REPORT

CAPACITY BUILDING WORKSHOP

for Thalassaemia Patients & Healthcare Professionals

& 1st RAIN Summit for Patients with Rare Anaemias

16-19 May 2024

Venue: Grand Hotel Bucharest, Romania

THALASSAEMIA INTERNATIONAL FEDERATION

Organised by: Thalassaemia International Federation

In collaboration with: Asociatia Persoanelor cu Talasemie Majora - Romania

APTM

RAIN Rare Anaemias International Network

THALIA

Co-funded by the European Union

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Introduction

Faithful to its vision of ensuring equal access to quality health care for every patient with thalassaemia and other haemoglobin disorders across the world, and in the framework of its strategic partnership with the European Commission for the "THALassaemia In Action 2024 (THALIA 2024)" programme, the Thalassaemia International Federation (TIF) organised a Capacity Building Workshop for Thalassaemia patient and other haemoglobinopathies & Healthcare Professionals entitled ''Capacity Building Workshop for Patients with Thalassaemia and Healthcare Professionals & the First RAIN Summit FOR Patients with Rare Anaemias'' on 16 – 19 May 2024 in Bucharest, Romania.

This Capacity Building Workshop is part of the Federation's consistent efforts for the life-long education and empowerment of patients with Haemoglobin Disorders and Healthcare Professionals and for the reinforcement of their networking and collaborations. The workshop aim for the immediate need of accurate knowledge and education on many aspects pertaining to the effective prevention, control and management of thalassaemia and SCD and on the new scientific advances occurring in the field, and to provide guidance and support, in order for patients and supporting organisations and groups to be able to create and sustain fruitful partnerships to better advocate for their rights, whilst actively participating in decision-making processes as equal partners with healthcare professionals and their national health authorities.

OBJECTIVES

Through its educational endeavours in Romania, part of which is this Capacity Building Workshop, TIF seeks to raise awareness and understanding about thalassaemia and haemoglobin disorders. In particular, the Capacity Building Workshop's main objectives were to highlight the value of patient contribution to the improvement of provided healthcare services for thalassaemia and haemoglobin disorders, through the meaningful participation in decision-making processes at national, regional and international levels, as well as to incapacitate patients and parents' associations to:

- Empower and build capacities for patients and their families through strengthening their disease specific knowledge.

- Inform the medical and patient communities about the state-of-art clinical management of haemoglobin disorders, the latest scientific advancements in the field, and regulatory
- developments for new drugs and therapies.
- Extend the knowledge gained in experienced countries to other European countries through sharing experiences and best practices.
- Develop the skills and capacities of patients for the advocacy and productive participation in decision making at the country and regional levels.
- Educate and create core groups of patients in

SUMMARY

More than 140 patient organizations' representatives and patient leaders from 34 countries attended TIF's **''Capacity Building Workshop for Patients with Thalassaemia and Healthcare Professionals & the First RAIN Summit FOR Patients with Rare Anaemias'"**, coming mainly from European countries, such as Romania, Belgium, Bulgaria, Albania, Germany, Cyprus, Greece, France, Italy, England, Austria, Denmark, Portugal, Hungary, Sweden, the Netherlands, and the UK. Other countries represented in the Workshop were the US, Algeria, Brazil, Azerbaijan, Egypt, Lebanon, Malaysia, Mauritius, Maldives, Sri Lanka, Vietnam, Jordan, Canada, India, Saudi Arabia and Turkey.

The Workshop comprised of an opening ceremony followed by 7 different sessions, which were delivered by national and international experts from the US, Cyprus, Italy, Canada, Germany, Greece and the UK, and included presentations on key elements that Healthcare Professionals and patients should be striving to fulfil, delivered by international experts. More particularly, the sessions focused on the following themes:

- The fundamentals of clinical management of Haemoglobin Disorders
- Multidisciplinary Care in Haemoglobin Disorders
- National Challenges in Addressing Haemoglobin Disorders
- New Era in the Care & Cure of Haemoglobin Disorders: Patient Testimonials
- Recently approved innovative therapies and medicinal products for Thalassaemia
- New Advances in the Pipeline for the Care & Cure of Haemoglobin Disorders
- Treatment Beyond the Haematologist & Paediatrician: Emerging Concerns
- Living with Thalassaemia - Opportunities, Challenges & Dilemmas
- Achieving Healthcare Reforms through Patient Advocacy

PROGRAMME

THALASSAEMIA INTERNATIONAL FEDERATION

DAY 1 THURSDAY 16 MAY 2024	
15.00 – 16.30	Meeting of patient leaders - Central Europe (By invitation only)
17.00 – 18.30	Meeting of patient leaders - Eastern Europe (By invitation only)

DAY 2 FRIDAY 17 MAY 2024	
ROOM Name:	**Ronda**
08.30 – 09.00	Registration
	OPENING CEREMONY
09.00 – 09.30	Welcome Messages Panos Englezos, President, Thalassaemia International Federation (TIF) - Pre-recorded George Constantinou, – Vice President of the Thalassaemia International Federation, UK Radu Ganescu, President, Asociatia Persoanelor cu Talasemie Majora Dr Horatiu-Remus Moldovan – Secretary of State at the Ministry of Health, Romania Prof. Dr. Daniel Coriu - Head of Department, Fundeni Clinical Institute, Hematology and Bone Marrow Transplantation, Bucharest.
Session 1	**Clinical Management for Thalassaemia – Progress & advances**
	Chairs: J. Porter & E. Vlachaki
09.30 – 09.55	Blood transfusion therapy – E. Vlachaki
09.55 – 10.20	Iron chelation Management – J. Porter
10.20 – 10.45	Iron Overload: the value of reliable monitoring and assessment – J. Porter
10.45 – 11.00	Q&A
11.00 – 11.30	**Coffee break**
Session 2	**Haemoglobin Disorders: New Developments**
	Chairs: D. Rees & R. Grosse
11.30 – 12.00	Authorized medicinal products – D. Rees
	Q&A
12.00 - 12.35	**The Patients' Perspective**
	Testimonial: Person with Thalassaemia – A. Ahmad Testimonial: Person with Rare Anaemia – T. Schryver Testimonial: Person with SCD – G. Vindigni Q&A

THALASSAEMIA INTERNATIONAL FEDERATION

12.35 – 13.00	Overview of ongoing clinical trials in Haemoglobin Disorders – R. Grosse
13.00 – 14.00	**Lunch**
Session 3	**New Advances for the Cure of Haemoglobin Disorders**
	Chairs: P. Sodani & C. Lederer
14.00 – 14.25	Gene Editing for thalassaemia & SCD: where we stand today. Lecture: P. Kottaridis Patient Testimonial: E. Hikmet Demiray
14.25 – 14.50	Hematopoietic Stem Cell Transplantation in thalassaemia – Benefits & Complications – P. Sodani
14.50 – 15.15	Gene addition for thalassaemia & SCD: latest information – C. Lederer
15.15 – 15.30	Q&A
15.30 – 16.00	**Coffee break**
Session 4	**Multidisciplinary care: The third essential cornerstone in the management of Haemoglobin Disorders**
	Chairs: D. Farmakis & N. Skordis
16.00 – 16.25	Cardiac complications in thalassaemia – D. Farmakis
16.25 – 16.50	Impact of bone disease and pain in thalassaemia – N. Skordis
16.50 – 17.15	Management of Fertility and Pregnancy in thalassaemia – N. Skordis
17.15 – 17.30	Q&A
17.30 – 19.00	**Round-Table Discussion: Multi-Generational and Multi-Cultural Dynamics in Haemoglobin Disorders**
	Moderators: G. Constantinou & E. Georganda
	Global Panel of Patients and Healthcare Professionals

DAY 3 – SATURDAY 18 MAY 2024	
Session 5	**Healthcare Professionals Session Case Presentations & Exchange of Best Practices**
ROOM Name:	**Rapsodia**
	Chairs: J. Porter & JL. Vives Corrons
09.30 – 09.45	Case presentation: E. Vlachaki
09.45 – 10.00	Q&A
10.00 – 10.15	Case presentation: D. Farmakis
10.15 – 10.30	Q&A
10.30 – 10.45	Case presentation: JL. Vives Corrons
10.45 – 11.00	Q&A

11.00 – 11.30	**Coffee break**
11.30 – 11.45	Case Presentation: D. Rees
11.45 – 12.00	Q&A
12.00 – 12.15	Case Presentation: N. Skordis
12.15 – 12.30	Q&A
12.30 – 12.45	Case Presentation: P. Kottaridis
12.45 – 13.00	Q&A
13.00 – 13.15	Case Presentation: K. Kuo
13.15 – 13.30	Q&A
13.30	**End of HCPs session**

1ST RAIN SUMMIT FOR PATIENTS WITH RARE ANAEMIAS

Joint Session with patient attendees of Capacity Building Workshop

Session 5	**Living with Rare Anaemias**
ROOM Name:	**Ronda**
	Chairs: T. Schryver & C. Ganescu
09.30 – 10.00	Patient Testimonials: Life with a rare anaemia Living with Thalassaemia – C. Ganescu Living with Rare Anaemia – A. Al Fahdi
10.00 – 10.20	Mental Health: Body image and relationships - E. Georganda
10.20 – 10.40	Nutrition in Thalassaemia & PKD - T. Schryver
10.40 – 11.00	How do I tell my child I have a rare anaemia and what it means - A. Watson
11.00 – 11.30	**Coffee break**
Session 6	**Capacity Building for Patient Associations - Breakout working groups**
	Moderator: S. Tuli & G. Constantinou
11.30 – 11.45	Intro presentation: K. Pelides
11.45 – 12.30	Table 1. Fundraising: How to gain supporters Facilitator: R. Ganescu
	Table 2. The Power of Collaboration: Building Effective Partner Networks Facilitator: L. Brunetta

	Table 3. Effective advocacy skills Facilitator: G. Constantinou
	Table 4. The importance of Communication and Visibility for Nonprofit Organizations Facilitator: B. Calabria
12.30 – 13.00	**Interaction & Reports**
13.00 – 14.30	**Lunch**
Session 7	**Management for Rare Anaemias: Guidelines**
	Chairs: D. Farmakis & G. Constantinou
14.30 – 15.00	Guidelines for the Management of Thalassaemia: History of development and methodology – M.D. Cappellini
15.00 – 15.30	New Guidelines for the Management of PKD: Unmet need & value – K. Kuo
15.30 – 16.00	Patients' involvement and perspective: Thal Patient: G. Constantinou PKD Patient: Carl Lander SCD Patient: J. James (Pre-recorded)

DAY 4 – SUNDAY 19 MAY 2024

ROOM Name:	**Rapsodia**
RAIN Members Strategy Session	
	Chairs: J. Porter & JL. Vives Corrons
09.30 – 10.00	Overview of RAIN accomplishments - L. Cannon (Pre-recorded)
10.00 – 10.30	Current Therapeutic Approaches for Rare Anaemias – J. Porter
10.30 – 11.00	New Development on diagnosis of Rare Anaemia and Thalassaemia – JL. Vives Corrons
11.00 – 11.30	**Coffee break**
11.30 – 12.00	Clinical Trials and New advances for the care and cure of Rare Anaemias – K. Kuo
12.00 – 13.00	Interactive Discussion: Future actions and activities **Moderators: G. Constantinou & JL. Vives Corrons**

SESSIONS OVERVIEW

Day 1: 17 May 2024

After the Welcome session where all the speakers have highlighted the scope of this workshop and the Under-Secretary of the Rumanian MoH expressed appreciation to TIF and the local Associatia persoanelor cu Talasemia Majora for the organization of the event in Bucharest, welcoming all the participants, showing full engagement to cooperate for ensuring patients' access to basic treatment and improving the knowledge of thalassemia across the country, the program of the Workshop started.

Preliminary considerations: The participation of local patients and more in general of patients coming from eastern and balcanic region were quite low, particularly on the first day, Friday, maybe due to the fact that Friday is a working day. The collaboration of the Asociatia Persoanelor cu Thalasemia Majora with TIF has been vital for the success of the event. TIF staff (Rawad and Katie) did a great job for ensuring everything went smooth.

Session 1: Clinical Management for Thalassaemia - Progress & advances

Blood transfusion therapy

Dr. Efthymia Vlachaki, Assistant Professor of Haematology-Hemoglobinopathies, Aristotle University, Thessaloniki, Greece, Adult Thalassemia Unit, Ippokration Hospital, Greece

Blood transfusion remains the essential lifesaving treatment for people living with transfusion-dependent thalassaemia (TDT), apart from the curative therapies which are only available to a minority of patients. Transfusion-related complications remain a source of morbidity but can be effectively prevented with optimal care and patients' monitoring. However, worldwide blood supplies are limited, particularly in several high-prevalence countries with limited resources, while increasing blood donation and safeguarding optimal processing and handling of blood products remain unmet needs. In this context, continuous efforts to raise community awareness, regular training of health-care workers and proper utilization of available resources are essential to ensuring safe blood transfusions for patients with thalassaemia. On the other hand, recent insights into the pathophysiology of β-thalassaemia had led to advances in therapeutic

approaches and access to the new treatments at an affordable cost may reduce the blood transfusion burden.

Iron chelation management and the value of reliable monitoring and assessment of iron overload

Prof. John Porter, Professor of Haematology, Head of the Joint Red Cell Unit for UCLH & Whittington Hospitals, Haematology Department, University College London, London, UK

Chelation therapy can be a highly effective modality in preventing organ toxicity from iron overload that results from repetitive blood transfusions and increased intestinal iron absorption. Reversal of iron-mediated toxicity has been clearly demonstrated for the heart, but it is less clear for other organ systems. Monitoring the trends in iron load should aim at identifying under- and over-treatment before irreversible organ damage occurs. Treatment adherence remains an important issue and healthcare systems should be designed to help patients adhere to chelation regimen so that rescue chelation therapy becomes less necessary.

Session 2: Haemoglobin Disorders - New Developments

Authorized medicinal products

Prof. David Rees, Paediatric Haematologist at King's College Hospital, London, UK

There is generally a limited range of drug treatments for sickle cell disease (SCD) and thalassaemia. For SCD, hydroxyurea, acting mainly by increasing haemoglobin F levels, has shown a convincing reduction in disease-related complications in nearly all clinical studies. Crizanlizumab, a P-selectin blocker that aims at preventing vaso-occlusive episodes, has demonstrated modest and variable benefits in reducing pain in SCD and the same is true for L-glutamine, an essential amino acid required in red cell metabolism. Voxelotor, an orally active benzaldehyde derivative that increases oxygen affinity of haemoglobin thus aiming at preventing haemoglobin S polymerization, shows a consistent increase in haemoglobin, but clinical benefits remain yet unclear. In thalassaemia, luspatercept, a recombinant fusion protein binding to transforming growth factor b superfamily ligands that promotes maturation of erythroid precursor cells, provides a moderate increase in haemoglobin levels in both TDT and non-transfusion-dependent thalassaemia (NTDT). Hydroxyurea remains widely used for thalassaemia patients in

several countries, despite lack of authorization. Finally, mitapivat and etavopivat are pyruvate kinase activators that aim at improving red cell survival and are currently under investigation in SCD and thalassaemia with promising initial evidence.

After Prof. Rees's presentation there was a Patients' testimonial session where three patients were involved: Giorgio Vindigni, sickle-□thal patient from Italy, Anwar Ahmad a thalassemia patient from Saudi Arabia and Tamara Schryver a PKD patient, but also researcher on nutrition from US.

Giorgio Vindigni, a young Italian patient currently living in Bulgaria, explained his life with the sickle cell condition and the strong willingness to live far from any conditioning due to his disease. He showed to be open-minded, indeed he is, currently, studying medicine in Bulgaria but periodically travel back to Sicily for receiving his treatment. He expressed his gratitude to his family for teaching him the important values of life beyond his disease.

Mr Anwar Ahmed a thalassaemic patient, talked about his experience with Luspatercept, a medicine recently approved both in Europe and US for reducing transfusion burden in patients with transfusion dependent thalassemia. Very genuinely, he's explained all the difficulties of thriving with thalassemia in his country therefore as soon as Luspatercept came up he was eager to have it in order to reduce his need of frequent blood transfusions. Unfortunately, he had to halt the treatment after a period due to issues in managing the medicine's side effects.

Tamara Schryver has pre-recorded her witness as she was traveling to Bucharest that day. She talked about her experience with her PKD condition describing all the troubles she went through for being diagnosed (it's a very rare haemolytic anaemia not known in the area of US where she lives) and then for receiving the appropriate care, mainly the splenectomy and a blood transfusion regimen. She expressed how difficult is living with this condition but despite that she was able to live a normal life and to be excellent in her professional life, being an expert in nutrition. She developed these competencies as she considers relevant the diet to tackle some problems due to her anaemia. Recently, thanks to the approval of a medicine able to target specifically the deficiency of the enzyme that provokes the disease (Mitapivat) her life changed completely in a positive way.

Overview of ongoing clinical trials in Haemoglobin Disorders

The session was concluded with the presentation of Dr. Regine Grosse, former paediatric haematologist and oncologist at the University of Hamburg, recently moved to Koblenz. She gave a very interesting overview of the ongoing clinical trials in haemoglobin disorders. Her presentation was mainly focused on Luspatercept for TDT and NTDT patients, on the Piruvate Kinase activators like Mitapivat and Etavopivat for NTDT and TDT, showing the preliminary results of all these studies. Also, Gene Editing, recently approved both in Europe and US, was discussed and some considerations on this treatment has been shared with the audience.

Session 3: New Advances for the Cure of Haemoglobin Disorders

Gene Editing for thalassaemia & SCD: where we stand today

Dr Panos Kottaridis, Consultant Haematologist at University College London Hospital,Medical Director at HCA Healthcare UK at UCLH, UK

Gene editing strategies for thalassaemia and SCD include (i) the correction of the causative mutation of the beta-globin gene, (ii) the inhibition of the alpha-globin gene expression to balance the alpha/beta globin ratio and (iii) the induction of foetal haemoglobin expression. Gene editing with the CRISPR approach, using the Exa-cel product, has been studied in TDT and severe SCD in pivotal phase 3 trials. According to these trials, in TDT, 93% of patients remained transfusion-independent for ≥12 consecutive months, while in SCD, 97% of patients achieved ≥12 consecutive months without a severe vaso-occlusive crisis. The initial results were also positive in terms of safety, including no evidence of off-target effects, chromosomal abnormalities or carcinogenesis. Future research should focus on improving regimens to reduce myeloablation and immunosuppression, reducing the delays in the production line, extending the age and dropping additional exclusion criteria, reducing the cost and managing inequalities and patients' expectations.

Hematopoietic Stem Cell Transplantation in thalassaemia – Benefits & Complications

Prof. Pietro Sodani, Scientific Consultant, Haemato- Oncology Paediatric Section, Bone Marrow Transplant Unit, Charitè Hospital Berlin Germany & Azienda Ospedali Riuniti Marche Nord, Pesaro, Italy

Haematopoietic cell transplantation (HCT) from a matched related or unrelated donor remains the only established curative therapy for haemoglobinopathies. However, approximately 30% of patients have a matched related donor and less than 52% of non-Europeans can find a matched unrelated donor. In addition, the lack of donor registries and the cost of recruiting unrelated donors make matched unrelated donor transplants unaffordable for developing countries where many patients with hemoglobinopathies reside. A haploidentical related donor is often available and represents an alternate source of stem cells and haploidentical HCT (haplo-HCT) is a viable treatment option for selected patients with haemoglobinopathies who lack a suitable donor. CD34+-selected haplo-HSCT for patients with haemoglobinopathies was associated with a high graft failure rate and low disease-free survival, while TCRαβ+/CD19+-depleted grafts is associated with significantly reduced graft failure. However, viral reactivation and infectious complications are frequent, graft-versus-host is not totally avoided, while a major challenge of haplo-HSCT in haemoglobinopathies is the delayed immune reconstitution.

Gene addition for thalassaemia & SCD: latest information

Dr. Carsten Werner Lederer, Associate Professor and Head, Molecular Genetics Thalassaemia Department, The Cyprus Institute of Neurology & Genetics, Nicosia, Cyprus

Gene addition concerns the addition of a normal globin gene to patient's haematopoietic stem cells. Patient's own haematopoietic stem cells are harvested through mobilization and apheresis and are manipulated ex vivo to express the normal beta--globin gene. More specifically, the beta-globin gene is transferred to these cells by a lentivirus vector. After quality and safety checks, the manipulated cells are transfused to the patient following myeloablative conditioning with busulfan that aims at preparing patient's bone marrow for hosting the new cells. These cells are engrafted in the bone marrow and begin to multiply and produce normal red cell. The patient is followed more closely for

a 2-year period and then enters a long-term follow-up period. Pooled data from clinical studies with the beti-cell regimen have overall shown transfusion independence in 82% of patients. However, the considerably high cost of this therapy limits substantially patients' access. Beyond access, additional safety, efficacy and ethical issues still remain to be addressed. Future development in the field may include in vivo or even in utero gene addition or editing.

Session 4: Multidisciplinary care - The third essential cornerstone in the management of Haemoglobin Disorders

Cardiac complications in thalassaemia

Prof. Dimitrios Farmakis, National and Kapodistrian University of Athens Medical School, Athens, Greece

Heart disease has traditionally been the leading cause of death in patients with thalassaemia. Over the past few years, however, therapeutic advances have dramatically reduced the rate of cardiovascular disease and mortality and, in thalassaemia populations with access to proper care, heart disease is not anymore, the leading cause of death. Not only the epidemiology but also the forms of heart disease have progressed following therapeutic advances, from high-output heart failure and pulmonary hypertension, in patients not adequately transfused, to iron overload cardiomyopathy, in patients transfused but not properly chelated, and to minimal or no heart disease in well-treated patients. With the ageing of the thalassaemia populations, by virtue of optimal multidisciplinary care, "new" forms of heart disease related to increasing age may appear, such as atrial fibrillation, atherosclerosis, aortic stenosis or heart failure with preserved left ventricular ejection fraction. Advances in cardiovascular care, such new drugs for heart failure, novel percutaneous interventions for valvular disease and new imaging modalities will help cope with these new challenges, but the key remains the prevention of heart disease with access to proper, multidisciplinary, thalassaemia-specific care.

Impact of bone disease and pain in thalassaemia

Prof. Nicos Skordis, Paediatric Endocrinologist School of Medicine, University of Nicosia, Department of Paediatric and Adolescent Endocrinology Paedi Centre for Specialized Paediatrics, Cyprus

Low mineral mass and reduced bone strength with increased fracture risk are the main causes of morbidity in thalassaemia. The pathogenesis of bone disease in thalassaemia is multifactorial, involving the pathophysiology of the main disease, as well as disease-related and treatment-related complications. Dual-energy X-ray absorptiometry (DEXA) is the “gold standard” for the evaluation of bone mineral density (BMD) and assessment should be performed every 24 months after the age of 10 years. Treatment is required in patients with (i) very low BMD values, (ii) progressive significant BMD loss and/or fragility fractures and (iii) concomitant use of agents known to affect bone turnover (e.g., corticosteroids). Biphosphonates constitute the most widely used treatment but additional clinical research is required to draw more meaningful conclusions for their use in thalassaemia. Treatment should always be individually tailored, balancing benefits and risks. Preventive measures are also of paramount importance, including sufficient blood transfusions, optimal iron chelation, regular physical activity, adequate calcium and vitamin D intake and proper hormonal replacement therapy in cases of hypogonadism.

Management of fertility and pregnancy in thalassaemia

Prof. Nicos Skordis, Paediatric Endocrinologist School of Medicine, University of Nicosia, Department of Paediatric and Adolescent Endocrinology Paedi Centre for Specialized Paediatrics, Cyprus

Pregnancy is feasible, safe and usually has a favorable outcome in women with thalassaemia with normal resting cardiac performance and optimized iron overload, when performed and monitored in specialised centres and by a multidisciplinary team. Assessment before pregnancy ensures eligibility of pregnancy, optimal thalassaemia treatment, absence of iron overload, normal cardiac and endocrine function and absence of abdominal pathology or active infections. During pregnancy, proper management of medications along with multidisciplinary monitoring is required. Special care is also needed during delivery. In addition, in women with hypogonadotrophic hypogonadism, fertility is usually retrievable with proper care. Above all, the desire of thalassaemia women to achieve motherhood should be recognized, respected and approached with special caution and sensitivity and medical and other issues should be addressed prudently, ensuring the best outcomes for mother and baby.

Global Panel of Patients and Healthcare Professionals

Round-Table Discussion: Multi-Generational and Multi-Cultural Dynamics in Haemoglobin Disorders
Moderators: G. Constantinou & E. Georganda

In conclusion of the first day, there was a Round-Table discussion with a global panel of patients and Healthcare Professionals for an interactive discussion chaired by Mr. George Constantinou and Dr. Evgenia Georganda, patient and psychotherapist in Athens, Greece. The discussion has involved all the panellists and was focused mainly on psychosocial aspects of living with thalassemia or other rare anaemias.

Day 2: 18 May 2024

Session 5: Healthcare Professionals Session Case Presentations & Exchange of Best Practices

In this session, a series of interesting cases of patients with thalassaemia were presented, showing the broad spectrum of clinical challenges that can be faced while managing thalassaemia and engaging audience in fruitful discussions on optimal clinical practices and persisting unmet needs. Cases were presented by Dr. Vlachaki, Prof. Farmakis, Dr. Vives Corrons, Prof. Rees, Prof. Skordis, Dr. Kottaridis and Prof. Kuo and the session was coordinated by Prof. Porter and Dr. Vives Corrons.

1st RAIN Summit for people living with Rare Anaemias
Joint Session with patient attendees of Capacity Building Workshop

Session 5: Living with Rare Anaemias

The session started with patients' testimonials about living with a rare anaemia. Mr. Costin Ganescu talked about his personal experience with beta-thalassemia major, from the difficult diagnosis in late '70s in Romania 'till the access to the basic treatment and what this meant for him and his family, moving from a small town in the province to a big city like Bucharest for having the proper treatment under the complex political situation at that time, for those countries lying beyond the "Iron Curtain". Then, Mr. Abdullah Al Fahdi from Oman Hereditary Blood Disorders Association and father of a child with Fanconi Anaemia, talked about his family experience with t such a life-threatening disease, from the desperation that raised soon after his son was diagnosed to the hope for a better future when he realized that Fanconi Anaemia can be treated, back to the desperation when his wife, who lived totally devoted to her son, felt sick for a severe form of cancer and passed away few months later. Now all his energies are focused on ensuring a good quality of life to his son, and he joined the organization with this purpose.

Dr. Evgenia Georganda gave a very interesting presentation about mental health, mainly related to body image and relationships and the ability of people to adjust and to cope with circumstances that are difficult and beyond any control; the ability to function in important areas of our existence, like relationship and work; the ability to change circumstances that can be changed and are harmful, painful or distressing. She further highlighted the existential concerns that give origin to our angst that are: death, existential aloneness, freedom, and responsibility and meaning as despite the life is a miracle people struggle to find purpose and meaning especially when people suffer.

Tamara Schryver talked about nutrition in thalassemia and PKD, being herself a patient suffering from PKD. Her presentation was mainly focused on the Guidelines for nutrition officially disseminated during the TIF International Conference in Kuala Lumpur, Malaysia. She explained why to prepare a guideline for nutrition in rare anemias, just to give a holistic approach to support the medical treatment as nutrition, and more in general the lifestyle, are part of the patient care and malnutrition and nutrition deficiencies have been identified in many settings. Therefore, she suggested to consult a trained expert in nutrition rather than listening advises through social media influencers or consulting unreliable sources on the web. In a very appropriate way, she went through the factors affecting the nutrient intake in thalassemia and PKD, showing the building blocks for nutrition and growth made by macro and micro-nutrients and plant-based bioactive plus tables of nutrients that are particularly important for improving the calorie intake in children and adolescents.

Then, Alejandra Watson, president of PKD Foundation of USA and parent of a child with PKD, talked about how to explain to a child the diagnosis of a rare anemia and how to deal with it. She provided lot of important suggestions on how to approach such a delicate conversation with a child, trying to assess initially the level of maturity and confidentiality with the medical terminology, having the patience to explain with no hurry and being prepared to answer specific questions. Addressing emotional reactions is important as well as building a supportive environment being creative in expressing the situation as a normal one. It's fundamental to understand that everyone with a rare disease doesn't have to struggle alone but supported by parents, friends, peers, doctors and the specific patients' organizations.

Session 6: Capacity Building for Patient Associations - Breakout working groups

Moderators: Mr George Constantinou & Mr Ganescu Costin Radu

This was a breakout session. A brief introduction by Mrs. Katia Pelides from TIF Office, explained the objectives of this session: strengthening competencies and capacities of patients' representatives and to empower leaders with skills to enhance their partnership with medical community and national health authorities. The patients' reps were divided into 4 different tables with 4 different topics: 1) Fundraising: how to gain supporters; 2) The powers of collaborations: Building an effective partner network; 3) Effective advocacy skills; 4) The importance of communication and visibility for non-profit organizations.

Table 1. Fundraising: How to gain supporters
Facilitator: Mr Ganescu Costin Radu
Rapporteur: Mr Ganescu Costin Radu

In order to operate organizations need to have proper funds and for that each organization should consider their fundraising strategy. So, we started the discussion with what we are currently doing to raise funds and what are the main sources of financing.

Among the main fundraising methods are the banquets organized for the benefit of the association or food fairs where all sales are collected by the association and of course the projects carried out with the support of pharma or non-pharma companies.

We have determined that in order to increase the visibility of these activities and bring as many supporters as possible from outside our community, we must be as transparent as possible and transmit the information about the activities carried out by the association on as many social information channels as possible, mass media, radio TV, and use if it's possible blogger or vlogger that which are already very well known to support out association.

We also discussed the possibility of supporting the organization not only from a financial point of view, but even sponsorships such as IT equipment, IT licenses, free access to a space of the municipality would be a financial advantage because it would save certain expenses that should be made from their own funds and thus a part of them will be redirected to other important activities of the organization.

We have come to the conclusion that the funding of the organization must be diversified in order to have stability from the point of view of the association's functioning and thus any possibility to access local or national funds or if we look at the European level, European funds would increase both the level of the organization and the importance of the organization as a decision-making partner.

But as a conclusion, an increase in funding and in our community can only be done if we change the activity of the association from one based on volunteering to an activity carried out with professionals who can bring financing and implementation of projects to a higher level.



Table 2. The Power of Collaboration: Building Effective Partner Networks
Facilitator: Mr Loris Brunetta
Rapporteur: Ms Alejandra Watson

During this roundtable on collaboration, the team highlighted the fundamental role of collaboration in nonprofit organizations (NPOs) and addressed several challenges, including:

Capacity Building: Limited human and financial resources hinder growth and effectiveness.

Lack of Volunteers: Volunteers often feel frustrated by the slow progress in developing patient programs, further exacerbated by resource constraints.

The team emphasized the need for collaboration to create support programs for low socioeconomic groups and recognized the efforts of the Thalassemia International Federation (TIF) in addressing rare anaemias. Despite TIF's respect for NPO individuality, there is a critical need to improve awareness and access to haematologists for rare anaemias.
The discussion underscored the severe social and economic disparities in developing countries, leading to high mortality rates from rare anaemias due to a lack of medical opportunities. Through collaboration, we aim to close these inequity gaps by:

• Creating educational programs for healthcare communities.
• Advocating for government financial support for treatments, especially for the most vulnerable populations.

Collaboration empowers organizations by pooling their collective influence, enabling them to achieve their missions and broader systemic changes more effectively. By leveraging joint efforts, NPOs can experiment with various solutions and align their activities with other stakeholders.

Given the unprecedented complexity of current social, political, and economic challenges, isolated efforts are insufficient. Effective collaboration is essential for creating significant social impact. NPOs should focus on why collaboration is necessary and explore various methods, including cooperation and coordination to maximize the impact. Ultimately, by combining forces we can lead to transformative changes, making a substantial difference in the world.

"If we add the numbers, we can make it to the headlines."

Table 3. Effective advocacy skills
Facilitator: Mr George Constantinou
Rapporteur: Ms Imola Moricz

1) meaning of advocacy
- Represent needs and rights
- Advocate patients' rights
- Market the Patient voice
- Raise awareness
- Process of supporting and enabling ppl to express their voice through us

2) what does an advocate look like:
- Like a lawyer representing someone else's voice
- Has knowledge and understands.
- Promote on requirements.
- Personal experience (disease)

3) Thalassemia advocate should have:

- Personal experience.
- Listening stills, empathy, charisma, credibility ability to socialize
- Expert on the treatments, developments, research that needed for financial support
- Expert of how the health care system (hospital) both s locally internationally
- Experts know of how the government and political system works - what would they be willing to do, Qualities, credible, knowledgeable (facts, details), Has support of society and of the patients not afraid to raise their voice, good comm. Skills, persistent & professional collaboration.
- Widespread knowledge (health care, government, disease)

4) What do we advocate for?

- To have a life quality
- Advocating for the doctors - new medications
- To have: Blood transfusion, adequate supply of blood
- To have: Haemoglobin Disorders centres
- Multidisciplinary care
- International treatment protocols
- Mental care
- Analyse the needs you need but you don't have
- Another type of advocacy from the health care to the patients try new drugs that can improve quality of life

5) Who do we advocate to?

- Patient associations
- Young doctors
- Insurance companies
- National decision maker
- Ward nurse
- Specialist nurse of the ward
- Treating doctor
- Hospital manager
- government officials.

PRIORITIES

1. Must have (having blood)
2. Should have (selection of all the medicines)
3. Could haves (unit opening on the weekend)
4. Would like to have (in the future)
5. if everything is a priority nothing is a priority

Table 3. Communication & visibility
Facilitator: Bessie Calabria
Rapporteur: Dore Peereboom

Non-profit organisations communicate with patients, parents and society

- LinkedIn.
- Twitter.
- Facebook.
- Newsletter.
- (Conference; patients and parents but hard to reach society);
- Tiktok: challenge is that not many people want to be visible in video's so use non-identification material.
- Website: Challenges of a website is to keep it up to date. Way to do this is to give volunteer accountability and responsibility for their content creation: deploy having ownership of a program.
- E-mail: to reach out if people need for example fill in a survey. Challenge is to reach younger population because they often do not check their inbox.

Visibility of an organisation or disease

- Potential donors.
- Connect with new patients.

Suggestion is to connect with blood operators / blood banks to organise something for national blood donor week to reach out to donors and the public to find more donors. Or visit a blood bank in your city.

It's also important to be visible towards your governments or doctors. You can join national alliances for rare anaemias or rare diseases to have one strong voice. You can organise National Thalassemia Day or National Pyruvate Kinase Deficiency Day.

Funding

- Find funding for research (if this is allowed in your country of residence);
- Try to find a (Rare) Blood Disorders organisation to find funding together.

Challenges:

- How to combine or find balance between foundation and paid job;
- Call for volunteers often.
- Other ideas for visibility:
- Organise a fundraiser.
- Hold a walk for support / visibility.
- Researchers collaborations.
- Invite collaborators / stakeholders (doctors, researchers, government, blood bank) to attend your conferences.

Session 7: Management for Rare Anaemias: Guidelines

Guidelines for the management of thalassaemia: History of development and methodology

Prof. Dimitrios Farmakis, National and Kapodistrian University of Athens Medical School, Athens, Greece

The development and publication of clinical practice guidelines for the management of thalassaemia and other haemoglobinopathies represents one of the key activities of Thalassaemia International Federation (TIF). Guidelines aim at promoting optimal management for all people living with haemoglobinopathies through standardization and harmonization of care across the globe, while acting as a strong educational tool and supporting advocacy. The development of guidelines follows a well-defined stepwise process, involving experts from all over the world covering different aspects of the diagnosis, monitoring and treatment of the disease and its complications, and ensuring inclusion of state-of-art and up-to-date recommendations. The first guideline document was issues by TIF in 2000 and since then, a long list of publications and updates have been released. The latest documents include the 2021 guidelines for the management of transfusion-dependent thalassaemia (4th edition) and three new publications in 2023, covering the management of non-transfusion-dependent beta-thalassaemia, the management of alpha-thalassaemia and a nutrition guide, respectively. TIF also ensures the proper dissemination of its guideline documents, making them available totally free of charge through TIF's website as well as by congress and workshop sessions, dedicated webinars and with the online courses of the TIF e-Academy.

The New Guidelines for the Management of pyruvate kinase deficiency (PKD).

Prof. Kuo, Ass. Professor at the Division of Haematology at the University of Toronto presented

The goal of the International Guidelines for the Diagnosis and Management of Pyruvate Kinase Deficiency was to develop evidence-based guidelines for the clinical care of patients with PK deficiency. These clinical guidelines were developed by use of GRADE methodology and the AGREE II framework. Experts were invited after consideration of area of expertise, scholarly contributions in PK deficiency, and country of practice for global representation. The expert panel included 29 expert physicians (including adult and paediatric haematologists and other subspecialists), geneticists, laboratory specialists, nurses, a guidelines methodologist, patients with PK deficiency, and caregivers from ten countries. Five key topic areas were identified, the panel prioritised key

questions, and a systematic literature search was done to generate evidence summaries that were used in the development of draft recommendations. The expert panel then met in person to finalise and vote on recommendations according to a structured consensus procedure. Agreement of greater than or equal to 67% among the expert panel was required for inclusion of a recommendation in the final guideline. The expert panel agreed on 31 total recommendations across five key topics: diagnosis and genetics, monitoring and management of chronic complications, standard management of anaemia, targeted and advanced therapies, and special populations. These new guidelines should facilitate best practices and evidence-based PK deficiency care into clinical practice.

At the end of this session, three patients' representatives talked about their perspective to live with a rare anaemia, Mr. Constantinou for thalassemia, Mr. Lander for PKD and Mr. James, pre-recorded for SCD. Every one of them talked about the importance of having patients involved in the preparation of the scientific Guidelines as it's something very useful for the healthcare professionals as well, often clinicians and nurses rely on the information gathered from the patients on how to live with a rare disease, patients know much better than anyone else what is better for themselves and how to incorporate their treatments within their lives without losing quality. Clinicians understand the importance of having patients involved. Mr. Constantinou went through the TIF Publications as important tools for patients' advocates, as being knowledgeable make the advocates reliable for the stakeholders. He also explained the experience done with the UKTS, many years ago, for implementing the so called "Standards for the Management of Thalassemia".

Day 3: 19 May 2024

RAIN Summit (RAIN Members Strategy Session)

Chaired by Prof. John Porter and Prof. Joan-Lluis Vives Corrons

The 1st RAIN Summit for Patients with Rare Anaemias, held on May 19, 2024, at the Grand Hotel Bucharest, Romania, was part of the “Capacity Building Workshop for Thalassaemia Patients & Healthcare Professionals”. The Session was chaired by Prof. John Porter and Prof. Joan-Lluis Vives Corrons and aimed to foster dialogue between healthcare professionals and patients, providing a comprehensive overview of current research, treatment options, and patient care strategies for rare anaemias The programme included a series of sessions designed to address both scientific advancements and patient-centric approaches in managing rare anaemias. The programme included a combination of lectures, panel discussions, and interactive Q&A segments.

Lectures

J. Porter opened the Session with welcome remarks and an introduction to the summit’s objectives: 1. Implement the importance of collaborative efforts between researchers, clinicians, and patient advocacy groups and 2. Overview of the session structure and key topics to be covered.

L.Cannon presented a pre-recorded overview of the several significant milestones achieved by RAIN in its mission to support individuals with rare and ultra-rare anaemias worldwide.: Global advocacy, educational initiatives, awareness campaigns, patient support and collaborative projects. These accomplishments reflect RAIN's dedication to improving the lives of individuals with rare anaemias through advocacy, education, and collaboration.

J.Porter presented the current therapeutic approaches for rare anaemias encompassing a variety of advanced and emerging treatments tailored to the specific needs of these complex conditions: Gene therapy, targeted drug treatments, haematopoietic stem cell transplantation (HSCT), m-RNA therapies, next-generation sequencing (NGS), artificial intelligence (AI), supportive care and symptom management. These approaches reflect the significant advancements in the understanding and treatment of rare anaemias, emphasizing the importance of continued research and international collaboration to improve patient outcomes and ensure access to these innovative therapies

JL Vives Corrons presented new developments in the diagnosis of rare anaemias and thalassaemias. With a focus on advanced genetic, metabolomics and ektacytometric technologies, which significantly enhance the accuracy and speed of diagnosis, as well as provide insights into the underlying pathophysiological mechanisms. Outstanding achievements have been the new-generation sequencing (NGS), which has become a cornerstone in the diagnosis of rare anaemias and thalassaemias and artificial intelligence including machine learning. These tools are particularly useful in interpreting results from high-throughput sequencing and

identifying patterns that may be missed by traditional methods, and facilitate personalized medicine approaches

Finally, K. Kuo presented the recent clinical trials and advancements in the care and cure of rare anaemias with the significant strides being made in various therapeutic approaches. Numerous clinical trials are ongoing to test the efficacy and safety of these advanced therapies. For example, trials for new gene therapies in beta thalassemia and SCD, and for novel drug treatments like Mitapivat and Luspatercept, are showing promising early results These advancements reflect a rapidly evolving landscape in the treatment of rare anaemias, driven by innovative therapies and improved diagnostic tools. Continued research and collaboration among scientists, clinicians, and patient advocacy groups are essential to further these developments and bring effective treatments to patients worldwide

Each session concluded with an interactive Q&A segment, allowing participants to engage directly with the speakers. Key discussions during these segments included: the impact of new research on clinical guidelines, practical challenges in managing rare anaemias in different healthcare settings and opportunities for patient involvement in research and clinical trials.

CONCLUSION

This **1st** RAIN Summit provided a comprehensive and interactive platform for discussing the latest advancements and challenges in the field of rare anaemias, highlighting the importance of collaboration among researchers, clinicians, and patients to improve care and treatment outcomes. The insights and discussions from these sessions are expected to drive future research and enhance patient care strategies.

ACKNOWLEDGEMENTS

The success of these sessions was made possible by the contributions of all speakers, participants, and organizers. Special thanks to Prof. John Porter and Prof. Joan-Lluis Vives Corrons and for their leadership and dedication to advancing the field of rare anaemias.

NEXT STEPS

1. Strengthening research collaborations and networks.
2. Enhancing educational initiatives for healthcare providers and patients.
3. Advocating for increased funding and support for rare anaemia research and treatment

4. Post-Conference Questionnaire Analysis

Capacity Building Workshop for Thalassaemia Patients & Healthcare Professionals & 1st RAIN Summit for Patients with Rare Anaemias
16-19 May 2024
Grand Hotel Bucharest, Romania
POST WORKSHOP QUESTIONNAIRE

TOTAL NUMBER OR RESPONSES: 52

Q1. What were the main reasons for your participation in this Workshop?	**RESPONSES**
- The level and content of the programme	28
- The expertise of the faculty speakers/presenters	31
- The need to be updated with reliable information on Haemoglobin Disorders	34
- Networking with and/or meeting colleagues	35
- Other (please specify)	

I received sponsorship to attend

Q2. How did you find the following?

(Please rate on a scale from 1 to 5 with: 1 = not at all useful ; 5 = very useful)						**AVERAGE**
- Quality of the presentations	1	2	3	4	5	**4.38**
REPLIES	**1**	**0**	**6**	**16**	**29**	
PERCENTAGE	**2%**	**0%**	**12%**	**31%**	**56%**	

- Format of the conference		1	2	3	4	5	**4.23**
	REPLIES	**1**	**1**	**8**	**17**	**25**	
	PERCENTAGE	**2%**	**2%**	**15%**	**33%**	**48%**	
- Relevance to your concerns and to the reasons for your participation		1	2	3	4	5	**4.08**
	REPLIES	**1**	**0**	**11**	**22**	**18**	
	PERCENTAGE	**2%**	**0%**	**21%**	**42%**	**35%**	
- Usefulness to your Association's work		1	2	3	4	5	**4.26**
	REPLIES	**1**	**0**	**6**	**21**	**22**	
	PERCENTAGE	**2%**	**0%**	**12%**	**40%**	**42%**	
- Opportunities for networking		1	2	3	4	5	**4.35**
	REPLIES	**1**	**0**	**6**	**18**	**27**	
	PERCENTAGE	**2%**	**0%**	**12%**	**35%**	**52%**	

- Comments

Very useful
Nicely managed & informative
Topics related to scientific issues were not satisfactory
Very successful workshop, informative, excellent organisation and fantastic networking. Keep it up :). It would be great if you shared the presentations on TIF website
Too much information in short time. We need more time for questions
More breakout groups-sessions
Too much input. Too many topics
Thank you for this workshop
The presentations are very professional, too medical for the simple patient mind
Thank you

Q3. Would you like to have had more, or less of the following? (Please rate on a scale of: 1=less ; 5=more)							**AVERAGE**
- Lectures		1	2	3	4	5	**3.53**
	REPLIES	**4**	**6**	**14**	**13**	**14**	
	PERCENTAGE	**8%**	**12%**	**27%**	**25%**	**27%**	
- Question and answer sessions		1	2	3	4	5	**4.14**
	REPLIES	**1**	**0**	**10**	**18**	**20**	
	PERCENTAGE	**2%**	**0%**	**19%**	**35%**	**38%**	
- Discussions in small groups		1	2	3	4	5	**3.9**
	REPLIES	**2**	**2**	**13**	**15**	**18**	
	PERCENTAGE	**4%**	**4%**	**25%**	**29%**	**35%**	
- Case studies		1	2	3	4	5	**3.78**
	REPLIES	**2**	**1**	**18**	**14**	**15**	
	PERCENTAGE	**4%**	**2%**	**35%**	**27%**	**29%**	

Q4. Did the Workshop meet your expectations in the following ways? (Please rate on a scale from 1 to 5 with: 1 = not at all ; 5 = a great deal)							**AVERAGE**
- I received useful information		1	2	3	4	5	**4.31**
	REPLIES	**0**	**1**	**8**	**17**	**26**	
	PERCENTAGE	**0%**	**2%**	**15%**	**33%**	**50%**	
- I updated my knowledge		1	2	3	4	5	**4.12**
	REPLIES	**0**	**2**	**11**	**18**	**21**	
	PERCENTAGE	**0%**	**4%**	**21%**	**35%**	**40%**	

- I established new contacts		1	2	3	4	5	**4.27**
	REPLIES	**2**	**0**	**6**	**18**	**26**	
	PERCENTAGE	**4%**	**0%**	**12%**	**35%**	**50%**	

- Other (please specify):

Not so much time to establish new contacts due to the intense schedule

The information was perfect but the consecutive programs are far too tiring for the patients to keep up with because of the fatigue you hardly absorb the programme. Personally I think is is a much to full agenda in 1 day

Not many sickle cell presentations and subjects unfortunately

Not enough space, time and opportunity to network and discuss. Tight schedule. Very busy

Q5. What is the key information or knowledge that you have obtained during the Workshop?

Q6. In your opinion, what was the highlight/ best part of the Workshop?

Q7. Would you like to participate in another TIF event in the future?	**RESPONSES**
- Yes, but only if the costs of travel and accommodation will be covered	45
- Yes, even if I have to cover my own costs	
- No, because (please explain	

I came first time at my own expenses & would like to visit in future if TIF help to cover some or whole expenses of travel/accommodation

(If I can afford the cost)

Q8. How satisfied were you with the following organisational aspects? (Please rate on a scale from 1 to 5 with: 1 = not satisfied ; 5 = extremely satisfied)							**AVERAGE**
- Registration process		1	2	3	4	5	**4.66**
	REPLIES	**0**	**1**	**3**	**7**	**36**	
	PERCENTAGE	**0%**	**2%**	**6%**	**13%**	**69%**	
- Workshop materials provided		1	2	3	4	5	**4.39**
	REPLIES	**0**	**2**	**6**	**12**	**29**	
	PERCENTAGE	**0%**	**4%**	**12%**	**23%**	**56%**	
- Secretariat – helpful and courteous (before and during the conference		1	2	3	4	5	**4.6**
	REPLIES	**0**	**0**	**4**	**12**	**34**	
	PERCENTAGE	**0%**	**0%**	**8%**	**23%**	**65%**	
- Audio-visual facilities		1	2	3	4	5	**4.27**
	REPLIES	**0**	**2**	**11**	**8**	**28**	
	PERCENTAGE	**0%**	**4%**	**21%**	**15%**	**54%**	
- Location		1	2	3	4	5	**4.46**
	REPLIES	**0**	**0**	**7**	**13**	**30**	
	PERCENTAGE	**0%**	**0%**	**13%**	**25%**	**58%**	
- Accommodation		1	2	3	4	5	**4.66**
	REPLIES	**0**	**0**	**3**	**10**	**34**	
	PERCENTAGE	**0%**	**0%**	**6%**	**19%**	**65%**	
- Overall level and quality of communication during your preparation for participation at the Workshop		1	2	3	4	5	**4.48**
	REPLIES	**0**	**1**	**5**	**12**	**30**	

	PERCENTAGE	**0%**	**2%**	**10%**	**23%**	**58%**

- Other (please specify): *The daily programming was a little long and it was so consolidated in 2 days. This might be as light more comfortable the 4th session each day reduced to have a half day more*
It will be interesting for me to have translation in French
Thanks to Rawad Merhi and Catherine Skari
Everything was greatly organised. Thank you so much
The time was short to discuss more after the presentation
More time. Not so full agenda. For patients it is very exhausting
Because I don't find anything in Italian language
IASG- Side meeting 1
Great effort for TIF. I'd like to send you many thanks to all members

Central European Patient Leaders' Network

Meeting Details:

- Date: 16 May 2024
- Venue: Opera Room, Grand Hotel Bucharest, Romania
- Time: 17:00 – 18:30

Participants:

- Sabrina Souaifi (France)
- Mohamed Ghoubali (France)
- Adel Mansouri (France)
- Stamatula Trianti (Germany)
- Enzar Hikmet Demiray (Germany)
- Nicola De Nittis (Germany)
- Kaplan Mehtap (Germany)
- Kostas Tsurlis (Germany)
- Maria-Lucia Nieddu (Germany)
- Giorgio Pio Vindigni (Italy)
- Faried Guman (Netherlands)

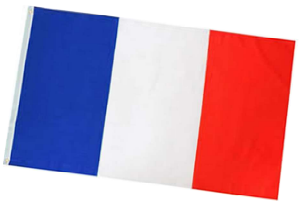
Background:
This meeting is part of Thalassaemia International Federation's overall strategic plan of creating regional networks worldwide for the purpose of:

- Peer support and empowerment.
- Collaboration through networking and exchange of best practices fostering advocacy and empowering initiatives with the potential of meaningful change.
- Creating a platform for capacity building and skill development
- Enhancing visibility and representation in decision-making processes at the national and international levels, by engaging policymakers, healthcare providers, and other stakeholders.

The purpose of this inaugural meeting was to gather information about the current situation in each country and identify common and specific national needs. Quarterly meetings will follow, with agendas based on the identified unmet needs, for the purpose of planning tailored national and regional activities.

TIF Presentation:
A presentation was given by Mr Loris Brunetta, one of TIF's founders, Board Member and experienced patient advocate, on the importance of patient leaders' networks.

FRANCE



Report was provided by Adel Mansouri.
Current Healthcare Status

- **Prevalence:**
  865 tients recorded on national registry.
  866
- **Screening:**

No specific screening programme for thalassaemia is in place however, there is a screening programme for sickle cell disease which could diagnose thalassaemia cases if individuals are originally from the same region as SCD patients.

Screening could be recommended by the doctor, based on family assessment

Healthcare provided:
For children the care that is provided is adequate however, when one transitions to adult care there are some challenges, such as the patient needing to make their own arrangements to see specialist doctors and comply with the appropriate diagnostic/monitoring tests.

- **Blood Donation Issues:**
  - Adequate however, there are regions in France where the number of thalassaemia patients is low, blood supply needs are covered by other regions' reserves.

- **Diagnostics:**
  - Cardiac T2* measurement is done from the age of 6 years old.
  - LIC is done once a year.

- **Multidisciplinary Care**
  - Available in specialised centres, at least once a year.

- **Reimbursement**
  - 100% of the cost of services are covered by the NHS.

- **Employers' Flexibility**
  - Patients are eligible for one day off per month, upon request by the employee.

- **Needs:**
  - Prevent isolation of patients. There are national programs to promote patient involvement and patient groups to help with transition from child to adult.

GERMANY

Information provided by:
- Kostas Tsourlis

Patient Demographics:
- Estimated number is between 600-1000 including NTDT patients.
  - Approximately 500 of these patients are TDT patients.
  - Note that not all NTDT patients are registered.

Blood Supplies:
- Blood supplies are adequate.

Screening:
- No prenatal or antenatal screening programme in place.

- Screening is available after recommendation from the doctor.

Diagnostics:

- Cardiac iron levels measured yearly.
- In most university hospitals and private hospitals measure LIC with FerriSmart which is measured twice a year.
- Bone tensity test is not offered on an annual basis.

Reimbursement:

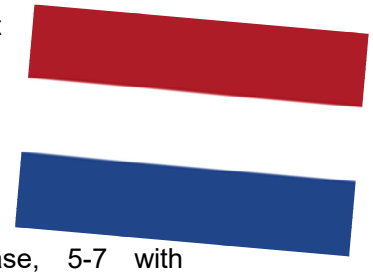
- There is reimbursement however, on many occasions the diagnostic/monitoring testing for thalassaemia need to be advocated for reimbursement to the gov/insurance.

Needs & Challenges in Thalassaemia Management

- **Multidisciplinary Care**
  - **Access to Specialists:**
    - Once a patient is transitioned from child to adult care, he/she needs to plan on his own his appointments with specialists and/or remind his haematologist that an appointment is needed with a specialist. Whereas for children, specialists will see the child on the day they receive their transfusion.

- **Lack of consistency of following national and international guidelines on treatment.**
- **Employment and Social Challenges**
  - Workplace Flexibility:
    - Employers are not always flexible. At times an employee is asked to take time off from their annual leave or work overtime to be able to receive their treatment once a month.
  - Need the flexibility of receiving transfusions on weekends.
- **Raise awareness among new doctors or medical students of various specialisations about haemoglobinopathies.**

NETHERLANDS:



Information provided by Faried Guman, Patient Representative, OSCAR Nederland

Demographics:

- Sickle Cell Disease: 1800-2000 individuals
- Thalassaemia: 275-300 individuals
- Carriers of hemoglobinopathies: Over 200,000
- Yearly newborns: 50-60 births with severe hemoglobinopathy (35-40 with sickle cell disease, 5-7 with thalassaemia)
- Trends:
  - Despite being rare, the prevalence of these diseases is increasing due to migration, new refugees, and consanguineous marriages.

Treatment in the Netherlands:

- Expertise Centers (Treatment & Research):
  - Amsterdam, Rotterdam, Utrecht, Leiden, Den Haag
- Shared Care Centers (Treatment):
  - Nijmegen, Groningen

- National Guidelines:
    - Sickle Cell Disease: National guidelines available since 2017 for both children and adults, to be updated.
    - Thalassaemia: Previously used TIF guidelines; Dutch national guideline available since 2023.

Access to Treatment and Care:

- Insurance:
    - Mandatory standard health insurance for all residents, costing between 125-200 euros with an own-risk amount of 385 euros monthly.
    - Additional insurance available for more coverage.
- Referral System:
    - Patients need referrals for hospital visits, except for emergency cases of sickle cell pain crises.
    - Everyone is entitled to one second opinion.
- Primary Care:
    - First-line care through General Practitioners (GPs); each resident has a personal GP.
- Emergency Services:
    - Emergency line must be contacted first; non-critical cases are referred to GPs

Prevention:

- Newborn Screening:
  Since 2007, all newborns are screened for hemoglobinopathies via the heel prick test.
  Severe forms of thalassaemia included separately since 2017.
- Carrier Testing:
  Available through GPs based on necessity.

Treatments:

- Sickle Cell Disease:
  Casgevy (CRISPR-CAS 9) in collaboration with Vertex and local doctors.
  Voxelotor for severe anemia, in collaboration with Pfizer for conditional admission.
  Stem cell transplantation for adults and children.
- Thalassaemia:
  Casgevy (CRISPR-CAS 9) and collaboration with Vertex and local doctors.
  Reblozyl (Luspatercept) for beta-thalassaemia major and intermedia.
  Stem cell transplantation for children.

Meeting Details:

- Date: 16 May 2024
- Venue: Opera Room, Grand Hotel Bucharest, Romania
- Time: 15:00 – 16:30

Participants:

- Anduela Pinguli (Albania)
- Denisa Malo (Albania)
- Lili Andreeva (Bulgaria)
- Athanasios Mylonas (Greece)
- Ahmet Varoğlu (Turkey)
- Tayfun Aksoy (Turkey)
- Costin Radu Ganescu(Romania)
- Ioana Olteanu Radu (Romania)

Background:

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- Peer support and empowerment.
- Collaboration through networking and exchange of best practices fostering advocacy and empowering initiatives with the potential of meaningful change.
- Creating a platform for capacity building and skill development
- Enhancing visibility and representation in decision-making processes at the national and international levels, by engaging policymakers, healthcare providers, and other stakeholders.

The purpose of this inaugural meeting was to gather information about the current situation in each country and identify both common and specific national needs. Quarterly meetings will follow, with agendas based on the identified unmet needs, and will include planning tailored national and regional activities.

TIF Presentation:

A presentation was given by Mr Loris Brunetta, one of TIF's founders, Board Member and experienced patient advocate, on the importance of patient leaders' networks.

ALBANIA

Report was provided by Ms Anduela Pinguli, Albanian Thalassaemia & Oncho-Haemoglobinopathie Association (AT&OHA) representative. The Association is based in Tirana, and it is the first and only patient-created association for thalassaemia in Albania.

Demographics

- **Prevalence:** 7.1% of the population are carriers of Beta Thalassaemia and Sickle Cell Disease (SCD).
- **Patient Distribution:** Approximately 500 patients across three centres:
    - Tirana: 150 patients
    - Lushnja: 250 patients
    - Fier: 100 patients

Screening

No government policy for preventing new births of Thalassaemia and SCD.

Healthcare Services Challenges

- **Inadequate Facilities:**
    - There are three “centres” in Albania for at least 500 patients.
        - Lushnje
            - 2 doctors (paediatrician and adult haematologist) and 3 nurses serving 200 doctors.
        - Tirana
            - Tirana’s thalassaemia centre is overcrowded and inadequately staffed serving patients of all ages.
            - Patients must take time off from work or school for treatments as the centre’s working hours are only between 8:30 and 14:30.
            - Insufficient transfusion facilities (3 small transfusion room serving 12 patients per day) and medical staff (1 doctor and 2 nurses).

- **Blood Donation Issues:**
    - Blood needs are mainly met in Tirana, with shortages in other cities.
    - Efforts by the Ministry of Health and NGOs are insufficient.

- **Protocol Implementation:**
    - Lack of multidisciplinary care for adults.
    - Patients must navigate a bureaucratic healthcare system.
    - Critical diagnostic tests like MRI are unavailable.

Patient Treatment and Medication

- **Treatment Gaps:**
    - Removal of Exjade from the national reimbursement list three years ago.
    - Current medications include four types of generic deferasirox which are reimbursed.

- **Patient Education:**
    - Efforts are ongoing but hampered by insufficient resources and patient engagement.
    - Previous NGO failures have led to mistrust and lack of volunteerism.

Successes and Challenges

- **Successes:**
    - Memorandum with the National Blood Bank.

    - Partnership with the Network of Organization of Patients with Chronic Diseases.
    - Free osteoporosis and vitamin D testing for 250 patients supported by Novartis.
- **Challenges:**
    - Need for a dedicated thalassaemia centre.
    - Application of protocols for adult patients.
    - Improvement in the quality and quantity of blood donations.
    - Enhanced collaboration with other associations and health institutions.
    - Creation of an online platform and patient database.
    - **Future Collaboration Needs**
- **Support from TIF:**
    - Training for doctors and nurses.
    - Creation of a consultation platform for critical patient cases.
    - Advocacy for a dedicated thalassaemia centre.

BULGARIA

Information provided by:

- Assoc. Prof. Atanas Banchev, Dr.Denka Stoyanova “Pediatric center for Haemophilia and Thalassaemia”, Sofia;
- Dr. Mirella Rangelova “Hematology and Transfusiology Center for Thalassaemia”, Sofia.
- Lili Andreeva - Patient Leader” Bachelor of Pharmacy”

General Care:

Patient Demographics:

- 290 TDT patients
    - Thalassaemia major patients are 270,
    - TDT Intermedia patients are approximately 20.
    - Less than 100, out of the 290 patients, are under 18 years old.

Treatment Centres:

- Patients are managed in centres located in Sofia, Plovdiv, Varna, Stara Zagora, and Pleven.
- Available chelators (Exjade, Ferriprox, Desferal) are reimbursed by the National Health Insurance Fund (NHIF).

Challenges in Thalassaemia Management

- **Blood Supplies:**
    - Generally adequate but delays occur, especially for negative blood groups and around public holidays.

- **Screening:**
    - No national prenatal or antenatal screening program for carriers.
    - Screening is occasionally available in private cases at the discretion of doctors.

- **Diagnostics:**

- Cardiac and liver iron levels are monitored via MRI T2* annually or biannually based on clinical indicators.
- Serum ferritin is tested quarterly.
- Ferriscan access is limited to clinical trials or specific programs.

Multidisciplinary Care

- **Access to Specialists:**
  - Patients have access to a multidisciplinary team including cardiologists, endocrinologists, gastroenterologists, hepatologists, psychologists, and reproductive medicine doctors.
  - NHIF covers treatments and regular visits to specialists.

Employment and Social Challenges

- **Workplace Flexibility:**
  - Employer flexibility for time off due to transfusions varies.
  - Some job positions for disabled individuals are facilitated by the Agency of Employment.
  - The Ministry of Labor and Social Policy provides tax breaks for companies employing disabled individuals.

Comprehensive Treatment Centres

- **New Centre:**
  - In April 2021, a new Comprehensive Treatment Centre for Haemophilia and Thalassaemia was established in Sofia.
  - The centre provides services for about 200 patients, including 45 children under 18 with thalassaemia major.
  - There are three comprehensive centres in Bulgaria: two in Sofia and one in Varna.

Pending Issues and Needs

- **Screening Programs:**
  - Lack of a national mandatory screening program for severe hemoglobinopathies.
- **Blood Donation:**
  - Shortage of red blood cell concentrates, particularly for rare blood groups.
  - Need for government-encouraged blood donation programs.
- **FerriScan Access:**
  - Not all patients have access to FerriScan, as it is not covered by NHIF.
- **Medical Staff Turnover:**
  - High turnover of medical personnel in thalassaemia centres.
- **Patient Registry:**
  - Absence of a comprehensive national registry for thalassaemia patients.

Recommendations and Future Directions

- **Psychological Support:**
  - Increased psychological support for thalassaemia patients, with recent appointments of psychologists in Sofia centres.
- **Public Awareness:**
  - Greater efforts in advertising and publicizing the importance of prenatal and antenatal screening.
- **Blood Donation Initiatives:**
  - More initiatives and programs to enhance blood donation and supplies.
- **Collaboration:**

- Enhanced collaboration among NGOs to address these needs effectively through meetings, conferences, and events.

GREECE:

Information provided by Athanasios Mylonas

Demographics:
- Approximately 3,000 patients in the country.

Blood Donations:
- Adequate supply however, better management is needed.
- A shortage is noticed during the month of August and holidays. When there is a shortage of blood supply, it can be supplemented by reserves of other countries.

Multidisciplinary Care:
- Access to all specialists.
- Reimbursement by the government for all treatments and visits to various specialists.

Needs:
- Raise awareness among blood donors.
- Better collaboration between multidisciplinary medical team.
- Accessibility to new treatments.

ROMANIA

Information provided by Mr Costin Radu and a parent, Ms Ioana Olteanu Radu

Demographics:
- Approximately 200 patients. 100 to 140 of these patients live in Bucharest.
- Birth rate has decreased to 1-2 per year.
- The number of patients in the past 10 years is stable.
- 25 years ago, there were 340 patients.

Blood Supply:
- Adequate supply

Diagnostics:
- Only few hospitals have MRI.

Needs for children specifically ones who are not living in Bucharest:
- Children are treated in the same centre with oncology patients.
- Lack of multidisciplinary care for children.
- Children and adults are not treated in the same centre.
- Treating doctors for children should be permanent staff on the unit.
- Lack of consistency with chelators used for children. Every month the chelator is changed.

- Children need to travel and stay approximately 3 days a month in the hospital to receive their treatment.
- International clinical guidelines are not consistently followed.
- Lack of specialised haematologists.

Needs for adults:
- International clinical guidelines are not consistently followed.
- Increase availability of MRI T2* and reimbursement.
- Availability of a multidisciplinary healthcare team.
- Support in the transition from child to adult care.

TURKEY:

Information provided by Mr Ahmet Varoglu.

Demographics:
- There are about 10,000 thousand people living with thalassaemia.
- 12% of patients need to travel to another city to receive care.

Screening:
- Screening tests are done in all the provinces for all couples.

Blood Supplies:
- Red Crescent helps the patients to find the required blood supplies.
- Patient Federation and societies carry out awareness projects to recruit more donors.

Diagnostics:
- T2* is available in 6 provinces such as Ankara, Adana, and other big cities.
- The Mediterranean part of the country, up to the north and east parts of Turkey, there are problems with the availability of T2*. Patients who reside in these areas, need to travel to one of the 6 provinces where T2* is available. The traveling, many times is financially difficult and inconvenient. due to long distances.

Multidisciplinary Care
- Not available in all provinces. Patient federation is making efforts to promote multidisciplinary care through educational programs and the creation of a directory of doctors.
- The cost of treatment is mainly covered by the government.

Pending issues and Needs:
- Need for awareness and education on thalassaemia among the Palestinian refugees living in Turkey.
- Need for consistency from doctors across the country in applying the international clinical guidelines for the management of TDT.
- Need for equal access to new treatments and medications across the country.
- Raise awareness among patients regarding the importance of psychological support.
- Need of multidisciplinary care across the country.
- Difficulty in advocating to policy makers.