Bulletin recherche

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FILIÈRE SANTÉ MALADIES RARES

Table des matières

Le point sur ...

L'hématopoïèse clonale dans la drépanocytose

Contexte et objectifs

La drépanocytose est une maladie génétique qui touche les érythrocytes matures et qui s'exprime par de nombreuses complications cliniques dont les causes sont encore mal connues. Les patients drépanocytaires ont notamment davantage de risques de développer une leucémie aiguë, et deux échecs de greffes précoces ont été rapportés chez des patients drépanocytaires qui avaient eu une transplantation de cellules souches hématopoïétiques (greffe de moelle osseuse) et qui ont par la suite développé des néoplasmes myéloïdes avec mutation du gène TP53 (impliqué dans le fonctionnement cellulaire). L'hématopoïèse clonale confère à la cellule souche où elle est apparue une activité de multiplication accrue par rapport aux autres et donc une expression augmentée et que l'on peut mesurer. Ces cellules souches hématopoïétiques sont généralement porteuses d'une mutation somatique (c'est-à-dire acquise) dans des gènes associés aux hémopathies malignes (cancers des cellules sanguines et de leurs précurseurs), telles que les leucémies, les syndromes myélodysplasiques et les lymphomes. L'hématopoïèse clonale est aussi associée à un plus grand risque de maladies cardiovasculaires et de mortalités toutes causes confondues. Cet article visait à mieux caractériser l'hématopoïèse clonale dans la drépanocytose et l'influence de ces deux pathologies l'une sur l'autre.

Méthode

Afin de caractériser l'hématopoïèse clonale chez les patients drépanocytaires, le séquençage whole exome (WES), qui permet de séquencer en une seule étape l'ensemble des régions codantes du génome humain, a été utilisé pour rechercher les variations somatiques chez 1459 patients drépanocytaires issus de 5 cohortes différentes. La moyenne d'âge était de 23.6 +/- 14 ans et 1439 patients étaient SS ou S/ß° thalassémiques et 20 SC. Un groupe contrôle a été constitué à partir de la cohorte BioMe par 6848 individus Africains américains non-drépanocytaires (dont 517 étaient porteurs d'un trait drépanocytaire) dont l'âge moyen au moment du prélèvement sanguin était 50.1+/- 15.1 ans.

Résultats

Quinze mutations d'hématopoïèse clonale ont été identifiées chez 15 patients drépanocytaires (SS ou S/B°) dont 13 variants mono-nucléotidiques et 2 délétions. Ces variants ont été annotés faux-sens (n=10), non-sens (n=2), décalé (n=2) ou site d'épissage essentiel (n=1) et la fréquence de variation allélique allait de 0.025 à 0.26. Les variants ont été retrouvés sur les gènes DNMT3A (n=10), TP53 (n=2), TET2 (n=1), ETV6 (n=1) et IKZF1 (n=1).

Les mutations d'hématopoïèse clonale apparaissaient davantage chez les individus plus âgés mais de manière plus précoce chez les patients drépanocytaires que chez les individus contrôles. La prévalence (nombre de cas dans une population à un moment donné) de l'hématopoïèse clonale était plus importante chez les patients drépanocytaires par rapport aux individus contrôles.

L'analyse de régression logistique a montré que le fait d'être atteint de drépanocytose était associé à une augmentation du risque de l'apparition de l'hématopoïèse clonale indépendamment de l'âge, du sexe ou de la méthode de l'analyse.

Etant donné que les patients drépanocytaires étaient issus de 5 cohortes distinctes, des analyses portant uniquement sur la cohorte BioMe (29 patients drépanocytaires et 6848 individus contrôles) ont été réalisées pour s'affranchir de l'hétérogénéité de la profondeur de séquençage entre les cohortes. L'apparition de l'hématopoïèse clonale était plus importante chez les patients drépanocytaires mais elle n'apparaissait pas plus tôt chez les patients drépanocytaires par rapport aux individus contrôles.

Le suivi clinique était disponible pour 13 patients drépanocytaires atteints d'une hématopoïèse clonale avec un suivi médian de 6,1 ans après le prélèvement sanguin. Les 3 patients les plus âgés sont décédés de tumeurs malignes solides et 4 ont développé des pathologies cardiovasculaires mais aucun n'a déclaré de cancer hématologique. Par ailleurs, ces 13 patients ont présenté d'autres complications liées à la drépanocytose telles qu'une rétinopathie (n=8) ou une maladie rénale chronique (n=6). Le nombre de patients était trop faible pour déterminer si la présence de l'hématopoïèse clonale a contribué à ces complications. Aucune association n'a été trouvée entre la présence d'hématopoïèse clonale et les paramètres hématologiques.

D'autres études utilisant des techniques de séquençage ciblé plus approfondies permettront d'améliorer la connaissance de la prévalence liée à l'âge de l'hématopoïèse clonale et ses caractéristiques génétiques dans la drépanocytose.

L'étude en quelques chiffres :

1459 patients atteints de drépanocytose (dont **1439 SS et S/ß° thalassémiques** et **20 SC**) et **6848** contrôles ont été inclus dans cette étude

15 mutations d'hématopoïèse clonale identifiées chez 15 patients SS ou S/ß° dont 13 variantes mono-nucléotidiques et 2 délétions

199 mutations d'hématopoïèse clonale identifiées chez **189** contrôles

7,1 % : c'est le risque qu'a un individu drépanocytaire âgé de 50 ans d'avoir une mutation d'hématopoïèse clonale alors qu'il n'est que de 0,7 % pour un individu non drépanocytaire.

13,5 fois plus de risque pour les patients drépanocytaires de développer une hématopoïèse clonale par rapport aux individus contrôles

Cette étude a fait l'objet d'une publication en novembre 2021 dans le journal Blood : https://doi.org/10.1182/blood.2021011121

Échange avec ...

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→ Pouvez-vous nous décrire le contexte clinique de l'étude ?

Notre laboratoire cherche à comprendre l'influence des mutations génétiques sur le développement des maladies cardiovasculaires et hématologiques (dont la drépanocytose). Des études ont récemment démontré que la prévalence des mutations somatiques était plus importante chez les patients atteints de pathologies sanguines par rapport aux individus contrôles. Nous avons donc souhaité déterminer la prévalence des mutations de l'hématopoïèse clonale chez les patients atteints de drépanocytose à partir de données issues d'un précédent projet de séquençage génomique réalisé chez un grand nombre d'individus. Lorsque nous avons soumis notre étude au comité éditorial il y a près d'un an, les articles sur les échecs précoces de greffe de moelle osseuse chez les patients drépanocytaires n'avaient pas encore été publiés. Depuis, plusieurs travaux de recherche sont parus sur cette thématique et attestent que la caractérisation de l'hématopoïèse clonale dans la drépanocytose est un sujet d'intérêt majeur et d'actualité.

Notre étude a établi de façon robuste l'existence de l'hématopoïèse clonale chez les patients drépanocytaires. La démonstration de l'apparition de mutations somatiques dans les gènes p53, DNMT3, TET2... est un résultat en soi très intéressant d'un point de vue clinique.

Nous avons de plus observé un âge plus précoce des patients drépanocytaires comparés aux individus contrôles concernant la prévalence des mutations de l'hématopoïèse clonale. Cependant, ce résultat ne semble pas être confirmé par un plus grand échantillon. En effet, la dernière étude parue ce mois-ci dans la revue Journal of Clinical Investigation (https://doi.org/10.1172/JCI156060) intègre un nombre beaucoup plus élevé d'individus et ne montre pas de différence d'âge dans la prévalence de l'hématopoïèse clonale chez 3090 patients drépanocytaires comparés à 71100 individus contrôles.

Le résultat de notre étude montrant que les patients drépanocytaires ont davantage de mutations somatiques est cohérent avec ce que l'on connait de la biologie des cellules souches drépanocytaires. Nous restons toutefois prudents sur le résultat suggérant que ces mutations apparaitraient de manière plus précoce chez les patients car il est en contradiction avec d'autres études et nécessite d'être reproduit avec des techniques plus précises et une plus grande cohorte avant de pouvoir être considéré comme valide.

→ Avez-vous rencontré des difficultés particulières dans la réalisation de cette étude ?

Dans la présente étude, 13 patients ont pu être suivis. Bien que ceci ait permis d'obtenir des résultats intéressants, ce n'est pas suffisant pour apporter des conclusions robustes d'un point de vue statistique. Nous n'avons pas mis en évidence d'effet néfaste de l'hématopoïèse clonale chez les patients drépanocytaires du point de vue de la sévérité clinique de leur maladie, mais je ne suis pas convaincu que cette conclusion soit maintenue

avec une plus grande cohorte. Il faudrait donc valider nos observations par un suivi épidémiologique sur une très grande cohorte qui serait de l'ordre de plusieurs milliers de patients (entre 5000 et 10 000) et en utilisant une technique de séquençage très précise qui repose sur une technologie développée spécifiquement pour l'étude de l'hématopoïèse clonale. Au lieu de séquencer le génome ou l'exome, cette technique consiste à séquencer les gènes déjà identifiés comme porteurs de mutations somatiques (tels que p53, DNMT3...) et à se concentrer uniquement sur les séquences concernées, permettant ainsi un séquençage très précis. Ce type d'analyse réduit fortement le risque d'erreur de séquençage et permet de distinguer une mutation somatique rare d'une erreur de séquençage (on parle en anglais de error corrected DNA sequencing). Nous pourrions ainsi obtenir une très bonne idée des mutations somatiques de l'hématopoïèse clonale, même les plus rares. Ces projets sont en discussion.

→ Quel pourrait être l'impact de vos recherches dans la prise en charge des patients drépanocytaires ?

Ces recherches pourraient avoir un impact pour les patients à deux niveaux.

Premièrement, et ceci est valable aussi bien pour les patients drépanocytaires que pour tous les patients traités par thérapie génique, il y a un intérêt à caractériser la présence de mutations somatiques d'hématopoïèse clonale chez le donneur et chez le receveur afin d'évaluer leur impact sur le succès du traitement. Il est important de bien comprendre que nos résultats ne mettent aucunement en doute l'utilité de la thérapie génique. Les traitements ablatifs ne sont pas la cause de ces mutations, cela fait partie du processus normal de prolifération, de différenciation des cellules souches hématopoïétiques.

Deuxièmement, l'amélioration de la connaissance de l'hématopoïèse clonale chez les patients drépanocytaires nous permettra de déterminer si ces mutations ont un impact sur la sévérité clinique des patients, comme l'apparition de maladies cérébrovasculaires, des crises de douleurs, des syndromes thoraciques aigus... Seul un suivi longitudinal sur une plus grande cohorte nous permettra de répondre à cette question.

Appels à projets

FRM – Appel à projets 2022 « espoirs de la recherche » – Aides individuelles - Poste de thèse pour internes et assistants

Budget	50 000 €/an pour un interne ou un ancien interne, correspondant au coût du salaire du bénéficiaire en CDD 60 000 €/an pour un chef de clinique assistant (CCA) ou un assistant hospitalo-universitaire (AHU) correspondant au coût du salaire du bénéficiaire en CDD 3000 € correspondant aux éventuels de frais de mission
Durée	1 an et demi (01/10/ 2022 – 01/04/ 2023)
Date limite de dépôt des dossiers	9 mars 2022 à 16 heures (heure de Paris)
Eligibilité	Demandeur : doctorant inscrit en 1ere ou en 2nde année de thèse de sciences durant l'année 2022-2023 Profil du demandeur : à la date de début du financement, le demandeur doit être titulaire d'un Master 2 recherche. Il doit également être soit interne ou ancien interne soit chef de clinique assistant (CCA) / assistant hospitalo-universitaire (AHU). NB : La thèse d'exercice n'est pas requise. • Inscription obligatoire pour l'année universitaire 2022-2023 soit en 1ère soit en 2ème année de thèse de sciences dans une université française. • Laboratoire d'accueil situé en France ou à l'étranger.
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/chercheurs/appel-a-projets-frm/espoirs-de-la-recherche

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Appel à projets SPA 2022 – Lutter contre les usages et les addictions aux substances psychoactives

Budget	En fonction de la durée du projet
Durée	Projet de recherche complet : 36 à 48 mois Projets pilote : 18 à 24 mois Projet de recherche « d'amorçage » : 12 à 18 mois
Date limite de dépôt des dossiers	10 mars 2022 (16H00)
Eligibilité	Les projets déposés doivent s'inscrire dans l'un des 3 volets suivants : Volet 1 : Substances psychoactives et population générale Volet 2 : Substances psychoactives et cancers Volet 3 : Substances psychoactives et pathologies autres que les cancers.
Objectif	outenir la recherche et à produire des connaissances dans le domaine des consommations de substances psychoactives et la lutte contre les addictions : en priorité sur tabac, alcool et cannabis, identifiés notamment comme facteurs à risques avérés de cancers, mais aussi sur les autres substances psychoactives ainsi que sur les poly-consommations.

→ Plus d'informations :
https://iresp.net/appel_a_projets/appel-a-projets-spa-2022-lutter-contre-les-usages-et-les-addictions-aux-substances-psychoactives/

PHC Maghreb 2023

Budget	Moyens attribués par la partie française : 22 000 € par an/projet Moyens attribués par la partie marocaine : environ 35750 MAD Moyens attribués par la partie tunisienne : 3680€ par an
Durée	3 ans
Date limite de dépôt des dossiers	17 mars 2022
Eligibilité	• Les projets conjoints doivent impliquer la Tunisie, le Maroc et la France • La présence d'un ou plusieurs partenaires socio-économiques ayant des activités au Maghreb est fortement encouragée. • Un coordinateur du projet doit être désigné parmi les responsables des équipes engagées dans chacun des différents pays • Les échanges sud-sud (Tunisie-Maroc) devront représenter au moins 30% des mobilités
Objectif	Encourager les coopérations multilatérales en finançant des projets conjoints impliquant la Tunisie, le Maroc et la France en répondant entre autres aux questions de santé publique.

→ Plus d'informations : https://www.campusfrance.org/fr/maghreb#Conditions

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FRM – Appel à projets 2022 « espoirs de la recherche » – Amorçage de jeunes équipes

Budget	1 800 000€ (450 000 euros maximum par projet)
Durée	3 ans
Date limite de dépôt des dossiers	Session 1 : 13 avril 2022 Session 2 : 8 septembre 2022 Session 3 : 3 novembre 2022
Eligibilité	La structure d'accueil doit avoir sélectionné le/la candidat.e dans le cadre d'un appel à candidatures finalisé par des auditions par un jury international.
Objectif	Soutien à de jeunes chercheurs, français ou étrangers, désireux de rejoindre une structure de recherche française pour mettre en place et animer une nouvelle équipe de recherche.

→ Plus d'informations :
https://www.frm.org/upload/chercheurs/pdf/ao_amorcage2022.pdf

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

Budget	52 000 €/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) ou d'un PhD (soutenu à l'étranger) soutenu au plus tôt le 1er avril 2021 (soit depuis 18 mois maximum à la date du conseil scientifique) et au plus tard le 31 décembre 2022. • Le demandeur doit être auteur d'au moins une publication (à l'exclusion des articles de revues) acceptée ou en révision en lien avec ses travaux de thèse. • Laboratoire d'origine situé en France ou à l'étranger • Laboratoire d'accueil situé en France et différent du laboratoire de thèse. • L'encadrant dans le laboratoire d'accueil doit être différent du directeur ou du codirecteur de thèse
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

Fondation Groupama – Prix de l'Innovation Sociale

Budget	20 000 €
Durée	24 mois
Date limite de dépôt des dossiers	Tout au long de l'année
Eligibilité	Starts-up, associations et institution structure justifiant de son existence en France métropolitaine
Objectif	Soutenir des initiatives novatrices marquant une avancée significative dans la lutte contre les maladies rares en : - Contribuant à rompre l'isolement, - Favorisant le lien social et/ou l'insertion professionnelle, Aidant au développement de l'éducation thérapeutique du patient, isolé par la maladie.

→ Plus d'informations :
https://www.fondation-groupama.com/recherche-innovation/le-prix-de-linnovation-sociale/

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 concerne les articles parus entre octobre 2021 et janvier 2022.

Information COVID-19 : la bibliographie inclut une partie des articles concernant la maladie COVID-19 et les pathologies du globule rouge.
Pour consulter la liste complète, régulièrement mise à jour, de ces articles, rendez-vous sur le site MCGRE :

https://filiere-mcgre.fr/actualites/veille-bibliographique-covid-19-et-maladies-du-globule-rouge/

Anémie dysérythropoïétique congénitale

SEC23A rescues SEC23B-deficient congenital dyserythropoietic anemia type II

King R, Lin Z, Balbin-Cuesta G, *et al.*
Sci Adv. 2021 Nov 26;7(48):eabj5293. doi: 10.1126/sciadv.abj5293

Anomalies de la membrane du globule rouge

Laboratory Indices in Patients with Positive and Borderline Flow-cytometry EMA-screening Test Results for Hereditary Spherocytosis

Azoulay D, Levov I, Shaoul E, Kuperman AA.
J Pediatr. 2022 Jan 13:S0022-3476(21)01186-0. doi: 10.1016/j.jpeds.2021.11.074

Diagnosis and clinical management of red cell membrane disorders

Kalfa TA.
Hematology Am Soc Hematol Educ Program. 2021 Dec 10;2021(1):331-340. doi: 10.1182/hematology.2021000265

The diagnostic protocol for hereditary spherocytosis-2021 update

Wu Y, Liao L, Lin F. T.
J Clin Lab Anal. 2021 Dec;35(12):e24034. doi: 10.1002/jcla.24034

Hereditary Xerocytosis: Differential Behavior of PIEZO1 Mutations in the N-Terminal Extracellular Domain Between Red Blood Cells and HEK Cells

Yamaguchi Y, Allegrini B, Rapetti-Mauss R, *et al.*
Front Physiol. 2021 Oct 18;12:736585. doi: 10.3389/fphys.2021.736585

Pyoderma Gangrenosum in a Splenectomy Incision in a Patient with Haemolytic Anaemia due to Hereditary Spherocytosis: a Case Report and Literature Review

Krajewski PK, Chlebicka I, Szepietowski JC, Maj J.
Acta Derm Venereol. 2021 Nov 24;101(11):adv00599. doi: 10.2340/actadv.v101.438

Efficacy of cytochemical tests in gene analysis of hereditary spherocytosis: a case study of six patients with different disease subtypes

Shibuya A, Kawashima H, Tanaka M.
Hematology. 2021 Dec;26(1):827-834. doi: 10.1080/16078454.2021.1979184

A Systematic review on diagnostic methods of red cell membrane disorders in Asia

Silva R, Amarasinghe D, Perera S, Premawardhena A.
Int J Lab Hematol. 2022 Jan 23. doi: 10.1111/ijlh.13800

Déficit en glucose-6-phosphate déshydrogénase

Thirty-five males with severe (Class 1) G6PD deficiency (c.637G>T) in a North American family of European ancestry

Bahr TM, Agarwal AM, Meznarich JA, *et al.*
Blood Cells Mol Dis. 2021 Dec;92:102625. doi: 10.1016/j.bcmd.2021.102625

Acute Hemolytic Anemia Caused by G6PD Deficiency in Children in Mayotte: A Frequent and Severe Complication

Boyadjian M, Follenfant E, Chamouine A, *et al.*
J Pediatr Hematol Oncol. 2021 Dec 30. doi: 10.1097/MPH.0000000000002381

Hemolysis After Medication Exposure in Pediatric Patients With G6PD Deficiency

Doshi BS, Kamdar A, Lambert MP, Obstfeld AE.
J Pediatr Hematol Oncol. 2021 Oct 15. doi: 10.1097/MPH.0000000000002350

Acquired glucose 6-phosphate dehydrogenase (G6PD) deficiency in a patient with Chronic Myelomonocytic Leukemia

Naville AS, Lazaro E, Boutin J, *et al.*
Br J Haematol. 2022 Jan 6. doi: 10.1111/bjh.18037

Acute hemolysis and methemoglobinemia secondary to fava beans ingestion in a patient with G6PD deficiency: A case report of a rare co-occurrence

Al-Dubai H, Al-Mashdali A, Hailan Y.
Medicine (Baltimore). 2021 Nov 24;100(47):e27904. doi: 10.1097/MD.0000000000027904

Enhanced Eryptosis in Glucose-6-Phosphate Dehydrogenase Deficiency

Bouguerra G, Talbi K, Trabelsi N, *et al.*
Cell Physiol Biochem. 2021 Dec 11;55(6):761-772. doi: 10.33594/000000474

Influenza Virus Down-Modulates G6PD Expression and Activity to Induce Oxidative Stress and Promote Its Replication

De Angelis M, Amatore D, Checconi P, *et al.*
Front Cell Infect Microbiol. 2022 Jan 6;11:804976. doi: 10.3389/fcimb.2021.804976

Association between Glucose-6-Phosphate Dehydrogenase Deficiency and Asthma

Fois A, Dore MP, Manca A, Scano V, Pirina P, Pes GM.
J Clin Med. 2021 Nov 29;10(23):5639. doi: 10.3390/jcm10235639

Stabilization of glucose-6-phosphate dehydrogenase oligomers enhances catalytic activity and stability of clinical variants

Garcia AA, Mathews II, Horikoshi N, *et al*.
J Biol Chem. 2022 Jan 19:101610. doi: 10.1016/j.jbc.2022.101610

An International Survey of Glucose-6-Phosphate Dehydrogenase Laboratory Reporting Practices: Implications for Tafenoquine Eligibility Assessment

Genzen JR, Nwosu A, Long T, Murphy H, Alter DN.
Arch Pathol Lab Med. 2022 Jan 28. doi: 10.5858/arpa.2021-0276-CP

Incorporating G6PD genotyping to identify patients with G6PD deficiency

Morris SA, Crews KR, Hayden RT, et al.
Pharmacogenet Genomics. 2021 Oct 22. doi: 10.1097/FPC.0000000000000456

Concurrence of glucose-6-phosphate dehydrogenase deficiency in pregnancy

Sarkar A, Rohilla M, Kumari S.
J Obstet Gynaecol. 2022 Jan 24:1-4. doi: 10.1080/01443615.2021.2024157

Population pharmacogenomics: an update on ethnogeographic differences and opportunities for precision public health

Zhou Y, Lauschke VM.
Hum Genet. 2021 Oct 15. doi: 10.1007/s00439-021-02385-x

Hematological and molecular analysis of patients with G6PD deficiency revealed coexistent hereditary spherocytosis and alpha thalassemia

Rizo-delaTorre LDC, Herrera-Tirado IM, Hernández-Peña R, Ibarra-Cortés B, Perea-Díaz FJ.
Ann Hum Genet. 2021 Nov 29. doi: 10.1111/ahg.12451

Déficit en pyruvate kinase

Red Blood Cell Metabolism in Pyruvate Kinase Deficient Patients

Roy MK, Cendali F, Ooyama G, Gamboni F, Morton H, D'Alessandro A.
Front Physiol. 2021 Oct 21;12:735543. doi: 10.3389/fphys.2021.735543

Who should be eligible for gene therapy clinical trials in red blood cell pyruvate kinase deficiency (PKD)?: Toward an expanded definition of severe PKD

Schwartz JD, Barcellini W, Grace RF, *et al.*
Am J Hematol. 2022 Mar 1;97(3):E120-E125. doi: 10.1002/ajh.26458

Pyruvate Kinase Deficiency

Enegela OA, Anjum F.
2021 Dec 8. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan

Autres (ou plusieurs) enzymopathies érythrocytaires

Hereditary myopathies associated with hematological abnormalities.

Beecher G, Fleming MD, Liewluck T.
Muscle Nerve 2022 Jan 5. doi: 10.1002/mus.27474

Diagnosis and clinical management of enzymopathies.

Luzzatto L.
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