ARGUMENTAIRE

Protocole national de diagnostic et de soins (PNDS)

APLASIES MEDULLAIRES ACQUISES ET CONSTITUTIONNELLES

Version du 1er Mars 2023

Cet argumentaire a été élaboré par le centre de référence
«**APLASIES MEDULLAIRES ACQUISES ET CONSTITUTIONNELLES"**
Il a servi de base à l'élaboration du PNDS
« APLASIES MEDULLAIRES ACQUISES ET CONSTITUTIONNELLES »
Le PNDS est téléchargeable sur le site du centre de référence
www.aplasiemedullaire.com

APLASIE MEDULLAIRE centre de référence

centre de référence maladies rares

MaRIH Filière de santé Maladies Rares Immuno-Hématologiques

filière de santé maladies rares Reconnue par le Ministère de la Santé

Sommaire

Liste des abréviations

AMA	Aplasie médullaire acquise
AMS	Aplasie médullaire sévère
ATGAM	Sérum anti-lymphocytaires de cheval
CsA	Ciclosporine A
CSH	Cellules souches hématopoïétiques
CSP	Cellules souches périphériques
ELT	Eltrombopag
GMO	Greffe de moelle osseuse
GVHa	Acute Graft versus host disease (maladie du greffon contre l'hôte aigu)
GVHc	Chronic Graft versus host disease (maladie du greffon contre l'hôte chronique)
HAS	Haute Autorité de Santé
PNDS	Protocole national de diagnostic et de soins
SAL	Sérum anti-lymphocytaire
SFGM-TC	Société francophone de greffe de moelle et de thérapie cellulaire

Préambule

Le PNDS « aplasies médullaires acquises et constitutionnelles a été élaboré selon la « Méthode d'élaboration d'un protocole national de diagnostic et de soins pour les maladies rares » publiée par la Haute Autorité de Santé en 2012 (guide méthodologique disponible sur le site de la HAS : www.has-sante.fr). Le présent argumentaire comporte l'ensemble des données bibliographiques analysées pour la rédaction du PNDS

Champ du PNDS

Ce PNDS ne concerne pas les pancytopénies survenant dans les suites immédiates d'une chimiothérapie antimitotique, ainsi que les cytopénies isolées (anémies, thrombopénies et neutropénies) acquises ou congénitales non en rapport avec une insuffisance médullaire globale et les cytopénies réfractaires acquises (syndromes myélodysplasiques) qui sont en dehors du champ d'expertise reconnu pour le centre de référence maladies rares « aplasies médullaires acquises et constitutionnelles ». Les évolutions clonales vers des syndromes myélodysplasiques et leucémies aigues des insuffisances médullaires constitutionnelles et acquises sont par contre traitées, de même que les complications extra-hématologiques en rapport avec ces pathologies.

Argumentaire

Stratégie de recherche bibliographique

La recherche documentaire a été réalisée en utilisant successivement les mots-clefs suivants :

- Aplastic anemia
- Paroxysmal nocturnal hemoglobinuria
- Fanconi anemia
- Blackfan-Diamond anemia
- Short telomeres syndrome (dyskeratosis congenita)
- *SAMD9/SAMD9L* syndrome
- *MECOM* syndrome
- Amegacaryocytic thrombopenia ,
- *MPL*
- *THPO*
- *ERCC6L2*
- *LIG4 genes*

Les bases de données consultées :

- PubMed (https://pubmed-ncbi-nlm-nih-gov.proxy.insermbiblio.inist.fr/)
- Orphanet (https://www.orpha.net/consor4.01/www/cgi-bin/?lng=FR

Les sources principales utilisées par le groupe de travail pluridisciplinaire (Annexe 1) pour la rédaction de ce guide ont été les suivantes :

- le texte du 1er PNDS élaboré en 2009, le 2nd en 2019 qui a servi de base de travail à cette mise à jour (période 1990-2019),
- les principales recommandations internationales de prise en charge de l'aplasie médullaire acquise et constitutionnelle (table 1)
- les méta-analyses, essais cliniques et études de cohortes (revue non exhaustive) publiés dans la littérature (PubMed) entre 2019 et 2023 concernant les modalités du diagnostic, les caractéristiques et le traitement des aplasies médullaires acquises et constitutionnelles et de l'Hémoglobinurie Paroxystique Nocturne (HPN) de l'adulte et de l'enfant (Table 2)

- les documents des sites Internet de la Société Française d'Hématologie, de la Société d'Immuno-Hématologie Pédiatrique (SHIP) et du groupe pédiatrique de la Société Francophone de Greffe de moelle et de Thérapie Cellulaire (SFGM-TC)

Langues retenues : anglais, français

Table 1 : Recommandations de Bonnes Pratiques ou protocoles existants

Auteur, Année Référence Pays	Objectif	Stratégie de recherche bibliographique renseignée (oui/non)	Recueil de l'avis des professionnels (non, oui, lesquels)	Recueil de l'avis des patients (non, oui)	Populations et techniques (ou produits) étudiées	Résultats (avec grade des recommandations si disponible)
Peffault de Latour R, Soulier J. Blood. 2016. doi: 10.1182	Identifier rapidement et prédire la progression clonale de la moelle osseuse ; prise en charge thérapeutique dans cette situation	Recherche Pubmed sur la thématique	oui	non	Patient atteint de la maladie de Fanconi	PNDS AM , versions 2019/ 2023
Risitano AM, Peffault de Latour R.Br J Haematol 2022. doi: 10.1111	Prise en charge d'une HPN dans le futur	Recherche Pubmed sur la thématique	Groupe européen de travail sur l'aplasie médullaire (SAAWP of EBMT)	non	Patient atteint d'HPN	
5ème édition, 2020 Fanconi anemia research fund	Maladie de Fanconi : Guide de la prise en charge clinique		Oui Experts du CR aplasies médullaires Fanconi Anemia Research Fund	Oui	Patient atteint de la maladie de Fanconi	Guide Fanconi version 2020
Adrianna Vlachos & al.	Anémie de Blackfan-Diamond; diagnostic et prise en charge		Consensus international dont expert du CR	Non	Patient atteint de l'anémie de Blackfan - Diamond	Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference

Da Costa, Narla & Leblanc, 2020	Anémie de Blackfan-Diamond; diagnostic et prise en charge		Recommandations (avis d'expert) dont experts du CR (Da Costa, Leblanc)	Non	Patient atteint de l'anémie de Blackfan - Diamond	Diamond-Blackfan anemia
Adrianna Vlachos, Ellen Muir. Blood 2010. doi: 10.1182	Prise en charge diagnostic, thérapeutique (transfusions et chélation du fer, corticostéroides, GMO)			Non	Patient atteint de l'anémie de Blackfan - Diamond	
Diaz-de-Heredia, C et al., Bone Marrow Transplant. 2021. doi: 10.1038	Recommendations sur la prise en charge de l'allogreffe de CSH dans l'anémie de Blackfan Diamond-		Groupe européen de travail sur l'aplasie médullaire (SAAWP of EBMT)	Non	Patient atteint de l'anémie de Blackfan - Diamond	
Strahm & al, Blood Advances 2020	Recommandations sur la prise en charge de l'allogreffe de CSH dans l'anémie de Blackfan Diamond-		Recommandations Franco-Allemandes	Non	Patient atteint de l'anémie de Blackfan - Diamond	
Flore Sicre de Fontbrune, Thierry Leblanc Thierry Leblanc	Fiches urgences « ORPHANET »		Oui Experts du CR aplasies médullaires			*Bonnes pratiques en cas d'urgence / HPN* *Bonnes pratiques en cas d'urgence / Anémie de Fanconi* *Bonnes pratiques en cas d'urgence / Anémie de Blackfan-Diamond*

Table 2 : Études cliniques référencées

Pour les aspects thérapeutiques, les différents grades de recommandations indiqués suivent les niveaux de preuves explicités dans le tableau ci-dessous (référence de la Haute Autorité de Santé - HAS 2013)

Grade des recommandations	Niveau de preuve scientifique fourni par la littérature
A Preuve scientifique établie	Niveau 1 - essais comparatifs randomisés de forte puissance ; - méta-analyse d'essais comparatifs randomisés ; - analyse de décision fondée sur des études bien menées.
B Présomption scientifique	Niveau 2 - essais comparatifs randomisés de faible puissance ; - études comparatives non randomisées bien menées ; - études de cohortes.
C Faible niveau de preuve scientifique	Niveau 3 - études cas-témoins.
	Niveau 4 - études comparatives comportant des biais importants ; - études rétrospectives ; - séries de cas ; - études épidémiologiques descriptives (transversale, longitudinale).

Tableau 3. Etudes cliniques

Auteur, année, référence, pays	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et signification
Scheinberg P, et al., N Engl J Med. 2011 . doi: 10.1056/	Identifier le meilleur type de sérum anti lymphocytaire	Grade A	Aplasie médullaire sévère	Comparaison SAL lapin /SAL cheval (ATGAM)	Survie et réponse hématologique	Traitement par SAL –cheval en 1ère en termes de taux de réponse hématologique et de survie
de Latour et al., Blood. 2008	Histoire naturelle de l'HPN	Grade C	HPN	-	Evènements thrombotiques/ facteurs de risque Complications hémorragiques Infections Evolution clonale	2 sous-catégorie de maladie : HPN classique et HPN associée à une AMA Mêmes complications pour les 2 sous-catégories
Peffault de Latour R et al., Am J Hematol. 2018 . doi: 10.1002	Sérum anti-lymphocytaire de cheval	Etude rétrospective portée par le CR aplasies médullaires -Grade C	AMA	-	Survie à 5 ans	90 % pour les formes non sévères
Peffault de Latour et al., NEJM 2022 .doi: 10.1056	Ajout Eltrombopag au SAL-cicloporine en traitement de première ligne dans l'AMS chez l'adulte	Grade A	AMS, adulte	SAL de cheval) + CsA *versus* ATGAM + CsA + ELT		Augmentation réponse globale d'environ 30% à 6 mois (médiane réponse à 3 mois (versus 9 mois avec le traitement standard ATGAM-CsA

Groarke et al., BJH 2021. doi: 10.1111	Intérêt de l'ajout de Eltrombopag en première ligne dans l'AMS chez l'enfant	Grade C	AMS, pédiatrique	SALcheval+CSA+ELT versus cohorte historique SAL cheval-CsA	Survie globale et réponse hématologique	Pas de différence significative dans le taux de réponse globale ou le taux de réponse complète à 6 mois.
Contejean et al.,Haematol. 2018. doi: 10.3324	Evaluer l'efficacité et la tolérance et facteurs prédictifs de survie et de réponse	Grade C	AMA chez le sujet âgé	-	Survie et réponse hématologique	Traitement de référence SAL-CsA-ELT si pas comorbidités cardiovasculaires ou rénales ou un risque de perte d'autonomie trop élevée
Lengline et al., Haematologica. 2018. doi: 10.3324 Olnes et al., N Engl J Med. 2012. doi: 10.1056 Desmond et al., Semin Hematol 2015. doi: 10.1053	Intérêt à utiliser l'ELT dans AMS réfractaire	Grade C	AMA en rechute ou réfractaire , non éligible SAL ou greffe	-	Réponse hématologique	Bénéfice à utiliser ELT en monothérapie ou avec CsA chez les patients de plus de 65 ans
Fan X et al., Blood Adv 2020 doi: 10.1182		Grade B –étude de phase II	AM modérée	Escalade de dose de l'ELT	Taux de réponse à 20 semaines	Bonne tolérance. Pas de différence notable en efficacité par rapport au traitement standard SAL-CsA
Konopacki et al., Haematologica 2012 doi: 10.3324	Suivi à long terme patients allogreffés dans AMS	Grade C	AMS chez les patients de moins de 40 ans	Allogreffe de CSH, donneur géno-identique	Survie à long terme	> 80%, voire 90% chez les très jeunes
Champlin et al., Blood 2007. doi: 10.1182	Conditionnement pour greffe géno-identique	Grade A	AMS	Conditionnement cyclophosphamide seul *versus* cyclophosphamide + SAL -donneur génoidentique	Prise de greffe Survie à 5 ans	Diminution de 5% de la probabilité d e non pris /rejet avec conditionnement SAL + cyclophosphamide à forte dose

Schrezenmeier et al., Blood 2007. doi: 10.1182	Influence de la source de CSH sur la survenue de GvH	Grade A	AMS	CSP *versus* moelle Osseuse- greffe génoidentique	Taux de GVH	Réduction du risque de GVH, de même que la durée de la prophylaxie par ciclosporine avec les cellules de moelle osseuse
Bacigalupo et al., Haematologica 2012. doi: 10.3324	Influence de la source de CSH sur la survenue de GvH Y compris chez le sujet plus âgé	Grade A	AMA	CSP *versus* moelle Osseuse- greffe génoidentique	Survie	Cellules de moelle osseuse à utiliser quel que soit l'âge ; réduction du taux de GvHa et GVHc

Annexe 1 : Listes des participants

Ce travail a été coordonné par le Pr Régis Peffault de Latour, coordonnateur du centre de référence des aplasies médullaires acquises et constitutionnelles, Service d'Hématologie greffe, Hôpital Saint-Louis, Paris en collaboration avec le Pr Thierry Leblanc, site constitutif du centre de référence de l'Hôpital Robert Debré à Paris.

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Groupe de travail multidisciplinaire 2023 :

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Dr Maxime ESKENAZI, Médecin généraliste, Marseille
Mr Farid Ouabdesselam: Association AFMF
Mr Adel Ayad: Association HPN France –Aplasie Médullaire
Mr Marcel Hibert : Association ABD
Mme Isabelle Brindel : centre de référence des aplasies médullaires

Déclarations des conflits d'Intérêts

Tous les participants à l'élaboration du PNDS sur les aplasies médullaires acquises et constitutionnelles ont rempli une déclaration d'intérêt disponible sur le site internet du centre de référence. Les déclarations d'intérêt ont été analysées et prises en compte, en vue d'éviter les conflits d'intérêts, conformément au guide HAS « Guide des déclarations d'intérêts et de gestion des conflits d'intérêts » (HAS, 2010).

Modalités de concertation du groupe de travail pluridisciplinaire

La rédaction du PNDS a été réalisée lors de réunions physiques et par échanges de mails entre les membres du groupe de travail multidisciplinaire.

Annexe 2 : Bibliographie

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