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EDITED AND REVIEWED BY
Anna Rita Migliaccio,
Campus Bio-Medico University, Italy

*CORRESPONDENCE
Foued Maaoui
maaoui_foued@hotmail.com

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Editorial: Investigations and management of hereditary red blood cells diseases

Imen Moumni and Foued Maaoui*

Laboratory of Molecular and Cellular Hematology, Pasteur Institute of Tunis, Tunis, Tunisia

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Editorial on the Research Topic
Investigations and management of hereditary red blood cells diseases

Hereditary red blood cell disorders, such as sickle cell disease (SCD) and thalassemia, are major global health challenges, affecting millions, particularly in regions with a historical prevalence of malaria. These conditions, driven by hemoglobin defects or red cell structural abnormalities, cause chronic anemia, painful crises, and reduced life expectancy.

The Research Topic "*Investigations and management of hereditary red blood cells diseases*" seeks to advance diagnostic, epidemiological, and therapeutic approaches to better understand and manage these disorders. Four recent articles in *Frontiers in Hematology and Frontiers in Medicine*, published on August 4, 2023, November 8, 2023, March 17, 2025, and September 20, 2024, offer critical insights into pain management, population surveillance, screening innovations, and diagnostic imaging for liver iron overload.

This editorial contextualizes their contributions, highlighting their clinical significance and implications for global healthcare systems.

A key focus is clinical management, particularly pain control in SCD. The study by Ismail et al. published on August 4, 2023, investigates physicians' knowledge, attitudes, and practices in managing vaso-occlusive crises in Qatar. While 91% of clinicians prescribe opioids like morphine, aligning with NHLBI and ASH guidelines, only 21% reassess pain promptly, and 20% use patient-controlled analgesia. These gaps, partly driven by negative attitudes toward SCD patients, reveal a disconnect between guidelines and practice. This work underscores the need for targeted education and destigmatization to improve care delivery and patient quality of life.

Epidemiological surveillance is another cornerstone, informing health policy. The study by Plaxco et al. published on November 8, 2023, leverages newborn screening and Medicaid data to map SCD in Tennessee from 2008 to 2019. Identifying 3,439 cases, it reveals disparities: most patients reside in urban areas, but rural individuals, though a minority, face limited specialist access, with 69.2% lacking local hematologists.

High acute care utilization, especially among young adults, signals unmet needs. This population-level investigation demonstrates how data can pinpoint inequities and guide resource allocation, such as incentives to attract specialists to underserved regions.

Screening, a vital preventive tool, benefits from innovation. The study by Periyavan et al. published on March 17, 2025, evaluates high-performance liquid chromatography (HPLC) as a primary screening tool compared to the traditional complete blood count (CBC) method in India. Testing 6,549 pregnant women, HPLC detects more hemoglobinopathy carriers, missing 14.1% fewer cases than CBC, with minimal cost increases.

This rapid, effective approach suits high-prevalence settings, identifying diverse variants overlooked by red cell indices. It marks a significant advance in diagnostic investigation, strengthening prenatal prevention.

Diagnostic imaging for thalassemia management is also critical, particularly for assessing liver iron overload (LIO). The study by Luo et al. published on September 20, 2024, evaluates the accuracy of CMRtools software for measuring liver iron concentration (LIC) in 108 thalassemia patients. Comparing T2*-based LIC measurements to the FDA-approved MRI-R2/FerriScan, it finds that higher R^2 values and more echo time (TE) images improve diagnostic accuracy, though LIC severity does not significantly affect results.

This work highlights the potential of T2* relaxometry for guiding chelation therapy, despite challenges with severe LIO. Together, these studies, Ismail et al., Plaxco et al., Periyavan et al. and Luo et al. form a cohesive narrative: effective management of hereditary red blood cell diseases requires integrated clinical, epidemiological, diagnostic, and imaging efforts. Pain control falters without consistent follow-up, surveillance exposes access gaps, HPLC screening enhances early detection, and advanced imaging refines treatment monitoring. These findings emphasize that addressing these disorders demands collaboration across research, clinical practice, and policy.

Their impact extends globally. The stigma noted by Ismail et al. in Qatar reflects challenges in chronic disease management worldwide. The rural-urban divide highlighted by Plaxco et al. in Tennessee parallels access barriers in low-resource settings.

The screening innovation by Periyavan et al. in India offers a model for regions upgrading diagnostic capabilities. The imaging advancements by Luo et al. address a critical need in thalassemia care globally. Aligned with WHO objectives to reduce hemoglobinopathy burdens, these studies call for tailored strategies across diverse socioeconomic contexts.

Challenges remain. How can clinician training eliminate bias? What incentives best address specialist shortages? Can HPLC and advanced imaging become universally accessible? Future research could explore genomic sequencing, artificial intelligence, or ultrashort echo sequences to refine diagnostics and predict complications. Global cooperation is vital to standardize protocols and promote equitable care.

By addressing pain, surveillance, screening, and imaging, these articles pave the way for more equitable, effective care, offering hope to affected communities worldwide.

In conclusion, the four studies featured in this Research Topic collectively demonstrate that progress in managing hereditary red blood cell disorders hinges on a multi-dimensional approach, one that bridges clinical care, epidemiological insight, diagnostic innovation, and imaging precision. From optimizing pain management and addressing healthcare disparities to advancing screening and monitoring techniques, these contributions reflect the evolving landscape of care for sickle cell disease and thalassemia.

As health systems strive to meet WHO goals and reduce the global burden of hemoglobinopathies, sustained investment in research, training, and international collaboration is essential. Only by aligning clinical practices with scientific advances and social realities can we ensure that all patients, regardless of geography or socioeconomic status, receive timely, effective, and dignified care.

Author contributions

IM: Writing – review & editing. FM: Writing – original draft, Writing – review & editing.

Conflict of interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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The author(s) declare that no Generative AI was used in the creation of this manuscript.

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