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Down Syndrome

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Parents play a critical role in determining the children's health before and after they are born. Through gene hereditary, parents can pass certain conditions to their offspring. Since the gene is the structural block of hereditary, they have deoxyribonucleic acid, which generates proteins that perform critical roles in the cells. Down syndrome is among the common genetic Disorder instigated by abnormal cell division where there is more or incomplete copy of chromosome 21. CDC (2021) claimed that chromosomes are minor packages of genes in the human body. They regulate how the child's body appears and operates as it develops during pregnancy and after birth. Moreover, since children with Down Syndrome (DS) have more copies of chromosome 21, the word used to refer to this is trisomy hence known as trisomy 21. Although the Disorder affects children before and after birth, this paper will explore the down syndrome disorder by looking at its history, evaluating a case on the same, and explaining the treatment options and my reflection on the Disorder.

History of the Disorder

The concept of trisomy 21 originates nearly two thousand five hundred years ago when Briceno and Bernal reasoned that some sculptures indicate people with the Disorder. Although this was the initial experiential sign of the illness, Martinez-Frias recognized the syndrome in five hundred people with Alzheimer's disease where the facial characteristics of trisomy 21 exhibited evidently (Kazemi et al., 2016). The same source added that various experts used images to explain the profound demonstrations of the syndrome between the 15th and 16th centuries. Some of the critical scientists that helped improve the concept of DS are Esquirol, who developed the phenotypic explanation of trisomy 21, and John Langdon Down described the

phenotypic kids with typical characteristics identified from other progenies with mental obstruction.

Moreover, in 1866 John Down realized the syndrome, and that is why it was referred to as the Down Syndrome. Kazemi et al. (2016) explain that the causes of the Disorder were not recognized until the 20th century; nonetheless, the possibility that the syndrome can be caused by chromosomal irregularity was proposed in 1932. As more research continues to identify the cause of the Disorder, several experimental conditions conducted in 1956 permitted scientists to precisely describe the number of individuals' chromosomes as forty-six. However, in 1959, more research allowed scientists to recognize an additional copy of chromosome 21 in karyotypes organized in DS individuals, which were determined to be the cause of the Disorder.

A Case Found

The case concerning the Disorder is found in CBS News, where a mother provides her experience about her eighteen-month daughter living with DS disorder. Marguerite Reardon is among many mothers facing significant challenges in training their children to strengthen their muscles and develop as independent individuals. Reardon was shocked after realizing from an early prenatal blood test that her baby would have DS (CBSN, 2015). Furthermore, Reardon explains to the new media the excruciating choices made and the hopes and dreams she has for her daughter. The test was conducted by counting the chromosome of the baby. Although the child has low muscle ton, Reardon can train the child by starting an early therapy to train the muscles. For example, Reardon uses the chewing tube to make the muscles strong and move the tongue around. Together with the father, they work to ensure they provide the best help to the child and grow to be independent individuals.

Age the Disorder is Diagnosed

Children can be diagnosed with DS in both prenatal and postnatal. The prenatal diagnosis always occurs at 15 to 22 weeks of pregnancy. Kazemi et al. (2016) claimed that amniocentesis is the most conservative invasive prenatal diagnosis approach used and allowed across the globe. By using an integrated screening test, the physician can associate the results from the initial trimester screening and the second-trimester screening to increase the likelihood that the child may have DS. Therefore, the process is performed by taking the amniotic fluid for karyotyping starting from fifteen weeks. Nonetheless, the test can be conducted before fifteen weeks and will be referred to as early amniocentesis. Also, chorionic villus sampling (CVS) is done between 11 and 13 weeks of gestation and comprises of ambition or operation of placenta villi. Although CVS and amniocentesis are dependable, however, they enhance the danger of miscarriage.

Another age to diagnose a child with DS is at birth. During delivery, physicians can be aware of the likelihood of the Disorder on a child depending on the physical appearance. Although Bull (2020) states that physical inspection is the precise diagnostic evaluation, a qualified clinician will realize the body habitus and physiognomic characteristics, usually conveyed by muscular hypotonia. Similarly, Ostermaier (2019) stated that some of the physical features that can be identified to the child with DS are upward slant to the eyes, flat occiput and a flattened facial appearance, a solitary deep crease across the center of the palm, and a small chin, abnormal outer ears, and flat nasal bridge.

Treatment Options

There is a significant increase in the number of children born with DS across the world. For instance, CDC (2021) indicated that nearly 6000 children are born with DS across the United States every year. Therefore, this shows that DS happens to approximately one in every seven hundred children. Since the number of children born with DS is high, several treatment options

are developed to improve the quality of life of these children. Similarly, Kazemi et al. (2016) claimed that there is no medical cure for the Disorder. Nevertheless, kids with the Disorder can get help from early therapeutic support and advanced interventions during childhood.

The early support with help children and babies with DS to enhance their physical and rational capacities. The majority of the services concentrate on assisting kids with DS grow to their full potential. Some of the help used comprise occupation therapy, physical therapy, and speech-language therapy. Children under six years old can obtain the best results through interventions because they are developing mechanical habits and speech. Kazemi et al. (2016) stated that speech-language therapy assists children in communication and language skills by concentrating on articulation, reasoning skills and enhancing the oral muscles such as the tongue. Speech therapy can be used during the newborn and toddler phase because the general objective is to improve the kid's communication capacity. Also, DS has significant effects because it can lead to other conditions such as hearing loss (Ostermaier, 2019). Therefore, through speech therapy, one can align the communication forms based on the child's needs.

On the other hand, physical therapy is a critical intervention because it helps children enhance the way they move. Since children with DS always have poor muscle tone and smaller hands, this intervention can alleviate any challenges associated with the characteristics. The therapy routine will possibly comprise firming and toning muscles using a chewing tube, enhancing the general organization and balance, and adjusting posture. Furthermore, occupation therapy intervention will aid DS children because it was developed to enhance the daily activities of children as well as improve the skills required for staying a healthy and prosperous life in society (CDC, 2021). Thus, occupation therapy enhances fine motor abilities and the presentation of everyday roles such as brushing their teeth and dressing.

Reflection Piece

The field of children and youth need to have treatment options. Children and kids with DS need to practice physical therapy to enhance their movement and operate on their daily activities. I would bring physical therapy to the child and youth field by creating a group to guide them on developmental activities such as walking and crawling based on the growth stage. Also, I will recommend them on balance and coordination events, building strength on weaker muscles, and flexibility exercise. Since the session will be divided twice a week, before the start of every session, I will measure the participant's flexibility and strength, examine how they walk and run and recognize prevailing potential issues. I will also implement occupation therapy by visiting the child once a week and providing different strategies on different events. For instance, when a child does not like to brush their teeth, I can recommend trying to use a vibrating toothbrush and at the same time utilize unflavored toothpaste. Finally, I will implement speech and language therapy in the child and youth field by creating categories. The first is to develop a group intervention, and the second is individual intervention. All the groups will require support from parents or caregivers. Also, I will utilize different ways to enhance the patients' speech and language, including active listening and learning, powerful collaboration, attention and listening methods, and language for thinking.

In summation, Down Syndrome is a disorder where an individual has an extra chromosome. The Disorder is a congenital disability associated with high therapeutic and societal costs caused by trisomy of the entire or part of chromosome 21. The disease is named after John Langdon Down as an honor for explaining the medical description. The case of Marguerite Reardon indicates how children with DS are struggling and demanding and how parents are experiencing it. The Disorder can be diagnosed between 15-22 weeks of pregnancy

or at birth because physical appearance can be identified. Similarly, there is no medical cure for the Disorder, but several interventions are used, such as physical, occupational, and speech and language therapy. These interventions can be brought to youth and child care by creating groups of children with similar conditions and involving parents and caregivers in treatments.

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