Journal of Community Genetics (2023) 14:345–353
https://doi.org/10.1007/s12687-023-00661-z

REVIEW

Genetic counseling in sickle cell disease: Insights from the Indian tribal population

Pooja Aggarwal[1] · **Deepa Bhat**[1]

Received: 7 March 2023 / Accepted: 25 July 2023 / Published online: 4 August 2023

Abstract

Sickle cell disease (SCD) is an inherited disorder of hemoglobin. With an overall prevalence of 4.3%, India is the second-largest hub of SCD after Africa. Genetic counseling (GC) is the most cost-effective intervention to reduce the burden of a genetic disease including SCD. Even though GC's role in reducing SCD birth prevalence is well established, it is still not incorporated into Indian national policy and is unavailable to most Indians approaching their marriageable age and childbirth. GC perception and efficacy have also not been explored yet among young adults, especially in Indian tribal communities. Counseling in these communities requires careful consideration of their socioeconomic, cultural, and ethical values. Community engagement with local tribes and healthcare infrastructure in a multitier approach is essential for an effective GC. This review aims to provide healthcare providers and genetic counselors with the essentials of GC in the prevention and management of SCD among tribal communities based on the author's counseling experience in South India.

Keywords Sickle cell disease · Genetic counseling · Tribal communities · Screening · Guidelines

Introduction

Sickle cell disease (SCD) is a monogenic inherited disorder of hemoglobin (Hb). The beta-globin chain of an oxygen-carrying normal Hb (HbAA) molecule is coded by two alleles (βAβA) inherited one from each of the parents. SCD manifests with the homozygous inheritance of a point mutation (A → T) in the beta-globin gene (β-globin), which results in the substitution of negatively charged glutamic acid with hydrophobic valine at the 6th position of the β-globin chain (Pauling et al. 1949). The resulting defective globin tetramer is poorly soluble when deoxygenated and modulates the structural and functional properties of red blood cells (RBCs) leading to a variety of pathological manifestations such as infarction, anemia, inflammation, hypercoagulability, oxidative stress, hemolysis, vascular endothelial dysfunction, and vaso-occlusive and pain crises (Bunn 1997; Porter and Garbowski 2013; Serjeant 2016).

When only one allele of β-globin gene is affected with sickle cell mutation, it is called sickle cell trait or carrier and generally asymptomatic (Headings and Fielding 1975). The Indian sickle cell haplotype is closely associated with the Arab-Indian or Asian haplotype which is presumed to have high fetal hemoglobin (Hb F) with a milder clinical presentation (Serjeant 2016; Kulozik et al. 1986).

Over 70 years ago, the first Indian case of SCD was reported in a tribal community of Nilgiri Hills located in South India. With an overall prevalence of 4.3% and the second-highest predicted SCD births of 44,000 each year after Africa, India has a huge burden of SCD (Rees and Brousse 2016). Disconcertingly, many studies have found that the SCD prevalence is still higher among Indian tribal tribes, reaching up to 40–55% in some communities. The higher prevalence is correspondingly accompanied by a higher infant and childhood morbidity and mortality rate among Indian tribals given their poorer access to healthcare. With a population of about 100 million, India has the single largest tribal population in the world (Census of India, 2011). Hence, there is an urgent need to devise newer strategies for lowering SCD prevalence as well as morbidity among deprived tribal communities.

The Ministry of Health and Family Welfare in India has identified SCD as a disease of national importance. The

✉ Deepa Bhat
deepabhat@jssuni.edu.in

[1] Department of Anatomy, JSS Medical College, JSS Academy of Higher Education & Research, Mysore, Karnataka, India

Indian National Sickle Cell Mission (NSCM), launched in November 2022, aims to lower the prevalence of SCD through a coordinated strategy that includes widespread screening, raising awareness, and creating a national SCD registry. Recently, the Indian government has launched National Sickle Cell Mission, which "aims to eradicate sickle cell anemia from India in a mission mode by 2047" (National Sickle Cell Anemia Elimination Mission 2023). This will include awareness creation, universal screening of seventy million people in the age group of 0–40 years in tribal areas, and premarital counseling through collaborative efforts of central and state governments (National Health Mission 2023; National Sickle Cell Anemia Elimination Mission 2023). It may be a herculean task to "eradicate" SCD by 2047 in India, but with timely action such as awareness, carrier screening programs, and GC, the SCD burden can be greatly reduced in the Indian population (Sinha et al. 2020).

Given that SCD is an inherited disease, providing genetic counseling (GC) is essential in minimizing its burden. In this review, we describe the need for GC and the challenges faced by counselors. Next, we explain various levels of GC for SCD and their importance. We also provide healthcare workers and genetic counselors with the GC toolkit for the prevention and management of SCD among tribal communities based on the author's counseling experience in South India. Finally, we discuss how collaboration between the government and private sectors can herald a change in the field of GC & SCD management.

Genetic counseling in SCD

Who are genetic counselors?

GC is a communication process by which the counselor ensures clients gain a detailed understanding of genetic diseases while carefully considering their emotional state (Ellington et al. 2011). During counseling, genetic counselors analyze and provide information about how SCD might affect a patient or his/her family. A counselor collects personal and family health history and determines what are the chances of a person or his/her family member inheriting SCD. Based on this information, the counselor can help individuals decide which genetic test to undergo for a confirmed diagnosis. Finally, counselors help patients in interpreting and understanding the implications of genetic reports, especially if a patient is planning for future pregnancies. Post-diagnosis, counselors can also help in linking the patient with a relevant specialist for the management of the disease (Mohanty and Das 2011). However, at all stages of GC, the autonomy of individuals and their decisions must be ensured by the counselors (Schmidtke and Cornel 2020).

Why do we need them?

According to a WHO (2011) report, GC is the most cost-effective intervention to lessen the burden of SCD. Together with carrier screening and prenatal diagnosis, genetic counselors have been recognized as an essential part of SCD control efforts. The counselors also ensure that the etiology, clinical symptoms, and management of SCD—including the use of hydroxyurea and bone marrow transplant—are clearly communicated. Hence, it is imperative to intensify community and state efforts for the establishment of viable SCD genetic counseling clinics backed by equally robust screening facilities.

Current understanding of GC in SCD

Though the state health department and various NGOs have made significant progress in SCD screening across various parts of India, most of them provide limited GC. Even though GC's role in reducing SCD birth prevalence is well established, it is still not incorporated into Indian national policy and is unavailable to most Indians approaching their marriageable age and childbirth. The importance of GC itself appears to be poorly understood by many practitioners, nurses, and other healthcare workers. As a result, India trails other nations in the development of general GC, let alone SCD-specific counseling for tribal communities. According to Choi and Kim (2014), nurses with strong genetic counseling knowledge are better able to provide genetic information to patients and families which ultimately leads to better management of SCD. Imparting SCD-appropriate counseling modules and management guidelines to the existing health workers should be the first step in developing SCD screening and care model, especially for indigenous populations (Fig. 1).

Challenges among indigenous/tribal communities

Although substantial literature on biomedical and clinical aspects of SCD exists, limited data is available on the design and implementation of SCD control programs including GC in Indian tribal communities (Raman et al. 2021). There are many challenges faced by counselors while working with tribal communities, and a few of them are discussed here.

Education

As per the Census 2011, the literacy rate of Indian tribes has considerably improved from only 8.54% in 1961 to 63.1%

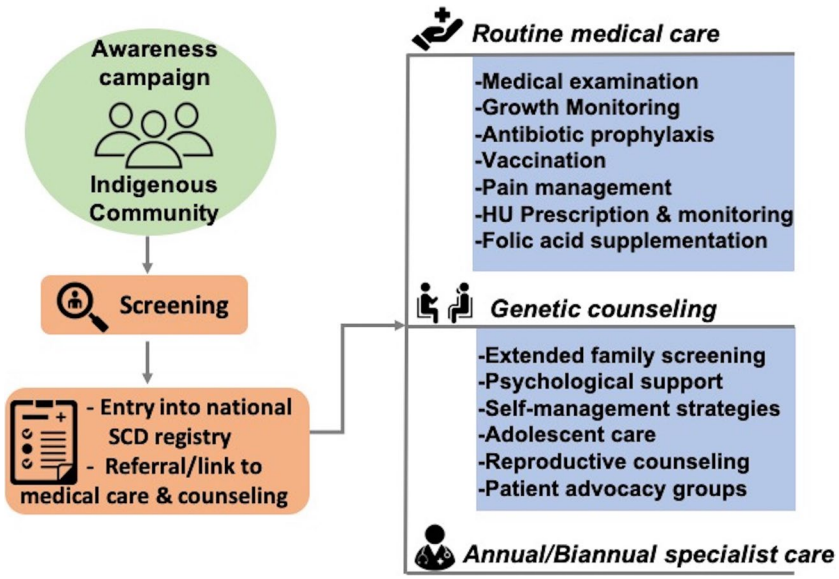

Fig. 1 The healthcare model for SCD screening, care, and genetic counseling. Awareness campaigns and SCD-related education should precede the routine screening of indigenous communities with their proper consent. The results of the screening shall be entered in "National SCD Registry" followed by referrals and linking of patients with SCD to routine medical care, genetic counselors, and annual/biannual specialist care

in 2011. However, female literacy of tribes is still at 54.4% compared to male literacy of 71.7%. Several tribal children also drop out of school: 55% at the primary level and a whopping 71% at the secondary level (Upmanyu 2016). As education does not yield any immediate economic return, the tribal parents prefer to engage their children in remunerative employment which supplements the family income (Hasnain 2004). Education and health awareness are clearly the transforming agents toward development and health equities, especially in marginalized tribal communities (Bagavandas 2021). Not only the clear understanding of genetic basis, preventative measures, reproductive options, and lifelong healthcare management of SCD, but also GC perception and acceptance could be enhanced with education.

Income

Most of the Indian tribal communities are spread across the country and reside in the forest and hilly regions of the country. They majorly depend on forests for food, fodder, firewood, and water. The data from the Socio Economic Caste Census (SECC) 2011 shows that 86.53% of the Scheduled Tribe (ST) households earn monthly a paltry sum of less than 5000 INR (equivalent to 61 USD). ST communities are still lagging behind in terms of their socioeconomic factors as compared to other backward classes (OBC) and upper castes in India (Desai and Dubey 2011). The socioeconomic status of a family significantly determines its response to a health crisis. The tribal people often fail to meet their basic needs due to irregularities in their income, let alone obtain healthcare (Yadav and Sahoo 2019). Furthermore, lack of information, literacy, etc. makes them forego available treatment options and seek counseling for genetic diseases.

Accessibility

The fourth National Family Health Survey of India (2015–2016) revealed that only 73% of tribal women received antenatal care against 86% of uncategorized groups. Similarly, only 55% of tribal children were fully immunized. This low access to healthcare services among tribal communities can be attributed to several factors such as poor access to transportation and health facilities, financial constraints, lack of awareness, and cultural factors. Genetic counselors practice in a variety of settings including government and private hospitals. Still, tribals staying in tough geographical terrain often have very limited or no access to genetic counselors as most of the public health centers (PHCs) in India do not employ one. It is mainly through a referral system or NGO help, SCD tribal patients get to consult a genetic counselor on a regular basis. Certification and regularization of genetic counselors by the Indian Council of Medical Research (ICMR) or other recognized governmental bodies will significantly create the necessary labor force and help to close the demand gap.

Social stigma

Patients with SCD or sickle cell carriers are sometimes given informational cards, which provide awareness about the disease and may be useful for premarital counseling (National Sickle Cell Anaemia Elimination Mission 2023; Shrikhande et al. 2014). However, from the Indian marriage perspective, the chief concern that emerged with this method is diminished marriage prospects for girls having a "carrier" gene due to the associated discrimination and stigmatization with SCD. Thus, any formal way of patient identification should be applied with caution as it could lead to unwanted

stigmatization. Carrier screening coupled with reproductive counseling is a feasible solution to avoid such stigmatization and ostracization. During pregnancy, pediatricians and obstetricians should suggest prenatal screening and, if found positive, refer to the genetic counselor. GC enable couples to form informed reproductive choices which could lead to a reduced SCD prevalence and stigmatization (Cowan 2009; Schmidtke and Cornel 2020).

Different levels of SCD counseling

Premarital

SCD is inherited in an autosomal recessive manner, which implies that carriers have a 25% chance of passing it on to their offspring. Prospective risk detection through carrier screening across a wide range of populations is an effective way to reduce SCD birth prevalence (Modell 2020). Premarital counseling by trained healthcare personnel or counselors should be made available to all carriers and families at risk to assist them in making informed reproductive choices. In Saudi Arabia and Middle Eastern regions, premarital genetic counseling and screening markedly decrease the prevalence of SCD and b-thalassemia by reducing the number of at-risk marriages by more than the 5-fold between 2004 and 2009 (Memish and Saeedi 2011). Creating awareness about genetic tests and screening at schools and educational institutions is found to be equally effective in SCD control programs (Moronkola and Fadairo 2006; Zlotogora 2009). Premarital screening is more successful than random screening in India for family members of patients with SCD (Tamhankar et al. 2009). Premarital GC, however, has low acceptance among Indian tribes mainly because of the deeply ingrained practice of endogamous marriage (Mohanty and Das 2011). A combination of hospital-based and community outreach approaches with the support of community leaders should be utilized to reach out to the affected population.

Prenatal

It is now possible to identify SCD genetic mutation in fetuses prenatally by using efficient and safe prenatal diagnostic procedures. Counselors should guide pregnant couples carrying the SCD mutation to make informed reproductive choices. In the UK adoption of the SCD and thalassemia program in the NHS Plan (2000), timely screening for both SCD and thalassemia is offered to all women in the prenatal period before the 12th week of gestation. Adopting the same model, screening should be made mandatory in all prenatal centers in high-risk areas in India (Colah et al. 2014; Thakur and Singh 2019). Based on our GC experience, both prenatal screening and counseling have a higher acceptance rate in India; hence, we recommend following this method whenever possible. However, critical issues associated with prenatal screening such as the exorbitant cost of testing and the lack of facilities, experts, and government support need to be duly addressed (Bain 2009).

Postnatal

Children with SCD may exhibit clinical manifestations in their first year of life (Brousse et al. 2014). The most common signs and symptoms are the severe form of pain normally referred to as “crisis” (Adewoyin 2015). Without effective health care, over 95% of children born with the disease die before the age of 5 years (Dennis-Antwi et al. 2008). In a study performed in tribal areas of India, around 20% of children with SCD died by 2 years of age and 30% of children died before they reached adulthood (Saxena et al. 2017). Though these children affected with SCD may be asymptomatic at birth, early diagnosis is the best measure to save children from life-threatening infections due to the functional asplenia present in these children. Counselors shall assist to diagnose early by facilitating appropriate screening tests. They should also inform parents about the benefits of prescribing hydroxyurea early in life and the judicious use of blood transfusions. Hydroxyurea benefits children by reducing painful crises, organ damage, and transfusion requirement and improving overall survival. As the majority of SCD cases are detected postnatally in India, an effective GC intervention could lead to better health-related outcomes through systematic planning and lifelong management of SCD (our unpublished results from an intervention study between 2019 and 2022, National Task Force SCD ICMR project, India).

Adults

Over the years, the early high mortality associated with SCD patients has been considerably reduced by routine health monitoring and parental counseling (Quinn et al. 2010). However, for a better quality of life, adult patients still need to be educated about the disease and its long-term symptoms (Cordovil 2018). Adolescent issues such as marriage and reproductive decision-making must be included in the individualized GC. Sickle cell carriers usually have milder or no symptoms, but some may need intervention for fever, pain, etc. and should be informed thus by genetic counselors. Because the SCD condition is persistent, patients incur high healthcare bills, which adds to social and emotional issues in addition to physical pain. Engaging young adults through multiple visits, emphatic responses, and communicating in the local language with a psychotherapeutic approach can help alleviate some of the issues faced by patients with SCD (Austin et al. 2014).

The benefits and drawbacks of counseling at various levels are described in Table 1.

Toolkit for successful GC among the tribal population

There have been several state-wide SCD screening programs in India but without a provision of counseling facilities. Lack of counseling can lead to unnecessary anxiety, misconceptions, and an impaired self-image. Some of the effective strategies for SCD counseling are described below.

One-to-one counseling

One-to-one counseling or individual counseling is a process through which patients work one-on-one with a trained genetic counselor in a safe, caring, and confidential environment. Often, the two form an alliance, relationship, or bond that enables trust and better outcomes, especially in tribal communities. Social stigmatization and ostracization can make SCD-affected patients wary of genetic counseling. Many of these concerns can be readily resolved in individual counseling where confidentiality and privacy can be guaranteed. Besides, longer commitment to complex issues such as clinical follow-up can also be easily obtained. Furthermore, assessment of an individual's health beliefs that influences SCD testing and counseling may be beneficial in designing approaches to individual counseling (Gustafson et al. 2007). In general, one-to-one counseling is more effective in a tribal setting where trust-building is paramount for the success of GC.

Group counseling

This form of counseling involves four to ten participants with one or two group therapists. Most groups meet regularly (weekly or monthly) for 1 to 2 h. During that time, the members of the group discuss the issues that are concerning them and offer each other support and feedback. A similar objective can also be achieved by establishing patient support groups (PSGs). A person or afflicted patient known to the community is usually recruited to conduct group counseling sessions or run PSGs. Not only it provides much-needed experiential advice, but it also helps in the further management of disease such as tertiary treatment and financial help (Nietert et al. 2002). However, it could also become a double-edged sword if the counselor or therapist belonging to the local community fails to safeguard the confidentiality of the patient, hence inadvertently exposing patients to community stigmatization or ostracization.

Psychological counseling

Beyond diagnosis, the SCD patient often requires counseling to deal with the new normal and make necessary life adjustments. It is essential to develop counseling strategies with the support of clinical psychologists to cope with the disease, health maintenance, psychological adjustment, and acceptance of extended family testing. Appropriate psychological intervention profoundly alters sets of beliefs, cultural perspectives, and health-seeking behavior of patients with SCD (Ilesanmi 2010). Through effective communication, a counselor should correct misconceptions and assist the counselee to express and manage his desires, expectations, concerns, anxieties, and any potential threats to self-image (Headings and Fielding 1975).

Table 1 Characteristics of different levels of genetic counseling (GC)

Levels of GC	Benefits	Drawbacks
Premarital	- Ideal for low-resource setting - Preventive and easily implemented in communities that practice free-choice marriages - Mandatory counseling helps during the eradication stage	- Need a basic understanding or awareness about SCD - Culturally not acceptable in many tribal communities that practice endogamy - Can create stigma/ society ostracization
Antenatal	- More acceptable in communities that practice arranged marriages - Early intervention leads to a better outcome (informed reproductive choice)	- High cost - Need trained professionals in fetal testing - Emotional trauma for the mother and requires expert counselors if the pregnancy needs to be terminated
Postnatal	- Early diagnosis and counseling lead to better disease management	- Requires considerable resources to run comprehensive government programs for screening, counseling, and management
Adult	- Provide counseling to patients missed at earlier levels - Provision to assess mental health and provide care to adult patients - Reproductive counseling can prevent future affected pregnancies	- Sometimes difficult to consul adult patients with fixed conceptions - Non-conformity with counselor due to stigma or prior commitments - Less acceptance of GC - Avoidable disabilities would already be established and hence poorer outcome

Auxiliary requirements

The first pillar of SCD care is to manage the important functional aspects of the disease, with the goal to lead near-healthy life. The second is to manage the cognitive, social, and neuropsychiatric aspects of the disease. To ensure the successful and sustained implementation of GC in indigenous communities, the following auxiliary factors must be considered.

Local socio-cultural values and behavior

In India, 635 biological tribes and sub-tribes are identified based on their specific biological constitution, anthropogenic characterization, distinct cultural and linguistic patterns, and confined geographical localization (Census 2011). Many of these tribes are rooted deeply in cultural and religious practices, which are unique to each tribe (Mohanty and Das 2011). Therefore, being aware of socio-cultural practices along with religious beliefs can help a counselor initiate and hold genetic discussions unalienated from their cultural context. It is also advisable to become familiar with the local terminology for SCD and its specifics. For example, sickle cell disease (SCD) is called by its local name *Kuḍagōlu kaṇa rōga* in tribal areas of South Karnataka. Using local names for SCD and symptoms immediately creates an atmosphere of familiarity and improves GC efficacy.

In Mediterranean countries such as Cyprus, religious bodies have played very significant roles in enforcing mandatory premarital testing and giving appropriate counseling to at-risk individuals with very remarkable results (Cowan 2009). Similarly, in India, by enlisting tribal elders and religious organizations, counselors can ensure better reception of their advice. Recognizing the underlying causes of GC rejection and acknowledging differences among tribal subgroups demand culturally competent counselors (Dheensa et al. 2016). Genetic counseling in tribals unlike the general population is a difficult task due to lower literacy and poor socioeconomic factors. However, with sustained efforts and close interaction in the local language with tribals, certain misbeliefs can be removed gradually considering their socio-cultural background.

Community awareness

In addition to individual counseling, counselors should provide community outreach through education and training to keep communities more informed, particularly about genetic diseases prevalent locally. Counselors often possess the necessary expertise that can help communities develop awareness programs. These programs would help communities in recognizing the symptoms and getting tested for genetic diseases prevalent within these communities. In addition, counselors should reach out to local legislators, tribal leaders, school teachers, and grassroots health workers such as ASHA and Anganwadi workers to work within their communities to develop such awareness programs. Counselors can also harness the support of traditional healers and local medical or healthcare providers at primary health centers (PHCs) to create more awareness about SCD.

Tribal trained counselors

There is a dearth of genetic counselors in India and it is impossible to recruit them for every tribal district. Based on the prevalence rate and anticipated annual pregnancies, therefore, each state may designate or recognize a certified genetic counselor. Every district shall select a competent educated individual from their own community who can be trained, supervised, and advised by the state genetic counselor in accordance with national GC guidelines (Kumar et al. 2020). Together, they would address the scarcity and bridge the gap between the beneficiaries and the health infrastructure.

Future perspectives

Globally, indigenous people have limited access to culturally appropriate care due to linguistic, geographic, and financial factors, which delays diagnosis and severely limits their access to improved medical care including genetic counseling (D'Angelo et al. 2020). With a tribal population of nearly 8.6%, cooperation between the public and commercial sectors is key to the success of SCD control initiatives in India (Fig. 2). We have outlined a few areas where such collaboration can take place.

Establishing GC centers

Preventive measures for SCD are far easier as they can be implemented with a tighter budget (Gupta 2009). The treatment for SCD such as gene therapy is quite expensive and out of reach for most of the Indian population. Both prenatal and premarital counseling for SCD has been successfully initiated in several areas across India. However, sustaining GC success in SCD prevention requires the setting up of GC permanent centers, particularly in the tribal areas of India (Mohanty and Das 2011). State governments should develop a comprehensive plan to set up at least two to three GC centers for disseminating awareness and education about genetic diseases. Furthermore, the guideline that counseling is not the sole responsibility of counselors at the local level must also be observed. Local healthcare workers must be actively

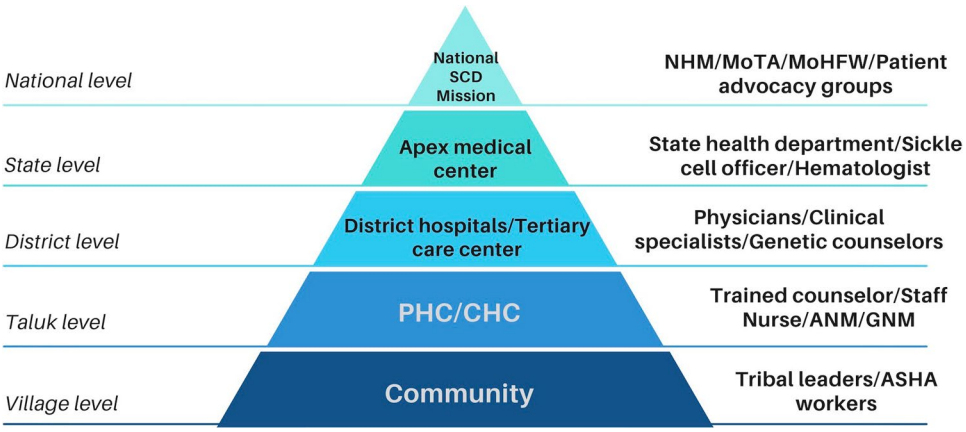

Fig. 2 Multi-level care model for SCD among Indian tribals. National SCD Mission is designed and guided by policies framed by National Health Mission (NHM), Ministry of Tribal Affairs (MoTA), and Ministry of Health and Family Welfare (MoHFW). Primary health center (PHC); community health center (CHC); auxiliary nurse and midwife (ANM); general nurse and midwife (GNM); Accredited Social Health Activist (ASHA)

involved in the counseling procedure, guided by certified genetic counselors. If a permanent GC center is not feasible, a public-private collaboration should be employed to identify existing health centers as counseling service providers to avoid any unnecessary delays. For the successful implementation of GC, duties should be clearly defined for each stakeholder.

Initiating a referral system

In India, under national programs, some of the local health centers already conduct genetic screenings for hemoglobinopathies such as SCD and thalassemia. However, such programs offer no referrals for follow-up and SCD management. Partnering with genetic counselors will allow state and district centers to provide their patients with referrals for easy access to GC. In our experience, a referral system proved to be a time-tested and cost-effective way to provide easy access to GC in remote tribal places.

Repurposing health wellness centers as genetic counseling centers

In 2018, the Indian government established 150,000 Health and Wellness Centers (HWCs) by transforming existing sub-centers and PHCs across India. Since then, these centers brought primary healthcare closer to the homes of people by providing both maternal and child health services and management of non-communicable diseases, including free essential drugs and diagnostic services (Lahariya 2020). The fundamental idea is to enable people and local communities to build high-quality health infrastructure. In the same milieu, Auxiliary Nursing Midwifery (ANM) and General Nursing Midwifery (GNM) can be trained by certified genetic counselors to provide quality GC at HWCs. This will not only bring credibility to counseling but would also ensure 24/7 access to people belonging to remote areas.

Conclusions

Counseling, community education, and awareness play a very important role in the successful implementation of SCD prevention programs. To ensure better acceptability, counseling and screening programs should be conducted in sync with cultural, social, and religious beliefs. GC can greatly reduce the SCD burden by seeking timely help and establishing lifelong treatment and care for patients with SCD. Importantly, counselors serve as a link between the patients and the healthcare system to ensure the effective management of SCD. Establishing genetic counseling services for the tribal communities at the premarital and prenatal levels would greatly help tribal populations at risk. Taking lessons from developed countries, India needs to recognize, accept, and integrate GC into the routine clinical management of SCD. Sustained implementation of GC coupled with the establishment of permanent centers is required to reduce the overall SCD burden in India. Clearly, SCD has social and health implications for the patients, so the treatment and counseling of the person with SCD needs an approach focused on the prevention of these complications in an individualized way.

Acknowledgements We acknowledge the support and cooperation of our patients who have taught us many valuable lessons mentioned in this manuscript. We also acknowledge financial support from the CTRITH research grant funded by DBT/Wellcome Trust India Alliance.

Author contribution P.A. made substantial contributions to the conception and wrote the first draft of the manuscript. D.B. revised it critically for important intellectual content. Both P.A. and D.B. read and approved the final manuscript to be published.

Funding P.A. is supported by DBT/Wellcome trust India Alliance grant no. IA/CRC/20/1/600007. D.B. is permanent faculty and supported by JSS Medical College.

Declarations

Ethical approval This is an observational study. The JSS medical college research ethics committee has confirmed that no ethical approval is required.

Conflict of interest The authors declare no competing interests.

References

Adewoyin AS (2015) Management of sickle cell disease: a review for physician education in Nigeria (sub-Saharan Africa). Anemia 2015:791498. https://doi.org/10.1155/2015/791498

Austin J, Semaka A, Hadjipavlou G (2014) Conceptualizing genetic counseling as psychotherapy in the era of genomic medicine. J Genet Couns 23(6):903–909. https://doi.org/10.1007/s10897-014-9728-1

Bagavandas M (2021) Development of multifactor index for assessing quality of life of a tribal population of India: multilevel analysis approach. BMC Public Health 21:383. https://doi.org/10.1186/s12889-021-10338-2

Bain BJ (2009) Neonatal/newborn haemoglobinopathy screening in Europe and Africa. J Clin Pathol 62(1):53–56. https://doi.org/10.1136/jcp.2008.060624

Brousse V, Makani J, Rees DC (2014) Management of sickle cell disease in the community. BMJ 10(348):g1765. https://doi.org/10.1136/bmj.g1765

Bunn HF (1997) Pathogenesis and treatment of sickle cell disease. N Engl J Med 337(11):762–769. https://doi.org/10.1056/NEJM199709113371107

Choi J, Kim H (2014) Effectiveness of the interventions utilized in genetic counselling. Adv Nurs 2014:1–9. https://doi.org/10.1155/2014/725968

Colah R, Mukherjee M, Ghosh K (2014) Sickle cell disease in India. Curr Opin Hematol 21(3):215–223. https://doi.org/10.1097/MOH.0000000000000029

Cordovil K (2018) Sickle cell disease: a genetic disorder of beta-globin. In: Thalassemia and Other Hemolytic Anemias. InTech. https://doi.org/10.5772/intechopen.74778

Cowan RS (2009) Moving up the slippery slope: mandated genetic screening on Cyprus. Am J Med Genet Part C Semin Med Genet 151C:95–103. https://doi.org/10.1002/ajmg.c.30202

D'Angelo CS, Hermes A, McMaster CR et al (2020) Barriers and considerations for diagnosing rare diseases in indigenous populations. Front Pediatr 8:579924. https://doi.org/10.3389/fped.2020.579924

Dennis-Antwi JA, Dyson SM, Ohene-Frempong K (2008) Healthcare provision for sickle cell disease in Ghana: challenges for the African context. Divers Health Soc Care 5(4):241–254

Desai S, Dubey A (2011) Caste in 21st century India: competing narratives. Econ Polit Wkly JSTOR 46(11):40–49. http://www.jstor.org/stable/41151970. Accessed 6 March 2023

Dheensa S, Fenwick A, Shkedi-Rafid S et al (2016) Health-care professionals' responsibility to patients' relatives in genetic medicine: a systematic review and synthesis of empirical research. Genet Med 18:290–301. https://doi.org/10.1038/gim.2015.72

Ellington L, Kelly K, Reblin M (2011) Communication in genetic counseling: cognitive and emotional processing. Health Commun 26(7):667–675. https://doi.org/10.1080/10410236.2011.561921

Government of India: State Primary Census Abstract for Individual Scheduled Caste and State Primary Census Abstract for Individual Scheduled Tribe: Census of India. In: Ministry of Home Affairs, Office of the Registrar General and Census Commissioner. (2011). https://censusindia.gov.in/census.website/data/census-tables. Accessed 28 July 2023

Gupta RB (2009) Sickle cell disease in Central India-need for micro level planning. In: Tribal health: proceedings on national symposium. RMRC (ICMR), Jabalpur, pp 109–115

Gustafson SL, Gettig EA, Watt-Morse M, Krishnamurti L (2007) Health beliefs among African American women regarding genetic testing and counseling for sickle cell disease. Genet Med 9(5):303–310. https://doi.org/10.1097/gim.0b013e3180534282

Hasnain (2004) Indian society and culture: continuity and change. Jawahar Publishers & Distributors, New Delhi

Headings V, Fielding J (1975) Guidelines for counseling young adults with sickle cell trait. Am J Public Health 65(8):819–827. https://doi.org/10.2105/ajph.65.8.819

Ilesanmi OO (2010) Pathological basis of symptoms and crises in sickle cell disorder: implications for counseling and psychotherapy. Hematol Rep 2(1):e2. https://doi.org/10.4081/hr.2010.e2

Kulozik AE, Wainscoat JS, Serjeant GR, Kar BC, Al-Awamy B, Esan GJF et al (1986) Geographical survey of βs-globin gene haplotype: evidence for an independent Asian origin of the sickle cell mutation. Am J Hum Genet 39:239–244

Kumar MM, Pathak VK, Ruikar M (2020) Tribal population in India: a public health challenge and road to future. J Family Med Prim Care 9(2):508–512. https://doi.org/10.4103/jfmpc.jfmpc_992_19

Lahariya C (2020) Health & wellness centers to strengthen primary health care in India: concept, progress and ways forward. Indian J Pediatr 87(11):916–929. https://doi.org/10.1007/s12098-020-03359-z

Memish ZA, Saeedi MY (2011) Six-year outcome of the national premarital screening and genetic counseling program for sickle cell disease and β-thalassemia in Saudi Arabia. Ann Saudi Med 31(3):229–235. https://doi.org/10.4103/0256-4947.81527

Modell B (2020) Haemoglobin disorders-a point of entry for community genetics services in India? J Community Genet 11:7–9. https://doi.org/10.1007/s12687-019-00441-8

Mohanty D, Das K (2011) Genetic counseling in tribals in India. Indian J Med Res 134(4):561–571

Moronkola OA, Fadairo RA (2006) University students in Nigeria: knowledge, attitude toward sickle cell disease, and genetic counseling before marriage. Int Q Community Health Educ 26(1):85–93. https://doi.org/10.2190/jn25-4353-75pk-3733

National Health Mission Ministry of Health & Family Welfare, Government of India (2023) Prevention and control of hemoglobinopathies in India-thalassemias, sickle cell disease and other variant hemoglobins. https://pib.gov.in/PressReleasePage.aspx?PRID=1896039. Accessed 19 July 2023

National Sickle Cell Anaemia Elimination Mission (2023) https://sickle.nhm.gov.in/. Accessed 18 July 2023

Nietert PJ, Silverstein MD, Abboud MR (2002) Sickle cell anaemia: epidemiology and cost of illness. Pharmacoeconomics 20(6):357–366. https://doi.org/10.2165/00019053-200220060-00001

Pauling L, Itano HA, Singer SJ, Wells IC (1949) Sickle cell anemia, a molecular disease. Science 110:543–548. https://doi.org/10.1126/science.110.2865.543

Porter J, Garbowski M (2013) Consequences and management of iron overload in sickle cell disease. Hematology Am Soc Hematol Educ Program 1:447–456. https://doi.org/10.1182/asheducation-2013.1.447

Quinn CT, Rogers ZR, McCavit TL, Buchanan GR (2010) Improved survival of children and adolescents with sickle cell disease. Blood 115:3447–3452. https://doi.org/10.1182/blood-2009-07-233700

Raman V, Seshadri T, Joice SV, Srinivas N (2021) Sickle cell disease in India: a scoping review from a health systems perspective to identify an agenda for research and action. BMJ Glob Health 6(2):e004322. https://doi.org/10.1136/bmjgh-2020-004322

Rees DC, Brousse VA (2016) Sickle cell disease: status with particular reference to India. Indian J Med Res 143(6):675–677. https://doi.org/10.4103/0971-5916.191916

Regional Committee for Africa, 60 (2011) Sickle-Cell Disease: a strategy for the WHO African Region. https://apps.who.int/iris/handle/10665/1682. Accessed 28 July 2023

Saxena D, Yasobant S, Golechha M (2017) Situational analysis of sickle cell disease in Gujarat, India. Indian J Community Med 42:218–221

Schmidtke J, Cornel MC (2020) Contentious ethical issues in community genetics: let's talk about them. J Community Genet 11:5–6. https://doi.org/10.1007/s12687-019-00444-5

Serjeant GR (2016) Evolving locally appropriate models of care for Indian sickle cell disease. Indian J Med Res 143(4):405–413. https://doi.org/10.4103/0971-5916.184282

Shrikhande AV, Arjunan A, Agarwal A, Dani A, Tijare J, Gettig E, Krishnamurti L (2014) Prevalence of the βS gene among scheduled castes, scheduled tribes and other backward class groups in Central India. Hemoglobin 38(4):230–235. https://doi.org/10.3109/03630269.2014.931287

Sinha S, Seth T, Colah RB et al (2020) Haemoglobinopathies in India: estimates of blood requirements and treatment costs for the decade 2017–2026. J Community Genet 11:39–45. https://doi.org/10.1007/s12687-019-00410-1

Tamhankar PM, Agarwal S, Arya V, Kumar R, Gupta UR, Agarwal SS (2009) Prevention of homozygous beta thalassemia by premarital screening and prenatal diagnosis in India. Prenat Diagn 29:83–88. https://doi.org/10.1002/pd.2176

Thakur S, Singh S (2019) Genetic counseling to prevent thalassemia and hemoglobinopathy in the Indian population. Perinatology 20(3):61–65

Upmanyu MC (2016) The tribal education in India, status, challenges, and issues. Int J Novel Res Education Learning 3(6):96–102

Yadav M, Sahoo AK (2019) An empirical study of determining factors of tribal household income: a case study of Tharu tribe in India. Humanit Soc Sci Stud 8:102–112

Zlotogora J (2009) Population programs for the detection of couples at risk for severe monogenic genetic diseases. Hum Genet 126(2):247–253. https://doi.org/10.1007/s00439-009-0669-y