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Advancements and Interventions in Down Syndrome: A Comprehensive Review of Current Research and Clinical Endeavors

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Abstract

Down syndrome, a chromosomal disorder characterized by an extra copy of chromosome 21, presents multifaceted challenges across cognitive, health, and social domains. This review synthesizes current research and interventions in Down syndrome, encompassing diverse areas. Cognitive interventions focus on educational approaches and behavioral therapies tailored to enhance learning and memory. Drug therapies aim to address cognitive function and associated health conditions like Alzheimer's disease. Genetic and molecular research explores targeted treatments by deciphering underlying genetic mechanisms. Health interventions aim to improve healthcare access and manage medical complications linked to Down syndrome. Behavioral and social interventions aim to enhance social skills and overall quality of life. Clinical trials span these areas, investigating cognitive, health, and genetic aspects, albeit with considerations of risks and informed consent. This review offers a comprehensive overview of ongoing efforts to improve outcomes and interventions for individuals with Down syndrome.

Keywords: Down Syndrome; Trisomy 21; Mosaicism; Translocation & Dr. John Langdon Down

Abbreviations: IEPs: Individualized Education Plans; DSFI: Down Syndrome Federation of India; ESDM: Early Start Denver Model

Introduction

Down syndrome is a genetic condition characterized by the presence of an additional or extra copy of chromosome 21 in an individual. This surplus genetic material results in various mental and physical health challenges. Individuals with Down syndrome often exhibit distinct physical features such as a flattened facial structure, almond-shaped eyes, and a shorter neck. The condition is also referred to as Trisomy 21 and has been historically known by names like Mongolism or Down's syndrome, named after Dr. John Langdon Down, who first detailed its characteristics in 1866. Dr. Down's efforts in describing and identifying Down syndrome marked a pivotal moment in recognizing and advocating for individuals with mental disabilities. His work not only highlighted the unique features but also emphasized the importance of care facilities and educational opportunities to support those affected [1]. Later research by Jerome Lejeune in 1958 established that an additional chromosome 21, known as Trisomy 21, causes Down syndrome. Lejeune's groundbreaking work linked chromosomal abnormalities to developmental disorders, influencing the scientific community's understanding of genetic conditions. He also played a role in ethical discussions, including those concerning abortion following the detection of chromosomal abnormalities.

Types of Down Syndrome

Three primary types of Down syndrome exist:

i. Trisomy 21, where there's an extra copy of chromosome 21 in every cell (the most common type).

ii. Mosaicism occurs when some but not all cells have an extra chromosome.

iii. Translocation, involving an extra part of chromosome 21 attaching to another chromosome.

Down syndrome's occurrence increases with maternal age, with varying frequencies across different populations (ranging from 1 in 319 to 1 in 1000 live births). Genetic information, typically inherited from both parents' 46 chromosomes, determines an individual's traits. In Down syndrome, the presence of an extra or attached chromosome 21 leads to characteristic physical features and developmental delays [2]. Children with Down syndrome commonly display physical traits like a flat facial profile, upward-slanting eyes, small ears, and slower developmental milestones such as sitting, crawling, and walking. Low muscle tone, smaller size at birth, and delays in speech and self-care skills are also typical. While intellectual disability ranges from mild to moderate, children with Down syndrome learn and develop skills at their own pace, requiring individualized support and avoiding comparisons to typical peers [3] (Figure 1).

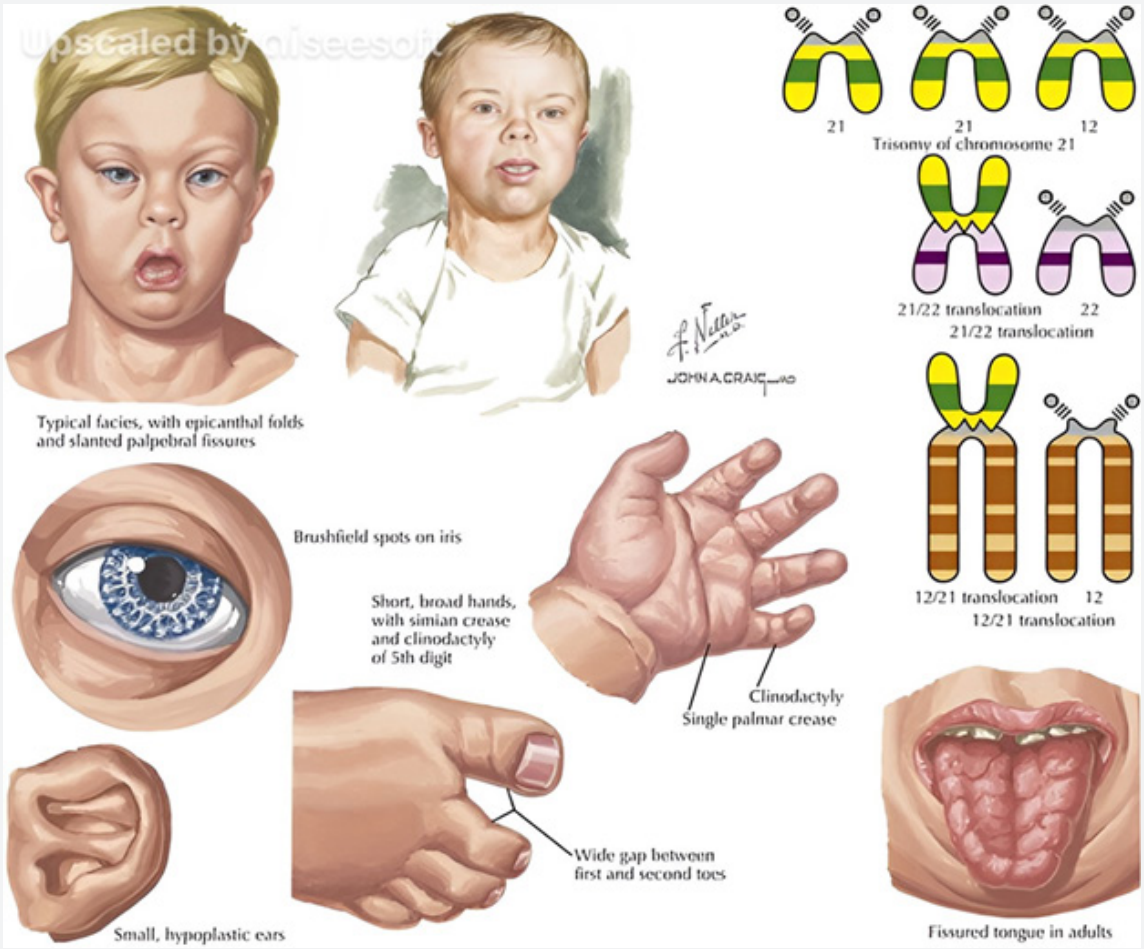

Figure 1: Down Syndrome (Image taken from: https://www.labtestsguide.com/down-syndrome).

Prevalence and Statistics of Down Syndrome in India

In India, Down syndrome prevalence varies across different regions and populations. However, statistical data on Down syndrome specifically in India can be challenging to pinpoint due to factors like underreporting, varying healthcare access, and limited comprehensive nationwide studies. Estimates suggest that the prevalence of Down syndrome in India might be around 1 in 800 to 1 in 1000 live births [4]. However, these figures are approximate and might not represent the complete picture due to the lack of extensive, nationwide data collection. Factors influencing the prevalence of Down syndrome in India include the population's size, maternal age (as the risk increases with older mothers), and regional variations in healthcare quality and access to prenatal screening and diagnostic services. Despite the absence of precise statistical figures, efforts have been made in India to enhance awareness, education, and support for individuals and families affected by Down syndrome. Organizations and healthcare initiatives strive to provide better healthcare access, early intervention programs, and educational opportunities for those with Down syndrome, contributing to improved quality of life and greater inclusion within society [5,6].

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Causes and Diagnosis of Down Syndrome

Causes

Down syndrome is a genetic disorder caused by an extra copy of chromosome 21, which results from an error in cell division called nondisjunction. This error can occur randomly during the formation of an egg or sperm, and no behavioral activity of the parents or environmental factor is known to cause Down syndrome [7].

Complications

The condition causes physical and mental developmental delays and disabilities, including mild to moderate developmental disability, impulsive behavior, poor judgment, short attention span, slow learning capabilities, and lifelong intellectual disability. People with Down syndrome may also have medical complications, such as congenital heart defects, hearing loss, cataracts, leukaemia, chronic constipation, sleep Apena, dementia, hypothyroidism, obesity, late tooth growth, and increased risk of infection [8].

Diagnosis

Diagnosis of Down syndrome can occur during pregnancy or after birth.

Prenatal Diagnosis:

i. Prenatal Screening Tests: Non-invasive tests like ultrasound and maternal blood screenings can indicate an increased likelihood of Down syndrome. These screenings do not provide a definitive diagnosis but identify individuals who might benefit from further diagnostic testing.

ii. Prenatal Diagnostic Tests: More definitive tests such as amniocentesis or chorionic villus sampling (CVS) involve obtaining samples of amniotic fluid or placental tissue for genetic analysis. These tests can confirm the presence of an extra chromosome 21 [9,10].

Postnatal Diagnosis

i. Physical Examination: After birth, doctors might identify physical features associated with Down syndrome, such as distinctive facial characteristics and low muscle tone.

ii. Genetic Testing: A blood test called karyotyping can confirm the diagnosis by examining the individual's chromosomes for the presence of an extra chromosome 21 [11].

While prenatal testing allows for early identification, it's important to note that these tests come with their own set of considerations, including the potential risks associated with invasive procedures like amniocentesis or CVS, as well as the emotional impact of receiving a diagnosis during pregnancy. Early diagnosis of Down syndrome is crucial for implementing appropriate medical care, interventions, and supportive strategies to enhance the individual's quality of life and development. This includes access to specialized healthcare, early intervention programs, educational support, and resources that cater to the specific needs of individuals with Down syndrome [12].

Medical and Developmental Characteristics of Down Syndrome

Down syndrome is characterized by a distinct set of medical and developmental features caused by the presence of an additional chromosome 21. These characteristics vary widely among individuals but commonly include [13]:

Physical Features: Individuals with Down syndrome often exhibit facial characteristics such as a flattened facial profile, upward-slanting eyes with epicanthic folds, a smaller nose, a protruding tongue, smaller and rounded ears, and a single crease across the palm (simian crease). They may also have shorter stature and lower muscle tone, contributing to a more relaxed posture [14].

Health Issues: There's an increased likelihood of certain health conditions among individuals with Down syndrome, including congenital heart defects, gastrointestinal issues like esophageal reflux or celiac disease, respiratory problems, hearing or vision impairments, thyroid issues, and a higher susceptibility to infections.

Intellectual and Developmental Disabilities: Most individuals with Down syndrome experience varying degrees of intellectual disability, ranging from mild to moderate. This affects cognitive abilities, learning, and language development. They may reach developmental milestones like walking or talking later than their peers [15].

Delayed Development: Children with Down syndrome often have delayed motor skills development, including crawling, walking, and fine motor skills like grasping objects or feeding themselves. They might also have delayed speech and language development.

Social and Behavioral Traits: People with Down syndrome typically exhibit warm and engaging personalities. They might face challenges in social interactions, including difficulty with social cues and communication, but often have a friendly and affectionate demeanor.

Potential Medical Conditions: Beyond the common health issues, individuals with Down syndrome have a higher risk of certain conditions later in life, such as early-onset Alzheimer's disease, obesity, and musculoskeletal problems like joint issues [16,17].

Physical Features and Health Issues Associated with Down Syndrome

Physical Features:

Facial Characteristics: Upward-slanting eyes, small or slanted palpebral fissures, epicanthic folds (skin folds on the inner

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corners of the eyes), flattened facial profile, a small nose with a low bridge, and a protruding tongue.

Ears: Smaller and sometimes shaped differently, with a lower placement [20].

Hands and Feet: A single crease across the palm (simian crease), shorter fingers, and a wider gap between the big toe and the second toe.

Muscle Tone: Low muscle tone (hypotonia), contributing to a more relaxed posture and delayed motor skills development [21].

Health Issues

Congenital Heart Defects: Approximately half of individuals with Down syndrome have heart defects, which might require surgical intervention.

Gastrointestinal Problems: Conditions like esophageal reflux, celiac disease, and constipation are more prevalent.

Respiratory Issues: Increased susceptibility to respiratory infections and conditions like sleep apnea.

Hearing and Vision Problems: Higher incidence of hearing loss, refractive errors, and conditions like cataracts or strabismus.

Thyroid Dysfunction: Greater likelihood of thyroid disorders, including hypothyroidism.

Obesity: There's an increased risk of obesity due to lower metabolic rates and sometimes dietary habits.

Musculoskeletal Problems: Joint problems, such as atlantoaxial instability, which requires careful monitoring in certain activities.

Increased Risk of Infections: Higher susceptibility to infections, especially respiratory infections [22,23] (Figure 2).

Figure 2: Physical characteristics of Down Syndrome. (Image taken from: https://www.medicoverhospitals.in/diseases/down-syndrome/).

Intellectual and Developmental Disabilities

Intellectual and developmental disabilities are common traits associated with Down syndrome. These characteristics encompass a range of cognitive, learning, and developmental challenges that individuals with Down syndrome may experience:

Intellectual Disability: Most individuals with Down syndrome have some level of intellectual disability, which can range from mild to moderate. This impacts their cognitive abilities,

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including reasoning, problem-solving, and learning. It might affect their ability to process information and learn at a slower pace compared to their peers.

Language and Communication: Speech and language development can be delayed or affected in varying degrees. Some individuals with Down syndrome might have difficulty articulating words, forming sentences, or understanding complex language structures. However, with early intervention and support, many individuals can develop effective communication skills using speech, sign language, or augmentative communication devices [24].

Motor Skills Development: Low muscle tone (hypotonia) can impact gross and fine motor skills development. This might result in delays in sitting up, crawling, walking, and fine motor tasks like holding a pencil or using utensils. Occupational and physical therapies are often utilized to assist in developing these skills.

Learning Differences: Individuals with Down syndrome often have different learning styles and strengths. They may benefit from visual learning techniques, hands-on activities, and repetitive practice to acquire and retain new skills. Tailored educational programs that accommodate these learning styles can significantly support their progress.

Memory and Attention: Some individuals with Down syndrome might have difficulties with short-term memory and attention span. Strategies involving repetition, visual aids, and structured routines can aid in memory retention and focus [25].

Challenges and Barriers Faced by Individuals with Down Syndrome in Accessing Education

Individuals with Down syndrome often encounter various challenges and barriers when accessing education. Some of these include:

Limited Accessibility to Inclusive Education: Despite efforts toward inclusive education, many schools and educational institutions lack the resources, training, or accommodations necessary to support the diverse learning needs of students with Down syndrome [26].

Lack of Specialized Support: Often, there's a scarcity of specialized educators or support staff trained in addressing the specific learning styles and needs of individuals with Down syndrome. This absence can hinder the implementation of effective teaching methods and tailored learning plans [27].

Social Stigma and Discrimination: Attitudinal barriers and societal misconceptions about intellectual disabilities can lead to stigma, exclusion, and discrimination, impacting the social integration and acceptance of students with Down syndrome within educational settings [28].

Inadequate Individualized Education Plans (IEPs): Developing comprehensive and effective Individualized Education Plans that cater to the unique strengths and challenges of each student with Down syndrome can be a challenge. Insufficient IEPs might not adequately address their learning needs [29].

Limited Access to Assistive Technologies: Access to assistive technologies and supportive resources that facilitate learning, such as speech and communication devices or specialized educational tools, may be limited in some educational settings [30].

Financial Constraints: Families might face financial barriers in accessing specialized education or services for their children with Down syndrome, particularly in regions with limited or no financial support for special education [31].

Transition and Post-School Opportunities: Planning for transition services and post-school opportunities, such as vocational training, employment, or independent living, might not be adequately addressed, limiting prospects for individuals with Down syndrome [32].

Unequal Educational Opportunities: Disparities in educational opportunities exist based on geographical location, socio-economic status, or access to resources, resulting in unequal access to quality education for individuals with Down syndrome [33].

Successful Inclusion Practices and Interventions

Successful inclusion practices and interventions for individuals with Down syndrome involve various strategies aimed at creating supportive and inclusive environments [34]. Some effective practices include:

Individualized Education Plans (IEPs): Developing comprehensive IEPs tailored to the specific strengths, learning styles, and needs of each student with Down syndrome. These plans outline personalized goals, accommodations, and strategies to support their academic, social, and developmental progress [35].

Inclusive Education Models: Implementing inclusive education models that promote the integration of students with Down syndrome into mainstream classrooms alongside their typically developing peers. This encourages social interaction, peer support, and positive relationships while fostering a sense of belonging.

Specialized Instruction and Support: Providing specialized instruction by trained educators familiar with strategies and techniques to address the learning needs of individuals with Down syndrome. Utilizing differentiated instruction, visual aids, hands-on activities, and assistive technologies can enhance learning outcomes [36].

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Collaboration and Professional Development: Encouraging collaboration among teachers, specialists, therapists, and parents to create a cohesive support system. Offering ongoing professional development and training for educators to enhance their understanding of Down syndrome and effective teaching methods is crucial.

Peer Support and Mentoring Programs: Establishing peer support and mentoring programs within schools to promote social interaction, peer learning, and positive relationships between students with Down syndrome and their peers. This fosters a sense of inclusion and understanding among all students [37].

Adaptive and Assistive Technologies: Providing access to adaptive technologies, such as speech and communication devices, educational apps, and assistive tools, to support learning and communication skills development [38].

Early Intervention Programs: Offering early intervention services, including speech therapy, occupational therapy, and developmental programs, to address developmental delays and provide crucial support during the formative years.

Community Engagement and Awareness: Encouraging community involvement and awareness through activities, events, and workshops that promote understanding, acceptance, and appreciation of individuals with Down syndrome. This reduces stigma and fosters a supportive environment [39].

Transition Planning and Post-School Opportunities: Implementing transition planning services that prepare individuals with Down syndrome for life after school, including vocational training, employment support, independent living skills, and community integration programs [40].

Family Involvement and Support: Involving families in the educational process, providing guidance, resources, and emotional support, and fostering collaboration between home and school environments [41].

Support Services and Resources for Individuals with Down Syndrome and Their Families in India

In India, several support services and resources are available to assist individuals with Down syndrome and their families. These resources aim to provide guidance, education, and support for various aspects of their lives. Some of these services include:

Down Syndrome Federations and Associations: Organizations like Down Syndrome Federation of India (DSFI) and Down Syndrome Support Groups (local or regional) offer information, advocacy, and support networks for individuals with Down syndrome and their families. They often provide guidance on education, healthcare, and community integration.

Special Education and Early Intervention Programs: Special schools, early intervention centers, and programs tailored for children with Down syndrome offer educational support, therapies, and developmental interventions to address specific learning needs and developmental delays.

Therapeutic Services: Access to speech therapy, occupational therapy, and physical therapy services to support speech, motor skills development, and overall well-being of individuals with Down syndrome.

Parent Support Groups: Support groups specifically for parents and caregivers of individuals with Down syndrome offer a platform to share experiences, information, and emotional support. These groups provide a sense of community and valuable resources for families.

Medical and Healthcare Facilities: Hospitals, clinics, and healthcare professionals specializing in Down syndrome offer medical care, screenings, and treatments for associated health conditions.

Awareness and Training Programs: Workshops, seminars, and training sessions conducted by various organizations aim to raise awareness, educate families and professionals, and provide guidance on supporting individuals with Down syndrome.

Online Resources and Information: Websites, forums, and online resources by organizations and support groups offer information, resources, and guidance on various aspects of Down syndrome, including education, healthcare, and advocacy.

Government Initiatives and Policies: Government programs, schemes, and policies for individuals with disabilities might offer financial support, accessibility provisions, and educational assistance. Local government bodies or disability departments might offer relevant services.

Employment and Vocational Training: Some organizations or vocational training centers provide skill development, job training, and employment support for individuals with Down syndrome to facilitate their integration into the workforce.

Negative Perceptions and Stereotypes

In India, negative perceptions and stereotypes about Down syndrome contribute to societal stigma. Misconceptions about intellectual disabilities often lead to underestimating the capabilities and potential of individuals with Down syndrome. These stereotypes perpetuate a lack of understanding about their abilities, resulting in marginalization and exclusion from various aspects of society.

Impact on the Lives of Individuals and Families

The social stigma and discrimination against individuals with Down syndrome have significant repercussions. Limited opportunities for education, employment, healthcare, and social engagement hinder their full participation in society. Such exclusion not only affects the self-esteem and sense of belonging for those with Down syndrome but also places added emotional and practical burdens on their families, who often face societal

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judgment and barriers in providing support.

Efforts to Promote Awareness and Acceptance

Efforts to combat stigma and promote acceptance involve multifaceted approaches. Awareness campaigns, educational initiatives, and advocacy work seek to dispel myths and increase understanding about Down syndrome. Inclusive practices in schools, workplaces, and communities are encouraged to foster environments that embrace diversity and value the contributions of individuals with Down syndrome. Empowerment programs, community engagement events, and support networks aim to showcase their talents, break stereotypes, and create a more inclusive society that respects the rights and dignity of individuals with Down syndrome in India.

Recommendations and Future aspects

Recommendations for Improving Quality of Life:

Enhanced Access to Education: Ensure inclusive education practices, specialized training for educators, and tailored curricula that cater to diverse learning needs.

Healthcare Accessibility: Improve access to specialized healthcare services, including early intervention programs, therapies, and regular health check-ups.

Vocational Training and Employment Opportunities: Develop vocational training programs and promote inclusive employment practices to facilitate meaningful employment opportunities for individuals with Down syndrome.

Community Support and Inclusion: Encourage community involvement, awareness campaigns, and support networks to foster social integration and inclusion.

Legal and Policy Frameworks: Advocate for policies that protect the rights and promote the inclusion of individuals with Down syndrome, ensuring equal opportunities and access to resources.

Future Research Directions and areas of Focus:

Healthcare Interventions: Research into more effective healthcare interventions and treatments for associated medical conditions, especially focusing on heart defects, cognitive development, and aging-related issues like Alzheimer's disease.

Educational Strategies: Further exploration of innovative educational approaches and technologies that cater to the diverse learning styles and needs of individuals with Down syndrome.

Social Inclusion and Well-being: Research on the impact of inclusive practices on the social integration, well-being, and mental health of individuals with Down syndrome.

Transition and Post-School Life: Studies focusing on transition planning, vocational training, employment outcomes, and independent living skills for individuals with Down syndrome.

Genomic Research: Continued genetic research to understand the mechanisms underlying Down syndrome, potential therapeutic interventions, and identifying factors influencing variability in cognitive abilities.

Long-Term Care and Aging: Research addressing the specific needs and challenges faced by individuals with Down syndrome as they age, including healthcare, support services, and quality of life in later stages. By prioritizing these recommendations and research areas, India can take significant strides in improving the lives of individuals with Down syndrome, fostering a more inclusive and supportive environment that enables them to thrive and contribute meaningfully to society [42].

Current Research

Current research in Down syndrome encompasses diverse areas. Studies investigate cognitive development, exploring novel educational approaches like the Early Start Denver Model (ESDM), showing promising results in enhancing language and cognitive skills. Medical research focuses on heart defects, with approximately 50% of individuals with Down syndrome having congenital heart issues; ongoing studies aim to improve surgical outcomes and management. Genomic studies continue, such as investigating the role of the DYRK1A gene in cognitive function, showcasing potential for targeted therapies. Social inclusion research emphasizes the impact of inclusive practices; however, only around 10% of adults with Down syndrome are employed, highlighting the need for greater employment inclusion efforts. Transition planning remains a key area, considering that about 20% of the population with Down syndrome over 35 years has early-onset Alzheimer's disease; research aims to improve aging support and long-term care. These diverse research pursuits contribute to an evolving understanding and betterment of the lives of the approximately 1 in 800 to 1 in 1000 live births affected by Down syndrome in India, though comprehensive data can be challenging to ascertain due to varied healthcare access and underreporting [43].

Novel FDA Approved Drug for Down Syndrome

There are currently no specific FDA-approved drugs for treating Down syndrome itself. However, research and clinical trials have been ongoing to explore medications targeting various aspects or associated conditions related to Down syndrome. These studies aim to address specific health issues or cognitive challenges individuals with Down syndrome may face, such as cognitive enhancement, neurological development, and associated health conditions like Alzheimer's disease. Some recent advances in Down syndrome research include:

i. A phase 1b clinical trial conducted by the Spanish Hospital del Mar Medical Research Institute to test the safety and efficacy of a new treatment to improve cognitive function in people with Down syndrome.

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ii. A study on the use of GnRH replacement therapy drug Lutrelef, which demonstrated improved cognitive performance in patients.

iii. The ELND005 drug trial, which aimed to prevent the accumulation of plaques that might contribute to Alzheimer's disease and improve working memory and cognitive functioning by regulating myo-inositol levels in the brain.

iv. The development of model trisomic cell lines, which could help advance understanding and potential therapies for Down syndrome.

These research efforts contribute to an evolving understanding of Down syndrome and the development of targeted therapies to address specific health issues and cognitive challenges faced by individuals with Down syndrome [44,45].

Ongoing Clinical Trials

The current research and clinical trials in Down syndrome encompass various areas, including cognitive development, potential treatments, and interventions. Some of the recent advancements and ongoing studies include:

i. **New Drug Trials:** A phase 1b clinical trial conducted by the Spanish Hospital del Mar Medical Research Institute to test the safety and efficacy of a new treatment to improve cognitive function in people with Down syndrome.

ii. **GnRH Replacement Therapy:** Trials were conducted on patients with Down syndrome, where the GnRH replacement therapy drug Lutrelef demonstrated improved cognitive performance in patients.

iii. **ELND005 Drug Trial:** This trial aimed to prevent the accumulation of plaques that might contribute to Alzheimer's disease and improve working memory and cognitive functioning in individuals with Down syndrome.

iv. **Model Trisomic Cell Lines:** The development of model trisomic cell lines are being explored to advance understanding and potential therapies for Down syndrome.

v. **Early Intervention Services:** Early intervention services, including occupational therapy, physical therapy, and speech therapy, are recommended for children with Down syndrome.

vi. **Assistive Devices and Therapies:** Interventions for children with Down syndrome involve assistive devices and specialized education services to enhance learning and development.

vii. **Future Therapies and Research Directions:** Research is focused on identifying genetic, neurological, and psychosocial factors, as well as potential pharmacological interventions to modify disease progression and improve cognitive function in individuals with Down syndrome.

These diverse research pursuits and clinical trials contribute to an evolving understanding of Down syndrome and the development of targeted therapies to address specific health issues and cognitive challenges faced by individuals with Down syndrome [46,47].

Conclusion

The comprehensive review underscores the multidimensional nature of Down syndrome, highlighting ongoing research and interventions across cognitive, health, and social spheres. Current endeavors encompass cognitive interventions tailored for learning enhancement, drug therapies targeting cognitive function and associated health conditions, genetic and molecular research seeking targeted treatments, and diverse health and social interventions. Clinical trials exploring these areas underscore the evolving landscape in Down syndrome research. However, while these initiatives show promise, careful considerations regarding risks, benefits, and informed consent remain imperative. The synthesis of these efforts signifies a concerted push toward enhancing outcomes and interventions for individuals with Down syndrome, offering a promising trajectory for improved quality of life and holistic support.

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References

1. Antonarakis SE, Skotko BG, Rafii MS, Strydom A, Pape SE, et al. (2020) Down syndrome. Nat Rev Dis Primers 6(1): 9.

2. Annerén G, Edman BB (1993) Down syndrome-a gene dosage disease caused by trisomy of genes within a small segment of the long arm of chromosome 21, exemplified by the study of effects from the superoxide-dismutase type 1 (SOD-1) gene. APMIS Suppl 40: 71-79.

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3. Roizen NJ, Patterson D (2003) Down's syndrome. Lancet 361(9365): 1281-1289.
4. Manikandan K, Seshadri S (2017) Down syndrome screening in India: are we there yet? J Obstet Gynaecol India 67(6): 393-399.
5. Lakhan R, Kishore MT (2016) Down syndrome in tribal population in India: A field observation. J Neurosci Rural Pract 7(1): 40-43.
6. Pankaj G, Avani K, Salil V (2014) Prevalence of Down syndrome in Western India: A cytogenetic study. Br J Med Med Res 5(10): 1255-1259.
7. (2011) National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, US.
8. What causes Down syndrome? (2023) US Department of Health and Human Services.
9. Prenatal Testing for Down Syndrome. UCSF Health.
10. Hasina Z, Wang CC (2022) Prenatal and postnatal therapies for Down's syndrome and associated developmental anomalies and degenerative deficits: A systematic review of guidelines and trials. Front Med (Lausanne) 9: 910424.
11. https://ndss.org/lifespan/understanding-a-diagnosis-of-down-syndrome.
12. Skotko BG, Capone GT, Kishnani PS (2009) Down Syndrome Diagnosis Study Group. Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news. Pediatrics 124(4): e751-758.
13. Akhtar F, Bokhari SRA (2023) Down syndrome e. StatPearls.
14. Mégarbané A, Ravel A, Mircher C, Sturtz F, Grattau Y, et al. (2009) The 50th anniversary of the discovery of trisomy 21: the past, present, and future of research and treatment of Down syndrome. Genet Med 11(9): 611-616.
15. What conditions or disorders are commonly associated with Down syndrome? (2023) US Department of Health and Human Services.
16. Down Syndrome (2023) Cleveland Clinic.
17. Down syndrome (2018) Diseases & Conditions. Mayo Clinic.
18. Down Syndrome (2023) Children's Health Guide.
19. Kathleen F (2022) What Are the Features of Down Syndrome? The Impact of Down Syndrome on a Person's Life is Variable. Very well Health.
20. Down Syndrome! It's high time to create a loud shout in India (2021) Health world.
21. Pankaj G, Kathiriya A, Vaniawala S (2014) Prevalence of Down Syndrome in Western India: A Cytogenetic Study. J Adv Med Res 5(10): 1255-1259.
22. Down Syndrome still fatal in India (2013) Deccan Hearald.
23. Over 30,000 Down's Syndrome children born in India a year (2018) Health Issues India.
24. Nina Powell-Hamilton N (2023) Down Syndrome (Trisomy 21). Chromosome and Gene Abnormalities.
25. Priyanka P, Rakesh Kumar V, Navneet K, Sciddhartha K (2018) Down syndrome: a cytogenetic study in North Indian population. Bio Med Res 29(19).
26. Laura B, Stephanie H, Rebecca B, Sarah H, Kelly B (2023) Views of educators working with pupils with Down syndrome on their roles and responsibilities and factors related to successful inclusion. Res Dev Disabil 142: 104617.
27. Wolpert G (2001) Successful Daily Practices of Inclusion Teachers of Children with Down Syndrome. Eric Rep Res p. 11.
28. Inclusive Education Guidelines. National Down syndrome society.
29. Julie H (2006) Inclusive education for individuals with Down syndrome. Down Syndrome Educational Trust UK 6(1): 1-3.
30. Inclusion. Down syndrome Resource Foundation.
31. How to Teach Students with Down Syndrome: 15 Effective Strategies (2023) Positive Action.
32. Heather White (2022) 6 Ways to Support Children with Down Syndrome in the Montessori Classroom. American Montessori Society.
33. Down Syndrome. Amrit Foundation of India.
34. Team Acko (2023) Health Insurance for a Down Syndrome Child in India. Child Health Insurance Plans.
35. Down syndrome about Family. Down syndrome Federation of India.
36. Down syndrome About DSFI. Down syndrome Federation of India.
37. https://www.ds-int.org/legal-capacity-in-india.
38. http://motherandchildschool.com/downsyndromeindia.html.
39. Down Syndrome. Developmental Disabilities. Uttarakhand, India.
40. Inclusive Education Guidelines. National Down syndrome society.
41. Roohi Mariam P (2023) 5 advancements in Down syndrome research over the past year. LABIOTECH.
42. Jonathan DS, Rebecca P, Runi T, Dania P, Mellad K, et al. (2022) Evidence of neuroinflammation and immunotherapy responsiveness in individuals with down syndrome regression disorder. J Neurodev Disorder 14(35).
43. Down Syndrome Program: The ELND005 Drug Trial Is a Success! Mass General for Children.
44. Wiseman FK, Alford KA, Tybulewicz VL, Fisher EM (2009) Down syndrome-recent progress and future prospects. Hum Mol Genet 18(R1): R75-R83.
45. Moria L (2022) Down Syndrome: Treatments, Therapies, Services. Everyday Health.
46. Dierssen M, Herault Y, Helguera P, Martínez de Lagran M, Vazquez A, et al. (2021) Building the future therapies for Down syndrome: the Third International Conference of the T21 Research Society. Mol Syndromol Third International Conference of the T21 Research Society. Mol Syndromol 12(4): 202-218.

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