ERN-EuroBloodNet

European Reference Network on Rare Hematological Diseases

CLINICAL CARE

2.2.1. The ERN has implemented a process for offering advice for complex patient cases provided by multidisciplinary healthcare teams

Annex II ERN-EuroBloodNet CPMS needs

Introduction

One of the key objectives established by ERN-EuroBloodNet is to provide inter-professional consultation by sharing of expertise and safe exchange of clinical information through the Clinical Patient Management System (CPMS), as the platform supporting European Reference Networks in facilitating the decision making for the diagnosis and treatment of rare disease or low prevalence complex diseases or conditions across national borders.

12 ERN-EuroBloodNet contributed to use the platform and provide feedback during the CPMS pilot phase (2017-2018). Following this period, ERN-EuroBloodNet aimed to identify the areas where CPMS is more needed in the context of ERN-EuroBloodNet, including the RHD area, type of advice (care/diagnosis) and age of patients (pediatrics/adults)

Analysis of CPMS needs by subnetwork and disease category

ERN-EuroBloodNet CPMS helpdesk and coordination team prepared a template circulated among the Subnetworks coordinators to provide their feedback including:

- A section to score each of the Categories from 0-5 to rank the need of the CPMS for each category (0- CPMS not needed, 5- CPMS highly needed).
- A section to indicate the type of advice more required for each of the Category: clinical care/diagnosis/both
- A section to indicate if advice more required for pediatrics/adults/both for each category
- A section for the identification of one or two reference persons for each category

A total of 6 experts participated. Some of the coordinators provided their view on more than one subnetworks. In these cases, the media of the answers was calculated for the analysis of the results (Table 1 Scores for CPMS needs and type of advice required by subnetwork and disease category)

	Score need for CPMS (0-5)	Type of advice needed: clinical care/diagnosis/Both	Advice more related to pediatrics/Adults/Both
Red Blood Cell disorders	**4,5**		
Sickle cell disorders	4	Both	Both
Thallassaemia disorders	4	Both	Both
Hereditary erythroenzymopathies and RBC membrane defects	5	Both	Both
Congenital Erythrocytosis and other rare RBC defects	5	Both	Both
Bone marrow failures and related disorders - Answer 1	**3,75**		
Congenital dyserythropoietic anemia	1	Both	Both
BMF Inherited (Fanconi anemia, Dyskeratosis congenital, GATA2 syndrome,Congenital	5	Both	Both
BMF Acquired (Aplastic Anaemia and Paroxysmal Nocturnal Hemoglobinuria)	5	Both	Both
Blackfan-Diamond anemia	4	Both	Both
Bone marrow failures and related disorders - Answer 2	**5**		
Congenital dyserythropoietic anemia	5	Both	Both
BMF Inherited (Fanconi anemia, Dyskeratosis congenital, GATA2 syndrome,Congenital amegakaryocytic thrombocytopenia and others)	5	Both	Both
BMF Acquired (Aplastic Anaemia and Paroxysmal Nocturnal Hemoglobinuria)	5	Both	Both
Blackfan-Diamond anemia	5	Both	Both
Bleeding and coagulation disorders - Answer 1	**3**		
Haemophilia A, B	3	Both	Both
The rarer congenital deficiencies of other coagulation factors (such as fibrinogen and	3	Both	Both
Von Willebrand disease	3	Both	Both
Inherited platelet defects	3	Both	Both
Bleeding and coagulation disorders - Answer 2	**4,5**		
Haemophilia A, B	5	Both	Both
The rarer congenital deficiencies of other coagulation factors (such as fibrinogen and factors II, V, VII, X, XI and XIII)	5	Both	Both
Von Willebrand disease	5	Both	Both
Inherited platelet defects	3	Both	Both
Haemochromotaosis and rare iron metabolism and heme synthesis - Answer 1	**5**		
Rare iron overload (hereditary hemochromatosis)	5	Both	Both
Rare Ferritinopathy	5	Both	Both
Porphyrias	5	Both	Both
Rare iron metabolism disorders (sideroblastic and non-sideroblastic)	5	Both	Both
Haemochromotaosis and rare iron metabolism and heme synthesis - Answer 2	**4,5**		
Rare iron overload (hereditary hemochromatosis)	4	Both	Adults
Rare Ferritinopathy	5	Diagnosis	Adults
Porphyrias	4	Both	Adults
Rare iron metabolism disorders (sideroblastic and non-sideroblastic)	5	Both	Adults
Lymphoid malignancies	**5**		
Acute lymphoblastic leukemia (ALL)	5	Both	Both
Marginal zone lymphomas	5	Both	Adult
Light chain Amyloidosis (AL amyloidosis)	5	Both	Adult
Rare lymphomas (hairy cell leukamia…)	5	Both	Adult
Myeloid malignancies	**5**		
Myelodysplastic syndrome (MDS)	5	Both	Adult
Acute myeloid leukemia (AML)	5	Both	Both
Chronic myelomonocytic leukemia (CMML)	5	Both	Adult
Chronic Myeloid Leukemia (CML)	5	Both	Adult
Myeloproliferative neoplasm (MPN)	5	Both	Adult
Myelofibrosis	5	Both	Adult
Systemic mastocytosis			
BMT			
Non oncological disorders	5	Both	(Answer for BMT)
Oncological disorders	5	Both	(Answer for BMT)

CPMS need - Analysis by subnetworks

Fig. 1 illustrates the need of the CPMS by subnetwork. For those subnetworks with more than one answer, the media of the answers have been calculated for the analysis of results. RBC: Red Blood Cell, BMF: Bone Marrow Failures, Bleeding: Bleeding and coagulation disorders, HH and Iron: Hereditary Hemochromatosis and Iron metabolism disorders, Lymphoid: Lymphoid malignancies, Myeloid: Myeloid malignancies.

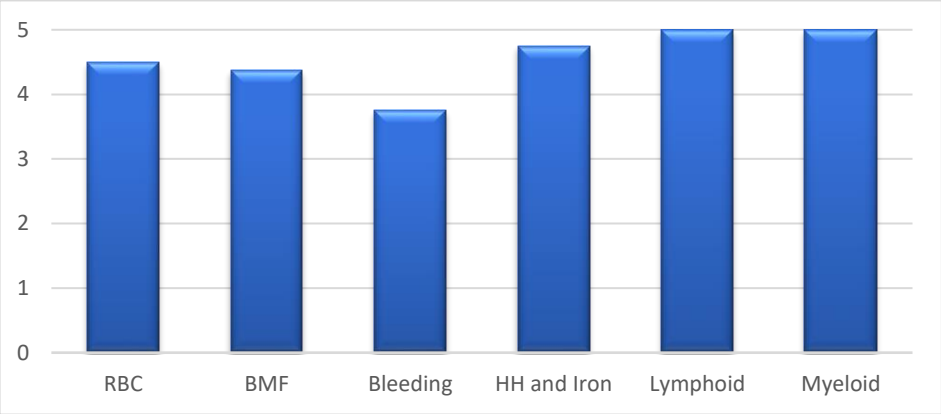

Fig. 1 CPMS need by subnetwork

According to subnetworks coordinators opinion, the CPMS is most required for the oncological disorders, where the need was total (5) for Lymphoid and Myeloid subnetworks. Regarding the non-oncological hub, the highest need was identified for the Hemochromatosis and other rare genetic disorders of iron metabolism and heme synthesis subnetwork (4,75), which encompasses a great number of very rare disorders that in many occasions require from a second advice from different multidisciplinary team for the correct management of the patients. Also a similar result was obtained for the Red blood cell and bone marrow failure subnetworks (4,5 and 4,375 respectively). On the other hand the lowest need identified was assigned to the Bleeding and coagulation subnetwork (3,75).

The analysis of the type of advice and age of patients for which the CPMS is most required has been performed by category (see below: CPMS need – Analysis by Category).

CPMS need - Analysis by Category

The media of the answers regarding the need of the CPMS according to the categories for each subnetwork are represented in the following figures:

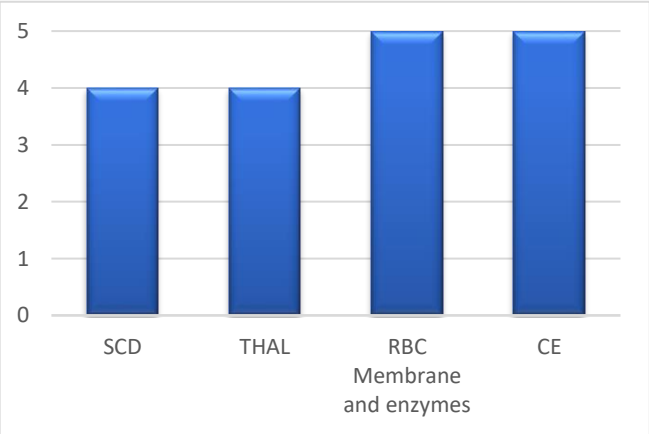

Fig. 2 CPMS Need for Red blood cell subnetwork categories. SCD: Sickle cell disorders, THAL: Thalassaemia syndromes, Red Blood Cell membrane and enzyme disorders, CE: Congenital Erythrocytosis.

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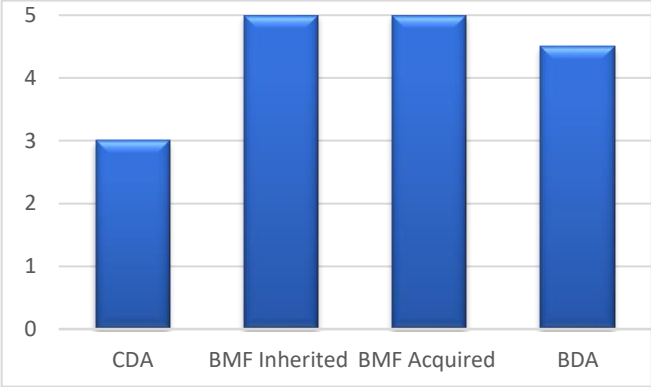

Fig. 3 CPMS Need for bone marrow failure subnetwork categories. CDA: Congenital Dyserythropoietic anaemia. BMF Inherited: Fanconi anemia, Dyskeratosis congenital, GATA2 syndrome,Congenital amegakaryocytic thrombocytopenia and others. BMF Acquired: Aplastic Anaemia and Paroxysmal Nocturnal Hemoglobinuria, BDA: Blackfand-Diamond Anaemia.

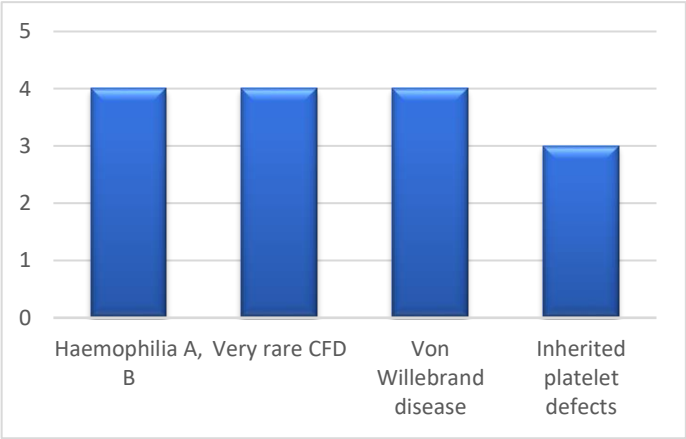

Fig. 4 CPMS Need for bleeding-coagulation subnetwork categories. Very rare CFD: The rarer congenital deficiencies of other coagulation factors (fibrinogen and factors II, V, VII, X, XI and XIII)

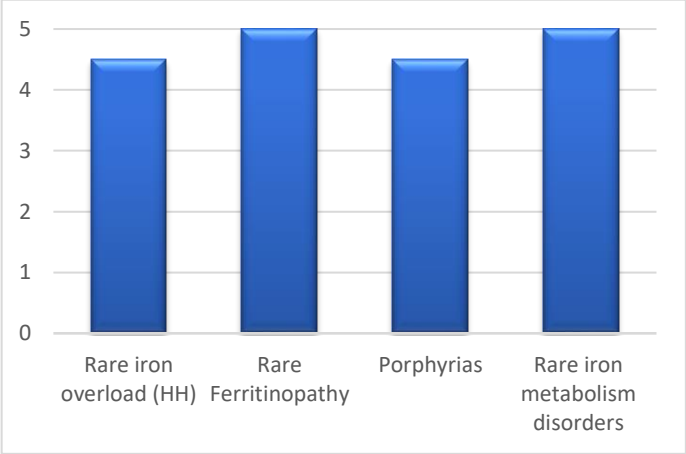

Fig. 5 CPMS Need for Haemochromatosis and rare iron metabolism and heme synthesis subnetwork categories. HH: Hereditary Hemochromatosis.

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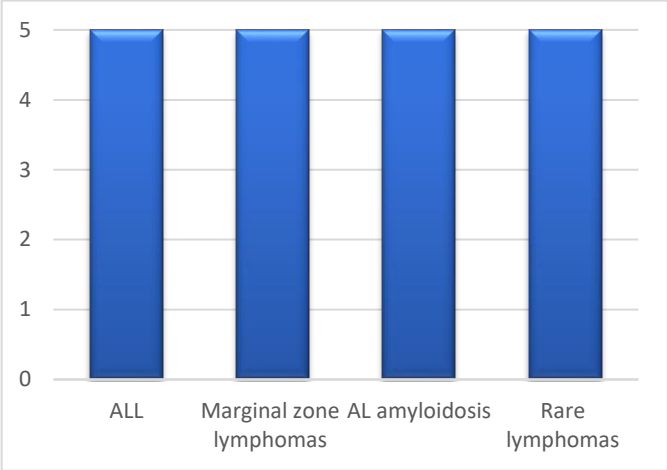

Fig. 6 CPMS Need for Lymphoid subnetwork categories. ALL: Acute lymphoblastic leukemia

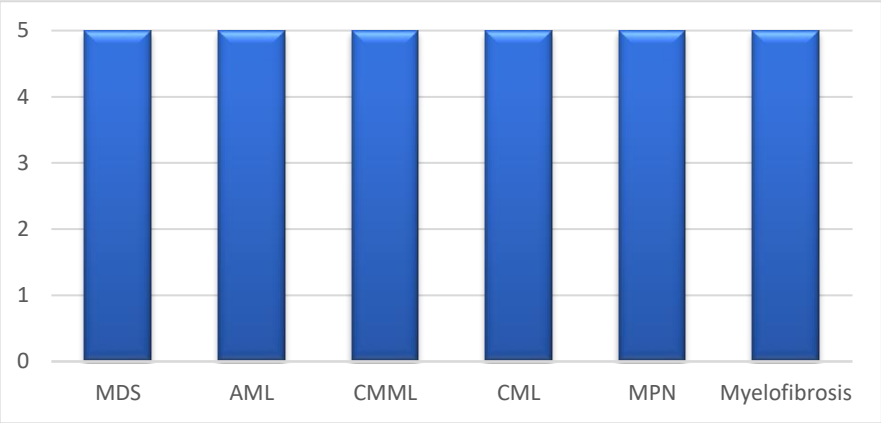

Fig. 7 CPMS Need for Myeloid subnetwork categories. MDS: Myelodysplastic syndrome, AML: Acute myeloid leukemia, CMML: Chronic myelomonocytic leukemia, CML: Chronic Myeloid Leukemia, MPN: Myeloproliferative neoplasm.

Regarding the need of the CPMS by category of disorders, it is important to highlight the difference between the answers provided by two experts on the BMF subnetworks in the Congenital dyserythropoietic anemia, being rated as essential for one expert (5) and not necessary by another (1). A similar case was also given for two answers on the Bleeding and coagulation disorders, where one expert rated the CPMS as highly necessary (5), and another expert finds it not so needed (3).

Regarding the type of advice for which CPMS is most required for each category, there is an unanimous opinion that the platform is needed for both, clinical care and diagnosis, with the only exception found by one expert on Rare Ferritinopathy, who highlighted its need for the Diagnosis.

Regarding the age of the patients for which CPMS is most required, paediatrics and adults advice have been identified the most needed for Red blood cell, bone marrow failure and bleeding and coagulation subnetworks. For the Haemochromotaosis and rare iron metabolism and heme synthesis subnetwork, the major need was identified for paediatrics and adults by one expert, while only for adults for another expert from their point of view. On the oncological hub the major need found was for adults, with the exception of Acute lymphoblastic leukemia (ALL) and Acute myeloid leukemia (AML), where the need was also highlighted for children.