



Bulletin recherche

Filière MCGRE

N° 22 - Septembre 2024

MCGRE

FILIERE SANTÉ MALADIES RARES

Table des matières

Le point sur.....	3
Contexte et objectif.....	3
Méthode.....	3
Résultats.....	3
L'étude en quelques chiffres.....	4
Echange avec	5
Appels à projets	7
Bibliographie.....	10
Anémie dysérythropoïétique congénitale	10
Anémie liée au métabolisme du fer et anémie sidéroblastique.....	10
Anomalies de la membrane du globule rouge.....	10
Déficit en glucose-6-phosphate déshydrogénase.....	11
Déficit en pyruvate kinase.....	12
Déficit enzymatique érythrocytaire (autre).....	12
Drépanocytose.....	12
Polyglobulies.....	28
Thalassémies.....	29
Hémoglobinopathies – Autres maladies du globule rouge.....	33

Prédicteurs de la qualité de vie des patients adultes drépanocytaires en France : l'étude Drépatient

Contexte et objectif

La drépanocytose est la maladie génétique la plus fréquente au monde dans laquelle une mutation de l'hémoglobine (molécule indispensable au transport de l'oxygène dans le sang) provoque des complications aiguës et chroniques qui retentissent sur le bien-être émotionnel et physique et donc sur la qualité de vie des patients.

Le SF-36 est un questionnaire permettant d'obtenir une mesure générique de la perception de la santé du patient à travers une auto-évaluation de la qualité de vie par 36 items notés de 0 (mauvaise qualité de vie) à 100 (très bonne qualité de vie). Ce questionnaire évalue la qualité de vie par 8 dimensions : le fonctionnement physique, la perception de la santé générale et mentale, la limitation des rôles en raison de problèmes physiques et/ou émotionnels, la vitalité, les douleurs corporelles et le fonctionnement social.

L'objectif de cette étude était d'estimer la qualité de vie et d'identifier les facteurs associés chez des patients drépanocytaires adultes en France.

Méthode

DREPatient est une étude transversale multicentrique réalisée en France métropolitaine et dans les DOM qui a inclus 1088 patients adultes et enfants (et leurs parents) entre juin 2020 et avril 2021. Les analyses du présent article se sont concentrées sur 570 patients drépanocytaires adultes dont la moyenne d'âge était de 33,3 ans (+/- 10,7). Le SF-36 a été complété en ligne par les patients. Les 8 dimensions du SF-36 ont été regroupées en 2 scores composites principaux : le score résumant la composante physique (PCS) et le score résumant la composante mentale (MCS). Les données sociodémographiques (sexe, âge, lieu de naissance, lieu de vie, avoir des enfants, niveau d'étude, activité professionnelle, auto-estimation de sa situation financière et aide reçue par les proches), cliniques et thérapeutiques des patients (hospitalisation pour crise vaso-occlusive (CVO) ou un syndrome thoracique aigu (STA), admission en soins intensifs, chirurgie, pause de prothèse ou transplantation médullaire, transfusion sanguine lors des 12 derniers mois, traitement contre la drépanocytose, suivi psychologique et complications chroniques ou aiguës) ont été recueillies et analysées en tant que facteurs prédictifs à la qualité de vie. Un modèle de régression linéaire univariée a permis d'identifier les facteurs associés à la qualité de vie ($p < 0.25$) puis un modèle de régression linéaire multivariée corrigé pour l'âge et le sexe a été réalisé pour identifier les facteurs prédictifs des scores de la qualité de vie.

Résultats

Le domaine du fonctionnement physique présente le score le plus élevé (67,5 +/- 21,8) tandis que le domaine de la santé globale a le plus faible (37,7 +/- 20,3). Les scores composites PCS et MCS obtiennent des scores de 40,6 +/- 8,9 et 45,8 +/- 9,8 respectivement.

Les scores composites PCS et MCS sont plus faibles chez les patients avec une situation financière instable, hospitalisés pour une CVO ou STA, admis en soins intensifs, sous anti-douleurs, ayant un suivi psychologique et ceux ayant des complications aiguës ou chroniques. Les scores composites PCS et MCS sont plus élevés chez les patients en activité professionnelle et ceux ayant le soutien de leurs proches.

Les analyses multivariées concernant le score composite de la composante physique ont montré que le score de PCS est plus faible chez les patients sous oxygénothérapie, ayant eu une ostéonécrose fémorale, hospitalisés pour CVO ou STA, avec des complications chroniques, de sexe féminin, avec un suivi psychologique, âgés de plus de 35 ans et recevant des anti-douleurs. Les patients avec un plus haut niveau d'étude et ceux avec une situation financière stable présentent les meilleurs scores de PCS.

Les analyses multivariées concernant le score composite de la composante mentale ont montré que le score de MCS est plus faible chez les patients ayant un suivi psychologique et ceux hospitalisés pour CVO ou STA au cours des 12 mois précédant l'étude. Les patients ayant le soutien de leurs proches et ceux avec une situation financière stable présentent les meilleurs scores de MCS.

Cette étude met en évidence que les patients drépanocytaires ont des scores de qualité de vie physique et mentale plus faibles que la population générale. Les femmes, les patients âgés et ceux hospitalisés pour CVO et /ou STA ou ayant eu des complications de la drépanocytose sont ceux présentant les scores de qualité de vie les plus faibles. Des recherches complémentaires sont nécessaires pour affiner les connaissances sur la qualité de vie durant les différentes phases de la maladie et identifier les facteurs qui peuvent l'améliorer afin d'adapter la prise en charge des patients drépanocytaires.

L'étude en quelques chiffres

570 patients adultes drépanocytaires dont :

- 68,9 % de femmes ;
- 62,6 âgés d'au moins de 35 ans ;
- 1/3 vit chez leur parent ou un proche et 1/3 vit en couple ;
- 85,2 % ont un niveau scolaire au moins supérieur au secondaire ;
- 53 % ont une activité professionnelle ;
- 38,6% ont une situation financière stable ;
- 38 % sont parents d'au moins un enfant (dont 17,6% d'un enfant drépanocytaire) ;
- 93,2 % a un soutien (financier, matériel, moral) ;
- 48,8% ont été hospitalisé pour une CVO ou STA, 1/5 admis en soins intensifs et 1/3 a reçu une transfusion sanguine dans les 12 mois précédant l'étude ;
- 78,2 % ont un traitement contre la drépanocytose, 1/3 a un suivi psychologique ;
- 96,6 % ont eu une complication aiguë, 3/4 ont une complication chronique.

Cette étude a fait l'objet d'une publication en mai 2024 dans *Frontiers in Public Health* (<https://10.3389/fpubh.2024.1374805>).

Issifou Yaya

PROQOL, URC-ECO, ECEVE, UMR-S 1123 Université Paris Cité



→ Pourriez-vous nous décrire le contexte et les principaux résultats de l'étude ?

La drépanocytose est un problème de santé publique majeur en France car c'est la première maladie génétique en terme d'incidence (nombre de nouveaux cas par an) et de prévalence (nombre total de cas par an). Cette maladie a des répercussions sur la vie quotidienne des patients. L'étude DREPatient est une étude observationnelle qui a été implémentée en 2020 et financée par le laboratoire BlueBird Bio pour évaluer certains aspects socio-économiques de la maladie et leurs répercussions notamment sur la scolarité, l'employabilité, la situation matérielle et la qualité de vie des patients. Le cabinet Argo Santé a été mandaté pour implémenter l'étude sur tout le territoire. Le principal résultat de cette étude est que la qualité de vie des patients est moins bonne que celle de la population générale. D'autres résultats portant sur les aspects socio-économiques, la scolarité des enfants feront l'objet de valorisations ultérieures.

→ Est-ce que vous avez rencontré des difficultés particulières pour mener cette étude ?

La pandémie de covid a perturbé le déroulement de l'étude car le personnel médical était mobilisé pour s'occuper des cas de covid et les restrictions de circulation mises en place ont été un frein pour les acteurs de la recherche qui devaient aller collecter les données. La vie quotidienne des patients que nous souhaitions interroger a elle aussi été fortement impactée par la pandémie. Cela a été très compliqué de pouvoir les rencontrer physiquement. La période de recrutement a dû être prolongée et la communication de l'étude a été renforcée par des annonces sur notre site internet et via les associations afin de rassurer les patients et pouvoir les recruter en toute sécurité. Tous les questionnaires se remplissaient en ligne mais il fallait d'abord rencontrer physiquement les patients pour pouvoir les recruter.

→ Pourquoi avez-vous demandé aux patients de rapporter les complications (CVO, hospitalisation...) seulement dans les 12 derniers mois qui précédaient l'étude ?

C'est pour éviter les biais de mémoire. Nous avons interrogé directement les patients et au-delà de 12 mois, les personnes ne se rappellent pas vraiment et les informations ne sont plus trop fiables.

→ Quelles sont les limites de votre étude ?

Le recrutement n'a pas pu se faire de manière homogène sur toute l'étendue du territoire. Ce sont surtout les patients situés dans les grandes villes et ceux en contact avec les associations les plus actives qui ont accepté de participer au protocole.

De plus, l'échantillon n'a pas pu inclure les patients qui ne parlent pas français, ceux qui ont des problèmes de littéracie ou encore ceux qui n'ont pas accès à internet ce qui constitue un biais de représentativité.

Il faut souligner que la méthodologie observationnelle doit être interprétée avec précaution. L'étude Drépatient est une étude transversale qui ne permet pas de déterminer la causalité c'est-à-dire que si on dit par exemple tel phénomène a amélioré la qualité de vie, cela ne veut pas forcément dire que ça en est la cause. Pour identifier la cause, il faudrait mener des études complémentaires notamment longitudinales. Cependant ce type d'étude est plus complexe à mettre en place car les populations peuvent avoir des problèmes de mobilité, et il faut prendre en compte dans le calcul de l'effectif qu'un certain nombre de participants sera perdu de vue au cours de l'étude.

—▶ **Quelles sont les perspectives de votre étude ?**

La réalisation d'une analyse qualitative permettra de comprendre certains aspects de la qualité de vie. Les scores obtenus peuvent certes donner des informations mais il est très important d'étudier la perception qu'ont les patients de leur maladie. La littérature montre qu'habituellement les patients ont tendance à penser que la maladie se limite aux crises, ce qui n'est pas vrai et cela empêche leur bonne prise en charge à long terme. Il faudrait effectuer une analyse plus fine de la perception et de la compréhension de la maladie, aussi par les patients que par les soignants. Il y a encore malheureusement des soignants qui méconnaissent la drépanocytose ce qui pose un problème d'accès à des soins adaptés aux patients.

—▶ **Avez-vous des pistes qui pourraient améliorer la qualité de vie ?**

C'est une question à laquelle il est difficile de répondre en raison du design de l'étude, qui ne permet pas de déterminer la causalité des différents facteurs étudiés sur la qualité de vie des patients. Cependant, il existe certains facteurs modifiables sur lesquels nous pouvons agir comme l'accès au traitement. Les patients sous hydroxyurée rapportent une meilleure qualité de vie, l'hydroxyurée permettant de prévenir les crises et d'éviter les hospitalisations. L'accès à des soins spécifiques semble être la clé pour améliorer la qualité de vie des patients drépanocytaires. Il faudrait mettre en place des études plus expérimentales afin de démontrer scientifiquement ce qui améliorerait la qualité de vie, notamment les bénéfices du suivi psychologique, la prise en charge des complications aiguës et chroniques.

Appel à projets GIRCI GO 2024 – PHRC Interrégional

Budget	Non communiqué
Durée	Non communiqué
Date limite de dépôt des dossiers	18 octobre 2024 (lettre d'intention) 28 février 2025 (dossier complet)
Eligibilité	Professionnels des établissements de santé du territoire, aux GCS, aux maisons de santé et centres de santé de l'interrégion.
Objectif	Soutenir des projets s'appuyant sur les réseaux du GIRCI Grand Ouest, impliquant plusieurs établissements de l'interrégion, s'intégrant dans une démarche de structuration de la recherche dans le Grand Ouest et touchant à toutes les thématiques et disciplines (à l'exception des infections liées aux VIH, VHB et VHC, SARS-coV-2 et aux maladies infectieuses émergentes).

➔ Plus d'informations [ici](#)

Genopole - Appel à idées innovantes # édition 7

Budget	jusqu'à 10 000 €
Durée	Non communiqué
Date limite de dépôt des dossiers	30 octobre 2024
Eligibilité	Etre membre d'un établissement de soins d'Ile-de-France
Objectif	Optimiser la pratique médicale et améliorer la qualité des soins et bénéficier de l'aide du Genopole qui aidera à réaliser une preuve de concept et étudier le potentiel du projet.

➔ Plus d'informations [ici](#)

Bpifrance - AAP Innovations en biothérapies et bioproductions

Budget	20 000 €
Durée	Non communiqué
Date limite de dépôt des dossiers	26 novembre 2024 à 12h00 (midi heure de Paris)

Eligibilité	Proposer des solutions innovantes portant sur 2 des thématiques suivantes : 1. Développement de biothérapies en santé humaine ou santé animale (lorsqu'un impact sur la santé humaine est envisagé) 2. Développement d'outils de R&D ou de stratification pour le développement de biothérapies. 3. Développement de nouveaux procédés de bioproduction, nouveaux outils, équipements et systèmes d'optimisation de technologies de bioproduction existantes. Précisions supplémentaires ici :
Objectif	Catalyser et maintenir l'excellence de la recherche en biothérapie en accélérant notamment le transfert technologique et en assurant un flux constant d'innovations, de la paillasse au lit du patient.



Plus d'informations [ici](#)

Fondation Groupama - Prix de l'Innovation sociale 2025

Budget	jusqu'à 10 000 €
Durée	Non communiqué
Date limite de dépôt des dossiers	22 décembre 2024 à minuit
Eligibilité	Associations, filières maladies rares, institutions maladies rares et équipes de recherche.
Objectif	Soutenir des actions innovantes qui constituent une avancée significative ou une rupture dans le quotidien des personnes atteintes de maladies rares et leurs familles.



Plus d'informations [ici](#)

ANR - Soutien aux Réseaux Scientifiques Européens ou Internationaux

Budget	17 000 €
Durée	12 mois
Date limite de dépôt des dossiers	31 décembre 2024, à 23h59 CET (dossier de soumission simplifié)
Eligibilité	Réseau scientifique constitués de collaborateurs européens ou internationaux avec au moins un partenaire français, couvrant des sujets de toutes les disciplines.
Objectif	Renforcer un réseau scientifique européen ou international, coordonné par une équipe française ayant été invité à poursuivre sa candidature à la dernière étape d'un appel Européen ou international en plusieurs étapes.



Plus d'informations [ici](#)

IReSP - Subventions hors appels à projets

Budget	5 000 €
Durée	NA
Date limite de dépôt des dossiers	Envoi de la demande de subvention complète au maximum 3 mois avant la date de l'évènement.
Eligibilité	Manifestations scientifiques en santé publique (hors recherche clinique)
Objectif	<p>Soutenir des projets d'envergure locale, nationale ou internationale (plusieurs pays, dont la France) répondant aux objectifs de valorisation et diffusion des résultats de travaux de recherche auprès de différents publics(chercheurs, institutionnels, professionnels de terrain, société civile, acteurs politiques locaux...).</p> <p>Les projets devront concerner des thématiques de recherche en santé publique, en particulier sur :</p> <ul style="list-style-type: none">- Les déterminants de la santé, la promotion de la santé et la prévention (soutien à une meilleure connaissance des déterminants de santé et des comportements à risques, soutien aux approches transversales prenant en compte les interactions entre les différents déterminants, soutien à la recherche interventionnelle...);- Le fonctionnement du système de santé (Health Services Research) ; <p>Les politiques publiques et de santé (impact sur la santé des populations, conditions d'élaboration, évaluation, évolution...).</p>



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://filieremcgre.fr/espace-professionnels-de-sante/appels-a-projets/>

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

Anémie dysérythropoïétique congénitale

Updates on clinical and laboratory aspects of hereditary dyserythropoietic anemias

Russo R, Iolascon A, Andolfo I, Marra R, Rosato BE.

Int J Lab Hematol. 2024 Aug;46(4):595-605. doi: 10.1111/ijlh.14307

Congenital dyserythropoietic anemia type II and ineffective erythropoiesis: Challenges of diagnosis and management

Akpan IJ, Bogyo K, Leeman-Neill RJ, Wattacheril J, Francis RO.

Front Hematol. 2024;3:1389820. doi: 10.3389/frhem.2024.1389820

Anémie liée au métabolisme du fer et anémie sidéroblastique

Hematologic Manifestations in Primary Mitochondrial Diseases

Selvanathan A, Teo J, Parayil Sankaran B.

J Pediatr Hematol Oncol. 2024 Jul 1;46(5):e338-e347. doi: 10.1097/MPH.0000000000002890

ATP-Binding Cassette Transporter of Clinical Significance: Sideroblastic Anemia

Ogunbileje JO, Harris N, Wynn T, Kashif R, Stover B, *et al.*

J Pers Med. 2024 Jun 14;14(6):636. doi: 10.3390/jpm14060636

Recommendations for diagnosis, treatment, and prevention of iron deficiency and iron deficiency anemia

Iolascon A, Andolfo I, Russo R, Sanchez M, Busti F, *et al.*; from EHA-SWG Red Cell and Iron.

Hemasphere. 2024 Jul 15;8(7):e108. doi: 10.1002/hem3.108

Novel TMPRSS6 variants and their impact on iron-refractory iron deficiency anaemia in pregnancy: A North Indian genotype phenotype study

Sharma A, Kumar A, Rawat K, Vij S, Sandhu A, *et al.*

Br J Haematol. 2024 Aug;205(2):686-698. doi: 10.1111/bjh.19616

Murine models of erythroid 5ALA synthesis disorders and their conditional synthetic lethal dependency on pyridoxine

Ducamp S, Sendamarai A, Campagna DR, Chin DWL, Fujiwara Y, *et al.*

Blood. 2024 Jun 20;blood.2023023078. doi: 10.1182/blood.2023023078

Anomalies de la membrane du globule rouge

Intragenic deletions in SPTB are associated with hereditary spherocytosis: Series of 12 cases

Evrard O, Billes A, Badens C, Cadet E, Mansour-Hendili L, *et al.*

Br J Haematol. 2024 Aug 7. doi: 10.1111/bjh.19692

Flow Cytometry as a New Accessible Method to Evaluate Diagnostic Osmotic Changes in Patients with Red Blood Cell Membrane Defects

Beltrán A, Sánchez-Villalobos M, Salido E, Alguero C, Campos E, *et al.*

Biomedicines. 2024 Jul 19;12(7):1607. doi: 10.3390/biomedicines12071607

Hemolysis during open heart surgery in patients with hereditary spherocytosis - systematic review of the literature and case study

Mendrala K, Czober T, Darocha T, Hudziak D, Podsiadło P, *et al.*

Perioper Med (Lond). 2024 Jun 10;13(1):54. doi: 10.1186/s13741-024-00411-w

Piezo1 and its inhibitors: Overview and perspectives

Thien ND, Hai-Nam N, Anh DT, Baecker D.

Eur J Med Chem. 2024 Jul 5;273:116502. doi: 10.1016/j.ejmech.2024.116502

Systematic review of phenotypes in McLeod syndrome and case report of a progressive supranuclear palsy in a female carrier

Braun AA, Jung HH.

Orphanet J Rare Dis. 2024 Aug 25;19(1):312. doi: 10.1186/s13023-024-03309-4

Déficit en glucose-6-phosphate déshydrogénase

New WHO classification of genetic variants causing G6PD deficiency

Luzzatto L, Bancone G, Dugué PA, Jiang W, Minucci A, *et al.*

Bull World Health Organ. 2024 Aug 1;102(8):615-617. doi: 10.2471/BLT.23.291224

Pathogenic G6PD variants: Different clinical pictures arise from different missense mutations in the same codon

Costa S, Minucci A, Kumawat A, De Bonis M, Prontera G, *et al.*

Br J Haematol. 2024 Sep 18. doi: 10.1111/bjh.19775

Implementation of Universal Screening for G6PD Deficiency in Newborns

Milburn S, Bhutani VK, Weintraub A, Guttman K.

Pediatrics. 2024 Aug 1;154(2):e2024065900. doi: 10.1542/peds.2024-065900

Glucose-6-phosphate dehydrogenase deficiency as a cause for nonimmune hydrops fetalis and severe fetal anemia: A systematic review

Iyer NS, Mossayebi MH, Gao TJ, Haizler-Cohen L, Di Mascio D, *et al.*

Mol Genet Genomic Med. 2024 Jul;12(7):e2491. doi: 10.1002/mgg3.2491

Medications and Acute Hemolysis in G6PD-Deficient Patients - A Real-World Study

Gronich N, Rosh B, Stein N, Saliba W.

Clin Pharmacol Ther. 2024 Jun 6. doi: 10.1002/cpt.3333

Metformin-Induced Hemolysis in a Glucose-6-Phosphate Dehydrogenase-Deficient Patient: A Case Report

Jumani A, Ibrahim H, Purra H, Alkhazraji AK, AlNajjar MS.

Cureus. 2024 Jul 22;16(7):e65081. doi: 10.7759/cureus.65081

The global role of G6PD in infection and immunity

Shah SS, Stone EF, Francis RO, Karafin MS.

Front Immunol. 2024 Jun 13;15:1393213. doi: 10.3389/fimmu.2024.1393213

Association of Rheumatoid Arthritis with Glucose-6-Phosphate Dehydrogenase Deficiency: Results from a Case-Control Study

Dore MP, Pes GM, Mereu S, Piroddu J, Cavagna L, Erre GL.

Mediterr J Hematol Infect Dis. 2024 Jul 1;16(1):e2024056. doi: 10.4084/MJHID.2024.056

Hydroxychloroquine Therapy and G6PD Genotype

Kane M.

2023 May 2 [updated 2024 Aug 22]. In: Pratt VM, Scott SA, Pirmohamed M, Esquivel B, Kattman BL, Malheiro AJ, editors. Medical Genetics Summaries [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 2012–

Primaquine Therapy and G6PD and CYP2D6 Genotype

Kane M.

2023 Jul 6 [updated 2024 Aug 21]. In: Pratt VM, Scott SA, Pirmohamed M, Esquivel B, Kattman BL, Malheiro AJ, editors. Medical Genetics Summaries [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 2012–

Sulfamethoxazole-Trimethoprim Prophylaxis in Pediatric Oncology Patients With Glucose-6-Phosphate Dehydrogenase Deficiency

Stone RM, Haidar CE, Kornegay NM, Barker PJ, Karol SE, *et al.*

Pediatr Infect Dis J. 2024 Aug 7. doi: 10.1097/INF.0000000000004515

Type 2 Diabetes in Patients with G6PD Deficiency

Israel A, Raz I, Vinker S, Magen E, Green I, *et al.*

N Engl J Med. 2024 Aug 8;391(6):568-569. doi: 10.1056/NEJMc2406156

Risk of diabetes mellitus based on the interactive association between G6PD rs72554664 polymorphism and sex in Taiwan Biobank individuals

Chang YL, Nfor ON, Chou YH, Hsiao CH, Zhong JH, *et al.*

Sci Rep. 2024 Jun 4;14(1):12802. doi: 10.1038/s41598-024-63361-9

Adaptive selection at G6PD and disparities in diabetes complications

Breeyear JH, Hellwege JN, Schroeder PH, House JS, Poisner HM, *et al.*; VA Million Veteran Program; Giri A, Phillips LS, Edwards TL.

Nat Med. 2024 Sep;30(9):2480-2488. doi: 10.1038/s41591-024-03089-1

Serum microRNAs as new biomarkers for detecting subclinical hemolysis in the nonacute phase of G6PD deficiency

Boonpeng K, Shibuta T, Hirooka Y, Kulkeaw K, Palasuwan D, Umemura T.

Sci Rep. 2024 Jul 11;14(1):16029. doi: 10.1038/s41598-024-67108-4

Prevalence of glucose-6-phosphate dehydrogenase (G6PD) deficiency among the fulminant hepatitis A virus infection patients

Ejtehadi F, Serpoosh MS, Shahramian I, Aminlari L, Niknam R, *et al.*

Caspian J Intern Med. 2024 Summer;15(3):451-458. doi: 10.22088/cjim.15.3.451

G6PD deficiency mediated impairment of iNOS and lysosomal acidification affecting phagocytotic clearance in microglia in response to SARS-CoV-2

Mondal A, Munan S, Saxena I, Mukherjee S, Upadhyay P, *et al.*

Biochim Biophys Acta Mol Basis Dis. 2024 Oct;1870(7):167444. doi: 10.1016/j.bbadis.2024.167444

Déficit en pyruvate kinase

Comorbidities and complications in adult and paediatric patients with pyruvate kinase deficiency: Analysis from the Peak Registry

Glenthøj A, Grace RF, Lander C, van Beers EJ, Glader B, *et al.*

Br J Haematol. 2024 Aug;205(2):613-623. doi: 10.1111/bjh.19601

Déficit enzymatique érythrocytaire (autre)

Neuromuscular dysfunction and pathogenesis in triosephosphate isomerase deficiency

Myers TD, Li Y, Taiclet S, Cabada-Aguirre P, Kuti E, *et al.*

Sci Rep. 2024 Aug 10;14(1):18575. doi: 10.1038/s41598-024-69618-7

Drépanocytose

Health Supervision for Children and Adolescents With Sickle Cell Disease: Clinical Report

Yates AM, Aygun B, Nuss R, Rogers ZR; Section on Hematology/Oncology; American Society of Pediatric Hematology/Oncology.

Pediatrics. 2024 Aug 1;154(2):e2024066842. doi: 10.1542/peds.2024-066842

The Risk of Kidney Injury in Patients With Sickle Cell Disease Treated With Ketorolac for Acute Pain

Harris EM, Oni MO, Donado C, Heeney MM, Solodiuk J, *et al.*

J Pediatr Hematol Oncol. 2024 Jul 1;46(5):e290-e295. doi: 10.1097/MPH.0000000000002857

Evaluation of Hemoglobin S Percent Threshold to Prevent Severe Pain Events: A Secondary Analysis of the SIT Trial

Mejias J, Gonzalez-Barreto AR, Rodeghier M, DeBaun MR.

Blood Adv. 2024 Sep 18:bloodadvances.2024013216. doi: 10.1182/bloodadvances.2024013216

SNAP: Supportive non-invasive ventilation for acute chest syndrome prevention for hospitalized children with sickle cell disease: Perspectives of patients, parents, and the healthcare team

Cohen RT, Burrowes SAB, Williams CJ, Neri CM, Klings ES, *et al.*

Pediatr Blood Cancer. 2024 Sep;71(9):e31142. doi: 10.1002/pbc.31142

Comparison of Measures of Pain Intensity During Sickle Cell Disease Vaso-Occlusive Episodes

Knisely MR, Barnhart HX, Ibemere SO, Kavanagh P, Paice JA, *et al.*

J Pain. 2024 Aug 21:104658. doi: 10.1016/j.jpain.2024.104658

Impact of an Individualized Pain Plan to Treat Sickle Cell Disease Vaso-Occlusive Episodes in the Emergency Department

Siewny L, King AA, Melvin CL, Carpenter CR, Hankins JS, *et al.*

Blood Adv. 2024 May 30:bloodadvances.2023012439. doi: 10.1182/bloodadvances.2023012439

Can we use biomarkers to identify those at risk of acute pain from sickle cell disease?

Bhat V, Sheehan VA.

Expert Rev Hematol. 2024 Aug;17(8):411-418. doi: 10.1080/17474086.2024.2372322

Applying Comfort Theory to Improve Outcomes for People in Sickle Cell Crisis

Elting JK, Di Cesare D, Layne J, Murthy M, Mysyuk O.

Nurs Sci Q. 2024 Jul;37(3):249-254. doi: 10.1177/08943184241247080

Homelessness and Its Impact on the Management of Vaso-Occlusive Crises in Sickle Cell Disease

Narain A, Weaver S, Kalu N.

Health Soc Work. 2024 Aug 1;49(3):204-206. doi: 10.1093/hsw/hlae017

Unraveling the Complexity of Vaso-Occlusive Crises in Sickle Cell Disease: Insights from a Resource-Limited Setting

Kaponda A, Muya K, Panda J, Koto KK, Bonnechère B.

J Clin Med. 2024 Apr 25;13(9):2528. doi: 10.3390/jcm13092528

Exploring the role of viscosity-vaso-occlusion and haemolysis-endothelial dysfunction in pain sensitization among Jamaicans with sickle cell disease

Ramsay Z, Ali A, Grant J, Asnani M.

Br J Haematol. 2024 Jul 31. doi: 10.1111/bjh.19667

Investigating home-based opioid use among youth with sickle cell disease using ecological momentary assessment

Valrie C, Crawford BS, Shipman-Lacewell J, Ajibade O, Fuh B, *et al.*

Pediatr Blood Cancer. 2024 Aug;71(8):e31116. doi: 10.1002/pbc.31116

Real-world implementation of the David-Carroll buprenorphine protocol for pain management in sickle cell disease

Torres C, Galadanci N, Osborn J, Kanter J.

Am J Hematol. 2024 Sep;99(9):1834-1836. doi: 10.1002/ajh.27407

Risks and benefits of cannabis as a pain control modality in patients with sickle cell disease

Jacobs JW, Adkins BD, Stephens LD, Woo JS, Booth GS.

Clin Hematol Int. 2023 Dec 18;5(4):47-50. doi: 10.46989/001c.90837

Δ^9 -Tetrahydrocannabinol alleviates hyperalgesia in a humanized mouse model of sickle cell disease

Mabou Tagne A, Fotio Y, Gupta K, Piomelli D.

J Pharmacol Exp Ther. 2024 Jul 2;JPET-AR-2024-002285. doi: 10.1124/jpet.124.002285

Low-intensity transcranial focused ultrasound suppresses pain by modulating pain-processing brain circuits

Kim MG, Yu K, Yeh CY, Fouda R, Argueta D, *et al.*

Blood. 2024 Sep 5;144(10):1101-1115. doi: 10.1182/blood.2023023718

High-impact chronic pain in sickle cell disease: insights from the Pain in Sickle Cell Epidemiology Study (PiSCES)

Jagtiani A, Chou E, Gillespie SE, Liu K, Krishnamurti L, *et al.*

Pain. 2024 Oct 1;165(10):2364-2369. doi: 10.1097/j.pain.0000000000003262

Evaluating the feasibility of delivering a pain management programme for adults living with sickle cell disease

McLoughlin R, Love J, Smith JG, Scott W, Noblet T.

Br J Pain. 2024 Jun;18(3):257-273. doi: 10.1177/20494637231202744

Unmet Need: Mechanistic and Translational Studies of Sickle Cell Disease Pain as a Whole-Person Health Challenge

Belfer I, Chen W, Weber W, Edwards E, Langevin HM.

J Pain. 2024 Oct;25(10):104603. doi: 10.1016/j.jpain.2024.104603

Pain sensitisation in patients with sickle cell disease: A preliminary study

Garcia-Hernandez A, de la Coba P, Martinez-Triana RJ, Reyes Del Paso GA.

J Eval Clin Pract. 2024 Jul 18. doi: 10.1111/jep.14101

Provider Implicit Racial Bias in Pediatric Sickle Cell Disease

Mulchan SS, Theriault CB, DiVietro S, Litt MD, Sukhera J, *et al.*

J Racial Ethn Health Disparities. 2024 Jul 17. doi: 0.1007/s40615-024-02086-x

Feasibility of In-Home Virtual Reality for Chronic Pain in Sickle Cell Disease

Matthie N, Higgins M, Doorenbos A, Maddox T, Jenerette C.

Pain Manag Nurs. 2024 Aug;25(4):425-431. doi: 10.1016/j.pmn.2024.04.007

Limited access to transcranial Doppler screening and stroke prevention for children with sickle cell disease in Europe: Results of a multinational EuroBloodNet survey

Voi V, Gutierrez-Valle V, Cuzzubbo D, McMahon C, Casale M, *et al.*

Pediatr Blood Cancer. 2024 Oct;71(10):e31190. doi: 10.1002/pbc.31190

Quantifying dilated perivascular spaces in children with sickle cell disease

Karkoska KA, Gollamudi J, Sawyer RP, Woo D, Hyacinth HI.

Pediatr Blood Cancer. 2024 Sep;71(9):e31150. doi: 10.1002/pbc.31150

Intracranial aneurysms in sickle cell disease are associated with hemodynamic stress and anemia

Wang Y, Garland JS, Fella S, Reis MN, Parsons MS, *et al.*

Blood Adv. 2024 Sep 24;8(18):4823-4831. doi: 10.1182/bloodadvances.2024013928

Sickle Cell Disease Related Vasculopathies and Early Evaluation in a Pediatric Population

Panosyan DE, Panosyan WS, Corral I, Hanudel MR, Pak Y, Gotesman M.

In Vivo. 2024 May-Jun;38(3):1203-1212. doi: 10.21873/invivo.13556

Reversibility of Cognitive Deficits and Functional Connectivity With Transfusion in Children With Sickle Cell Disease

Power LC, Mirro AE, Binkley MM, Wang J, Williams KP, *et al.*

Neurology. 2024 May 28;102(10):e209429. doi: 10.1212/WNL.0000000000209429

Indications for cerebral revascularization for moyamoya syndrome in pediatric sickle cell disease determined by Delphi methodology

Robert AP, Hanel RA, Adelson PD, Lang SS, Grabb P, *et al.*
J Neurosurg Pediatr. 2024 Jul 19;1-12. doi: 10.3171/2024.5.PEDS2426

Genetic Modifiers of Stroke in Patients with Sickle Cell Disease-A Scoping Review

Oni MO, Brito M, Rotman C, Archer NM; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 Jun 7;25(12):6317. doi: 10.3390/ijms25126317

Rates of Strokes in Californians with Sickle Cell Disease in the Post-STOP Era

Oluwole OB, Brunson A, Adesina OO, Willen S, Keegan TH, *et al.*
Blood. 2024 Sep 20;blood.2023023031. doi: 10.1182/blood.2023023031

Incidence and Risk Factors for New and Recurrent Infarcts in Adults With Sickle Cell Disease

Jordan LC, King AA, Kanter J, Lebensburger J, Ford AL, *et al.*
J Am Heart Assoc. 2024 Jun 18;13(12):e033278. doi: 10.1161/JAHA.123.033278

Perinatal risk factors and neurocognitive outcomes in children and adolescents with sickle cell disease

Longoria JN, Dandar CM, Semko JH, Liyanage JSS, Kang G, *et al.*
Pediatr Blood Cancer. 2024 Oct;71(10):e31209. doi: 10.1002/pbc.31209

Prediction of Functional Academic Outcomes by Fine Motor Skills in Individuals With Sickle Cell Disease

Kearson L, Dandar C, Hoyt C, Longoria J, Okhominina V, *et al.*
Am J Occup Ther. 2024 Sep 1;78(5):7805205180. doi: 10.5014/ajot.2024.050684

Cognitive outcomes of children and adults with sickle cell anaemia: A contemporary cohort

Turner AD, Power LC, Yan Y, Binkley MM, Hood AM, *et al.*
Br J Haematol. 2024 Jul 9. doi: 10.1111/bjh.19642

Atrial Arrhythmia in Sickle Cell Anemia Adults: A missing link towards understanding and preventing strokes

d'Humières T, Sadraoui Z, Savale L, Boyer L, Guillet H, *et al.*
Blood Adv. 2024 Jul 5;bloodadvances.2024013208. doi: 10.1182/bloodadvances.2024013208

Measurement of left atrial size as a predictor of severity of illness in sickle cell disease

Nasrollahi FS, Gutierrez DJ, Nocek J, Folami J, Ekram S, *et al.*
Am J Emerg Med. 2024 Sep;83:126-128. doi: 10.1016/j.ajem.2024.07.001

Detection of Subclinical Cardiac Dysfunction in Patients With Sickle Cell Disease Using Speckle-Tracking Echocardiography

Kim MJ, Lee G, Lima G, Mukarram O, Crooks S, *et al.*
Am J Cardiol. 2024 Oct 15;229:28-35. doi: 10.1016/j.amjcard.2024.08.006

Pulmonary Hypertension in Sickle Cell Disease: Novel Findings of Gene Polymorphisms Related to Pathophysiology

Chatzidauid S, Flevari P, Tombrou I, Anastasiadis G, Dimopoulou M; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 Apr 27;25(9):4792. doi: 10.3390/ijms25094792

Diagnosis and management of chronic thromboembolic pulmonary hypertension (CTEPH) in sickle cell disease: A review

Hersi K, Ramani GV, Law JY, Sadek AS, Vaidya A, *et al.*
Pulm Circ. 2024 May 27;14(2):e12362. doi: 10.1002/pul2.12362

Prognostic value of exercise longitudinal right ventricular free wall strain in patients with sickle cell disease

Mansour MJ, De Marco C, Haddad K, Potter BJ, Argentin S, *et al.*
Int J Cardiovasc Imaging. 2024 Jul;40(7):1413-1421. doi: 10.1007/s10554-024-03116-9

PM2.5 exposure and household income are associated with lung function abnormalities in children with sickle cell disease

Kelchtermans J, Allen J, Bhandari A.
Pediatr Pulmonol. 2024 Aug 22. doi: 10.1002/ppul.27222

Effects of Air Pollution on Respiratory Events and Pain Crises among Children with Sickle Cell Disease in New York City

De A, Jung KH, Davis H, Siddiqi A, Kattan M, *et al.*
Ann Am Thorac Soc. 2024 Aug 28. doi: 10.1513/AnnalsATS.202310-860OC

Venoarterial Extracorporeal Membrane Oxygenation Therapy in Patients with Sickle Cell Disease: Case Series and Review for Intensive Care Physicians

Grazioli A, Rabin J, Rector RP, Wu ZJ, Burke AP, *et al.*
J Intensive Care Med. 2024 Aug 14;8850666241260605. doi: 10.1177/08850666241260605

Updates in Pediatric Sickle Cell Lung Disease

Gillespie M, Afolabi-Brown O, Machogu E, Willen S, Kopp BT.
Clin Chest Med. 2024 Sep;45(3):749-760. doi: 10.1016/j.ccm.2024.02.022

Prevalence, patterns, and factors associated with abnormal lung function among children with sickle cell disease in Uganda: a cross-sectional study

Aol PM, Fahdil G, Bongomin F, Okello B, Batte C, *et al.*
BMC Pediatr. 2024 Aug 10;24(1):516. doi: 10.1186/s12887-024-04988-5

Prevalence, Mortality, and Access to Care for Chronic Kidney Disease in Medicaid-Enrolled Adults With Sickle Cell Disease in California: Retrospective Cohort Study

Valle J, Lebensburger JD, Garimella PS, Gopal S.
JMIR Public Health Surveill. 2024 Jul 15;10:e57290. doi: 10.2196/57290

Persistent albuminuria and chronic kidney disease in adults with sickle cell anaemia: Results from a multicenter natural history study

Zhou LY, Derebail VK, Desai PC, Elsherif L, Patillo KL, *et al.*
Br J Haematol. 2024 Jul 8. doi: 10.1111/bjh.19636

Association of biomarkers of endothelial function, coagulation activation and kidney injury with persistent albuminuria in sickle cell anaemia

Elsherif L, Tang Y, Patillo KL, Wichlan D, Ogu UO, *et al.*
Br J Haematol. 2024 Sep 3. doi: 10.1111/bjh.19743

Systematic Review of Genetic Modifiers Associated with the Development and/or Progression of Nephropathy in Patients with Sickle Cell Disease

Labarque V, Okocha EC; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 May 16;25(10):5427. doi: 10.3390/ijms25105427

Relevance of repeated analyses of albuminuria and glomerular filtration rate in African children with sickle cell anaemia

Nkoy AB, Mumaka FM, Ngonde A, Mafumba SK, Matoka TT, *et al.*
Br J Haematol. 2024 Jun 27. doi: 10.1111/bjh.19603

Sex-disaggregated analysis of acute kidney injury in hospitalized children with sickle cell anemia in Uganda

Weckman A, McDonald CR, Naggayi SK, Soranno DE, Conroy AL, Batte A.
Am J Physiol Renal Physiol. 2024 Aug 1;327(2):F304-F313. doi: 10.1152/ajprenal.00385.2023

Roxadustat, an hypoxia-inducible factor-prolyl hydroxylase inhibitor induce sickle cell crisis: A case report

Chabannes M, Vagnet A, Benjemia L, Seibel J, Tristant M, *et al.*
Br J Haematol. 2024 Sep 18. doi: 10.1111/bjh.19783

Advances in sickle cell retinopathy screening techniques, tests, and practices: A systematic review

Cummings OW, Rahman S, Fletcher L, Scott AW.

Am J Hematol. 2024 Jul 23. doi: 10.1002/ajh.27439. Epub ahead of print. PMID: 39041108.

The Association of Sickle-Cell Disorders With Diabetic Retinopathy: A Large Database Study

Chauhan MZ, Elhousseiny AM, Sallam AB.

Ophthalmol Sci. 2024 Feb 13;4(4):100490. doi: 10.1016/j.xops.2024.100490

Surgical outcomes in concurrent sickle cell and diabetic retinopathy

Rohowetz LJ, Shaheen A, Magraner M, Flynn HW Jr, Williams BK Jr, *et al.*

Retina. 2024 Sep 1;44(9):1560-1564. doi: 10.1097/IAE.0000000000004156

Retrospective Cohort Study of Sickle Cell Disease and Large Vessel Retinal Vascular Occlusion Risk in a National United States Database

Kaufmann GT, Russell M, Shukla P, Singh RP, Talcott KE.

Ophthalmol Retina. 2024 Jul 19:S2468-6530(24)00336-1. doi: 10.1016/j.oret.2024.07.013

Macular neurodegenerative and vascular changes on OCTA in sickle cell disease are not related to its ocular and systemic complications

Brandsen RP, Biemond BJ, Nasim GZ, Nur E, Schlingemann RO, Diederens RMH.

Retina. 2024 Aug 13. doi: 10.1097/IAE.0000000000004247

Surgical outcomes of retinal detachment associated with proliferative sickle cell retinopathy

Ahmed I, Wakabayashi T, Gonzales AF, Ong SS, Light JG, Handa JT, *et al.*

Retina. 2024 Sep 1;44(9):1565-1571. doi: 10.1097/IAE.0000000000004145

Optical Coherence Tomography Angiography of Macular Microangiopathy in Children With Sickle Cell Disease

Raffa EH, Raffa L, Almadani S, Murad W, Alshanti H.

J Pediatr Hematol Oncol. 2024 Aug 2. doi: 10.1097/MPH.0000000000002934

The molecular mechanism responsible for HbSC retinopathy may depend on the action of the angiogenesis-related genes *ROBO1* and *SLC38A5*

da Silva Costa SM, Ito MT, da Cruz PRS, De Souza BB, Rios VM, *et al.*

Exp Biol Med (Maywood). 2024 Jul 24;249:10070. doi: 10.3389/ebm.2024.10070

Bone and Joint Infections in Children With Sickle Cell Disease in French Guiana: A 13-Year Retrospective Multicenter Review

Furgier A, Goutines J, Dobian S, Zappa M, Demar M, *et al.*

Pediatr Infect Dis J. 2024 Oct 1;43(10):946-952. doi: 10.1097/INF.0000000000004416

Total Hip Arthroplasty Complications in Sickle Cell Disease: Systematic Review and Meta-Analysis

Alfaya FF, Ghazy RM, Hammouda EA, Mahfouz AA, Faya HK, *et al.*

J Clin Med. 2024 Jul 15;13(14):4129. doi: 10.3390/jcm13144129

Lumbar Fusions in Patients with Sickle Cell Disease: A Propensity-Matched Analysis of Postoperative Complications

Liu KG, Ton AT, Brown M, Mertz K, Patel S, *et al.*

World Neurosurg. 2024 Jul 25:S1878-8750(24)01292-0. doi: 10.1016/j.wneu.2024.07.152

Bone Disease among Children with Sickle Cell Disease: A Scoping Review of Incidence and Interventions

Mulyana AM, Rakhmawati W, Pramukti I, Lukman M, Wartakusumah R, Mediani HS.

J Multidiscip Healthc. 2024 Jul 9;17:3235-3246. doi: 10.2147/JMDH.S475371

The many faces of sickle cell disease in children: complications in the appendicular skeleton

De La Hoz Polo M, Hudson VE, Adu J, Chakravorty S, Haque S.

Pediatr Radiol. 2024 Aug;54(9):1437-1450. doi: 10.1007/s00247-024-05913-9

Therapy with bone marrow mesenchymal stem cells in bone regeneration in children with osteonecrosis secondary to sickle cell disease

Teixeira TRG, Daltro GC, Sberge FL, Barreto ESR, da Silva AF.
Front Cell Dev Biol. 2024 May 6;12:1410861. doi: 10.3389/fcell.2024.1410861

Current and emerging drug treatment strategies to tackle sickle cell anemia

Persaud Y, Leonard A, Rai P.
Expert Opin Emerg Drugs. 2024 Jul 22:1-20. doi: 10.1080/14728214.2024.2379260

Hydroxyurea in the sickle cell disease modern era

Riley C, Kraft WK, Miller R.
Expert Rev Clin Pharmacol. 2024 Sep;17(9):777-791. doi: 10.1080/17512433.2024.2390915

Effects of hydroxyurea on fertility in male and female sickle cell disease patients. A systemic review and meta-analysis

Sewaralthahab S, Alsubki LA, Alhrabi MS, Alsultan A.
PLoS One. 2024 Jun 7;19(6):e0304241. doi: 10.1371/journal.pone.0304241

Impact of Hydroxyurea on follicle density in patients with sickle cell disease

Diesch-Furlanetto T, Sanchez C, Atkinson A, Pondarre C, Dhedin N, *et al.*
Blood Adv. 2024 Jul 18:bloodadvances.2023011536. doi: 10.1182/bloodadvances.2023011536

Hydroxyurea in sickle cell disease and invasive bacterial infections: a case-control study

Pascault A, Koehl B, Brousse V, Benkerrou M, Gaschignard J.
J Pediatric Infect Dis Soc. 2024 Jul 6:piae071. doi: 10.1093/jpids/piae071

A Pharmacist-Managed Hydroxyurea Prescribing Protocol Improves Uptake and Optimization among Patients with Sickle Cell Disease

Roessner C, Sale T, Uminski K, Goodyear D, Rydz N.
Adv Hematol. 2024 May 30;2024:4753349. doi: 10.1155/2024/4753349

Feasibility of a clinic-based, multicomponent hydroxyurea adherence intervention for adolescents and young adults with sickle cell disease

Murphy BN, Keenan-Pfeiffer ME, Lane A, Crosby LE, Walsh KE.
Pediatr Blood Cancer. 2024 Sep;71(9):e31144. doi: 10.1002/pbc.31144

Hydroxyurea maintains working memory function in pediatric sickle cell disease

Lai J, Zou P, Dalboni da Rocha JL, Heitzer AM, Patni T, *et al.*
PLoS One. 2024 Jun 27;19(6):e0296196. doi: 10.1371/journal.pone.0296196

Hydroxyurea to prevent brain injury in children with sickle cell disease (HU Prevent)-A randomized, placebo-controlled phase II feasibility/pilot study

Casella JF, Furstenau DK, Adams RJ, Brambilla DJ, Lebensburger JD, *et al.*
Am J Hematol. 2024 Jul 2. doi: 10.1002/ajh.27423

Reviewing the impact of hydroxyurea on DNA methylation and its potential clinical implications in sickle cell disease

Lewis J, Guilcher GMT, Greenway SC.
Eur J Haematol. 2024 Sep;113(3):264-272. doi: 10.1111/ejh.14247

Assessing multilevel barriers to hydroxyurea adherence in youth with sickle cell disease using pharmacy-based refill records

Smaldone A, Manwani D, Aygun B, Appiah-Kubi A, Smith-Whitley K, Green NS.
Pediatr Blood Cancer. 2024 Sep;71(9):e31170. doi: 10.1002/pbc.31170

Determinants of the haemoglobin level in patients with sickle cell disease living in sub-Saharan Africa: Major impact of the country of residence and independent effects of leucocyte and platelet counts and haemolysis

Rossi M, Belinga S, Tolo A, Diop S, Diagne I, *et al.*

Br J Haematol. 2024 Aug;205(2):664-673. doi: 10.1111/bjh.19576

Towards genomic medicine: a tailored next-generation sequencing panel for hydroxyurea pharmacogenomics in Tanzania

Nkya S, Nzunda C, Saukiwa E, Kaywanga F, Buberwa E, *et al.*

BMC Med Genomics. 2024 Jul 18;17(1):190. doi: 10.1186/s12920-024-01924-5

Hydroxyurea mobile directly observed therapy versus standard monitoring in patients with sickle cell anemia: a phase 2 randomized trial

Sasi P, Makubi A, Sangeda RZ, Ngaeje MY, Mmbando BP, *et al.*

Commun Med (Lond). 2024 Aug 9;4(1):160. doi: 10.1038/s43856-024-00552-5

Understanding patient-related barriers to hydroxyurea use among adolescent and adult patients with sickle cell disease in Mulago and Kiruddu hospitals, Uganda, a qualitative study

Namaganda P, Nantume P, Mubiru KR, Twimukye A, Wiltshire CS.

BMC Health Serv Res. 2024 May 27;24(1):666. doi: 10.1186/s12913-024-11125-6

Pharmacokinetic-Guided Hydroxyurea to Reduce Transfusions in Ugandan Children with Sickle Cell Anemia: Study Design of the Alternative Dosing And Prevention of Transfusions Trial

Power-Hays A, Namazzi R, Kato C, McElhinney KE, Conroy AL, *et al.*

Acta Haematol. 2024 Jun 5:1-12. doi: 10.1159/000539541

Clinical Practice Patterns in Sickle Cell Disease Treatment: Disease-modifying and Potentially Curative Therapies

Sánchez LM, Morrone K, Frei-Jones M, Fasipe TA.

J Pediatr Hematol Oncol. 2024 Jul 1;46(5):e277-e283. doi: 10.1097/MPH.0000000000002869

Crizanlizumab in adult patients with sickle cell disease: a retrospective German analysis

Poppenborg F, Röth A, Yamamoto R, Reinhardt HC, Alashkar F.

Front Hematol. 2024;3:1374181. doi: 10.3389/frhem.2024.1374181

Cost-effectiveness of l-glutamine versus crizanlizumab for adults with sickle cell disease: model focused on reducing pain episode costs from Qatar's healthcare perspective

Adel AM, Abushanab D, Al-Badriyeh D, Hamad A, Alshurafa A, Yassin MA.

SAGE Open Med. 2024 May 6;12:20503121231224551. doi: 10.1177/20503121231224551

Evaluation of the effect of voxelotor and darbepoetin alfa on hemoglobin levels in patients with sickle cell disease

Weaver SB, Akinwale H, Nonyel NP, Wingate LT.

Expert Rev Hematol. 2024 Jun;17(6):255-260. doi: 10.1080/17474086.2024.2352497

Changes in indicators of cerebral metabolic stress following treatment with voxelotor in children and adolescents with sickle cell anemia

Heitzer AM, Zou P, Hodges J, Brown C, Davis M, *et al.*

eJHaem. Published online Aug 27, 2024. doi: 10.1002/jha2.1001

Metabolic blood profile and response to treatment with the pyruvate kinase activator mitapivat in patients with sickle cell disease

van Dijk MJ, Ruiters TJJ, van der Veen S, Rab MAE, van Oirschot BA, *et al.*

Hemasphere. 2024 Jun 25;8(6):e109. doi: 10.1002/hem3.109

Activating pyruvate kinase improves red blood cell integrity by reducing band 3 tyrosine phosphorylation

Le K, Wang X, Chu J, Lundt M, Lee YY, *et al.*

Blood Adv. 2024 Sep 12:bloodadvances.2024013504. doi: 10.1182/bloodadvances.2024013504

A pharmacokinetic-pharmacodynamic analysis of l-glutamine for the treatment of sickle cell disease: Implications for understanding the mechanism of action and evaluating response to therapy

Sadaf A, Dong M, Pfeiffer A, Korpik J, Kalfa TA, *et al.*
Br J Haematol. 2024 Jul 8. doi: 10.1111/bjh.19632

Safety and efficacy of L-Glutamine in reducing the frequency of acute complications among patients with sickle cell disease: A randomized controlled study

Ebeid FSE, Aly NH, Shaheen NM, Abdellatif SMA, Okba AAM, *et al.*
Ann Hematol. 2024 Sep;103(9):3493-3506. doi: 10.1007/s00277-024-05877-8

Advances in pharmacotherapy for sickle cell disease: what is the current state of play?

Alan S, Kanter J.
Expert Opin Pharmacother. 2024 Jul;25(10):1325-1334. doi: 10.1080/14656566.2024.2377711

Budget Impact of Disease-Modifying Treatments and a CRISPR Gene-Edited Therapy for Sickle Cell Disease

Abdallah K, Huys I, Claes KJ, Simoens S.
Clin Drug Investig. 2024 Aug;44(8):611-627. doi: 10.1007/s40261-024-01384-w

Folic Acid in the Treatment of Sickle Cell Disease: A Systematic Review

Arrey Agbor DB, Panday P, Ejaz S, Gurugubelli S, Prathi SK, *et al.*
Cureus. 2024 Apr 10;16(4):e57962. doi: 10.7759/cureus.57962

A molecular glue degrader of the WIZ transcription factor for fetal hemoglobin induction

Ting PY, Borikar S, Kerrigan JR, Thomsen NM, Aghania E, *et al.*
Science. 2024 Jul 5;385(6704):91-99. doi: 10.1126/science.adk6129

PGC-1 α agonism induces fetal hemoglobin and exerts antisickling effects in sickle cell disease

Sun Y, Benmhammed H, Al Abdullatif S, Habara A, Fu E, *et al.*
Sci Adv. 2024 Aug 2;10(31):eadn8750. doi: 10.1126/sciadv.adn8750

Exploring the genetic mechanisms: SELP gene's contribution to alleviating vaso-occlusive crisis in sickle cell disease

Gupta P, Choudhari V, Kumar R.
Gene. 2024 Nov 30;928:148805. doi: 10.1016/j.gene.2024.148805

Targeted therapeutic management based on phytoconstituents for sickle cell anemia focusing on molecular mechanisms: Current trends and future perspectives

Islam MR, Rauf A, Akash S, Sharkar M, Mahreen M, *et al.*
Phytomedicine. 2024 Oct;133:155936. doi: 10.1016/j.phymed.2024.155936

Safety and efficacy of monthly high-dose vitamin D₃ supplementation in children and adolescents with sickle cell disease

Hanna D, Kamal DE, Fawzy HM, Abd Elkhalek R.
Eur J Pediatr. 2024 Aug;183(8):3347-3357. doi: 10.1007/s00431-024-05572-w

Current and Future Therapeutics for Treating Patients with Sickle Cell Disease

Barak M, Hu C, Matthews A, Fortenberry YM.
Cells. 2024 May 16;13(10):848. doi: 10.3390/cells13100848

Automated red blood cell exchange with a post-procedure haematocrit targeted at 34% in the chronic management of sickle cell disease

M Ross J, Forté S, Mercure-Corriveau N, Lemay AS, Rioux-Massé B, *et al.*
Br J Haematol. 2024 Jul 30. doi: 10.1111/bjh.19674

Chronic automated red cell exchange therapy for sickle cell disease

Zakieh A, Mercure-Corriveau N, Lanzkron S, Feng X, Vozniak S, *et al.*
Transfusion. 2024 Aug;64(8):1509-1519. doi: 10.1111/trf.17924

Red cell exchange modulates neutrophil degranulation responses in sickle cell disease

Lee GM, Boyle K, Batchvarova M, Delahunty M, Suggs MA, *et al.*

Transfusion. 2024 Jul 9. doi: 10.1111/trf.17947

Eculizumab for management of hyperhemolysis syndrome in pediatric patients with sickle cell disease: A single-center case series

Lapite A, Bhar S, Fasipe T.

Pediatr Blood Cancer. 2024 Aug;71(8):e31061. doi: 10.1002/pbc.31061

Genotyped RHD+ red cells for D-positive patients with sickle cell disease with conventional RHD and unexpected anti-D

Chou ST, Mewha J, Friedman DF, Lazariu V, Makrm S, *et al.*

Blood. 2024 Aug 20;blood.2024025602. doi: 10.1182/blood.2024025602

Position paper on advancing sickle cell disease management in France by bridging the clinical practices and guidelines through expert insights

Benmoussa K, Bernaudin F, Connes P, Héquet O, Joseph L, *et al.*

Transfus Apher Sci. 2024 Oct;63(5):103988. doi: 10.1016/j.transci.2024.103988

Molecular genotyping versus serological diagnosis for RH blood group typing in sickle cell patients

Eftekhari Z, Oodi A, Amirzadeh N, Mohammadipour M, Keikhaei Dehdezi B, Jalali Far MA.

Expert Rev Clin Immunol. 2024 Aug 8:1-6. doi: 10.1080/1744666X.2024.2388700

Iron absorption in adults with sickle cell anemia: a stable-isotope approach

Omena J, Bezerra FF, Voll VM, Braz BF, Santelli RE, *et al.*

Eur J Nutr. 2024 Sep;63(6):2163-2172. doi: 10.1007/s00394-024-03417-8

Significant pituitary siderosis is common in transfusion dependent sickle cell disease

Vadivelan A, Doyle E, Carson S, Denton CC, Veluswamy S, *et al.*

Blood. 2024 Jul 24;blood.2024025462. doi: 10.1182/blood.2024025462

Long-term quality of life after hematopoietic cell transplant for sickle cell disease in childhood: A STELLAR interim analysis

Arnold SD, Bakshi N, Ross D, Smith C, Sinha C, *et al.*

Am J Hematol. 2024 Aug 6. doi: 10.1002/ajh.27436

Reproductive Health Assessment and Reports of Fertility Counseling in Pediatric and Adolescent Patients with Sickle Cell Disease After Hematopoietic Cell Transplantation

George SA, Veludhandi A, Xiang Y, Liu K, Stenger E, *et al.*

Transplant Cell Ther. 2024 Sep;30(9):912.e1-912.e13. doi: 10.1016/j.jtct.2024.06.029

Posterior reversible encephalopathy syndrome post stem cell transplantation in sickle cell disease: case series and literature review

BinAmir HA, AlAhmari A, AlQahtani A, Mohamed G, Alotaibi F, *et al.*

Front Med (Lausanne). 2024 May 1;11:1330688. doi: 10.3389/fmed.2024.1330688

Assessing acute toxicity profiles of HLA-identical hematopoietic stem cell transplantation in pediatric patients with sickle cell anemia: A comprehensive analysis on behalf of the SFGM-TC

Delafof M, Dalle JH, Pondarre C, Andrieu GP, Fahd M, *et al.*; Société Francophone de Greffe de Moelle et de Thérapie Cellulaire.

Am J Hematol. 2024 Sep 24. doi: 10.1002/ajh.27486

Neurocognitive outcome in children with sickle cell disease after myeloimmunoablative conditioning and haploidentical hematopoietic stem cell transplantation: a non-randomized clinical trial

Braniecki S, Vichinsky E, Walters MC, Shenoy S, Shi Q, *et al.*

Front Neurol. 2024 May 22;15:1263373. doi: 10.3389/fneur.2024.1263373

Pulmonary Function After Non-Myeloablative Hematopoietic Cell Transplant for Sickle Cell Disease

Ruhl AP, Shalhoub R, Jeffries N, Limerick EM, Leonard A, *et al.*

Ann Am Thorac Soc. 2024 Aug 27. doi: 10.1513/AnnalsATS.202309-771OC

Therapeutic plasma exchange for sickle cell disease acute complications: A systematic review

Denoon RB, Soares Ferreira Junior A, Tuttle B, Onwuemene OA.

Transfusion. 2024 Aug;64(8):1570-1587. doi: 10.1111/trf.17932

Assessing Psychosocial Risk and Resilience to Support Readiness for Gene Therapy in Sickle Cell Disease: A Consensus Statement

Hardy SJ, Crosby LE, Porter JS, Sil S, Valrie CR, *et al.*

JAMA Netw Open. 2024 Aug 1;7(8):e2429443. doi: 10.1001/jamanetworkopen.2024.29443

Consensus-driven target product profiles for curative sickle cell disease gene therapies

Bukini D, Makani J, McCune J, Lee D, Bansbach C, *et al.*

Mol Ther Methods Clin Dev. 2024 Jun 22;32(3):101287. doi: 10.1016/j.omtm.2024.101287

A budget impact analysis of gene therapy for sickle cell disease: an updated analysis

DeMartino PC, Haag MB, Caughey AB, Roth JA.

Blood Adv. 2024 Sep 10;8(17):4658-4661. doi: 10.1182/bloodadvances.2024013093

Gene therapies on the horizon for sickle cell disease: a clinician's perspective

Butt H, Tisdale JF.

Expert Rev Hematol. 2024 Sep;17(9):555-566. doi: 10.1080/17474086.2024.2386366

Safety and efficacy studies of CRISPR-Cas9 treatment of sickle cell disease highlights disease-specific responses

Fрати G, Brusson M, Sartre G, Mlayah B, Felix T, *et al.*

Mol Ther. 2024 Jul 22:S1525-0016(24)00470-2. doi: 10.1016/j.ymthe.2024.07.015

Non-viral DNA delivery and TALEN editing correct the sickle cell mutation in hematopoietic stem cells

Moiani A, Letort G, Lizot S, Chalumeau A, Foray C, *et al.*

Nat Commun. 2024 Jun 11;15(1):4965. doi: 10.1038/s41467-024-49353-3

Bone-marrow-homing lipid nanoparticles for genome editing in diseased and malignant haematopoietic stem cells

Lian X, Chatterjee S, Sun Y, Dilliard SA, Moore S, *et al.*

Nat Nanotechnol. 2024 Sep;19(9):1409-1417. doi: 10.1038/s41565-024-01680-8

Development and IND-enabling studies of a novel Cas9 genome-edited autologous CD34⁺ cell therapy to induce fetal hemoglobin for sickle cell disease

Katta V, O'Keefe K, Li Y, Mayuranathan T, Lazzarotto CR, *et al.*

Mol Ther. 2024 Jul 31:S1525-0016(24)00477-5

Preclinical safety assessment of modified gamma globin lentiviral vector-mediated autologous hematopoietic stem cell gene therapy for hemoglobinopathies

Shadid M, Shrestha A, Malik P.

PLoS One. 2024 Jul 8;19(7):e0306719. doi: 10.1371/journal.pone.0306719

UM171 enhances fitness and engraftment of gene modified hematopoietic stem cells from sickle cells disease patients

Liu B, Klatt D, Zhou Y, Manis JP, Sauvageau G, *et al.*

Blood Adv. 2024 Sep 18:bloodadvances.2024013932. doi: 10.1182/bloodadvances.2024013932

Sickle cell disease and infertility risks: implications for counseling and care of affected girls and women

Pecker LH, Cameron K.

Expert Rev Hematol. 2024 Aug;17(8):493-504. doi: 10.1080/17474086.2024.2372320

The impact of sickle cell disease and its treatment on ovarian reserve in reproductive-aged Black women

Shandley LM, Fasano RM, Spencer JB, Mertens AC, McPherson LJ, *et al.*

Br J Haematol. 2024 Aug;205(2):674-685. doi: 10.1111/bjh.19582

Prophylactic red cell transfusions for sickle cell disease pregnancy: increased use of therapy could transform outcomes

Alan S, Sharma D, Pecker LH.

Curr Opin Hematol. 2024 Aug 15. doi: 10.1097/MOH.0000000000000837

Prophylactic exchange transfusion in sickle cell disease pregnancy: a TAPS2 feasibility randomized controlled trial

Oteng-Ntim E, Oakley LL, Robinson V, Brien S, Joseph J, *et al.*

Blood Adv. 2024 Aug 27;8(16):4359-4369. doi: 10.1182/bloodadvances.2024012923

Contraceptive Attitudes and Beliefs of Women With Sickle Cell Disease: A Qualitative Study

Roe AH, Wu J, McAllister A, Aragoncillo S, Nunyi E, *et al.*

Womens Health Issues. 2024 Jul-Aug;34(4):409-416. doi: 10.1016/j.whi.2024.03.007

La drépanocytose en Guyane : bilan de 30 années de dépistage néonatal (1992-2021)

Elenga N, Ro V, Mafema Missindu J, Thomas Boizan N, Vaz T, *et al.*

Med Trop Sante Int. 2024 Feb 13;4(1):mtsiv4i1.2024.488. French. doi: 10.48327/mtsiv4i1.2024.488

Screening for sickle cell disease: focus on newborn investigations

Mosca A, Paleari R, Palazzi G, Pancaldi A, Iughetti L, *et al.*; SIBioC Working Group on Diabetes and of the SIBioC-SIMMESN Working Group on Neonatal Clinical Biochemistry and Metabolic Diseases.

Clin Chem Lab Med. 2024 Jun 19;62(9):1804-1813. doi: 10.1515/cclm-2024-0478

Differential sensitivity to hypoxia enables shape-based classification of sickle cell disease and trait blood samples at point of care

D'Costa C, Sharma O, Manna R, Singh M, Singh S, *et al.*

Bioeng Transl Med. 2023 Dec 27;9(4):e10643. doi: 10.1002/btm2.10643

Definition of an algorithm to identify patients with sickle-cell disease in the French National Health Database

Walter O, Cougoul P, Zadrou Y, Moulis G, Lafaurie M.

Eur J Intern Med. 2024 May 31;S0953-6205(24)00218-8. doi: 10.1016/j.ejim.2024.05.012

The Ugandan sickle Pan-African research consortium registry: design, development, and lessons

Nsubuga M, Mutegeki H, Jjingo D, Munube D, Namazzi R, *et al.*

BMC Med Inform Decis Mak. 2024 Jul 29;24(1):212. doi: 10.1186/s12911-024-02618-9

Standardization of coding definitions for sickle cell disease complications: A systematic literature review

Ericksen PN, Dabbous F, Ghosh R, Shah S, Sun X, *et al.*

Pharmacoepidemiol Drug Saf. 2024 Sep;33(9):e5769. doi: 10.1002/pds.5769

The clinical spectrum of HbSC sickle cell disease-not a benign condition

Nelson M, Noisette L, Pugh N, Gordeuk V, Hsu LL, *et al.*

Br J Haematol. 2024 Aug;205(2):653-663. doi: 10.1111/bjh.19523

The German sickle cell disease registry reveals a surprising risk of acute splenic sequestration and an increased transfusion requirement in patients with compound heterozygous sickle cell disease HbS/ β -thalassaemia and no or low HbA expression

Allard P, Tagliaferri L, Weru V, Cario H, Lobitz S, *et al.*; German Sickle Cell Disease Study Group.

Eur J Haematol. 2024 Jun 30. doi: 10.1111/ejh.14259

Acute Pancreatitis in Individuals with Sickle Cell Disease: A Systematic Review

Dike CR, DadeMatthews A, DadeMatthews O, Abu-El-Haija M, Lebensburger J, *et al.*

J Clin Med. 2024 Aug 11;13(16):4712. doi: 10.3390/jcm13164712

Presentation of Acute Pancreatitis in Sickle Cell Disease Patients: A Single Hospital Experience

Al-Hindi S, Khalaf Z, Al-Sousi AN.

Afr J Paediatr Surg. 2024 Jul 1;21(3):151-154. doi: 10.4103/ajps.ajps_133_22

The enigma of sickle cell hepatopathy: Pathophysiology, clinical manifestations and therapy

Rizvi I, Solipuram D, Kaur N, Komel A, Batool S, Wang J.

Br J Haematol. 2024 Jul 8. doi: 10.1111/bjh.19620

Controversies in the pathophysiology of leg ulcers in sickle cell disease

Catella J, Guillot N, Nader E, Skinner S, Poutrel S, *et al.*

Br J Haematol. 2024 Jul;205(1):61-70. doi: 10.1111/bjh.19584

Impaired microvascular function in patients with sickle cell anemia and leg ulcers improved with healing

Catella J, Turpin E, Connes P, Nader E, Carin R, *et al.*

Br J Haematol. 2024 Sep 24. doi: 10.1111/bjh.19785

Clinical Characteristics, Management, and Outcomes of Patients with Renal Medullary Carcinoma: A Single-center Retrospective Analysis of 135 Patients

Lebenthal JM, Kontoyiannis PD, Hahn AW, Lim ZD, Rao P, *et al.*

Eur Urol Oncol. 2024 Jul 15:S2588-9311(24)00175-5. doi: 10.1016/j.euo.2024.07.002

Comorbid ADHD and Pediatric Sickle Cell Disease: Prevalence and Risk Factors

Bills SE, Schatz J, Varanasi S, Johnston JD, Gillooly E.

J Clin Psychol Med Settings. 2024 Jul 9. doi: 10.1007/s10880-024-10027-3

Concomitant sickle cell disease and systemic lupus erythematosus: A single-center case series

Lapite A, Sánchez LM, Altaffer AL, Rae M, Ramirez AA, *et al.*

Pediatr Blood Cancer. 2024 Oct;71(10):e31194. doi: 10.1002/pbc.31194

Targeting heme in sickle cell disease: new perspectives on priapism treatment

Silveira THR, Calmasini FB, de Oliveira MG, Costa FF, Silva FH.

Front Physiol. 2024 Jul 17;15:1435220. doi: 10.3389/fphys.2024.1435220

End Organ Affection in Sickle Cell Disease

Bathla T, Lotfollahzadeh S, Quisel M, Mehta M, Malikova M, Chitalia VC.

Cells. 2024 May 29;13(11):934. doi: 10.3390/cells13110934

Clearance of pathogenic erythrocytes is maintained despite spleen dysfunction in children with sickle cell disease.

Sissoko A, Cissé A, Duverdier C, Marin M, Dumas L, *et al.*

Am J Hematol. 2024 Sep 17. doi: 10.1002/ajh.27481

Splenic complications in pediatric sickle cell disease: A retrospective cohort review

George A, Conneely SE, Mangum R, Fasipe T, Lupo PJ, Scheurer ME.

Pediatr Blood Cancer. 2024 Oct;71(10):e31219. doi: 10.1002/pbc.31219

Rescue splenic artery embolization in an adult patient of sickle cell disease presented with acute splenic sequestration crisis

Mohapatra S, Das PK, Rao PB, Nayak MK, Mane K, Sahoo B.

Emerg Radiol. 2024 Aug;31(4):613-617. doi: 10.1007/s10140-024-02246-w

Routine hemostasis profile in steady state sickle cell disease adult patients compared to laboratory values, between phenotypes and during pregnancy: A retrospective, descriptive study

Affo C, Schmidt C, Bosquet A, Dumont B, Mahé I.

eJHaem. Published online Aug 20, 2024. doi: 10.1002/jha2.876

Hospital acquired venous thromboembolism in children with sickle cell disease

Agarwal S, Foster KL, Anum SJ, Shapiro MC, Han H, *et al.*

Pediatr Hematol Oncol. 2024 Oct;41(7):459-469. doi: 10.1080/08880018.2024.2362147

Disease severity drives risk of venous thrombotic events in women with sickle cell disease in a single-center retrospective study

Light J, Abrams CM, Ilich A, Huang S, Zhu H, *et al.*

Res Pract Thromb Haemost. 2024 Jun 10;8(4):102471. doi: 10.1016/j.rpth.2024.102471

Influenza vaccine uptake among youth with sickle cell disease who are seen in clinic before and after the COVID-19 pandemic

Walden J, Stanek JR, Young J, Griffith MM, Nahata L, Creary SE.

Vaccine. 2024 Sep 17;42(22):126212. doi: 10.1016/j.vaccine.2024.126212

Clinical outcomes of COVID-19 in patients with sickle cell disease in French Guiana

Elenga N, Ntab B, Mafema Missindu J, Thomas Boizan N, Abassi A.

Heliyon. 2024 May 31;10(11):e32017. doi: 10.1016/j.heliyon.2024.e32017

Double Trouble: COVID-19 Infection Exacerbates Sickle Cell Crisis Outcomes in Hospitalized Patients- Insights from National Inpatient Sample 2020

Bodla ZH, Hashmi M, Niaz F, Auyeung AB, Oyetoran A, *et al.*

Hematol Rep. 2024 Jun 29;16(3):421-430. doi: 10.3390/hematolrep16030041

Seroprevalence of SARS-CoV-2 in pediatric hematology-oncology patients

Phan V, Richards T, Kang K, Sheridan M, Levorson R, *et al.*

Pediatr Blood Cancer. 2024 Oct;71(10):e31212. doi: 10.1002/pbc.31212

COVID-19 mRNA vaccination responses in individuals with sickle cell disease: an ASH RC Sickle Cell Research Network Study

Anderson AR, Strouse JJ, Manwani D, Brandow AM, Vichinsky E, *et al.*

Blood Adv. 2024 Sep 10;8(17):4549-4553. doi: 10.1182/bloodadvances

Attitudes, Beliefs, and Intention to Receive a COVID-19 Vaccine for Pediatric Patients With Sickle Cell Disease

Shook LM, Rosen BL, Mara CA, Mosley C, Thompson AA, *et al.*

J Pediatr Hematol Oncol. 2024 Jul 1;46(5):e305-e312. doi: 10.1097/MPH.0000000000002877

Outcome of pregnancy in sickle cell anemia patients with COVID-19 infection

Gowri V, Al Dughaisi T, Geetha D, Al Riyami M, Alburaidi R, Al Kindi S.

Asian J Transfus Sci. 2024 Jan-Jun;18(1):144-147. doi: 10.4103/ajts.ajts_164_21

Emergency department management of patients with sickle cell disease

Newman B, Wilkerson RG.

Emerg Med Pract. 2024 Aug 1;26(8):1-24.

Shift in emergency department utilization by frequent attendees with sickle cell disease during the COVID-19 pandemic: A multicentre cohort study

Rech JS, Cohen A, Bartolucci P, Santin A, Chantalat Auger C, *et al.*

Br J Haematol. 2024 Aug;205(2):463-472. doi: 10.1111/bjh.19556

Emergency department utilization before and during the COVID-19 pandemic among individuals with sickle cell disease

Attell BK, Plaxco AP, Zhou M, Valle J, Reeves SL, *et al.*

BMC Emerg Med. 2024 Jul 29;24(1):134. doi: 10.1186/s12873-024-01043-5

Emergency department 30-Day emergency department revisits among people with sickle cell disease: Variations in characteristics

Rushing M, Horiuchi S, Zhou M, Kavanagh PL, Reeves SL, *et al.*
Pediatr Blood Cancer. 2024 Oct;71(10):e31188. doi: 10.1002/pbc.31188

Gaps during pediatric to adult care transfer escalate acute resource utilization in sickle cell disease

Howell KE, Kayle M, Smeltzer MP, Nolan VG, Mathias JG, *et al.*
Blood Adv. 2024 Jul 23;8(14):3679-3685. doi: 10.1182/bloodadvances.2023011268

"Everyone screens to some extent": Barriers and facilitators of developmental screening among children with sickle cell disease: A mixed methods study

Hoyt CR, Luo L, Rice HE, Shivakumar N, Houston AJ, *et al.*
Pediatr Blood Cancer. 2024 Aug;71(8):e31060. doi: 10.1002/pbc.31060

A Mixed-Methods Evaluation of a Project ECHO Program for the Evidence-Based Management of Sickle Cell Disease

Mosley C, Farrell CB, Quinn CT, Shook LM.
Int J Environ Res Public Health. 2024 Apr 25;21(5):530. doi: 10.3390/ijerph21050530

Healthcare Professionals' Knowledge, Attitudes, and Practices in the Assessment, and Management of Sickle-Cell Disease: A Meta-Aggregative Review

Druye AA, Agyare DF, Akoto-Buabeng W, Zutah J, Offei FO, *et al.*
Diseases. 2024 Jul 14;12(7):156. doi: 10.3390/diseases12070156

Empowering sickle cell disease care: the rise of *TechnoRehabLab* in Sub-Saharan Africa for enhanced patient's perspectives

Boma PM, Ngoy SKK, Panda JM, Bonnechère B.
Front Rehabil Sci. 2024 Jun 27;5:1388855. doi: 10.3389/fresc.2024.1388855

Update on SickleInAfrica: a collaborative and multidimensional approach to conduct research and improve health

Nkya S, Masamu U, Kuona P, Kiguli S, Guindo A, *et al.*; SickleInAfrica.
Lancet Haematol. 2024 Aug;11(8):e565-e566. doi: 10.1016/S2352-3026(24)00219-9

Predictors of health-related quality of life in a large cohort of adult patients living with sickle cell disease in France: the DREPAntient study

Yaya I, Pourageaud A, Derbez B, Odièvre MH, Oudin Doglioni D, *et al.*; DREPAntient study group.
Front Public Health. 2024 May 20;12:1374805. doi: 10.3389/fpubh.2024.1374805

Exploring health-related quality of life, exercise and alcohol use in adolescents with sickle cell disease and healthy siblings

Constantinou C, Payne N, van den Akker O, Inusa B.
Psychol Health. 2024 Jun 26:1-21. doi: 10.1080/08870446.2024.2371018

Nothing about us without us: Advocacy and engagement in genetic medicine

Olayiwola O, Castillejo A, Louella M, Supercharger M, Harlow E, *et al.*
Sci Transl Med. 2024 May 8;16(746):eadn2401. doi: 10.1126/scitranslmed.adn2401

"Whatever is bad goes back to the woman": The gendered blame game of sickle cell disease in Malawi and Uganda

Svege S, Rujumba J, Robberstad B, Lange S.
Soc Sci Med. 2024 Aug;355:117119. doi: 10.1016/j.socscimed.2024.117119

Exercise and training in sickle cell disease: Safety, potential benefits, and recommendations

Connes P, Stauffer E, Liem RI, Nader E.
Am J Hematol. 2024 Aug 12. doi: 10.1002/ajh.27454

Understanding Exercise (in)tolerance in Sickle Cell Disease: Impacts of Hemolysis and Exercise Training on Skeletal Muscle Oxygen Delivery

Irwin DC, Calvo ETN, Belbis MD, Ehrenfort SKC, Noguer M, *et al.*
J Appl Physiol (1985). 2024 Aug 29. doi: 10.1152/jappphysiol.00390.2024

The highest-cost Medicaid enrollees with sickle cell disease had annual health care expenditures nearing \$200 000

Speller J, Rayel S, Hayashi K, Kirby M, Munevar D, *et al.*
Health Aff Sch. 2024 Mar 11;2(4):qxae029. doi: 10.1093/haschl/qxae029

Impacts of oxidative stress and anti-oxidants on the development, pathogenesis, and therapy of sickle cell disease: A comprehensive review

Pavitra E, Acharya RK, Gupta VK, Verma HK, Kang H, *et al.*
Biomed Pharmacother. 2024 Jul;176:116849. doi: 10.1016/j.biopha.2024.116849

Plasma monomeric ApoA1 and high-density lipoprotein bound ApoA1 are markedly decreased and associated with low levels of lipophilic antioxidants in sickle cell disease: A potential new pathway for therapy

Niesor EJ, Perez A, Rezzi S, Hodgson A, Canarelli S, *et al.*
Eur J Haematol. 2024 Aug 20. doi: 10.1111/ejh.14288

Heme-induced loss of renovascular endothelial protein C receptor promotes chronic kidney disease in sickle mice

Chen Q, Hazra R, Crosby D, Lenhart D, Lenhart SC, *et al.*
Blood. 2024 Aug 1;144(5):552-564. doi: 10.1182/blood.2023023528

Prospective study of complement activation and thromboinflammation within sickle cell disease and its complications

Varelas C, Vlachaki E, Klonizakis P, Pantelidou D, Minti F, *et al.*
Hemisphere. 2024 Jul 25;8(7):e135. doi: 10.1002/hem3.135

Microbiome in sickle cell disease: Pathophysiology and therapeutic insights

Gupta CL, Jaganathasamy N, Madkaikar M.
Br J Haematol. 2024 Aug 29. doi: 10.1111/bjh.19736

Endothelial dysfunction in Sickle Cell Disease: Strategies for the treatment

Pavan AR, Terroni B, Dos Santos JL.
Nitric Oxide. 2024 Aug 1;149:7-17. doi: 10.1016/j.niox.2024.05.003

Less Deformable Erythrocyte Subpopulations Biomechanically Induce Endothelial Inflammation in Sickle Cell Disease

Caruso C, Cheng X, Michaud ME, Szafraniec HM, Thomas BE, *et al.*
Blood. 2024 Aug 23;blood.2024024608. doi: 10.1182/blood.2024024608

Heme- and iron-activated macrophages in sickle cell disease: an updated perspective

Navaneethabalakrishnan S, An X, Vinchi F.
Curr Opin Hematol. 2024 Jul 17. doi: 10.1097/MOH.0000000000000836

Ferroptosis as an emerging target in sickle cell disease

Fortuna V, Lima J, Oliveira GF, Oliveira YS, Getachew B, *et al.*
Curr Res Toxicol. 2024 Jun 18;7:100181. doi: 10.1016/j.crtox.2024.100181

Iron scavenging and myeloid cell polarization

Ludwig N, Cucinelli S, Hametner S, Muckenthaler MU, Schirmer L.
Trends Immunol. 2024 Aug;45(8):625-638. doi: 10.1016/j.it.2024.06.006

Genetic variants associated with white blood cell count amongst individuals with sickle cell disease

Cinθο Ozahata M, Guo Y, Gomes I, Malta B, Belisário A, *et al.*; International Component of the NHLBI Recipient Epidemiology and Donor Evaluation Study (REDS-III) and for the TOPMed (NHLBI TransOmics for Precision Medicine) SCD working.

Br J Haematol. 2024 Sep 15. doi: 10.1111/bjh.19758

Impact of intravascular hemolysis on functional and molecular alterations in the urinary bladder: implications for an overactive bladder in sickle cell disease

Silveira THRE, Pereira DA, Pereira DA, Calmasini FB, Burnett AL, *et al.*

Front Physiol. 2024 Jul 19;15:1369120. doi: 10.3389/fphys.2024.1369120

Telomere Dynamics in Sickle Cell Anemia: Unraveling Molecular Aging and Disease Progression

Obeagu EI, Obeagu GU.

J Blood Med. 2024 Jul 26;15:313-323. doi: 10.2147/JBM.S462758

Informing parents of newborns with sickle cell trait detected at neonatal screening: A northern France experience

Freppel M, Mention K, Renom G, Lambilliotte A, Barbati M.

Pediatr Blood Cancer. 2024 Oct;71(10):e31174. doi: 10.1002/pbc.31174

Update on the practice of premarital screening for sickle cell traits in Africa: a systematic review and meta-analysis

Dilli PP, Obeagu E, Tamale A, Ajugwo A, Pius T, Makeri D.

BMC Public Health. 2024 May 31;24(1):1467. doi: 10.1186/s12889-024-19001-y

Ancestry-Independent Risk of Venous Thromboembolism in Individuals with Sickle Cell Trait vs. Factor V Leiden

Lin KH, Granka JM, Shastri AJ, Bonham VL, Naik RP.

Blood Adv. 2024 Sep 12;bloodadvances.2024014252. doi: 10.1182/bloodadvances.2024014252

Hypertonicity and/or acidosis induce marked rheological changes under hypoxic conditions in sickle trait red blood cells

Ellsworth P, Pawlinski IJ, Sielaty R, Ilich A, Prokopenko Y, *et al.*

Br J Haematol. 2024 Jul 25. doi: 10.1111/bjh.19669

Risk and Protective Factors for COVID-19 Infection among Pregnant Women with Sickle Cell Trait

Aldecoa KAT, Arsene C, Krishnamoorthy G, Chng T, Cherry G, *et al.*

Adv Hematol. 2024 Jun 12;2024:1595091. doi: 10.1155/2024/1595091

Pachychoroid neovascular membrane in a patient with sickle cell disease trait

Córdoba-Ortega CM, Arias Aristizabal JD, Suta Figueroa M, Pérez-Vergara V, Pulgarin DM.

Eur J Ophthalmol. 2024 Aug 28;11206721241278392. doi: 10.1177/11206721241278392

Polyglobulies

Erythrocytosis: Diagnosis and investigation

Noumani I, Harrison CN, McMullin MF.

Int J Lab Hematol. 2024 May;46 Suppl 1:55-62. doi: 10.1111/ijlh.14298

Olympic Games: When the haematocrit does not fit, the athlete is not always a cheat

Maaziz N, Martin L, Marchand A, Gardie B, Girodon F.

J Intern Med. 2024 Aug;296(2):213-214. doi: 10.1111/joim.13822

von Hippel- Lindau syndrome-related congenital polycythemia and response to belzutifan

Do Amaral PS, Mohan SR, Beckermann KE.

Haematologica. 2024 Aug 8. doi: 10.3324/haematol.2024.285724

Alpha-Thalassemia

Tamary H, Dgany O.

2005 Nov 1 [updated 2024 May 23]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2024

Disease burden, management strategies, and unmet needs in α -thalassemia due to hemoglobin H disease

Lal A, Viprakasit V, Vichinsky E, Lai Y, Lu MY, Kattamis A.

Am J Hematol. 2024 Jul 22. doi: 10.1002/ajh.27440

Global, regional, and national burden of thalassemia during 1990-2019: A systematic analysis of the Global Burden of Disease Study 2019

Wang R, Ma XH, Qin ZZ, Hu XX, Mo ZY, *et al.*

Pediatr Blood Cancer. 2024 Sep;71(9):e31177. doi: 10.1002/pbc.31177

Global, regional, and national burden of thalassemia, 1990-2021: a systematic analysis for the global burden of disease study 2021

Tuo Y, Li Y, Li Y, Ma J, Yang X, *et al.*

EClinicalMedicine. 2024 May 6;72:102619. doi: 10.1016/j.eclinm.2024.102619

Clinical burden and healthcare resource utilization associated with managing transfusion-dependent β -thalassemia in France

Baldwin J, Udeze C, Li N, Boulmerka L, Dahal L, *et al.*

Curr Med Res Opin. 2024 Aug;40(8):1289-1295. doi: 10.1080/03007995.2024.2368197

Anemia and iron overload as prognostic markers of outcomes in β -thalassemia

Musallam KM, Sheth S, Cappellini MD, Forni GL, Maggio A, Taher AT.

Expert Rev Hematol. 2024 Sep;17(9):631-642. doi: 10.1080/17474086.2024.2383420. Epub 2024 Jul 26. PMID: 39037857.

Evaluation of cardiac findings using speckle tracking in fetuses with hemoglobin Bart's disease

Anuwutnavin S, Russameecharoen K, Ruangvutilert P, Viboonchard S, Yaiyiam C, *et al.*

Ultrasound Obstet Gynecol. 2024 May 6. doi: 10.1002/uog.27676

Effects of thalassemia minor on umbilical artery blood flow and perinatal outcomes in pregnancy: A retrospective cohort study

Zhang L, Fan E, Chen X, Zhuo Y, Lee Y, Yan R.

Int J Gynaecol Obstet. 2024 Jul 27. doi: 10.1002/ijgo.15822

HbA2 levels in children with β -thalassemia trait associated with iron deficiency: A perspective for pediatricians

Değermenci Ş, Aslan D.

Am J Clin Pathol. 2024 Jul 23;aqae085. doi: 10.1093/ajcp/aqae085

A novel discriminant algorithm for differential diagnosis of mild to moderate thalassemia and iron deficiency anemia

Pan L, Li L, Qiu Y, Ling X, Wang C, *et al.*

Medicine (Baltimore). 2024 May 17;103(20):e38205. doi: 10.1097/MD.00000000000038205

Comprehensive analysis of thalassemia alleles (CATSA) based on third-generation sequencing is a comprehensive and accurate approach for neonatal thalassemia screening

Long J, Yu C, Sun L, Peng M, Song C, *et al.*

Clin Chim Acta. 2024 Jun 15;560:119749. doi: 10.1016/j.cca.2024.119749

A systematic review on thalassaemia screening and birth reduction initiatives: cost to success

Azrin Syahida AB, Nour El Huda AR, Safurah J.
Med J Malaysia. 2024 May;79(3):348-359.

Development of pre-implantation genetic testing protocol for monogenic disorders (PGT-M) of Hb H disease

Somboonchai P, Charoenkwan P, Piyamongkol S, Lattiwongsakorn W, Pantasri T, Piyamongkol W.
BMC Genomics. 2024 Jul 3;25(1):668. doi: 10.1186/s12864-024-10578-7

Loss-of-Function Variants in *SUPT5H* as Modifying Factors in Beta-Thalassemia

Harteveld CL, Achour A, Fairuz Mohd Hasan NF, Legebeke J, Arkesteijn SJG, *et al.*; International Hemoglobinopathy Research Network (INHERENT).
Int J Mol Sci. 2024 Aug 16;25(16):8928. doi: 10.3390/ijms25168928

Moderate-severe beta-thalassemia intermedia phenotype caused by sextuplicated alpha-globin gene allele in two beta-thalassemia carriers

Achour A, Knijnenburg J, Koopmann T, Raz A, Tischkowitz M, *et al.*
Am J Hematol. 2024 Aug;99(8):1655-1658. doi: 10.1002/ajh.27386

Beta-thalassemia intermedia due to a complex alpha-globin rearrangement and a heterozygous beta thalassemia mutation

Marin V, Huguenin Y, Bessi L, Weinmann L, Augis V, *et al.*
Br J Haematol. 2024 Aug 18. doi: 10.1111/bjh.19715

α -Globin mutations and Genetic Variants in γ -globin Promoters are Associated with Unelevated Hemoglobin F Expression of Atypical β^0 -thalassemia/HbE

Satthakarn S, Panyasai S.
Arch Med Res. 2024 Sep;55(6):103055. doi: 10.1016/j.arcmed.2024.103055

Prenatal Counseling on the Alpha Thalassemia Allele HBA2:c.*94A>G

Alderei L, Alshkeili N, Alnaqbi D, Shehab OA, Vijayan R, Soud AK.
Thalass Rep. 2024; 14(2):44-48. <https://doi.org/10.3390/thalassrep14020006>

Hb H Disease Caused by Uniparental Disomy: First Report of the $\alpha^{T-Saudi}\alpha$ Mutation in the Chinese Population

Wang G, Xie H, Zhang J, Huang P, Liang M, *et al.*
Mediterr J Hematol Infect Dis. 2024 Sep 1;16(1):e2024069. doi: 10.4084/MJHID.2024.069

Clinical characteristics, laboratory features and genetic profile of hemoglobin E (HBB:c.79 G > A)/ β (nucleotide -28 A > G) (HBB:c.-78 A > G) -thalassemia subjects identified from community- and hospital-recruited cohorts

Chumnumsiriwath P, Charoenporn P, Jermnim S, Suannum P, Samaisombat M, *et al.*
Blood Cells Mol Dis. 2024 Nov;109:102883. doi: 10.1016/j.bcmed.2024.102883

Causes of Death and Mortality Trends in Individuals with Thalassemia in the United States, 1999-2020

Tan JY, Yeo YH, Chan KH, Shaaban HS, Guron G.
J Blood Med. 2024 Aug 5;15:331-339. doi: 10.2147/JBM.S470177

Left and right atrioventricular coupling index in patients with beta-thalassemia major

Meloni A, Saba L, Positano V, Pistoia L, Spasiano A, *et al.*
Int J Cardiovasc Imaging. 2024 Aug;40(8):1631-1640. doi: 10.1007/s10554-024-03146-3

Magnetic Resonance Evaluation of Tissue Iron Deposition and Cardiac Function in Adult Regularly Transfused Thalassemia Intermedia Compared with Thalassemia Major Patients

Meloni A, Pistoia L, Ricchi P, Longo F, Cecinati V, *et al.*
J Clin Med. 2024 Aug 14;13(16):4791. doi: 10.3390/jcm13164791

Global longitudinal strain by cardiac magnetic resonance is associated with cardiac iron and complications in beta-thalassemia major patients

Meloni A, Saba L, Positano V, Pistoia L, Campanella A, *et al.*
Int J Cardiol. 2024 Oct 15;413:132319. doi: 10.1016/j.ijcard.2024.132319

Association between Epicardial Adipose Tissue and Atrial Fibrillation in Patients with Transfusion-Dependent β -Thalassemia

Malagù M, Tonet E, Orazio G, Longo F, De Raffe M, *et al.*
J Clin Med. 2024 Jun 14;13(12):3471. doi: 10.3390/jcm13123471

Global and regional cardiac magnetic resonance feature tracking left ventricular strain analysis in assessing early myocardial disease in β thalassemia major patients

Batouty NM, Tawfik AM, Sobh DM, Gadelhak BN, El-Ashwah S, *et al.*
J Cardiovasc Imaging. 2024 Aug 3;32(1):18. doi: 10.1186/s44348-024-00026-1

Soluble Fms-like tyrosine kinase-1 as an endothelial dysfunction biomarker associated with pulmonary hypertension in adult patients with beta-thalassemia major

Abozeid SF, Elkholy RA, Elbedewy TA, Seliem MF.
J Investig Med. 2024 Aug 19:10815589241270616. doi: 10.1177/10815589241270616

The relationship between liver stiffness by two-dimensional shear wave elastography and iron overload status in transfusion-dependent patients

Puttawibul P, Kritsaneepaiboon S, Chotsampancharoen T, Vichitkunakorn P.
Pediatr Hematol Oncol. 2024 Sep;41(6):409-421. doi: 10.1080/08880018.2024.2353900

Brain perfusion changes in beta-thalassemia

Manara R, Ponticorvo S, Contieri M, Canna A, Russo AG, *et al.*
Orphanet J Rare Dis. 2024 May 21;19(1):212. doi: 10.1186/s13023-024-03194-x

Evaluation of microstructural changes in the brain in transfusion dependent thalassemia patients with advanced magnetic resonance imaging techniques

Genç B, Aslan K, Atay MH, Akan H.
Neuroradiology. 2024 Jul 8. doi: 10.1007/s00234-024-03414-y

Liver disease in patients with transfusion-dependent β -thalassemia: The emerging role of metabolism dysfunction-associated steatotic liver disease

Fragkou N, Vlachaki E, Goulis I, Sinakos E.
World J Hepatol. 2024 May 27;16(5):671-677. doi: 10.4254/wjh.v16.i5.671

Bone mineral density in adult thalassaemias: a retrospective longitudinal study

Algodayan S, Balachandar R, Papathanasiou N, Bomanji J, Porter JB, Waung J.
Nucl Med Commun. 2024 Aug 1;45(8):658-665. doi: 10.1097/MNM.0000000000001864

Prevalence and risk factors predisposing low bone mineral density in patients with thalassemia

Ananvutisombat N, Tantiworawit A, Punnachet T, Hantrakun N, Piriyaikhuntorn P, *et al.*
Front Endocrinol (Lausanne). 2024 Jun 24;15:1393865. doi: 10.3389/fendo.2024.1393865

Appropriateness of the EQ-5D-5L in capturing health-related quality of life in individuals with transfusion-dependent β -thalassemia: a mixed methods study

Boateng-Kuffour A, Skrobanski H, Drahos J, Kohli P, Forster K, *et al.*
Health Qual Life Outcomes. 2024 Jul 11;22(1):54. doi: 10.1186/s12955-024-02265-8

Quality of Life in Transfusion-Dependent Thalassemia Patients in Greece Before and During the COVID-19 Pandemic

Klonizakis P, Klaassen RJ, Roy N, Papatsouma I, Mainou M, *et al.*
Value Health Reg Issues. 2024 Jul;42:100986. doi: 10.1016/j.vhri.2024.100986

Iron chelation therapy for children with transfusion-dependent β -thalassemia: How young is too young?

Forni GL, Kattamis A, Kuo KHM, Maggio A, Sheth S, *et al.*
Pediatr Blood Cancer. 2024 Aug;71(8):e31035. doi: 10.1002/pbc.31035

Differential effects of iron chelators on iron burden and long-term morbidity and mortality outcomes in a large cohort of transfusion-dependent β -thalassemia patients who remained on the same monotherapy over 10 years

Musallam KM, Barella S, Origa R, Ferrero GB, Lisi R, et al; Webthal[®] project.
Blood Cells Mol Dis. 2024 Jul;107:102859. doi: 10.1016/j.bcmd.2024.102859

Skin complications during iron chelation therapy for beta-thalassemia: overview and treatment approach

Saeidnia M, Shadfar F, Sharifi S, Babashahi M, Ghaderi A, Shokri M.
Int J Hematol. 2024 Sep;120(3):271-277. doi: 10.1007/s12185-024-03825-w

In Vivo Silencing of Intestinal DMT1 Mitigates Iron Loading in β -thalassemia Intermedia (Hbbth3/+) Mice

Yu Y, Woloshun RR, Lee JK, Ebea-Ugwuanyi P, Shine JS, et al.
Blood Adv. 2024 Sep 9;bloodadvances.2024013333. doi: 10.1182/bloodadvances.2024013333

Manuka combinations with nigella sativa and hydroxyurea in treating iron overload of pediatric β -thalassemia major, randomized clinical trial

Gamaleldin MM, Abraham IL, Meabed MH, Elberry AA, Abdelhalim SM, et al.
Heliyon. 2024 Jun 26;10(13):e33707. doi: 10.1016/j.heliyon.2024.e33707

Real-world efficacy and safety of luspatercept and predictive factors of response in patients with transfusion-dependent β -thalassemia

Panzieri DL, Consonni D, Scaramellini N, Ausenda G, Granata F, et al I.
Am J Hematol. 2024 Sep 12. doi: 10.1002/ajh.27474

Luspatercept for non-deletional hemoglobin H disease

Yang YN, Chou YY, Cheng CN.
Pediatr Blood Cancer. 2024 Oct;71(10):e31224. doi: 10.1002/pbc.31224

Luspatercept's use in a patient with transfusion-dependent beta-thalassemia and intrathoracic extramedullary hematopoiesis (EMH)

Seijari MN, Alshurafa A, Yassin MA.
Clin Case Rep. 2024 May 10;12(5):e8795. doi: 10.1002/ccr3.8795

Clinical efficacy of thalidomide for various genotypes of beta thalassemia

Yang WJ, Kang QP, Zhou Q, Lin T, Gong XM, et al.
BMC Med Genomics. 2024 Jul 18;17(1):191. doi: 10.1186/s12920-024-01963-y

Long-Term Follow-Up of Patients Undergoing Thalidomide Therapy for Transfusion-Dependent β -Thalassaemia: A Single-Center Experience

Zhu W, He Y, Huang M, Fu S, Liu Z, et al.
Int J Gen Med. 2024 Apr 30;17:1729-1738. doi: 10.2147/IJGM.S462991

Thalidomide and Hydroxyurea in Transfusion-Dependent Thalassemia: Efficacy, Safety Profile and Impact on Quality of Life

Bhattacharjee S, Ghosh S, Shaw J, Bhattacharjee S, Bhattacharyya M.
Hemoglobin. 2024 Aug 2:1-8. doi: 10.1080/03630269.2024.2386076

Safety and efficacy of human apotransferrin infusion in patients with β -thalassemia intermedia: the AIM study

Konté K, Swinkels DW, Budde IK, Nur E, Biemond BJ.
Haematologica. 2024 Aug 22. doi: 10.3324/haematol.2024.285045

The Effects and Safety of Silymarin on β -thalassemia in Children and Adolescents: A Systematic Review based on Clinical Trial Studies

Rahimi-Dehkordi N, Heidari-Soureshjani S, Mt Sherwin C.
Rev Recent Clin Trials. 2024 May 30. doi: 10.2174/0115748871305325240511122602

Increased Expression of α -Hemoglobin Stabilizing Protein (AHSP) mRNA in Erythroid Precursor Cells Isolated from β -Thalassemia Patients Treated with Sirolimus (Rapamycin)

Zurlo M, Zuccato C, Cosenza LC, Gamberini MR, Finotti A, Gambari R.
J Clin Med. 2024 Apr 24;13(9):2479. doi: 10.3390/jcm13092479

Mitoxantrone ameliorates ineffective erythropoiesis in a β -thalassemia intermedia mouse model

Zhang H, Liu R, Fang Z, Nie L, Ma Y, *et al.*
Blood Adv. 2024 Aug 13;8(15):4017-4024. doi: 10.1182/bloodadvances.2024012679

Therapeutic Relevance of Inducing Autophagy in β -Thalassemia

Gambari R, Finotti A.
Cells. 2024 May 25;13(11):918. doi: 10.3390/cells13110918

Nutritional and Body Composition Changes in Paediatric β -Thalassemia Patients Undergoing Hematopoietic Stem Cell Transplantation: A Retrospective Study Using Bioelectrical Impedance Analysis

Zhang L, Wang L, Long J, Yin Y, Patil S.
J Multidiscip Healthc. 2024 May 9;17:2203-2214. doi: 10.2147/JMDH.S463796

Development of a nomogram to predict the risk of secondary failure of platelet recovery in patients with β -thalassemia major after hematopoietic stem cell transplantation: a retrospective study

Xie Y, Yang G, Pan L, Gan Z, Huang Y, *et al.*
Ther Adv Hematol. 2024 May 9;15:20406207241245190. doi: 10.1177/20406207241245190

Casegy: Innovative Medicinal Products Require Innovative Approaches to Regulatory Assessment

Kerwash E, Johnston JD.
Pharmaceutics. 2024 Jul 6;16(7):906. doi: 10.3390/pharmaceutics16070906

Modified lentiviral globin gene therapy for pediatric β^0/β^0 transfusion-dependent β -thalassemia: A single-center, single-arm pilot trial

Li S, Ling S, Wang D, Wang X, Hao F, *et al.*
Cell Stem Cell. 2024 Jul 5;31(7):961-973.e8. doi: 10.1016/j.stem.2024.04.021

Precise correction of a spectrum of β -thalassemia mutations in coding and non-coding regions by base editors

Prasad K, Devaraju N, George A, Ravi NS, Paul J, *et al.*
Mol Ther Nucleic Acids. 2024 May 3;35(2):102205. doi: 10.1016/j.omtn.2024.102205

Use of HSC Targeted LNP to Generate a Mouse Model of Lethal α -Thalassemia and Treatment via Lentiviral Gene Therapy

Chappell ME, Breda L, Tricoli L, Guerra A, Jarocha DJ, *et al.*
Blood. 2024 Jul 1;blood.2023023349. doi: 10.1182/blood.2023023349

Exploring the bone marrow micro environment in thalassemia patients: potential therapeutic alternatives

Li Z, Yao X, Zhang J, Yang J, Ni J, Wang Y.
Front Immunol. 2024 Aug 5;15:1403458. doi: 10.3389/fimmu.2024.1403458

Hémoglobinopathies – Autres maladies du globule rouge

Novel therapeutic approaches in thalassemias, sickle cell disease, and other red cell disorders

Pinto VM, Mazzi F, De Franceschi L.
Blood. 2024 Aug 22;144(8):853-866. doi: 10.1182/blood.2023022193

Understanding Rare Anemias: Emerging Frontiers for Diagnosis and Treatment

Vives Corrons JL.

J Clin Med. 2024 May 29;13(11):3180. doi: 10.3390/jcm13113180

Management of iron overload: lessons from transfusion-dependent hemoglobinopathies

Coates TD.

Blood. 2024 Sep 18;blood.2023022502. doi: 10.1182/blood.2023022502

Haemoglobinopathies and other rare anemias in Spain: ten years of a nationwide registry (REHem-AR)

Marco Sánchez JM, Bardón Cancho EJ, Benítez D, Payán-Pernía S, Collado Gimbert A, *et al.*

Ann Hematol. 2024 Aug;103(8):2743-2755. doi: 10.1007/s00277-024-05788-8

Catalase, Glutathione Peroxidase, and Peroxiredoxin 2 in Erythrocyte Cytosol and Membrane in Hereditary Spherocytosis, Sickle Cell Disease, and β -Thalassemia

Melo D, Ferreira F, Teles MJ, Porto G, Coimbra S, *et al.*

Antioxidants (Basel). 2024 May 22;13(6):629. doi: 10.3390/antiox13060629

Current Status of Newborn Bloodspot Screening Worldwide 2024: A Comprehensive Review of Recent Activities (2020-2023)

Therrell BL, Padilla CD, Borrajo GJC, Khneisser I, Schielen PCJI, *et al.*

Int J Neonatal Screen. 2024 May 23;10(2):38. doi: 10.3390/ijns10020038

Prevention and management of indwelling catheter-related thrombosis in sickle cell disease and thalassaemia: A British Society for Haematology Good Practice Paper

Woodward G, Drasar E, Pancham S, Sadasivam N, Thachil J, Brewin J; BSH Committee.

Br J Haematol. 2024 Jul 31. doi: 10.1111/bjh.19650

Hematopoietic Stem Cell Transplantation in Children with Sickle Cell Disease and Thalassemia Major: A National Database Study

De Avila C, Martinez PA, Sendi P, Galvez Silva JR, Maher OM, Totapally BR.

Pediatr Hematol Oncol. 2024 Oct;41(7):489-503. doi: 10.1080/08880018.2024.2378282

COVID-19 in patients affected by red blood cell disorders, results from the European registry ERN-EuroBloodNet

Velasco Puyó P, Christou S, Campisi S, Rodríguez-Sánchez MA, Reidel S, *et al.*

Authorea. August 25, 2024. doi: 10.22541/au.172462030.03839690/v1

Exploring the Interplay between Asthma and Hemoglobinopathies: A Comprehensive Review

Indolfi C, Dinardo G, Grella C, Klain A, Perrotta A, *et al.*

J Clin Med. 2024 May 31;13(11):3263. doi: 10.3390/jcm13113263

Circulating free heme induces cytokine storm and pulmonary hypertension through the MKK3/p38 axis

Valuparampil Varghese M, James J, Bharti D, Rischard F, Rafikova O, Rafikov R.

Am J Physiol Lung Cell Mol Physiol. 2024 Aug 28. doi: 10.1152/ajplung.00422.2022

Post-GWAS Validation of Target Genes Associated with HbF and HbA₂ Levels

Caria CA, Faà V, Porcu S, Marongiu MF, Poddie D, *et al.*

Cells. 2024 Jul 12;13(14):1185. doi: 10.3390/cells13141185

Advances in Hemoglobinopathies and Thalassemia Evaluation

Agarwal AM, Rets AV.

Clin Lab Med. 2024 Sep;44(3):441-453. doi: 10.1016/j.cl.2024.04.006

International Society for Cell & Gene Therapy Stem Cell Engineering Committee report on the current state of hematopoietic stem and progenitor cell-based genomic therapies and the challenges faced

Gupta AO, Azul M, Bhoopalan SV, Abraham A, Bertaina A, *et al.*

Cytotherapy 2024 Jun 6:S1465-3249(24)00735-7. doi: 10.1016/j.jcyt.2024.06.002

Regulatory Assessment of Casgevy for the Treatment of Transfusion-Dependent β -Thalassemia and Sickle Cell Disease with Recurrent Vaso-Occlusive Crises

Kerwash E, Sajic M, Rantell KR, McBlane JW, Johnston JD, *et al.*

Curr Issues Mol Biol. 2024 Jul 30;46(8):8209-8225. doi: 10.3390/cimb46080485

ACT To Sustain: Adoptive Cell Therapy To Sustain Access to Non-Commercialized Genetically Modified Cell Therapies

Gardner RA, White C, Elsallab M, Farnia S, Frait E, *et al.*

Transplant Cell Ther. 2024 Aug;30(8):776-787. doi: 10.1016/j.jtct.2024.05.010

A roadmap for affordable genetic medicines

Kliegman M, Zaghlula M, Abrahamson S, Esensten JH, Wilson R, *et al.*

Nature. 2024 Jul 17. doi: 10.1038/s41586-024-07800-7

A Review of Gene Therapies for Hemoglobinopathies

Jones-Wonni B, Kelkar AH, Achebe MO.

Hemoglobin. 2024 Aug 15:1-12. doi: 10.1080/03630269.2024.2369534

Therapeutic Gene Editing for Hemoglobinopathies

Testa U, Leone G, Cappellini MD.

Mediterr J Hematol Infect Dis. 2024 Sep 1;16(1):e2024068. doi: 10.4084/MJHID.2024.068

Machine Learning-Based Prediction of Hemoglobinopathies Using Complete Blood Count Data

Schipper A, Rutten M, van Gammeren A, Harteveld CL, Urrechaga E, *et al.*

Clin Chem. 2024 Aug 1;70(8):1064-1075. doi: 10.1093/clinchem/hvae081

Optimisation of the Danish national haemoglobinopathy screening programme – A prospective intervention study

Gravholt EAE, Jørgensen FS, Holm C, Petersen J, Nardo-Marino A, *et al.*

eJHaem. Published online Aug 08, 2024. doi: 10.1002/jha2.980

Ovarian Tissue Cryopreservation for Fertility Preservation in Patients with Hemoglobin Disorders: A Comprehensive Review

Haering C, Coyne K, Daunov K, Anim S, Christianson MS, Flyckt R.

J Clin Med. 2024 Jun 21;13(13):3631. doi: 10.3390/jcm13133631

The Characteristics of Compound Heterozygosity for Hemoglobin G-Makassar with Hb E in Malaysia

Hamzah R, Mohamad AS, Mohd Yasin N, Esa E, Chen G, Selvaratnam V.

J Blood Med. 2024 May 29;15:255-264. doi: 10.2147/JBM.S432849

Modulation of the allosteric and vasoregulatory arms of erythrocytic oxygen transport

Wise TJ, Ott ME, Joseph MS, Welsby IJ, Darrow CC, McMahon TJ.

Front Physiol. 2024 Jun 10;15:1394650. doi: 10.3389/fphys.2024.1394650

A moonlighting job for α -globin in blood vessels

Abbineni PS, Baid S, Weiss MJ.

Blood. 2024 Aug 22;144(8):834-844. doi: 10.1182/blood.2023022192

Designing a single-arm phase 2 clinical trial of mitapivat for adult patients with erythrocyte membranopathies (SATISFY): a framework for interventional trials in rare anaemias - pilot study protocol

Glenthøj A, van Beers EJ, van Wijk R, Rab MAE, Groot E, *et al.*

BMJ Open. 2024 Jul 30;14(7):e083691. doi: 10.1136/bmjopen-2023-083691

Erythroid-intrinsic activation of TLR8 impairs erythropoiesis in inherited anemia

Liang J, Wan Y, Gao J, Zheng L, Wang J, *et al.*

Nat Commun. 2024 Jul 6;15(1):5678. doi: 10.1038/s41467-024-50066-w

DAHEAN: A Danish nationwide study ensuring quality assurance through real-world data for suspected hereditary anemia patients

Glenthøj A, Rasmussen AØ, Bendtsen SK, Hasle H, Hoffmann M, *et al.*
Orphanet J Rare Dis. 2024 Jul 31;19(1):284. doi: 10.1186/s13023-024-03298-4

Impact of Iron Overload on Incidence of Diabetes Mellitus, Cardiac Disease, and Death in Congenital Hemolytic Anemias

Cheng A, Al-Samkari H.

Blood Adv. 2024 Aug 27;bloodadvances.2024013666. doi: 10.1182/bloodadvances.2024013666

Longitudinal analysis of clinical and laboratory biomarkers in a patient with familial lecithin: cholesterol acyltransferase deficiency (FLD) and accelerated eGFR decline: A case study

Alfaro G, Pendyala J, Sulewski M, Miller M, Vitali C, Cuchel M.

J Clin Lipidol. 2024 Jul-Aug;18(4):e636-e643. doi: 10.1016/j.jacl.2024.03.002

Rescue of Familial Lecithin:Cholesterol Acyltransferase Deficiency Mutations with an Allosteric Activator

Manthei KA, Tremonti GE, Chang L, Niemelä A, Giorgi L, *et al.*

Mol Pharmacol. 2024 Sep 17;106(4):188-197. doi: 10.1124/molpharm.124.000932

Novel transferrin gene mutations in four new cases of congenital Atransferrinaemia: Functional and diagnostic pathogenicity despite negative bioinformatics

Romero-Cortadellas L, Ferrer-Cortès X, Calvo-López L, Olivella M, Barqué A, *et al.*

Br J Haematol. 2024 Jul 31. doi: 10.1111/bjh.19675

Histology, Howell Jolly Bodies

Fakoya AO, Amraei R.

2024 Jul 27. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan–

Les numéros précédents du Bulletin Recherche sont disponibles sur la page :
<https://filiere-mcgre.fr/le-bulletin-recherche/>

Filière de santé maladies rares MCGRE - Hôpital Henri Mondor
1 rue Gustave Eiffel, 94000 Créteil
contact@filiere-mcgre.fr - www.filiere-mcgre.fr