EMA
EUROPEAN MEDICINES AGENCY

Workshop - Challenges in drug development, regulation and clinical practice in haemoglobinopathies

1st July 2024

Virtual meeting

Background and Objectives

Hemoglobinopathy is a group of inherited blood disorders and diseases that primarily affect red blood cells. They are single-gene disorders and, in most cases, they are inherited as autosomal co-dominant traits.

There are two main groups: abnormal structural haemoglobin variants caused by mutations in the haemoglobin genes, and the thalassemias, which are caused by an underproduction of otherwise normal haemoglobin molecules. The main structural haemoglobin variants are HbS, HbE and HbC. The main types of thalassemia are alpha-thalassemia and beta thalassemia.

The two conditions may overlap because some conditions which cause abnormalities in haemoglobin proteins also affect their production. Some haemoglobin variants do not cause pathology or anaemia, and thus are often not classed as hemoglobinopathies.

Recently, new therapies, including gene therapies, have been authorised in the EU and the US and further products are in development.

This workshop is organised in order to have a multi-stakeholder's perspectives on these diseases before initiating the drafting of new scientific guidelines both in sickle disease and thalassaemia in line with the haematology work plan for 2024. These guidelines are aimed to define the data needed and clinical requirements for benefit-risk evaluation and obtaining marketing authorisation.

The aims of the workshop are:

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• To present the epidemiology and disease background in adults and children with sickle cell and thalassemia, the current international treatment guidelines, the unmet medical need, and the overview of the authorised medicines/treatments in the EU and in US for sickle cell and thalassemia.

• To present the challenges in treatment/drug development from a clinicians' perspective with regards to study design and endpoints used in clinical trials as well as the introduction of new therapies such as gene therapy.

• To present additional perspectives from Health Technology Assessment bodies, bioethics for gene editing, and the use of registries in these diseases.

Practical information:

The workshop can be accessed via WebEx with the link provided in the meeting invitation.

Challenges in drug development, regulation and clinical practice in hemoglobinopathies

Chaired by: Daniela Philadelphy, CHMP member for Austria and Haematology working party chair

13:30 Joining and technical checks

14:00 Welcome and opening speech

Opening remarks from EMA Executive Director **10'**
Emer Cooke, Executive Director of EMA

Outline of the day and objectives **5'**
Caroline Voltz-Girolt, Office of advanced therapies and haematological diseases, EMA

14:15 Session 1: Patients and clinical perspectives and management of sickle cell and thalassemia diseases

Diagnosis, epidemiology and patient characteristics of thalassemia, clinical management and treatment guideline **15'**

Prof. Ali Taher, Professor of Medicine, Hematology & Oncology and Director – Naef K. Basile Cancer Institute American University of Beirut Medical Center Beirut – Lebanon

Diagnosis, epidemiology and patient characteristics of sickle cell disease and clinical management and treatment guideline **15'**

Prof. John Porter, University College London, Research Department of Haematology, UK

Paediatric perspectives and treatment goals **15'**

Pr. Mariane de Montalembert, Hôpital Universitaire Necker-Enfants maladies, France

Pr. Raffaella Colombatti, Associate Professor of Pediatrics at the Department of Women's and Child's Health SDB, University of Padova, Italy

Patients perspective on sickle cell and (beta) thalassemia **15'**

Mr. Loris Brunetta, Thalassaemia International Federation (TIF)

15:15 Coffee Break

15:30 Session 2: Overview and regulatory consideration of authorised medicines in Sickle Cell Disease and Thalassemia diseases

Authorised medicines and regulatory considerations by FDA CDER **10'**

Patricia Oneal, Division of Non-Malignant Hematology, FDA

Authorised medicines and regulatory considerations by FDA CBER **10'**

Megha Kaushal, Benign Hematology Branch, FDA

Authorised medicines and regulatory considerations by EMA **10'**

Johanna Lähteenvuo, CHMP member, Finland, EMA

EMA project OPEN **10'**

Radhouane Cherif, Senior International Liaison officer, EMA

16:10 Session 3: Additional perspectives

Bioethics considerations on gene editing **10'**

Laurence Lwoff, Human Rights, Directorate, Council of Europe

HTA perspectives **10'**

Anja Schiel, Special Advisor, Norwegian Medical Products Agency

Experience from registries **10'**

Dr María del Mar Mañú Pereira, Vall d'Hebron Institute of Research (VHIR) - Vall d'Hebron Institut de Recerca, Spain

Challenges in drug development for hemoglobinopathies **10'**

Antonella Isgrò, Haematology working party member, AIFA, Italy

16:50 Coffee Break

17:00 Panel discussion

Chairs: Daniela Philadelphy, CHMP member, Austria and Haematology working party chair
Caroline Voltz, EMA

Additional panelist: Jenica Leah interim president of European Sickle Cell Federation ESCF

Discussion on relevant parameters and outcome measures to be collected in clinical trials by considering treatment goals, patient characteristics and differences in patient populations, study designs aspects for confirmatory trials and specific safety and efficacy considerations.

17:55 Closing remarks

Wrap up **5'**
Daniela Philadelphy