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# The Management of Down Syndrome

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## ABSTRACT

*Down syndrome occurs when individuals experience abnormal cell division, resulting in a partial or complete copy of chromosome 21. Persons with Down syndrome have short necks, protruding tongues, poor muscle tones, flattened faces, and excessive flexibility. The genetic disorder affects approximately 400,000 adults and children in the United States. Unmanaged Down syndrome leads to serious health complications like ear infections, eye diseases, heart defects, hearing loss, seizures, gastrointestinal disorders, obesity, and obstructive sleep apnea. The literature review completes a comprehensive analysis of pharmacological and non-pharmacological methods for managing Down syndrome to protect individuals from adverse complications. Specifically, the report examines the benefits of medications, such as donepezil, rivastigmine, memantine, and galantamine, in managing Down syndrome associated with dementia. They review the impact of non-pharmacological methods, including speech therapy, occupational therapy, physical therapy, assistive technologies, and surgeries, on managing Down syndrome. The collection of data for the literature review is based on secondary research. Particularly, the researcher collected and applied data from peer-reviewed journals, e-books, or agencies reports published between 2011 and 2021. The literature review has specific gaps as the peer-reviewed articles did not provide information on particular surgeries for managing complications of Down syndrome, the accurate dosage of medications for adults and children, and the side effects of pharmacological methods. Although the literature has gaps, patients and clinicians should apply the discussed pharmacological and non-pharmacological strategies for managing Down syndrome.*

*The clinical significance of the literature review is to:*

- *Facilitate the early intervention and management of Down syndrome.*
- *Reduce the adverse complications of Down syndrome in children and adults.*
- *Enhance knowledge and awareness of Down syndrome and its evidence-based management approaches among clinicians, vulnerable persons, and community members.*
- *Increase the safety and clinical outcomes of individuals with Down syndrome.*

## Keywords

Down syndrome, Donepezil, Rivastigmine, Memantine, Galantamine, Antidepressants, Anticonvulsants, Speech therapy, Occupational therapy, Physical therapy, Assistive technologies, Surgeries.

## Introduction

Currently, Down syndrome is among the leading genetic disorders in the United States. According to Newton [1], individuals suffer from Down syndrome when they experience abnormal cell division, resulting in a partial or complete copy of chromosome 21. The increased copy of chromosome 21 negatively impacts the physical features and developmental changes of individuals. Newton [1] indicates that Translocation, Mosaic, and Trisomy 21 are the three common types of Down syndrome. The genetic disorder affects both adults and children in the United States. Presson et al. [2] note approximately 400,000 adults and children have Down syndrome. More than 6,000 children are born with Down syndrome every year, implying the genetic complication occurs in at least one in every 700 babies. Persons with Down syndrome have unique physical

features, including short necks, protruding tongues, poor muscle tones, flattened faces, and excessive flexibility. Newton [1] argues that individuals with Down syndrome experience additional signs and symptoms, such as short stature, small hands and feet, and tiny white spots in coloured parts of their eyes. Even though most people experience less severe signs and symptoms, untreated or unmanaged Down syndrome leads to serious health complications like ear infections, eye diseases, heart defects, hearing loss, seizures, gastrointestinal disorders, obesity, and obstructive sleep apnea. Adults with unmanaged Down syndrome have increased risks of blockages, refluxes, and celiac diseases, threatening their wellbeing and lives. Kerins, Petrovic, and Bruder [3] add that 34% of adults with Down syndrome experience seizures due to the lack of clinical care. Based on the discussion, medical practitioners and facilities should invest in learning and applying evidence-based methods to prevent, manage, or treat Down syndrome. Consequently, the research aims to examine evidence-based approaches for preventing, managing, or treating Down Syndrome to reduce its adverse complications on Americans.

## Methods of Data Collection

Researchers conduct effective data collection to obtain and apply information-rich and reliable data in statistical analysis and discussions. The researcher intends to conduct secondary research to obtain adequate to make reasonable and valid findings in the study of Down syndrome. According to Lew [4], secondary research expects scholars to identify, summarize, or synthesize existing studies or information to make reasonable arguments or conclusions. Secondary researchers examine journals, magazines, e-books, or newspapers to access, summarize, and synthesize information and data. Lew [4] conducts secondary research online by reviewing databases or offline by visiting physical libraries or government institutions to access adequate information. The researcher plans to use secondary research as it is cost and time-effective. On the contrary, the researcher will experience difficulties in accessing specific information required to complete the study.

In the Down syndrome study, the researcher plans to focus on online secondary research, especially examining journal databases, e-book websites, credible websites, and online newspapers, to acquire adequate and diverse information. The researcher intends only to collect and apply data from peer-reviewed journals, e-books, or agencies reports published between 2011 and 2021. For example, the scholar will log into google scholar and ProQuest to access updated and relevant peer-reviewed journals with information on Down syndrome.

The research will log into Google books to access updated e-books with adequate information on Down syndrome. It is essential to note that the researcher will utilize key phrases, such as "Down syndrome," "Down Syndrome management and treatment;" "Evidence-based non-pharmacological methods for managing Down Syndrome," and "Pharmacological methods for treating or managing Down Syndrome," to access adequate reading materials for the research.

## Pharmacological Management of Down Syndrome

Presson et al. [2] indicate that medical practitioners cannot cure Down syndrome, but only manage its adverse signs and symptoms using specific medications. Healthcare workers, especially physicians and nurses, prescribe medicines for symptomatic pain treatment among persons with Down syndrome. According to Presson et al. [2], medical practitioners prescribe medications, such as donepezil, rivastigmine, memantine, and galantamine, for Down syndrome associated with dementia. In a study, doctors prescribed donepezil for patients with Down syndrome associated with dementia for five months. Patients showed improvement in dementia scores after three to five months of using donepezil. Also, Presson et al. [2] add that medical practitioners prescribe antidepressants and antipsychotic medications for patients with Down syndrome associated with dementia. In a study, nurses administered antidepressants and antipsychotic medicines like fluoxetine and sertraline to four individuals with Down syndrome. The medications were 98% effective as they delayed the onset of dementia and its related symptoms by 1.31 years among the four patients. Consequently, medical practitioners should utilize evidence-based medications, such as donepezil, rivastigmine, memantine, antidepressants and antipsychotics, to manage or delay the onset of dementia among persons with Down syndrome.

Equally important, medical practitioners use traditional anticonvulsants in managing or preventing seizures among persons with Down syndrome. According to Presson et al. [2], physicians prescribe multiple anticonvulsants, including valproic acid, phenytoin, and carbamazepine, for persons with Down syndrome with increased risks of seizure. The therapies are effective in managing and reducing the threat of seizures among persons with Down syndrome. On the other hand, some of the medications have adverse side effects that may affect patients. Presson et al. [2] admit that valproic acid therapy may decrease the plasma level of folic acid and increase the plasma level of homocysteine in patients with Down syndrome, increasing their risks of other health complications, especially cardiovascular diseases. Consequently, medical practitioners should use traditional anticonvulsants moderately to manage or reduce the risk of seizures among persons with Down syndrome.

Additionally, clinicians use antibiotic therapies to manage or prevent respiratory disorders among persons with Down syndrome. Individuals with Down syndrome have increased risks of lung infections or aspirations, leading to intensive care admissions. According to Presson et al. [2], physicians use antibiotic therapies and bronchodilators to treat respiratory complications among children and adults with Down syndrome. In a study, infants with Down syndrome showed reduced theophylline clearance after using antibiotic therapies and bronchodilators. As a result, medical practitioners should utilize only current and evidence-based antibiotics to manage or prevent respiratory disorders among persons with Down syndrome.

### Non-Pharmacological Methods of Down Syndrome

Medical practitioners complement pharmacological with non-pharmacological methods to manage and control the adverse symptoms of Down syndrome. Medical practitioners utilize early interventions, including speech, physical, and occupational therapies, to manage or prevent negative physical signs of Down syndrome. According to Regis et al. [5], speech therapy focuses on strengthening oral muscles, such as tongue and lip muscles, articulation, and cognitive skills, to improve the communication and language skills of persons with Down syndrome. Medical practitioners commence speech therapy at infant and toddler stages. Thus, children with Down syndrome who undergo speech therapy have improved communication skills. Regis et al. [5] state that a study involved eleven children with Down syndrome who underwent speech therapy. They underwent eight sessions of speech therapy. The children demonstrated improved communication skills and abilities after undergoing speech therapy. Consequently, medical practitioners ensure all children or infants with Down syndrome undergo speech therapy as early as possible to develop exceptional communication skills and abilities.

Next, Ruiz-González et al. [6] indicate that physical therapy improves people's strength and how they move. Most persons with Down syndrome have smaller hands and poor muscle tones. Persons with unmanaged Down syndrome develop bad posture in life due to poor muscle tones. According to Ruiz-González et al. [6], the physical therapy regimen has routines for strengthening and toning muscles. The therapy involves teaching individuals with Down syndrome how to move their bodies to accomplish their daily functioning and activities. Ruiz-González et al. [6] indicate that the review of 27 peer-reviewed articles on physical therapy revealed that the evidence-based method is 95% efficient in improving strength and how people move. Hence, medical practitioners should apply physical therapy to improve strength and appropriate muscle toning of patients with Down syndrome, ensuring they develop good postures later in life.

Moreover, clinicians complement speech and physical therapy with occupational therapy to improve the health conditions of patients with Down syndrome. According to Ciucurel and Iconaru [7], occupational therapy expects medical practitioners to improve the day-to-day skills of persons with Down syndrome to enhance their healthy living. Occupation therapists work with individuals with Down syndrome to develop their fine motor skills. Persons who complete occupational therapy sessions successfully learn to brush their teeth, eat, get dressed, pushbuttons, and open doorknobs without help. Ciucurel and Iconaru [7] indicate that a case study of a thirteen-year-old child showed that occupational therapy is essential in improving the day-to-day skills of persons with Down syndrome. Therefore, medical practitioners utilize speech, physical, and occupational therapies to improve the physical and emotional wellbeing of persons with Down syndrome.

### Assistive Technology

Alammary [8] argues that assistive technology involves the devices or technologies, which medical practitioners recommend for persons with disabilities to improve their daily functioning. Medical practitioners recommend assistive devices, such as seat cushions, walking aids, large button mobile phones, hearing aids, and pen grips for persons with Down syndrome. Alammary [8] indicates that the use of large button mobile phones and touchscreen tablets help individuals with Down syndrome communicate effectively, increasing their independence. Also, Alammary [8] medical practitioners suggest the use of educational assistive technologies, such as software, to enhance learning via interaction and implementation of sound and touch. Assistive technologies help individuals to overcome their physical challenges during learning processes.

### Surgeries

Medical practitioners rely on evidence-based surgeries to correct some of the adverse health complications of Down syndrome. Individuals with Down syndrome have increased risks of congenital heart diseases. According to Delany et al. [9], 50% of children with Down syndrome develop congenital heart diseases. Delany et al. [9] state that medical practitioners use surgery to correct an atrioventricular septal defect, a heart problem that interferes with normal blood flow in patients with Down syndrome because of a hole in the heart. Surgery helps physicians seal the hole in the heart and repair damaged valves to enhance normal blood flow. Thus, patients with Down syndrome must work with cardiologists to reduce their risks of atrioventricular septal defects and leaky valves.

## Prevention of Down Syndrome

### Health Education

Medical practitioners should increase health education about Down syndrome to reduce its prevalence and adverse complications. According to Ostermaier [10], medical practitioners should educate community members, especially parents, about Down syndrome's physical signs and symptoms among infants and adults. For example, they should inform community members that infants with Down syndrome have flattened faces, whereas adults have flattened heads in the back. The knowledge will help communities or parents to identify individuals with Down syndrome and take them for urgent medical care. Ostermaier [10] states that medical practitioners should inform community members about the adverse complications of Down syndrome in children and adults. Specifically, they should explain how unmanaged Down syndrome leads to heart defects, blood disorders, hormonal disorders, immune system problems, stomach and digestive system problems, and intellectual disability. Medical practitioners should explain to community members the long term implications of the complications on patients' lifestyle, physical, and emotional health. The information will influence community members to collaborate with healthcare workers in managing Down syndrome. Also, Ostermaier [10] suggest that medical practitioners should inform vulnerable persons about the risks of Down syndrome. Particularly, they should encourage pregnant women above 35 years to go for Down syndrome screening. Nurses should encourage couples with children with Down syndrome to go for genetic counselling before making pregnancy-related decisions.

Consequently, health education will increase the prevention and early interventions for patients with Down syndrome, reducing their risks of adverse complications.

### Compare and Contrast Themes and Gaps in Literature

The e-books and peer-reviewed journals included in this literature focus on the main subject of Down syndrome. The articles define Down syndrome, signs and symptoms, and management or treatment methods of the genetic complication. Mainly, Newton [1] and Presson et al. [2] provide detailed definitions of Down syndrome and signs and symptoms of the complication. The articles concur that Down syndrome occurs when individuals experience abnormal cell division, resulting in a partial or complete copy of chromosome 21. They add that individuals with Down syndrome have short necks, protruding tongues, poor muscle tones, flattened faces, and excessive flexibility.

Equally important, some of the peer-reviewed articles provide detailed information on the pharmacological and non-pharmacological methods for managing or preventing Down syndrome. Presson et al. [2] focus only on pharmacological strategies for managing Down syndrome's adverse signs or complications. The article indicates that medical practitioners utilize donepezil, rivastigmine, memantine, and galantamine to manage Down syndrome associated with dementia. Presson et al. [2] add that physicians prescribe traditional anticonvulsants for managing or preventing seizures among persons with Down syndrome. The peer-reviewed journal notes that clinicians use antibiotic therapies to control or prevent respiratory disorders among persons with Down syndrome. Although Regis et al. [5], Ruiz-González et al. [6], and Ciucurel and Iconaru [7] examine treatment methods, they differ from Presson et al. [2] as they look at non-pharmacological methods only. Ruiz-González et al. [6] discusses the effectiveness of physical therapy in improving the strength and movement of persons with Down syndrome. Regis et al. [5] examine the importance of speech therapy in enhancing the communication skills of individuals with Down syndrome. Ciucurel and Iconaru [7] analyze the benefits of occupational therapy in promoting the fine motor skills of individuals with Down syndrome. Therefore, the articles focus on the theme of Down syndrome treatment and management, but examine contrasting pharmacological and non-pharmacological methods.

It is undeniable that the articles have exhausted the theme of Down syndrome treatment and management, but they have significant gaps. The article by Presson et al. [2] provides the benefits of medication therapies and overlook the side effects of some of the evidence-based drugs. Presson et al. [2] fail to discuss the side effects of donepezil, rivastigmine, memantine, galantamine, antibiotics, antidepressants, and antipsychotic medications on patients with Down syndrome. For example, the side effects of antidepressants and antipsychotic drugs include weight gain, constipation, sleepiness, slowness, and akathisia. The peer-reviewed journal should highlight the side effects of pharmacological therapies to enable medical practitioners and patients to make rational decisions during care planning. Presson et al. [2] do not provide the recommended or accurate dosage of pharmacological therapies for adults and children with Down syndrome. Next, the peer-reviewed article by Delany et al. [9] has not discussed the specific surgery required for correcting the atrioventricular septal defect in patients with Down syndrome. The article has not highlighted the adverse complications related to the recommend surgical methods for managing Down syndrome in infants.

### Questions to Further Research

- Are health informatics, especially Electronic Health Records, effective in providing Down syndrome health education for high-risk populations?
- What are the evidence-based surgical methods for managing or addressing Down syndrome-related complications in children and adults?
- What are the accurate dosages of donepezil, rivastigmine, memantine, galantamine, antibiotics, antidepressants, and antipsychotic medications for children and adults with Down syndrome?

## Conclusion

In summary, Down syndrome occurs when individuals experience abnormal cell division, resulting in a partial or complete copy of chromosome 21. Persons with Down syndrome have short necks, protruding tongues, poor muscle tones, flattened faces, and excessive flexibility. The genetic disorder affects approximately 400,000 adults and children in the United States. Unmanaged Down syndrome leads to serious health complications like ear infections, eye diseases, heart defects, hearing loss, seizures, gastrointestinal disorders, obesity, and obstructive sleep apnea. Peer-reviewed articles recommend evidence-based pharmacological and non-pharmacological methods to reduce complications and the prevalence of Down syndrome among Americans. The articles reveal that medical practitioners need to donepezil, rivastigmine, memantine, and galantamine to manage Down syndrome associated with dementia. They add that non-pharmacological methods, including speech therapy, occupational therapy, physical therapy, assistive technologies, and surgeries, are beneficial in improving the lives and health of persons with Down syndrome. Finally, the peer-reviewed journals note that clinical facilities or medical practitioners should provide health education to promote early intervention and prevention of Down syndrome among vulnerable populations.

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