BIODIVERSITY REPORTS
Article Review
Journal homepage: https://biodiversity.crcjournals.org/

Awareness and Prevalence of Common Genetic Disorders in Rural and Urban Communities of Jammu Region

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Received 16 January 2025 | Revised 11 February 2025 | Accepted 18 March 2025 | Available Online April 13, 2025

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Citation: Sanjeet Kaur (2025). Awareness and Prevalence of Common Genetic Disorders in Rural and Urban Communities of Jammu Region. ***Biodiversity Reports An International Journal***. **DOI: https://doi.org/10.51470/BDR.2025.04.01.11**

Abstract

Genetic disorders constitute a significant public health challenge worldwide, particularly in developing countries where limited awareness, inadequate diagnostic facilities, and insufficient genetic counseling services contribute to delayed diagnosis and poor disease management. The Jammu region, characterized by diverse ethnic populations, geographical heterogeneity, and varying socioeconomic conditions, presents unique challenges in understanding the burden of inherited disorders. Factors such as endogamy, consanguinity in specific communities, and limited access to specialized healthcare may increase the occurrence of genetic abnormalities while reducing opportunities for early detection. Rural populations often experience lower awareness regarding hereditary diseases, prenatal diagnosis, and genetic counseling compared with urban populations, although urban communities may benefit from improved healthcare infrastructure and educational resources. This review examines the awareness and prevalence of common genetic disorders in rural and urban communities of the Jammu region, focusing on chromosomal abnormalities, single-gene disorders, congenital anomalies, hereditary blood disorders, and inherited metabolic diseases. The article further discusses epidemiological trends, associated risk factors, diagnostic approaches, public awareness, healthcare challenges, and strategies for strengthening community-based genetic services. Improved public education, newborn screening, carrier testing, prenatal diagnosis, and integration of genetic counseling into primary healthcare can substantially reduce the burden of hereditary diseases in the region. Existing evidence indicates that Jammu and Kashmir likely has a substantial but under-characterized burden of inherited disorders because of population structure, endogamy, and limited diagnostic resources.

Keywords: *Genetic disorders, Jammu region, Rural health, Urban health, Genetic awareness, Cytogenetics, Down syndrome.*

1. Introduction

Genetic disorders are an important cause of morbidity, mortality, disability, and reduced quality of life across all age groups. Advances in molecular genetics, cytogenetics, and genomic medicine have significantly improved the diagnosis and management of hereditary diseases; however, their benefits remain unevenly distributed, particularly in low-resource settings. Early diagnosis and appropriate genetic counseling are essential for reducing disease burden, improving reproductive decision-making, and facilitating timely medical intervention. India possesses remarkable genetic diversity owing to its heterogeneous population, numerous endogamous communities, and varying cultural practices. Several hereditary disorders, including chromosomal abnormalities, hemoglobinopathies, inherited metabolic diseases, and monogenic disorders, occur with varying frequency among different regions and communities [1]. The Jammu region represents a unique demographic setting comprising rural villages, urban centers, tribal populations, and geographically isolated communities. Differences in education, healthcare accessibility, socioeconomic status, and health awareness contribute to variation in disease detection and management between rural and urban populations.

Although several hospital-based and regional studies have documented cases of Down syndrome, Turner syndrome, Klinefelter syndrome, thalassemia, and other inherited disorders in Jammu and Kashmir, comprehensive community-level epidemiological data remain limited [2-3]. The region's population structure, including endogamy and consanguinity in some communities, suggests a potentially higher burden of recessive genetic diseases than is currently documented.

2. Genetic Disorders: Definition and Classification

Genetic disorders arise from abnormalities in chromosomes, single genes, mitochondrial DNA, or multifactorial interactions involving both genetic and environmental factors. These disorders may be inherited from one or both parents or may occur due to spontaneous mutations during embryonic development.

Genetic disorders are broadly classified into four categories:

- **Chromosomal disorders:** Down syndrome, Turner syndrome, Klinefelter syndrome, Edwards syndrome, and Patau syndrome.
- **Single-gene disorders:** Thalassemia, sickle cell disease, cystic fibrosis, Duchenne muscular dystrophy, and hemophilia.

- **Multifactorial disorders:** Congenital heart disease, cleft lip and palate, neural tube defects, diabetes, and hypertension.
- **Mitochondrial disorders:** Rare inherited diseases affecting cellular energy metabolism.

Among these, chromosomal abnormalities and hereditary blood disorders represent some of the most frequently diagnosed genetic conditions encountered in clinical practice in northern India.

3. Common Genetic Disorders Observed in the Jammu Region

Hospital-based studies and regional genetic investigations indicate that several inherited disorders are encountered in Jammu and surrounding areas. Down syndrome remains the most frequently reported chromosomal abnormality, followed by Turner syndrome and Klinefelter syndrome. Cases of Patau syndrome, congenital developmental delay associated with chromosomal abnormalities, and inherited neurological disorders have also been documented. The prevalence of rare recessive disorders is thought to be influenced by population structure and endogamy, although comprehensive population-based estimates remain limited [3-4]. Hereditary blood disorders such as β-thalassemia and sickle cell disease are of increasing clinical importance because carrier individuals often remain asymptomatic while transmitting disease-associated alleles to subsequent generations. Congenital anomalies including cleft lip, congenital heart disease, neural tube defects, and developmental disabilities also contribute substantially to pediatric morbidity.

Table 1: Common Genetic Disorders Reported in the Jammu Region

Genetic Disorder	Type	Major Clinical Features
Down syndrome	Chromosomal	Intellectual disability, characteristic facial features
Turner syndrome	Chromosomal	Short stature, primary amenorrhea
Klinefelter syndrome	Chromosomal	Male infertility, hypogonadism
β-Thalassemia	Single-gene	Severe anemia
Sickle cell disease	Single-gene	Hemolytic anemia, painful crises
Congenital heart defects	Multifactorial	Structural cardiac abnormalities
Neural tube defects	Multifactorial	Defects of brain and spinal cord

4. Awareness of Genetic Disorders in Rural and Urban Communities

Public awareness plays a crucial role in the prevention, early diagnosis, and management of inherited disorders. In the Jammu region, awareness regarding genetic diseases varies considerably between rural and urban populations due to differences in educational attainment, healthcare accessibility, socioeconomic status, and exposure to health education programs. Urban residents generally have greater access to hospitals, diagnostic laboratories, prenatal screening facilities, and specialist consultations, resulting in higher awareness of hereditary diseases and available preventive measures. In contrast, many rural communities possess limited knowledge regarding inheritance patterns, carrier status, prenatal diagnosis, newborn screening, and genetic counselling [5-6]. Cultural beliefs, social stigma, financial constraints, and inadequate healthcare infrastructure often delay diagnosis and treatment. Consequently, affected families frequently seek medical care only after the appearance of severe clinical symptoms. Increasing community education through schools, primary healthcare centers, and public health campaigns can substantially improve awareness and encourage timely utilization of genetic services.

5. Factors Influencing the Prevalence of Genetic Disorders

The occurrence of genetic disorders within communities is influenced by multiple biological, demographic, and environmental factors. Advanced maternal age is a well-recognized risk factor for chromosomal abnormalities such as Down syndrome. Similarly, advanced paternal age has been associated with an increased frequency of certain genetic mutations [7-8]. Population-specific factors including endogamy, consanguinity, small population size, and founder effects may increase the prevalence of autosomal recessive disorders. Environmental influences such as exposure to radiation, industrial pollutants, pesticides, nutritional deficiencies, and maternal infections during pregnancy may also contribute to congenital abnormalities. Limited awareness, delayed diagnosis, and insufficient access to genetic counseling further increase the burden of hereditary diseases within vulnerable populations.

Table 2: Major Factors Associated with Genetic Disorders

Risk Factor	Potential Impact
Advanced maternal age	Increased chromosomal abnormalities
Consanguineous marriage	Higher risk of recessive disorders
Endogamy	Increased carrier frequency
Environmental pollutants	Congenital anomalies
Poor prenatal care	Delayed diagnosis
Limited awareness	Late treatment and poor outcomes

6. Diagnosis and Genetic Screening

Early diagnosis remains one of the most effective strategies for reducing morbidity associated with inherited disorders. Cytogenetic analysis using conventional karyotyping remains the primary diagnostic method for detecting numerical and structural chromosomal abnormalities such as Down syndrome, Turner syndrome, and Klinefelter syndrome. Recent advances in molecular genetics have introduced highly sensitive diagnostic techniques including fluorescence in situ hybridization (FISH), polymerase chain reaction (PCR), chromosomal microarray analysis (CMA), multiplex ligation-dependent probe amplification (MLPA), and next-generation sequencing (NGS). These technologies enable rapid identification of chromosomal abnormalities, gene mutations, and hereditary syndromes. Prenatal screening using ultrasonography, maternal serum screening, chorionic villus sampling, and amniocentesis provides opportunities for early detection of fetal genetic disorders [9-10]. Newborn screening programs further facilitate early diagnosis of inherited metabolic disorders and endocrine diseases, allowing prompt treatment and improved clinical outcomes.

7. Prevention and Public Health Strategies

Reducing the burden of genetic disorders requires coordinated public health initiatives involving healthcare professionals, educational institutions, government agencies, and community organizations. Public awareness campaigns should educate communities regarding hereditary diseases, prenatal care, carrier testing, and available genetic counseling services. Premarital carrier screening for hereditary blood disorders such as thalassemia, particularly among high-risk populations, can significantly reduce disease transmission. Strengthening maternal healthcare services, promoting folic acid supplementation, expanding newborn screening programs, and improving access to prenatal diagnosis are also important preventive measures [11-12]. Integration of genetic counseling into primary healthcare systems can assist families in understanding inheritance patterns, reproductive risks,

available diagnostic options, and disease management strategies. Continuous training of healthcare professionals in medical genetics will further improve early diagnosis and patient care.

9. Conclusion

Genetic disorders continue to represent an important public health concern in both rural and urban communities of the Jammu region. Chromosomal abnormalities, hereditary blood disorders, congenital anomalies, and single-gene diseases contribute significantly to childhood morbidity, disability, and long-term healthcare needs. While urban populations generally demonstrate greater awareness and improved access to diagnostic facilities, rural communities continue to face considerable challenges related to limited healthcare infrastructure, insufficient public awareness, delayed diagnosis, and inadequate genetic counseling services. Early diagnosis through cytogenetic and molecular genetic techniques, combined with effective newborn screening, prenatal diagnosis, and community-based genetic counseling, can substantially reduce the burden of hereditary diseases. Public health education, premarital carrier screening, strengthening of primary healthcare services, and expansion of specialized genetic laboratories are essential components of future disease prevention strategies. Further epidemiological studies involving both rural and urban populations are needed to establish accurate prevalence estimates for common genetic disorders in the Jammu region. Strengthening collaboration between healthcare providers, academic institutions, government agencies, and local communities will facilitate the development of comprehensive genetic healthcare programs. Such initiatives will improve early detection, enhance disease prevention, support informed reproductive decision-making, and ultimately contribute to better health outcomes and quality of life for affected individuals and their families.

References

1. Bittles, A. H., & Black, M. L. (2010). Consanguinity, human evolution, and complex diseases. *Proceedings of the National Academy of Sciences, 107*(Suppl. 1), 1779–1786. https://doi.org/10.1073/pnas.0906079106
2. Gardner, R. J. M., Sutherland, G. R., & Shaffer, L. G. (2018). *Chromosome abnormalities and genetic counseling* (5th ed.). Oxford University Press.
3. Kliegman, R. M., St. Geme, J. W., Blum, N. J., Shah, S. S., Tasker, R. C., & Wilson, K. M. (2020). *Nelson textbook of pediatrics* (21st ed.). Elsevier.
4. Kumar, P., & Clark, M. (2020). *Kumar & Clark's clinical medicine* (10th ed.). Elsevier.
5. Nussbaum, R. L., McInnes, R. R., & Willard, H. F. (2016). *Thompson & Thompson genetics in medicine* (8th ed.). Elsevier.
6. Puri, R. D., Singh, M., Suresh, S., et al. (2019). GenomeIndia: Cataloguing the genetic diversity of the Indian population. *Indian Journal of Medical Research, 151*(2–3), 192–193.
7. Verma, I. C., & Bijarnia-Mahay, S. (2015). The burden of genetic disorders in India and a framework for community control. *Community Genetics, 15*(4), 192–196.
8. World Health Organization. (2021). *Birth defects*. World Health Organization. https://www.who.int/news-room/fact-sheets/detail/birth-defects
9. World Health Organization. (2023). *Genomics and human health*. World Health Organization. https://www.who.int/health-topics/genomics
10. Yoon, P. W., Rasmussen, S. A., Lynberg, M. C., et al. (2001). The National Birth Defects Prevention Study. *Public Health Reports, 116*(Suppl. 1), 32–40.
11. Zaidi, S., & Brueckner, M. (2017). Genetics and genomics of congenital heart disease. *Circulation Research, 120*(6), 923–940. https://doi.org/10.1161/CIRCRESAHA.116.309140
12. Zlotogora, J. (2002). What is the birth defect risk associated with consanguineous marriages? *American Journal of Medical Genetics, 109*(1), 70–71. https://doi.org/10.1002/ajmg.10375