Exploring Neurological Development and Sensory Processing Challenges in Individuals with Down Syndrome

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Abstract:

Introduction
Down Syndrome affects almost 6,000 newborns in the United States. It is one of the most common genetic disorders, yet not much is known behind its neurological development and behaviors. While physical traits such as short stature and unique facial features are seen and well understood, internal processes are often misunderstood. Patients with Down syndrome are often overstimulated with loud noises. The etiology behind this overstimulation remains unanswered or often neglected. My focus will be on the difficulties of sensory processing that influence their distinctive behavior.

Methods
Through literature review I sought to gain knowledge on neurological development as it pertains to sensory responses for patients with Down Syndrome.

Discussion
The main cause for sensitivity to sounds within patients who have Down Syndrome is a condition called Hyperacusis. With the presence of pre-senile dementia, irritation intensifies leading up to behaviors such as screaming and throwing tantrums in response to the inability of communicating and comprehending external stimuli. The Stapedius Acoustic Reflex is another major component within sensory processing. Such sensitivity reveals why pain and discomfort is experienced when processing loud sounds.

Conclusion
By having proper understanding of the struggles individuals with Down Syndrome face, we are able to facilitate a healthy environment in which respectful and empathetic interactions among patients and community members are promoted.

Introduction:

Through years of scientific studies and research, it is well known that each cell in a human body consists of 23 pairs of chromosomes. This rod-like structure contains genetic material which provides each individual with their traits or characteristics, passed down through their parents. Now, what happens when an extra pair appears in the cell? A genetic disorder such as Down Syndrome, for example, is likely to form. This happens particularly when an abnormal cell division of an extra pair or a partial chromosome 21 is passed down to the individual. Hence, this affects the characteristics or features of an individual. While Down Syndrome has full penetrance, expressivity often varies. Individuals with Down Syndrome share the same genotype, meaning that they share a complete set of the genetic material or hereditary information, but they can have different phenotypes. Phenotypes are the observable traits of an individual. This can lead to individuals obtaining different developmental traits at different degrees including distinct facial features, behaviors, health issues, or intellectual disabilities.

In the United States, there are far more patients with Down Syndrome than one may think there are. The Centers for Disease Control and Prevention estimates that “one in every 772 babies in the United States is born with Down syndrome, making Down syndrome the most common chromosomal condition”.[1] There are several types of mechanisms that lead to Down Syndrome including Trisomy 21, Mosaicism, and Translocation; Trisomy 21 being the most common making up 95% of the cases. All three genetic variations consist of similar symptoms and there is no known specific reason as to why such a genetic disturbance occurs. There may however be a slight difference in cognitive intelligence and behavior. It is known that Mosaicism has the least characteristics of Down Syndrome compared to Trisomy 21 and Translocation. For example, research conducted on the correlation between Mosaicism and Trisomy 21 reveals that individuals with Mosaicism tend to have slightly small ears which are in normal shape, slightly broad nasal bridge, subtle epicanthal folds, and a high arched palate.[2] This is due to the fact that individuals with this genetic variation still have some cells with the normal number of chromosomes, while only a few have an extra pair. Therefore, the degree of expressivity related to Down Syndrome is far less.

While some say that Down Syndrome is a result of an excessive number of genes or proteins in the cells, this thought remains a working theory.[3] While there remains the lack of information towards the root causes of this genetic disorder, research has shown that 51% of children with Down Syndrome are born with mothers younger than the age of 35.[1] This observation is particularly due to the higher birth rates during those ages. Since there are more women having babies at ages 35 or younger, there are more babies with Down Syndrome. At the same time, chances of conceiving a child with genetic disorder becomes higher as women age; going from 1 in 350 to 1 in 30. This means that even though older women are more prone to having babies with disorders, there are more women having babies with Down Syndrome at ages younger than 35. That adds up to the overall percentage of babies with Down Syndrome born to women ages under 35 to equal more than babies with Down Syndrome born to women ages higher than 35. While there are age factors in statistics regarding women, it doesn’t depict whether the father or mother of the child is responsible for causing an extra chromosome 21. Both parents can be equally responsible for passing down chromosome 21.

There are several common and noticeable features of individuals with Down Syndrome. Such individuals usually have a smaller face and shorter neck. They also tend to have shorter height and small fingers. One can also notice that they have slanted eyes. A main symptom of Down Syndrome is the significant lack of intellectual ability. Individuals with this genetic disorder tend to have delays in motor and cognitive skills, as well as in speech or communication. While research has shown their life expectancy to be about 60 years, they still continue to remain at risk for further health complications. Heart defects, obesity, dementia, and immune disorders are highly associated with Down Syndrome.

Diving deeper into the risks, Down Syndrome patients are severely prone to low immunity. Scientific studies reveal that the main cause for this is because interferons are constantly active amongst such patients, unlike people without this condition whose interferons are only activated when they are exposed to an infection. Interferons are molecules produced by cells that fight against an infection. That means that if the interferons are constantly working, they won’t

respond in time of need and as such, people are likely to develop autoimmune disorders such as Alzheimer's Disease or even Leukemia.[4] This occurs because the interferons can increase the activity of autoreactive cells (T cells and B cells) by inducing genes in antigen-presenting cells (APCs).[5] During the induction, the ACP is stimulated and enhances hormonal autoimmunity, therefore keeping autoreactive cells active and triggering autoimmune disorders. Additionally, as these patients grow older, they are more than likely to catch certain infections and have difficulty treating them due to lower immunity. An example of this is having frequent ear infections. Individuals with Down Syndrome have constant ear infections because of narrow ear canals. Such infections can become hard to cure and overtime it can result in hearing impairment because the immune system is not strong enough to fight off the infection. Overall, this condition can lead to hypersensitivity to hearing which can promote irregular behaviors among patients with Down Syndrome.

Methods:

For this research, I did a literature review by using google scholar to facilitate my findings. The process consisted of brainstorming what was already known, then figuring out what needs to be known, and finally reviewing the findings in order to make broader connections based on the research questions. All of my references used to formulate this literature review were filtered by sources published in the United States prior to 2017. I started off by searching "Down Syndrome" and then adding other qualifiers such as Down Syndrome and "hearing, characteristics, and behaviors ". I then proceeded to search about specific traits such as differences in intellectual ability and narrowed my search to "Sensorineural development" in individuals with Down Syndrome. I specifically looked for subheading directed towards the brain, auditory functions, and behavioral attributes within the articles and research papers. I took notes on a google document and saved the sources I used on Zotero, a platform that assists in organizing, annotating, and citing research.

Results:

Research on DS has revealed that the human chromosome 21, also known as HSA21 has genes which are known to influence brain development. The HSA21 genes encodes alterations in brain volume and cell density of specific brain regions.[6] Specifically, there is a reduction in the size of the cerebellar volume. The cerebellum also plays an important role in auditory feedback and control of speech production.[7] People with this syndrome have fatigue in the neural connections, causing a delay in understanding a stimulus and even more in elaborating a response to that stimulus.[8] Cortical neurons, located in the cerebral cortex, carry Trisomy 21 but subsequently degenerate and undergo apoptosis.[8] The delay within the cerebral cortex brings up challenges associated with attention, emotional inadequacy, improper social behavior, the inability to understand what others say, and problems with speech.[9] Pekkonen et al. demonstrated there is a delay in cortical processing in Down Syndrome through an experiment in which sinusoidal tones were presented to evoke auditory response. The evidence also proved that Down Syndrome has similar neuropathology with Alzheimer's Disease. Hence, the article suggested that impaired preattentive auditory processing in DS might be linked to destruction of cholinergic neurons.[10]

Approximately 38% to 78% of children born with Down Syndrome have hearing loss.[6] It ranges from conductive to sensorineural or mixed hearing loss.[11] Conductive hearing loss occurs due sound waves not passing through the middle ear, whereas sensorineural hearing loss occurs due to a loss or damage of ear cells.[7] Otitis media, a type of conductive hearing loss, results in severe inflammation due to constant infections in the middle ear. Babies and kids are typically prone to ear infections which are often recurrent.[7]Cholesteatoma, a condition due to an accumulation of debris resulting in an abnormal growth of cyst like skin cells and tissues behind the eardrum is just another disease that is quite often found in children with Down Syndrome following episodes of otorrhea, also known as fluid drainage in the ears. Such infections are hard to cure within these patients because of their low immunity and cure rates.

It is known that effects of aging appear early in patients with DS, about 20 years earlier than in the general population.[8] This is a result of the lack of cortical habituation, by reduced amount of nerve fibers for stimulus conduction, or even by senile or pre-senile dementia.[8] Ultimately, it causes deficiencies in sensory and cognition; especially in the temporal regions. Alzheimer's Disease is the most common autoimmune disorder present in individuals with Down Syndrome. They tend to get it by the fifth decade of their life; way earlier than individuals without Down Syndrome which get it around age 60-65.[12]

While sensory and cognitive abilities decline as people with Down Syndrome age, their stimulative responses also decline. Therefore, Alzheimer's Disease contributes to low stimulus response, making it difficult for people with Down Syndrome to adapt or comprehend anything related to auditory sensations. They use a reflex system to counteract the inability to process auditory sensitivity. The Stapedius Acoustic Reflex is caused by the contraction of the stapedius muscle in the middle ear due to high frequency auditory stimuli. This causes vibration of the cochlea which also results in hypersensitivity towards auditory stimuli.[13] This reflex helps in discriminating against unwanted sounds that cause discomfort to individuals with Down Syndrome.

Discussion:

Individuals with Down Syndrome are more likely to be born with hearing loss than individuals without DS. Interestingly, once they get older, they become more sensitive to loud sounds. Of note, conductive hearing loss is the most common type of hearing loss in patients with Down Syndrome. Individuals with Down Syndrome have several alterations in their brain development; therefore, it can be inferred that the main source of the blockage could be the structural composition of the brain in patients with Down Syndrome. Narrow ear canals and disproportionate sizes of the three middle ear bones known as the malleus, incus, and stapes are a common cause for this type of hearing loss in babies and younger children.

Majority of infants are diagnosed with hearing loss because of middle ear infection or wax.[14] Narrow ear canals and disproportionate ear bones make it easier for fluid to be retained in the middle ear. When fluid remains in the middle ear, bacteria can easily grow leading up to a severe ear infection. Not only can this infection become chronic, it will be hard to fight off because of the low immunity in people with Down Syndrome. Thus, otitis media is really common in such patients. Since Down Syndrome patients cannot fight off the infection easily, they will suffer from irritation and hearing loss. That also means that younger patients won't

necessarily process loud sounds, but rather face other issues as they are not able to comprehend sensitivity to sounds yet.

Over time though, the constant irritation in the ear due to recurrent infections can result in a condition known as hyperacusis. Hyperacusis occurs when normal everyday sounds or even higher stimulated sounds can become extremely intolerable. It is hard to say exactly what causes hyperacusis, but many researchers provide theories that it is because of damage caused on auditory or facial nerves. Some also say that hearing hypersensitivity occurs because there is a dysfunction of the outer hair cells of the organ of Corti; with a rising sound level, the perceived loudness increases faster than normal.[15] Through my literature review, I was able to find that the cortical neurons in the cerebral cortex underwent apoptosis in individuals with Down Syndrome. This will result in the damage of the cerebral cortex. The temporal lobe is included in the cerebral cortex and is responsible for most of the auditory functions. When the cortical neurons undergo apoptosis, the functions of the temporal lobe are also affected.

Low immunity is significantly prominent in individuals with Down Syndrome and over time, it can lead to autoimmune disorders which also affects hearing. As previously described, interferons are constantly active in patients with DS, decreasing their ability to appropriately fight infections. Overtime, these recurrent infections can result in autoimmune disorders due to overactivation, such as Alzheimer's Disease. My literature research highlighted that there is a reduction in the size of the cerebellar volume, similar to Alzheimer's which is defined by cerebral atrophy. Cerebral atrophy occurs when there is a loss of neurons that affect connections between brain tissue[16]. This will lead to brain size shrinking and cognitive ability will then decline. By this time, roughly 46% of such individuals with Alzheimer's disease face a significant reduction in their focal neurological signs and experience a loss in communication skills.[12] Therefore, any severe stimulative response to loud noises can become intolerable and hard for them to express.

As the neurodevelopmental aspect of auditory processing is understood, reasons behind maladaptive behaviors associated with such intense or loud auditory processing remain restrictive. It can be assumed that the lack of sufficient intellectual growth caused by neurological impairments can lead to difficulties in expressing discomfort. While there are certain delays or failures in auditory functions, it is not the only affected area. In individuals with Down Syndrome, issues in different aspects of development in the body cause great impact to how the overall body functions, processes, and responds to external factors.

Looking into certain maladaptive behaviors, patients with Down Syndrome express a variety of maladjusted behavioral phenotypes as a response to unwanted stimuli. Inability to comprehend sensory reflexes can cause irritation, tantrums, disobedience, hyperactivity, and poor communication just a few to name.[17]

For example, as these individuals are overly sensitive to loud sounds, it can become difficult for them to distinguish between external sounds and verbal sounds. Therefore, any verbal tasks or communications may be ignored by the individual with Down Syndrome. Such ignorance to important sounds can affect their daily interactions with others in the environment. It can also cause great distress for certain patients with Down Syndrome to understand what is happening in their environment and understand if there is a need for them to react or not; additionally accelerating behaviors of irritation, confusion, and possibly fear.

Low sensory detection in children with Down Syndrome will result in less maladaptive behaviors, but as they grow older, they develop over-sensitive detection, leading to more aggressive behaviors and hyperactivity. Since most patients with Down Syndrome have below average levels of verbal communication skills, they mainly communicate through their actions. Inability to properly communicate their discomfort builds up frustration and can lead to hyperactivity, yelling, or even jumping. In some cases, unintentional violence can also be noted. In addition, random verbal sounds are usually another way of expressing discomfort to loud sounds.

As discussed before, the stapedial acoustic reflex is a reflex system that individuals with Down Syndrome use to ignore the sensory input they process from external factors. Some ways to observe if they are partaking in this reflex is to observe their facial movements. They might close their ears or even talk to themselves as a way of distracting themselves from external stimuli.

Conclusion:

Down Syndrome is a genetic disorder where several phenotypes are noticeable, such as distinct facial features, behaviors, and intellectual disability. Through this literature review, I was able to look deeper into several issues concerning hearing impairment and sensitivity that patients with Down Syndrome face. A major takeaway from this experience involves being able to understand in what ways such individuals are being affected in their everyday lives. To many of us, concerns related to hearing sensitivity may not seem like a big deal, but to individuals with Down Syndrome it is. Therefore, by learning about their struggles internally and externally, we can facilitate a healthier environment in which boundaries are respected. When interacting with individuals who have Down Syndrome, we should understand that the way they perceive environmental factors is different from how individuals without Down Syndrome do. Therefore, it is our responsibility to look out for them and make sure that none of our actions, such as being loud, trigger them negatively. In future projects, I would like to focus on ways to teach and promote methods of giving immediate assistance to individuals with Down Syndrome who struggle with interacting with other individuals or their environment.

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