GOMAL UNIVERSITY
JOURNAL OF RESEARCH
Gomal University, Dera Ismail Khan, Khyber Pakhtunkhwa, Pakistan
ISSN:1019- 8180 (Print) ISSN: 2708- 1737 (Online)

Website	www.gujr.com.pk	HEC Recognized	Social Sciences	CrossRef	DOI:10.51380

THE CLINICAL AND HEALTHCARE ACCESS PROFILE OF CHILDREN WITH DOWN SYNDROME IN PAKISTAN: A DISTRICT-LEVEL ANALYSIS

Maryam Nazir[1], Nazia Ahmad[2] & Nihal Nisar[2]
[1]Government Post-Graduate College for Women, Mardan, Khyber Pakhtunkhwa (KP), Pakistan
[2]Special Education Department (SED), The Government of Khyber Pakhtunkhwa (KP), Pakistan

KEYWORDS
Down Syndrome, Healthcare Access, Demographic Characteristics, Comorbidities, Clinical Profile, Pakistan

ABSTRACT

Down syndrome (DS) is a common chromosomal condition associated with intellectual disability & multiple medical complications. In order to conduce the study, null hypotheses were formulated to precede the study. A descriptive cross-sectional study with the inferential analysis was conducted to assess the demographic characteristics, related health conditions as well as healthcare accessibility amid children with Down syndrome attending special education center in District Mardan. The total of 100 students diagnosed with DS were included using purposive sampling. The standardized 15-item questionnaire was used to collect data, which was then analysed using descriptive statistics and the chi-square test. Most participants were between 13 and 18 years old, and 70% were men. There was a statistically significant link ($p < 0.05$) between respiratory disorders, thyroid abnormalities, and sensory impairments. Still, congenital heart defects & neurological illnesses were not significant. There were also significant differences in healthcare access features, with high medical prices (60%) being the biggest problem. The findings of current study suggest that children with down syndrome in this context experience many health difficulties and inequitable access to medical care. Similarly, results reveal that these patterns are not random and have a statistically significant meaning in specific context. The improved care for those affected could come from earlier diagnosis, more awareness and better coordination of healthcare at district level.

Article History
Date of Submission: 20-02-2026
Date of Acceptance: 23-03-2026
Date of Publication: 30-03-2026

Corresponding Author: Nihal Nisar: nihalnisar787@gmail.com

DOI: https://doi.org/10.51380/gujr-42-01-07

INTRODUCTION

The down syndrome is one of the most frequent genetic disorders in the world. It is a genetic disorder caused by an extra copy of chromosome 21 and is associated with several connected medical issues, unique facial features, varied levels of cognitive impairment (Kazemi, Salehi & Kheirollahi, 2016). This condition affects all parts of life, including physical, mental, and developmental. The DS affects all societies; however, its prevalence and overall effects vary based on socioeconomic, environmental, and healthcare-related factors (Akhtar & Bokhari, 2023). The people with Down syndrome often deal with a variety of health issues that lower their quality of life. Congenital heart problems, gastrointestinal issues, pulmonary infections, dysfunctional thyroid, neurological disorders & sensory impairments are related conditions (Ijezie, Healy, Davies, Ballester & Heaslip, 2023). Average lifespan and functional results can be increased with early diagnosis and effective medical therapy. Still, access to specialized services remains limited in low and middle-income areas, controlling disease presents social and financial difficulties for many families (Marilyn, Trotter, Santoro, Christensen & Grout, 2022).

Due to advancements in the healthcare and research, people with Down syndrome have seen gains in the life expectancy and quality of life in recent times. Disparities in health outcomes persist, nonetheless, basically due to low knowledge and fewer resources. Research in these areas is also scarce, and many people live in low- and middle-income areas with not enough access to healthcare (Fortea, McGlinchey, Espinosa & Rafii, 2024). There is not a lot of district -level information about children with Down syndrome in Pakistan, especially in rural areas. District Mardan in KP has a mix of urban and rural country residents, and not everyone has same access to medical care. There are special education facilities, but there isn't any reliable information about how people with DS in this district get healthcare, what medical problems they have, or how many of them there are. The health planners, educators, and lawmakers encounter difficulties in formulating targeted measures or improving support networks for affected families without such data (Ali & Fatima-Haider, 2024). Therefore, the present study was conducted to assess demographic distribution of children in Down syndrome attending special education center in the District Mardan, to identify most common associated medical conditions, and to evaluate the accessibility and quality of the healthcare services available to them.

Research Objectives

1. To assess demographic distribution of those impacted by Down syndrome & ascertain condition's prevalence in District Mardan.
2. To categorize and evaluate the most communal diseases associated with the Down syndrome in the District Mardan, KP.
3. To assess quality, accessibility and availability of medical services in District Mardan to the people with Down syndrome.

Research Hypothesis

1. The study does not show any significant difference in demographic distribution & the prevalence of Down syndrome (H0).
2. The prevalence of the particular illnesses in the District Mardan and Down syndrome is not significantly associated (H0).
3. There is no significant difference in quality, accessibility and availability of the medical services availed to the individuals with Down syndrome in District Mardan with that of the general population (H0).

LITERATURE REVIEW

The Down syndrome is the most prevalent chromosomal disorder internationally, primarily linked with intellectual disability and a spectrum of additional clinical features. Occurring in approximately 1 in 800 individuals worldwide (Bull, 2020). The DS results from complete or partial trisomy of chromosome 21, results in overexpressions of some genes on chromosome. This gene causes an imbalance that underpins physical, intellectual, and health manifestation detected in affected individuals (Antonarakis, Lyle, Dermitzakis, Reymond & Deutsch, 2020). It took about a hundred years before the chromosomal basis was identified, so that, as early as 1866, John Langdon Down identified this condition in laboratory. In 1959, phenotype was linked with an additional chromosome 21, thereby describing the genetic nature of affected individuals (LeJeune, Gautier & Turpin, 1959). Down Syndrome occurs throughout world, though its incidence varies with mother's age during pregnancy and availability of prenatal screening. The annual number of births of infants with Down Syndrome in United States is about 5,000 infants, more than 200,000 individuals live with down syndrome (Marilyn & Bull, 2020).

According to a recent survey, one case of Down Syndrome reported per 779 new births in the USA (~12.8 per 10,000 live births) (Antonarakis et al., 2020). Prevalence of Down Syndrome in all parts of Europe ranges from 3.3 to 6.0 per 10,000 individuals, varying with the mother's age, prenatal screening uptake, other laws on pregnancy termination (Loane, Morris, Addor, Arriola, Budd & Doray, 2013). Benjak, Vuljanić, Draušnik, Barišić, Vuković and Vuletić (2025) reported on nationally registered data to estimate the Down syndrome ratio in Croatia. The data showed 3.7 to 5.3 per 10,000 persons, related with different levels of Down Syndrome, on the existing birth ratio. Among the linked diseases, Congenital cardiac abnormalities were the common associated issue, with a high ratio in men. The data suggest that longer survival period quality treatment may change existing condition. In 2015, 90% of projected 6-million people with Down Syndrome lived outside North America, Europe (McGlinchey et al., 2025). Ye et al. (2025) observed global impact of Down syndrome on mortality rate of children and adolescents utilizing GBD data. Their results mentioned that, despite a reduction in mortality rates with time, children with Down syndrome die at higher rates, in regions with the lower SDI.

The study manifested that mortality rate is negatively associated with socio-demographic conditions. These data indicate that prompt interventions and access to health care remain critical variables in improving survival rates. This global gap underscores the importance of improving diagnostic, therapeutic, and support services in resource-limited settings (Adachi et al., 2023). The existence of Down Syndrome affects almost all organ systems of individual. 50% of the cases had cardiac issues from infancy, most often atrioventricular septal defects (AVSD), which are still major cause of early death and illness (Paladini et al., 2000). Besides, DS is recognized as a risk factor for some hematological spites. The individuals with DS are substantially likely to get acute megakaryoblast leukemia and acute lymphoblastic leukemia (ALL), especially because they have trisomy 21 and GATA1 mutations (Asim et al., 2015). DS is associated to intellectual disability; the cognitive consequences might be very different and vast. While social and emotional performance surpasses expectations derived from cognitive scores most individuals experience mild to moderate intellectual & social issues (Grieco et al., 2015).

The developmental outcomes can be improved and stop problems to happen, by applying rehab activities like speech therapy, occupational therapy and physiotherapy. Compared to peers with other intellectual disabilities, children with the Down syndrome generally exhibit fewer externalizing psychiatric symptoms, so reducing familial stress (Otterman et al., 2019). Despite most of the countries working to develop different plans for reducing the impact of Down Syndrome but still a of work need to do. It is in practice, that DS clinics can help with diagnosis, treatment coordination, and observing health parameters, especially in children of low age (Skotko et al., 2013). A defining feature of clinical profile in Pakistan is the reliance on postnatal diagnosis, often based on the phenotypic presentation rather than confirmatory genetic testing. Within Pakistani context, Down syndrome is consistently identified as one of the most common chromosomal causes of intellectual disability, accompanied by a complex clinical profile involving multisystem comorbidities. Still a large group of people with DS in most parts of the world don't have access to such facilities, especially in poor & low-income nations.

The health problems are present even in developed countries, frequently influenced by many other factors like socioeconomic status, race, and residency. The cultural differences in life expectancy and healthcare availability among individuals with Down syndrome have been reported in United States (Krahn et al., 2006). Alanazi et al. (2025) conducted study to explore the financial expenses and healthcare access among Saudi Arabian societies of children with Down syndrome. The study found that families spend a lot of their money on care, and the main costs are for medical care and therapy. Similar hospital-based investigations across Pakistan reinforce this pattern, highlighting congenital cardiac anomalies as a leading contributor to morbidity and mortality among these children Significant differences in access to services between urban and rural areas were also found, with rural households facing the greater difficulties as a result of the less resources and more expensive transportation. This diagnostic delay knowingly impacts development trajectories, as early stimulation therapies and medical interventions are known to improve cognitive and physical outcomes. These results emphasize the need for better healthcare support systems also continuous burden of care.

RESEARCH METHODOLOGY

This study was conducted as descriptive cross-sectional research. It focused on exploring the existing situation of children with Down syndrome and their related health issues. The study was conducted at Centre of Intellectually Challenged Children, Special Education Complex, Mardan (Sheikh Maltoon), in District Mardan, Khyber Pakhtunkhwa. The research methods provides vomprehensive details about the methods and procedures supported by relevant tools and techniques for conducting the study on particular issues and attaining the desited outcomes.

Population and Sample

The population of the study included children between the ages of 5 to 19 years living in District Mardan. Still, due to practical limitations, the sample was selected from individuals diagnosed with Down syndrome who were enrolled in Special Education Complex Mardan. In this linking, it was difficult to access the individuals with Down syndrome in the general community. Consequently, the purposive sampling method was used in the study access the sample. A total of 100 students diagnosed with Down syndrome were included in the study sample. The selection was based on the availability and confirmed diagnosis records at the institution.

Research Instrument

A standardized questionnaire with fifteen items was used to gather data. In order to address every goal of the study, questionnaire was created in conjunction with subject matter experts. It contained five questions about distribution of demographics, five questions about linked medical issues, five questions about reach and availability of healthcare services. The split-half statistics was applied for finding the reliability of instrument and it was 0.87 which was satisfactory.

Data Collection Procedure

Data was collected from the students with Down Syndrome enrolled in the special education complex Mardan. In this connection, the classroom teachers helped in data collection and through provision of clinical record of each student in particular area and jurisdiction used in study.

RESULTS OF STUDY

The collected data were tabulated and interpreted by using both descriptive and inferential statistical methods. Variables were arranged in three groups and responses were expressed in percentages and then chi-square was applied for testing hypothesis. The calculated value of chi-square (χ^2) analyzed relationships among the variables. This test helped figure out if differences in demographic characteristics, related conditions, and healthcare access patterns were real or just random. In this investigation, a p-value of less than 0.05 was considered statistically significant. The questionnaire was pilot tested on small group prior to gathering final data to ensure that questions were understandable and straightforward. After receiving input from experts, few adjustments were made to make questions easier for respondents to understand.

Table 1

Demographic Distribution

1	Age	Frequency	Percentage	Chi-Square χ^2)	df	P-value
	0-5 years	9	9	42.96	3	<0.001
	6-12 years	33	33			
	13-18 years	48	48			
	19 years & above	10	10			

	Total	100	100			
2	Gender	Frequency	Percentage	Chi-Square (χ^2)	df	P-value
	Male	70	70	16	1	<0.001
	Female	30	30			
	Total	100	100			

Table 1A

Demographic Distribution

3	Residency	Frequency	Percentage	Chi-Square (χ^2)	df	P-value
	Urban	40	40	3.84	1	
	Rural	60	60			
	Total	100	100			
4	Down Syndrome in family	Frequency	Percentage	Chi-Square χ^2)	df	P-value
	Yes	30	30	16	1	<0.001
	No	70	70			
	Total	100	100			
	Time of Diagnosis	Frequency	Percentage	Chi Square (χ^2)	df	P-value
5	By birth	45	45	24.49	2	<0.001
	1-4 years	45	45			
	5 year and above	10	10			
	Total	100	100			

The chi-square analysis revealed statistically significant differences in distributions of age, gender, family history, and time of diagnosis ($p < 0.001$). The residency showed a marginally significant association ($p \approx 0.05$). Hence, null hypothesis, H0: The study does not show any significant difference in demographic distribution and prevalence of Down syndrome in has been rejected that leads to findings that demographic and diagnostic factors play important role in understanding prevalence and identification patterns of Down Syndrome in the study population.

Table 2

Common Illness Associated With Down Syndrome

1	Congenital Heart Defect	FRE	PER	Chi-Sq (χ^2)	df	P-Value	Result
	Yes	49	49	0.04	1	0.84	Not Significant
	No	51	51				
	Total	100	100				
2	Respiratory Issues	FRE	PER	Chi-Sq (χ^2)	df	P-Value	Result
	Yes	20	20	36	1	<0.001	Significant
	No	80	80				
	Total	100	100				
3	Thyroid related Issues	FRE	PER	Chi-Sq (χ^2)	df	P-Value	Result
	Yes	25	25	25.00	1	<0.001	Significant
	No	75	75				
	Total	100	100				

4	Hearing or Vision Impair	FRE	PER	Chi Sq (χ^2)	df	P-Value	Result
	Yes	25	25	25.00	1	<0.001	Significant
	No	25	75				
	Total	100	100				
5	Neurological Conditions	FRE	PER	Chi Sq (χ^2)	df	P-Value	Result
	Yes	52	52	0.16	2	0.69	Not Significant
	No	48	48				
	Total	100	100				

The Chi-square analysis indicates that respiratory issues, thyroid-related issues, and hearing or vision impairments are significantly associated with Down syndrome in District Mardan. In contrast, the congenital heart defects and the neurological conditions show no significant association. In this connection, this implies that while some medical disorders are not much different from the overall distribution, others are more common among people with Down syndrome.

Table 3

Quality, Accessibility And Availability of Medical Services

1	conditions to Down syndrome	FRE	PER	Chi-Sq (χ^2)	df	p-value	Result
	Regularly	40	40	30.0	3	0.000	Significant
	Occasionally	25	25				
	Rarely	35	35				
	Never	0	0				
	Total	100	100				
2	Healthcare Facilities in Mardan	FRE	PER	Chi-Sq (χ^2)	df	p-value	Result
	Yes	60	60	6.0	2	0.049	Significant
	No	30	30				
	Not Sure	30	30				
	Total	100	100				
3	Challenges in medical care	FRE	PER	Chi Sq (χ^2)	df	p-value	Result
	Lack of doctors	10	10	80	3	0.000	Significant
	High medical costs	60	60				
	awareness about services	20	20				
	distances to healthcare centers	10	10				
	Total	100	100				

Table 3A

Quality, Accessibility And Availability of Medical Services

4	Access to therapy services	FRE	PER	Chi Sq (χ^2)	df	p-value	Result
	Yes (Easily)	50	50	14	2	0.0001	Significant
	Yes (Difficultly)	30	30				
	No access	20	20				
	Total	100	100				

5	Quality of healthcare	FRE	PER	Chi Sq (χ²)	df	p-value	Result
	Very satisfied	38	38	59.3	4	0.000	Significant
	Somewhat satisfied	35	35				
	Neutral	25	25				
	Dissatisfied	2	2				
	Somewhat dissatisfied	0	0				
	Total	100	100				

The results for every variable are statistically significant ($p < 0.05$). In this connection, as a result, the null hypothesis, Ho: There is no significant correlation between the prevalence of specific illnesses in District Mardan and Down syndrome, is rejected as per the results from analysis.

DISCUSSION

The study aims to explore healthcare access, health problems, and demographic distribution of children diagnosed Down syndrome in District Mardan. It applied chi-square to interpret data, which leads to the rejection of null hypotheses formulated for prediction. the overall results were statistically significant ($p < 0.05$). This means that there are reasons behind the patterns we see and that they aren't random. The demographic characteristics that showed a significant variation in age, gender, family history, and timing of diagnosis ($p < 0.05$). The age limit of students was 13 and 18 years, which manifests that enrolment and diagnose in institutions happen later. This may be lack of facilities to provide early screenings or because the diagnosis took too late. This view is validated by the concept that most of the children in study were not diagnosed at birth. Bull (2020) admitted that while early diagnosis is essential for desired developmental results, but it remains delayed in most of the regions. There ratio of Down Syndrome is higher in men as compared to women. Which is also manifested from the results of current study which a statistically significant difference ($p < 0.001$) among the genders.

It must be noted that down syndrome is not a sex-linked disease but still the ration is high in men. Social background may influence developmental results, as male individual are more frequently referred to institutions of special education, healthcare facilities (Dyck et al., 2004). Krahn et al. (2006) noted that socioeconomic and demographic situations can make it harder to get proper care and rehabilitation, especially in the regions where resources are scare. The results at linked health issues, results were not constant. Some issues showed a strong link, whereas others did not. For example, Thyroid related issues, hearing or visual impairments, chest and respiratory problems were all shown be statistically significant ($p < 0.05$). These results align with earlier research. According to Grieco et al. (2015), endocrine related issues and lung problems are worldwide in distribution among individuals with Down syndrome. Similarly, According to Antonarakis et al. (2020), thyroid problems are commonly noted in people with Down syndrome and needs medical treatment. However, neurological disorders ($p = 0.69$) and congenital cardiac problems ($p = 0.84$) were not statistically significant in our investigation.

In contrast, congenital heart disease is described as a prominent problem among children with Down syndrome in numerous international results. For example, Paladini et al. (2000) found high prevalence of heart defects in such cases. The difference in the present study may be due to underdiagnosis or limited access to proper diagnostic tools. It is possible that some conditions were not properly recorded. So, non-significant result here should be interpreted carefully, not as the absence of the condition. In third part of the study, all variables related to healthcare access and quality showed statistically significant results ($p < 0.05$). This clearly indicates that healthcare access is not equal among participants. Only a portion of children reported regular medical check-ups, while many faced difficulties. The most common barrier reported was high medical cost, which is expected in low-income settings. Alkhushi (2024) also noted that financial burden is a major challenge in managing chronic conditions in such regions. Other factors, lack of specialized doctors, long travel distances, and low awareness, were reported. While some individuals reported having access to treatment services, many others said it was challenging. There have been reports of such worries in other developing nations.

According to Ahmed and Tamim (2025), social stigma, a lack of early diagnosis, and a lack of healthcare structure continue to have an impact on how Down syndrome is managed. The study find out that persons with Down Syndrome often have more than one disorder, and the life quality of that person depends on the provision of health care facilities. Some people were satisfied and happy with results, while others were not. This means that quality of healthcare services isn't always the same. The facilities, the resources that are available, or the families' financial situation all play a role. According to Fried (2017), not everyone is treated equally by the healthcare system. When we consider all of results, it is evident that children with Down syndrome in this environment suffer variety of hitches. The delays in diagnosis, specific medical issues, and challenges in obtaining the right medical care are some of these. The conclusion these differences are significant and demand attention is further supported by failure of all three hypotheses. Ye et al. (2025) confirmed that more research is required to elucidate epidemiologic designs and tendencies among individuals with Down syndrome, to facilitate the development of effective strategies to prevent issues improve survival and good health.

CONCLUSION

The major aim of the current study was to know that how individuals with Down syndrome in Mardan district get medical treatment, what medical issues they face, and what trend is manifested in demographical distributions. The findings confirmed that there are significant differences in the distribution of the above characteristics.The participants under observation were older, which confirmed that they were likely diagnosed and enrolled very late in the institutions. Which means that early diagnose still an issue to be addressed. The sample size showed that number of male students were more in number as compared to female students due to social factors rather biological influence. It was observed that some health problems like respiratory issues, thyroid and endocrine problems are strongly connected to the Down Syndrome.

This could mean that there are problems like not getting enough diagnoses or not having enough places to get medical care, but it doesn't mean that these conditions aren't real. It was concluded that access to healthcare facilities was not same. The common problem diagnosed was a high medical expense, and many children were not having regular medical checkups. Access to diverse therapies was out of reach to most of students. This shows that accessibility to facility is important and that just having services available is not the solution. The findings revealed that participants showed systemic as well medical problems. It was concluded early diagnose, family awareness and access to health facilities is very important especially in rural areas.

Recommendations

Based on the findings enlisted above following conclusions are drawn to improve the living conditions of the students with Down Syndrome. Development of a mechanism to diagnose the kids at the early stage. It should be possible to run an awareness campaign along with health department to diagnose new born kids in Down Syndrome characters. It is suggested to improve screening services at the basic health units. Second, the care takers and parents should be aware through media, through cell phone ringtones. In this way, basic awareness efforts be performed by special school, social workers, and medical professional. Societal taboos may be encountered through religious speakers and school teachers. Students with DS be paid by government or provision be provided in Sehat Card facility for treatment and therapies.

Trained personals may be produced to deal children with Down Syndrome as market is deficient with experts in this field. Training programs must be initiate for healthcare workers and teachers. t is a good idea to create suitable medical record systems. Some of the disorders in this study may not have been diagnosed or recorded correctly. Better recordkeeping and regular health checks can help us learn more about real health of kids with Down syndrome. Finally, additional research with larger samples and in diverse regions should be carried out. This study was limited to one institue, thus further research is needed to get a better picture. This can help lawmakers come up with better programs to help the people and improve their health.

REFERENCES

Adachi, T., El-Hattab, W., Jain, R., Crespo, A., Lazo, C. I., Scarpa, M., & Wattanasirichaigoon, D. (2023). Enhancing equitable access to rare disease diagnosis and treatment around world: A review of evidence, policies & challenges. *International journal of environmental research and public health*, *20*(6), 4732.

Ahmed, R., & Tamim, M. T. R. (2025). Understanding Down Syndrome: Global Perspectives, Advances, and Challenges in Context of Bangladesh. *THRIVE Health Science Journal,* *2*(1), 16-28.

Akhtar, F., & Bokhari, S. R. A. (2023).The Down syndrome. In: Stat Pearl Treasure Island (FL): Available fromhttps://www.ncbi.nlm.nih.gov/books/NBK526016/.

Alanazi, A. S., Alanazi, A. S., & Benlaria, H. (2025). Balancing costs and care: a healthcare cost analysis for families of children with Down syndrome in Saudi Arabia. *Frontiers in Public Health*, *13*, 1651534.

Ali, S., & Haider, S. K. (2024). Exploring the impact of interventions on the psychological well-being of parents of children with down syndrome in khyber pakhtunkhwa, pakistan. *Journal of Ayub Medical College, JAMC*, 36 (1): 78–82.

Alkhushi, N. (2024). The detailed profile of congenital heart diseases in 254 children with Down syndrome in Saudi Arabia. *Cardiothorac Surgery*, 32: 1.

Antonarakis, E., Lyle, R., Dermitzakis, E. T., Reymond, A., & Deutsch, S. (2020). Chromosome 21 and Down syndrome: from genomics to pathophysiology. *Nature Reviews Genetics*, 21(9): 560–574.

Asim, A., Kumar, A., Muthuswamy, S., Jain, S., & Agarwal, S. (2015). "Down syndrome: an insight of the disease". *Journal of biomedical science,* 22(1): 41.

Benjak, T., Vuljanić, A., Draušnik, Ž., Barišić, I., Vuković, D., & Vuletić, G. (2025). Prevalence of Down Syndrome in Croatia in the Period from 2014 to 2024. *Medicina*, *61*(11), 1934.

Bull, M. J. (2020). Down Syndrome. *The New England journal of medicine*, 382(24): 2344–2352. https://doi.org/10.1056/NEJMra1706537

Fortea, J., McGlinchey, E. E., Espinosa, J. M., & Rafii, M. S. (2024). Addressing challenges in health care and research for people with Down syndrome. *The Lancet*, *403*(10439), 1830-1833.

Fried, C. (2017). Equality and rights in medical care. In *Health Rights* (pp. 3-8). The Routledge.

Grieco, J., Pulsifer, M., Seligsohn, K., Skotko, B., & Schwartz, A. (2015). Down syndrome: Cognitive and behavioral functioning across the lifespan.

Ijezie, O. A., Healy, J., Davies, P., Ballester, E., & Heaslip, V. (2023). Quality of life in adults with Down syndrome: A mixed methods systematic review. *Plos One*, *18*(5), e0280014.

Kazemi, M., Salehi, M., & Kheirollahi, M. (2016). Down syndrome: current status, challenges and future perspectives. *International journal of molecular and cellular medicine*, *5*(3), 125.

Krahn, G. L., Hammond, L., & Turner, A. (2006). A cascade of disparities: health and health care access for people with intellectual disabilities. *Mental Retardation and Developmental Disabilities Research Reviews,* 12(1): 70– 82.

LeJeune J, Gautier M & Turpin R. (1959). Study of somatic chromosomes from 9 mongoloid children [French]. *C. R. Hebd. Seances. Academic Sciences,* 248: 1721– 1722.

Loane, M., Morris, J. K., Addor, C., Arriola, L., Budd, J., & Doray, B. (2013). The Twenty-year trends in the prevalence of Down syndrome and other trisomies in Europe. *European Journal of Human Genetics,* 21(1): 27–33.

Marilyn, J., Trotter, T., Santoro, S.L., Christensen, C., & Grout, R.W.(2022). The Council On Genetics; Health Supervision for Children and Adolescents With Down Syndrome. *Pediatrics,* 149 (5): e2022057010.

McGlinchey, E., Tewolde, S., He, A., Fortea, J., Mbakile-Mahlanza, L., Tefera, A., ... & Merali, Z. (2025). Down syndrome in Africa: Challenges, opportunities, and future directions. *Alzheimer's & Dementia*, *21*(6), e70388

Otterman, D.L., Koopman-Verhoeff, M.E., & White, T.J. (2019). Executive functioning and neuro-developmental disorders in early childhood: a prospective population-based study. *Child Adolesc Psychiatry Ment Health* 13: 38.

Paladini, D., Volpe, P., Russo, G., Vassallo, M., & Sglavo, G. (2000). The association between congenital heart disease and Down syndrome. *Ultrasound in Obstetrics & Gynecology*, 16(3): 199–203.

Santoro, J. D., Pagarkar, D., Chu, D. T., Rosso, M., Paulsen, K. C., Levitt, P., & Rafii, M. S. (2021). Neurologic complications of Down syndrome: a systematic review. *Journal of Neurology*, 268*:* 4495-4509.

Van Dyck, P. C., Kogan, M. D., McPherson, M. G., Weissman, G. R., & Newacheck, P. W. (2004). Prevalence & characteristics of children with special health care needs. *Archives of pediatrics & adolescent medicine*, *158*(9), 884-890.

Ye, E., Wu, E., & Han, R. (2025). Global, regional, and national impact of Down syndrome on child and adolescent mortality from 1980 to 2021, with projections to 2050: a cross-sectional study. *Frontiers in Public Health*, *13*, 1554589.