Bulletin recherche

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FILIÈRE SANTÉ MALADIES RARES

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Evolution de la pathologie de l'artère carotide interne extracrânienne chez les enfants drépanocytaires

Contexte et objectif

L'artériopathie cérébrale est une complication fréquente de la drépanocytose qui survient tôt dans l'enfance et augmente le risque d'accident vasculaire cérébral (AVC) et d'infarctus cérébral silencieux. Les atteintes de la circulation antérieure intracrânienne sont les plus fréquentes et peuvent être détectées de manière précoce par le Doppler transcrânien, examen d'imagerie qui mesure la moyenne des vitesses maximales au cours du temps (TAMV) des globules rouges circulants dans les artères dont le seuil pathologique est de 200 cm/s. Grâce à la surveillance des artères par le Doppler transcrânien et l'utilisation de thérapeutiques adaptées telles que les programmes transfusionnels, recommandés en cas de TAMV intracrânienne pathologique et l'hydroxyurée, le nombre d'AVC a considérablement été réduit mais le nombre d'infarctus cérébraux silencieux demeure élevé. Il a récemment été montré que le segment cervical des artères carotides internes (ACIc) peut également être sténosé et être responsable d'AVC et d'infarctus silencieux. Une TAMV >=160 cm/s de l'ACIc est associée à une sténose visible en angiographie IRM (ou ARM, examen d'imagerie par résonance magnétique permettant de visualiser les artères ou les veines), s'accompagnant souvent d'une angulation localisée de l'artère appelée plicature. L'objectif de cette étude était de décrire l'évolution de l'artériopathie de l'ACI cervicale entre 2011 et 2021 dans la cohorte d'enfants drépanocytaires suivis à l'hôpital universitaire Robert Debré APHP en fonction de la thérapie (simple surveillance, programme transfusionnel ou hydroxyurée). Les patients porteurs d'une drépanocytose HbSS ou d'une S-β° thalassémie bénéficient en routine d'un écho-Doppler transcrânien et cervical annuel dès la 2ème année de vie et d'une IRM incluant l'étude des artères intracrâniennes et des ACI cervicales en cas de Doppler anormal.

Méthode

Cette étude rétrospective a inclus 54 patients présentant une TAMV de l'ACIc >=160 cm/s et qui ont eu un suivi par Doppler et IRM supérieur à 1 an. L'âge médian du premier Doppler cervical anormal était de 4,9 ans (1,6 – 17,1 ans). Le suivi médian entre le premier et dernier examen IRM était de 4,7 ans. Durant le suivi, 4 patients n'ont eu aucune intensification du traitement, 10 ont été placés exclusivement sous hydroxyurée, 17 exclusivement sous programme transfusionnel et 27 ont reçu un traitement séquentiel (pas d'intensification, hydroxyurée puis programme transfusionnel ou hydroxyurée et programme transfusionnel si la vitesse ne diminuait pas). La sténose de l'ACIc a été évaluée selon l'échelle du NASCET (North American Symptomatic Carotid Endarterectomy Trial).

Résultats

La première angiographie a révélé que 89 % des patients présentaient une sténose de l'ACIc contre 72 % lors de la dernière angiographie. L'occlusion de l'ACIc s'est produite chez 5 patients malgré le programme transfusionnel, dont 3 patientes avec une sténose ostiale pseudomembraneuse. Les patients n'ayant eu aucune intensification du traitement ont été 8 % à avoir un score de sténose amélioré, ceux sous hydroxyurée 20 % et ceux sous programme transfusionnel 48 %. La variation moyenne annuelle du score de sténose était de 0,4 pour les patients sans intensification, 0,2 sous hydroxyurée et -0,18 sous programme transfusionnel, la valeur négative reflétant une diminution de la sténose et donc une amélioration clinique. Les analyses statistiques de régression de Cox ont montré que le score initial de la sténose de l'ACIc était un facteur prédictif du risque de la survenue d'infarctus cérébral silencieux.
Cette étude met en évidence que la prévention des AVC et des infarctus cérébraux silencieux chez les enfants drépanocytaires ayant des sténoses de l'ACI cervicale peut être améliorée par la surveillance des artères cervicales et la mise en place de programmes transfusionnels.

L’étude en quelques chiffres :

- 54 patients inclus dont 8 ayant déjà un AVC
- 4,7 an de suivi médian par patient entre 2011 et 2021
- 89 % des patients présentaient une sténose à la première angiographie du cou contre 72 % lors du dernier examen

Amélioration du score de sténose chez :

- 8 % des patients sans intensification du traitement
- 20 % des patients sous hydroxyurée
- 48 % des patients sous programme transfusionnel

Cette étude a fait l’objet d’une publication en avril 2022 dans le journal Stroke (https://doi.org/10.1161/STROKEAHA.121.037980)

Dr Suzanne Verlhac
Service d'Imagerie Pédiatrique,
Centre de référence de la drépanocytose
Hôpital Universitaire Robert Debré, APHP, Paris, France

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

On sait depuis les années 1990 que l'AVC qui est une complication particulièrement redoutée et précoce de la drépanocytose, est lié à une artériopathie sténo-occlusive des artères de gros et moyen calibre de la circulation carotidienne intracrânienne. Nous avons rapporté en 2014 que le segment cervical des artères carotides internes pouvait aussi être atteint et qu'une anomalie pouvait être détectée par écho-Doppler en utilisant une technique particulière. Une vitesse ≥160 cm/s était associée à une sténose visible sur la séquence d'angiographie de l'examen IRM, s'accompagnant souvent d'une angulation localisée de l'artère appelée plicature. Cette artériopathie cervicale risque de provoquer un AVC mais aussi des infarctus dits silencieux qui sont des lésions ischémiques visibles en IRM, ne se traduisant pas par des signes neurologiques moteurs ou sensitifs, mais pourvoyeurs d'une atteinte cognitive. Depuis 2011, nous étudions systématiquement l'étage cervical par écho-Doppler et IRM chez les enfants SS et Sβ0, en même temps que les artères intracrâniennes. Alors que le traitement par programme transfusionnel des patients ayant une vitesse intracrânienne anormale et donc à haut risque d'AVC est admis par tous et fait l'objet de recommandations internationales, la prise en charge des anomalies cervicales n'est pas codifiée.

Dans cette étude rétrospective d'une série de cas, nous nous sommes intéressés aux patients suivis à l'hôpital Robert Debré ayant une vitesse anormale à l'écho-Doppler du segment cervical d'une ou des deux artères carotides internes et nous avons analysé l'évolution des paramètres d'écho-Doppler transcrânien et d'IRM en fonction du traitement.

Nous avons fait trois constatations notables :

1°) Le rétrécissement de l'artère est localisé et situé le plus souvent au 1/3 moyen du trajet artériel. Il est souvent associé à une plicature, comme si la paroi de l'artère avait perdu son élasticité. Fait notable, nous avons constaté chez quelques patients sous programme transfusionnel que cette déformation pouvait se corriger en même temps que la sténose disparaissait.

2°) Cette artériopathie cervicale risque de provoquer un AVC, puisque 5 patients sur les 8 avec AVC de cette cohorte avait une atteinte cervicale isolée et plus fréquemment encore des infarctus silencieux qui concernaient 31 % des patients au moment de la découverte du Doppler cervical anormal, donc précoces puisque l'âge médian des patients étaient de 5 ans. Et au cours du suivi, un patient a eu un nouvel AVC et six de nouveaux infarctus silencieux.

3°) Le programme transfusionnel est plus efficace sur les sténoses artérielles cervicales que l'hydroxycarbamide : 48 % des patients ont été améliorés sous programme transfusionnel contre 20 % avec hydroxycarbamide et 8 % avec la prise en charge habituelle.

Comment expliquez-vous que les transfusions soient plus efficaces que l'hydroxycarbamide pour réduire les sténoses de l'ACI cervicale ?

Pour essayer de répondre à cette question, il faut se pencher sur le mécanisme d'action de ces traitements. Les transfusions apportent des globules rouges contenant de l'hémoglobine adulte normale HbA et diluent en quelque sorte l'hémoglobine mutée HbS qui a l'effet nocif de modifier les propriétés des globules rouges en réduisant leur durée de vie et leur capacité à circuler dans les vaisseaux sanguins. Avec les transfusions, l'anémie est améliorée, les conditions circulatoires dans les vaisseaux aussi, donc les parois artérielles subissent moins d'agressions, ce qui stoppe la progression de la sténose de l'artère, voire la fait disparaitre. L'hydroxycarbamide quant à elle agit en stimulant la production de l'hémoglobine fœtale F, dont la production normalement chute après la naissance. La coexistence d'hémoglobine foetale diminue la polymérisation de l'HbS et l'hémolyse et l'obstruction des petits vaisseaux. L'hydroxycarbamide agit aussi par d'autres mécanismes qui sont la diminution de la production des globules blancs et des réticulocytes et la diminution de l'adhérence des globules rouges aux cellules de l'endothélium vasculaire. L'efficacité de l'hydroxycarbamide sur la prévention et/ou l'atténuation des crises vaso-occlusives et des syndromes thoraciques est démontrée. Concernant la prévention de l'AVC, il a été montré qu'elle diminuait un peu les vitesses circulatoires cérébrales grâce à l'amélioration de l'anémie, mais qu'elle était moins efficace que le programme transfusionnel pour la prévention des récidives des AVC avec un risque persistant de 20 %. Elle est réservée aux situations d'impasse transfusionnelle et en l'absence de donneur HLA compatible. Notons que la greffe de moelle osseuse est encore plus efficace sur l'artériopathie cérébrale que le programme transfusionnel comme l'a montré le protocole DREPAGREFFE, comparant la greffe et le programme transfusionnel chez 60 patients sans antécédent d'AVC mais ayant une vitesse pathologique à l'écho-Doppler transcrânien et cervical, randomisés en deux groupes : 28 enfants ayant un donneur HLA identique ont bénéficié d'une greffe et 32 enfants ont été placés sous programme transfusionnel.

Vous avez publié avec le Dr Bernaudin un article en septembre 2022 dans Frontiers in Neurology (DOI 10.3389/fneur.2022.846596**) issu d'une étude prospective qui étudie les facteurs de risque des artériopathies cérébrales intra- et extracrâniennes chez des enfants drépanocytaires à l'aide de l'écho-Doppler. Pourriez-vous nous présenter les principaux résultats ?**

L'objectif de cette étude était de définir le risque pour un enfant porteur d'une drépanocytose dépistée à la naissance d'avoir une anomalie des artères carotides internes à l'étage cervical à l'âge de 10 ans et d'identifier les facteurs participant au développement de cette artériopathie. Pour cela nous avons utilisé la base de données qui permet le suivi des patients drépanocytaires du centre de référence de l'hôpital intercommunal de Créteil, après accord du comité d'éthique local. Il s'agit d'une recherche non interventionnelle dans laquelle tous les actes et traitements font partie de la prise en charge habituelle et qui ne comporte ni risque ni contrainte pour les patients. L'étude a montré que le risque, appelé en terme statistique l'incidence cumulée, pour un patient d'avoir une accélération anormale avec une vitesse ≥160 cm/s dans au moins une des artères carotides internes (ACI) à l'étage cervical était de 17,4 % à l'âge de 10 ans et n'augmentait plus dans les années suivantes et que la survenue de l'anomalie était très précoce dès la deuxième année de vie. Cette vitesse anormale a permis de dépister une sténose anatomique par IRM dans la majorité des cas. L'incidence cumulée d'une sténose vue en IRM était de 12,3 % à 10 ans. Le plus souvent l'anomalie de l'ACI cervicale était isolée sans anomalie associée des artères intracrâniennes. Ce qui fait que l'étude des ACI cervicales par écho-Doppler et IRM a permis de détecter 13,5 % de patients à risque d'AVC, qui n'étaient pas détectés par Doppler transcrânien seul.

Cette étude a permis aussi de rapporter la valeur prédictive du bilan hématologique basal réalisé au cours de la deuxième année de vie du patient. Une hémoglobinémie basse, un nombre élevé de réticulocytes et une hyperleucocytose étaient des facteurs de risque d'une artériopathie intracrânienne, alors qu'une hémoglobinémie basse et l'haplotype Sénégal étaient des facteurs de risque d'une artériopathie cervicale.

Quelle est la prise en charge des cas litigieux ou atypiques ?

L'indication d'un programme transfusionnel nous semble indiscutable en cas de vitesse cervicale de 200 cm/sec ou plus. En cas de vitesses cervicales comprises entre 160 et 200 cm/sec, on sait que le risque de sténose est important. Jusqu'à présent, notre pratique était de faire une IRM avec étude des artères cervicales et de débuter un programme transfusionnel en cas de sténose cervicale objectivée en IRM et de mettre les patients sans sténose sous hydroxycarbamide si ce traitement n'était pas encore entrepris. Néanmoins le Dr Bernaudin vient de présenter à l'ASH le risque très important d'infarctus silencieux chez ces enfants avec une vitesse >=160 même sans sténose et nous pensons désormais qu'il serait plus approprié de les traiter d'emblée par programme transfusionnel, ce qui permet de plus de réaliser l'IRM en toute sécurité chez des enfants de moins de 5 ans nécessitant une sédation.

Pensez-vous que la prise en charge par Doppler et programme transfusionnel peut être adoptée pour les patients ayant une vitesse anormale des artères carotides internes extracrâniennes ?

Oui. Actuellement, un examen annuel écho-Doppler est recommandé dans la prise en charge d'un enfant porteur d'une drépanocytose SS ou S-β0 dès la deuxième année de vie jusqu'à l'âge de 16-18 ans et est pratiqué de manière plus fréquente en cas de vitesse proche du seuil d'anomalie. L'exploration de l'étage cervical ne prolonge l'examen que de quelques minutes. L'IRM n'est pas indiquée de manière systématique. Elle est faite chez les enfants dont le Doppler est anormal (intracrânien ou cervical) et bien entendu en cas de troubles faisant suspecter un AVC comme un déficit moteur brutal ou des difficultés à parler. Mais il me semble de plus pertinent de faire une IRM systématiquement aux âges clés de 5-6 ans, 10 et 15 ans, pour rechercher les infarctus cérébraux silencieux dont la présence incite à intensifier le traitement. L'étude des artères cervicales est faite par une séquence supplémentaire lors de l'examen d'IRM qui ne rallonge que très peu la durée de l'examen. Le fait que l'artériopathie cervicale soit le plus souvent isolée, sans atteinte intracrânienne associée incite à la rechercher de manière systématique du fait de son risque élevé d'AVC et d'infarctus silencieux. Ce protocole est appliqué depuis de nombreuses années à l'hôpital intercommunal de Créteil et à l'hôpital Robert Debré et nous conseillons lors des réunions de concertation pluridisciplinaire (RCP) à nos collègues français de l'appliquer également. La prise en charge de l'artériopathie cervicale est un enjeu de taille pour préserver le cerveau de ces enfants afin qu'ils puissent parvenir à l'âge adulte sans séquelle neuro-cognitive.

Appels à projets

Appel à Projets « Genomics of rare diseases

Budget	Budget : non précisé
Durée	NC
Date limite de dépôt des dossiers	19 janvier 2023, 17h (heure de Paris)
Eligibilité	L'investigateur principal doit appartenir à une équipe de recherche française, affiliée au milieu universitaire (équipe de recherche travaillant dans des universités, d'autres établissements d'enseignement supérieur ou d'instituts de recherche) et/ou au secteur clinique/secteur de santé publique (équipe de recherche travaillant dans les hôpitaux/organismes de santé publiques). Les scientifiques en début de carrière sont encouragés à postuler en tant qu'investigateur principal.
Objectif	Soutenir des projets de recherche utilisant le séquençage de nouvelle génération pour élucider les bases génétiques et moléculaires des maladies rares.

→ Plus d'informations : https://fondation-maladiesrares.org/wp-content/uploads/2022/11/2022-GenOmics_call_text.pdf

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Programme conjoint européen sur les maladies rares (EJP RD) - Appel à propositions 2023
« Études d'histoire naturelle répondant aux besoins non satisfaits dans les maladies rares »

Budget	NC
Durée	NC
Date limite de dépôt des dossiers	15 février 2023
Eligibilité	Les propositions de recherche doivent couvrir au moins un des domaines suivants : - Estimation de la prévalence de la maladie ; - Identification de biomarqueurs/compagnons pour le diagnostic/pronostic d'une RD ; - Identification de biomarqueurs/indicateurs/prédicteurs d'une maladie rare ou d'un groupe de troubles (par exemple ayant la même étiologie) d'apparition/progression (y compris la collecte de données ou de variables génétiques, physiologiques, environnementales…) ; - Identification des critères d'évaluation pertinents pour les études futures qui incluent des biomarqueurs potentiels, l'interrogation des résultats rapportés par les patients (PRO) et des mesures de la qualité de vie ; - Identification de biomarqueurs/variables pour des approches thérapeutiques (pharmacologie, repositionnement de médicaments, thérapie génique, thérapie ARN, thérapie cellulaire, dispositifs médicaux…).
Objectif	Collecter et analyser des données complètes sur les patients afin de définir des cibles pour de futures thérapies, en tenant compte de l'innovation, de la sécurité et de l'efficacité.

→ Plus d'informations : https://www.ejprarediseases.org/joint-transnational-call-2023/

Fonds de dotation CSL Behring pour la recherche

Budget	• 1 à 3 bourse(s) de recherche en Immunologie : 20 000 euros chacune • 1 à 2 bourse(s) de recherche en Hémostase : 20 000 euros chacune • 1 bourse de mobilité internationale en immunologie ou en hémostase : 25 000 euros
Durée	NC
Date limite de dépôt des dossiers	15 février 2023
Eligibilité	Immunologie : programme de recherche sur le rôle des anticorps pathogènes et/ou des immunoglobulines thérapeutiques dans les maladies auto-immunes ou inflammatoires, les déficits immunitaires, les greffes ou la transplantation. Hémostase : programme de recherche dans les maladies hémorragiques constitutionnelles ou acquises. Mobilité internationale : chercheur travaillant sur les maladies auto-immunes ou inflammatoires, les déficits immunitaires, les greffes ou la transplantation, les maladies hémorragiques constitutionnelles ou acquises.
Objectif	Promouvoir et aider des initiatives exemplaires et d'intérêt général visant la recherche générale visant la recherche médicale et notamment de soutenir financièrement des programmes de recherche dans le domaine des maladies rares et en particulier en immunologie et en hémostase.

→ Plus d'informations :
https://www.fondsdedotationcslbehring.fr/wp-content/uploads/2022/12/CSL_22_35_DOTATION_REGLEMENT_N7_10.pdf

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Prix Alnylam pharmaceutical 2023 « ARN interférent et maladies rares »

Budget	20 000 €
Durée	3 ans
Date limite de dépôt des dossiers	30 mars 2023, 17h (heure de Paris)
Eligibilité	Chercheur confirmé, responsable scientifique d'une équipe rattachée à un organisme de recherche français contribuant par ses recherches à des avances majeures dans le domaine des maladies rares.
Objectif	Récompenser un projet de recherche innovant dans le domaine des maladies rares utilisant la technologie de l'ARN interférent, portant sur l'un des thèmes suivants : • Recherche translationnelle développant une approche d'ARN interférence et utilisant un modèle animal ou cellulaire. • Vectorisation innovante d'ARN interférents ciblant un organe ou un tissu.

→ Plus d'informations :
https://filiere-mcgre.fr/wp-content/uploads/2022/12/Prix-Alnylam-Pharmaceutical-2023-ARN-interferent-et-maladies-rares-.pdf

Instituts Hospitalo-Universitaires (IHU 3) – Appel à projets – 2022

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://anr.fr/fr/detail/call/instituts-hospitalo-universitaires-ihu-3-appel-a-projets-2022/?utm_source=Newsletter&utm_medium=Email&utm_campaign=avril-2022

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 des articles parus de juillet à novembre 2022 inclus (c'est-à-dire entrés dans PubMed entre le 4 juillet et le 30 novembre 2022, sauf exceptions).

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Filière de santé maladies rares MCGRE - Hôpital Henri Mondor
1 rue Gustave Eiffel, 94010 Créteil
contact@filiere-mcgre.fr- www.filiere-mcgre.fr