# Bulletin recherche

## Filière MCGRE

**N° 23 - Février 2025**

MCGRE

FILIÈRE SANTÉ MALADIES RARES

MCGRE
FILIÈRE SANTÉ MALADIES RARES

# Table des matières

# Arythmie atriale chez les patients drépanocytaires adultes : un lien manquant pour comprendre et éviter les accidents vasculaires cérébraux

## Contexte et objectif

La fonction circulatoire du cœur est un aspect majeur de la compensation des anémies. Le sang passe successivement de la cavité cardiaque atriale (ou oreillette) droite au ventricule droit, se fait oxygéner en passant par les poumons puis circule de la cavité atriale gauche vers le ventricule gauche avant d'être envoyé par l'aorte vers l'ensemble des organes. La contraction anarchique des oreillettes (fibrillation) perturbe la séquence de remplissage des ventricules ce qui peut provoquer une insuffisance cardiaque et exposer à un risque de formation de caillot, et donc d'accident vasculaire.

En réponse à l'anémie hémolytique chronique, les patients drépanocytaires présentent une dilatation des oreillettes dès le plus jeune âge, les exposants précocement à un risque d'arythmie atriale et la survenue de complications thromboemboliques. Les accidents vasculaires cérébraux (AVC) sont l'une des principales complications aiguës de la drépanocytose et sont fréquemment associés à la présence d'une vasculopathie cérébrale. Néanmoins, une part non négligeable des AVC pourrait être en lien avec un mécanisme cardio-embolique, résultant possiblement d'arythmies atriales non diagnostiquées. Les auteurs de cette étude ont émis l'hypothèse que ce trouble du rythme cardiaque contribuerait à la survenue de complications emboliques, d'hypertension pulmonaire post-capillaire et à des décompensations cardiaques aiguës, influençant la qualité de vie et le pronostic des patients drépanocytaires. L'objectif de cette étude était d'évaluer de manière prospective la prévalence des arythmies atriales, leurs causes et leurs liens avec la survenue d'AVC chez les patients drépanocytaires adultes.

## Méthode

Cette étude a été réalisée chez 130 patients drépanocytaires adultes (SS ou Sβ0) suivis au centre de référence adulte de Créteil du GHU Henri Mondor inclus dans le registre français prospectif multicentrique DREPACOEUR entre novembre 2018 et novembre 2022. Cette étude a été déclarée à la CNIL (n◦7830264) et approuvée par un comité d'éthique (2013/NICB).

Les patients étaient adressés au service de Physiologie Cardio-vasculaire pour une suspicion d'anomalie cardiaque, faisant suite à une évaluation clinique ou paraclinique.

Les patients ont bénéficié d'un Holter ECG de 24 heures réalisé la veille d'une hospitalisation de jour incluant un examen clinique, un électrocardiogramme, une échocardiographie transthoracique, un test de marche de 6 minutes et des tests biologiques. Les dossiers complexes ont été discutés lors de réunions de concertation pluridisciplinaire (RCP) et tous les patients présentant un antécédent d'AVC ont également été inclus dans le registre PCDREP et discutés en RCP par des spécialistes en neuro-radiologie afin d'en classifier la cause : vasculopathie cérébrale, thrombose vasculaire ou AVC cardio-emboligène probable.

Le critère d'évaluation principal était la survenue d'une arythmie atriale, définie par la présence d'une activité supraventriculaire ectopique excessive au Holter ECG, une fibrillation atriale de novo ou un

antécédent récent de fibrillation atriale (< 2ans). Les patients présentant une arythmie atriale ont été comparés au groupe de patients n'en présentant pas par des analyses statistiques adaptées.

## Résultats

L'arythmie atriale a été retrouvée chez 34 patients ce qui représente 26 % des patients inclus. L'âge (p=0.001), la dilatation atriale gauche (p<0,001) et les antécédents d'AVC (p=0,009, OR = 6.6) sans vasculopathie cérébrale sous-jacente étaient indépendamment associés à l'arythmie atriale. Un âge supérieur à 47 ans ou une dilatation atriale gauche >55mL/m² étaient prédictifs de l'arythmie atriale avec une valeur positive prédictive à 33 % et une valeur négative prédictive à 92 %.

Cette étude met en évidence que l'arythmie atriale est fréquente chez les patients drépanocytaires adultes et est associée à un risque accru d'AVC. Ces conclusions sont en faveur de la mise en place d'un suivi rythmologique adapté dès le plus jeune âge afin d'éviter de potentielles complications thromboemboliques.

## L'étude en quelques chiffres :

**130** patients drépanocytaires ou thalassémiques $S\beta^0$ (âge moyen **45 ± 12 ans** ; **48 %** d'hommes) inclus dans le registre DREPACOEUR entre novembre 2018 et Novembre 2022 et ayant fait un Holter ECG la veille de l'examen dont :

- **34** patients avec une arythmie atriale de **52 ± 9 ans** dont **26 %** ayant eu un AVC sans vasculopathie cérébrale sous-jacente et présentant un volume atrial gauche de **71 ± 24 mL/m².**
- **96** patients sans arythmie atriale de **42 ± 12 ans** dont **5 %** ayant eu un AVC lié sans vasculopathie cérébrale sous-jacente et présentant un volume atrial gauche de **52 ± 14 mL/m².**

Cette étude a fait l'objet d'une publication en novembre 2024 dans *Blood advances* (https://doi.org/10.1182/bloodadvances.2024013208).

Dr Thomas d'Humières

Département de physiologie, FHU SENEC, Hôpital Henri Mondor, APHP, Créteil

**Pouvez-vous nous décrire le contexte de l'étude et les principaux résultats ? ?**

Une filière de soins DREPACOEUR a été mise en place en 2018 pour les patients drépanocytaires adultes adressés vers l'hôpital de jour du CHU Henri Mondor, par le centre de référence ou par d'autres centres, pour une suspicion d'atteinte cardiovasculaire. Ces patients bénéficient d'une approche cardiovasculaire complète et standardisée incluant une échographie transthoracique, un Holter ECG de 24 heures ainsi qu'une évaluation biologique. Compte tenu du fait que les patients drépanocytaires présentent une anémie et une inflammation chroniques, avec des cavités atriales très dilatées et ce à un jeune âge, il existe un rationnel physiopathologique assez fort pour supposer que ces patients sont exposés de manière plus précoce et plus fréquente à des arythmies que la population générale et donc plus susceptibles de faire des complications thromboemboliques (telles que les AVC).

Le registre DREPACOEUR est avant tout une filière de soins sur lequel nous avons adossé une thématique de recherche. L'étude rapportée ici avait pour but de déterminer s'il y avait bien une association entre l'arythmie atriale et les complications emboliques chez ces patients. Au moment de la soumission de l'article (début 2024), 130 patients étaient inclus, il y en a près de 350 à ce jour.

Cette étude a montré qu'un patient sur quatre faisait un nombre conséquent d'extrasystoles atriales par jour, voire même des troubles du rythme établi (fibrillation atriale). Cet état d'hyperexcitabilité atriale a été fortuitement observé chez 26 % des patients de la cohorte. Ce simple critère est déjà associé dans la population générale à un sur-risque d'AVC et d'arythmie constituée. Les analyses multivariées ont montré que l'âge et la dilatation de l'oreillette étaient deux facteurs de risque indépendamment associés à cette hyperexcitabilité atriale. Cela signifie qu'un patient jeune avec une oreillette dilatée et un patient plus âgé avec une oreillette moins volumineuse seront tous deux exposés à un risque de survenue d'arythmie atriale. En comparant à la population non drépanocytaire, les anomalies électriques surviennent 20 à 30 ans plus tôt.

De plus, les patients présentant une arythmie atriale présentaient un risque d'antécédent d'AVC non lié à une vasculopathie cérébrale de 6,6. Nous ne pouvons pas conclure à un lien de cause à effet, néanmoins c'est un chiffre qui est alarmant car en termes de facteur d'exposition, c'est majeur au sein d'une population très à risque de faire des AVC. Ces résultats suggèrent la mise en place d'un suivi rythmologique adapté de ces patients pour dépister plus précocement ces arythmies atriales et possiblement éviter des complications emboliques. De futures études prospectives et multicentriques permettront d'aboutir à de véritables recommandations.

Nous avons par ailleurs proposé un algorithme pour dépister les arythmies atriales chez les patients drépanocytaires. En bref, si le patient a déjà fait un AVC sans vasculopathie cérébrale, il convient de programmer une surveillance longue durée des signaux électriques avec le Holter ECG pendant 24

heures puis 14 jours et éventuellement un reveal (surveillance sous-cutanée, parfois pendant plusieurs années). Si le patient n'a pas fait d'AVC, il faut se fier à des critères liés à l'oreillette et à l'âge. Pour les patients ayant plus de 47 ans (seuil déterminé par l'étude mais le risque est important dès la quarantaine) et/ou une dilatation de l'oreillette d'au moins 55 mL/m² (sachant que la valeur limite dans la population non drépanocytaire est de 34 mL/m²), il faut systématiquement réaliser un Holter ECG de 24 heures. Si l'examen est rassurant, la surveillance est réduite à un contrôle annuel mais s'il y a déjà beaucoup d'extrasystoles, la surveillance est prolongée pour essayer de dépister l'arythmie avec des Holter ECG de 14 jours.

### → Quelles sont les conclusions thérapeutiques de cette étude ?

La fibrillation atriale est bien prise en charge chez les patients non drépanocytaires, en revanche, les patients drépanocytaires non jamais été inclus dans les essais thérapeutiques. Leur risque thromboembolique, leur tolérance aux anticoagulants et aux traitements spécifiques cardiologiques de l'arythmie ne sont pas connus. Notre étude permet une meilleure identification des patients qui font de l'arythmie atriale. Depuis la création du registre DREPACOEUR, nous commençons à avoir une bonne expérience et connaissance des atteintes cardiaques de la drépanocytose qui étaient jusqu'à présent peu explorées. Cela a permis d'organiser une Réunion de Concertation Pluridiciplinaire nationale mensuelle où les cas de patients sévères sur le plan cardio-vasculaire venant de nombreuses villes sont discutés de manière collégiale afin de choisir la meilleure option thérapeutique. Le traitement anticoagulant pour prévenir le risque thrombotique semble bien toléré à ce jour et nous n'avons pas observé de récidive d'AVC sous traitement. Cela nous conforte dans notre approche mais nous ne pouvons pas affirmer qu'un traitement est supérieur à un autre. La prochaine étape sera de conduire un essai thérapeutique sur cette question.

### → Comment ce remodelage atrial apparait ? Y a-t-il des thérapeutiques pour limiter son évolution ?

Plus les patients sont anémiés, plus ils vont être exposés à une dilatation précoce et importante de l'oreillette gauche. L'association de l'anémie chronique avec l'hémolyse (qui altère le fonctionnement des ventricules) provoque un embouteillage de la circulation sanguine entre l'oreillette et le ventricule et une surcharge de pression au niveau de l'oreillette, tout ceci dépendant de la durée d'évolution de la maladie et de sa sévérité. L'impact des traitements actuels sur le remodelage de l'oreillette n'a jamais été évalué. Il va falloir des années de surveillance car c'est un processus long. On peut supposer que les traitements qui augmentent l'hémoglobine et baissent l'hémolyse vont possiblement limiter la dilatation atriale.

### → Est-ce que l'arythmie atriale doit faire l'objet d'une prise en charge particulière dans d'autres pathologies du globule rouge telles que la thalassémie ?

Il y a une spécificité drépanocytaire avec la profondeur de l'anémie et l'hémolyse intravasculaire qui est quand même très particulière surtout chez les drépanocytaires SS. Les mêmes problématiques existent dans d'autres pathologies du globule rouge, notamment la thalassémie, mais ce n'est pas tout à fait pareil car même en présence d'anémies transfusion dépendantes extrêmement sévères, la dilatation des oreillettes ne sera pas aussi importante. Nous recevons à l'hôpital de jour tous les patients atteints d'une maladie du globule rouge (dont plusieurs patients thalassémiques) pour lesquels une atteinte cardio-vasculaire est suspectée. Nous pouvons supposer que ces patients thalassémiques sont plus exposés à un risque rythmique que la population générale mais moins que la population drépanocytaire.

## Pourquoi l'arythmie atriale est-elle sous-diagnostiquée ?

Les AVC survenant chez les enfants drépanocytaires ont été rapidement liés à la vasculopathie cérébrale qui a été bien étudiée et contre laquelle le bénéfice des échanges transfusionnels a largement été établi. Cependant, en regardant les séries d'AVC publiées, tous les AVC ne s'expliquent pas par une vasculopathie sous-jacente et le contingent restant est peu étudié car la vasculopathie est souvent mise au premier plan et aussi parce que peu de cardiologues se sont intéressés aux troubles rythmiques dans la drépanocytose. Les AVC sont plus attendus chez des patients diabétiques de 65 ans que chez des jeunes de 25 ans. Cette vision nouvelle de surveiller la rythmologie de patients même très jeunes permettra une meilleure prise en charge des troubles rythmiques et une prévention des AVC chez les patients drépanocytaires.

# Appels à projets

## ANR Chaires industrielles (CHIN) - Appel à projets - Edition 2025

| | |
|---|---|
| Budget | 50 000 € |
| Durée | NC |
| Date limite de dépôt des dossiers | 18 mars 2025 à 13h00 |
| Eligibilité | Le projet doit impérativement être mené dans le cadre d'interactions avec une (des) entreprise(s) ; l'AAP est ouvert à toutes les thématiques de recherche et à tous les niveaux de maturité technologique (projets de recherche fondamentale, projets de recherche appliquée ou développement expérimental). |
| Objectif | Stimuler des coopérations entre acteurs publics et acteurs privés de la recherche. Cette stimulation se traduit notamment par le programme « Chaires Industrielles » dédié à la recherche partenariale.<br>• Construire et structurer des projets de recherche collaborative ;<br>• Permettre à un(e) enseignant(e)-chercheur(e) ou à un(e) chercheur(e) de notoriété internationale, français(e) ou étranger(ère), en mobilité ou non, de travailler sur un programme de recherche ambitieux, innovant et de portée industrielle indiscutable.<br>• Assurer une formation par la recherche, en offrant aux doctorants et aux post-doctorants des travaux de recherche menés selon une vision à long terme dans les laboratoires de recherche académique combinés à l'expérience et à l'approche de la recherche des acteurs du monde économique. |

→ Plus d'informations : ICI

---

## MCGRE - Appel à projets généraliste - Edition 2025

| | |
|---|---|
| Budget | 50 000 € |
| Durée | 24 mois maximum |
| Date limite de dépôt des dossiers | 31 mars 2025, à 17h |
| Eligibilité | AAP ouvert aux projets portés par des centres de référence, compétence, laboratoires de recherche et associations membres de la filière ainsi qu'aux étudiants de M2 et aux doctorants en 3e ou 4e année de thèse. |
| Objectif | Soutenir le développement de projets améliorant la connaissance des pathologies du périmètre de la filière et/ou leur prise en charge. |

→ Plus d'informations : ICI

## IReSP : Appel à projets « analyse des politiques de l'autonomie (APOLAU) »

| | |
|---|---|
| Durée et Budget | • Projets de recherche complets : 48 mois / 30 000-250000€<br>• Projets d'amorçage : 18 mois maximum / 15 000-50 000 €<br>• Aide à la mise en place d'un projet européen :<br>18 mois maximum / 15 000-50 000 € |
| Date limite de dépôt des dossiers | 4 avril 2025 à 12h (heure de Paris) |
| Eligibilité | Projets de recherche visant à produire de nouvelles connaissances scientifiques relevant des sciences humaines et sociales (SHS) et de la santé publique.<br>Les projets devront appartenir aux disciplines de sciences humaines et sociales (SHS) et de santé publique. Les travaux relevant de toutes autres disciplines demeurent finançables dans le cadre de travaux interdisciplinaires. |
| Objectif | Soutenir les recherches sur les politiques de l'autonomie à destination des personnes en situation de handicap à tous les âges de la vie et des personnes âgées, leurs proches et les professionnels, dans le contexte français (France métropolitaine et d'outre-mer). |

→ Plus d'informations : ICI

---

## MCGRE - Bourse de prise en charge au 30e congrès de l'EHA

| | |
|---|---|
| Budget | Non applicable |
| Date limite de dépôt des dossiers | 7 avril 2025 |
| Eligibilité | Etre interne au moment de l'EHA dans un service d'hématologie, de médecine interne ou de pédiatrie (orienté hématologie). |
| Objectif | Favoriser la participation d'internes (hématologie, médecine interne et pédiatrie) au 30th EHA Congress ( 12 au 15 juin 2025 à Milan). |

→ Plus d'informations : ICI

## Fondation maladies rares - Appel à candidatures 2025 « Innovation en Intelligence Artificielle & e-Santé pour les Maladies Rares »

| | |
|---|---|
| Budget | Chaque projet sélectionné bénéficiera d'un soutien financier allant jusqu'à 5 000 €, exclusivement dédié à la gratification du stagiaire de master 2. |
| Durée | Non communiqué |
| Date limite de dépôt **des dossiers** | Début mai 2025 |
| Eligibilité | Projets innovants posant une question de recherche et qui répondent à des problématiques spécifiquement liées aux maladies rares Le financement couvrira exclusivement la gratification du stagiaire de master 2. |
| Objectif | Financement de bourses de master 2 en IA et/ou e-santé pour des projets innovants spécifiquement liés aux maladies rares. |

→ Plus d'informations : ICI

---

## Bpifrance : AAP Data Challenges en santé

| | |
|---|---|
| Budget | 200 000 € |
| Durée | Non communiqué |
| Date limite de dépôt des dossiers | 30 mai 2025 |
| Eligibilité | Projets de Data Challenges en santé sur des thématiques médicales d'intérêt portés par des sociétés savantes, des établissements de santé, des groupements de coopération sanitaire ou des structures labellisées « Institut Hospitalo-Universitaire » souhaitant organiser de telles compétitions. |
| Objectif | Valoriser des données de santé par l'intelligence artificielle au bénéfice de la santé des patients par l'organisation de data challenges en santé. Répondre à des problématiques médicales précises à l'aide de l'analyse de jeux de données anonymisées de haute qualité, mis à la disposition de l'ensemble des participants de l'AAP. |

→ Plus d'informations : ICI

## Fondation Mustela - Bourses de Recherche pour l'enfance

| | |
|---|---|
| Budget | 12 000 € (à répartir entre 2 bourses) |
| Durée | Les recherches doivent être menées dans le courant de l'année civile. |
| Date limite de dépôt des dossiers | 6 juin 2025 |
| Eligibilité | Les candidats doivent avoir soutenu leur Master 2 Recherche avant octobre de l'année de la candidature. Ils doivent être en mesure de présenter une attestation le jour de la réunion de sélection qui se déroule en octobre de l'année de la candidature. |
| Objectif | Financer des recherches doctorales/post-doctorales appliquées à l'enfance et à la parentalité ; encourager les travaux sur le développement bio-psycho-social de l'enfant dans ses aspects normaux et pathologiques (à l'exception des maladies somatiques). |

→ Plus d'informations : ICI

---

## Fondation Mustela - Prix de Recherche Action

| | |
|---|---|
| Budget | 8 000 € (La dotation du prix doit permettre la mise en place de l'action primée.) |
| Durée | Les recherches doivent être menées dans le courant de l'année civile. |
| Date limite de dépôt des dossiers | 6 juin 2025 |
| Eligibilité | La démarche de Recherche Action peut être conduite à titre individuel ou par une équipe exerçant en pédiatrie, pédopsychiatrie, maternité, PMI, réseaux scolaires, établissements médico-sociaux, recherche universitaire.<br>Le projet doit constituer une Recherche Action « pilote » pouvant être reproduite sur d'autres sites ou encore inspirer d'autres initiatives à plus grande échelle. Les retombées de la Recherche Action sous forme de diffusion d'information ou de formation seront appréciées. |
| Objectif | Financer des actions innovantes menées par des praticiens de terrain et/ou des chercheurs, autour du développement du jeune enfant dans un environnement familial ou institutionnel dans la thématique Les accueils de l'enfance : approches innovantes (maison de l'enfance, dispositifs périscolaires, pouponnières, autres). |

→ Plus d'informations : ICI

## Fondation Mustela - Bourse de recherche Vulnérabilités

| | |
|---|---|
| Budget | 3 000 € |
| Durée | Les recherches doivent être menées dans le courant de l'année civile. |
| Date limite de dépôt des dossiers | 6 juin 2025 |
| Eligibilité | Tous professionnels intervenant auprès du jeune enfant : pédiatres, pédopsychiatres, médecins, psychologues, neuropsychologues, sociologues, psychomotricien(ne)s, orthophonistes... |
| Objectif | Financer des projets de recherche fondamentale ou appliquée pour lutter contre la vulnérabilité des jeunes enfants |

→ Plus d'informations : ICI

---

Les appels à projets sont régulièrement mis à jour sur le site internet de la filière MCGRE, à l'adresse suivante :

https://filiere-mcgre.fr/espace-professionnels-de-sante/appels-a-projets/

# Bibliographie

La bibliographie proposée dans ce bulletin recherche est une sélection d'articles dont la majorité sont entrés dans PubMed/parus de septembre à décembre 2024 inclus.

## Anémie dysérythropoïétique congénitale

**Majeed syndrome: first description in a patient of central-European ancestry**
Drago E, Bertoni A, Grossi A, Damasio MB, Anfigeno L, *et al.*
Rheumatology (Oxford). 2024 Sep 10:keae480. doi: 10.1093/rheumatology/keae480

## Anémie liée au métabolisme du fer et anémie sidéroblastique

**An erythroid-specific lentiviral vector improves anemia and iron metabolism in a new model of XLSA**
**Castruccio Castracani C, Breda L, Papp TE, Guerra A, Radaelli E, et al.**
**Blood. 2025 Jan 2;145(1):98-113. doi: 10.1182/blood.2024025846**

## Anomalies de la membrane du globule rouge

**Overview on Hereditary Spherocytosis Diagnosis**
Polizzi A, Dicembre LP, Failla C, Matola TD, Moretti M, *et al.*
Int J Lab Hematol. 2025 Feb;47(1):18-25. doi: 10.1111/ijlh.14376

Hereditary stomatocytosis in the general population: A genetically based prevalence estimate from a 109 039 individual Danish cohort
Mottelson M, Helby J, Petersen J, Nordestgaard BG, Bojesen SE, *et al.*
Am J Hematol. 2025 Jan;100(1):152-157. doi: 10.1002/ajh.27508

The efficacy of partial versus total splenectomy in the treatment of hereditary spherocytosis in children: a systematic review and meta-analysis
Tang X, Xue J, Zhang J, Zhou J.
Pediatr Surg Int. 2024 Oct 29;40(1):280. doi: 10.1007/s00383-024-05879-7

**Iron overload in hereditary spherocytosis: Are genetic factors the cause?**
Donaty L, Giansily-Blaizot M, Bertchansky I, Cunat S, Azoury V, *et al.*
Br J Haematol. 2024 Dec 4. doi: 10.1111/bjh.19941

Gallbladder preserving cholelithotomy in children with hereditary spherocytosis complicated by gallstones: a single-center retrospective study
Tang R, Zhou CX, Yang Y, Bian J, Meng LX, *et al.*
Front Pediatr. 2024 Nov 20;12:1457927. doi: 10.3389/fped.2024.1457927

**Evaluation of endocrine changes and insulin release in patients with hereditary spherocytosis**
Ağırman Z, Temiz F, Acıpayam C, Akkececi N.
J Paediatr Child Health. 2024 Dec 12. doi: 10.1111/jpc.16744

**Relevance of the E756del common variant in the PIEZO1 gene for haemolytic anaemia and hepatic iron overload**
Esposito FM, D'Onofrio V, Rosato BE, Marra R, Nostroso A, *et al.*
Br J Haematol. 2025 Jan;206(1):337-341. doi: 10.1111/bjh.19886

**RAS signaling pathway is essential in regulating PIEZO1-mediated hepatic iron overload in dehydrated hereditary stomatocytosis**
Rosato BE, D'Onofrio V, Marra R, Nostroso A, Esposito FM, *et al.*
Am J Hematol. 2025 Jan;100(1):52-65. doi: 10.1002/ajh.27523

## Déficit en glucose-6-phosphate déshydrogénase

**Biochemical screening of glucose-6-phosphate dehydrogenase deficiency in borderline cases: Complementary inputs of standardization enzymes and comparison with genetic status**
Raynor A, Jacquel B, François S, Fellahi S, Mouri N, *et al.*
Br J Haematol. 2025 Jan 6. doi: 10.1111/bjh.19990

**Glucose-6-phosphate dehydrogenase deficiency detection using fluorocytometric assay: Evaluation after 1 year of clinical implementation**
Souissi M, Bera E, Boutet C, Chatellier C, Conte C, *et al.*
Cytometry B Clin Cytom. 2024 Oct 2. doi: 10.1002/cyto.b.22207

**Diagnostic accuracy of the point-of-care standard G6PD test™ (SD Biosensor) for glucose-6-phosphate dehydrogenase deficiency: a systematic review and meta-analysis**
Martínez JC, Vélez-Marín V, Lopez-Perez M, Patiño-Lugo DF, Florez ID.
Malar J. 2024 Nov 2;23(1):327. doi: 10.1186/s12936-024-05144-1

**Functional analysis of G6PD variants associated with low G6PD activity in the All of Us Research Program**
Powell NR, Geck RC, Lai D, Shugg T, Skaar TC, Dunham MJ.
Genetics. 2024 Nov 28;228(4):iyae170. doi: 10.1093/genetics/iyae170

**Molecular epidemiological characteristics, variant spectrum and genotype-phenotype correlation of glucose-6-phosphate dehydrogenase deficiency in China: A population-based multicenter study using newborn screening**
Tan M, Liu X, Zhang Y, Yin Y, Chen T, *et al.*
PLoS One. 2024 Oct 22;19(10):e0310517. doi: 10.1371/journal.pone.0310517

**Controversies in our understanding of extreme hyperbilirubinemia in glucose-6-phosphate dehydrogenase-deficient neonates**
Kaplan M, Kassirer Y, Hammerman C.
Pediatr Res. 2024 Oct 6. doi: 10.1038/s41390-024-03611-8

**Evaluation of Glucose 6-Phosphate Dehydrogenase, Pyruvate Kinase, and New Generation Inflammation Biomarkers in Prolonged Neonatal Jaundice**
Okuyan O, Dumur S, Elgormus N, Uzun H.
Medicina (Kaunas). 2024 Sep 12;60(9):1491. doi: 10.3390/medicina60091491

**Genetic variation features of neonatal hyperbilirubinemia caused by inherited diseases**
You JY, Xiong LY, Wu MF, Fan JS, Fu QH, Qiu MH.
World J Clin Pediatr. 2024 Dec 9;13(4):98462. doi: 10.5409/wjcp.v13.i4.98462

**G6PD trait: societal importance to ensure well-being of female heterozygotes for health and childbirth**
Bhutani VK.
Pediatr Res. 2024 Nov 13. doi: 10.1038/s41390-024-03712-4

**Health disparities in diabetes treatment: The challenge of G6PD deficiency**
Israel A, Raz I, Green I, Golan-Cohen A, Berkovitch M, *et al.*
Diabetes Res Clin Pract. 2025 Jan;219:111965. doi: 10.1016/j.diabres.2024.111965

**The Impact of the G6PD Gene Mutations in Patients with Chronic Hepatitis C Infection Treated with Direct-Acting Antivirals: A Multicenter Observational Study**
Smirne C, Crobu MG, Gerevini C, Berton AM, Rapetti R, *et al.*
Genes (Basel). 2024 Aug 24;15(9):1116. doi: 10.3390/genes15091116

**Case report: Dichloroacetate-induced methaemoglobinaemia in a G6PD-deficient neonate**
Tan ZL, Ng NBH, Ong JSM.
Pediatr Blood Cancer. 2025 Jan;72(1):e31408. doi: 10.1002/pbc.31408

**Use of trimethoprim-sulfamethoxazole for treating *Pneumocystis jirovecii pneumonia* in a patient with glucose-6-phosphate dehydrogenase deficiency: a case report**
Wang L, Xie X, Li Z, Li Y.
Front Med (Lausanne). 2024 Sep 10;11:1443645. doi: 10.3389/fmed.2024.1443645

**Increased exercise tolerance in humanized G6PD-deficient mice**
Cendali FI, Lisk C, Dzieciatkowska M, LaCroix IS, Reisz JA, *et al.*
Blood Adv. 2025 Jan 28;9(2):321-334. doi: 10.1182/bloodadvances.2024013968

## Déficit en pyruvate kinase

**How We Treat Hemolytic Anemia Due to Pyruvate Kinase Deficiency**
Tama-Shekan S, Moreno V, Saba L, Chaulagain CP.
Hematol Rep. 2024 Aug 31;16(3):559-567. doi: 10.3390/hematolrep16030054

**Case report: Modified transplantation for pediatric patients with pyruvate kinase deficiency**
Pang Y, Qi X, Qin J, Zhai X, Wang R, *et al.*
Front Immunol. 2024 Nov 20;15:1493398. doi: 10.3389/fimmu.2024.1493398

## Drépanocytose

**Variability in Notification of Positive Newborn Screening Results for Sickle Cell Trait Across the United States**
Scott JL, Christian J, Plazas Montana M, Miller YM, Naik RP.
Adv Hematol. 2024 Dec 19;2024:3854629. doi: 10.1155/ah/3854629

**Evaluation of a New Tandem Mass Spectrometry Method for Sickle Cell Disease Newborn Screening**
Renoux C, Roland E, Ruet S, Zouaghi S, Michel M, *et al.*
Int J Neonatal Screen. 2024 Nov 26;10(4):77. doi: 10.3390/ijns10040077

**A Modular Genetic Approach to Newborn Screening from Spinal Muscular Atrophy to Sickle Cell Disease-Results from Six Years of Genetic Newborn Screening**
Bzdok J, Czibere L, Burggraf S, Pauly N, Maier EM, *et al.*
Genes (Basel). 2024 Nov 13;15(11):1467. doi: 10.3390/genes15111467

**Newborn Screening for Sickle Cell Disease in Catalonia between 2015 and 2022-Epidemiology and Impact on Clinical Events**
González de Aledo-Castillo JM, Argudo-Ramírez A, Beneitez-Pastor D, Collado-Gimbert A, Almazán Castro F, *et al.*, On Behalf Of The Sickle Cell Disease Newborn Screening Group Of Catalonia.
Int J Neonatal Screen. 2024 Oct 3;10(4):69. doi: 10.3390/ijns10040069

**Newborn screening initiatives for sickle cell disease in Africa**
Nnodu OE, Okeke CO, Isa HA.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):227-233. doi: 10.1182/hematology.2024000548

**Sustainability of newborn screening for sickle cell disease in resource-poor countries: A systematic review**
Okeke CO, Okeke C, Asala S, Ofakunrin AOD, Ufelle S, Nnodu OE.
PLoS One. 2024 Sep 6;19(9):e0305110. doi: 10.1371/journal.pone.0305110

**Acceptability, barriers and facilitators of using dried blood spots-point-of-care testing for sickle cell disease in Africa: an implementation science protocol for a multinational qualitative study**
Nnodu OE, Munung NS, Chirande L, Chunda-Liyoka C, Kiguli S, *et al.*; SickleInAfrica Consortium.
BMJ Open. 2024 Nov 7;14(11):e089056. doi: 10.1136/bmjopen-2024-089056

**Point-of-care diagnostic test accuracy in children and adolescents with sickle cell disease: A systematic review and meta-analysis**
Bagnall R, Guy D, Morgan RL, Babatunde I, Nevière A, *et al.*
Blood Rev. 2025 Jan;69:101243. doi: 10.1016/j.blre.2024.101243

**Systemic Inflammatory Diseases in Children with Sickle Cell Disease: A French Multicenter Observational Study on Diagnostic and Therapeutic Issues**
Vinit C, Guitton C, De Montalembert M, Benhaim P, Amor-Chelihi L, *et al.*
Pediatr Blood Cancer. 2025 Jan 27:e31563. doi: 10.1002/pbc.31563

**Trends in Sickle Cell Disease Mortality: 1979-2020**
Karkoska KA, McGann PT.
Pediatrics. 2024 Dec 1;154(6):e2024067341. doi: 10.1542/peds.2024-067341

**The Prevalence of Sickle Cell Disease in Colorado and Methodologies of the Colorado Sickle Cell Data Collection Program: Public Health Surveillance Study**
Miller JI, Hassell KL, Kellar-Guenther Y, Quesada S, West R, Sontag M.
JMIR Public Health Surveill. 2024 Dec 9;10:e64995. doi: 10.2196/64995

**Infrequent Resolution of Vaso-Occlusive Crises in Routine Clinical Care Among Patients Mimicking the Exa-Cel Trial Population: A Cohort Study of Medicaid Enrollees**
Mahesri M, Lee SB, Levin R, Imren S, Zhang L, *et al.*
Clin Pharmacol Ther. 2024 Dec;116(6):1572-1579. doi: 10.1002/cpt.3449

**Clinical Burden and Health Care Resource Utilization Associated With Managing Sickle Cell Disease With Recurrent Vaso-occlusive Crises in England**
Udeze C, Ly NF, Ingleby FC, Fleming SD, Conner SC, *et al.*
Clin Ther. 2025 Jan;47(1):29-36. doi: 10.1016/j.clinthera.2024.09.023

**Trends and hospital practice variation for analgesia for children with sickle cell disease with vaso-occlusive pain episodes: An 11-year analysis**
Eltorki M, Hall M, Ramgopal S, Chaudhari PP, Badaki-Makun O, *et al.*
Am J Emerg Med. 2024 Dec;86:129-134. doi: 10.1016/j.ajem.2024.10.028

**Safety and efficacy of ketamine use in patients with vaso-occlusive crisis: A systematic review and meta-analysis**
Calderon Martinez E, Zachariah Saji S, Campos Carmona T, Abbagoni V, Salman M, *et al.*
EJHaem. 2024 Nov 25;5(6):1312-1321. doi: 10.1002/jha2.1050

**Efficacy of a vein visualisation device for facilitating peripheral venous line placement in adult patients with sickle cell disease: A randomised clinical trial**
Corbasson A, Fischer E, Vilfaillot A, Da Costa AB, Charmettan M, *et al.*
Int J Nurs Stud. 2024 Dec 19;163:104988. doi: 10.1016/j.ijnurstu.2024.104988

**Lactated Ringer vs Normal Saline Solution During Sickle Cell Vaso-Occlusive Episodes**
Alwang AK, Law AC, Klings ES, Cohen RT, Bosch NA.
JAMA Intern Med. 2024 Nov 1;184(11):1365-1372. doi: 10.1001/jamainternmed.2024.4428

**Development of a Calculator to Determine Individualized Opioid Doses for Treatment of Vaso-Occlusive Episodes for Sickle Cell Disease in the Emergency Department**
Kavanagh PL, Strouse JJ, Paice JA, Ibemere SO, Tanabe P.
Jt Comm J Qual Patient Saf. 2025 Jan;51(1):74-79. doi: 10.1016/j.jcjq.2024.10.002

**A Telemedicine Triage Model for Infusion Center-Based Management of Adult Sickle Cell Pain Episodes**
Bliamptis J, You A, Groesbeck J, Sacknoff S, Byrne S, Gopal S.
Blood Adv. 2024 Dec 6:bloodadvances.2024013973. doi: 10.1182/bloodadvances.2024013973

**Protocol for a multicentric, double-blind, randomised controlled trial of hyperbaric oxygen therapy (HBOT) versus sham for treating vaso-occlusive crisis (VOC) in sickle cell disease (SCD) in patients aged 8 years or older (HBOT-SCD study)**
Stirnemann J, Serratrice J, Mann T, Louge P, Christophe C, *et al.*
BMJ Open. 2024 Nov 28;14(11):e084825. doi: 10.1136/bmjopen-2024-084825

**Asthma Is an Independent Risk Factor for Acute Chest Syndrome in Children with Sickle Cell Disease in French Guiana**
Bafunyembaka G, Nacher M, Maniassom C, Birindwa AM, Elenga N.
Children (Basel). 2024 Dec 19;11(12):1541. doi: 10.3390/children11121541

**Exhaled breath analysis during vaso-occlusive events in sickle cell disease**
Vuong C, Brinkman P, Heijboer H, Nur E, Eckhardt CL, *et al.*
Br J Haematol. 2024 Dec 15. doi: 10.1111/bjh.19942

**Inpatient Management of Pain Episodes in Children with Sickle Cell Disease: A Review**
Barnawi Z, Thomas R, Peddinti R, Abou Baker N.
Children (Basel). 2024 Sep 10;11(9):1106. doi: 10.3390/children11091106

**Burden of vaso-occlusive crisis, its management and impact on quality of life of Indian sickle cell disease patients**
Seth T, Udupi S, Jain S, Bhatwadekar S, Menon N, *et al.*
Br J Haematol. 2025 Jan;206(1):296-309. doi: 10.1111/bjh.19829

**Sex-stratified association of variants in the serotonin 1A receptor gene with acute crisis pain among African American patients with sickle cell disease**
Sadhu N, He Y, Kashyap Y, Ilktach G, Wang MA, *et al.*
Exp Hematol. 2024 Nov 28;142:104692. doi: 10.1016/j.exphem.2024.104692

**Elevated posterior insula glutamate in patients with sickle cell disease**
Zhou X, Ichesco E, Pucka AQ, Liu Z, O'Brien AR, *et al.*
J Pain. 2024 Nov 28;27:104743. doi: 10.1016/j.jpain.2024.104743

**Safety and Tolerability of Intravenous Lidocaine Infusions as Opioid Adjunct for Children Hospitalized With Sickle Cell Vaso-Occlusive Pain**
Agbakwuru U, AuBuchon JD, Toebe B, LaBarge A, Di Paola J, Hulbert ML.
Pediatr Blood Cancer. 2025 Mar;72(3):e31458. doi: 10.1002/pbc.31458

**Development, implementation, and acceptability of a bedside mindfulness intervention for adults with sickle cell disease**
Pavlacic JM, Meredith LR, Boylan AM, Kilpatrick RL, Abrams CM, Rheingold AA.
Psychol Health Med. 2025 Jan;30(1):108-118. doi: 10.1080/13548506.2024.2411637

**Biomarkers to Differentiate Acute Chest Syndrome From Vaso-Occlusive Crisis in Children With Sickle Cell Disease**
Wang K, Olave N, Aggarwal S, Oh JY, Patel RP, *et al.*
Eur J Haematol. 2025 Feb;114(2):325-333. doi: 10.1111/ejh.14342

**Beyond IV push: alternative methods for management of acute pain in SCD**
Azul M, Brandow AM.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):611-617. doi: 10.1182/hematology.2024000585

**Cognitive considerations for adults with sickle cell disease completing the brief pain inventory**
O'Brien JA, Drake JA, Bearden DJ, Ono KE, Sil S, *et al.*
Pain Rep. 2024 Dec 9;10(1):e1189. doi: 10.1097/PR9.0000000000001189

**Emergency department intravenous fluid resuscitation and renal outcomes among adults with sickle cell disease**
Carden MA, Lebensburger J, Rosamond W, Tanabe P, Derebail VK.
Am J Hematol. 2025 Jan;100(1):144-148. doi: 10.1002/ajh.27509

**Characteristics and outcomes of children and young adults with sickle cell disease supported with extracorporeal membrane oxygenation (ECMO): An updated analysis of the ELSO registry**
Remy T, Jegard J, Chenouard A, Maminirina P, Liet JM, *et al.*
Artif Organs. 2024 Oct 3. doi: 10.1111/aor.14880

**Respiratory management of acute chest syndrome in children with sickle cell disease**
Ahmed B, Arigliani M, Gupta A.
Eur Respir Rev. 2024 Sep 18;33(173):240005. doi: 10.1183/16000617.0005-2024

**Secretory Phospholipase A2 as a Promising Biomarker for Predicting Acute Chest Syndrome in Children With Sickle Cell Disease: A Systematic Review and Meta-Analysis**
Alsabri M, Elsnhory AB, Alattar O, Ahmed N, Ashraf S, Alassri R.
Cureus. 2024 Sep 10;16(9):e69053. doi: 10.7759/cureus.69053

**Physician perspectives about the diagnosis and management of acute chest syndrome**
Bhasin N, LeBlanc DM, Yates S, Eichbaum Q, Pham A, *et al.*
Transfusion. 2024 Nov;64(11):2095-2103. doi: 10.1111/trf.18034

**Prevent Acute Chest Syndrome checklist (PACScheck): A quality improvement initiative to reduce acute chest syndrome**
Morrone K, Strumph K, Pisacano C, Briggs J, Zipper R, *et al.*
Pediatr Blood Cancer. 2025 Jan;72(1):e31378. doi: 10.1002/pbc.31378

**Children with sickle cell disease and fever but no respiratory symptoms: Is a chest x-ray needed?**
Khan NI, Coleman KD, Nimmer M, Brazauskas R, Brousseau DC.
Am J Emerg Med. 2024 Dec 13;89:75-77. doi: 10.1016/j.ajem.2024.12.019

**Lung ultrasound score to predict development of acute chest syndrome in children with sickle cell disease**
Vieira PPMG, Braga JAP, Regacini R.
Hematol Transfus Cell Ther. 2024 Nov;46 Suppl 5(Suppl 5):S239-S245. doi: 10.1016/j.htct.2024.07.003

**The prevalence of opioid misuse diagnostic codes in children with sickle cell disease**
Tantoco AM, Badawy SM, Lee CK, Merz J, Steed M, *et al.*
Pediatr Hematol Oncol. 2024 Dec 11:1-10. doi: 10.1080/08880018.2024.2437045

**Parental Perceptions and Practices Regarding Pain Management and Medical Marijuana Use in Patients With Sickle Cell Disease**
Cinquepalmi L, Ayeni A, Melville L, Kelly C.
Clin Pediatr (Phila). 2024 Dec 27:99228241304464. doi: 10.1177/00099228241304464

**Temporal summation of pain in sickle cell disease: comparison of adolescents and young adults with chronic vs. infrequent pain**
Karlson C, Dickens H, Williams-Kirkwood W, Mascaro M, Jackson E, *et al.*
J Pediatr Psychol. 2024 Dec 1;49(12):882-890. doi: 10.1093/jpepsy/jsae090

**The use of abstract animations and a graphical body image for assessing pain outcomes among adults with sickle cell disease**
O'Brien JA, Jonassaint CR, Parchuri E, Lalama CM, Badawy SM, *et al.*
J Pain. 2024 Oct 22;26:104720. doi: 10.1016/j.jpain.2024.104720

**Knowledge and Experience of In-Home Virtual Reality for Chronic Pain in Sickle Cell Disease**
Matthie N, Jenerette C.
J Pain. 2024 Dec;25(12):104668. doi: 10.1016/j.jpain.2024.104668

**Osteopathic Manipulative Treatment: A Safe and Feasible Adjunct in Sickle Cell Disease Pain Management**
Brown A, Goubeaux D, Jacob SA, Belsky JA.
Pediatr Blood Cancer. 2025 Jan;72(1):e31433. doi: 10.1002/pbc.31433

**Mast cell extracellular trap formation underlies vascular and neural injury and hyperalgesia in sickle cell disease**
Argueta DA, Tran H, Goel Y, Nguyen A, Nguyen J, *et al.*
Life Sci Alliance. 2024 Sep 6;7(11):e202402788. doi: 10.26508/lsa.202402788

**Impact of transcranial Doppler screening on stroke prevention in children and adolescents with sickle cell disease: A systematic review and meta-analysis**
Guy D, Bagnall R, Morgan RL, Babatunde I, Nevière A, *et al.*
Blood Rev. 2025 Jan;69:101253. doi: 10.1016/j.blre.2024.101253. Erratum in: Blood Rev. 2025 Jan 17:101258. doi: 10.1016/j.blre.2025.101258

**Posterior cerebral circulation in children with sickle cell anemia: an uncharted territory**
Salimbeni AF, De Rosa L, Volpato A, Viaro F, Pieroni A, *et al.*
Haematologica. 2024 Oct 24. doi: 10.3324/haematol.2024.285773

**Residual Cerebrovascular Morbidity Despite Treatment in Pediatric Sickle Cell Disease Highlights Opportunities for Earlier, Intensified Monitoring and Treatment**
Reggiani G, Coppadoro B, Boaro MP, Trapanese R, Baido I, *et al.*
Am J Hematol. 2025 Jan 16. doi: 10.1002/ajh.27596

**Unruptured intracranial aneurysms in pediatric sickle cell disease: clinical and MR imaging follow-up of 296 patients**
Khatri D, Zampolin R, Fortunel A, Lee SK.
J Neurosurg Pediatr. 2024 Sep 27;34(6):585-590. doi: 10.3171/2024.7.PEDS24231

**Cerebral Oxygen Metabolic Stress in Children and Adults With Large Vessel Vasculopathy Due to Sickle Cell Disease**
Wang Y, Fellah S, Reis M, Guilliams KP, Fields ME, *et al.*
Neurology. 2024 Dec 10;103(11):e210032. doi: 10.1212/WNL.0000000000210032

**Cerebral vascular shunting and oxygen metabolism in sickle cell disease**
Song AK, Richerson WT, Aumann MA, Waddle SL, Jones RS, *et al.*
Blood Adv. 2025 Jan 28;9(2):386-397. doi: 10.1182/bloodadvances.2024014201

**Longitudinal outcomes of chronically transfused adults with sickle cell disease and a history of childhood stroke**
Jones JM, Wool J, Crowe EP, Bloch EM, Pecker LH, Lanzkron S.
Transfusion. 2024 Dec;64(12):2260-2269. doi: 10.1111/trf.18041

**Stroke Prevention in Hispanic Children with Sickle Cell Anemia: the SACRED trial**
Nieves R, Latham T, Marte NM, Berges Morales M, Sanchez LM, *et al.*
Blood Adv. 2025 Jan 16:bloodadvances.2024014327. doi: 10.1182/bloodadvances.2024014327

**Children and Adolescents With Sickle Cell Disease and Skull Infarction: A Systematic Review**
Perez AM, Garcia-Guaqueta DP, Setty BN, Neri C, Torres AR.
J Child Neurol. 2024 Oct;39(11-12):433-439. doi: 10.1177/08830738241274352

**Management of Spontaneous Epidural Hematoma in the Setting of Vaso-occlusive Crisis Among Pediatric Patients With Sickle Cell Disease: A Case Series and Scoping Literature Review**
Patel R, Reisert H, Keymakh M, Drakou E, Briggs J, *et al.*
J Pediatr Hematol Oncol. 2025 Jan 1;47(1):38-46. doi: 10.1097/MPH.0000000000002975

**Cerebral Hemodynamic Responses to Disease-Modifying and Curative Sickle Cell Disease Therapies**
Aumann MA, Richerson W, Song AK, Martin D, Davis LT, *et al.*
Neurology. 2025 Jan 14;104(1):e210191. doi: 10.1212/WNL.0000000000210191

**Detectability of white matter cerebral blood flow using arterial spin labeling MRI in patients with sickle cell disease: Relevance of flow territory, bolus arrival time, and hematocrit**
Richerson WT, Aumann M, Song AK, Eisma JJ, Davis S, *et al.*
J Cereb Blood Flow Metab. 2024 Sep 10:271678X241270283. doi: 10.1177/0271678X241270283

**Associations of haematological and inflammatory biomarkers with brain volume in patients with sickle cell anaemia: A cross-sectional retrospective study**
Majrashi NA, Hendi AM, Dhayihi TM, Khamesi AM, Masmali MA, *et al.*
Trop Med Int Health. 2024 Dec;29(12):1026-1040. doi: 10.1111/tmi.14056

**Stroke Prevention and Treatment for Youth with Sickle Cell Anemia: Current Practice and Challenges and Promises for the Future**
Creary S, Chung MG, Villella AD, Lo WD.
Curr Neurol Neurosci Rep. 2024 Nov;24(11):537-546. doi: 10.1007/s11910-024-01372-9

**A contemporary evaluation of the frequency & factors associated with overt stroke across the lifespan: A Ghanaian sickle cell disease registry analysis**
Sarfo FS, Paintsil V, Nyanor I, Asafo-Adjei EK, Ahmed EA, *et al.*
J Neurol Sci. 2024 Nov 15;466:123263. doi: 10.1016/j.jns.2024.123263

**The Association Between Sickle Cell Anemia and Cognitive Dysfunction: A Systematic Review**
Alpakra M, Hamed NF, Almakki ZE, Al Bakrah E.
Cureus. 2024 Sep 10;16(9):e69104. doi: 10.7759/cureus.69104

**A clinical evaluation program to monitor neurocognitive risk in children and adolescents with sickle cell disease**
Longoria JN, Schreiber JE, Potter B, Raches D, MacArthur E, *et al.*
Clin Neuropsychol. 2024 Sep 4:1-21. doi: 10.1080/13854046.2024.2399861

**Utility of Caregiver Signaling Questions to Detect Neurocognitive Impairment in Children with Sickle Cell Disease**
Dandar CM, Kearson L, Longoria JN, Potter B, King AA, *et al.*
Blood Adv. 2024 Nov 13:bloodadvances.2024013958. doi: 10.1182/bloodadvances.2024013958

**Depression, sleep and pain affect instrumental activities of daily living through cognitive functioning in adults with sickle cell disease: A report from the Sickle Cell Disease Implementation Consortium**
Longoria JN, Howell KE, Porter JS, Treadwell M, King AA, *et al.*; Sickle Cell Disease Implementation Consortium.
Br J Haematol. 2024 Nov 20. doi: 10.1111/bjh.19881

**Brain Age Modeling and Cognitive Outcomes in Young Adults With and Without Sickle Cell Anemia**
Ford AL, Fellah S, Wang Y, Unger-Levinson K, Hagan M, *et al.*
JAMA Netw Open. 2025 Jan 2;8(1):e2453669. doi: 10.1001/jamanetworkopen.2024.53669

**The Interplay Between Ophthalmic and Systemic Outcomes in Patients With Sickle Cell Disease and Concurrent Retinopathy-A Population-Based Study**
Nangia P, Wai KM, Scott AW, Rahimy E, Mruthyunjaya P.
Am J Hematol. 2025 Jan 4. doi: 10.1002/ajh.27552

**Clinical and laboratory risk factors for sickle cell retinopathy and maculopathy: a scoping review of the current evidence**
Brandsen RP, Diederen RMH, Kocabas G, Nur E, Malekzadeh A, *et al.*
Haematologica. 2024 Dec 19:0. doi: 10.3324/haematol.2024.286420

**Healthcare Utilization Patterns in Sickle Cell Patients and Their Association With Sickle Cell Retinopathy**
Minniti CP, Suman PD, Dahlan K, Crouch A, Goodman M, Mian UK.
Eur J Haematol. 2024 Dec 23. doi: 10.1111/ejh.14369

**Sickle Cell Retinopathy Lost to Follow-up Study**
Wong JCY, Sivalingam MD, Griffin M, Magagna J, Momenaei B, *et al.*
Retina. 2024 Dec 9. doi: 10.1097/IAE.0000000000004369

**Computer-assisted evaluation of retinal vessel tortuosity in children with sickle cell disease without retinopathy**
Raffa LH, Raffa EH, Hervella ÁS, Ramos L, Novo J, *et al.*
Microvasc Res. 2025 Jan;157:104752. doi: 10.1016/j.mvr.2024.104752

**Clinical Outcomes of Sickle Cell Disease Patients With Myocardial Infarction Undergoing Percutaneous Coronary Intervention: A Nationwide Analysis**
Akinti OM, Perry JC, Ediale T, Rehman M, Aiwuyo HO.
Cureus. 2024 Sep 15;16(9):e69465. doi: 10.7759/cureus.69465

**Respiratory phenotype and health care utilization patterns by adults with sickle cell disease**
Agawu A, Nortey N, Jacobs C, Zebrowski A, Lin MP, Glassberg J.
Blood Adv. 2025 Jan 14;9(1):143-150. doi: 10.1182/bloodadvances.2023010808

**Sickle cell anaemia therapy in 2025**
Schechter AN.
Br J Haematol. 2024 Dec 22. doi: 10.1111/bjh.19933

**The modern use of hydroxyurea for children with sickle cell anemia**
Quinn CT, Ware RE.
Haematologica. 2025 Jan 9. doi: 10.3324/haematol.2023.284633

**Adherence outcomes of a liquid hydroxyurea delivery program in a pediatric population**
Sacta MA, Sarvode S, Kehasse A, Mothi SS, Shah B, Sobota A.
Pediatr Blood Cancer. 2024 Dec;71(12):e31348. doi: 10.1002/pbc.31348

**Hydroxyurea for Children and Adults with Hemoglobin SC Disease**
Dei-Adomakoh YA, Segbefia CI, Latham TS, Lane AC, Dzefi-Tettey K, *et al.*
NEJM Evid. 2025 Feb;4(2):EVIDoa2400402. doi: 10.1056/EVIDoa2400402

**Impact of hydroxycarbamide treatment on the whole-blood transcriptome in sickle cell disease**
Bhat V, Potdar AA, Yu GK, Gibson G, Sheehan VA.
Br J Haematol. 2024 Nov 17. doi: 10.1111/bjh.19839

**CYB5R3 T117S tempers fetal hemoglobin induction by hydroxyurea in patients with sickle cell disease**
Chowdhury FA, Sharma M, Schafer D, Nouraie SM, Gladwin MT, *et al.*
Blood Adv. 2024 Dec 10;8(23):6098-6103. doi: 10.1182/bloodadvances.2024013801

**Hydroxyurea Pharmacokinetic Evaluation in Patients with Sickle Cell Disease**
Di Grazia D, Mirabella C, Chiara F, Caudana M, Shelton Agar FMA, *et al.*
Pharmaceuticals (Basel). 2024 Oct 17;17(10):1386. doi: 10.3390/ph17101386

**CureSCi Metadata Catalog-finding and harmonizing studies for secondary analysis of hydroxyurea use for sickle cell disease**
Wu X, Stratford J, Kesler K, Ives C, Hendershot T, *et al.*
bioRxiv [Preprint]. 2024 Aug 19:2024.08.15.608203. doi: 10.1101/2024.08.15.608203

**Hydroxyurea blunts mitochondrial energy metabolism and osteoblast and osteoclast differentiation exacerbating trabecular bone loss in sickle cell mice**
Tripathi AK, Dabeer S, Song J, Vikulina T, Roser-Page S, *et al.*
Cell Death Dis. 2024 Dec 18;15(12):907. doi: 10.1038/s41419-024-07296-z

**Transcriptional regulators of fetal hemoglobin**
Santos GPD, Rabi LT, Bezerra AA, da Cunha MR, Iatecola A, Fernandes VAR.
Hematol Transfus Cell Ther. 2024 Nov;46 Suppl 5(Suppl 5):S258-S268. doi: 10.1016/j.htct.2024.06.001

**Identification of small molecule agonists of fetal hemoglobin expression for the treatment of sickle cell disease**
Yang JP, Toughiri R, Gounder AP, Scheibe D, Petrus M, *et al.*
PLoS One. 2024 Nov 6;19(11):e0307049. doi: 10.1371/journal.pone.0307049

**Practical guide for disease-modifying medication management of children and adolescents with sickle cell disease**
Walden J, Creary S.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):604-610. doi: 10.1182/hematology.2024000587

**Fetal Hemoglobin Decrease During Voxelotor Treatment**
De Luna G, Habibi A, Moutereau S, Martino S, Alhamrouni J, *et al.*
Eur J Haematol. 2025 Feb;114(2):264-266. doi: 10.1111/ejh.14332

**Voxelotor-induced pulmonary embolism**
Schiro P, Barus R, Chouchana L, Sari R, Cougoul P, *et al.*
Therapie. 2024 Oct 19:S0040-5957(24)00166-5. doi: 10.1016/j.therap.2024.10.050

**The Voxelotor Effect: Decreased Affinity for New Drugs for Sickle Cell Disease?**
Quinn CT, Ware RE.
Pediatr Blood Cancer. 2025 Mar;72(3):e31475. doi: 10.1002/pbc.31475

**Effect of voxelotor on cerebral perfusion and cerebral oxygen metabolism and cardiac stress in adult patients with sickle cell disease**
Konté K, Afzali-Hashemi L, Baas KPA, Schrantee A, Wood JC, *et al.*
Am J Hematol. 2025 Jan;100(1):78-84. doi: 10.1002/ajh.27522

**Promising role of Voxelotor in managing sickle cell disease in children: a narrative review**
Agrawal A, Jadon G, Singh J, Janjua D.
Clin Exp Pediatr. 2024 Nov 13. doi: 10.3345/cep.2024.00500

**Effects of GBT1118, a voxelotor analog, on bone disease in sickle cell disease mice**
Xiao L, He W, Hurley MM.
Sci Rep. 2024 Sep 27;14(1):22330. doi: 10.1038/s41598-024-69589-9

**GBT1118, a Voxelotor Analog, Ameliorates Hepatopathy in Sickle Cell Disease**
Haroun E, Lim SH, Dutta D.
Medicina (Kaunas). 2024 Sep 26;60(10):1581. doi: 10.3390/medicina60101581

**Therapeutic potential of the latest oxygen affinity-modifying agent, GBT021601, for treating sickle cell disease is questionable**
Alaimo B, Metaferia B, Cellmer T, Thein SL, Bunn HF, Eaton WA.
Br J Haematol. 2024 Dec;205(6):2527-2529. doi: 10.1111/bjh.19697

**Safety and efficacy of mitapivat in sickle cell disease (RISE UP): results from the phase 2 portion of a global, double-blind, randomised, placebo-controlled trial**
Idowu M, Otieno L, Dumitriu B, Lobo CLC, Thein SL, *et al.*
Lancet Haematol. 2025 Jan;12(1):e35-e44. doi: 10.1016/S2352-3026(24)00319-3

**Newer Modalities and Updates in the Management of Sickle Cell Disease: A Systematic Review**
Patel ZV, Prajjwal P, Bethineedi LD, Patel DJ, Khullar K, *et al.*
J Blood Med. 2024 Sep 12;15:435-447. doi: 10.2147/JBM.S477507

**Real-World Evidence of Crizanlizumab Showing Reductions in Vaso-Occlusive Crises and Opioid Usage in Sickle Cell Disease**
DeBonnett L, Joshi V, Silva-Pinto AC, Colombatti R, Pasanisi A, *et al.*
Eur J Haematol. 2025 Feb;114(2):293-302. doi: 10.1111/ejh.14323

**Pharmacokinetics, pharmacodynamics, safety, and efficacy of crizanlizumab in patients with sickle cell disease: final results from the phase II SOLACE-adults study**
Kanter J, Mennito S, Nair SM, Manwani D, Kutlar A, *et al.*
Ther Adv Hematol. 2024 Nov 3;15:20406207241292508. doi: 10.1177/20406207241292508

**Memantine treatment in sickle cell disease: A 1-year study of its effects on cognitive functions and neural processing**
Raz S, Koren A, Bogdanova AY, Gassmann M, Levin C.
Br J Haematol. 2024 Nov 5. doi: 10.1111/bjh.19866

**Low-dose iron chelation as anti-oxidative therapy in patients with sickle cell disease: A single-centre pilot study**
Gaartman AE, Beuger BM, de Ligt LA, Veldthuis M, Matlung HL, *et al.*
Br J Haematol. 2024 Dec 6. doi: 10.1111/bjh.19943

**Globalization in clinical drug development for sickle cell disease**
Costa E, Ware RE, Tshilolo L, Makani J, Leufkens HGM, Luzzatto L.
Am J Hematol. 2025 Jan;100(1):4-9. doi: 10.1002/ajh.27525

**Enhanced vasoconstriction in sickle cell disease is dependent on ETA receptor activation**
Allan JM, Fox BM, Kasztan M, Kelly GC, Molina PA, *et al.*
Clin Sci (Lond). 2024 Dec 4;138(23):1505-1520. doi: 10.1042/CS20240625

**Inhibition of sodium-glucose cotransporter-2 improves anaemia in mice and humans with sickle cell disease, and reduces infarct size in a murine stroke model**
Wang J, Silaghi P, Guo C, Harro D, Eitzman DT.
J Cell Mol Med. 2024 Sep;28(17):e70091. doi: 10.1111/jcmm.70091

**Targeting P-selectin and interleukin-1β in mice with sickle cell disease: effects on vaso-occlusion, liver injury and organ iron deposition**
Gotardo ÉMF, Torres LS, Zaidan BC, Gushiken LFS, Brito PL, *et al.*
Haematologica. 2024 Nov 21. doi: 10.3324/haematol.2024.286418

**Transfusions, disease-modifying treatments, and curative therapies for sickle cell anemia in Africa: where are we now?**
Odame I, Bazuaye GN.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):234-239. doi: 10.1182/hematology.2024000550

**Quality of life in people with sickle cell disease treated with automated red blood cell exchange**
Dierick K, Rodriguez-Grande B, Navarro-Aragall AG, Beraud M.
Vox Sang. 2025 Jan;120(1):4-12. doi: 10.1111/vox.13757

**Post-exchange neutrophil count, but not post-hematocrit, predicts endogenous erythropoiesis in patients with sickle cell disease undergoing chronic red cell exchange**
Yurtsever N, Tong N, Geetha S, Nandi V, Shi PA.
Transfusion. 2024 Dec;64(12):2270-2278. doi: 10.1111/trf.18044

**Impact of Red Cell Exchange Transfusion on Inflammatory Markers in Sickle Cell Disease**
Costa A, Mendes IF, Lage J, Moniz M, Amorim C, *et al.*
Mediterr J Hematol Infect Dis. 2025 Jan 1;17(1):e2025011. doi: 10.4084/MJHID.2025.011

**Adding hydroxyurea to chronic transfusion therapy for sickle cell anemia reduces transfusion burden**
Nickel RS, Margulies S, Panchapakesan K, Chorvinsky E, Nino G, *et al.*
Transfusion. 2025 Jan;65(1):38-49. doi: 10.1111/trf.18073

**How do we manage hyperhemolysis syndrome**
Adkins BD, Kaylani SZ, Nero A, Ibrahim IF, Shen YM, Sarode R.
Transfusion. 2024 Oct;64(10):1822-1829. doi: 10.1111/trf.17995

**The American Rare Donor Program: 25 years supporting rare blood needs**
Keller MA, Nance ST, Maurer J, Kavitsky V, Babariya SP.
Immunohematology. 2024 Oct 4;40(3):100-121. doi: 10.2478/immunohematology-2024-015

**Late effects of hemopoietic stem cell transplant for sickle cell disease: monitoring and management**
Goldenberg M, Lanzkron S, Pecker LH.
Expert Rev Hematol. 2024 Dec;17(12):891-905. doi: 10.1080/17474086.2024.2423368

**Hematopoietic Cell Transplant compared with Standard Care in Adolescents and Young Adults with Sickle Cell Disease**
Walters MC, Eapen M, Liu Y, El Rassi F, Waller EK, *et al.*
Blood Adv. 2024 Oct 29:bloodadvances.2024013926. doi: 10.1182/bloodadvances.2024013926

**Incremental eligibility criteria for the BMT CTN 1507 haploidentical trial for children with sickle cell disease**
John TD, Walters MC, Rangarajan HG, Rahim MQ, McKinney C, *et al.*
Blood Adv. 2024 Dec 10;8(23):6055-6063. doi: 10.1182/bloodadvances.2024014078

**A machine learning-based workflow for predicting transplant outcomes in patients with sickle cell disease**
Li H, Sachdev V, Tian X, Nguyen ML, Hsieh M, *et al.*
Br J Haematol. 2024 Oct 22. doi: 10.1111/bjh.19842

**Systemic Inflammatory Response Post Alemtuzumab and Low-Dose Total Body Irradiation in Pediatric Patients With Sickle Cell Disease: A Case Series**
Chok R, Girgulis K, Nickel RS, Guilcher GMT.
Pediatr Blood Cancer. 2025 Feb;72(2):e31448. doi: 10.1002/pbc.31448

**Assessment of Biventricular Systolic and Diastolic Function Using Conventional and Strain Echocardiography in Children with Sickle Cell Disease Surviving 1-year After Hematopoietic Stem Cell Transplant**
Harrington JK, DiLorenzo MP, Bhatia M, Boscamp N, Krishnan US.
Pediatr Cardiol. 2024 Oct 4. doi: 10.1007/s00246-024-03646-y

**High-risk allogeneic stem cells transplantation in advanced age patients with uncontrolled sickle cell disease**
Garban F, Makowski C, Bulabois CE, Bouillet L, Meunier M.
Eur J Intern Med. 2024 Dec 8:S0953-6205(24)00481-3. doi: 10.1016/j.ejim.2024.11.017

**Effect of ABO Mismatch and Red Blood Cell Alloimmunization on the Outcome of Hematopoietic Cell Transplantation for Sickle Cell Disease**
Saib I, Alahmari B, Alsadi H, Alaskar A, Hejazi A, *et al.*
Transplant Cell Ther. 2025 Jan;31(1):30.e1-30.e8. doi: 10.1016/j.jtct.2024.11.003

**Disparity in HLA typing rates among hospitalized pediatric patients with sickle cell disease**
Ayuk-Arrey AT, Martin OY, Darbari I, Abraham A, Nickel RS.
Transplant Cell Ther. 2023 Oct 28:S2666-6367(23)01641-X. doi: 10.1016/j.jtct.2023.10.020

**"Being alone for a whole year [is hard]" Families reflect on the emotional toll of stem cell transplantation for sickle cell disease**
Steinberg DM, Mulinda C, Castaño K, DiCola K, Tanenbaum ML, *et al.*
J Pediatr Psychol. 2024 Nov 28:jsae101. doi: 10.1093/jpepsy/jsae101

**Is Allogeneic Transplantation for Sickle Cell Disease Still Relevant in the Era of Gene Therapy?**
Jones RJ, Kassim AA, Brodsky RA, DeBaun MR.
Blood Adv. 2024 Nov 27:bloodadvances.2024013693. doi: 10.1182/bloodadvances.2024013693

**Treating Sickle Cell Disease: Gene Therapy Approaches**
Cavazzana M, Corsia A, Brusson M, Miccio A, Semeraro M.
Annu Rev Pharmacol Toxicol. 2025 Jan;65(1):397-413. doi: 10.1146/annurev-pharmtox-022124-022000

**Gene therapy for sickle cell disease: recent advances, clinical trials and future directions**
Ballantine J, Tisdale JF.
Cytotherapy. 2024 Dec 26:S1465-3249(24)00925-3. doi: 10.1016/j.jcyt.2024.11.006

**CRISPR-based gene therapy for the induction of fetal hemoglobin in sickle cell disease**
McManus M, Frangoul H, Steinberg MH.
Expert Rev Hematol. 2024 Dec;17(12):957-966. doi: 10.1080/17474086.2024.2429605

**How I treat sickle cell disease with gene therapy**
Sharma A.
Blood. 2024 Dec 26;144(26):2693-2705. doi: 10.1182/blood.2024024519

**Zinc finger nuclease-mediated gene editing in hematopoietic stem cells results in reactivation of fetal hemoglobin in sickle cell disease**
Lessard S, Rimmelé P, Ling H, Moran K, Vieira B, *et al.*
Sci Rep. 2024 Oct 16;14(1):24298. doi: 10.1038/s41598-024-74716-7

**Cyclosporin H Improves the Transduction of CD34$^{+}$ Cells with an Anti-Sickling Globin Vector, a Possible Therapeutic Approach for Sickle Cell Disease**
Mormin M, Rigonnot L, Chalumeau A, Miccio A, Fournier C, *et al.*
Hum Gene Ther. 2024 Nov;35(21-22):896-903. doi: 10.1089/hum.2024.098

**Introducing a hemoglobin G-Makassar variant in HSCs by in vivo base editing treats sickle cell disease in mice**
Li C, Georgakopoulou A, Paschoudi K, Anderson AK, Huang L, *et al.*
Mol Ther. 2024 Dec 4;32(12):4353-4371. doi: 10.1016/j.ymthe.2024.10.018

**Financial Impacts of Paying for Gene Therapy for Sickle Cell Disease Under Alternative Pricing and Financing Mechanisms**
Basu A.
Value Health. 2024 Nov 10:S1098-3015(24)06756-1. doi: 10.1016/j.jval.2024.10.3848

**Managing the challenges of paying for gene therapy: strategies for market action and policy reform in the United States**
Phares S, Trusheim M, Emond SK, Pearson SD.
J Comp Eff Res. 2024 Dec;13(12):e240118. doi: 10.57264/cer-2024-0118

**The CMS Sickle Cell Gene Therapy Access Model**
Ballreich J, Odouard IC, Socal MP.
JAMA Pediatr. 2024 Dec 16. doi: 10.1001/jamapediatrics.2024.5536

**Prediction of obstetric outcomes in sickle cell patients based on tricuspid regurgitant velocity**
Lacasse A, Williams V, Ganguli P, Grand'Maison S, Wo B, *et al.*
Obstet Med. 2024 Jul 25:1753495X241263135. doi: 10.1177/1753495X241263135

**Patient-reported pregnancy loss and maternal complications: Insights from the sickle cell disease implementation consortium**
Mathias JG, Masese RV, King AA, Bulgin D, Stevenson E, *et al.*
Int J Gynaecol Obstet. 2024 Oct 25. doi: 10.1002/ijgo.15974

**A Cross-Sectional Study Comparing Placental Characteristics in Pregnancy-Induced Hypertension and Sickle Cell Anaemia**
Bhanarkar U, Potdar P.
Cureus. 2024 Sep 23;16(9):e70034. doi: 10.7759/cureus.70034

**Trends in Pregnancy Outcomes in People with Sickle Cell Disease and Medicaid Insurance (2006-2018)**
O'Brien SH, Stanek JR, House A, Cronin RM, Creary SE, *et al.*
J Womens Health (Larchmt). 2025 Jan;34(1):21-26. doi: 10.1089/jwh.2023.1109

**Impact of scheduled partial exchange transfusions on outcomes in pregnant patients with severe sickle cell disease: a retrospective study**
Ananthaneni A, Jones S, Ghoweba M, Grant V, Leethy K, *et al.*
Hematol Transfus Cell Ther. 2024 Nov;46 Suppl 5(Suppl 5):S109-S114. doi: 10.1016/j.htct.2024.07.001

**The role of prophylactic transfusion on the maternal and fetal outcomes in pregnant women with sickle cell disease: A systematic review and meta-analysis**
AlMoshary M, Arabdin M.
Medicine (Baltimore). 2024 Sep 6;103(36):e39475. doi: 10.1097/MD.0000000000039475

**Impact of Etonogestrel-releasing contraceptive implant use in cisgender women with Sickle cell disease**
Brito MB, Boas AQV, Silva AMN, Miranda FP, Matos CBB, *et al.*
J Natl Med Assoc. 2024 Oct;116(5):569-575. doi: 10.1016/j.jnma.2024.09.003

**Functional ovarian reserve in women with Sickle Cell disease: A systematic review**
Silva CS, Passos NM, Ribeiro APMR, Mattos RR, Torquato E, *et al.*
JBRA Assist Reprod. 2024 Dec 2;28(4):683-690. doi: 10.5935/1518-0557.20240067

**Prognostic significance of early acute splenic sequestration in children with severe sickle cell genotypes: A comprehensive longitudinal neonatal cohort study**
Soulié A, Arnaud C, Pissard S, Hau I, Shum M, *et al.*
Am J Hematol. 2025 Jan;100(1):176-179. doi: 10.1002/ajh.27517

**Splenic filtration of red blood cells in physiology, malaria and sickle cell disease**
Sissoko A, Othmene YB, Buffet P.
Curr Opin Hematol. 2024 Nov 1;31(6):307-314. doi: 10.1097/MOH.0000000000000839

**Partial Splenectomy Versus Total Splenectomy in Sickle Cell Disease: A Systematic Review and Meta-Analysis**
Roshdy MR, Botros M, Mokhles A, Aldemerdash MA, Sabet H, *et al.*
J Pediatr Surg. 2025 Feb;60(2):162058. doi: 10.1016/j.jpedsurg.2024.162058

**Length of Stay, Readmission Rates, and Mortality Are Similar Between Hospitalized Individuals With Sickle Cell Disease With and Without COVID-19**
Clarke KA, Moussa M, Hall MAK, Shin YM.
Cureus. 2024 Sep 30;16(9):e70567. doi: 10.7759/cureus.70567

**Children with sickle cell disease: are they protected from serious COVID-19?**
Shahin WA, Aldeeb H, Alsulami M, Tammas A, Albatniji F, *et al.*
Front Pediatr. 2024 Oct 7;12:1337377. doi: 10.3389/fped.2024.1337377

**Clinical effects of dexamethasone among patients with sickle cell disease hospitalized with COVID-19: Outcomes from a single academic health system**
Garneau WM, Lankiewicz MJ, Lesko CR, Lauriello AP, Gebo KA, Lanzkron SM.
PLoS One. 2024 Nov 26;19(11):e0313289. doi: 10.1371/journal.pone.0313289

**Infections in sickle cell disease**
Scourfield LEA, Nardo-Marino A, Williams TN, Rees DC.
Haematologica. 2024 Nov 21. doi: 10.3324/haematol.2024.285066

**Prevention of Invasive Pneumococcal Disease Among Black or African American Children With and Without Sickle Cell Disease in the United States After Introduction of 13-Valent Pneumococcal Conjugate Vaccine, 2005 Through 2019**
Payne AB, Gierke R, Xing W, McGee L, Hulihan M, *et al.*; Active Bacterial Core Surveillance Team.
Pediatr Blood Cancer. 2025 Mar;72(3):e31467. doi: 10.1002/pbc.31467

**Immune Cell Profiles of Patients with Sickle Cell Disease during Parvovirus B19-Induced Transient Red Cell Aplasia**
Allen EK, Penkert RR, Hankins JS, Surman SL, Van de Velde LA, *et al.*
Vaccines (Basel). 2024 Aug 29;12(9):984. doi: 10.3390/vaccines12090984

**Weekly dihydroartemisinin-piperaquine versus monthly sulfadoxine-pyrimethamine for malaria chemoprevention in children with sickle cell anaemia in Uganda and Malawi (CHEMCHA): a randomised, double-blind, placebo-controlled trial**
Idro R, Nkosi-Gondwe T, Opoka R, Ssenkusu JM, Dennis K, *et al.*
Lancet Infect Dis. 2024 Dec 20:S1473-3099(24)00737-0. doi: 10.1016/S1473-3099(24)00737-0

**Should we use bisphosphonates to treat bone complications in sickle cell disease?**
Gollamudi J, Adesina O.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):623-626. doi: 10.1182/hematology.2024000671

**Subtrochanteric shortening osteotomy in adult sickle cell disease patients with cemented total hip arthroplasty for hip deformities secondary to childhood osteonecrosis: is healing a challenge?**
Hernigou P, Homma Y, Bastard C, Yoon BC, Lachaniette CF.
Int Orthop. 2025 Feb;49(2):407-419. doi: 10.1007/s00264-024-06394-z

**Postoperative complications and cost implications in sickle cell disease patients undergoing total hip arthroplasty: A national inpatient sample study**
Miskiewicz M, Madera R, Pesselev I, Gallagher J, Komatsu D, Nicholson J.
J Orthop. 2024 Nov 23;64:68-72. doi: 10.1016/j.jor.2024.11.016

**Vitamin D Deficiency and Avascular Necrosis in Patients With Sickle Cell Disease: A Retrospective Cohort Study**
Tammas AJ, Albehlal LB, Alabbas F.
Cureus. 2024 Oct 24;16(10):e72269. doi: 10.7759/cureus.72269

**Long-term outcomes of avascular necrosis in sickle cell disease using joint-specific patient-reported outcome measures: Results from a multicentre study**
Casale M, Toro G, Porcelli F, Quota A, Rosso R, *et al.*
Br J Haematol. 2025 Jan;206(1):310-319. doi: 10.1111/bjh.19802

**Alendronate preserves bone mineral density in adults with sickle cell disease and osteoporosis**
Adesina OO, Jenkins IC, Galvão F, de Moura AC, Fertrin KY, *et al.*
Osteoporos Int. 2025 Jan;36(1):93-102. doi: 10.1007/s00198-024-07268-1

**Vestibular Dysfunction in Patients With Sickle Cell Disease: A Systematic Review**
Laredo J, Torres-Small S, Wu L, Makishima T, Richard C.
Otol Neurotol. 2024 Dec 1;45(10):1098-1107. doi: 10.1097/MAO.0000000000004327

**A contemporary review of the management strategies for sickle cell disease related ischaemic and stuttering priapism**
Emmanuel A, Moussa A, Kesse-Adu R, Shabbir M.
Int J Impot Res. 2024 Dec 21. doi: 10.1038/s41443-024-01008-z

**The endocannabinoid system's genetic polymorphisms in sickle cell anemia patients**
Berti ACM, de Castro VDSR, Arcanjo GS, da Silva Araujo A, Lucena-Araujo AR, *et al.*
Sci Rep. 2024 Dec 30;14(1):31562. doi: 10.1038/s41598-024-76480-0

**Sympathetic hypoactivity leads to hypocontractility of the corpus cavernosum in sickle cell mice: a mechanism contributing to priapism**
Silveira THR, Pereira DA, Calmasini FB, Costa FF, Burnett AL, Silva FH.
Int J Impot Res. 2024 Nov 26. doi: 10.1038/s41443-024-00997-1

**Conservative management of segmental testicular infarction in a patient with sickle cell anemia: A case report**
Wu SY, Gibbs EM, Klose C, Qosja N, Zganjar A, Baird BA.
Urol Case Rep. 2024 Jun 21;56:102775. doi: 10.1016/j.eucr.2024.102775

**L'hématopoïèse extramédullaire, une complication rare de la drépanocytose : à propos de six cas et revue de la littérature**
Boccadifuoco U, Cheminet G, Morino B, Arlet JB.
Rev Med Interne. 2025 Jan 7:S0248-8663(24)01344-4. doi: 10.1016/j.revmed.2024.12.006

**Systemic lupus erythematosus-associated autoantibodies in sickle cell disease: Spontaneous emergence in a patient and in transgenic sickle mice**
Sinha U, Setty S, Pilon C, Brown JJ, Ruiz MA, *et al.*
Br J Haematol. 2025 Jan 2. doi: 10.1111/bjh.19972

**Black Americans With Sickle Cell Disease (SCD) Demonstrate Accelerated Epigenetic Pace of Aging Compared to Black Americans Without SCD**
Garrett ME, Le B, Bourassa KJ, Dennis MF, Hatch D, *et al.*; VA Mid-Atlantic MIRECC Workgroup.
J Gerontol A Biol Sci Med Sci. 2024 Nov 1;79(11):glae230. doi: 10.1093/gerona/glae230

**Adolescents and young adults with sickle cell disease exhibit accelerated aging with elevated T-cell p16$^{INK4a}$ expression**
Wilson SR, Mitin N, Miller VLA, Smitherman AB, Carden MA.
Aging (Albany NY). 2024 Nov 14;16(21):13225-13236. doi: 10.18632/aging.206152

**Novel clinical care models for patients with sickle cell disease**
Bartolucci P.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):618-622. doi: 10.1182/hematology.2024000586

**Evolving Landscape of Sickle Cell Anemia Management in Africa: A Critical Review**
Musuka HW, Iradukunda PG, Mano O, Saramba E, Gashema P, *et al.*
Trop Med Infect Dis. 2024 Nov 29;9(12):292. doi: 10.3390/tropicalmed9120292

**Fostering a healthier generation of children with sickle cell disease through advancements in care**
**Franco E, Nimura C, McGann PT.**
Pediatr Res. 2024 Sep 13. doi: 10.1038/s41390-024-03566-w

**Standardized Management of Sickle Cell Disease Patients and the Effects on Care Utilization and Costs**
Ross DH, Wozniak AW, Markossian T, Kellberg G, Gazi SK, Smith K.
Am J Med Qual. 2024 Sep-Oct 01;39(5):201-208. doi: 10.1097/JMQ.0000000000000199

**Exploring machine learning algorithms in sickle cell disease patient data: A systematic review**
Machado TF, Neto FDCB, Gonçalves MS, Barbosa CG, Barreto ME.
PLoS One. 2024 Nov 11;19(11):e0313315. doi: 10.1371/journal.pone.0313315

**End-of-life care for people with sickle cell disease: barriers to and facilitators of high-quality care**
Patel RV, Johnston EE.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):355-362. doi: 10.1182/hematology.2024000561

**Improving Sickle Cell Patient Appointment Compliance in a Hospital-based Pediatric Ophthalmology Clinic**
Louie E, Tang A, Patel A, Srinivasan R, Bohn S, Wilson M.
J Pediatr Hematol Oncol. 2025 Jan 1;47(1):e44-e47. doi: 10.1097/MPH.0000000000002970

**Results of a healthcare transition learning collaborative for emerging adults with sickle cell disease: the ST3P-UP study transition quality improvement collaborative**
Osunkwo I, Cornette JS, Noonan L, Courtlandt C, Mabus S, *et al.*
BMJ Qual Saf. 2024 Nov 21:bmjqs-2024-017725. doi: 10.1136/bmjqs-2024-017725

**Developing a transition workshop for adolescents with sickle cell disease**
Bradley J, Lee Z, Cheong M.
Health Care Transit. 2024 Jan 12;2:100040. doi: 10.1016/j.hctj.2023.100040

**Initial outcomes in a sickle cell disease transition clinic**
Madrazo-Aguirre K, Ribeil JA, Sobota AE.
Health Care Transit. 2024 Mar 15;2:100056. doi: 10.1016/j.hctj.2024.100056

**Development of the iManage SCD mobile health application for transition**
Steinway C, Shilly S, Belton T, Smith-Whitley K, Jan S, *et al.*
Health Care Transit. 2024 Oct 28;2:100074. doi: 10.1016/j.hctj.2024.100074

**Training young adults as community health workers specializing in pediatric to adult health care transition to support emerging adults with sickle cell disease**
Belton TD, Wu K, Steinway CM, Trachtenberg SW, Tchume-Johnson T, *et al.*
Health Care Transit. 2024 Feb 19;2:100050. doi: 10.1016/j.hctj.2024.100050

**Characterizing medical decision-making in sickle cell disease during childhood: Qualitative perspectives of caregivers**
Blakey AO, Amaro CM, Eilenberg JS, Brochier A, Kavanagh PL, *et al.*
Pediatr Blood Cancer. 2024 Dec;71(12):e31307. doi: 10.1002/pbc.31307

**"I was very scared when I found out my son had sickle cell": Caregiver knowledge and attitudes toward early intervention for young children with sickle cell disease: Implications for policy and practice from a multi-site study**
Hoyt CR, MacArthur E, Leaver H, Wilson AL, Davis K, *et al.*
Pediatr Blood Cancer. 2024 Nov;71(11):e31308. doi: 10.1002/pbc.31308

**Improving the Lives of People with Sickle Cell Disease: Community Organizations and Epidemiologists Working Together**
Young AJ, Maresh A, Pope S, Blaylark R, Lakshmanan S, *et al.*
Prog Community Health Partnersh. 2024;18(3):371-380

**Clinical burden and healthcare resource utilization associated with managing sickle cell disease with recurrent vaso-occlusive crises in France**
Baldwin J, Udeze C, Li N, Boulmerka L, Dahal L, *et al.*
Curr Med Res Opin. 2024 Dec;40(12):2117-2124. doi: 10.1080/03007995.2024.2421287

**Economic burden and quality of life of caregivers of patients with sickle cell disease in the United Kingdom and France: a cross-sectional study**
Besser M, O'Sullivan SB, Bourke S, Longworth L, Barcelos GT, Oluboyede Y.
J Patient Rep Outcomes. 2024 Sep 26;8(1):110. doi: 10.1186/s41687-024-00784-y

**Qualitative Assessment of Health-Related Quality of Life Impacts Associated with Sickle Cell Disease in the United States and United Kingdom**
Drahos J, Boateng-Kuffour A, Calvert M, Valentine A, Mason A, *et al.*
Adv Ther. 2024 Dec 16. doi: 10.1007/s12325-024-03038-x

**Health-related quality of life of children and adolescents with sickle cell disease: An evolutionary concept analysis**
Pandarakutty S, Arulappan J.
Appl Nurs Res. 2024 Dec;80:151862. doi: 10.1016/j.apnr.2024.151862

**Social Vulnerability and Sickle Cell Disease Mortality in the US**
Tan JY, San BJ, Yeo YH, Chan KH, Shaaban HS, *et al.*
JAMA Netw Open. 2024 Sep 3;7(9):e2440599. doi: 10.1001/jamanetworkopen.2024.40599

**Social determinants of health affect disease severity among preschool children with sickle cell disease**
Khan H, Kang G, Porter JS, Ding J, Wang WC, *et al.*
Blood Adv. 2024 Dec 10;8(23):6088-6096. doi: 10.1182/bloodadvances.2023012379

**A Scoping Review of Social Determinants of Health and Pain Outcomes in Sickle Cell Disease**
Darby JE, Akpotu IC, Wi D, Ahmed S, Doorenbos AZ, Lofton S.
Pain Manag Nurs. 2024 Oct 5:S1524-9042(24)00258-3. doi: 10.1016/j.pmn.2024.09.002

**Pain-Related Injustice Appraisals, Sickle Cell Stigma, and Racialized Discrimination in the Youth with Sickle Cell Disease: A Preliminary Investigation**
Miller MM, Kissi A, Rumble DD, Hirsh AT, Vervoort T, *et al.*
J Racial Ethn Health Disparities. 2024 Dec 9. doi: 10.1007/s40615-024-02247-y

**A Winding Road to Health Care Equity in Sickle Cell Disease**
Walia R, Fertrin KY, Sabath DE.
Clin Lab Med. 2024 Dec;44(4):693-704. doi: 10.1016/j.cll.2024.07.005

**Impact of Neighborhood Disadvantage on Preventive and Acute Care Utilization in Sickle Cell Disease**
Catanzarite A, Bouck JR, Matthes M, Goubeaux DL, Carter A, *et al.*
Pediatr Blood Cancer. 2025 Jan;72(1):e31422. doi: 10.1002/pbc.31422

**Association of Unmet Social Needs With Disease-Related Outcomes in Pediatric Patients With Sickle Cell Disease**
Sonenklar M, Marks S, Donald C, Valrie C, Smith W, Sisler I.
Pediatr Blood Cancer. 2025 Mar;72(3):e31478. doi: 10.1002/pbc.31478

**Mitochondrial retention in mature red blood cells from patients with sickle cell disease is associated with stress erythropoiesis but not with proinflammatory state**
Romana M, Laurance S, Hardy-Dessources MD, Claer L, *et al.*
Hemasphere. 2024 Oct 28;8(11):e70030. doi: 10.1002/hem3.70030

**Plasma inflammatory and angiogenic protein profiling of patients with sickle cell disease**
de Ligt LA, Gaartman AE, Konté K, Thakoerdin S, Fijnvandraat K, *et al.*
Br J Haematol. 2025 Jan 1. doi: 10.1111/bjh.19970

**Inflammatory pathways and anti-inflammatory therapies in sickle cell disease**
Tozatto-Maio K, Rós FA, Weinlich R, Rocha V.
Hemasphere. 2024 Nov 28;8(12):e70032. doi: 10.1002/hem3.70032

**Targeting ICAM1 to Ameliorate Vaso-Occlusion and Inflammation in Sickle Cell Disease**
Gupta P, Kumar R.
Eur J Haematol. 2024 Dec;113(6):730-737. doi: 10.1111/ejh.14313

**Relationships Between Markers of Iron Status and Hematological Parameters in Patients With Sickle Cell Disease**
Parrow NL, Doherty JM, Conrey A, Thein SL, Fleming RE.
Adv Hematol. 2024 Dec 3;2024:9872440. doi: 10.1155/ah/9872440

**The Calcium Homeostasis of Human Red Blood Cells in Health and Disease: Interactions of PIEZO1, the Plasma Membrane Calcium Pump, and Gardos Channels**
Lew VL.
Annu Rev Physiol. 2024 Oct 30. doi: 10.1146/annurev-physiol-022724-105119

**Transcriptomic atlas reveals organ-specific disease tolerance in sickle cell mice**
Grunenwald A, Peliconi J, Lavergne J, Revel M, Voilin E, *et al.*
Blood Adv. 2025 Jan 22:bloodadvances.2024013435. doi: 10.1182/bloodadvances.2024013435

**Gene-environmental influence of space and microgravity on red blood cells with sickle cell disease**
**Igbineweka NE, van Loon JJWA.**
NPJ Genom Med. 2024 Sep 30;9(1):44. doi: 10.1038/s41525-024-00427-7

**Novel single-cell measurements suggest irreversibly sickled cells are neither dense nor dehydrated**
Reese TJ, Worth EH, Fugate MK, Levitt MT, Ferrone FA.
Biophys J. 2024 Nov 5;123(21):3750-3758. doi: 10.1016/j.bpj.2024.09.024

**IFN-I promotes T-cell-independent immunity and RBC autoantibodies via modulation of B-1 cell subsets in murine SCD**
Su S, Bao W, Liu Y, Shi PA, Manwani D, *et al.*
Blood. 2025 Jan 16;145(3):334-347. doi: 10.1182/blood.2024025175

**Challenges and opportunities for conceiving genetically diverse sickle cell mice**
Agoro R, Churchill GA.
Trends Mol Med. 2024 Dec 5:S1471-4914(24)00305-8. doi: 10.1016/j.molmed.2024.11.004

**Adverse clinical outcomes associated with sickle cell trait at high altitude**
Stafford KA, Osataphan S, Patell R, Key NS.
Haematologica. 2024 Oct 3. doi: 10.3324/haematol.2024.285832

**Sickle Cell Trait, Inequity, and the Need for Change**
Carney BJ, Achebe MO, Haspel RL.
N Engl J Med. 2024 Nov 28;391(21):1972-1974. doi: 10.1056/NEJMp2408196

**Alterations of T Cell Subsets Associated with Sickle Cell Trait**
Reiner AP, Raffield LM, Ekunwe L, Olson NC, Auer PL, Doyle MF.
Blood Genom Discov. 2025;9(1):10001. doi: 10.70322/bgd.2025.10001

**Sickle cell trait in non-firearm arrest-related deaths of Black persons**
Kroll MW, Wolf DA, Hail SL, Zemrus TL, Kunz S, Williams HE.
J Forensic Sci. 2025 Jan;70(1):179-185. doi: 10.1111/1556-4029.15668

## Polyglobulie

**EGLN1 mutations in Cis can induce congenital erythrocytosis with thromboses by increasing protein instability**
Carillo S, Delamare M, Henry L, Maaziz N, Safraou H, *et al.*
Br J Haematol. 2025 Jan 2. doi: 10.1111/bjh.19932

**Identification of EPOR and JAK2 double heterozygous variants in twin cases with familial erythrocytosis**
Li W, Han G, Wang X, Shen K, Xie Y.
Sci China Life Sci. 2024 Dec 11. doi: 10.1007/s11427-024-2786-5

**Utility of next-generation sequencing in identifying congenital erythrocytosis in patients with idiopathic erythrocytosis**
Anžej Doma S, Kraljić N, Kristan A, Debeljak N, Maver A, *et al.*
Front Med (Lausanne). 2024 Sep 6;11:1440712. doi: 10.3389/fmed.2024.1440712

**Real-World Impact of Deep Targeted Sequencing on Erythrocytosis and Thrombocytosis Diagnosis: A Reference Centre Experience**
Blanco-Sánchez A, Gil-Manso R, de Nicolás R, López-Muñoz N, Colmenares R, *et al.*
Cancers (Basel). 2024 Sep 14;16(18):3149. doi: 10.3390/cancers16183149

**Impact of *HFE* mutations on thrombotic risk in patients with idiopathic erythrocytosis: A single-centre study**
Bertozzi I, Benetti A, Cosi E, Zerbinati M, Fortino C, *et al.*
EJHaem. 2024 Oct 4;5(5):1086-1088. doi: 10.1002/jha2.1019

***TFR2* gene alterations in idiopathic erythrocytosis reinforce a possible relation between erythrocytosis and iron metabolism**
Benetti A, Bertozzi I, Randi ML.
Genes Dis. 2024 Apr 5;12(1):101291. doi: 10.1016/j.gendis.2024.101291

**Global Globin Network and adopting genomic variant database requirements for thalassemia**
Halim-Fikri H, Zulkipli NN, Alauddin H, Bento C, Lederer CW, *et al.*
Database (Oxford). 2024 Sep 4;2024:baae080. doi: 10.1093/database/baae080

**SNPscan Combined With CNVplex as a High-Performance Diagnostic Method for Thalassemia**
Wei X, Wang X, Xiong F, Zhang X, Liu D, *et al.*
Prenat Diagn. 2024 Oct;44(11):1310-1317. doi: 10.1002/pd.6661

**Technically feasible solutions to challenges in preimplantation genetic testing for thalassemia: experiences of multiple centers between 2019 and 2022**
Ren Z, Huang P, Wang Y, Yao Y, Ren J, *et al.*
J Assist Reprod Genet. 2024 Nov;41(11):3225-3235. doi: 10.1007/s10815-024-03240-4

**Application of third-generation sequencing technology in the genetic testing of thalassemia**
Li W, Ye Y.
Mol Cytogenet. 2024 Dec 18;17(1):32. doi: 10.1186/s13039-024-00701-4

**Heterozygous Beta-Thalassaemia in Pregnancy: Two Rare Causes of Severe Fetal Anemia Requiring Intrauterine Blood Transfusions**
van der Meij E, Smiers FJW, Koopmann TT, Krapels I, LePoole K, *et al.*
Prenat Diagn. 2024 Nov 2. doi: 10.1002/pd.6695

**β-Thalassemia minor is associated with high rates of worsening anemia in pregnancy**
Langer AL, Goggins BB, Esrick EB, Fell G, Berliner N, Economy KE.
Blood. 2024 Dec 9:blood.2024026736. doi: 10.1182/blood.2024026736

**Reproductive Health in Women with Major β-Thalassemia: Evaluating Ovarian Reserve and Endocrine Complications**
Tsilionis V, Moustakli E, Dafopoulos S, Zikopoulos A, Sotiriou S, *et al.*
Metabolites. 2024 Dec 20;14(12):717. doi: 10.3390/metabo14120717

**Anti-Mullerian Hormone Evaluates Ovarian Function in Patients with Non-Transfusion-Dependent Thalassemia**
Li Y, Zhang X, Yin X, Fu Z, Xie W, *et al.*
Mediterr J Hematol Infect Dis. 2025 Jan 1;17(1):e2025007. doi: 10.4084/MJHID.2025.007

**Trans-acting genetic modifiers of clinical severity in heterozygous β-Thalassemia trait**
Loh JB, Ross JM, Musallam KM, Kuo KHM.
Ann Hematol. 2024 Nov;103(11):4437-4447. doi: 10.1007/s00277-024-06007-0

**Mutations in AMBRA1 aggravate β-thalassemia by impairing autophagy-mediated clearance of free α-globin**
Long Y, Zhang Q, Ling L, Zhuang Y, Wei X, *et al.*
Blood. 2024 Dec 18:blood.2023022688. doi: 10.1182/blood.2023022688

**Alpha Thalassemia Screening in Multiethnic Population in Northern Europe Using Hb Bart's Immunochromatographic Test**
Nik Mohd Hasan NFF, Arkesteijn SJG, Ter Huurne J, Verschuren M, Bhagwandien-Bisoen S, *et al.*
Int J Lab Hematol. 2024 Oct 9. doi: 10.1111/ijlh.14381

**Leveraging Multi-Omics Approaches and Advanced Technologies to Unravel the Molecular Complexities, Modifiers, and Precision Medicine Strategies for Hemoglobin H Disease**
Pahelkar A, Sharma D, Vohra P, Sawant S.
Eur J Haematol. 2024 Dec;113(6):738-744. doi: 10.1111/ejh.14319

**Impact of HbE mutation on the clinical severity of HbH disease: A multicentre study from Thailand**
Songdej D, Teawtrakul N, Laoaroon N, Komvilaisak P, Sripornsawan P, *et al.*; Red Blood Cell Disorders Study Group.
Br J Haematol. 2024 Oct 30. doi: 10.1111/bjh.19869

**Erythropoiesis and Gene Expression Analysis in Erythroid Progenitor Cells Derived from Patients with Hemoglobin H/Constant Spring Disease**
Wongkhammul N, Khamphikham P, Tongjai S, Tantiworawit A, Fanhchaksai K, *et al.*
Int J Mol Sci. 2024 Oct 19;25(20):11246. doi: 10.3390/ijms252011246

**Erythrocytapheresis as a strategy to manage anemia and iron overload in nondeletional hemoglobin H disease**
Zhang K, Bliamptis J, Park J, Kopko P, Sanchez AP, Gopal S.
eJHaem. Published online Jan 21, 2025;6:e21089. doi: 10.1002/jha2.1089

**Efficient and *in situ* correction of hemoglobin Constant Spring mutation by prime editing in human hematopoietic cells**
Shao C, Liu Q, Xu J, Zhang J, Zhang C, *et al.*
Mol Ther Nucleic Acids. 2024 Oct 28;35(4):102371. doi: 10.1016/j.omtn.2024.102371

**Beta Thalassemia in Children: Established Approaches, Old Issues, New Non-Curative Therapies, and Perspectives on Healing**
Origa R, Issa L.
J Clin Med. 2024 Nov 19;13(22):6966. doi: 10.3390/jcm13226966

**Demographics, clinical characteristics, and real-world treatment patterns among patients with beta-thalassemia: a retrospective medical record abstraction study.**
Cappellini MD, Glassberg MB, Meyers J, Jimenez M, Nham T, *et al.*
Ther Adv Hematol. 2024 Dec 15;15:20406207241298088. doi: 10.1177/20406207241298088

**Impaired physical ability in patients with transfusion-dependent β-thalassaemia: Can regular physical activity be a countermeasure?**
Noguer M, Berthon P, Makowski C, Messonnier LA.
Br J Haematol. 2025 Jan;206(1):86-93. doi: 10.1111/bjh.19847

**Cardiovascular magnetic resonance in β-thalassemia major: beyond T2**
Meloni A, Saba L, Cademartiri F, Positano V, Pistoia L, Cau R.
Radiol Med. 2024 Dec;129(12):1812-1822. doi: 10.1007/s11547-024-01916-6

**Early Detection of Myocardial Involvement in Thalassemia Intermedia Patients: Multiparametric Mapping by Magnetic Resonance Imaging**
Meloni A, Pistoia L, Garamella D, Parlato A, Positano V, *et al.*
J Magn Reson Imaging. 2024 Oct 3. doi: 10.1002/jmri.29625

**Speckle-tracking echocardiography as screening tool for myocardial fibrosis and Iron overload in transfusion-dependent beta-thalassemia**
Guaricci AI, Monitillo F, Basile P, Di Gennaro D, Dadamo ML, *et al.*
Int J Cardiol. 2025 Jan 1;418:132616. doi: 10.1016/j.ijcard.2024.132616

**Convolutional neural networks for automatic MR classification of myocardial iron overload in thalassemia major patients**
Positano V, Meloni A, De Santi LA, Pistoia L, Borsellino Z, *et al.*
Eur Radiol. 2024 Dec 10. doi: 10.1007/s00330-024-11245-x

**Early detection of myocardial iron overload in patients with β-thalassemia major using cardiac magnetic resonance T1 mapping**
Selim OMHZ, Ibrahim ASAH, Aly NH, Hegazy SNA, Ebeid FSE.
Magn Reson Imaging. 2024 Dec;114:110250. doi: 10.1016/j.mri.2024.110250

**Thalassemia and iron overload cardiomyopathy: Pathophysiological insights, clinical implications, and management strategies**
Ansharullah BA, Sutanto H, Romadhon PZ.
Curr Probl Cardiol. 2025 Jan;50(1):102911. doi: 10.1016/j.cpcardiol.2024.102911

**Assessing the accuracy of CMRtools software for diagnosing liver iron overload in thalassemia patients: influencing factors and optimisation strategies**
Luo C, Peng F, Xu F, Tang C, Zhang Y, *et al.*
Front Med (Lausanne). 2024 Sep 20;11:1424294. doi: 10.3389/fmed.2024.1424294

**Hypoparathyroidism in adults with iron overload diseases (IOD): evidence of a subclinical phenotype**
De Vincentis S, Evangelisti S, Rossi B, Decaroli MC, Locaso M, *et al.*
Endocrine. 2024 Dec 4. doi: 10.1007/s12020-024-04124-4

**Organ-Specific Iron Overload in Non-Transfusion-Dependent Thalassemia Patients: Insights from Quantitative MRI Evaluation**
Ning X, Tan S, Peng F, Luo C, Tang C, *et al.*
Eur J Radiol. 2024 Dec;181:111750. doi: 10.1016/j.ejrad.2024.111750

**Real-world experience with iron chelation therapy in transfusion-dependent thalassemia: impact of the oral chelators' era**
Pines M, Kleinert D, Thomas C, Mensah C, Musallam KM, Sheth S.
Ann Hematol. 2024 Dec;103(12):5229-5234. doi: 10.1007/s00277-024-06092-1

**Assessing the auditory effects of oral chelation therapy drug Deferasirox in individuals with β-thalassemia major**
Tiwana BS, Aggarwal A, Bhagat S, Singh H, Sahni D, Yadav V.
World J Otorhinolaryngol Head Neck Surg. 2023 Dec 10;10(4):309-314. doi: 10.1002/wjo2.150

**Assessing Disability in Thalassaemia: A Position Statement by the Thalassaemia International Federation**
Farmakis D, Angastiniotis M, Papingiotis G, Cannon L, Eleftheriou A.
Eur J Haematol. 2024 Dec 23. doi: 10.1111/ejh.14367

**Association of age to nutritional status and muscle mass in children with transfusion-dependent β-thalassemia: a cross-sectional study**
Wang L, Zhang L, Yang Y, Luo Y, Wang L, Patil S.
Front Nutr. 2024 Oct 25;11:1493502. doi: 10.3389/fnut.2024.1493502

**Renal Findings in Patients with Thalassemia at Abdominal Ultrasound: Should We Still Talk about "Incidentalomas"? Results of a Long-Term Follow-Up**
Fatigati C, Meloni A, Costantini S, Spasiano A, Ascione F, *et al.*
Diagnostics (Basel). 2024 Sep 15;14(18):2047. doi: 10.3390/diagnostics14182047

**Adrenal Insufficiency in Patients with Beta Thalassemia: A Meta-Analysis**
Savvidis C, Ragia D, Delicou S, Xydaki A, Rizzo M, Ilias I.
Medicina (Kaunas). 2024 Sep 25;60(10):1571. doi: 10.3390/medicina60101571

**A Multicenter ICET-A Survey on Adherence to Annual Oral Glucose Tolerance Test (OGTT) Screening in Transfusion-Dependent Thalassemia (TDT) Patients -The Expert Clinicians' Opinion on Factors Influencing the Adherence and on Alternative Strategies for Adherence Optimization**
de Sanctis V, Canatan D, Daar S, Kattamis C, Banchev A, *et al.*
Mediterr J Hematol Infect Dis. 2025 Jan 1;17(1):e2025008. doi: 10.4084/MJHID.2025.008

**Serum lipid profile abnormalities among beta-thalassemia patients: a systematic review and meta-analysis**
Mohamed SOO, Mohamed AEA, Salih MSK, Salih KSK, Abdelrahman ASEE, *et al.*
Lipids Health Dis. 2024 Nov 25;23(1):388. doi: 10.1186/s12944-024-02377-6

**A Comparison of Pain Before and After Transfusion in Adult transfusion-dependent thalassemia (TDT) Using BPI-SF**
Gamayani U, Dianathasari Santoso S, Nugraha Hermawan A, Irani Fianza P, Panigoro R.
F1000Res. 2024 Aug 9;12:1425. doi: 10.12688/f1000research.75952.2

**Whole-genome Sequencing Association Analysis of Quantitative Platelet Traits in A Large Cohort of β-thalassemia**
Wang X, Zhang Q, Chen X, Huang Y, Zhang W, *et al.*
Genomics Proteomics Bioinformatics. 2024 Sep 27:qzae065. doi: 10.1093/gpbjnl/qzae065

**Serum ferritin is a risk factor for pulmonary dysfunction in young patients with transfusion-dependent thalassemia**
Liang X, Yang G, Shi L, Liu L, Wei Z, *et al.*
Heliyon. 2024 Sep 19;10(19):e38069. doi: 10.1016/j.heliyon.2024.e38069

**Luspatercept: a treatment for ineffective erythropoiesis in thalassemia**
Musallam KM, Taher AT.
Hematology Am Soc Hematol Educ Program. 2024 Dec 6;2024(1):419-425. doi: 10.1182/hematology.2024000567

**Luspatercept in β-thalassemia: Who and when. Strengths and weaknesses points of a real-world evidence**
Torti L, Ardu N, Maffei L, De Fabritiis P, Sorrentino F.
EJHaem. 2024 Nov 4;5(6):1354-1358. doi: 10.1002/jha2.1032

**Identifying thresholds for meaningful improvements in NTDT-PRO scores to support conclusions about treatment benefit in clinical studies of patients with non-transfusion-dependent beta-thalassaemia: analysis of pooled data from a phase 2, double-blind, placebo-controlled, randomised trial**
Taher AT, Musallam KM, Viprakasit V, Kattamis A, Lord-Bessen J, *et al.*
BMJ Open. 2024 Nov 14;14(11):e085234. doi: 10.1136/bmjopen-2024-085234

**Comparison of Efficacy and Safety Outcomes of Different Doses Schedules of Thalidomide for Treating Moderate-to-Severe β-Thalassemia Patients**
Yang WJ, Shi LD, Liang Y, Liang LM, Zhang H, *et al.*
Ther Clin Risk Manag. 2024 Nov 29;20:799-809. doi: 10.2147/TCRM.S481128

**Efficacy and safety of thalidomide in patients with β-thalassemia intermedia and major**
Ju W, Din G, Huang J, Zheng M, Wang X, *et al.*
Medicine (Baltimore). 2024 Oct 25;103(43):e40328. doi: 10.1097/MD.0000000000040328

**Evaluating the Role of Red Blood Cell Lifespan in Transfusion-Dependent β-Thalassemia and Impact of Thalidomide Treatment**
Yang K, Gong Y, Xiao J.
Am J Hematol. 2024 Dec 9. doi: 10.1002/ajh.27557

**Antioxidative effects of N-acetylcysteine in patients with β-thalassemia: A quick review on clinical trials**
Ghazaiean M, Aliasgharian A, Karami H, Ghasemi MM, Darvishi-Khezri H.
Health Sci Rep. 2024 Oct 7;7(10):e70096. doi: 10.1002/hsr2.70096

**Haploidentical Hematopoietic Stem Cell Transplantation in Pediatric Transfusion-Dependent Thalassemia: A Systematic Review and Meta-Analysis**
Xiao H, Huang Q, Lai Y, Liu R.
Transplant Cell Ther. 2024 Dec 6:S2666-6367(24)00802-9. doi: 10.1016/j.jtct.2024.12.001

**A novel conditioning regimen with pre-transplantation immunosuppression reduces the complication rates in hematopoietic stem cell transplantation in transfusion-dependent β-thalassemia**
Yang H, Li X, Que L, Chen H, Zhan L, *et al.*
Stem Cell Res Ther. 2024 Dec 21;15(1):495. doi: 10.1186/s13287-024-04103-6

**Early Engraftment and Immune Kinetics Following Allogeneic Transplant Using a Novel Reduced-Toxicity Transplant Strategy in Children/Adolescents with High-Risk Transfusion-Dependent Thalassemia: Early Results of the ThalFAbS Trial**

Raffa EH, Harris TM, Choed-Amphai C, Kirby-Allen M, Odame I, *et al.*
Transplant Cell Ther. 2024 Dec 24:S2666-6367(24)00835-2. doi: 10.1016/j.jtct.2024.12.016

**ATG-Thymoglobulin Versus ATG-Fresenius for Conditioning in Thalassemia Patients Who Underwent Allogenic Stem Cell Transplantation from Matched-Sibling Donor: A Tertiary Cancer Care Center Short-Term Experience**
Singh R, Halder R, Hemant Gupta V, Rainchwar S, Bhatia N, *et al.*
Hemoglobin. 2024 May;48(3):186-191. doi: 10.1080/03630269.2024.2398244

**Uniform Graft-versus-Host Disease Prophylaxis using Post-Transplantation Cyclophosphamide, Methotrexate, and Cyclosporine following Peripheral Blood Hematopoietic Stem Cell Transplantation from Matched and Haploidentical Donors for Transfusion-Dependent Thalassemia: A Retrospective Report from the Bone Marrow Failure Working Group of Hunan Province, China**
Gong S, Tian X, Yang R, Yang L, Wang Z, *et al.*
Transplant Cell Ther. 2024 Dec;30(12):1213.e1-1213.e12. doi: 10.1016/j.jtct.2024.08.022

**Immediate hypersensitivity reactions to parenteral cyclosporine in patients with thalassemia major undergoing hematopoietic stem cell transplantation: a case report and review of the literature**
Mahmoudi PS, Kaveh-Ahangaran R, Rostami T, Hosseini AS, Vaezi M, Shahrami B.
J Med Case Rep. 2024 Nov 11;18(1):544. doi: 10.1186/s13256-024-04910-8

**Using T-lymphocyte subsets at engraftment to predict the risk of acute graft-versus-host disease in patients with thalassemia major: development of a new predictive nomogram**
Xiao H, Yang G, Huang Q, Wei Z, Gan Z, *et al.*
Ther Adv Hematol. 2024 Dec 10;15:20406207241294054. doi: 10.1177/20406207241294054

**Outcomes of Hematopoietic Stem Cell Transplantation in Patients With Thalassemia Major: How Do Anti-HLA Antibodies Impact?: The Impact of Anti-HLA Antibodies on Transplantation Outcomes in Thalassemia Major**
Ersoy GZ, Aksoy BA, Erdem M, Karataş L, Aydoğdu S, *et al.*
Clin Transplant. 2024 Dec;38(12):e70035. doi: 10.1111/ctr.70035

**Drug-drug interaction of phenytoin sodium and methylprednisolone on voriconazole: a population pharmacokinetic model in children with thalassemia undergoing allogeneic hematopoietic stem cell transplantation**
Wu Y, Niu LL, Ling YY, Zhou SR, Huang TM, *et al.*
Eur J Clin Pharmacol. 2024 Dec 23. doi: 10.1007/s00228-024-03795-2

**Risk Factors and Prognostic Markers for Posterior Reversible Encephalopathy Syndrome in Patients With Thalassemia Major After Allogeneic Hematopoietic Stem Cell Transplantation**
Bu M, Deng K, Wu M, Shi L, Qin Y, *et al.*
Am J Hematol. 2024 Dec 19. doi: 10.1002/ajh.27566

**The growth and development of children with β-thalassemia major one year after allogeneic hematopoietic stem cell transplantation**
Luo H, Lin Y, Kuang C.
Transl Pediatr. 2024 Nov 30;13(11):1972-1984. doi: 10.21037/tp-24-202

**Germ Cell Dysfunction is Universal in Male Patients with β-Thalassemia Following Hematopoietic Stem Cell Transplantation During Childhood and Adolescence**
Piriyapokin N, Mahachoklertwattana P, Poomthavorn P, Anurathapan U, Chiangjong W.
J Clin Res Pediatr Endocrinol. 2024 Nov 13. doi: 10.4274/jcrpe.galenos.2024.2024-6-5

**Betibeglogene autotemcel gene therapy in patients with transfusion-dependent, severe genotype β-thalassaemia (HGB-212): a non-randomised, multicentre, single-arm, open-label, single-dose, phase 3 trial**
Kwiatkowski JL, Walters MC, Hongeng S, Yannaki E, Kulozik AE, *et al.*
Lancet. 2024 Nov 30;404(10468):2175-2186. doi: 10.1016/S0140-6736(24)01884-1

**Gene therapy in transfusion-dependent non-β0/β0 genotype β-thalassemia: first real-world experience of beti-cel**
Mirza A, Ritsert ML, Tao G, Thakar H, Lobitz S, *et al.*
Blood Adv. 2025 Jan 14;9(1):29-38. doi: 10.1182/bloodadvances.2024014104

**Reframing thalassaemia syndrome as a benign haematopoietic stem cell disorder**
Maggio A, Napolitano M, Taher AT, Bou-Fakhredin R, Ostuni MA.
Br J Haematol. 2024 Dec 15. doi: 10.1111/bjh.19919

**Clinical Burden and Healthcare Resource Utilization Associated With Managing Transfusion-dependent β-Thalassemia in England**
Udeze C, Ly NF, Ingleby FC, Fleming SD, Conner SC, *et al.*
Clin Ther. 2025 Jan;47(1):37-43. doi: 10.1016/j.clinthera.2024.09.024

**Systematic literature review of the indirect costs and humanistic burden of β-thalassemia**
Aydinok Y, Purushotham S, Yucel A, Glassberg M, Deshpande S, *et al.*
Ther Adv Hematol. 2024 Sep 18;15:20406207241270872. doi: 10.1177/20406207241270872

**The effect of applying emotional intelligence components on coping strategies in adolescents with beta-thalassemia major: a randomized clinical trial**
Ahmadian B, Ramezani M, Badiei Z, Vaghee S.
BMC Pediatr. 2024 Sep 17;24(1):591. doi: 10.1186/s12887-024-05057-7

**The effect of mobile application based genetic counseling on the psychosocial well-being of thalassemia patients and caregivers: A randomized controlled trial**
Setiawan H, Xu W, Wang C, Li C, Ariyanto H, *et al.*
Patient Educ Couns. 2025 Jan;130:108457. doi: 10.1016/j.pec.2024.108457

**The gut microbiota metabolite trimethylamine-N-oxide in children with β-thalassemia: potential implication for iron-induced renal tubular dysfunction**
Ghalwash AA, El-Gohary RM, El Amrousy D, Morad LM, Kassem SS, *et al.*
Pediatr Res. 2024 Oct 25. doi: 10.1038/s41390-024-03639-w

**Exploring alterations of gut/blood microbes in addressing iron overload-induced gut dysbiosis and cognitive impairment in thalassemia patients**
Suparan K, Trirattanapa K, Piriyakhuntorn P, Sriwichaiin S, Thonusin C, *et al.*
Sci Rep. 2024 Oct 23;14(1):24951. doi: 10.1038/s41598-024-76684-4

**Differential gut microbiota composition in β-Thalassemia patients and its correlation with iron overload**
Nonejuie P, Wilantho A, McDonald D, Htoo HH, Chalerm J, *et al.*
Sci Rep. 2024 Oct 11;14(1):23858. doi: 10.1038/s41598-024-75456-4

**Integrated metabolomic and microbiome analysis identifies Cupriavidus metallidurans as a potential therapeutic target for β-thalassemia**
Guo X, Lin S, Zhang X, Li M, Wang Z, *et al.*
Ann Hematol. 2024 Dec;103(12):5169-5179. doi: 10.1007/s00277-024-06016-z

**Genetic modifications of *EGLN1* reactivate HbF production in $\beta^0$-thalassemia/HbE**
Jan-Ngam V, Boontha S, Tubsuwan A, Wongpalee SP, Fanhchaksai K, *et al.*
Heliyon. 2024 Sep 18;10(18):e38020. doi: 10.1016/j.heliyon.2024.e38020

## Hémoglobinopathies – Autres maladies du globule rouge

**Global burden of anemia and cause among children under five years 1990-2019: findings from the global burden of disease study 2019**
Liu Y, Ren W, Wang S, Xiang M, Zhang S, Zhang F.
Front Nutr. 2024 Oct 15;11:1474664. doi: 10.3389/fnut.2024.1474664

**Standardization of hemoglobin $A_2$ and hemoglobin F: Achievements and perspectives**
Mosca A, Arsene C, Paleari R, Kaiser P, Harteveld K, *et al.*; IFCC-ICSH Joint Working Group for the Standardization of HbA2 and of the IFCC Working Group for the Standardization of HbF.
Clin Chim Acta. 2025 Feb 1;567:120087. doi: 10.1016/j.cca.2024.120087

**Prevalence and mortality trends of hemoglobinopathies in Italy: a nationwide study**
Gianesin B, Piel FB, Musallam KM, Barella S, Casale M, *et al.*; Italian Hemoglobinopathies National Survey Group.
Haematologica. 2025 Jan 23. doi: 10.3324/haematol.2024.286886

**Mortality, Clinical Complications, and Healthcare Resource Utilization Associated with Managing Transfusion-Dependent β-Thalassemia and Sickle Cell Disease with Recurrent Vaso-occlusive Crises in Italy**
Udeze C, Dovizio M, Veronesi C, Esposti LD, Li N, *et al.*
Pharmacoecon Open. 2025 Jan;9(1):115-124. doi: 10.1007/s41669-024-00532-4

**Genetic Modifiers of Hemoglobin Expression from a Clinical Perspective in Hemoglobinopathy Patients with Beta Thalassemia and Sickle Cell Disease**
Diamantidis MD, Ikonomou G, Argyrakouli I, Pantelidou D, Delicou S; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 Nov 5;25(22):11886. doi: 10.3390/ijms252211886

**Hypercoagulability in hemoglobinopathies: Decoding the thrombotic threat**
Bou-Fakhredin R, Cappellini MD, Taher AT, De Franceschi L.
Am J Hematol. 2025 Jan;100(1):103-115. doi: 10.1002/ajh.27500

**Livedoid vasculopathy in hemoglobinopathy-associated chronic leg ulcers**
Eswaran H, Wilson S, Little J, Moll S.
Ann Hematol. 2024 Dec;103(12):5235-5239. doi: 10.1007/s00277-024-06112-0

**Cerebral hemodynamics and oxygen metabolism in patients with milder and severe forms of sickle cell disease and thalassemia**
Afzali-Hashemi L, Baas KPA, Schrantee A, Nur E, Vu C, *et al.*
Hemasphere. 2024 Dec 2;8(12):e70022. doi: 10.1002/hem3.70022

**Relationship between hemoglobinopathies and male infertility: a scoping review**
Al-Jubouri AM, Eliwa A, Haithm Y, Al-Qahtani N, Jolo L, Yassin M.
Int J Hematol. 2024 Nov;120(5):566-574. doi: 10.1007/s12185-024-03844-7

**Exploring Novel Strategies to Alleviate Symptoms of β-Globinopathies: Examining the Potential Role of Embryonic ε-globin Induction**
Liu J, Park K, Shen Z, Ye Y, Lee E, *et al.*
Transfus Med Rev. 2024 Oct;38(4):150861. doi: 10.1016/j.tmrv.2024.150861

**Red cell specifications for blood group matching in patients with haemoglobinopathies: An updated systematic review and clinical practice guideline from the International Collaboration for Transfusion Medicine Guidelines**
Wolf J, Blais-Normandin I, Bathla A, Keshavarz H, Chou ST, *et al.*; International Collaboration for Transfusion Medicine Guidelines (ICTMG).
Br J Haematol. 2025 Jan;206(1):94-108. doi: 10.1111/bjh.19837

**Recent advancements in gene therapy for sickle cell disease and β-thalassemia**
Li L, Mandal PK.
Front. Hematol. Published online Sep 27, 2024; 3:1468952. doi: 10.3389/frhem.2024.1468952

**An evaluation of exagamglogene autotemcel for the treatment of sickle cell disease and transfusion-dependent beta-thalassaemia**
Handgretinger R, Mezger M.
Expert Opin Biol Ther. 2024 Sep;24(9):883-888. doi: 10.1080/14712598.2024.2399134.

**Genome Editing Therapy for the Blood: *Ex Vivo* Success and *In Vivo* Prospects**
George CA, Sahu SU, de Oñate L, Souza BSF, Wilson RC.
CRISPR J. 2024 Oct;7(5):231-248. doi: 10.1089/crispr.2024.0036

**The role of the conditioning regimen for autologous and ex vivo genetically modified hematopoietic stem cell-based therapies: recommendations from the ISCT stem cell engineering committee**
Oved JH, Russell A, DeZern A, Prockop SE, Bonfim C, *et al.*
Cytotherapy. 2025 Jan;27(1):78-84. doi: 10.1016/j.jcyt.2024.09.001

**Pediatric Advances in Gene Therapies for Hemoglobinopathies**
Thompson C, Ferdjallah A, Gupta AO.
JAMA Pediatr. 2024 Dec 9. doi: 10.1001/jamapediatrics.2024.5366

**Auto-expansion of *in vivo* HDAd-transduced hematopoietic stem cells by constitutive expression of tHMGA2**
Wang H, Georgakopoulou A, Nizamis E, Mok KW, Eluère R, *et al.*
Mol Ther Methods Clin Dev. 2024 Aug 13;32(3):101319. doi: 10.1016/j.omtm.2024.101319

**Gene editing without ex vivo culture evades genotoxicity in human hematopoietic stem cells**
Zeng J, Nguyen MA, Liu P, da Silva LF, Levesque S, *et al.*
Cell Stem Cell. 2024 Dec 11:S1934-5909(24)00400-4. doi: 10.1016/j.stem.2024.11.001

***BCL11A* +58/+55 enhancer-editing facilitates HSPC engraftment and HbF induction in rhesus macaques conditioned with a CD45 antibody-drug conjugate**
Demirci S, Zeng J, Palchaudhuri R, Wu C, Abraham DM, *et al.*
Cell Stem Cell. 2024 Dec 5:S1934-5909(24)00376-X. doi: 10.1016/j.stem.2024.10.014

**Enhanced fetal hemoglobin production via dual-beneficial mutation editing of the HBG promoter in hematopoietic stem and progenitor cells for β-hemoglobinopathies**
Chandraprabha PB, Azhagiri MKK, Venkatesan V, Magis W, Prasad K, *et al.*
Stem Cell Res Ther. 2024 Dec 31;15(1):504. doi: 10.1186/s13287-024-04117-0

**Vaccination practices and knowledge among adults with hemoglobinopathies in Greece: a nationwide survey**
Delicou S, Manganas K, Xydaki A, Evliati L, Myrilla I, *et al.*
Ther Adv Vaccines Immunother. 2024 Sep 17;12:25151355241278869. doi: 10.1177/25151355241278869

**Activation of γ-globin expression by a common variant disrupting IKAROS-binding motif in β-thalassemia**
Luo H, Wang J, Qin L, Zhang X, Liu H, *et al.*
J Genet Genomics. 2024 Nov 8:S1673-8527(24)00289-3. doi: 10.1016/j.jgg.2024.10.015

**Genetic Polymorphisms Associated with Fetal Hemoglobin (HbF) Levels and F-Cell Numbers: A Systematic Review of Genome-Wide Association Studies**
Stephanou C, Menzel S, Philipsen S, Kountouris P; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 Oct 23;25(21):11408. doi: 10.3390/ijms252111408

**A Variable Clinical Presentation of Hemoglobin City of Hope**
Brik Simon D, Filon D, Meiner V, Krasnov T, Noy-Lotan S, *et al.*
Clin Genet. 2024 Dec 18. doi: 10.1111/cge.14675

**Molecular Characterization of δβ Thalassemia/Hereditary Persistence of Fetal Hemoglobin and Its Correlation With Clinical and Hematological Profile; a Single Center Study in North India**
Gupta R, Shah A, Gupta K, Chandra D, Sharma A, *et al.*
Int J Lab Hematol. 2024 Dec 27. doi: 10.1111/ijlh.14419

**Hb Monza: A novel extensive HBB duplication with preserved α-β subunit interaction and unstable hemoglobin phenotype**
Civettini I, Zappaterra A, Corti P, Messina A, Aroldi A, *et al.*
Med. 2024 Dec 18:100554. doi: 10.1016/j.medj.2024.11.007

**TR4 and BCL11A repress γ-globin transcription via independent mechanisms**
Wang Y, Myers G, Yu L, Deng K, Balbin-Cuesta G, *et al.*
Blood. 2024 Dec 26;144(26):2762-2772. doi: 10.1182/blood.2024024599

**Hemoglobinopathies in the Neonate**
Blankenhorn K, Strumph K.
Neoreviews. 2024 Nov 1;25(11):e720-e728. doi: 10.1542/neo.25-11-e720

**A Multi-Center Retrospective Database Evaluation of Pediatric Subjects Diagnosed With Methemoglobinemia**
Sinha N, Lichak B, Thomas NJ, Krawiec C.
Clin Med Insights Pediatr. 2024 Sep 19;18:11795565241271678. doi: 10.1177/11795565241271678

**Management of Congenital Methemoglobinemia in the Perioperative Setting: A Case Report and Review of Current Literature**
Saad MBGB, Karmakar A, Salih TSM, Arshad W, Khan MJ.
J Blood Med. 2024 Aug 29;15:395-405. doi: 10.2147/JBM.S468072

**Neonatal/perinatal diagnosis of hemolysis using ETCOc**
Christensen RD, Bahr TM, Ohls RK, Moise KJ Jr.
Semin Fetal Neonatal Med. 2024 Oct 22:101547. doi: 10.1016/j.siny.2024.101547

**Elucidating loss-of-function mechanisms of monoallelic EPAS1 mutations underlying congenital hypoplastic anaemia in a paediatric anaemia cohort**
Zhang J, Sun J, Huai W, Tang J, Chen J, *et al.*
Br J Haematol. 2024 Nov 29. doi: 10.1111/bjh.19930

**Marked microcytosis and increased transferrin saturation: Think about variants in SLC11A2 (DMT1)**
Raynor A, Peoc'h K, Boi C, Manceau H, Pissard S, *et al.*
Blood Cells Mol Dis. 2025 Feb;110:102898. doi: 10.1016/j.bcmd.2024.102898

**Whole Blood Transcriptome Analysis in Congenital Anemia Patients**
Sanchez-Villalobos M, Campos Baños E, Martínez-Balsalobre E, Navarro-Ramirez V, Videla MAB, *et al.*
Int J Mol Sci. 2024 Oct 31;25(21):11706. doi: 10.3390/ijms252111706

**Recent advances and clinical applications of red blood cell lifespan measurement**
Ji D, Peng Y, Zhang Y, Tang X, Zhao M, *et al.*
Heliyon. 2024 Aug 22;10(17):e36507. doi: 10.1016/j.heliyon.2024.e36507

**Prenatal Hemoglobin Concentration and Long-Term Child Neurocognitive Development**
Mireku MO, Boivin MJ, Zoumenou R, Garrison A, Cot M, *et al.*
Am J Trop Med Hyg. 2024 Dec 17:tpmd240643. doi: 10.4269/ajtmh.24-0643

**Impact of Vitamin A Deficiency on Iron Metabolism and Anemia: A Historical Perspective and Research Advances**
Mejia LA, Erdman JW Jr.
Nutr Rev. 2024 Dec 12:nuae183. doi: 10.1093/nutrit/nuae183

**Immunoregulation role of the erythroid cells**
Niu C, Zhang J.
Front Immunol. 2024 Oct 15;15:1466669. doi: 10.3389/fimmu.2024.1466669

**Beyond ATP: Metabolite Networks as Regulators of Physiological and Pathological Erythroid Differentiation**
Joly A, Schott A, Phadke I, Gonzalez-Menendez P, Kinet S, Taylor N.
Physiology (Bethesda). 2025 Jan 1;40(1):0. doi: 10.1152/physiol.00035.2024

**Biomechanics of phagocytosis of red blood cells by macrophages in the human spleen**
Li H, Qiang Y, Li X, Brugnara C, Buffet PA, *et al.*
Proc Natl Acad Sci U S A. 2024 Oct 29;121(44):e2414437121. doi: 10.1073/pnas.2414437121

**Red blood cell passage through deformable interendothelial slits in the spleen: Insights into splenic filtration and hemodynamics**
Li G, Li H, Ndour PA, Franco M, Li X, *et al.*
Comput Biol Med. 2024 Nov;182:109198. doi: 10.1016/j.compbiomed.2024.109198

**Development of a novel test of splenic function for use in a clinical diagnostic laboratory**
Khan AA, Laas CE, Brewin JN, Potter V, Cao H, *et al.*
Br J Haematol. 2025 Jan;206(1):320-330. doi: 10.1111/bjh.19950

**10-Year Risk of Gallstones in Congenital Red Blood Cell Disorder Patients: A Nationwide Cohort Study**
Naamansen AB, Hansen DL, Petersen J, Glenthøj A, Sørensen HT, Frederiksen H.
Am J Hematol. 2025 Feb;100(2):229-235. doi: 10.1002/ajh.27558

**Advances in the generation of erythrocytes from stem cells in vitro**
Ling Y, Du H, Zhang Y, Fu Q, He C.
Hematology. 2024 Dec;29(1):2427932. doi: 10.1080/16078454.2024.2427932

**The Roadmap to Integrate Diversity, Equity, and Inclusion in Hematology Clinical Trials: An American Society of Hematology Initiative**
Kuaban A, Croker A, Keefer J, Valentino LA, Bierer BE, *et al.*
Blood Adv. 2024 Dec 9:bloodadvances.2024013945. doi: 10.1182/bloodadvances.2024013945

## Toutes maladies rares

**PFMG2025–integrating genomic medicine into the national healthcare system in France**
PFMG2025 contributors group.
Lancet Reg Health Eur. Available online 6 January 2025, 101183. doi.org/10.1016/j.lanepe.2024.101183

Les numéros précédents du Bulletin Recherche sont disponibles sur la page :

https://filiere-mcgre.fr/le-bulletin-recherche/.3

Filière de santé maladies rares MCGRE - Hôpital Henri Mondor

51 avenue du Maréchal de Lattre de Tassigny, 94000 Créteil

contact@filiere-mcgre.fr - www.filiere-mcgre.fr