Bulletin recherche

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Le point sur ...

Des études in silico et fonctionnelles permettent d'étudier le caractère pathogène des mutations génétiques de EPAS1/HIF2A identifiées chez des patients atteints de polyglobulie.

Contexte et objectifs

Les polyglobulies (ou érythrocytoses) se caractérisent par une production excessive de globules rouges. Des mutations générant un gain de fonction du gène *EPAS1/HIF2A* ont été associées au développement de polyglobulies héréditaires, mais également au développement de tumeurs cancéreuses (paragangliomes, phéochromocytome et somatostatinomes).

Le gène *EPAS1/HIF2A* code le facteur de transcription HIF2A (Hypoxia Inducible Factor-2 alpha), impliqué dans la régulation de la voie de l'hypoxie. En condition d'hypoxie, les sous-unités HIF-α s'associent à la sous-unité HIF-1β. Sous l'action de co-activateurs, l'hétérodimère HIF α/β se lie aux éléments de réponse à l'hypoxie (HRE) dans l'ADN et active l'expression des gènes cibles de HIF comme le gène de l'EPO (érythropoiétine) qui va stimuler la prolifération et la différentiation des progéniteurs érythroïdes aboutissant à la production de globules rouges. HIF induit également l'expression de nombreux gènes impliqués dans la tumorigenèse et l'activation de la voie de l'hypoxie est la signature de nombreuses tumeurs.

L'objectif de cette étude était de déterminer l'implication des mutations génétiques gain de fonction de *EPAS1/HIF2A* dans la pathogenèse de la polyglobulie et leur potentielle contribution dans le développement de tumeurs.

Méthode

Après obtention de l'accord éthique et du consentement de chaque participant, un prélèvement sanguin a été réalisé et l'ADN a été extrait pour être séquencé par Next Generation Sequencing (NGS) chez 1450 patients ne présentant pas de causes évidentes de polyglobulie, issus de 7 centres de diagnostic (dont 4 européens). Le NGS a révélé la présence de 33 mutations faux-sens chez 43 patients dont 41 patients avec une mutation hétérozygote germinale et 2 patients avec des mutations mosaïques (retrouvées en faible quantité) et 28 proches, constituant ainsi la plus grande collection de mutation *EPAS1/HIF2A* étudiée à ce jour. Les données cliniques et les antécédents familiaux de ces patients ont été étudiés.

Pour déterminer le caractère pathogène de ces mutations, des analyses in silico ont été réalisées sur les mutations génétiques présentes à une fréquence inférieure à 5.10-4 sur le gène *EPAS1* à l'aide du serveur web MetaDome afin de déterminer la tolérance des mutations à chaque position de la protéine HIF-2α par des modélisations informatiques. L'attribution du caractère pathogène de chaque mutation a été permise par la plateforme Mobidetail et l'outil de prédiction intégrative PROVEAN. Des analyses fonctionnelles ont été menées pour mesurer avec précision le gain de fonction des nouveaux variants de la protéine HIF-2α. Pour ce faire, un nouveau vecteur rapporteur contenant un fragment du promoteur de l'EPO plus sensible à HIF-2α a été construit et utilisé pour mesurer son expression en temps réel en fonction des mutants HIF-2α utilisés. La classification finale des mutations a reposé sur les analyses globales *in silico* et fonctionnelles selon les critères de ACMG (American College of Medical Genetics and Genomics) : classe 1 : bénine, classe 2 : plutôt bénine, classe 3 : mutation de signification inconnue, classe 4 : plutôt pathogène, classe 5 : pathogène.

Résultats

Les études *in silico* et fonctionnelles ont permis d'identifier 11 mutations pathogènes chez 17 patients et 23 proches. Parmi ces 11 mutations, quatre nouvelles mutations (D525G, L526F, G527K, A530S) proches de la proline clé P531 ont été identifiées ce qui élargit le spectre des mutations impliquées dans la polyglobulie. Une jeune patiente porteuse de mutation pathogène à l'état mosaïque a développé de multiples paragangliomes. Cependant les patients porteurs de mutations pathogènes présentent rarement des complications associées ce qui souligne la complexité de la corrélation entre le génotype et le phénotype de cette pathologie.

Cette étude a démontré l'importance de combiner des analyses fonctionnelles et *in silico* pour améliorer le diagnostic et le suivi des patients atteints de polyglobulie porteurs d'une mutation sur *EPAS1* dont le caractère pathogène n'a pas été établi.

L'étude en quelques chiffres :

L'ADN de 1450 patients atteints de polyglobulie a été séquencé sur un panel de gènes candidats

33 mutations génétiques de *EPAS1* ont été identifiées chez 43 patients dont 2 porteurs de mutations mosaïques et 28 apparentés

11 mutations pathogènes identifiées chez 40 individus (17 patients et 23 proches)

4 nouvelles mutations pathogènes découvertes

Cette étude a fait l'objet d'une publication en janvier 2023 dans le journal *Haematologica* (https://doi.org/10.3324/haematol.2022.281698)

Échange avec ...

Betty Gardie

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Section : Sciences de la Vie et de la Terre

Thématiques : Génétique médicale

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

Cette étude a été mise en place en collaboration avec le Pr Girodon, hématologue au CHU de Dijon qui fédère depuis 2016 les hématologues de France pour travailler sur la polyglobulie. Comme c'est une pathologie rare, les centres travaillant séparément sur la polyglobulie découvrent souvent des mutations rares avec des fréquences qui ne sont toutefois pas nulles dans la population. Cela aboutit alors à leur classification en variants de signification inconnue. Ceci est dû à la grande difficulté de déterminer si un variant génétique est un polymorphisme rare ou s'il est en cause dans la pathologie. C'est dans ce but que j'ai développé la partie recherche de ce projet au travers d'études fonctionnelles permettant de distinguer les mutations génétiques pathogènes des polymorphismes rares. Le Pr Girodon est également le coordinateur d'un réseau européen sur les érythrocytoses (European Congenital Erythrocytosis Consortium, ECEC) qui nous a permis de rassembler des échantillons issus de quatre centres de référence en Europe travaillant sur cette pathologie. Nous avons ainsi pu regrouper plusieurs cas issus de pays différents et nous assurer de la non-apparentée entre les patients. Le rassemblement de ces cas nous a permis d'identifier plusieurs familles avec le même variant rare et le même phénotype ce qui a constitué un argument fort pour établir la pathogénicité de ces mutations.

→ Pourquoi avoir utilisé plusieurs outils de classification dans vos analyses *in silico* ?

Dans cette étude, les analyses sont issues de sept laboratoires qui n'ont pas utilisé les mêmes outils, ce qui a été l'occasion de les comparer. Lorsque deux outils *in silico* ont donné le même résultat, cela a constitué un argument supplémentaire pour classer le variant en tant que pathogène.

→ Comment expliquez-vous que certaines mutations classées comme pathogènes d'après les études in *silico* (comme E538K et F450L) n'engendrent finalement pas de complications telles que le développement de tumeurs?

Nous n'avons identifié aucun patient polyglobulique porteur d'une mutation germinale de HIF-2α et présentant des tumeurs. Les mutations de HIF2A impliquées dans les tumeurs sont uniquement retrouvées à l'état somatique, ou mosaïque chez les patients atteints du syndrome polyglobulie/paragangliome/pheochromocytome/somatostatinome (aussi appelé syndrome Zhuang/Pacak). L'hypothèse est que les mutations associées aux tumeurs sont trop sévères pour être viables. Dans beaucoup de cas de tumeurs par exemple, la Proline P531 est mutée. Cet acide aminé joue un rôle central dans la stabilité de HIF-2α , ce qui ne serait pas viable à l'état germinal. Cependant, la question se pose pour les acides aminés situés autour de la Proline P531 comme la Tyrosine 532 qui est présente dans les tumeurs et qui se retrouve à l'état germinal chez des patients polyglobuliques qui devront bénéficier d'une surveillance accrue. Cette hypothèse est valable pour tous les gènes de la voie l'hypoxie (EPAS1, VHL, PHD2): plus l'altération est forte, plus le risque de cancer est élevé. Il existe une sorte de continuum de la voie de l'activation de l'hypoxie au pied duquel se situent les polyglobulies.

C'est pour cela qu'il est très difficile de classer les variants associés aux polyglobulies qui présentent souvent une très faible dérégulation (mutants hypomophes). A l'autre bout de ce continuum se trouvent les mutations très sévères qui donnent des tumeurs. Le problème se pose donc pour les mutations situées entre les deux. Les outils in silico et fonctionnels sont très importants pour déterminer le caractère pathogène de ces variants. Pour les deux variants F540L et E538K, nous avons eu du mal à mettre en évidence le gain de fonction. Le risque de développer des tumeurs chez ces patients est donc très faible. Une surveillance est malgré tout fortement recommandée car ce sont des cas très rares.

→ Les patients porteurs de mutations mosaïques peuvent être sous-diagnostiqués, comment améliorer la détection de ces mutations ?

C'est la première fois que des taux de mosaïcisme aussi bas ont été détectés. Les protocoles d'analyses des variants par NGS ne prennent pas du tout en compte ces variants. Les seuils dépendent des laboratoires. Dans notre étude, ces variants étaient inférieur à 2 % ce qui est quasiment au même niveau que le reste du bruit de fond donc ces variants n'ont pas été sélectionnés par le logiciel. En présence d'un phénotype très particulier comme des enfants jeunes qui ont un taux d'EPO très élevé, nous recommandons de procéder à une analyse manuelle du séquençage NGS pour vérifier qu'il n'y a pas une mutation en mosaïque dans le gène. Il faut ensuite effectuer une PCR Digitale (droplet PCR) spécifique de la mutation, qui pourra confirmer sa présence.

→ Pourriez-vous nous en dire un peu plus sur le potentiel traitement contre la polyglobulie liée à l'hypoxie, qui permettrait de cibler et inhiber spécifiquement la protéine HIF-2α ?

Il existe une nouvelle molécule (Belzutifan, Welireg®) qui a été validée dans le traitement de cancers du rein chez des patients porteurs de mutations VHL. Les mutations de ce gène sont associées à la maladie de von Hippel-Lindau caractérisée par le développement de cancers du rein et de phéochromocytomes héréditaires. Dans ces tumeurs, HIF-2α est surexprimée suite à la perte de fonction de VHL qui joue un rôle majeur dans sa dégradation. Chez les patients avec cancer du rein qui sont traités, le seul effet secondaire est l'anémie. Ce traitement pourrait donc être utilisé dans le traitement des polyglobulies liées à une stabilisation de HIF-2α. Ce traitement a en effet été testé et validé dans un modèle murin de la polyglobulie de Chuvash qui est due à une mutation homozygote du gène VHL (mutation hypomorphe qui prédispose aux érythrocytoses mais pas aux cancers). Le principe de ce traitement repose sur l'inhibition de l'assemblage de la sous-unité HIF-2α avec sa sous-unité β, empêchant ainsi la formation d'un facteur de transcription HIF2A actif. Il s'avère que cette drogue agit à un endroit de la protéine différent des sites des mutations gain de fonction de HIF-2α. Ce traitement pourrait donc être utilisé chez les patients qui présentent des mutations de HIF-2α.

Un essai a récemment été réalisé chez une jeune patiente atteinte du syndrome Zhuang/Pacak et l'inhibiteur de HIF-2α s'est révélé très efficace sur l'ensemble des symptomes (polyglobulie, hypertension, stomatostatinome). L'ensemble des patients porteurs de mutations de HIF-2α que nous décrivons dans la présente étude pourraient donc être éligibles à ce traitement, notamment les jeunes patients mosaïques à fort risque de développer des tumeurs.

Tous ces résultats sont des arguments forts en faveur de la généralisation de ce traitement aux patients porteurs de mutations germinales dans les gènes de la voie de l'hypoxie (*HIF-2A, VHL, PHD2*).

Ce traitement constitue un véritable espoir thérapeutique pour les patients atteints de polyglobulie et la poursuite de travaux collaboratifs à travers des réseaux internationaux permettra de faciliter la recherche sur toutes les maladies rares dont celles du globule rouge.

Appels à projets

FRM – Appel à projets 2022 « espoirs de la recherche » – Aides individuelles - Aide au retour en France

Budget	68 000 €/an correspondant au coût du salaire du bénéficiaire en CDD 3 000 € correspondant aux éventuels de frais de mission
Durée	2 ou 3 ans, non renouvelable
Date limite de dépôt des dossiers	11 mai 2022 à 16 heures (heure de Paris)
Eligibilité	• Demandeur : chercheur post doctorant • Profil du demandeur : chercheur titulaire d'un doctorat en sciences soutenu en France depuis 6 ans maximum à la date du conseil scientifique ET ayant effectué un stage postdoctoral à l'étranger d'au moins 2 ans dans le même laboratoire. • Le demandeur doit être auteur d'au moins une publication acceptée ou en révision en lien avec son stage postdoctoral à l'étranger. • Laboratoire d'accueil situé en France
Objectif	Par cet appel à projets, la Fondation pour la Recherche Médicale s'adresse aux étudiants inscrivant leur recherche en biologie et en santé.

→ Plus d'informations : https://www.frm.org/upload/chercheurs/pdf/frm-per2022.pdf

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Commission Européenne - Programme ERC Advanced Grant

Budget	2 500 000 €
Durée	3 ans
Date limite de dépôt des dossiers	23 mai 2023
Eligibilité	Les chercheurs principaux (CP) - doivent être des chercheurs actifs ayant fait leurs preuves en matière de recherche importantes au cours des 10 dernières années.
Objectif	Permettre à des scientifiques confirmés de proposer un sujet innovant, en rupture par rapport à leurs activités de recherche, avec toujours pour unique critère l'excellence scientifique.

→ Plus d'informations : https://erc.europa.eu/apply-grant/advanced-grant

Appel à manifestation d'intérêt (AMI) Fin de vie 2023 – Plateforme nationale pour la recherche sur la fin de vie

Budget	20 000 € maximum
Durée	12 mois
Date limite de dépôt des dossiers	31 mai 2023 (17h – heure française)
Eligibilité	Le descriptif du projet scientifique ne devra pas dépasser 3 pages; Être rédigés en langue française ; Démontrer une composante interdisciplinaire essentielle au projet ; Associer jeunes chercheur.e.s (étudiant.e.s de Master II, doctorant.e.s, post-doctorant.e.s) et chercheur.e.s confirmé.e.s
Objectif	Inviter les chercheuses et chercheurs à faire émerger de nouveaux projets et de nouvelles thématiques dans le domaine de la fin de vie et favoriser de nouvelles collaborations entre jeunes chercheur.e.s et chercheur.e.s confirmé.e.s et contribuer à de nouvelles interfaces disciplinaires. Accompagner les phases de conception et de faisabilité des projets, puis les étapes de soumission à des appels à projets nationaux ou internationaux.

→ Plus d'informations :
https://www.plateforme-recherche-findevie.fr/appel-manifestation-dinteret-fin-de-vie-2023

Aides à l'organisation de manifestations scientifiques– Plateforme nationale pour la recherche sur la fin de vie

Budget	4 bourses d'un montant maximum de 5000 € chacune
Durée	12 mois
Date limite de dépôt des dossiers	31 mai 2023 (17h – heure française)
Eligibilité	Les dossiers devront être rédigés en langue française et déposés par des porteurs de projet figurant dans l'annuaire national de la Plateforme pour la recherche sur la fin de vie.
Objectif	Ces aides s'adressent aux chercheur.e.s, et plus particulièrement aux jeunes chercheur.e.s souhaitant organiser des manifestations scientifiques relatives à la fin de vie.

→ Plus d'informations : https://www.plateforme-recherche-findevie.fr/aides-lorganisation-de-manifestations-scientifiques

anr – Montage de Réseaux Scientifiques Européens ou Internationaux – MRSEI 2023

Budget	Montant maximal : 35 k€
Durée	24 mois
Date limite de dépôt des dossiers	3e session : 01/06/2023 à 13h00 CEST 4e session : 09/10/2023 à 13h00 CEST
Eligibilité	Le projet devra viser la création d'un réseau scientifique, quelle que soit la disciplines de recherche, constitué de collaborateurs européens ou internationaux avec au moins une entité publique ou assimilée de la recherche française. Le réseau sera coordonné par cette entité publique ou assimilée, porteuse de la proposition MRSEI et du futur projet européen ou international. Cette entité française coordinatrice sera la seule bénéficiaire de la subvention ANR.
Objectif	• Pertinence, originalité et innovation du sujet, ainsi que son adéquation avec l'appel européen ou international visé. • Qualité et crédibilité du réseau envisagé. • Qualification du coordinateur. • Qualité de la planification de montage du réseau. • Impact potentiel du futur projet européen ou international.

→ Plus d'informations : https://anr.fr/fr/detail/call/montage-de-reseaux-scientifiques-europeens-ou-internationaux-mr-sei-2023/

Fondation APICIL - Appel à projets recherche clinique

Budget	NC
Durée	NC
Date limite de dépôt des dossiers	3e session : 5 juin 2023 4e session : 15 octobre 2023
Eligibilité	Etre soignant, chercheur, association
Objectif	Soulager la douleur à travers trois type de projets : • Dossier de recherche clinique sur la douleur • Projet pilote & formation, améliorer le soin, • Dossier d'information sur la douleur

→ Plus d'informations : https://fondation-apicil.org/deposer-un-projet/

Les appels à projets sont régulièrement mis à jour sur le site internet de la filière MCGRE, à l'adresse suivante :
https://filiere-mcgre.fr/espace-professionnels-de-sante/appels-a-projets/

Bibliographie ...

La bibliographie proposée dans ce bulletin recherche concerne des articles parus/entrés dans PubMed
de décembre 2022 à février 2023 inclus
(novembre 2022 à février 2023 pour la rubrique « Toutes maladies rares »).

Anémie dysérythropoïétique congénitale

Congenital dyserythropoietic anemia type IV in the genetic era: A rare neonatal case report of rapid identification with a review of the literature

King R, Lin Z, Balbin-Cuesta G, *et al.*
Deguise MO, Blain S, Simpson E, Liebman M, Ferretti E.
Pediatr Blood Cancer. 2023 May;70(5):e30245. doi: 10.1002/pbc.30245

Anomalies de la membrane du globule rouge

Splenectomy improves erythrocyte functionality in spherocytosis based on septin abundance but not maturation defects

Cloos AS, Pollet H, Stommen A, e*t al.*
Blood Adv. 2023 Feb 8:bloodadvances.2022009114. doi: 10.1182/bloodadvances.2022009114

Treatment of asymptomatic gallstones in children with hereditary spherocytosis requiring splenectomy

Liu Y, Jin S, Li Y, *et al.*
J Pediatr Surg. 2023 Apr;58(4):756-761. doi: 10.1016/j.jpedsurg.2022.11.012

Hereditary spherocytosis associated with Noonan syndrome mimicking a dyserythropoietic anaemia

Faggetter S, Ferster A, Dedeken L, *et al.*
Pediatr Blood Cancer. 2023 Apr;70(4):e30121. doi: 10.1002/pbc.30121

Proteome alterations in erythrocytes with PIEZO1 gain-of-function mutations

Andolfo I, Monaco V, Cozzolino F, *et al.*
Blood Adv. 2023 Jan 3:bloodadvances.2022008673. doi: 10.1182/bloodadvances.2022008673

Red blood cell proteomics reveal remnant protein biosynthesis and folding pathways in PIEZO1-related hereditary xerocytosis

Caulier A, Jankovsky N, Gautier EF, *et al.*
Front Physiol. 2022 Dec 1;13:960291. doi: 10.3389/fphys.2022.960291

How we approach transfusions in a patient with high risk of alloimmunization from McLeod phenotype

Addams J, Hasan RA, Saifee NH.
Pediatr Blood Cancer. 2023 Feb;70(2):e30119. doi: 10.1002/pbc.30119

Déficit en glucose-6-phosphate déshydrogénase

Functional interpretation, cataloging, and analysis of 1,341 glucose-6-phosphate dehydrogenase variants

Geck RC, Powell NR, Dunham MJ.
Am J Hum Genet. 2023 Feb 2;110(2):228-239. doi: 10.1016/j.ajhg.2023.01.003

Favism: Clinical Features at Different Ages

Beretta A, Manuelli M, Cena H.
Nutrients. 2023 Jan 10;15(2):343. doi: 10.3390/nu15020343

Glucose-6-Phosphate Dehydrogenase Deficiency and COVID-19 Mortality, ICU Admission, and Length of Hospitalization

Parnasa E, Perzon O, Klinger A, Ezkoria T, Fischer M.
Isr Med Assoc J. 2023 Feb;25(2):88-90

Safety of age-dosed, single low-dose primaquine in children with glucose-6-phosphate dehydrogenase deficiency who are infected with Plasmodium falciparum in Uganda and the Democratic Republic of the Congo: a randomised, double-blind, placebo-controlled, non-inferiority trial

Taylor WR, Olupot-Olupot P, Onyamboko MA, et al.
Lancet Infect Dis. 2023 Apr;23(4):471-483. doi: 10.1016/S1473-3099(22)00658-2

Primaquine-induced Severe Hemolysis in the Absence of Concomitant Malaria: Effects on G6PD Activity and Renal Function

Douglas NM, Piera KA, Rumaseb A, Ley B, Anstey NM, Price RN.
Am J Trop Med Hyg. 2022 Dec 12;108(1):76-80. doi: 10.4269/ajtmh.21-0834

Anesthetic management of glucose 6-phosphate dehydrogenase deficiency

Gómez Gómez S, Ruano Santiago M, Rodríguez Morillo A, Pérez Muñoz AM, Echevarría Moreno M.
Rev Esp Anestesiol Reanim (Engl Ed). 2023 Feb 25:S2341-1929(23)00050-1. doi: 10.1016/j.redare.2021.11.010

Relationship between Glucose-6-Phosphate Dehydrogenase Deficiency, X-Chromosome Inactivation and Inflammatory Markers

Errigo A, Bitti A, Galistu F, Salis R, Pes GM, Dore MP.
Antioxidants (Basel). 2023 Jan 31;12(2):334. doi: 10.3390/antiox12020334

The potential role of vitamin E in patients with glucose-6-phosphate dehydrogenase deficiency: A systematic review and meta-analysis

Abdelwahab OA, Akil K, Seif A, Allam M, Sherif ME, Al-Alfy MN.
Medicine (Baltimore). 2023 Feb 10;102(6):e32937. doi: 10.1097/MD.0000000000032937

Effects of Two Different Doses of Ursodeoxycholic Acid on Indirect Hyperbilirubinemia in Neonates with Glucose-6-phosphate Dehydrogenase Deficiency Treated with Phototherapy: A Randomized Controlled Trial

Farhadi R, Keyhanian E, Naderisorki M, Nadi Ghara A.
Glob Pediatr Health. 2023 Feb 17;10:2333794X231156055. doi: 10.1177/2333794X231156055

G6PD : un déficit homozygote révélé par une macrocytose au décours d'un épisode d'éthylisme aigu

Souissi M, Morel-Cahoreau A, Daliphard S, Lahary A, Bobée V.
Ann Biol Clin (Paris). 2022 Nov 1;80(6):545-549. doi: 10.1684/abc.2022.1767

Déficit en pyruvate kinase

Early-onset reduced bone mineral density in patients with pyruvate kinase deficiency

Al-Samkari H, Grace RF, Glenthøj A, et al.
Am J Hematol. 2023 Mar;98(3):E57-E60. doi: 10.1002/ajh.26830

Déficit enzymatiques érythrocytaires (autres)

Newly discovered roles of triosephosphate isomerase including functions within the nucleus

Myers TD, Palladino MJ.
Mol Med. 2023 Jan 31;29(1):18. doi: 10.1186/s10020-023-00612-x

Improving the Emergency Department Management of Sickle Cell Vaso-Occlusive Pain Crisis: The Role and Options of Sublingual and Intranasally Administered Analgesia

Ojo AS, Odipe OG, Owoseni O.
J Clin Med Res. 2023 Jan;15(1):10-22. doi: 10.14740/jocmr4841

A Qualitative Systematic Review of Pediatric Patient and Caregiver Perspectives on Pain Management for Vaso-Occlusive Episodes in the Emergency Department

Lapite A, Lavina I, Goel S, Umana J, Ellison AM.
Pediatr Emerg Care. 2023 Mar 1;39(3):162-166. doi: 10.1097/PEC.0000000000002913

Intranasal fentanyl and discharge from the emergency department among children with sickle cell disease and vaso-occlusive pain: A multicenter pediatric emergency medicine perspective

Rees CA, Brousseau DC, Ahmad FA, et al.; SCD Arginine Study Group and PECARN.
Am J Hematol. 2023 Apr;98(4):620-627. doi: 10.1002/ajh.26837

Time to pain relief: A randomized controlled trial in the emergency department during vaso-occlusive episodes in sickle cell disease

Tanabe P, Bosworth HB, Crawford RD, *et al.*
Eur J Haematol. 2023 Jan 5. doi: 10.1111/ejh.13924

IF IM in a crisis: Intranasal fentanyl versus intravenous morphine in adult vaso-occlusive crisis

Assad O, Zamora R, Brown K, Melnitsky L, Moses J, Sherman V.
Am J Emerg Med. 2023 Feb;64:86-89. doi: 10.1016/j.ajem.2022.11.026

Patient-Controlled Analgesia vs Intravenous Push Hydromorphone for Pain Management of Vaso-Occlusive Crisis Associated With Sickle Cell Disease

Russo K, Chhunchha P.
J Pain Palliat Care Pharmacother. 2023 Jan 26:1-7. doi: 10.1080/15360288.2023.2167035

Design of an adaptive randomized clinical trial of intravenous citrulline for sickle cell pain crisis in the emergency department

Majumdar S, McKinley KW, Chamberlain J, *et al.*
Contemp Clin Trials Commun. 2023 Jan 16;32:101077. doi: 10.1016/j.conctc.2023.101077

The complex association of daily opioid dose with visits for pain in sickle cell disease: tolerance or treatment refractory pain?

Prince EJ, Pecker LH, Lanzkron S, Carroll CP.
Pain Med. 2022 Dec 2:pnac187. doi: 10.1093/pm/pnac187

Diagnostic Test Accuracy of Lung Ultrasound for Acute Chest Syndrome in Sickle Cell Disease: A Systematic Review and Meta-Analysis

Omar M, Jabir AR, Khan I, Novelli EM, Xu JZ.
Chest. 2022 Dec 9:S0012-3692(22)04217-9. doi: 10.1016/j.chest.2022.11.042

Inhaled bronchodilators for acute chest syndrome in people with sickle cell disease

Knight-Madden JM, Hambleton IR.
Cochrane Database Syst Rev. 2022 Dec 2;12(12):CD003733. doi: 10.1002/14651858.CD003733.pub5

Intraindividual pain variability metrics for youth with sickle cell disease: Relations to health outcomes

Pascale A, Sisler I, Smith W, Valrie C.
Pediatr Blood Cancer. 2023 Apr;70(4):e30194. doi: 10.1002/pbc.30194

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