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Understanding Down syndrome: A comprehensive review of cognitive, and parental perspectives

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Abstract

Down syndrome (DS), also known as trisomy 21, is one of the most common chromosomal disorders, affecting individuals worldwide. This paper provides a comprehensive overview of Down syndrome, covering its genetic etiology, cognitive characteristics, and behavioural manifestations. This additional genetic material alters developmental trajectories and contributes to the phenotypic features associated with DS. Despite advances in genetic research, the precise mechanisms linking genotype to phenotype in DS remain complex and multifaceted.

Cognitive profiles of individuals with Down syndrome exhibit variability, but commonly include intellectual disabilities ranging from mild to moderate severity. Challenges in cognitive functioning impact language development, memory, and executive functioning abilities. Understanding the cognitive strengths and weaknesses of individuals with DS is crucial for tailoring educational and therapeutic interventions to support their unique needs.

This paper synthesizes current research findings to elucidate the multifaceted nature of Down syndrome, emphasizing the importance of interdisciplinary approaches in promoting the well-being and inclusion of individuals with DS. By fostering greater understanding and awareness of Down syndrome, society can advocate for equitable opportunities and support systems that empower individuals with DS to thrive and contribute meaningfully to their communities.

Keywords: Communities, emphasizing, Down syndrome

Introduction

Down syndrome first described by John Langdon Down in 1866, this condition is characterized by an extra copy of chromosome 21, leading to a range of physical, cognitive, and behavioral differences. Despite its prevalence, Down syndrome remains misunderstood by many, leading to stigma and misconceptions. This comprehensive review aims to shed light on Down syndrome from genetic, cognitive, and behavioral perspectives. By delving into the underlying genetic mechanisms, we can gain a deeper understanding of how this condition manifests and its implications for development. Furthermore, exploring the cognitive and behavioral aspects provides insights into the unique strengths and challenges faced by individuals with Down syndrome.

Throughout this review, we will examine current research findings, discuss theoretical frameworks, and explore practical implications for individuals with Down syndrome, their families, and society at large. By fostering a better understanding of this complex condition, we can promote inclusivity, support optimal development, and enhance quality of life for individuals with Down syndrome.

Literature review

The attitude of common people towards Down syndrome is a complex, multifaceted construct influenced by several factors, including personal experiences, education, media representation, and cultural beliefs, ranging from acceptance and inclusion to stigma and discrimination. Individual differences: People's personal experiences with individuals with Down syndrome (positive or negative) can significantly impact their overall attitude.

Knowledge and education: Lack of knowledge and understanding about Down syndrome can lead to misconceptions and stereotypes, contributing to negative attitudes. Conversely, accurate information and educational efforts can promote acceptance and inclusion.

- The indirect evidence for the hypothesis of Developmental Dyscalculia being a feature of Down syndrome lies in the pervasive nature of calculation difficulties for this population across the world and over decades of research in the field. Direct evidence to confirm the hypothesis is needed and may come from fMRI studies, particularly to examine if the IPS is affected. Alternatively, clinical assessment tools, such as task-based interviews need to be developed that are designed for learners with DS and explicitly probe areas of number development observing strategy use. With the recent advances in the medical practice, development of surgical techniques for the correction of congenital disabilities, and improvement in general care, there has been a tremendous increase in the survival of infants and life expectancy of patients with Down syndrome. A Birmingham (United Kingdom) study done almost 60 years ago showed that 45% of infants survived the first year of life, and only 40% would be alive at 5 years. A later study conducted about 50 years after that showed that 78% of patients with Down syndrome plus a congenital heart defect survived for 1 year, while the number went up to 96% in patients without the anomalies. This rise in the life expectancy of these patients should continue to rise significantly because of the developments in medical science. Healthcare facilities aim to provide proper and timely management to these patients and to help them to have a fulfilled and productive life.
- The lack of statistically significant differences in attitudes towards inclusion between early childhood and preschool education teachers and students, it is hypothesized that there may be a shared perception and understanding of the challenges and successes associated with the inclusion of children with Down syndrome in kindergartens. However, the study suggests a need for additional education in the formal and non-formal training of teachers to address gaps in knowledge and promote a more inclusive environment. Further research with a larger and more diverse sample, using a sensitive and comprehensive instrument, may provide deeper insights into the dynamics of attitudes towards inclusion in early childhood education. In the context of mainstream primary schools, it can be hypothesized that non-disabled children's attitudes towards their peers with Down Syndrome may be influenced more by age and gender, with older female participants exhibiting more positive sociability attitudes. The consistently negative attitudes towards inclusion, despite exposure to audiovisual material promoting inclusion, suggest a potential resistance or lack of immediate impact on overall attitudes. Further research is warranted to explore the underlying factors shaping non-disabled children's attitudes towards peers with Down Syndrome, with a focus on identifying effective strategies to promote inclusion in mainstream school environments.
- It is hypothesized that targeted training materials and curricula addressing inclusive education, workplace inclusion, and medical care for individuals with Down syndrome could lead to a more positive and uniform attitude across the medical profession. The identified factors, such as race, previous relationships with individuals with Down syndrome, specialization

(Especially in pediatrics), and practice setting, may serve as key influencers shaping physicians' attitudes. Further research and development of tailored interventions could enhance the understanding and acceptance of Down syndrome within the medical community. The over-inhibition hypothesis in Down syndrome (DS), as outlined in the abstract, suggests that an imbalance in excitatory and inhibitory mechanisms, particularly related to GABAergic neurotransmission, plays a key role in cognitive deficits associated with DS. It is hypothesized that targeting this over-inhibition through pharmacological interventions, such as GABAA antagonists and negative allosteric modulators of $\alpha 5$ -containing GABAA receptors, may hold therapeutic potential for reversing cognitive deficits in individuals with DS.

- Further research and clinical trials are needed to validate these findings and explore the translational applications of pharmacological reduction of inhibition in DS. Based on the findings that adolescents with Down syndrome in this study demonstrated limited knowledge and behaviors related to health aspects, it is hypothesized that there exists a need for targeted interventions and educational programs addressing hygiene, substance use, exercise, and healthy eating in this population. Implementing comprehensive support systems and educational strategies may bridge the knowledge gap, fostering better health-related practices and enhancing the preparation of individuals with DS for independent living. Further research and tailored interventions could contribute to improved health outcomes for this group. The "amplified developmental instability" hypothesis was proposed to explain two observations: 1) the commonalties in the phenotypes of different aneuploid states and 2) all of the phenotypic traits in DS are also seen in the general population, albeit at lower frequency, with less severity and usually only present as a single trait. The hypothesis is based on the idea that environmental variance is a measure of the degree of homeostasis, and that those traits which are highly variable in the normal population (due to a greater environmental influence) are the traits most frequently and severely disturbed in aneuploidy. Proponents of the "amplified developmental instability" hypothesis have measured various physical parameters such as palate length, palmer and angle, tooth size, tooth eruption rates and stature (reviewed in Shapiro, 1983) and found these to be more variable in DS compared to the normal population, and concluded that this is due to a greater disruption of homeostasis.

Methodology

1. Study Design

The research utilized a mixed-methods approach to investigate various aspects of Down syndrome, including genetic factors, cognitive functioning, and behavioral characteristics. This approach allowed for the integration of quantitative data analysis with qualitative insights from clinical observations and interviews.

2. Participants

Participants were recruited from diverse settings, including specialized clinics, educational institutions, and community organizations serving individuals with Down syndrome. The

sample comprised individuals of different ages, socioeconomic backgrounds, and functional abilities to ensure a representative cross-section of the Down syndrome population.

All demographics were included in the following questionnaire including men, women, young adults and old people.

Sample size of the survey is approx 180 participants.

3. Questionnaire

The questionnaire was based on four keywords mainly such as parenting, knowledge, attitude, and behaviour which is related mainly to our topic. A study on down syndrome. The questionnaire was prepared as such in a manner that was mainly emphasising regarding the topic and in the beginning of the questionnaire brief introduction was given regarding our topic so that the one who is responding has some basic knowledge about the topic.

Data Analysis

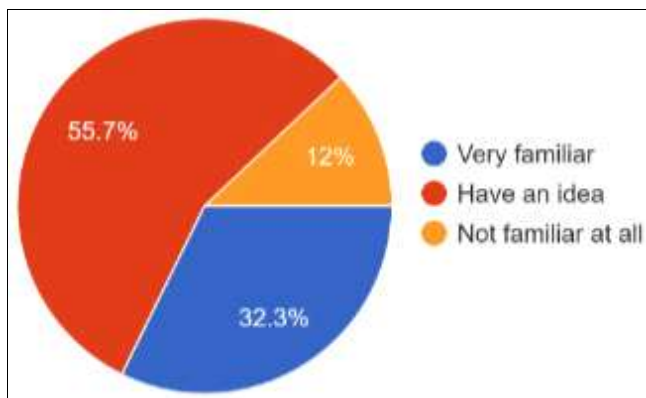


Fig 1: How familiar are you with the term down syndrome?

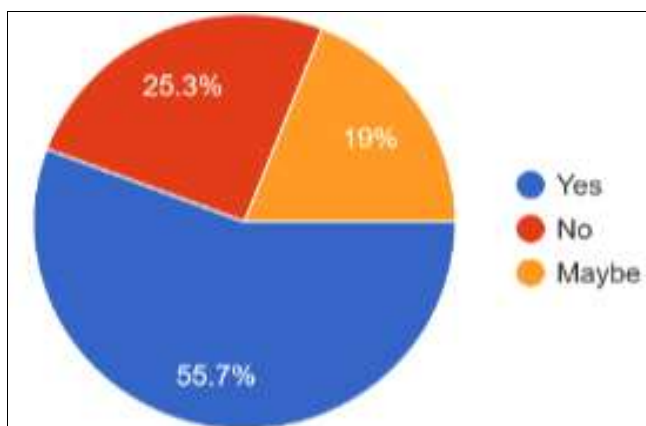


Fig 2: Are you aware of the common characteristics or physical features associated with down syndrome?

When we talk about the first question, which is how familiar are people with down syndrome so 55% of the people have an idea about it and 12% of them don't even know about it. A lot of people have an idea about it as they might have read it somewhere or they have seen people around them. Talk about these things. But around 32% of the people that responded to a questionnaire are very familiar about it. So they know what is down syndrome how it is caused and how you treat a person who has Down syndrome. The second question that we asked was related to the characteristics

associated with the syndrome. How people look like how did they talk? What is the reason that it is caused and what all things we have to keep in mind when spreading awareness about Down syndrome. Around 56% of the people have an idea about how a down syndrome person is treated and how do we spread awareness about it and around 25% of the people don't even know what are the characteristics associated with down syndrome as they are not aware about it and they might have not heard it anywhere.

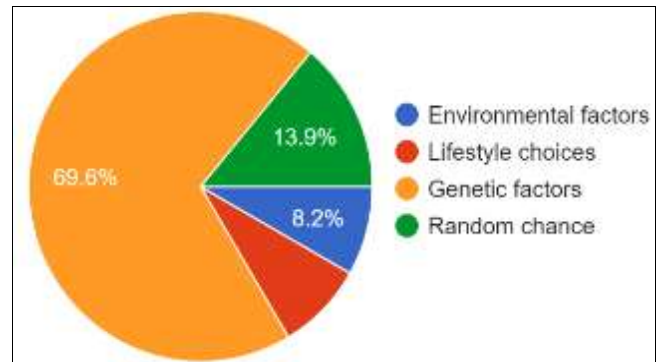


Fig 3: What is the primary cause of down syndrome?

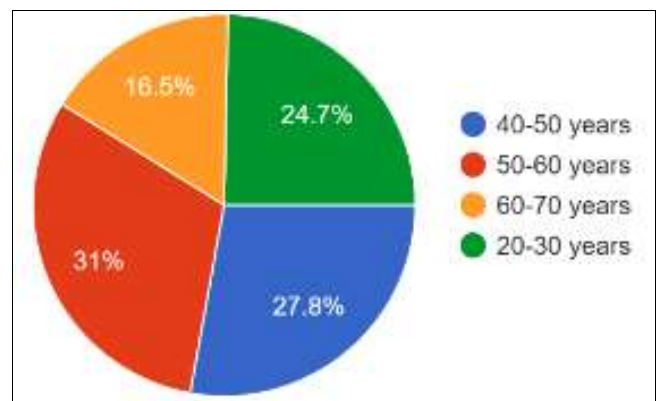


Fig 4: What is the life expectancy of individuals with down syndrome?

The pie chart illustrates students' responses regarding the primary causes of Down syndrome. A significant majority, 69.6%, attribute it to genetic factors, highlighting the prevailing understanding of its genetic basis. Environmental factors are cited by 8.2%, underscoring their perceived lesser role. Surprisingly, 13.9% attribute it to random chance. The remaining percentage reflects varied, unspecified causes. This underscores the need for further education on the complexities of Down syndrome etiology. The pie chart illustrates students' perceptions of the life expectancy of children with Down syndrome. While 24% believe it to be between 20-30 years, a significant portion (27.8%) anticipates a longer lifespan of 40-50 years. Surprisingly, 31% expect a life expectancy of 50-60 years, suggesting a more optimistic outlook compared to previous beliefs. The remaining respondents (Approx. 17.2%) anticipate even longer lifespans of 60-70 years, highlighting increasing awareness of improved healthcare and quality of life for individuals with Down syndrome.

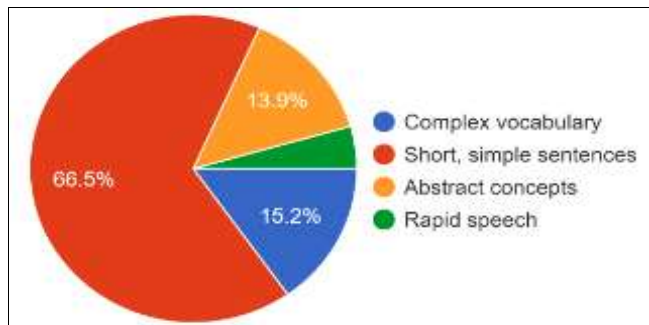


Fig 5: What is a key aspect of effective communication when parenting a child with down syndrome?

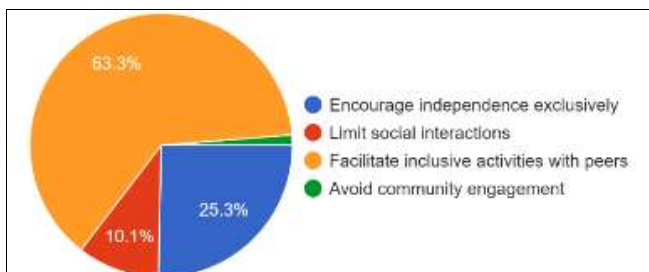


Fig 6: How can parents promote social inclusion for their child with down syndrome?

Most parents surveyed (66.5%) recognize that short, simple sentences are the most effective way to communicate with a child who has Down syndrome. Far fewer respondents chose complex vocabulary, abstract concepts, or rapid speech as key communication aspects.

Most parents (63.3%) understand the importance of facilitating inclusive activities with peers to promote their child's social inclusion. Encouraging independence exclusively was the second most popular answer (25.3%). It appears that a minority of parents may still believe it's best to limit social interactions or avoid community engagement for a child with Down syndrome.

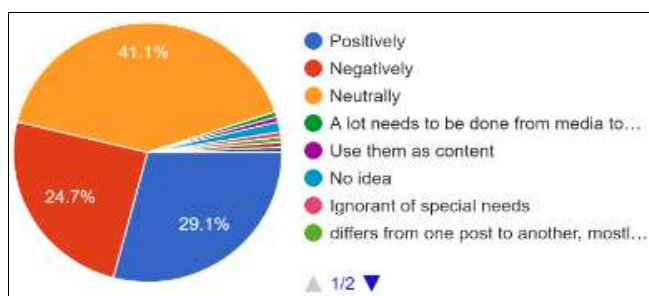


Fig 7: How do you think social media portrays individuals with down syndrome?

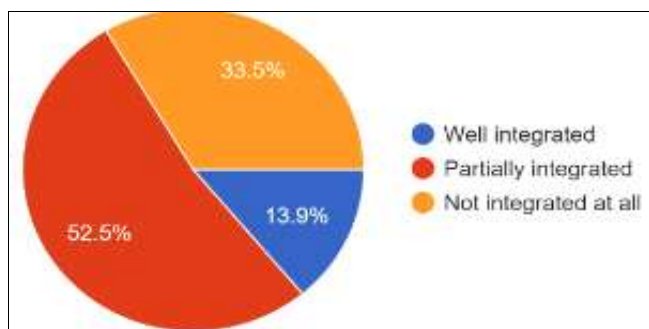


Fig 8: In your opinion, how well are individuals with down syndrome integrated with workforce?

The integration of individuals with Down syndrome into the workforce varies depending on factors like societal attitudes, accessibility, and support systems. Progress has been made in many places, with increasing awareness of the value of diversity and inclusion in the workplace, but there's still room for improvement in ensuring equal opportunities and support for all individuals.

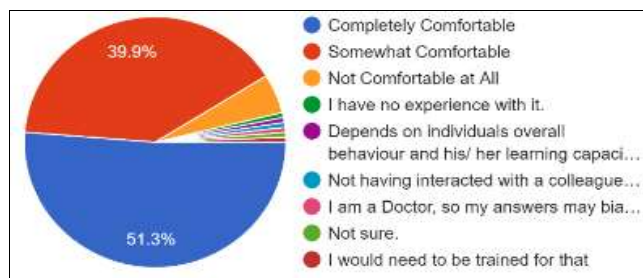


Fig 9: Would you feel comfortable working with a colleague who has down syndrome?

Working with someone who has Down syndrome can bring unique perspectives and strengths to the team. It's important to focus on individuals' capabilities and contributions rather than any perceived limitations. Building a supportive and inclusive workplace culture benefits everyone involved and fosters a more positive and productive environment.

A survey conducted among future healthcare providers to evaluate their knowledge and attitudes regarding Down syndrome (DS) revealed that 74.4% of the respondents reported positive knowledge of DS, with a median knowledge score of 14.0 (IQR 11.0–17.0). Additionally, 67.2% of the respondents had positive attitudes toward people with DS. Most students recognized that DS is a genetic disorder and is caused by an extra copy of chromosome 21. Furthermore, a significantly greater proportion of older respondents believed that people with DS should be integrated into society, had positive attitudes toward prenatal testing of DS, and agreed that treatment can improve the quality of life for individuals with DS. (Taibah 2023 Apr 7)

The study also found that medical students had significantly higher knowledge scores compared to students from other health science disciplines. The questionnaire used in the survey was field-tested and validated, and the study reported a Cronbach alpha reliability score of 0.807 for the knowledge scale and 0.842 for the attitude scale. (Akhtar; Rizwan, Bokhari)

In addition to the survey, it is important to note that individuals with Down syndrome typically present with mild to moderate intellectual disability, growth retardation, and characteristic facial features. They may also experience a wide range of congenital abnormalities, medical conditions, and dysmorphic features. Common physical signs of Down syndrome include a flattened face, upward slanting eyes, small ears, and poor muscle tone. Furthermore, individuals with Down syndrome usually have an IQ in the mildly-to-moderately low range and may experience both mental and physical challenges. (Oct 2023) This information provides valuable insights into the knowledge and attitudes of future healthcare providers regarding Down syndrome, which are important determinants of care, support, and inclusion for individuals with DS.

Discussions

Studies have shown that parents of individuals with Down syndrome in India face several challenges related to healthcare, including limited awareness, insufficient access to medical care, financial strain, stigma and prejudice, and varying degrees of government support. These challenges can lead to significant concerns and psychological impacts on parents, including high levels of distress, anxiety, and depression. Despite these challenges, initiatives such as early intervention programs, special care centres, and nonprofit-sponsored schools aim to increase access to quality education and employment opportunities for individuals with Down syndrome in India. Advocacy efforts and raising public awareness about Down syndrome are crucial steps in ensuring that individuals with Down syndrome receive the support and resources they require to succeed in education, employment, and healthcare.

The challenges and effects of behaviors on individuals with Down syndrome are multifaceted. Individuals with Down syndrome may experience a wide range of congenital abnormalities, medical conditions, and dysmorphic features, which can impact their behavior and mental health. Some of the commonly seen negative effects include irritability, agitation, aggressive behaviors, anxiety, and sleep-related problems. It is important to recognize that individuals with Down syndrome experience the full spectrum of emotions, including both positive and negative feelings. Additionally, at least half of all children and adults with Down syndrome will face a major mental health concern during their life, with common mental health concerns including anxiety, obsessive-compulsive behaviors, oppositional behaviors, depression, and tic disorder.

It is essential for caregivers, educators, and healthcare providers to have a comprehensive understanding of the behavioral challenges faced by individuals with Down syndrome. By being aware of these challenges, appropriate support and interventions can be implemented to promote the mental health and well-being of individuals with Down syndrome. This includes providing access to mental health resources, behavioral therapies, and creating inclusive environments that support their unique needs.

Overall, it is important to recognize the challenges faced by parents of individuals with Down syndrome in India and work towards creating a more inclusive and supportive society for individuals with Down syndrome and their families. (Zouine, Mehdaoui, Khabbache, atl July 18)

Conclusion

Parenting a child with Down syndrome requires patience, love, and understanding. Knowledge about the condition can empower parents to provide appropriate care and support, while maintaining a positive attitude can greatly enhance the child's development and overall well-being. It's essential for parents to educate themselves about Down syndrome, seek support from healthcare professionals and support groups, and approach their child's unique needs with empathy and acceptance.

Certainly! Parents who are well-informed about Down syndrome can better advocate for their child's needs, navigate healthcare systems, and access early intervention services. A positive attitude towards parenting a child with Down syndrome can foster a nurturing environment where the child feels valued and supported, leading to improved outcomes in areas such as cognitive development, social

skills, and emotional well-being. Embracing the journey with an open heart and a willingness to learn can enrich both the parent-child relationship and the family's overall experience.

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