Exploring Speech Disfluencies: An Investigation into Atypical and Typical Patterns, Genetic and Neurobiological Factors, and Psychological Responses

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Abstract:

With stuttering's vast spectrum of different factors and forms, this review paper focuses on the impact of genetic, neurobiological, and psychological factors on this phenomenon, with varying brain activity among stutterers. With respect to the types of disfluencies, their frequencies, and patterns, this paper examines their developmental, neurogenic, and psychogenic aspects. The causes of stuttering, encompassing genetic factors and neurobiological effects, are explored with an emphasis on the role of non-coding regions in DNA. The implications of neuroimaging technologies display altered patterns within the neural circuitry associated with speech production and motor control, and highlight the impact of emotional stress on brain regions. Exploring family heredity with a specific focus on twins investigates the role of specific genes that influence speech development and contribute to stuttering. Presenting a review of the current literature neuroimaging, genomic, and brain studies, this paper aims to contribute to a broader spectrum of other speech disorders, overall raising awareness of stuttering among both professionals and the public. This paper aims to deliver a deeper understanding of a topic that is not as researched as others and sheds light on the various components that affect stuttering in many ways.

I. Introduction

Stuttering, a speech disorder characterized by disruptions in speech motor behavior resulting in sound and syllable repetitions, along with prolongation of certain sounds, encapsulates the vast nature of stuttering, highlighting its impact on both verbal communication and the broader dimensions of affected individuals. (Max et al., 2004)

The prevalence and impact of stuttering underscores the significance of understanding the far-reaching consequences of this speech disorder. Stuttering is not a phase of language development; rather, it affects individuals across their lifespan, presenting unique challenges in various aspects of daily life in academic, professional, and social spheres. Here, the paper will discuss speech disfluencies and the types of stuttering, along with the various factors that cause them.

I. Understanding Speech Disfluencies

A. Definition and Types of Disfluencies

Speech disfluencies refer to breaks or disruptions in speech experienced by a speaker. A common example is stuttering; overall, such disfluencies are often categorized as typical and atypical. Atypical disfluencies occur when individuals experience abnormal speech rhythms, behavioral changes, or certain interruptions. It mainly covers the rhythm and repetition of speech, hesitations with speech, and prolongations. (American Speech-Language-Hearing Association [ASHA], 1993) An example of atypical rate/rhythm is the repetition of a sound, syllable, or word during speech. Similarly, hesitation, often intensified by tension and secondary behaviors (facial grimaces, eye blinking, abnormal body movements, avoiding speaking in situations they find difficult to say) during a conversation, may be marked by fillers and

interjections such as “um,” “uh,” and “like” as well. Along with repetitions, the use of fillers, and hesitations while speaking, prolongations involve the stretching or extent of sounds, syllables, or vowels for a period of time.

Typical disfluencies, often recognized through revisions (where the speaker rephrases or corrects themselves mid-sentence), repetitions (repeating short phrases for emphasis or clarification), and incomplete phrases, can occur or last for any period and usually do not raise particular concerns in terms of treatment or the need for intervention. Common among children are typical disfluencies because of fatigue, excitement, or unhappiness.

B. Frequency and Patterns

One fundamental aspect of studying speech disfluencies is focusing on the frequency in spoken language. Frequency analyses showed that fillers (e.g. "uh" and "um"), repetitions, and revisions, often appear 50-100 times in conversions with atypical disfluencies, also visualized in Figure 1. (Gregg et. Al., 2012) If we refer to Figure 1, we can see the correlation between age and the percentage of stuttered syllables in a stuttering episode. Different types of disfluency may exhibit distinct patterns across contexts and discussions. For instance, filled pauses are more prevalent in spontaneous, unscripted speech, while repetitions and revisions are more common in speech that requires problem-solving or storytelling.(Redford, 2012)

Additionally, the distribution of speech disfluencies varies among specific areas, especially with studies indicating that disfluencies tend to cluster at specific points in a sentence such as the beginning, end, or even during specific points of a sentence in a different language due to their orthographic transparency.

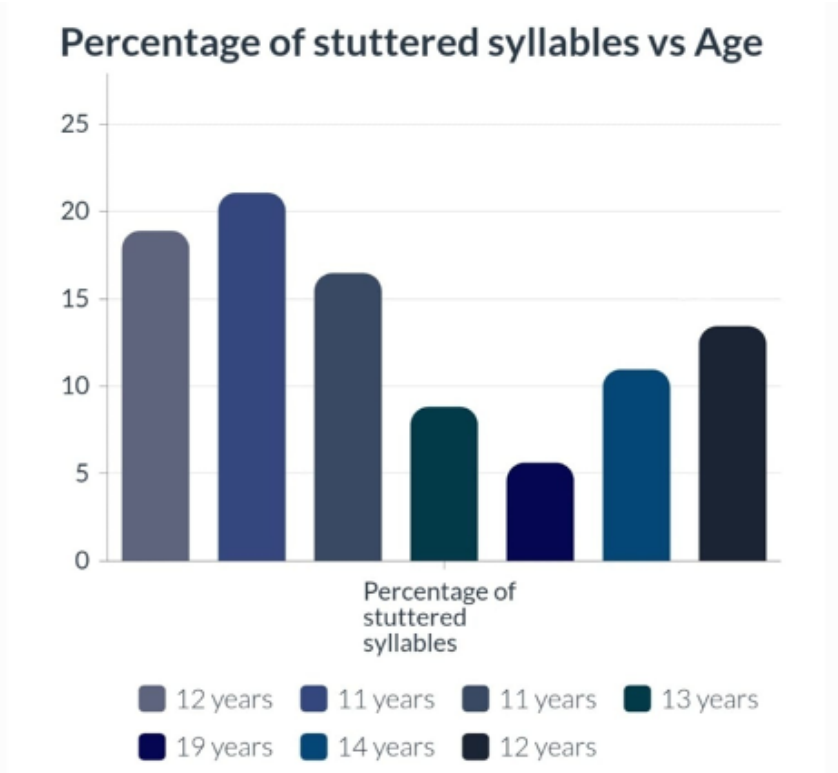

Figure 1: (Jani et al., 2013)

Age	Gender	Percentage of stuttered syllables
12	F	18.9%
11	F	21.09%
11	M	16.48%
13	M	8.83%
19	M	5.56%
14	M	10.97%
12	M	13.45%

II. Types of Stuttering

Referencing Figure 2, all the speech disfluencies and can be portrayed in an organized manner.

A. Developmental Stuttering

1. Characteristics

As developmental stuttering is an atypical disfluency, many of its characteristics are along the lines of disruptions in typical speech flow, repetitions, prolongations, or blocks (which correlate with hesitation and tension). Stuttering may vary from child to child in severity and can be influenced by many situational factors. Some examples include language, where certain languages require different phonetic abilities; environment, where negative feelings or emotions increase tension; and brain function, where areas of the brain responsible for language and speech look and work differently in people who stutter. (Singer et al., 2020)

2. Prevalence in Children and Adults
Developmental stuttering is most common among young children, especially new speakers, because it occurs when a child's speech and language abilities do not meet their verbal capabilities. While many children outgrow stuttering naturally, some may require intervention to reduce its impact on communication and social interaction.

Developmental stuttering is an atypical disfluency that is not as prominent during adulthood; thus, there have been major differences between stuttering in adults and children. Reduced or abnormal activity in auditory-associated areas, increased activity in the right frontal and left cerebellar regions (related to stuttering), deficiencies in left gray matter volume, reduced white matter in the left hemisphere, and irregular timing between motor regions in the left hemisphere of the brain are all present in persistent stuttering seen in adults. (Chang et al.,2007) Asymmetries in the brain structure of adults may result in functional differences and play a role in handedness (Buchel et al., 2004), language literacy, bilingualism (Nucifora et al., 2005), and instrument practice (Gaser and Schlaug, 2003).

B. Neurogenic Stuttering
Neurogenic stuttering is defined as a type of stuttering in which the disfluencies associated with brain damage or injury lead to disturbances in the typical function of the brain in those who do not stutter. (Junuzovic-Zunic et al., 2021) Unlike developmental stuttering, which mostly occurs in children, neurogenic stuttering can occur at any age because of neurological impairment. Distinct characteristics of neurogenic stuttering, such as uncontrollable speech rate, long pauses, increase in repetitions (especially those of syllables and words), prolongation, and rapid speech outbursts, are similar to those of other communication disorders, including aphasia, dysarthria, apraxia of speech, palilalia, anomia, and confusion.

1. Association With Brain Lesions and Injuries (Extraneous causes)
Based on the definition of neurogenic stuttering, it is not possible for it to be associated with damage to a particular part of the brain, as rather it takes part in the neural network for fluent speech production. Similarly, the occurrence of neurogenic stuttering, especially from a stroke, is not limited to a lesion in a specific region of the brain but also correlates with the basal ganglia, lower frontal cortex, upper temporal cortex, and white matter, all of which are related to neural sensory and motor network speech.

C. Psychogenic Stuttering
1. Psychological Trauma
The roots of psychogenic stuttering are often traced back to psychological trauma, in which individuals experience events or circumstances that have a lasting impact on their mental well-being. Traumatic experiences resulting in severe emotional distress, can manifest in psychogenic stuttering as a psychosomatic response, especially since individuals grapple with their emotional and cognitive challenges associated with such memories. As a rare speech condition, this form is characterized by rapid repetition of sounds and words during speech.

Psychological trauma often involves the formation of traumatic memories that may be linked to speech and communication. The structured response to trauma can manifest in speech

disruptions, as individuals grapple with the emotional and cognitive challenges associated with such memories.

2. Association with Stress and Anxiety
Stress and anxiety can exacerbate or trigger episodes of psychogenic stutter. The disorder often presents itself during periods of heightened stress or emotional turmoil. The association between stress and psychogenic stuttering suggests a direct link between an individual's psychological state and the occurrence of speech disruption. The anticipation of potential stressors or anxiety-inducing situations, such as increased muscle tension and altered respiratory patterns, can directly influence articulatory and phonatory aspects of speech.

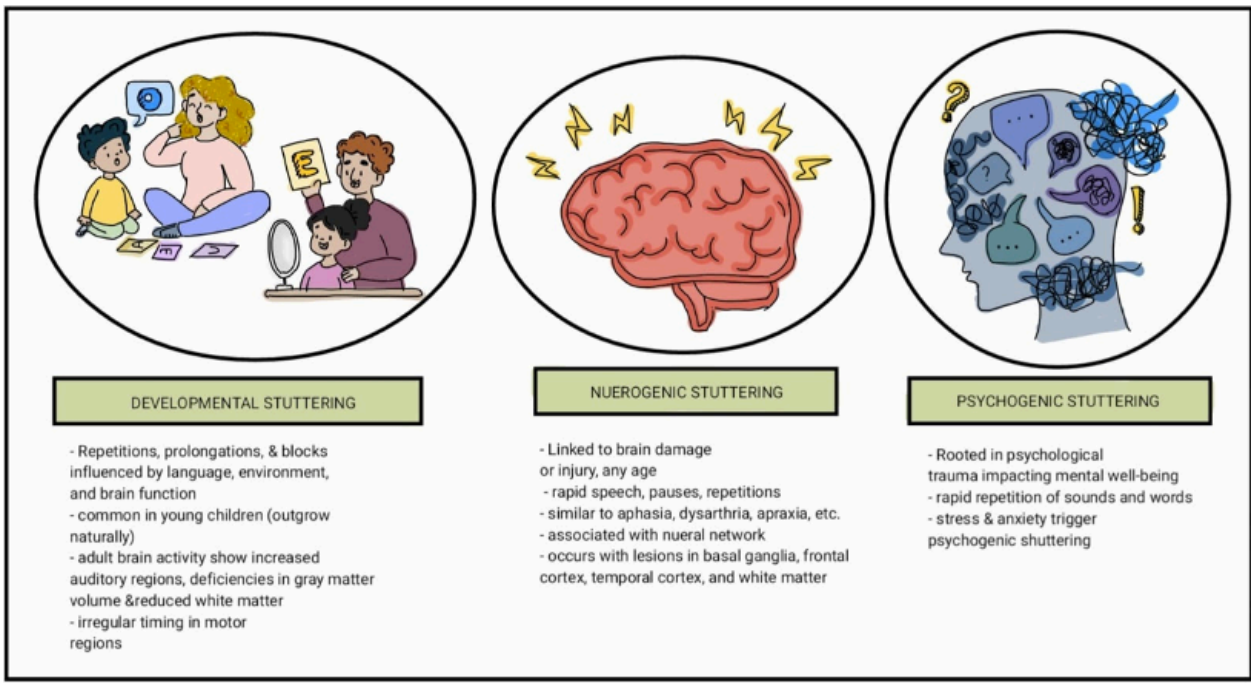

Figure 2: The figure shows the kinds of stuttering (Developmental, Neurogenic, and Psychogenic) and shares their characteristics

III. Causes of Stuttering

A. Genetic Factors

Heredity plays a major component in stuttering, allowing for a deeper outlook into its mechanisms. Family and twin inheritance has shown a higher prevalence of stuttering among first-degree relatives, indicating a genetic predisposition. Genetic factors consist of many components, many of which are causative mutations and ancestry of the population.

Exploring the genetic factors underlying stuttering has revealed a novel dimension with a focus on non-coding regions of DNA. Recent research has revealed that these non-coding regions harbor a substantial proportion of risk alleles associated with stuttering. (Benchek et al. 2021) In the context of genetic regulation, non-coding regions play a pivotal role in the intricate regulation

of gene expression. Notably, the risk alleles identified in these noncoding regions manifest incomplete effects and do not adhere to traditional patterns of inheritance.

This discovery challenges the conventional genetic model, as it underscores the significance of parts of the DNA that do not explicitly provide instructions for protein synthesis. The variations housed in these noncoding regions have been linked to susceptibility to certain conditions or diseases, shedding light on why some individuals are more prone to stuttering. Recognizing the impact of noncoding regions in our genetic makeup not only deepens our understanding of the genetic basis of stuttering but also underscores the broader implications of these regions in influencing health and predisposition to speech sound disorders.

1. Family Heredity

In twin families, twins are prone to stutter more frequently than single-sibling members, as established by one twin child in 11 cases of stuttering, whereas only one in 35 cases (in single siblings). Since identical twin pairs share all of their DNA, while non-identical twins share only half (similar to non-twin siblings), looking into identical and fraternal twins, the identical twin concordance for stuttering is 20% higher than that of fraternal twins. This relationship means that the factor in germplasm-producing twins may be the gene that results in the correlation between abnormal development of polarity in an embryo, which affects the production of normal speech. (Kraft et al. 2011)

Extended families (including parents, siblings, and grandparents) have a higher probability of stuttering than nuclear families (only parents and children), and many of the genes that are affected by speech impediment include FOXP2, a known gene that is associated with speech disorders (e.g. verbal dyspraxia), CNTNAP2, and KIAA, which are genes that affect phonological/expressive aspects, DACT, an excitatory synapse organization and dendrite formation during neuronal differentiation, and chromosome 18, which has the most relevant impact on those who stutter. (Benchek et al. 2021)

Chromosome 18, specifically, has a relatively large area, as it consists of clusters of genes belonging to desmoglein and cadherin. These two genes play important roles in cell adhesion, intercellular communication, and hearing impairment, all of which are relevant to the production of speech.

B. Neurobiological Aspects

Stuttering is closely linked to aberrations in the structure and function of the brain. Neuroimaging techniques, such as Magnetic Resonance Imaging (MRI) and Functional Magnetic Resonance Imaging (fMRI), have revealed differences in the neural circuitry of individuals who stutter. Figure 3 displays the relationship between all neurobiological aspects, along with the roles they take part in.

1. Brain Structure and Function

Investigating the neurobiological aspects of stuttering has revealed intricate relationships between brain structure and function. Stuttering, characterized by disruptions in speech fluency, is associated with a breakdown in the integration of auditory data during the planning of speech motor activities. Notably, connectivity analyses showed a significant reduction in the connections

between the left inferior frontal gyrus (IFG) and basal ganglia in individuals with stuttering. (Gajbe et al., 2022) This reduction points towards a key neural substrate for the manifestation of stuttering, suggesting that compromised communication between these brain regions contributes to the disorder's characteristic speech interruptions. (Benchek et al. 2021)

Further exploration into the brain regions implicated in stuttering extends to Broca's region, where decreased perfusion and indicators of neuronal density have been identified. (Desai et al,. 2016) This finding suggests that the neural underpinnings of stuttering go beyond mere connectivity issues involving alterations in regional blood flow and neuronal composition. Interestingly, the impact of stuttering on speech fluency is not uniform across all linguistic activities, with spontaneous speech being affected more than conversation, reading, sentence repetition, or singing.

The dual-process theory of language formation offers a valuable framework for understanding the neurobiological basis of stuttering. According to this theory, the basal ganglia plays a crucial role in handling formulaic phrases, while the left hemisphere is essential for constructing innovative, freshly constructed sentences. (Gajbe et al., 2022) fMRI studies have supported these theoretical foundations by revealing decreased frontal lobe activity in the inferior frontal gyrus of adults with stuttering.

In addition, the left language-dominant brain hemisphere, which is traditionally active during speech and language tasks, exhibits abnormal dominance in individuals who stutter. Early studies using electroencephalogram (EEG) lateralization techniques strongly suggest atypical brain lateralization patterns in stutterers.

2. Neuroimaging:

Functional brain imaging studies have revealed altered patterns of activation and connectivity within regions closely tied to speech production and motor control. (Chang et al., 2009) These images not only capture the dynamic aspects of stuttering but also provide a foundation for developing interventions grounded in neurobiological understanding.

Neurochemical investigations have linked stuttering to disorders within a network of structures that control movement, particularly in the basal ganglia. (Gajbe et al., 2022) Notably, an increase in the neurotransmitter dopamine, which is associated with movement disorders such as Tourette syndrome, has been identified, suggesting a role for the dopaminergic neurotransmitter system in stuttering. (Büchel et al., 2004)

3. Correlations with Psychological Factors

fMRI scans have revealed altered activation patterns in regions associated with emotion regulation and speech production in individuals who stutter. Specifically, heightened emotional stress has been correlated with increased activity in the amygdala, a key brain region involved in emotional processing. (Zengin-Bolatkale et al., 2018)

Additionally, the prefrontal cortex, a region associated with executive function and emotional regulation, has shown differential activation in response to emotional stress in individuals who stutter. The intricate balance between the prefrontal cortex and limbic system, which includes

the amygdala, appears to be disrupted during emotionally charged episodes, potentially contributing to increased speech disfluency. (Girotti et al., 2017)

Studies using positron emission tomography (PET) scans have indicated alterations in neurotransmitter systems, particularly those involving dopamine, in individuals who stutter under emotional stress. Dopaminergic pathways, which play a role in emotional responses and motor control, may be dysregulated in individuals with stuttering. (Alm, 2021)

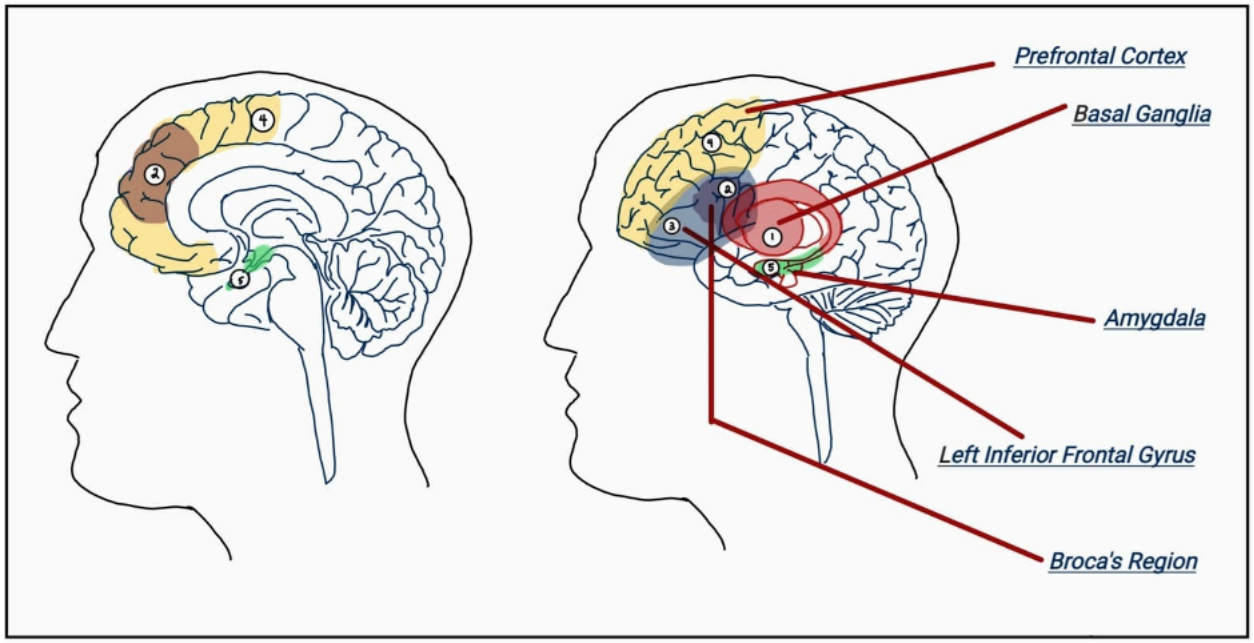

Figure 3: The Left Inferior Frontal Gyrus (IFG) is mainly involved in planning speech motor activities. It also has a reduced connectivity in individuals with stuttering, indicating a key neural substrate for the disorder. The Basal Ganglia is crucial for handling formulaic phrases. There is decreased connectivity with the left IFG observed in stuttering individuals, suggesting its involvement in speech disruptions. The Broca's Region predominantly associated with language formation. Similarly, it also has decreased perfusion and altered neuronal density in individuals with stuttering, indicating its role in speech fluency. The Amygdala is involved in processing emotions and has heightened activity during emotional stress in individuals who stutter, suggesting a correlation with increased speech disfluency. The Prefrontal Cortex is associated with executive functions and emotional regulation. It shows differential activation in response to emotional stress in individuals who stutter, potentially contributing to speech disfluency during emotionally charged episodes.

C. Psychological Factors

1. Behavioral and Environmental Influences

Avoidance behaviors such as word substitutions or circumlocutions (the use of many words where fewer would do) may become ingrained over time and affect neural plasticity. Functional neuroimaging reveals distinct neural signatures associated with these learned behaviors, indicating that the brain undergoes adaptive changes as individuals navigate stuttering challenges.

Early experiences that include negative responses to stuttering may contribute to the development and persistence of stuttering behavior. Additionally, environmental stressors such as high-pressure communication situations can trigger or worsen stuttering. Stigmatization and negative societal attitudes can induce biological changes in individuals. Chronic exposure to negative reactions leads to heightened stress responses that trigger changes in the neuroendocrine system. Elevated cortisol levels, which are indicative of increased stress, have also been observed in individuals who stutter during communicative situations. (Blood et al., 1997)

D. Psychological responses to stuttering

1. Emotional and Psychological Stress

Individuals who stutter may experience heightened anxiety and emotional tension, particularly in communicative situations, which contributes to increased speech dysfluency. The fear of stuttering, commonly known as anticipatory anxiety, can become a pervasive emotional undercurrent that affects individuals in communicative situations. This emotional and psychological stress can create a self-reinforcing cycle, where the fear of stuttering leads to increased anxiety, subsequently exacerbating speech disorders.

Individuals who stutter may develop maladaptive thought patterns and coping mechanisms in response to the challenges posed by their speech disorders. Negative self-perceptions and social anxiety can further intensify the psychological stress experienced by individuals who stutter.

With these challenges, the mechanisms underlying stuttering and its psychological ramifications is crucial to understand. The brain's capacity for plasticity and adaptive learning comes to the forefront in understanding how psychological responses to stuttering mold neural pathways over time. Behavioral responses to emotional stress, such as avoidance behaviors, become ingrained through adaptive learning mechanisms that shape the neural circuits associated with speech production and emotional regulation (Figure 4).

Figure 4: The feedback loop above shows the cycle of psychological responses towards a stuttering episode

Conclusion

The examination of frequency and patterns reveals distinct characteristics of atypical disfluencies, such as filled pauses, repetitions, and revisions, occurring more frequently in specific contexts. Frequency analyses revealed distinct patterns in atypical disfluencies that varied across contexts. Stuttering, which takes many forms, is often organized as developmental, neurogenic, and psychogenic stuttering, presenting various factors influencing its occurrence.

Genetic exploration, particularly in noncoding regions, also challenges the role of noncoding DNA in individuals who stutter. With the employment of neuroimaging technologies, altered patterns of activation and connectivity within the neural circuitry are displayed, along with the impact of emotional stress on brain regions. Family heredity, with a focus on twin and extended families, sheds light on specific genes, such as FOXP2 and Chromosome 18, implicated in stuttering.

Psychological responses to stuttering, including emotional stress and learned behaviors, contribute to a self-reinforcing cycle that shapes the neural pathways. With a specific focus on neuroimaging techniques, they showed the correlation between the basal ganglia and the left hemisphere throughout a stuttering episode.

As a whole, the role of genetic, neurobiological, and psychological factors in speech disfluencies provides a comprehensive perspective of stuttering, offering valuable insights for future research and interventions.

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