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Editorial

Hemoglobinopathies in India: Current Landscape and Future Directions in Screening, Genomics, and Curative Therapies

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India lies within the global thalassaemia belt, with a large tribal population (8.6%) [1] and beta thalassaemia frequencies ranging from 3% to 17% [2,3]. The prevalence of sickle cell disease also ranges from 1% to 44% [4]. Approximately ten thousand children are born with beta thalassaemia major every year in India [5]. HbS is the most common, with a frequency of 4.3% [6], followed by HbE, which has a carrier frequency of 50-60% [7]. The genetic diversity of Indian haemoglobinopathies is mainly due to endogamy, consanguinity, and the large tribal population [8,9], as shown in regional and caste-specific analyses of haemoglobinopathy mutation data [10-12].

Screening and diagnostic strategies for hemoglobinopathies in India have evolved over time to address this disease burden. The diagnostic approaches have transitioned from manual, time-consuming methods to automated, high-resolution techniques such as HPLC for preliminary screening and PCR for confirming diagnosis and prenatal diagnosis [13,14]. Screening programmes are tailored to regional variants: HbS is common in the tribal populations of Central, Western, and Southern India; HbE is prevalent in North-Eastern states; and HbD is frequently found in Northern states, particularly Punjab [15,16].

Children with haemoglobinopathies are particularly vulnerable to severe anaemia, which significantly increases the risk of morbidity and mortality. Key health impacts and complications include reduced oxygen supply, developmental delay, impaired cognitive development, organ damage and life-threatening crises [17]. WHO indicates that without early diagnosis and proper management, many children with these conditions die within their first few years of life. India has the world's largest burden of transfusion-dependent β-thalassaemia in children (1-1.5 lakh, with 10000 new cases annually). The Sankalp programme demonstrated that targeted antenatal screening is a highly feasible, effective and scalable strategy for preventing haemoglobinopathies in India [18]. In paediatric sickle cell disease, newborn screening, prophylactic antibiotics, vaccination and disease-modifying therapy (hydroxyurea) reduce vaso-occlusive crises and improve survival [19].

Therapeutic advances in the management of hemoglobinopathies in India have significantly improved patient survival, though equitable access to these treatments remains a challenge. Advancements include optimised transfusion protocols [20], advanced iron chelation, hydroxyurea therapy and expanding curative approaches, including hematopoietic stem cell transplantation [21]. The Indian government is addressing this through the National Health Mission guidelines on hemoglobinopathies, focusing on nationwide standards, neonatal screening and improved day care facilities for transfusion. Novel therapies and gene-based interventions for hemoglobinopathies have emerged as curative options in India, but their high cost has made them inaccessible [22]. The National Sickle Cell Anaemia Elimination Mission, launched in 2023, aims to eliminate the genetic transmission of sickle cell disease by 2047, with a focus on universal screening. Key strategies include community awareness, prenatal diagnosis, counselling for carriers and comprehensive care for affected individuals.

In conclusion, hemoglobinopathies in India exhibit complex epidemiological and molecular landscapes driven by population genetics and socio-cultural practices. Strengthening integrated screening programs, expanding molecular diagnostics, improving pediatric care, and ensuring equitable access to advanced therapies are critical for reducing disease burden, particularly among high-risk tribal populations.

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