Bulletin recherche

Filière FilRougE FILIÈRE SANTÉ MALADIES RARES MCGRE

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FilRougE FILIÈRE SANTÉ MALADIES RARES MCGRE

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Identification d'une EPO de type hépatique comme cause de polyglobulies constitutionnelles

Contexte et objectif

Les polyglobulies regroupent les pathologies caractérisées par un excès de globules rouges conduisant à une augmentation du volume globulaire total. Elles peuvent être d'origine acquise ou constitutionnelle (héréditaire).

Des mutations pathogènes du gène de l'érythropoïétine (EPO), hormone glycoprotéique impliquée dans la régulation de la production des globules rouges (érythropoïèse) ont été identifiées chez des patients atteints de polyglobulie constitutionnelle. Au stade fœtal, l'EPO est produite par les cellules de la crête neurale (futures cellules du rein) et les hépatocytes (cellules du foie). A la naissance, la production de l'EPO est réduite dans le foie et est principalement localisée dans le rein qui assure 90 % de sa production à l'âge adulte.

Les mécanismes moléculaires induits par les mutations pathogènes du gène de l'EPO aboutissant à la surproduction des globules rouges restent méconnus en raison du profil complexe présenté par ces patients qui ont des taux élevés d'ARN messager (ARNm), permettant la synthèse d'EPO, des taux normaux d'EPO circulant dans le sang et une augmentation du nombre de globules rouges.

L'objectif de cette étude était d'identifier des mutations dans les régions non-codantes du gène de l'EPO et d'en caractériser les effets sur l'érythropoïèse de patients atteints de polyglobulie constitutionnelle en errance diagnostique.

Méthode

Cette étude a été approuvée par le comité éthique local (NCT03957863) et réalisée chez les membres de 6 familles atteints de polyglobulie constitutionnelle pour lesquels la cause moléculaire et fonctionnelle n'avait pas encore été identifiée et dont les taux d'EPO circulant dans le sang étaient normaux. Les échantillons des nouveau-nés et des enfants ont été fournis par le CHU de Dijon et les prélèvements des cordons ombilicaux provenaient du centre de ressources biologiques de l'hôpital Saint-Louis (Paris). Les échantillons plasmatiques ont été obtenus après centrifugation du sang et l'ADN a été extrait à partir du sang total et séquencé selon un panel spécifique à l'érythropoïèse et incluant les gènes liés à la voie de l'hypoxie. Les mutations ont été classées selon les critères de l'American College of Medical Genetics and Genomics (ACMG). Les effets des mutations pathogènes sur l'expression du gène de l'EPO ont été étudiés à l'aide d'un gène rapporteur de la luciférase induite par le promoteur de l'*EPO*. Les cellules souches pluripotentes induites (IPS) de ces patients ont été différenciées en cellules productrices d'EPO de type hépatique. Des échantillons d'EPO circulante provenant des patients atteints de polyglobulie constitutionnelle et de nouveau-nés sains ont été analysés par focalisation isoélectrique (électrophorèse permettant de séparer les molécules selon leur charge pH) et l'activité de l'EPO a été évaluée.

Résultats

Trois nouvelles mutations génétiques ont été identifiées dans des régions non-codantes de l'EPO chez ces 6 familles atteintes de polyglobulie constitutionnelle : c.−252C→T au niveau du promoteur principal

et à proximité du site de liaison de facteurs régulant la production d'EPO (GATA, HIF et WT1) et les deux autres mutations c.14–28T→C et c.14–26A→G étaient situées dans l'intron 1.

Les analyses à l'aide des gènes rapporteurs et des cellules productrices d'EPO de type hépatique ont montré que les mutations ciblaient des éléments régulateurs du gène jusque-là non caractérisés, qui réagissaient fortement en cas hypoxie et notamment le facteur de transcription GATA. Les échantillons d'EPO de tous les patients présentaient un profil de focalisation isoélectrique modifié, identique à celui de l'EPO hépatique exprimée chez les nouveau-nés prématurés et chez les patients atteints de polyglobulie acquise associée à des maladies hépatiques.

Cette étude a permis d'identifier chez des patients atteints de polyglobulie constitutionnelle jusqu'alors non expliquée l'existence de 3 nouvelles mutations dans des régions non-codantes du gène de l'EPO, responsables d'une activité accrue de l'EPO dont le profil en iso-électrofocalisation est semblable à celui de l'EPO hépatique du nouveau-né.

L'étude en quelques chiffres :

29 patients issus de **6** familles différentes atteintes de polyglobulie constitutionnelle

3 nouvelles mutations de l'EPO ont été identifiées dans des régions non codantes

L'EPO de type hépatique a une activité au moins **10** fois supérieure chez les patients atteints de ces mutations comparée aux donneurs sains (contrôles).

Cette étude a fait l'objet d'une publication en mai 2025 dans *The New England Journal of Medecine* DOI: 10.1056/NEJMoa2414954

Echange avec

Pr François Girodon

Service d'hématologie Biologique du centre hospitalo-universitaire de Dijon, membre du France Intergroupe Myéloprolifératifs (FIM)

Responsable du CRMR FilRougE-MCGRE de Dijon

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

Le Pr Betty Gardie (directrice à l'Ecole Pratique des Hautes Etudes à Nantes et rattachée à l'INSERM) est l'investigatrice principale de cette étude pluridisciplinaire qui a fait l'objet d'une collaboration entre différentes équipes : celle de Nantes (avec Salam Idriss et Marine Delamare), le laboratoire antidopage français, expert dans l'étude de l'EPO (avec Laurent Martin et Alexandre Marchand), une équipe de recherche en Suisse (avec David Hoogewijs et Darko Maric), Yaël Zermati à Paris et l'équipe de Dijon.

En collaboration avec Betty Gardie, nous menons depuis une dizaine d'années des recherches sur les polyglobulies familiales en errance diagnostique notamment par un séquençage de l'ADN avec un panel de gènes qui comprend le gène de l'EPO, impliqué dans la régulation de la production des globules rouges. Deux publications faisaient mention de l'implication du gène de l'EPO chez deux familles atteintes de polyglobulies et nous ont incités à séquencer en plus des régions codantes, les régions non codantes de l'EPO.

Notre étude a identifié trois types de mutations dans des zones non codantes du gène de l'EPO chez 6 familles atteintes de polyglobulie constitutionnelle. Tout d'abord nous avons été surpris de constater des valeurs normales d'EPO circulant dans le sang chez ces patients avec une mutation du gène *EPO*, car nous nous attendions à une stimulation de la production de l'EPO. Par la suite, en collaboration avec le laboratoire antidopage français, une étude de focalisation isoélectrique de l'EPO circulante a révélé un profil basique très atypique chez toutes les familles atteintes, laissant supposer que la protéine de l'EPO serait insuffisamment glycosylée. Cela reste une hypothèse difficile à prouver, mais ce profil est similaire à celui de l'EPO des nouveau-nés et des prématurés chez qui le foie est responsable de la production de l'EPO alors que chez l'adulte, c'est le rein qui en fabrique la quasi-totalité. Autrement dit, l'EPO de ces patients partage les caractéristiques de l'EPO hépatique sans que nous puissions affirmer qu'elle est vraiment produite par le foie (une biopsie hépatique serait nécessaire, difficilement réalisable à visée de recherche).

Le laboratoire de Betty Gardie a montré que cette EPO avait un gain de fonction, c'est-à-dire qu'elle est une sorte de « super EPO » par rapport à l'EPO normale d'un adulte sain, qui stimule davantage les progéniteurs érythroblastiques et donc la production de globules rouges. L'activité de l'EPO a été mesurée en étudiant la voie JAK-STAT qui est la voie de signalisation activée par l'EPO pour produire des globules rouges. En comparant l'EPO extraite à partir des saignées des patients et l'EPO normale physiologique de donneurs de sang nous avons constaté que l'activité de l'EPO basique était significativement supérieure à celle de l'EPO des donneurs sains.

Comment expliquez-vous le paradoxe entre le taux normal d'EPO circulant et la surproduction des globules rouges ?

L'une des hypothèses serait que l'EPO de type hépatique serait plus instable, car elle est insuffisamment glycosylée et se dégraderait donc plus vite. Or le dosage ne reflèterait que la quantité d'EPO circulant dans le sang à l'instant T. D'autres questions restent en suspens et des travaux complémentaires seront nécessaires pour améliorer la compréhension des mécanismes sous-jacents à ce paradoxe. En particulier, il reste à comprendre pourquoi le foie est à nouveau stimulé dans sa production d'érythropoïétine dans certaines pathologies hépatiques.

Quelles sont les pistes d'amélioration diagnostique ?

Le bilan de débrouillage de la polyglobulie vraie pourrait intégrer en plus de la mesure quantitative de l'EPO, sa caractérisation qualitative par la focalisation isoélectrique : ainsi, selon le profil obtenu, le bilan diagnostique pourrait s'orienter vers des explorations complémentaires plus ciblées, au moins dans un premier temps et faire l'économie d'explorations complexes et coûteuses.

Est-ce que cela sera facile à mettre en place dans les laboratoires de diagnostic ?

A l'heure actuelle en France, seul le laboratoire antidopage français est en mesure de réaliser cette analyse. Ce laboratoire essaye de développer différentes techniques comme la spectrométrie de masse ou d'autres techniques qui pourraient être facilement utilisées par les différents laboratoires de diagnostic mais pour le moment ces techniques ne sont pas adaptées à la détection du profil de l'EPO. C'est donc le facteur limitant. Néanmoins, il faut savoir que cette analyse spécifique de l'EPO concerne relativement peu de malades. Il pourrait donc être envisagé de confier toutes les demandes d'examens au laboratoire antidopage français qui les réaliserait à termes, à l'aide d'un automate.

La seconde piste d'amélioration diagnostique est le séquençage des régions non codantes pour ne pas risquer de manquer la détection de certaines mutations. J'incite fortement, notamment lors des RCP (Réunion de concertation pluridisciplinaire) diagnostiques, mes collègues généticiens et biologistes à élargir leur panel du gène de l'EPO aux régions non codantes. Cela est déjà en cours de mise en place pour certains laboratoires.

Quelles sont les pistes thérapeutiques de votre étude ?

La poursuite de ces travaux de recherche pourra apporter des connaissances plus fines sur la régulation de l'EPO et sa caractérisation. Cela conduira, nous l'espérons, à des pistes thérapeutiques comme par exemple permettre de réactiver la production de l'EPO par le foie chez les patients atteints d'insuffisance rénale sévère.

Appels à projets

FilRougE-MCGRE/CGRF - Appel à projets Impulsion Recherche 2025

Budget	Budget total : 140 000 € 20 000 € pour les projets mono site 60 000 € pour les projets multisites
Durée	24 mois maximum
Date limite de dépôt des dossiers	5 septembre 2025, à 17h
Eligibilité	Être membres des laboratoires et des centres de référence (CRMR) et de compétence (CCMR) partenaires de la filière FilRougE-MCGRE ainsi que les laboratoires de recherche impliqués dans les thématiques de la filière FilRougE-MCGRE et du CGRF peuvent candidater à cet appel à projets.
Objectif	Encourager les domaines de la recherche fondamentale, translationnelle, clinique, en lien avec les pathologies rares de la filière

→ Plus d'informations : ICI

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FilRougE-MCGRE/Novo Nordisk/Fondation maladies rares – Sciences humaines et sociale - « Amélioration de la qualité de vie des patients atteints de drépanocytose ou de β-thalassémie »

Budget	30 000 €
Durée	24 mois maximum
Date limite de dépôt des dossiers	12 septembre 2025, à 12h
Eligibilité	Projet de recherche se focalisant sur des problématiques Sciences humaines et soiales et utilisant des méthodologies de SHS ; • Porté par un responsable scientifique chercheur en SHS avec un poste permanent dans une équipe de recherche française, affiliée au monde universitaire (équipe de recherche au sein des universités, autres établissements d'enseignement supérieur ou instituts de recherche) et/ou au secteur clinique ou de la santé publique (équipe de recherche travaillant dans les hôpitaux/les organismes de santé publique) ; • Démontrant l'implication et la complémentarité de l'équipe SHS, de l'équipe médicale experte de la pathologie et de l'association de patients. • Détaillant le bénéfice concret attendu pour les malades, en présentant par exemple les supports d'application de la recherche.
Objectif	Améliorer le parcours de vie des patients atteints de drépanocytose ou de la β-thalassémie.

→ Plus d'informations : ICI

FilRougE-MCGRE - Appel à projets Expertise-Formation

Budget	50 000 €
Durée	24 mois maximum
Date limite de dépôt des dossiers	30 septembre 2025, à 17h
Eligibilité	Cet AAP est ouvert à l'ensemble des acteurs de la filière : • Professionnels de santé ; • Associations membres.
Objectif	Soutenir la formation des professionnels de santé et sociaux afin de mieux identifier et prendre en charge les maladies constitutionnelles rares du globule rouge et de l'érythropoïèse et à poursuivre la contribution des associations de malades à cet objectif de la filière.

→ Plus d'informations : ICI

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FilRougE-MCGRE - Appel à projets Education thérapeutique du patient

Budget	50 000 €
Durée	24 mois maximum
Date limite de dépôt des dossiers	30 septembre 2025, à 17h
Eligibilité	Cet appel à projet est ouvert à l'ensemble des centres de compétence et de référence de la filière. Toutefois, une attention particulière sera portée : - Aux projets déposés par les centres de compétence ; - Aux projets pouvant s'adapter à toutes les pathologies rares du globule rouge et de l'érythropoïèse.
Objectif	Faciliter la mise en place de nouveau programme d'ETP ou l'amélioration de programmes existants.

→ Plus d'informations : ICI

Bpifrance - Grand Défi : AAP dispositifs médicaux numériques en santé mentale

Budget	Au moins 100 000 € => Bénéficiaire académique Au moins 500 000€ => Grande entreprise 60 % de l'aide attribuée sous la forme de subventions ; 40 % de l'aide attribuée sous la forme d'avances remboursables.
Durée	Les recherches doivent être menées dans le courant de l'année civile.
Date limite de dépôt des dossiers	30 septembre 2025
Eligibilité	Projets portant sur la prévention, dépistage/diagnostic, prise en charge ou encore suivi à distance par des dispositifs médicaux numériques (DMN) dans le cadre de la santé mentale incluant, notamment, les troubles psychiques, les troubles addictifs, les troubles neurodéveloppementaux.
Objectif	Soutenir des projets innovants dans le domaine du numérique en santé mentale et en psychiatrie, et à favoriser l'émergence accélérée d'entreprises leaders dans leur domaine, pouvant prétendre à une envergure mondiale en accélérant une des phases de développement et d'accès au marché.

→ Plus d'informations : ICI

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Fondation recherche médicale : Espoirs de la Recherche : Amorçage de jeunes équipes

Budget	450 000 euros maximum
Durée	3 ans
Dépôt des dossiers	5 novembre 2025 (La FRM se réserve le droit de clôturer cet appel à projets de façon anticipée, en cas d'utilisation complète des fonds).
Eligibilité	La structure d'accueil doit avoir sélectionné le/la candidat(e) dans le cadre d'un appel à candidatures finalisé par des auditions menées par un jury international. Cette sélection par la structure de recherche d'accueil doit avoir eu lieu depuis moins de 24 mois à la date de la réunion du Comité FRM
Objectif :	Renforcer le potentiel de recherche de structures qui ont, dans le cadre d'un appel à candidatures international, déjà sélectionné le/la chef d'équipe qu'elles souhaitent accueillir.

→ Plus d'informations : ICI

IReSP - Subventions hors appels à projets

Budget	5000 €
Durée	NC
Date limite de dépôt des dossiers	NC
Eligibilité	Manifestations scientifiques en santé publique (hors recherche clinique)
Objectif	Soutenir des projets d'envergure locale, nationale ou internationale 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...). Thématiques de recherche en santé publique : -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) ; Les politiques publiques et de santé (impact sur la santé des populations, conditions d'élaboration, évaluation, évolution...).

→ Plus d'informations : ICI

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Institut Cerba - Développer des actions de santé solidaire

Budget	NC
Durée	NC
Date limite de dépôt des dossiers	Permanent
Eligibilité	Être porteur ou soutenir un projet caritatif lié à la santé
Objectif	- Contribuer à l'information des professionnels de santé et à l'éducation sanitaire du public, - Soutenir et promouvoir la recherche et l'innovation en biologie médicale, - Permettre à tous le même accès à une biologie médicale innovante.

→ Plus d'informations : ICI

Viva Lab : Soutenir l'innovation au service du « Bien vieillir » !

Budget	NC
Durée	NC
Date limite de dépôt des dossiers	Permanent
Eligibilité	Les projets présentés doivent être innovants et répondre à des besoins dans les domaines suivants : • Habitat et cadre de vie • Autonomie numérique • Mobilité • Bien-être et prévention • Vie quotidienne • Soutien aux aidants
Objectif	Soutenir l'innovation dans le champ de la prévention et du vieillissement actif et en santé.

→ Plus d'informations : ICI

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EMBO - Bourses d'échanges scientifiques

Budget	Contribution aux frais de voyage et de subsistance du boursier
Durée	7 jours à 3 mois
Date limite de dépôt des dossiers	Postuler trois mois avant la date de début proposée. Soumission des demandes minimum30 jours avant la date de début de la visite de recherche.
Eligibilité	Être un chercheur actif à n'importe quelle étape ; Posséder au moins un an d'expérience en recherche aux cycles supérieurs ou l'équivalent au moment de la demande. Les échanges de recherche doivent avoir lieu entre les laboratoires des États membres de l'EMBC et les partenaires mondiaux de l'EMBC (Chili, Inde, Singapour, Taïwan). Les subventions ne sont pas attribuées pour des échanges entre deux laboratoires au sein d'un même pays.
Objectif	Soutenir les collaborations internationales, permettre le transfert d'expertise

→ Plus d'informations : ICI

Université de Poitiers - Soutien à l'accueil de chercheurs ou de chercheuses au sein des UMR du site de Poitiers

Budget	130 000€
Durée	3 ans
Dépôt des dossiers	Projet permanent
Eligibilité	Recrutement ou mobilité d'un chercheur titulaire CNRS ou Inserm (CR ou DR) au sein d'une UMR de Poitiers. Les thématiques et/ou projets d'intégration doivent concerner l'un des trois ODD du programme Excellences (Santé bien-être, Education de qualité, Villes et communautés durables). Installation sur le site universitaire de Poitiers
Objectif :	Allocation de thèse (130 k€) éventuellement divisible en deux pour favoriser la recherche de cofinancements - jusqu'à 45 k€ en dotation de crédits de fonctionnement - jusqu'à 50 k€ en dotation de crédits d'équipements - accompagnement individualisé par Grand Poitiers

→ Plus d'informations : ICI

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

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

Bibliographie

La bibliographie proposée dans ce bulletin recherche est une sélection d'articles entrés dans PubMed de janvier à mai 2025 inclus (plus une vingtaine d'articles plus récents).

Anémie dysérythropoïétique congénitale

Severe Neonatal Anemia with Multi-Organ Failure, Extreme Placentomegaly, and Placental Megaloblastic Erythroblastosis as Features in Identifying Congenital Dyserythropoietic Anemia Type 1: A Case Report
Roose O, Gengler C, Stoykova S, Good JM, Tolsa JF, Beauport L.
Neonatology. 2025 Jun 5:1-4. doi: 10.1159/000546794

Anémie liée au métabolisme du fer et anémie sidéroblastique

How I treat iron-refractory iron deficiency anaemia-An expert opinion-based treatment guidance for children and adults
Hoving V, Donker AE, Schols SEM, Swinkels DW.
Br J Haematol. 2025 Apr;206(4):1067-1076. doi: 10.1111/bjh.20030

X-linked sideroblastic anemia in females
Ducamp S, Campagna DR, Sendamarai AK, Schmidt PJ, Tsai HK, *et al.*
Blood. 2025 Apr 3;145(14):1583-1587. doi: 10.1182/blood.2024024475

SLC25A38 is required for mitochondrial pyridoxal 5'-phosphate (PLP) accumulation
Pena IA, Shi JS, Chang SM, Yang J, Block S, *et al.*
Nat Commun. 2025 Jan 24;16(1):978. doi: 10.1038/s41467-025-56130-3

P2 Receptor Antagonists Rescue Defective Heme Content in an In Vitro SLC25A38-Associated Congenital Sideroblastic Anemia Cell Model
Santoro A, De Santis S, Palmieri F, Vozza A, Agrimi G, *et al.*
Int J Mol Sci. 2024 Dec 12;25(24):13314. doi: 10.3390/ijms252413314

The role of genetic testing in accurate diagnosis of X-linked sideroblastic anemia: novel ALAS2 mutations and the impact of X-chromosome inactivation
Jové-Solavera D, Rámila M, Ferrer-Cortés X, Olivella M, Venturi V, *et al.*
Sci Rep. 2025 Apr 7;15(1):11843. doi: 10.1038/s41598-025-95590-x

Myopathy, lactic acidosis and sideroblastic anemia syndrome 1 (MLASA1) : clinical hallmarks in a large pedigree with a novel *PUS1* R144Q mutation, remarkable response to somatropin, and review of the literature
Parisi L, Escher R.
Haematologica. 2025 May 29. doi: 10.3324/haematol.2025.287393

Antenatal presentations of congenital sideroblastic anaemia as severe fetal anaemia
Ng K, Rougerie M, Rolnik DL.
BMJ Case Rep. 2025 May 26;18(5):e257172. doi: 10.1136/bcr-2023-257172

X-Linked Sideroblastic Anaemia Caused by Intronic *ALAS2* Variant Resulting in Highly Variable Expressive Phenotype in Male Siblings, a Case Report
O'Connor J, Mannion N, McKenna C, Sweeney K, Niblock A.
EJHaem. 2025 May 19;6(3):e70060. doi: 10.1002/jha2.70060

Functional characterisation of missense ceruloplasmin variants and real-world prevalence assessment of Aceruloplasminemia using population data
Ziliotto N, Lencioni S, Cirinciani M, Zanardi A, Alessio M, *et al.*
EBioMedicine. 2025 Mar;113:105625. doi: 10.1016/j.ebiom.2025.105625

Ceruloplasmin administration in the preclinical mouse model of aceruloplasminemia reveals a sex-related variation in biodistribution
Belloli S, Monterisi C, Rainone P, Coliva A, Zanardi A, *et al.*
Commun Biol. 2025 Feb 19;8(1):264. doi: 10.1038/s42003-025-07714-8

Expanding the Phenotype of DNA Ligase 1 Deficiency: First Report of Macrocytic Sideroblastic Anemia
Jiang D, Sampino EV, Rosenlind K, Campagna DR, DiTroia S, *et al.*
Am J Hematol. 2025 May;100(5):941-943. doi: 10.1002/ajh.27649

Anomalie de la membrane du globule rouge

An overview of hereditary spherocytosis and the curative effects of splenectomy
Turpaev K, Bovt E, Shakhidzhanov S, Sinauridze E, Smetanina N, *et al.*
Front Physiol. 2025 Feb 11;16:1497588. doi: 10.3389/fphys.2025.1497588

Correlation of Genetic Mutation With Outcomes in Children With Hereditary Spherocytosis Undergoing Partial Splenectomy: A Multicentre Study
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J Pediatr Surg. 2025 Apr;60(4):162229. doi: 10.1016/j.jpedsurg.2025.162229

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BMC Pediatr. 2025 May 19;25(1):404. doi: 10.1186/s12887-025-05760-z

Loss of Function SPTA1 Variants Causes Neonatal Liver Failure and Fetal Anemia
Brewin J, Clark B, Smith F, Parkin N, Nardo-Marino A, *et al.*
Am J Hematol. 2025 Jun 26. doi: 10.1002/ajh.27751

The evolving landscape of hereditary stomatocytosis
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Dehydrated Hereditary Stomatocytosis in a Very Preterm Twin
Horibe S, Isshiki K, Tamura S, Watanabe T, Miyamoto Y, Ikeda K.
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Ong ST, Nam YW, Nasburg JA, Ramanishka A, Ng XR, *et al.*
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Déficit en glucose-6-phosphate déshydrogénase

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