ORIGINAL ARTICLE

Impact of genetic counselling on awareness and mental health in sickle cell families

Sagar Bayaskar[1*], Aishwarya Mahalle[1]

ABSTRACT

Background: Sickle cell disease (SCD) is a hereditary blood disorder with high prevalence among tribal populations in India. This study evaluates the impact of genetic counselling on disease awareness and mental health in India's tribal communities, where limited access to such services exacerbates disease burden.

Methods: A qualitative, cross-sectional study involved 35 participants aged 20-44 years in a high-risk tribal region of Maharashtra, India. Participants included one SCD patient, one caregiver, one community health worker, two healthcare professionals (a hematologist and a psychiatrist), and 30 community members. Semi-structured interviews, developed through literature review and pilot-tested, explored SCD knowledge, preventive strategies, and psychological well-being. Thematic analysis compared outcomes between counseled (n = 18) and non-counseled (n = 17) participants.

Results: Counseled participants demonstrated greater knowledge of SCD's genetic basis (94% *vs.* 24%), symptoms (89% *vs.* 35%), and preventive measures (83% *vs.* 12%), such as premarital screening. They also showed higher engagement in community initiatives (83% *vs.* 12%) and reported reduced anxiety (78% *vs.* 29%), improved coping, and greater emotional resilience. Non-counseled participants often misidentified SCD as an infection (59%) or general anemia (41%) and reported higher stress (71%) and sleep disturbances (53%).

Conclusion: Genetic Counselling significantly enhances SCD awareness and psychological resilience in tribal communities. Expanding culturally tailored counselling services is critical to reducing SCD burden in underserved areas.

Keywords: Sickle cell disease, genetic counselling, mental health, preventive health services, health knowledge.

Introduction

Sickle cell disease (SCD) is a hereditary hemoglobinopathy characterized by abnormally shaped red blood cells, causing anemia, recurrent pain crises, organ damage, and other complications (1). In India, SCD prevalence ranges from 10% to 40% among tribal populations, such as the Gond and Bhil tribes, posing significant health and social challenges (2,3). Despite screening programs, low awareness in these communities leads to delayed diagnoses, suboptimal management, and psychosocial distress (3). While genetic counselling has been studied globally, its impact on awareness and mental health in India's tribal populations remains underexplored, representing a critical research gap.

Genetic counselling provides families with information on SCD's inheritance patterns, management strategies, and reproductive options, empowering informed decision-making (4). It promotes preventive measures, such as premarital and family screening, and community-based initiatives (5). Additionally, counselling mitigates the psychological burden of SCD, including anxiety, depression, and social isolation, by reducing uncertainty and fostering coping strategies (4).

This study assesses the impact of genetic counselling on SCD awareness and mental health outcomes in a high-risk tribal region. Through in-depth interviews with diverse stakeholders, it underscores counselling's role in enhancing knowledge, promoting preventive practices,

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and supporting psychosocial well-being in underserved communities.

SCD is a major public health concern in India, particularly in tribal regions of Maharashtra, Odisha, and Chhattisgarh, where prevalence is highest among marginalized groups (3). India's National Sickle Cell Anemia Elimination Mission (2023) aims to screen 70 million people in high-prevalence districts and eliminate SCD as a public health issue by 2047 (6).

Genetic counselling enhances understanding of SCD's genetic basis, transmission risks, and preventive measures, such as premarital and antenatal screening (7). It improves knowledge, facilitates informed reproductive decisions, and increases screening participation (8). However, cultural barriers (e.g., stigma around hereditary diseases) and logistical challenges (e.g., limited healthcare access) often hinder their effectiveness in tribal settings (5).

Counselling also alleviates SCD's psychological toll, including stress, stigma, and anxiety due to limited disease understanding (9). A systematic review found that comprehensive counselling reduces uncertainty and improves mental health outcomes (10). An Indian study by Patel et al. (11) reported reduced anxiety and enhanced emotional preparedness following counselling in high-risk regions, though it focused on urban settings. Community-based approaches integrating counselling with awareness campaigns and digital tools have proven effective in addressing knowledge gaps and psychosocial concerns (5,12). These findings highlight the need for accessible, culturally sensitive counselling to manage SCD in vulnerable populations.

This study assesses the impact of genetic counselling on SCD awareness and mental health outcomes in a high-risk tribal region. Through in-depth interviews with diverse stakeholders, it underscores counselling's role in enhancing knowledge, promoting preventive practices, and supporting psychosocial well-being in underserved communities.

Methodology

Study design

This qualitative, cross-sectional study used in-depth interviews to explore genetic counselling's impact on SCD awareness and mental health outcomes in tribal families.

Study setting and participants

The study was conducted in a high-risk tribal region in Maharashtra, India, with elevated SCD prevalence. Thirty-five participants aged 20-44 years were purposively selected to represent diverse SCD experiences: one SCD patient, one caregiver, one community health worker (CHW), two healthcare professionals (a hematologist and a psychiatrist), and 30 community members. The limited number of patients and caregivers reflects the region's low diagnosis rate, as only one confirmed SCD patient was accessible through local health centers.

Participant recruitment

Participants were recruited through community health centers, SCD screening programs, and tribal outreach networks. CHWs and healthcare professionals facilitated introductions to patients, caregivers, and community members. Eligibility criteria included age (20-44 years), residence in the high-risk area, and varying SCD exposure (e.g., diagnosis, family history, or community involvement). Of the 35 participants, 18 received genetic counselling via screening programs or health centers, and 17 did not. Recruitment continued until thematic saturation, confirmed when no new themes emerged after three consecutive interviews (achieved after ~30 interviews, with five additional interviews for confirmation).

Data collection

Semi-structured interviews were conducted in person (at community centers or participants' homes) or via telecommunication, based on accessibility. The interview guide, developed through literature review and pilot-tested with two community members for clarity, explored SCD knowledge, genetic counselling experiences, preventive strategies (e.g., premarital screening), and psychological impacts (e.g., stress, coping strategies). Interviews, conducted in Hindi and regional tribal dialects by a bilingual researcher with real-time translation for non-native speakers, lasted 30-60 minutes and were audio-recorded with consent. Transcriptions in local languages were translated to English and back-translated for accuracy. An English-speaking co-researcher ensured quality. Neutral, open-ended questions (e.g., "Can you describe what you know about SCD?") minimized social desirability bias.

Data analysis

Thematic analysis was applied using an inductive approach. Transcripts were manually coded to identify themes: (i) SCD awareness and knowledge, (ii) engagement with preventive initiatives, and (iii) mental health impacts. Responses were compared between counseled (n = 18) and non-counseled (n = 17) participants. Two researchers independently coded transcripts, resolving discrepancies through discussion to ensure inter-coder reliability. Key themes were quantified (e.g., percentage of participants reporting specific outcomes) to enhance clarity.

Ethical considerations

Ethical approval was obtained from the Institutional Ethics Committee of the Datta Meghe Institute of Higher Education and Research, Wardha (Reference no. DMIHER(DU)/IEC/2025/795). The study was conducted in full compliance with the Declaration of Helsinki. Participation was voluntary, anonymity was maintained, and participants could withdraw at any time without penalty. All participants provided written informed consent prior to their inclusion in the study. They were informed about the study's purpose, procedures, potential risks, and their right to withdraw at any time without consequences.

Results

Awareness and knowledge of sickle cell disease

Counseled participants ($n = 18$) showed robust knowledge, with 94% (17/18) understanding SCD's genetic basis and 89% (16/18) accurately describing symptoms (e.g., anemia, pain crises, fatigue) as illustrated in Figure 1. The SCD patient stated, "Counselling clarified that SCD is inherited from both parents' genes, not caused by food or water." The hematologist noted, "Counselling helps families grasp the importance of screening." Conversely, only 24% (4/17) of non-counseled participants recognized SCD's hereditary nature, with 59% (10/17) mistaking it for an infection and 41% (7/17) for general anemia. A non-counseled community member said, "I thought it was weakness from a poor diet."

Awareness of preventive measures and community initiatives

Counseled participants demonstrated greater awareness of preventive strategies, with 83% (15/18) familiar with premarital screening, family testing, and community initiatives (e.g., school campaigns, health center screenings) as shown in Figure 1 and Table 1. The caregiver remarked, "Counselling taught us to test before marriage; we now encourage others to do the same." The CHW reported, "Counselling's increases family participation in screening camps." Only 12% (2/17) of non-counseled participants knew about preventive measures, with one stating, "I didn't know tests could prevent this; we just manage the pain."

Impact on mental health

Counseled participants reported improved mental health, with 78% (14/18) experiencing reduced anxiety, better coping strategies, and greater emotional resilience, as summarized in Table 1. The SCD patient shared, "Counselling made me feel less scared about my child's future." The psychiatrist noted, "Counselling helps families process stigma and build resilience." Conversely, non-counseled participants reported higher stress (71%, 12/17), sleep disturbances (53%, 9/17), and isolation. A non-counseled community member said, "I can't sleep worrying about why this disease hit us; it feels like a curse."

Discussion

This study underscores genetic counselling's pivotal role in enhancing SCD awareness and mental health in India's tribal communities. Counseled participants' superior knowledge of SCD's genetic basis (94% *vs.* 24%) and preventive measures (83% *vs.* 12%) aligns with global evidence that counselling promotes informed decision-making and screening participation (7,8). However, cultural barriers, such as stigma surrounding hereditary diseases, and logistical challenges, like limited healthcare access in tribal areas, may reduce counselling's effectiveness (5).

The mental health benefits, with 78% of counseled participants reporting reduced anxiety and improved

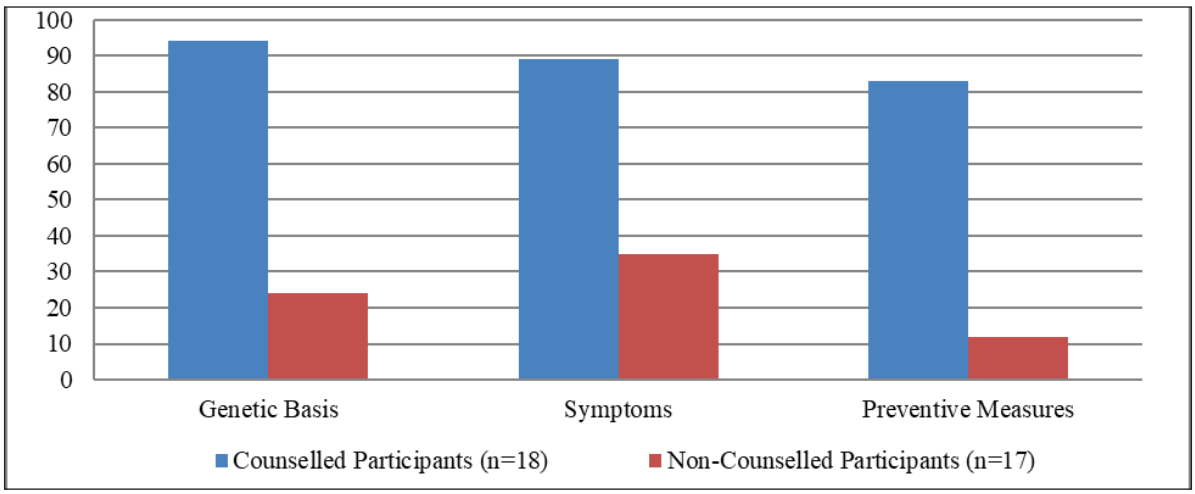

Figure 1. *Awareness of sickle cell disease among participants bar chart comparing awareness levels between counselled (n = 18) and non-counselled (n = 17) participants across three knowledge areas: genetic basis (94% vs. 24%), symptoms (89% vs. 35%), and preventive measures (83% vs. 12%).*

Table 1. *Summary of key outcomes: counselling versus no counselling.*

Outcome	Counselled (n = 18)	Non-counselled (n = 17)
Knowledge of SCD	94% (17/18) understood genetic basis; 89% (16/18) knew symptoms	24% (4/17) understood genetic basis; 35% (6/17) knew symptoms
Preventive measures	83% (15/18) aware of screening and community initiatives	12% (2/17) aware of preventive strategies
Mental health	78% (14/18) reported reduced anxiety, better coping strategies, and greater emotional resilience	71% (12/17) reported stress; 53% (9/17) had sleep disturbances

coping, corroborate findings that counselling mitigates uncertainty and stigma (9,10). The contrast with non-counseled participants (71% reporting stress, 53% with sleep disturbances) highlights SCD's psychosocial toll without support. This study extends Patel et al.'s urban-focused findings, emphasizing counselling's impact in tribal contexts (11).

The results support India's National Sickle Cell Anemia Elimination Mission (2023), which prioritizes screening and prevention (6). Integrating counselling into routine screening camps, training CHWs to deliver basic counselling in local dialects, and developing mobile-based audio-visual counselling apps for low-literacy populations could enhance the Mission's impact. Addressing tribal-specific challenges, such as nomadic lifestyles and low literacy, is critical for scalability (5).

Limitations include the qualitative design's limited generalizability and the small sample of patients (n = 1) and caregivers (n = 1), reflecting low diagnosis rates but restricting stakeholder perspectives. Selection bias may exist, as participants recruited via health centers may be more health-engaged. The cross-sectional design precludes assessing long-term impacts. Although neutral questioning minimized social desirability bias, self-reported data may still be influenced.

Future research should use mixed-methods designs to validate findings across diverse populations and longitudinal studies to evaluate sustained impacts. Exploring digital or community-based counselling models tailored for low-literacy tribal populations could identify scalable strategies.

Conclusion

Genetic counselling significantly enhances SCD awareness, promotes preventive practices, and fosters psychological resilience in tribal communities. Expanding culturally tailored counselling services, integrated with national SCD programs, is essential to reduce disease burden and improve quality of life in underserved areas.

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List of Abbreviations

CHW Community health worker
SCD Sickle cell disease

Conflict of interest

The authors of this article have no affiliations with or involvement in any organization or entity with any financial interest or non-financial interest in the subject matter or materials discussed in this manuscript.

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None.

Author contributions

Both authors contributed equally to the study design, data collection, analysis, and manuscript preparation.

Consent to participate

Informed consent was obtained from all participants, ensuring voluntary participation and the right to withdraw at any stage.

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