Placing thalassaemia on the agenda of European policy makers

By Tresja Bolt and Nicoletta Bertelli

Saturday 8th May 2021 marks the 27th annual International Thalassaemia Day. It is an important day for the global thalassaemia community to not only raise public awareness of the disease, but also to recognise its immeasurable impact on the lives of patients and their loved ones. It is also a potential wake-up call for EU health policy makers to recognise a rare disease of growing prevalence across the region.

Understanding thalassaemia

Thalassaemia is a rare inherited blood disorder estimated to affect 1 in 10,000 people in the European Union and 1 in 100,000 globally.[1] People with thalassaemia have a genetic mutation which affects their red blood cells. A diagnosis of the disease is typically made within the first two years of life, often passed on by parents who may be healthy or simply unaware of their own status as a carrier of the disease. An estimated 1.5% of the global population are carriers of the most severe form of thalassaemia.[2]

Those with severe cases of thalassaemia require frequent, lifelong transfusions to manage their haemoglobin levels, and are reliant on blood donations to survive.[3] This dependence on transfusions can cause iron to build up in the body, which in turn can cause problems in the heart, liver, and hormone levels if left untreated.[4] The complicated nature of the disease, and its connection to other comorbidities, mean that thalassaemia still claims too many lives prematurely. A recent UK study found that the median age of death for people with severe thalassaemia is just 45 years of age.[5]

Fortunately, medical advancements have made living with thalassaemia more manageable, allowing some patients to live into their fifth decade and beyond.[6] At the same time, however, people with thalassaemia must grapple with the long-term physical and medical challenges posed by the disease. The lifelong monitoring required to manage their complex condition can also cause them to miss out from precious time spent at school, at work and with family, which may also take a toll on their mental health.

Recognising the patient experience

Historically, thalassaemia is most common in certain regions along the so-called 'thalassaemia belt', which runs along the Mediterranean, the Middle East, Africa, or Asia. In recent years, however, shifting population movements is causing the disease to become more prevalent in countries where thalassaemia was incredibly rare. This means that it is no longer just prevalent in Italy, Greece, and

[1] Galanello and Origa, Beta-thalassemia, Orphanet Journal of Rare Diseases 2015, 5:11

[2] Galanello and Origa, Beta-thalassemia, Orphanet Journal of Rare Diseases 2015, 5:11

[3] NORD. Beta Thalassemia. 2018 [ONLINE] Available at: https://rarediseases.org/rare-diseases/thalassemia-major/ Last accessed April 2021

[4] NORD. Beta Thalassemia. 2018 [ONLINE] Available at: https://rarediseases.org/rare-diseases/thalassemia-major/ Last accessed April 2021

[5] Jobanputra, M., Paramore, C., Laird, S.G., McGahan, M. and Telfer, P. (2020), Co-morbidities and mortality associated with transfusion-dependent beta-thalassaemia in patients in England: a 10-year retrospective cohort analysis. Br. J. Haematol., 191: 897-905.

[6] Shamshirsaz, A.A., Bekheirnia, M.R., Kamgar, M. et al. Metabolic and endocrinologic complications in beta-thalassemia major: a multicenter study in Tehran. BMC Endocr Disord 3, 4 (2003).

Cyprus, but we are also seeing cases in places like Germany, the Netherlands, Belgium, and the Nordics. The lack of public awareness to this disease in these countries often creates challenges of access for patients to receive the quality treatment and care they need to survive.

Drawing attention to the many and multiple unmet needs of patients with thalassaemia worldwide is the focus of this year's International Thalassaemia Day, organized by the Thalassaemia International Foundation. To address the health inequities witnessed across the global thalassaemia community, the real-life experiences of those living with thalassaemia around the world are important stories that must continue to be told.

Illustrating the mosaic of experiences of those living with thalassaemia was the focus of a video project recently initiated by bluebird bio and developed in collaboration with several European patient organisations and clinicians. *'Living with Thalassaemia in Europe: Stories from patients of different ages'* spotlights the stories of nine patients from seven European countries (Italy, France, Germany, Greece, Cyprus, Belgium, and the Netherlands), each representing a different life stage between the ages of 4 and 61.

Many of the challenges people living with thalassaemia face are unfortunately all too common. The video project highlighted the struggles of adjusting to thalassaemia from a young age. Some highlighted the difficulties experienced in missing school, overcoming social stigma, and struggling to make friends. Having to come to terms with the regular hospital visits, blood transfusions, and iron chelation therapy appointments is also mentioned as a difficult experience from a young age.

Managing the long-term impact of thalassaemia and its comorbidities on daily life are also a commonality. Extreme fatigue caused by severe anaemia, for example, makes it difficult for those with thalassaemia to maintain a job, or cope with the demands of parenthood. Experiencing the premature loss of a friend or loved one due to thalassaemia is also a constant anguish, reinforcing the significant psychological impact on those living with the disease. One participant in the video project put it plainly: "There's nothing you can do if you don't first think of thalassaemia."

A call to action for EU health systems

Greater awareness in countries where thalassaemia is more common has resulted in the adoption of treatments to help meet the needs of thalassaemia patients. But shifting demographics in Europe mean that thalassaemia is quickly becoming a growing consideration for policy makers, and the national and regional health systems they represent. More should therefore be done to reduce the barriers to access for patients, irrespective of where they live.

On this International Thalassaemia Day and beyond, policy makers across the globe should take the opportunity to listen to the thalassaemia community. To address their unmet needs, the experiences of patients should be prioritised in policy making. At the most basic level, this includes a more concerted effort to protect blood supply levels across Europe, ensuring the availability of necessary transfusions for all patients amidst the challenges posed by COVID-19. In the long-term, however, patient organizations should be involved in greater collaborative efforts with researchers and policy makers to address the burden of living with thalassaemia and to identify appropriate treatment pathways.

NP-EU-00084
V1
APRIL 2021

About the Authors

Tresja Bolt is Head of Public Affairs, Europe at bluebird bio

Nicoletta Bertelli is Associate Director, Patient Advocacy, Europe at bluebird bio

NP-EU-00084
V1
APRIL 2021