causes to environment, some studies related to genetics as cited from behavioral genetic studies (Louis, 2011). Lowe, Menzies, Packman, O'Brian & Onslow (2015) found that there are high chances for a child to be born stuttering if a family member stutters or a person in the lineage had the disorder. In fact, numerous studies attribute genetic variation to the condition of stuttering. For example, a study among three families revealed that about 15% of people whose direct relatives of stuttering reported a history of the condition (Quesal & Manning, 2016). In comparison, the researchers carried out a control experiment among the families that did not stuttering in their lineage. They found that 1 to 3% of the people reported stuttering in

those families (Coalson & Byrd, 2016). It is sufficient therefore, to state that stuttering is hereditary, that is, it caused by some genes that are passed on in the family.

There are linkages between stuttering and environment. This is because some people who are predisposed to stutter do not develop the condition in their adulthood. There are life events in a person that makes it difficult for a child to overcome the disorder and communicate fluently (Raza, Mattera, Morell, Sainz, Rahn, Gutierrez, Drayna, 2015). For instance, the development of grammar skills can trigger stuttering in child. Özdemir & Topbaş (2011) show that children learn many grammar skills in school between the ages of two and five years. The rules for mastering grammar allows children to adopt longer sentences and leave their short immature messages learnt from their mothers. Further, sentences that consist one or two words may not be difficult for a child who is predisposed to stutter (Sassi, Matas, Mendonça & Andrade, 2011). However, Tompkins (2016) found that a child becomes discouraged to produce longer sentences that are mostly complex. Such a child does not practice enough to overcome stuttering. They say that practice makes perfect. In this case, the child will not be practicing thus may end up living with the disorder.

The factors that can propel stuttering in the child include emotions. Children get frustrated when they think of the bad things that they go through or directly contact with a frustrating subject (Trichon & Tetnowski, 2011). For example, domestic violence can cause a child to develop emotional problems. These emotional factors, which are environmental, escalates stuttering problem even if the child could have outgrown it. A child who is emotionally disturbed is likely to tense the teeth muscles when speaking and in most cases, when stuttering occurs during a conversation (Daniels et al, 2012). The tension increases the condition and the child gets even more frustrated. How the audience listens respond to stuttering can also affect a

child's learning efforts. If the audiences respond with teases and jeers, the child might develop fear of public and hate making a speech which aggravates the level of stuttering. On the other hand, people when the public reacts with appreciation, the child becomes motivated to learn, which enhances the chances to outgrow stuttering (Vincent, Grela & Gilbert, 2012).. It is therefore important for the public, through teachers and parents, to appreciate the child's effort to learn fluency in speech.

Genetic Relation.

There is a relationship between stuttering and genetics in that a family whose first degree relative had the stuttering is more likely to inherit it. Genetics as a cause of stuttering is the most advocated cause of stuttering. Numerous studies have outlined its effects which include a continued lineage that carries stuttering genes. At this point, it is important to note that it is not the fault of those who stutter nor their parents or the ancestors. It is a condition and can hit everyone. The effect of stuttering from genetics is inherent in the way it is passed on. In a study conducted from the Australian Twin Registry shows that one or both members were affected by stuttering. In the study, 70% of the variance attributed stuttering. In this case, the twins inherited stuttering from the parents (Vincent et al, 2012). However, Zraick et al (2011) found that there is variance between male and females who stutters. It seems the genes for stuttering follows males more than females. In the study, the ration of male to females among stuttering population was 3.3: 1. This means that males are three times more likely to stutter than females. Another relationship between stuttering and genetics is that people who stutter tend to have an emotionally sensitive temperament (Özdemir et al, 2011).

However, studies do not distinguish which comes first; stuttering or emotions. It is true that emotions can result from the inability to express a point fluently without stuttering. On the

other hand, a child can inherit a temperamental attitude from the parents which in effect causes stuttering (Alm, 2014). Focusing on genetic effect, stuttering can arise from inherited temperamental attitude, which leads to anxiety. There is an association between stuttering and social anxiety disorder, which is also genetic (Arenas et al, 2012). Therefore, a person who has a social anxiety disorder is more likely to develop stuttering. On the other hand, stuttering can also cause social anxiety. Bauerl & Nil (2011) stated social anxiety involves looking at things from another people's perspective. If images of rejection cross a person's mind, then the person can have social withdrawal symptoms. Adults who stutter are therefore likely to experience annoying encounters in their lives (Benditt, Hoit & Britton, 2016).

Raza, Mattera, Morell, Sainz, Rahn, Gutierrez & Drayna (2015) shows that there is an association between intracellular trafficking and stuttering. Where there are deficits in intracellular trafficking, the person persists in stuttering. The authors define intracellular trafficking as an exocytic pathway that links the endoplasmic reticulum, Golgi apparatus and the plasma membrane. Raza et al. (2015) found that the exocytic pathway might be deficient which causes stuttering. The study conducted among the Cameroon, Pakistan and North America found a significantly higher rate of rare variants in AP4E1 in these individuals. The use of twin and adopted studies clearly show the relationship between stuttering and genetics. Further, more than 50% of the cases reported attribute genetics to stuttering. The approximate ratio in most studies between male and females is 1.5:1 (Daniels et al, 2012). This means that families who transmit genes for stuttering for every 1.5 males reported, there is 1 female who stutters. However, other studies found a higher ratio between males and females, 7 or 8:1 who stutter from families that have no history of stuttering (Quesal & Manning, 2016). This means that from a family that has no history of stuttering, there are 7 or 8 males for every 1 female. This study

confirms the statement that males are more at risk of having the genes for stuttering than females.

Environmental Relation

Gerlach & Subramanian (2016) show that there is a relationship between stuttering and the environment. The environment has an effect of either initiating stuttering or aggravating it. Most of the people avoid speaking, are affected by anxiety and are more likely to withdraw from the society (Andy & Bhatnagar, 2012). Further, the environment affects the severity of stuttering among children. The first person that teaches a child how to speak is the mother whereby the child starts pronouncing one word at a time (Hinckley, 2016). In school the child is required to learn complex sentences and grammar rules. The complexity of the words can have an effect of a child. Where the children have difficulty in learning complex sentences and grammar rules, they might decide to remain silent and avoid speaking (Beaver, 2010). Moreover, Cox & Kidd (2003) state that avoidance of speech have a profound effect on stuttering because the child will not be practicing. The teacher therefore should encourage children to speak and appreciate their effort.

Stuttering causes social withdrawal both in children and adults. A stuttering person, who receives disapproval from the public may retreat and be an introvert. Fagnani, Fibiger, Skytthe & Hjelmborg (2011) show, males are at a higher risk of stuttering than females. The genetic variation between male and females confirms the notion. Women tend to be more emotional than men (Xu, Zhou, Zhang, Espinosa-Artiles, Wang, Zhang, Molnar, 2014). However, some studies cite that the emotional state between men and women is just the same and that men tend to keep emotions inside while women tend to express them (Holland, 2016). Consequently, keeping emotions inside affects the speech process which results to stuttering. On the other hand, women

will cry, even in public, to express their emotions. Crying makes them get over the problem and focus on other things thus reducing the risk of stuttering.

There is a relationship between stuttering and social anxiety. In the article, “Observer perspective imagery with stuttering”, Lowe, Menzies, Packman, O’Brian, & Onslow (2015) state that social anxiety involves viewing images as if looking at the self from the perspective of another. In contrast, the field perspective involves looking out from the self at the surrounding environment. When the images are those of disapproval, a person becomes uncomfortable and experiences social anxiety (Lebrun, 2007). According to Lasalle & Wolk (2011), stuttering group tends to view images of themselves from an observer’s perspective. Conversely, non-stuttering group tended to view images from the field perspective. In both groups, the images are associated with distorted and negative content. The interesting observation is that the stuttering group was more likely to remember the images that were distorted and negative than the non-stuttering group (Özdemir, Louis & Topbaş, 2011). The results indicate that stuttering group are affected by anxiety disorder.

The environment therefore, has a profound effect on stuttering people from childhood. Negative experiences that people go through in life are more likely to increase the severity of stuttering (Leon-Sarmiento, Paez & Hallett, 2013). For instance, negative emotions that arise from frustrations can be too strong for a person, which then appears in form of anxiety and speech problem. Nonetheless, expectations from the society also aggravates the problem. Where the family is so much concerned about the stuttering problem of a child is more likely to increase the problem because children feel that the attention is all on them (Logan & Willis, 2011).

Further, the concern makes a child to develop fears. Therefore, it is worth noting that the environment propels an already underlying problem that is genetic.

Treatment

First. Controlling and monitoring of speech rate; this is where the Speech Language Pathologist (SLP), helps the affected person to practice talking fluently using short sentences at a very slow rate. The therapists teaches the stutter to control words by stretching vowels (Lalor, 2008). Further, Mcallister, Collier & Shepstone (2012) state that the person is taught to maintain a continuous airflow while talking which might take time depending on the person. As the person continues to practice, gradually becomes able to talk smoothly at high speed. Further practice makes the person to use longer sentences at a higher speed (Trichon & Tetnowski, 2011). Regular practice with the therapist increases the success rate. Contrary, the stutter may take a significantly longer time. Second. Breathing control; regulating the breathing process also treats stuttering in the long term. In this method, the patient is conditioned to respond to the breathing mechanism (Moncur, 1951). The stutter learns to control breath, phonation and articulation. Articulation involves controlling the lips, jaw and tongue. Finally, stuttering modification; this involves a therapy that modifies stuttering rather than the previous methods that aim at eliminating it. The principle of stuttering modification therapy aims at balancing stuttering and anxiety. In this case, if anxiety causes stuttering, alleviating stuttering will require the use of easier stuttering accompanied by less avoidance and fear (Quesal & Manning, 2016).

There are four stages of Block Modification Therapy developed by Charles Van Riper in 1973. First. Identification. At this stage the stutter and the therapist work together to identify the cause of stuttering. For instance, this involves core behaviors, attitudes and feelings that arise

during stuttering (Roth & Bibeau, 2011). The therapist and the patient then determine whether the feelings are negative or positive. Second, desensitization; this is whereby the patient tries to freeze stuttering behavior to reduce fear and anxiety. The patient confronts sound difficulties instead of avoiding them. At this point, the patient stutters intentionally to remove fear and anxiety (Sassi, Matas, Mendonça & Andrade, 2011). When the patient stutters intentionally, it means he or she has accepted the condition and is ready to change. The third stage is modification. This is where the patient now learns easy stuttering. Sassi et al (2011) explain that the stutter learns to cancel disfluency in words by pausing and saying the word again. Further, the patient detects words that are difficult to say and uses words that are considered easy for stuttering. The final stage is stabilization. At this stage, the patient stabilizes in the process of controlling stuttering. This involves preparation of assignments, making preparatory sets and automatic pull-outs (Smits-Bandstra, Nil & Saint-Cyr, 2006). The person changes from stuttering severing to occasional mild stuttering.

Future Research on Treatment.

Future research on treatment should take the course of technology and its effectiveness. This includes use of drug therapy and electronic devices. So far, no drug has been approved by the U.S Food and Drug Administration (FDA) (Speller et al, 2013). However, since epilepsy, anxiety and depression have a relationship with stuttering, drugs that are used to treat these conditions are used to treat stuttering (Eggers & Leahy, 2011). Therefore, there is need for FDA to conduct further research on the drugs recommended by physicians before approving. Further, there should be further research on electronic devices available for treatment. There has been a remarkable breakthrough in this field, for example, there are electronic devices that are fitted into

the ear canal of a person, which acts as a hearing aid (Abdalla, St. Louis, Schuele & Kelly, 2014). The device replays the message so that the wearer gets the stutter effectively. However, there should be future research on the effectiveness of these devices. This is because according to Andy & Bhatnagar (2012) a stutter can only use the device in a formal setting like in a conference. Future research should therefore aim at addressing the problem in an informal setting.

Conclusion

Studying stuttering is very important because it helps to know the effects from genetics and from the environment. Knowing these effects are crucial for the purpose of diagnosing treatment. The incidence of the disorder is about 2% among males and 5% among females. Further, studies show that males are at a high risk to develop stuttering females. The real cause of the difference in the variation is not known, but most studies link the study to the difference in the emotional well-being of males and females. So far, the real cause of stuttering is known, but studies link the disorder to genetic and environment. On genetics, stuttering is passed on from one generation to the next. However, studies show that there are higher chances for males to develop stuttering than females. The treatment of stuttering involves the assistance of Speech – Language Pathologist (SLP). However, there is need for future research on the disorder to determine the effectiveness of electronic devices used by SLPs.

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