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Assessing public awareness and understanding of genetic disorders in Chhindwara District, Madhya Pradesh, India

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Abstract
Genetic disorders result from gene mutations that impair body functions, and most remain untreatable. India experiences a high occurrence of these conditions due to its large population, high birth rates, and consanguineous marriages within certain communities. A cross-sectional study in the Chhindwara district of Madhya Pradesh examined comprehension and awareness of genetic diseases and available healthcare. The research found that Chhindwara inhabitants possess moderate knowledge about genetic disorders. Among 958 participants surveyed, epilepsy and sickle cell anaemia were more frequently recognized, while thalassemia and Down syndrome were less familiar. Misconceptions persisted, including the erroneous belief that genetic diseases are contagious. Only 25% of respondents were aware of local treatment options, yet 85.7% endorsed the establishment of local genetic counseling services. These results underscore the necessity for enhanced education, awareness, and accessibility to genetic counseling to boost public health outcomes.

Keywords: Genetic diseases, awareness, knowledge, Chhindwara, India

Introduction
Genetic diseases present significant economic, emotional, and health challenges for patients and their families. These disorders result from DNA sequence mutations that deviate from the norm [9] and [11]. While genetic diseases occur globally, India experiences a particularly high prevalence. They represent the second leading cause of infant mortality in the country, affecting 25-60 per 1,000 births [5]. Social factors, such as high rates of consanguineous marriages in certain communities, contribute to this prevalence [10]. Cousin marriages are common not only among Muslims but also in Hindu [1, 4] and tribal populations [6]. Many genetic conditions remain undiagnosed due to inadequate data collection and awareness [5]. The scarcity of specialized medical professionals and testing facilities further impedes the diagnosis of rare genetic disorders [2]. Research indicates that a substantial number of infants are born annually with genetic conditions, emphasizing the urgent need for improved diagnostic and management approaches. In developing nations such as India, prevention remains the most viable strategy for addressing the majority of genetic diseases. Preventive measures include community education, population screening, premarital genetic counseling, and prenatal diagnosis [7, 8]. Public awareness and understanding of genetic diseases are crucial for implementing effective treatments. This study aims to assess the knowledge and awareness of genetic diseases among residents of the Chhindwara district in Madhya Pradesh, India

Materials and Methods
Situated in the southwestern part of the Satpura Mountain range, Chhindwara district in Madhya Pradesh spans from 21.28 to 22.49 degrees North latitude and 78.40 to 79.24 degrees East longitude, covering an area of 11,815 square kilometres. According to the 2011 Census [3], the district's population totals 2,090,922, with 24.2% residing in urban areas and 75.8% in rural areas. The literacy rate stands at 85.7% in urban regions and 66.4% in rural areas. Hindus comprise 83.84% of the population (146,761 individuals), while Muslims account for 11.87% (20,772 individuals). The Census 2011 also reported that Scheduled Castes and Scheduled Tribes constituted 11.11% and 36.82% of the population, respectively [3].

Our research employed a cross-sectional approach using an electronic survey. We created the survey using Google Forms, translated it into Hindi and distributed it via WhatsApp and email to Chhindwara residents in Madhya Pradesh. The study was conducted over two months from March to April 2024. Participants were encouraged to take part through an introductory note highlighting the significance of their input. A total of 958 individuals participated in the study. We transferred all responses to Microsoft Excel spreadsheets for statistical analysis. The data were examined using descriptive statistics, primarily percentages, and all statistical figures were generated using Microsoft Excel tools.

Results and Discussion

The survey results provide insights into participants' awareness and experiences regarding genetic disorders. The study included 958 participants, with a higher proportion of females (57.5%) compared to males (42.5%). Most participants (60.8%) were between 20-40 years old, followed by 38.4% in the 15-20 age range. Most participants were from rural areas of Chhindwara district (47.5%), with smaller representations from the city (40.8%), tehsils (8%) and tribal regions (3.7%). A large majority (88.8%) of participants were unmarried. In terms of education, 75.4% had a university degree, while 21.6% had completed high school or higher secondary education. A detailed breakdown of the demographic profile of the study respondents is shown in Figure 1.

Family history of genetic conditions ranged from 0.8% for Thalassemia to 8.20% for Epilepsy. Sickle Cell Anemia (SCA) was the second most common genetic disease reported among participants (4.9%). Thalassemia (0.8%) and Down Syndrome (1%) were the least common genetic diseases reported. 11.3% of participants had someone in their family suffering from a physical or mental impairment or abnormality**.** The percentage of Colorblindness and Haemophilia was found to be 2.4 and 1.4 respectively (Table 1).

Regarding treatment availability, 25.8% believed local hospitals could treat all mentioned diseases, while 51.6% thought treatment was available for some diseases. Knowledge of prevention methods was limited, with only 21% knowing how to prevent all mentioned diseases. A significant majority (85.7%) supported government-provided genetic counselling at the local level. Interestingly, there was a misconception about disease transmission, with 44.8% believing genetic diseases could spread through infection. 79.9% agreed that if a parent has a genetic disease, there is a strong possibility of children inheriting it (Table 1).

These results suggest a relatively low prevalence of genetic disorders among participants' families. However, there appears to be a lack of comprehensive knowledge about treatment availability and prevention methods. The majority of participants recognize the importance of genetic counselling and understand the hereditary nature of genetic diseases, but there is some misconception about disease transmission through infection. These findings highlight the need for improved genetic education and counselling services to address misconceptions and enhance prevention efforts. However, the integration of genetic services into primary healthcare remains a challenge, necessitating further research and funding.

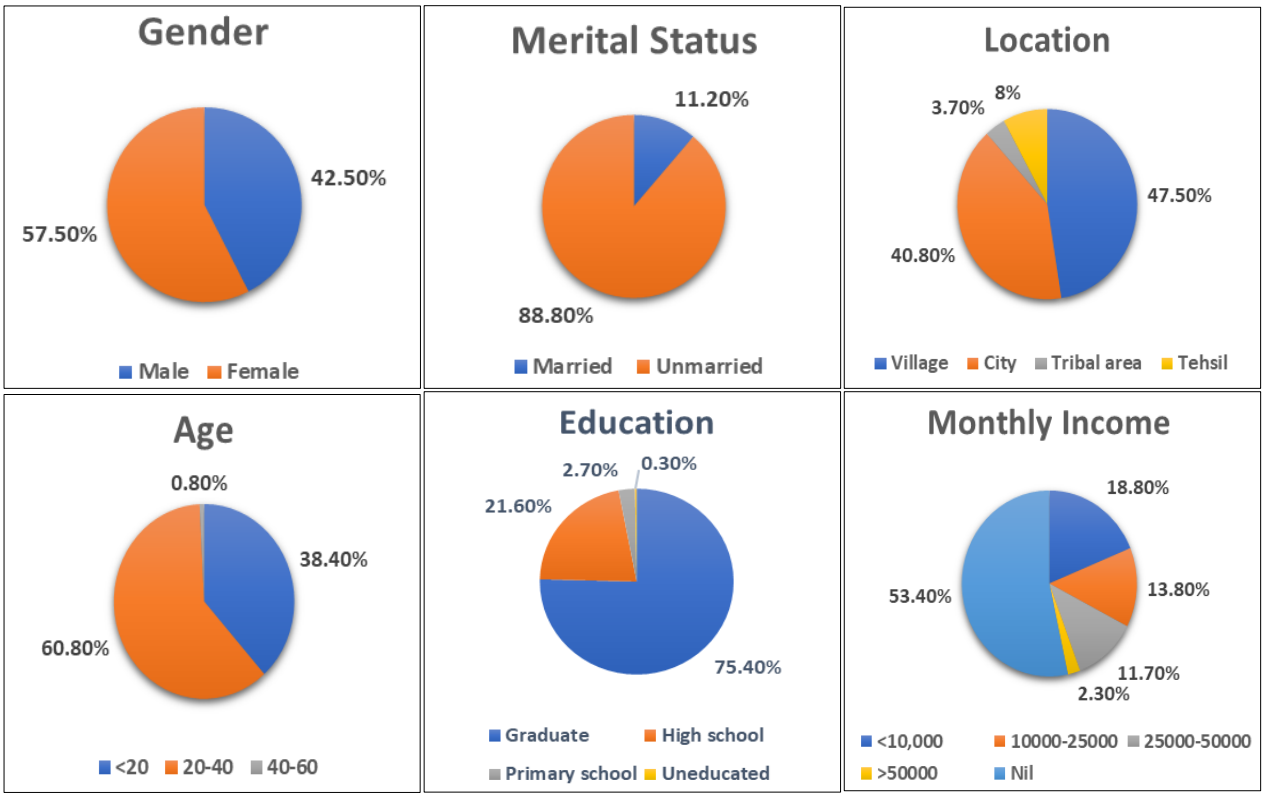

Fig 1: Demographic characteristics of the participants

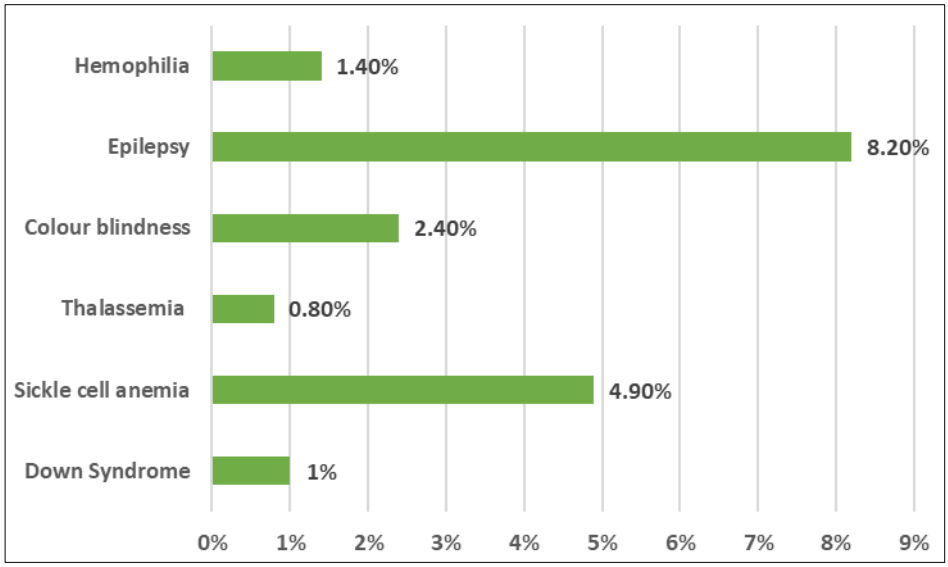

Fig 2: Family history of genetic conditions

Table 1: Participants' awareness regarding genetic disorders

Survey questions	Responses	%
Does any member of your family suffer from epilepsy?	Yes	8.20
	No	91.8
Do your family members have any genetic disease present in previous generations?	Yes	4.20
	No	95.8
Is there any person in your family who is mentally or physically weak or abnormal?	Yes	11.3
	No	88.7
Is anyone in your family suffering from Down Syndrome?	Yes	1.00
	No	99.0
Is anyone in your family or relative suffering from sickle cell anemia?	Yes	4.90
	No	95.1
Is anyone in your family or relative suffering from Thalassemia disease?	Yes	0.80
	No	99.2
Is anyone in your family or relative suffering from colour blindness?	Yes	2.40
	No	97.6
Is anyone in your family or relative suffering from Hemophilia?	Yes	1.40
	No	98.6
Are treatment facilities for all the above diseases available in the local hospital?	Yes	25.8
	No	22.7
	Treatment is available for some diseases	51.6
Do you know how to prevent all the above diseases?	Yes	21.0
	No	24.3
	There is some information	54.7
Do you think it is necessary that there should be a provision of counselling by the government at the local level to provide information related to the prevention of genetic diseases?	Yes	85.7
	No	14.3
Can genetic diseases be spread from one person to another through infection?	Yes	44.8
	No	55.2
Do you agree that if the mother or father has a genetic disease then there is a strong possibility of the children getting the same disease?	Yes	79.9
	No	20.1
Do you agree that there is a possibility of this disease developing in children even if the mother or father is healthy?	Yes	61.4
	No	38.6
Should the boy and girl be tested for genetic diseases before marriage?	Yes	71.2
	No	28.8
Is a testing facility for genetic diseases available in your city?	Yes	58.8
	No	41.2

Conclusion

The survey results reveal varying levels of awareness and understanding of genetic disorders across different demographic groups among participants in the Chhindwara district. While genetic conditions have a relatively low reported prevalence, significant knowledge gaps exist regarding treatment options and prevention methods. The majority of respondents recognise the importance of genetic counselling and the hereditary nature of genetic diseases. These findings underscore the need for targeted educational initiatives and improved genetic services in the region to address knowledge gaps and misconceptions, ultimately enhancing genetic healthcare delivery and awareness.

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