



Understanding Down Syndrome: Global Perspectives, Advances, and Challenges in the Context of Bangladesh

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Abstract: Down syndrome (DS), or Trisomy 21, is a genetic disorder characterized by intellectual disabilities, developmental delays, and various health complications. This comprehensive review examines the causes, symptoms, and management strategies of DS, emphasizing global research advancements and their relevance to Bangladesh. This article highlights emerging therapies, genetic studies, and social advocacy efforts, proposing strategies to bridge the gap in healthcare and research. It aims to inspire collaborative efforts to improve the quality of life for individuals with DS globally and within Bangladesh. Down Syndrome (DS), or Trisomy 21, is a genetic disorder characterized by developmental delays, intellectual disabilities, and a variety of health complications. This review explores the causes, symptoms, and management strategies of DS, emphasizing the latest global advancements such as CRISPR gene editing and neurodevelopmental imaging. Despite these breakthroughs, Bangladesh continues to face significant challenges in diagnosing and managing DS due to inadequate healthcare infrastructure, limited diagnostic resources, and persistent societal stigma. The paper highlights disparities in outcomes for individuals with DS between developed and developing nations, focusing on the stark differences in healthcare access, early intervention programs, and societal acceptance. Case studies illustrate the successes of advanced therapies in developed countries while underscoring the gaps in resources and awareness in Bangladesh. These findings emphasize the urgent need for targeted interventions, including improved diagnostic tools, enhanced healthcare systems, and inclusive social practices in developing nations. Additionally, the article examines the role of emerging therapies and advocacy efforts in bridging these gaps. By leveraging global advancements, promoting public-private partnerships, and fostering international collaborations, the study proposes actionable strategies to improve the quality of life for individuals with DS.

Keywords: Genetic Factors; Public Awareness; Syndrome; Genetic dysfunction; Strategies; Trisomy21

Introduction

Down Syndrome (DS), or Trisomy 21, is a genetic condition caused by the presence of an extra copy of chromosome 21. This disorder leads to developmental delays, intellectual disabilities, and various health complications (Esbensen et al., 2024; Roizen & Patterson, 2003; Ta et al., 2022). Globally, it is one of the most common chromosomal disorders, with an estimated prevalence of 1 in 700 live births (World Health Organization (WHO), 2022). While significant advancements have been made in diagnosing and

managing DS in developed nations, resource constraints, cultural beliefs, and social stigma continue to challenge its effective management in developing countries like Bangladesh (Jahan et al., 2024; Koul et al., 2023)

Bangladesh has recently seen an increase in research and awareness campaigns to address DS, but significant gaps remain in early diagnosis and therapeutic interventions (Al Mamun et al., 2022; Munsu et al., 2015). Understanding the causes, types, and implications of Down Syndrome is crucial for implementing targeted interventions and improving the quality of life for affected individuals. Down syndrome,

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also known as trisomy 21, is a genetic disorder caused by the presence of an extra copy of chromosome 21. It is one of the most common chromosomal abnormalities, affecting approximately 1 in 700 live births worldwide (Therrell et al., 2024). The condition results in developmental delays, intellectual disabilities, and a range of physical features and health issues. Down syndrome (DS), also known as Trisomy 21, is a genetic condition caused by the presence of an extra chromosome 21. It is one of the most prevalent chromosomal abnormalities, affecting approximately 1 in 700 live births globally. DS is characterized by distinct physical traits, intellectual disabilities, and a predisposition to various medical conditions.

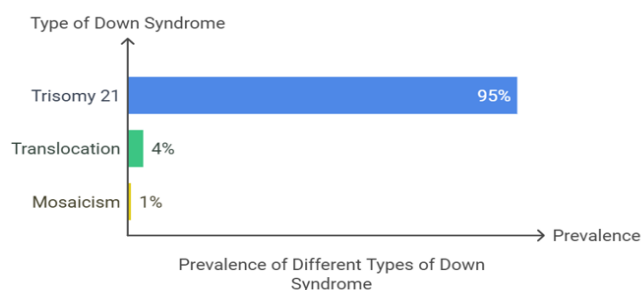


Figure 1. Shows the disease prevalence due to the chromosomal abnormalities

While significant advancements in understanding and managing the condition have been made worldwide, disparities remain, particularly in developing nations like Bangladesh. The article focuses on understanding Down Syndrome (DS) through a global and regional lens, particularly in Bangladesh. The primary objectives are to: Examine the causes, symptoms, and management strategies of DS, emphasizing advancements in global research such as CRISPR gene editing and neurodevelopmental imaging; Highlight disparities in diagnosis, treatment, and social integration between developed nations and Bangladesh, driven by factors like limited healthcare resources, societal stigma, and infrastructure gaps; Propose targeted strategies to bridge these gaps, including improving early detection, advocating for inclusive societal practices, and leveraging global advancements to enhance outcomes for individuals with DS in Bangladesh; Inspire collaborative research efforts and advocacy to improve the quality of life for individuals with DS globally and within the Bangladeshi context (Maure-Blesa et al., 2024; Niyibizi et al., 2023).

Method

Online Research

Online research refers to the use of digital resources and tools to gather information, data, or knowledge on a

particular topic or subject matter. It leverages the internet and various online platforms to access, analyze, and synthesize information for research purposes. Online research involves using the internet to gather information and data for your research project. It includes collecting data from websites, databases, online publications, and various digital sources. I used search engines, academic databases, and various online tools. Online research involves utilizing digital technologies and internet resources to collect data, information, or knowledge relevant to a research question or investigation. This method has become increasingly prevalent due to the accessibility and vast amount of information available online. I reviewed existing online sources, academic articles, and relevant websites to gather background information. Here are key aspects of online research Steps:

Identify research objectives and questions Accessing Information

Researchers can access a wide range of sources online, including academic databases, digital libraries, institutional repositories, websites, blogs, social media platforms, and online forums. These sources provide access to scholarly articles, books, reports, datasets, and other types of information. I used these sources to conduct my research.

Use search engines and databases to find relevant sources Searching and Retrieving Data

Online research typically begins with formulating search queries using search engines (e.g., Google Scholar, PubMed) or specific databases. Researchers use keywords, Boolean operators, and advanced search techniques to retrieve relevant information efficiently. I used these sources and methods to conduct my research.

Evaluate the credibility and reliability of online source Evaluating Sources

Critical evaluation of online sources is crucial to ensure credibility, accuracy, and relevance. Researchers assess factors such as authorship, publication date, peer-review status (for academic articles), and bias to determine the reliability of the information. I critically evaluated the online sources to ensure credibility, accuracy, and relevance to my research.

Extract and compile data from websites, articles, or digital resources

Tools and Techniques

Various tools and techniques support online research, such as reference management software (e.g., EndNote, Zotero), data analysis software (e.g., SPSS, NVivo), and online survey platforms (e.g., SurveyMonkey, Qualtrics). These tools facilitate the

organization of references, streamline data processing, and improve the accuracy of research findings. I used these tools to enhance efficiency in data collection, analysis, and documentation, ensuring a more systematic and reliable research process.

Analyze and synthesize the gathered information for research Analyzing and Synthesizing Data

Once data and information are gathered, researchers analyze and synthesize findings to address research questions or objectives. This involves comparing different sources, identifying patterns or trends, and drawing conclusions based on the evidence collected. I used these data and information that were gathered analyzed and synthesized the findings to address research questions or objectives.

Ethical Considerations

Ethical considerations in online research include respecting intellectual property rights, obtaining permissions for data use where necessary, ensuring data privacy and confidentiality, and maintaining transparency in reporting findings. Adhering to these principles helps uphold the integrity and credibility of the research process. I took these ethical considerations seriously and conducted the research in full compliance with ethical guidelines to ensure responsible and trustworthy outcomes.

Reporting and Disseminating Results

Researchers document their findings in reports, academic papers, articles, or presentations. They cite sources accurately to acknowledge contributions from previous studies and provide context for their research. I followed proper reporting and disseminating of results to conduct my research. Online research offers numerous advantages, including accessibility to a vast amount of information, convenience, and the ability to connect with a global network of researchers and experts. However, researchers must also navigate challenges such as information overload, quality control of online sources, and ethical considerations in data collection and use. By employing rigorous methodologies and critical thinking, researchers can leverage online resources effectively to advance knowledge and contribute to their respective fields.

Literature Review

A literature review is a comprehensive summary and analysis of the existing research on a particular topic. It identifies key themes, debates, and gaps in the literature, and provides a context for new research. Its main purposes are to provide an overview of what is already known, to identify patterns and trends, to highlight areas where knowledge is lacking, and to

establish a theoretical foundation for further research. The process of conducting a literature review involves several steps:

Identifying Research Questions and Objectives

I began by clearly defining the purpose of literature review. Ask what specific questions are trying to answer or what objectives are aimed to achieve. This helps to stay focused and guide search for relevant literature. It defines the research questions and objectives.

Searching for Relevant Literature

I used a variety of sources to gather relevant literature, including academic databases (e.g., PubMed, JSTOR, Google Scholar), library catalogs, and reference lists from relevant articles. Employ search strategies like keyword searches, Boolean operators (AND, OR, NOT), and filtering by date or type of publication to narrow down the search results. Make sure to include seminal works and recent studies to get a comprehensive view of the topic. Searched through databases and other sources for relevant literature using keywords and filters.

Screening and Selecting Sources

Once having a list of potential sources, I evaluated each one for its relevance and quality. Look at the abstract, introduction, and conclusion to determine if the source is pertinent to the research question. Assess the credibility of the authors and the publication, and ensure the methodology and findings are sound. Exclude sources that are outdated, irrelevant, or of low quality. Evaluate and select high-quality, relevant sources from search results.

Organizing the Literature

I Organized selected sources into categories based on themes, theories, methodologies, or chronological order. This can help to identify patterns, trends, and gaps in the research. Use reference management tools like EndNote, Zotero, or Mendeley to keep track of the sources and format citations properly. Create an outline to structure the review logically. Categorize the sources and use reference management tools to organize them.

Analyzing and Synthesizing the Literature

I critically analyze the content of the selected sources. Compare and contrast the findings, methodologies, and theoretical frameworks. Identify key themes, debates, and gaps in the literature. Synthesize the information by integrating the insights from different studies, highlighting how they contribute to the understanding of the research question. Look for consensus and disagreements among researchers. Critically evaluate and integrate findings from the sources.

Writing the Literature Review

I structured the literature review into three main sections: introduction, body, and conclusion. In the introduction, provide an overview of the topic and state the purpose of the review. In the body, discuss the literature in organized themes or categories, presenting a critical analysis and synthesis of the findings. Use subheadings for clarity. In the conclusion, I summarized the key findings, highlight gaps in the research, and suggest areas for future study. Ensure that writing is clear, concise, and free of jargon. Write the literature review with a structured introduction, body, and conclusion.

Revising and Editing

After completing the draft of the literature review, I took time to revise and edit it. Checked for clarity, coherence, and logical flow. Ensured that the arguments are well-supported by the literature. Looked for grammatical errors, typos, and proper citation of sources. Considered getting feedback from peers or advisors to improve the quality of the review. Making sure it adheres to the required formatting and style guidelines. Revise and edit the literature review for clarity, coherence, and correctness. A literature review is a critical appraisal of existing research that helps to build a foundation for new research projects. It demonstrates the researcher's knowledge of the field, identifies where further research is needed, and situates new research within the context of existing knowledge.

Result and Discussion

Causes of Down Syndrome

Down Syndrome results from a genetic anomaly in which an individual inherits an extra copy of chromosome 21, leading to developmental alterations and characteristic physical and intellectual disabilities. This additional genetic material affects cognitive function, growth patterns, and overall health, often resulting in distinct facial features, delayed milestones, and an increased risk of certain medical conditions. While the severity of symptoms varies, early intervention, specialized education, and medical support can significantly enhance the quality of life for individuals with Down Syndrome (Esbensen et al., 2024; Roizen & Patterson, 2003; Ta et al., 2022). This chromosomal disorder arises due to errors during cell division, and its occurrence is influenced by several factors: Maternal Age: Advanced maternal age is a significant risk factor for DS. Women above 35 have a higher likelihood of producing eggs with chromosomal abnormalities, increasing the chances of trisomy 21 (Korlimarla et al., 2021; Mainville et al., 2025).; Genetic

Mechanisms: Trisomy 21: Accounts for 95% of cases and is caused by nondisjunction during meiotic cell division, resulting in an extra chromosome in all cells of the body (Bull et al., 2022; Weijerman & de Winter, 2010); Translocation: Found in about 4% of cases, where an extra part or whole chromosome 21 attaches to another chromosome (Frazão da Silva et al., 2025; Stephanie et al., 2024); Mosaicism: Occurs in approximately 1% of cases, where only some cells carry the extra chromosome due to a post-fertilization error during (Costa, 2011; Costa et al., 2022); Environmental and Societal Factors: Limited prenatal screening, inadequate healthcare infrastructure, and social determinants, particularly in countries like Bangladesh, contribute to the underdiagnosis of DS (Mainville et al., 2025)

Types of Down Syndrome

Down syndrome arises due to chromosomal abnormalities, most commonly nondisjunction during cell division, leading to an extra chromosome 21. It can also occur due to mosaicism, where some cells have an extra chromosome, or translocation, where part of chromosome 21 attaches to another chromosome (Therrell et al., 2024)). These variations contribute to the severity of the condition. The primary cause of DS is the presence of an extra chromosome 21 due to abnormal cell division. It can manifest in three forms: Trisomy 21 (95% of cases): Caused by nondisjunction during cell division, resulting in three copies of chromosome 21 in all cells; Mosaicism (1-2%): Occurs when some cells have the extra chromosome while others do not, leading to milder symptoms; Translocation (3-4%): Part of chromosome 21 attaches to another chromosome, which may be inherited or occur spontaneously; Risk factors include advanced maternal age, genetic predisposition, and prior history of DS births; Down Syndrome can be categorized into three types based on genetic variations: Trisomy 21: The most prevalent form, occurring in approximately 95% of cases, where every cell contains an extra chromosome 21. This type is usually not inherited and results from random nondisjunction (Weijerman & de Winter, 2010); Translocation Down Syndrome: Occurs in about 4% of individuals. In this type, part or all of chromosome 21 becomes attached to another chromosome, such as chromosome 14 or 15. Translocation can be hereditary if one parent carries a balanced translocation without symptoms (Frazão da Silva et al., 2025; Stephanie et al., 2024); Down Syndrome: The rarest type, found in 1% of cases. In this form, some cells have the normal 46 chromosomes, while others have 47 due to an extra chromosome 21. This occurs due to an error during early embryonic cell division (Costa, 2011; Costa et al., 2022). Understanding these types aids in tailoring medical and educational

interventions to the specific needs of individuals with Down Syndrome.

Symptoms and Characteristics

Individuals with Down syndrome exhibit distinctive facial features such as almond-shaped eyes, a flat facial profile, and a small nose. Other common characteristics include low muscle tone, short stature, and a single deep crease across the palm (Grzadzinski et al., 2024; Tungate & Conners, 2021). Cognitive impairments vary from mild to moderate, often accompanied by speech and motor delays. Health complications may include congenital heart defects, respiratory issues, and a higher risk of infections. Individuals with DS often exhibit: Physical Features: Almond-shaped eyes, flat nasal bridge, small ears, and short stature; Developmental Delays: Mild to moderate intellectual disability, delayed speech, and motor skills; Health Complications: Congenital heart defects, hearing loss, vision problems, and higher risk of leukemia and Alzheimer's disease. Symptoms and Characteristics of Down Syndrome:

Physical Characteristics

People with Down syndrome frequently have a unique set of physical and facial characteristics. A short neck, tiny ears, almond-shaped eyes that tilt forward and a flattened facial profile are typical physical traits. They could also have a "Simian Crease"—a single wrinkle that runs across the palm of the hand. (Esbensen et al., 2024; Ta et al., 2022); A delayed development of motor abilities in neonates can result from the decreased muscle tone (Hypotonia) and shorter stature of many persons with Down syndrome (Weijerman & de Winter, 2010); Cognitive and Developmental Characteristics; Learning speed, reasoning and problem-solving skills are among the domains of mild to severe intellectual dysfunction linked to Down Syndrome. Language and motor abilities, including speaking and walking, are frequently delayed in children with Down syndrome. (Epstein & Nadel, 1993); They can do extraordinarily well on activities requiring social interaction and visual learning, even though many also have short-term memory problems. (Bull & Committee on Genetics, 2011)

Health-Related Issues

Heart Problems: Congenital heart problems, which may need surgical repair, are present in almost half of people with Down Syndrome from birth (Roizen & Patterson, 2003); Hearing and Vision Issues : They are more likely to experience hearing loss as well as visual disorders such as Cataracts and Strabismus or Crossed Eyes. (Weijerman & de Winter, 2010); Immune System Weakness : Individuals with Down Syndrome are often

more vulnerable to infections, such as Respiratory Infections and Autoimmune illnesses (Bull et al., 2022). These traits differ greatly and people with Down syndrome could have some or all of these symptoms, which emphasizes the necessity for individualized treatment.

Pathophysiology

The additional chromosome 21 disrupts normal development by altering the expression of multiple genes, leading to: Neurodevelopmental Changes: Impaired synaptic plasticity and reduced neuronal density cause cognitive delays; Cardiac Defects: Increased risk of congenital heart abnormalities, including atrioventricular septal defects; Immune Dysregulation: Increased susceptibility to infections and autoimmune diseases; Endocrine Dysfunction: Predisposition to hypothyroidism and diabetes.

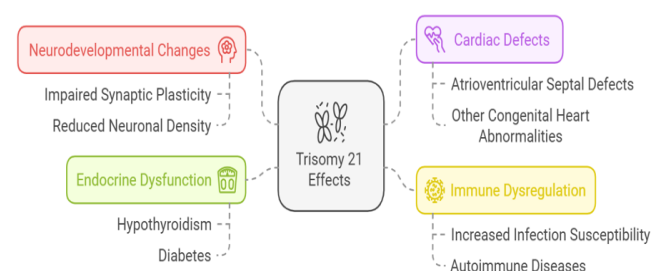


Figure 2. Pathophysiology of Down syndrome shows different pathways responsible for the disease

Diagnosis and screening Methods

Prenatal screening methods, such as nuchal translucency ultrasound, maternal serum screening, and cell-free DNA testing, estimate the likelihood of Down syndrome during pregnancy (American College of Obstetricians and Gynecologists [ACOG], 2022). Diagnostic procedures, including chorionic villus sampling (CVS) and amniocentesis, confirm the diagnosis with high accuracy. Postnatal diagnosis involves physical examination and chromosomal analysis through karyotyping.

Prenatal Screening

Non-invasive techniques for prenatal screening include nuchal translucency ultrasound, which measures the fluid at the back of the fetal neck to assess the risk of chromosomal abnormalities. Additionally, maternal serum markers such as pregnancy-associated plasma protein-A (PAPP-A) and human chorionic gonadotropin (hCG) provide biochemical indicators of potential fetal conditions. Cell-free DNA testing analyzes fetal DNA fragments in maternal blood, offering a highly sensitive method for detecting chromosomal anomalies. For diagnostic confirmation, invasive procedures such as chorionic villus sampling

(CVS) and amniocentesis are performed, allowing direct genetic analysis of fetal cells obtained from placental tissue or amniotic fluid, respectively.

Postnatal Diagnosis

Physical examination and confirmation through karyotyping.

Diagnosis

All pregnant women, regardless of age, should have access to Down syndrome screening and diagnostic tests, according to the American College of Obstetricians and Gynecologists; Screening tests may indicate the possibility that you are carrying a child with Down syndrome. However, these tests cannot definitively determine whether your child has Down syndrome; You can determine with certainty whether your child has Down syndrome with diagnostic testing.

Screening tests during pregnancy

Prenatal care, which includes screening for Down syndrome, is a standard component of care before the baby is born. Screening tests can assist you in determining whether diagnostic testing is necessary, even though they can only determine your chance of having a child with Down syndrome. The integrated screening test and the first trimester combination test are examples of screening tests. The first three months of pregnancy are referred to as the first trimester.

The first trimester combined test

The first trimester combined test is done in two steps. These include: Blood test: This blood test measures the levels of pregnancy-associated plasma protein-A (PAPP-A) and the pregnancy hormone known as human chorionic gonadotropin (HCG). Levels of PAPP-A and HCG outside the standard range may indicate a problem with the baby; Nuchal translucency screening test: During this test, an ultrasound is used to measure a specific area on the back of your baby's neck. When certain conditions caused by chromosome changes are present, more fluid than usual tends to collect in this neck tissue. Using your age and the results of the blood test and the ultrasound, your healthcare professional or genetic counselor can estimate the risk that your baby has Down syndrome.

Integrated screening test

The integrated screening test is done in two parts during the first and second trimesters of pregnancy. The results are combined to estimate the risk of your baby having Down syndrome; First trimester: Part one includes a blood test to measure PAPP-A and an ultrasound to measure nuchal translucency; Second trimester: The quad screen measures your blood level of

four substances present in pregnancy: alpha fetoprotein, estriol, HCG and inhibin A.

Cell-free DNA testing

The placenta releases a tiny quantity of DNA into the circulation of a pregnant woman. It is possible to check for the additional chromosome 21 material associated with Down syndrome using this cell-free DNA in the blood. The test can begin at 10 weeks of pregnancy for women who are at risk of having a child with Down syndrome. Diagnostic testing is typically required to determine that the infant has Down syndrome if the test results are positive.

Diagnostic tests during pregnancy

You can think about getting additional testing to confirm the diagnosis if your screening test results are positive or unclear, or if you are at a high risk of giving birth to a child who has Down syndrome. You can discuss the benefits and drawbacks of these tests with your healthcare provider. Diagnostic tests that can identify Down syndrome include:

Chorionic villus sampling (CVS)

Cells are extracted from the placenta during CVS. The chromosomes of the infant are examined using the cells. Typically, this test is performed between weeks 10 and 14 of pregnancy, during the first trimester. A CVS carries an extremely minimal chance of miscarriage, or pregnancy loss.

Amniocentesis

Cells are extracted from the placenta during CVS. The chromosomes of the infant are examined using the cells. Typically, this test is performed between weeks 10 and 14 of pregnancy, during the first trimester. A CVS carries an extremely minimal chance of miscarriage, or pregnancy loss.

Diagnostic tests for newborns

In the first 24 hours following birth, a physical examination is typically sufficient to diagnose Down syndrome in a baby. A chromosomal karyotype test is ordered by your healthcare provider if they believe your baby has Down syndrome in order to confirm the diagnosis. This test examines your child's chromosomes using a blood sample. Down syndrome is diagnosed if all or some cells have an extra copy of chromosome 21.

Management & Intervention Strategies of Down Syndrome

Management focuses on addressing medical and developmental needs through multidisciplinary care. Early intervention programs, encompassing physical, occupational, and speech therapy, significantly enhance cognitive and motor skills (Ptomey & Wittenbrook, 2015;

ToloueiRakhshan et al., 2024). Medical treatments address associated health conditions, such as heart surgery for congenital defects and routine monitoring for thyroid dysfunction. Effective management of DS requires a multidisciplinary approach, including: Early Intervention: Physical, occupational, and speech therapy to maximize developmental potential; Medical Care: Regular monitoring for congenital heart disease, hypothyroidism, and hearing impairments; Educational Support: Individualized education plans (IEPs) to enhance learning and social integration (Windsperger & Hoehl, 2021).

Early Diagnosis and Screening

Prenatal Testing: Low-cost methods of prenatal screening including nuchal translucency US and non-invasive prenatal test (NIPT); **Postnatal Diagnosis:** Provision of karyotyping and clinical evaluations for accurate diagnosis, Example: Diagnostic facilities are inadequate in the quantity of Dhaka Medical College Hospital and centres of excellence (Dhaka and Chittagong); **Parental Counseling and Education:** Treatment in families of once psychiatric illness with psychological modalities for improving the treatment of families both in relation to alleviating their sense of stigma, but also in relation to the management of reality and conflict. Community health workers (CHWs) can be recruited to educate rural communities on DSM.

Healthcare and Therapy Access

Medical Management: Establishment of a pediatric care unit to treat and care for diseases that are endemic to the pediatric population, e.g., congenital heart disease, thyroid disease, and so on; **Therapeutic Support:** Availability of physical, speech, and occupational therapy in the developmental enhancement process; **Inclusive Education:** Admission of children with Down's syndrome to regular schools as mandated by the policy framework. Training teachers to handle children with special needs. Example: Organizations, e.g., Society for the Welfare of Autistic Children (SWAC), taught education in Bangladesh; **Community Awareness and Advocacy:** Awareness campaigns to alleviate stigma and misinformation towards DS. Consider these materials as the family support network brokered activities or engagements with energizing NGOs (e.g., BRAC); **Policy Development:** Making and putting into effect disability policies (e.g., Persons with Disabilities Rights and Protection Act, 2013). Evidence for the social security interventions aimed at bettering the quality of life of children with DS has been compelling enough to be drawn;

Invention Strategies of Down Syndrome

Localized Research and Innovation: Design and development of culturally adapted model interventions based on good practice in other countries (e.g., transplanted interventions); **Affordable Assistive Technologies:** Development of applied and cheap assistive devices for the developmental modes (e.g., educational toys and devices). Collaborating with local manufacturers for accessibility; **Mobile Health Applications:** Bangla parent education and therapy management app development in the era of developing mobile internet application in Bangladesh; **Community-Based Rehabilitation (CBR):** CBR programs are being brought to the community to provide therapy and service to rural areas. Instructional direct work between CHWs and patients in the field; **Public-Private Partnerships (PPP):** Cooperation with the NGO and the private sector in order to set up centres and institutions for therapy and special education, Example: Coordination between government and non-government organizations (NGOs) such as CRP Center for Rehabilitation of Paralyzed; **International Collaboration:** Collaborating with other partners such as UNICEF, WHO, and Down Syndrome International for financing, capacity building and resource creation.

Treatment Algorithm

Congenital Heart Defects: Surgery for structural abnormalities; **Diuretics** (e.g., furosemide) for heart failure; **Thyroid Dysfunction:** Levothyroxine for hypothyroidism; **Seizures:** Antiepileptics (e.g., valproate, lamotrigine); **Behavioral Symptoms:** Selective serotonin reuptake inhibitors (SSRIs) like fluoxetine; **Cognitive Decline in Adults:** Experimental use of memantine or acetylcholinesterase inhibitors like donepezil.

Down Syndrome and Context in Bangladesh

Down Syndrome (DS), or Trisomy 21, is a genetic disorder caused by the presence of an extra chromosome 21. This condition leads to developmental delays and various health challenges. Globally, efforts to understand DS are expanding, with Bangladesh making notable contributions in research, awareness, and therapeutic innovations. The prevalence of Down Syndrome in Bangladesh is underreported due to limited diagnostic facilities and societal stigma. However, recent estimates suggest that around 1 in 700 live births may be affected, aligning with global statistics (World Health Organization (WHO), 2022). Down Syndrome (DS) is underreported in Bangladesh due to limited diagnostic resources and societal stigma. Contributing factors include advanced maternal age, lack of prenatal screening, and inadequate healthcare infrastructure. Efforts by researchers and NGOs aim to enhance early diagnosis, genetic studies, and awareness

programs to address these issues (Mainville et al., 2025; Niyibizi et al., 2023; World Health Organization (WHO), 2022)

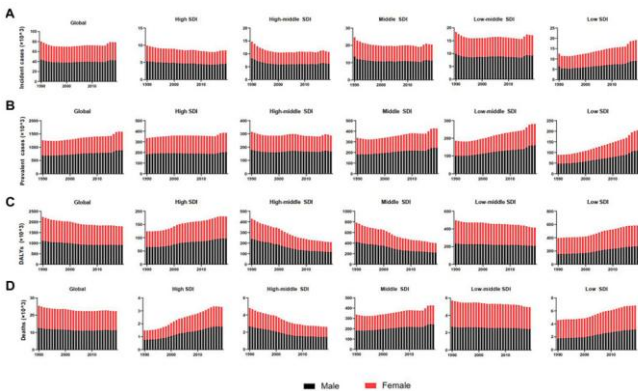


Figure 3. Burden and trends of Down syndrome globally and in five SDI quintiles from 1990 to 2019. (A) Incident cases. (B) Prevalent cases. (C) Disability-adjusted life-years (DALYs). (D) Deaths. Black bars represent males, and red bars represent females. Note: DALYs, disability-adjusted life-years; SDI, social-demographic index.

Current Research in Bangladesh

Bangladesh has seen a surge in research on DS over the last decade. In Bangladesh, research focuses on understanding genetic variations, health outcomes, and social integration. Studies reveal high rates of untreated co-morbidities like congenital heart defects and speech delays (Kwon et al., 2024; Srivastava & Bolia, 2019). Globally, cutting-edge research explores chromosome silencing through CRISPR and neurodevelopmental imaging to understand cognitive delays (Jiang et al., 2022; Smith et al., 2021). Advanced therapies, including pharmacological and behavioral interventions, are leading to improved outcomes (Mendola et al., 2021). Key areas include: Genetic Studies: Researchers are working on identifying regional genetic variations in trisomy 21 cases (Jahan et al., 2024); Health Outcomes: Studies examine co-morbidities like congenital heart defects and hypothyroidism among Bangladeshi DS patients (Kwon et al., 2024; Srivastava & Bolia, 2019); Social Impact: Sociological research highlights the challenges of inclusive education and societal acceptance (Ashrafun et al., 2024; Chowdhury et al., 2024; Islam et al., 2022)

Case Study in Bangladesh

A 2020 study in Bangladesh documented high rates of untreated congenital conditions, highlighting gaps in early intervention programs. Conversely, case studies in the USA and Europe show remarkable advancements, including cognitive improvements through combined therapies and insights into aging in DS populations (Chen et al., 2022; Fortea et al., 2021; Zammit et al., 2024)

The disparity in outcomes reflects differences in healthcare access, research infrastructure, and societal attitudes; Bangladesh: A study conducted at Dhaka Medical College (2020) tracked 50 children with DS, noting a high prevalence of untreated congenital heart disease and speech delays. These findings underscore the need for improved early intervention programs

Case Studies Worldwide

Country	Years	DS incidence (per 10,000 live births)	
		Country	Years
Canada	1993-1997	12.79	
United States	1985-1993	9.8-11.8	
Brazil	1993-1998	15.01	
Norway	1993-1998	10.28	
Finland	1993-1998	11.75	
England & Wales	1993-1998	5.38	
Europe	1990-2009	22.02	
France Paris	1981-2000	25.9	
Australia	1993-1997	13.14	
New Zealand	1994-1998	9.9	
United Arab Emirates	1996-1998	17.65	
Japan	1980-1997	5.82 (8.3-9.7*)	
Singapore	1993-1998	10.2	
Taiwan	2001-2010	7.9	

*. After correction according to the estimated ascertainment ratio: 60–70

Figure 4. Case Studies Worldwide

USA: A landmark case in New York demonstrated improved cognitive outcomes following early intervention using behavioral therapies combined with pharmacological support (Chen et al., 2022); Europe: A longitudinal study in the UK tracked individuals with DS over 30 years, providing insights into aging and Alzheimer’s disease in this population (Zammit et al., 2024)

Comparison of Case Studies: Bangladesh vs. Worldwide

A 2020 study in Bangladesh documented high rates of untreated congenital conditions, highlighting gaps in early intervention programs. Conversely, case studies in the USA and Europe show remarkable advancements, including cognitive improvements through combined therapies and insights into aging in DS populations (Chen et al., 2022; Fortea et al., 2021; Zammit et al., 2024). The disparity in outcomes reflects differences in healthcare access, research infrastructure, and societal attitudes.: Healthcare Access: While advanced countries provide free genetic counseling and therapy, Bangladesh struggles with affordability and accessibility; Therapeutic Focus: Global studies emphasize targeted therapies, while Bangladesh prioritizes basic healthcare and education initiatives; Outcomes: Life expectancy for individuals with DS in

developed nations exceeds 60 years, compared to 30-40 years in Bangladesh (Jahan et al., 2024; Koul et al., 2023).

Global Research Trends

Worldwide, research on Down Syndrome is advancing rapidly. Genetic research is utilizing cutting-edge techniques such as CRISPR-Cas9 to explore chromosome silencing, potentially opening new therapeutic avenues. Additionally, advancements in neurodevelopmental studies and early intervention strategies are improving cognitive outcomes and quality of life for individuals with Down Syndrome (Huang et al., 2024; Moon & Lawrence, 2022); Neurodevelopmental Studies: Understanding cognitive delays through brain imaging techniques has become a focal point (Beresford-Webb et al., 2025; Hartley et al., 2022); Therapeutic Innovations: Development of targeted therapies to address specific genetic pathways is gaining momentum.

New Trends in Down Syndrome Research

Globally, technologies such as CRISPR gene editing and RNA-based therapies show promise in managing the effects of DS. Non-invasive prenatal testing (NIPT) is transforming early diagnosis, though its accessibility remains a challenge in Bangladesh. Stem cell therapy, currently experimental, is another emerging avenue with potential for neurodegenerative damage repair (Cecerska-Heryć et al., 2023; Sivandzade & Cucullo, 2021); Genetic Advances: Innovations like gene editing and RNA therapies hold promise for mitigating some effects of DS by silencing the extra chromosome; Early Detection Technologies: Non-invasive prenatal testing (NIPT) is revolutionizing early detection. While NIPT is routine in developed countries, efforts are underway to make it affordable in Bangladesh; Emerging Therapies: Modern interventions include drugs like memantine for cognitive enhancement and SSRIs for behavioral improvements. These complement traditional therapies, such as occupational and speech therapy, offering a more comprehensive approach to DS management. While developed nations integrate these advanced therapies, Bangladesh still focuses on basic healthcare and education due to resource constraints (Abukhaled et al., 2024); New Therapies ;Pharmacological Interventions: Trials using drugs like memantine and selective serotonin reuptake inhibitors (SSRIs) show potential in enhancing cognitive abilities (Mendola et al., 2021).Stem Cell Therapy: Experimental studies suggest that stem cells could repair neurodegenerative damage (Cecerska-Heryć et al., 2023; Sivandzade & Cucullo, 2021)

Comparison of New vs. Old Therapies

Choose the most effective therapy approach for comprehensive care.



Figure 4. Comparative approach between old and new therapies

Genetic Counseling and Awareness

Awareness initiatives in Bangladesh, led by organizations like the Bangladesh Down Syndrome Society (BDSS), emphasize prenatal care and family support.

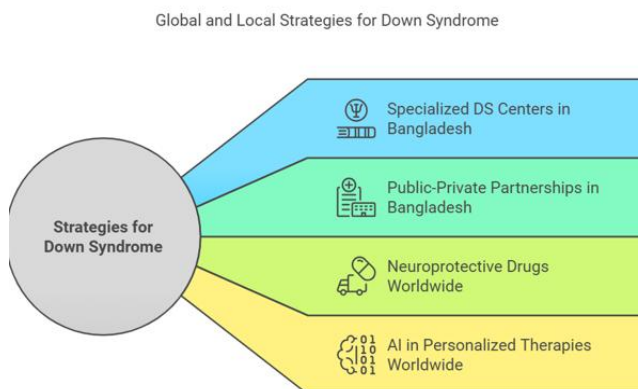


Figure 5. Shows future Strategies for the management of Trisomy 21

Globally, efforts by Down Syndrome International (DSi) promote education on genetic counseling, vital for early detection and improved care outcomes. Raising awareness about genetic counseling is vital for reducing DS prevalence and improving management: Bangladesh: Awareness campaigns by NGOs like Bangladesh Down Syndrome Society (BDSS) emphasize prenatal care and family support; Global Efforts: Organizations like Down Syndrome International (DSi) promote global solidarity and education on genetic counseling; Future Directions: In Bangladesh, priorities include establishing specialized DS centers, subsidizing diagnostic tools like NIPT, and fostering public-private partnerships. Globally, research aims to develop neuroprotective drugs and leverage AI for personalized therapies. These efforts could transform the quality of life for individuals with DS, narrowing the gap between developed and developing nations; Bangladesh: Establishing specialized centers for DS care and

research; Enhancing public-private partnerships to subsidize advanced diagnostic tools like NIPT; Worldwide; Exploring neuroprotective drugs to delay or prevent Alzheimer's disease in DS individuals; Expanding the use of artificial intelligence in developing personalized therapies.

Social viewpoint on Down syndrome in Bangladesh

In Bangladesh, cultural beliefs, stigma, and lack of awareness create significant social obstacles for individuals with Down syndrome, a genetic disorder resulting from an extra chromosome 21. In Bangladeshi culture, disability, such as Down syndrome, is often not properly understood and is commonly linked to superstitions or seen as punishment from higher powers (Sijuola & Davidova, 2022). These false beliefs lead to the marginalization of people with Down syndrome and their families.

Social Stigma and Marginalization

Social stigma and marginalization refer to the negative attitudes and treatment experienced by individuals or groups who are perceived as different or deviant from societal norms. In Bangladesh, cultural norms frequently uphold traditional perspectives on health and disabilities. Consequently, families of children with Down syndrome could encounter discrimination, as certain members of society may perceive the condition as a "curse" or consequence of previous wrongdoing (Hussain & Raihan, 2022; Nuri et al., 2022; Tushar et al., 2020). This societal expectation may result in seclusion, as parents may choose to conceal their children to avoid criticism.

Barriers in Education and Employment

Access to inclusive education for individuals with Down syndrome remains limited. Despite adopting policies like the Persons with Disabilities Rights and Protection Act (2013), practical implementation faces challenges due to inadequate resources and societal resistance. Additionally, the lack of trained educators, insufficient funding, and deeply rooted social stigmas further hinder the effectiveness of inclusive education initiatives (Islam et al., 2022). This lack of support continues into adulthood, where opportunities for meaningful employment are rare.

Efforts to Improve Awareness and Inclusion

Organizations such as the BDSS are crucial in raising awareness and providing support for individuals with Down syndrome. These organizations aim to educate families, provide caregivers with training, and advocate for policy changes. While their work has improved understanding and acceptance in

some urban areas, significant differences persist in rural regions.

Religious and Family Support

Religious beliefs in Bangladesh have the potential to impact views on Down syndrome in both beneficial and detrimental ways. Faith offers solace and inspires certain families to take care of their children as a sacred responsibility (Lakhani et al., 2025). On the other hand, some individuals may receive negative feedback from their community for perceiving the situation as a challenge to their beliefs. Despite some advancements in raising awareness and promoting inclusion, individuals with Down syndrome in Bangladesh still encounter significant social obstacles. Dealing with these problems necessitates a comprehensive strategy, which involves educating the public, improving the enforcement of disability rights, and providing more assistance to impacted families.

Conclusion

In Bangladesh, the presence of Down Syndrome is influenced by a combination of genetic, social, and healthcare complexities. Early detection and comprehensive care are impeded by limited diagnostic facilities, societal stigma, and insufficient resources. Even with these obstacles, there is noticeable development through more research, advocacy from groups, and initiatives to improve public knowledge. Nevertheless, there are still gaps in healthcare availability, inclusive education, and long-term assistance for people with Down Syndrome. Progressive developments in genetic research and cutting-edge treatments bring optimism, yet bringing them to Bangladesh necessitates fundamental enhancements. By combining worldwide approaches, promoting collaborations between the public and private sectors, and enhancing social welfare structures, Bangladesh has the potential to enhance results for individuals with Down Syndrome, leading to a more fair and all-encompassing society.

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Author Contributions

Conceptualization,; methodology,; validation,; formal analysis,; investigation,; resources,; data curation,; R. A.; writing—original draft preparation,; writing—review and editing, .: visualization, Md. T. R. T. All authors have read and agreed to the published version of the manuscript.

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Conflicts of Interest

The authors declare no conflict of interest.

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