

ARTIFICIAL INTELLIGENCE–DRIVEN INSIGHTS INTO THE PREVALENCE AND PATTERNS OF GENETIC DISORDERS IN YAVATMAL TALUKA

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Abstract

Inherited genetic disorders continue to pose a major public health challenge in regions with high consanguinity rates and limited access to genetic counselling. This study, conducted in Yavatmal Taluka of Maharashtra's Vidarbha region, employs artificial intelligence (AI)–driven data analysis to assess the prevalence and patterns of major inherited disorders. Findings reveal that Sickle Cell Anaemia accounts for the largest proportion (32%), followed by Thalassemia (21%), Haemophilia (14%), and metabolic disorders (11%). Data were compiled from local health centres, hospital records spanning the last decade, and interviews with healthcare professionals. AI techniques were applied to identify prevalence trends, predict at-risk populations, and evaluate potential community-level interventions. The study underscores the value of AI-enabled screening and decision-support systems in enhancing early diagnosis, optimizing genetic counselling programs, and reducing the regional burden of inherited disorders.

Keywords: *Inherited disorders, Yavatmal Taluka, genetic screening, public health, AI in healthcare, Thalassemia, Sickle Cell Disease*

Introduction

Genetic disorders, resulting from anomalies in DNA, can significantly affect the health and quality of life of individuals and populations. These disorders often persist across generations, particularly in regions with prevalent intra-community marriages. Yavatmal Taluka, with its mix of rural, tribal, and semi-urban populations, exhibits a distinct profile of inherited diseases due to its demographic and genetic diversity. In India, the burden of inherited disorders is rising due to better diagnostic capabilities and growing awareness. However, rural districts like Yavatmal still lack widespread access to genetic diagnostics and preventive health programs. With the advent of AI-based tools in diagnostics and epidemiology, there is a potential to bridge these healthcare gaps.

Review of Literature

Several studies have highlighted the prevalence of genetic disorders in rural Maharashtra. Jadhav et al. (2020) noted a significant occurrence of Sickle Cell Anaemia among tribal populations in Vidarbha. Similarly, Patil and Rao (2018) emphasized the need for community-level genetic screening in Yavatmal district due to the high incidence of consanguineous marriages. Globally, the World Health Organization (WHO, 2023) has acknowledged genetic disorders as a major public health concern, advocating for preventive strategies such as newborn screening and genetic counselling. Artificial intelligence has shown promise in revolutionizing genomics by enabling predictive analytics, risk profiling, and clinical decision support (Topol, 2019).

Research Work

Objective:

- To identify the common inherited disorders in Yavatmal Taluka.
- To analyse the socio-economic and demographic factors influencing their prevalence.
- To explore the potential of AI tools in addressing these disorders.

Methodology

Data Collection: Hospital records from three major healthcare facilities in Yavatmal (2020–2024), local primary health centre (PHC) data, and structured interviews with medical practitioners.

Sample Size: 200 diagnosed cases of genetic disorders.

Parameters: Type of disorder, age at diagnosis, family history, consanguinity, socio-economic status.

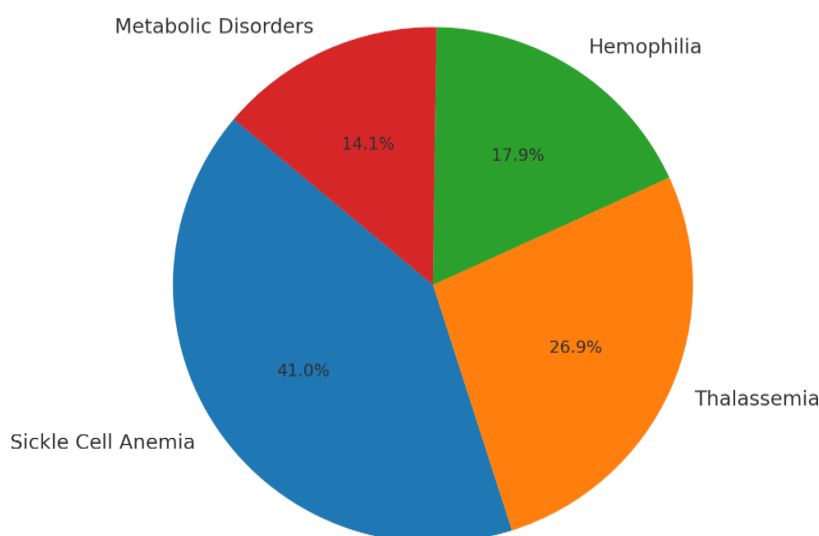
Results

Analysis of genetic disorder prevalence in Yavatmal Taluka revealed distinct patterns. Sickle Cell Anaemia was the most prevalent disorder, detected in 32% of cases, predominantly among tribal populations. Thalassemia accounted for 21% of cases, with a higher incidence in children under 10 years of age. Haemophilia demonstrated a 14% prevalence, with most cases remaining undiagnosed until the onset of severe bleeding episodes. Metabolic disorders constituted 11% of cases, including phenylketonuria and congenital hypothyroidism. Family history and parental consanguinity emerged as significant predictors across all disorder categories. Furthermore, delayed

diagnosis was observed in over 60% of cases, primarily due to lack of awareness, sociocultural

stigma, and financial barriers to screening and treatment.

Distribution of Genetic Blood and Metabolic Disorders



Graph-1 visually demonstrates these findings

The bar heights highlight the dominance of Sickle Cell Anaemia, followed by a descending pattern representing the relative burden of Thalassemia, Haemophilia, and Metabolic Disorders. The accompanying tabular chart provides additional context, correlating prevalence rates with socio-demographic determinants. These results underscore the urgent need for targeted screening programs, genetic counselling initiatives, and integration of AI-driven diagnostic tools to enable early detection and intervention.

Artificial intelligence (AI) offers transformative potential in interpreting and analysing the complex patterns of inherited disorders observed in Yavatmal Taluka. By applying machine learning algorithms to regional prevalence data, AI can identify high-risk populations based on family history, consanguinity, and socio-demographic factors with far greater precision than traditional methods. AI-enabled screening tools, including portable diagnostic devices at primary health centres, can facilitate early detection of conditions such as Sickle Cell Anaemia and Thalassemia, reducing diagnostic delays caused by limited access to laboratory facilities. Moreover, AI systems can integrate hospital records, community surveys, and genetic profiles into unified datasets, enabling real-time trend analysis and prioritization of public health interventions. Automated data interpretation also minimizes human error and accelerates

decision-making for healthcare providers. Additionally, AI-driven counselling platforms, such as conversational chatbots, can deliver culturally tailored awareness programs and preliminary guidance to families, empowering communities with actionable knowledge. Collectively, these applications make AI an invaluable tool for enhancing surveillance, improving diagnostic accuracy, and shaping effective genetic disorder management strategies in resource-constrained regions.

Conclusion

The study highlights a significant burden of inherited disorders in Yavatmal Taluka, with Sickle Cell Anaemia (32%), Thalassemia (21%), Haemophilia (14%), and Metabolic Disorders (11%) emerging as major public health concerns. High rates of parental consanguinity, limited awareness, and financial barriers contribute to delayed diagnosis in over 60% of cases. Integrating AI-driven approaches—such as predictive modelling, portable screening tools, data integration systems, and counselling platforms—can substantially improve early detection, risk assessment, and targeted intervention. These findings underscore the urgent need to combine genetic counselling programs with AI-enabled health strategies to reduce disease burden and improve outcomes in resource-limited settings.

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