Bulletin recherche

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FILIÈRE SANTÉ MALADIES RARES

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Effets à long terme de la thérapie génique lentivirale dans le traitement des β-hémoglobinopathies : l'essai HGB-205

Contexte et objectif

La β-thalassémie dépendante des transfusions (TDT) et la drépanocytose sont des β-hémoglobinopathies dues à la mutation du gène codant pour la sous-unité β de l'hémoglobine adulte (HbA). Ces pathologies se caractérisent toutes les deux par une anémie hémolytique (destruction des globules rouges) mais également par une érythropoïèse inefficace (impossibilité de fabriquer des globules rouges) pour les TDT et la polymérisation de HbS pour la drépanocytose, entrainant des crises vaso-occlusives, des syndromes thoraciques aigus et des atteintes d'organe. A ce jour, le seul traitement curatif ne peut être proposé qu'à moins de 20 % des patients car il repose sur la transplantation de cellules souches hématopoïétiques (qui produisent les cellules du sang) issues d'un donneur génétiquement compatible de la famille du patient (pour éviter le rejet de greffe). La thérapie génique par transplantation de cellules souches hématopoïétiques autologues (qui proviennent du patient) génétiquement modifiées est une piste thérapeutique prometteuse car elle résoudrait les problèmes de compatibilité et pourrait ainsi être accessible à tous les patients. Dans le cadre de la stratégie de l'essai thérapeutique HGB 205 de l'entreprise Bluebirdbio, les cellules souches hématopoïétiques sont génétiquement modifiées par l'intégration dans le génome du patient d'une globine dite thérapeutique (βT87Q) grâce à un vecteur lentiviral. L'hémoglobine produite présente l'avantage d'empêcher la polymérisation de l'hémoglobine S ou de compenser l'hémoglobine manquante dans le cas des β-thalassémies. L'étude publiée dans *Nature medicine* avait pour but d'analyser précisément les effets de la thérapie génique chez les patients français de l'étude HGB-205 (NCT02151526).

Méthode

L'essai clinique interventionnel HGB-205 de phase I/II a été réalisé de manière non-randomisée et ouverte (les patients et médecins avaient connaissance que tous les patients avaient reçu le traitement) à l'hôpital pédiatrique Necker (Paris) de 2013 à 2015 chez des patients atteints de TDT ou de drépanocytose. Cet essai s'est poursuivi par 2 études observationnelles permettant un suivi à long terme dont les données ont été collectées jusqu'en mars 2020 chez les 4 patients atteints de TDT et août 2020 chez les 3 patients drépanocytaires. Après le prélèvement de la moelle osseuse, la modification génétique des cellules souches hématopoïétiques et leur intégration dans le vecteur lentiviral BB305, les patients ont reçu un traitement myeloablatif par injection intraveineuse de Busulfan pour préparer la greffe. Les taux des neutrophiles et des plaquettes ont ensuite été normalisés à 32 et 51 jours respectivement et les patients ont eu un suivi médian de 4,5 ans.

Résultats

Aucun effet indésirable lié à l'utilisation du vecteur lentiviral n'a été observé chez les 7 patients.
Le besoin transfusionnel des patients TDT a rapidement été supprimé et le suivi à long terme a permis de constater une réduction de la surcharge ferrique et des signes morphologiques de dysérythropoïese.
L'amélioration des symptômes et la correction des paramètres biologiques ont pu être observées chez deux patients drépanocytaires mais le troisième a nécessité un traitement par hydroxyurée et des échanges transfusionnels du fait de la réapparition de crises vaso- occlusives.
La thérapie génique par addition de gène est une piste thérapeutique très prometteuse pour les patients TDT. Le suivi à long terme des patients drépanocytaires a prouvé l'efficacité de ce traitement chez 2 patients sur trois encourageant ainsi l'optimisation de cette technique.

L'étude en quelques chiffres :

Début de l'étude en 2013 et présentation des données collectées jusqu'en 2020

- 4 patients TDT âgés de 17 à 19 ans
- 3 patients drépanocytaires âgés de 13 à 21 ans
- 2/3 patients drépanocytaires n'ont plus de symptômes drépanocytaires
- 4/4 patients atteints de TDT n'ont plus besoin de transfusion

Cette étude a fait l'objet d'une publication en janvier 2022 dans le journal *Nature medecine* : https://doi.org/10.1038/s41591-021-01650-w.

Un dossier spécial sur la thérapie génique des hémoglobinopathies a été publié dans le *New Globinoscope n°4*, gratuitement disponible en ligne https://filiere-mcgre.fr/actualites/new-globinoscope-n4/ ou sur demande en version imprimée à contact@filiere-mcgre.fr.

Pr Pablo Bartolucci

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Pouvez-vous nous décrire le contexte de cette étude ?

La thérapie génique par addition de gène consiste à prélever des cellules souches des cellules sanguines du patient, à les modifier *in vitro* en insérant dans leur ADN le gène thérapeutique au moyen d'un vecteur lentiviral puis à réinjecter les cellules modifiées chez le patient. Le vecteur lentiviral est dérivé du virus du VIH débarrassé de tout effet pathogène. Dans le cas des β-thalassémies ou de la drépanocytose, il s'agit de maladies liées à des mutations de la chaîne β de l'hémoglobine qu'il faut traiter.

Le lentivirus utilisé dans cette thérapie génique inclut le gène normal de la β-globine auquel a été ajoutée une variation génétique de l'hémoglobine fœtale (βT87Q) pour obtenir un effet anti-polymérisant. Le but est de produire une « super-hémoglobine » pouvant à la fois i) augmenter la production de la β-globine qui va s'associer à la chaine α pour traiter la β-thalassémie et ii) entrer en compétition avec l'HbS pour empêcher sa polymérisation afin de traiter la drépanocytose.

Ces travaux ont débouché sur les essais de Bluebirdbio qui se sont révélés positifs après une phase d'amélioration des procédures à laquelle a contribué l'équipe du Pr Cavazzana.

Dans cette étude, les équipes de Marina Cavazzana, d'Annarita Miccio, de Wassim El Nemer et la mienne ont collaboré pour évaluer quels étaient les effets bénéfiques à plus long terme concernant la qualité des globules rouges et la distribution de cette hémoglobine thérapeutique chez les trois patients drépanocytaires et les quatre patients thalassémiques traités en France.

Quels sont les résultats majeurs de cette étude ?

Deux patients drépanocytaires ont été cliniquement guéris, mais ce fut un échec pour le troisième patient en raison de la faible quantité de transduction lentivirale (quantité de virus ayant intégré la cellule). Concernant les 2 patients drépanocytaires pour lesquels ce fut un succès, le premier avait un bon niveau de transduction avec une quantité d'hémoglobine thérapeutique suffisante. Le résultat le plus surprenant concerne le second patient drépanocytaire dont le niveau de transduction lentivirale était relativement médiocre (0,3 copies par cellule alors que le minimum probablement nécessaire est de 1 à 2 copies par cellule) mais qui avait par ailleurs une expression de l'HbF. Il y a donc eu chez ce patient une complémentarité entre l'HbF dans les cellules mal transduites et l'hémoglobine thérapeutique aboutissant à la disparition de ses symptômes drépanocytaires. L'efficacité de la thérapie génique ne se résume donc pas au nombre de copies du vecteur par cellule mais est en réalité beaucoup plus complexe. Grâce aux différentes techniques développées dans nos laboratoires, nous avons pu montrer que les patients « guéris » avaient une amélioration de tous les paramètres : déformabilité des globules rouges,

affinité de l'hémoglobine pour l'oxygène, falciformation, niveaux d'hémolyse. Le niveau de transduction lentivirale et l'expression globale des différentes hémoglobines ont aussi été évalués.
L'expérience s'est également révélée positive chez les patients thalassémiques pour lesquels les besoins transfusionnels ont été supprimés et les paramètres biologiques se sont améliorés avec une dysérythropoïèse contrôlée et une surcharge en fer diminuée.
Cette étude a été menée chez les premiers malades bénéficiant de cette technique thérapeutique. Depuis, de nouvelles procédures in vitro ont permis d'améliorer la capacité du virus à entrer dans la cellule.

Quelles sont les suites de l'étude ?

Il devait y avoir la suite de l'étude chez les patients drépanocytaires en France et la mise en place des premiers traitements médicamenteux chez les patients β-thalassémiques. Plusieurs centres avaient déjà été sélectionnés et des patients sollicités étaient sur liste d'attente. Mais en septembre 2021, la compagnie pharmaceutique Bluebirdbio, propriétaire de ce traitement a décidé de partir d'Europe. Ceci n'était pas étranger à l'apparition de deux cas de leucémie aigüe chez des patients qui avaient été traités aux Etats Unis.
Cependant les analyses de ces patients ont révélé que les gènes apportés par le virus n'étaient pas directement responsables de la leucémie mais que le conditionnement par chimiothérapie avait pu les favoriser.
En France, des programmes de dépistage ont été mis en place avant le traitement chez les patients en vue d'une thérapie génique, et de plus en plus pour les allogreffes, pour rechercher des clones myéloïdes circulants qui pourraient être des facteurs prédisposant à ce risque, en particulier après chimiothérapie. Celle-ci est indispensable pour « remettre à zéro » les cellules souches afin d'optimiser les chances de succès de la thérapie génique mais elle peut entrainer une sélection de certains clones qui pourraient avoir un avantage compétitif qu'ils n'ont pas en temps normal.
Bluebirdbio s'est également retiré de l'Europe en raison du problème du prix de remboursement. Une discussion sociétale de plus large ampleur avec des économistes de la santé, des personnes impliquées dans l'éthique, des professionnels de santé et des associations de patients, est indispensable pour réellement mesurer les bénéfices / risques de ces traitements et le coût pour la société en comparaison du bénéfice rendu. A ce jour, aucune étude socio-économique indépendante n'a été menée pour faire le calcul en termes d'économies de médicaments, d'hospitalisations, mais aussi concernant les arrêts de travail, les gardes d'enfant, les absentéismes, le niveau scolaire… Le développement de ce champ de recherche socio-économique et éthique est crucial pour l'avenir de la thérapie génique.

Quelles sont les perspectives de la thérapie génique ?

Un article sur le suivi de la thérapie génique mené au Boston Children's Hospital auquel nous avons collaboré avec l'équipe de David Williams -Harvard Medical School- devrait être prochainement publié. Il s'agit d'une thérapie génique d'addition qui cette fois-ci n'ajoute pas un gène d'hémoglobine mais vient empêcher la répression du gène de la gamma-globine augmentant ainsi la production de l'HbF qui a un puissant effet anti polymérisant sur l'HbS. Après la naissance, la désactivation du gène de la gamma-globine concomitante à l'activation du gène de la β-globine conduit à un effondrement des taux d'HbF au profit des taux de l'HbA. La compréhension des mécanismes moléculaires sous-jacents à cette commutation des hémoglobines F et A a permis le développement de nouvelles stratégies de thérapie génique. L'ajout d'un shRNA - qui est un ARN interférent - va réprimer l'expression du gène BCL11A qui

contrôle la commutation des hémoglobines F et A. Cette technologie n'est pas encore disponible en Europe.
Parallèlement, la société Vertex développe une stratégie basée sur la technologie CRISPR/Cas9. Elle consiste à diminuer l'expression de la protéine BCL11A par destruction ciblée dans le tissu érythroïde du gène correspondant afin d'augmenter les taux d'HbF chez les patients drépanocytaires et β-thalassémiques. On espère que cette stratégie arrivera en France prochainement.
Enfin l'équipe d'Annarita Miccio en France, ou la société Beam therapeutics aux Etats Unis, travaillent sur des stratégies « base editing » : au lieu de casser l'ADN, on modifie juste une base (molécule constituant l'ADN). Cela permet de modifier l'hémoglobine ou éventuellement d'augmenter les taux d'HbF sans avoir les risques liés à la cassure de l'ADN.

→ Y-a-t-il une technique plus prometteuse que les autres ?

Les stratégies reposant sur l'ajout d'une hémoglobine thérapeutique ou sur l'augmentation de l'expression de l'HbF par l'addition de type ShRNA ou en abimant directement le gène BCL11A par CRISPR/Cas9 ont toutes montré leur efficacité thérapeutique. Cependant, les risquent existent et ils sont probablement différents en fonction des stratégies utilisées.
La thérapie génique est donc une immense source d'espoir de guérison mais nous devons rester vigilants et prudents pour bien appréhender la balance entre les bénéfices et les risques.

Appels à projets

MESSIDORE 2022 - AAP

Budget	50 000€ minimum, sans maximum, dans la limite du budget total disponible pour le programme
Durée	12 à 48 mois
Date limite de dépôt des dossiers	14 octobre 2022, 16h
Eligibilité	Réseau scientifique constitué de collaborateurs européens ou internationaux avec au moins un partenaire français, couvrant des sujets de toutes les disciplines.
Objectif	• Soutenir le développement d'essais cliniques innovants reposant sur des méthodologies nouvelles ; • Soutenir des projets s'appuyant sur des études, des bases de données ou des collections biologiques existantes.

→ Plus d'informations :
https://pro.inserm.fr/nouveau-programme-strategique-de-recherche-collaborative-en-sante

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Prix de la recherche médicale de la Fondation de France/Jean Valade 2023

Budget	• 100 000 euros décerné à un chercheur sénior (45 ans et plus) • 50 000 euros pour un jeune chercheur (âgé de moins de 45 ans).
Durée	NC
Date limite de dépôt des dossiers	19 octobre 2022, 17h
Eligibilité	Chercheurs (DR, PU ou PU-PH) titulaires, rattachés à une équipe, exerçant dans un laboratoire de recherche à but non lucratif. Travaux de recherche dans les thématiques suivantes : • (…) ; • Douleur ou fin de vie ; Travaux ayant débouché sur des résultats originaux, (recherche fondamentale, clinique ou sciences humaines et sociales) ; Application possible à l'Homme : clinique, épidémiologique ou amélioration des pratiques.
Objectif	Faire progresser la découverte médicale

→ Plus d'informations :
https://www.fondationdefrance.org/fr/appels-a-projets/prix-de-la-recherche-medicale-de-la-fondation-de-france-jean-valade-2023

Montage de reseaux scientifiques europeens ou internationaux (MRSEI) - AAP, édition 2022

Budget	30000€ maximum
Durée	24 mois
Date limite de dépôt des dossiers	18 octobre 2022, 13h CEST
Eligibilité	Réseau scientifique constitués de collaborateurs européens ou internationaux avec au moins un partenaire français, couvrant des sujets de toutes les disciplines.
Objectif	Constituer un réseau scientifique européen ou international, coordonné par une équipe française.

→ Plus d'informations : https://anr.fr/fr/detail/call/appel-a-projets-montage-de-reseaux-scientifiques-europeens-ou-internationaux-mrsei-edition-20/

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	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 : frm_notex_aje2022.pdf

Instituts hospitalo-universitaires (IHU 3) – APPEL À PROJETS – 2022

Budget	300M€, pour six nouveaux Instituts Hospitalo-Universitaires (IHU)
Durée	120 mois
Date limite de dépôt des dossiers	7 novembre 2022
Eligibilité	Voir Texte de l'AAP
Objectif	• Viser l'excellence mondiale en matière de recherche, d'enseignement, de soin, de prévention dans une thématique définie ;

	• Mettre au cœur de chaque projet une dynamique du laboratoire vers le patient et du patient vers le laboratoire : • Disposer d'une file active de patients significative dans la thématique proposée et d'une prise en charge du patient en cohérence avec le projet scientifique ; • Impliquer de façon harmonieuse cliniciens et chercheurs dans l'ensemble des activités de l'IHU, en favorisant leur participation conjointe aux activités de recherche translationnelle ou clinique ; • S'assurer du caractère intégré des travaux de recherche fondamentale, clinique et translationnelle, au sein d'un périmètre géographique limité et autour d'un noyau central de ressources et de compétences au coeur de l'IHU, garantissant une continuité de fonctionnement ; • Intégrer un objectif de valorisation et de transfert de technologies ; • Avoir la capacité d'attirer une quantité significative de projets émanant de partenaires privés.

→ Plus d'informations :
https://anr.fr/fr/detail/call/instituts-hospitalo-universitaires-ihu-3-appel-a-projets-2022/

Appel à projets interrégional de recherche en soins primaires – RESP-IR

Budget	300 000€ par projet
Durée	120 mois
Date limite de dépôt des dossiers	Lettre d'intention : mardi 13 décembre 2022 – 14h00 Dossier complet : mardi 4 avril 2023 – 14h00
Eligibilité	Pas de priorisation de thématiques. • Tous les domaines de la recherche appliquée à la santé médicales et/ou paramédicales ; • Toutes pathologies en santé relevant d'une recherche dans les lieux d'exercice, nécessitant une mise en place de soins primaires.
Objectif	Favoriser la coopération entre les acteurs du premier recours et les acteurs de la recherche appliquée en santé.

→ Plus d'informations : https://girci-est.fr/resp-ir/

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 (entrés dans PubMed) entre début février et début juillet 2022. Pour chaque citation, un lien hypertexte est inclus dans le doi. Il permet, en principe, d'accéder à l'abstract de l'article (voire à son texte intégral) sur le site de l'éditeur.

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