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BOOK OF ABSTRACTS

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Disclaimer Statement

The abstracts contained herein have been reviewed by the Abstract Review 1st Pan-American Conference on Thalassaemia and Other Haemoglobin Disorders. The Committee members have assessed each abstract in relation to its scientific value, sound methodology, results and conclusions. The spelling, grammar and use of English language have not been reviewed.

CONTENTS

ORAL PRESENTATION ABSTRACTS[1]

Title: Real-world safety profile of twice-daily deferiprone for iron overload in us patients with thalassemia, sickle cell disease or other anemias

Abstract Category: Iron overload and management

Authors: Thomas Coates[1], Sujit Sheth[2], Ashutosh Lal[3], Abena Appiah-Kubi[4,5], Scott A. Peslak[6], Noemi Toiber Temin[7]

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 17.00 – 17.15

Abstract

Background: Iron overload is a significant cause of transfusion-related morbidity and mortality. Deferiprone (DFP) is an oral iron chelator with >28 years of safety and efficacy data. Initially approved for administration three times a day (TID), a twice-a-day (BID) formulation was developed to improve patient adherence. The United States (US) Food and Drug Administration (FDA) approved DFP BID for transfusional iron overload in patients with thalassemia in 2020, and DFP TID and BID for sickle cell disease (SCD) or other anemias in 2021. To evaluate the real-world safety profile of DFP, the Ferriprox® Total Care Registry was established in the US. Here, we assess the safety of DFP BID in patients with thalassemia, SCD, or other anemias in real-world clinical practice.

Method: Data were obtained from US patients receiving DFP BID between July 1st 2020–August 31st 2023. DFP exposure: time active in the registry. Frequency of adverse events (AEs) and serious AEs (SAEs) by Medical Dictionary for Regulatory Activities (MedDRA) Preferred Term was assessed.

Results: A total of 425 patients were referred to the Ferriprox® Total Care Registry. Primary diagnoses included "thalassemia" (n=133, 31.3%), "SCD" (n=197, 46.4%), "other anemias" (n=30, 7.1%), and "other indications" (n=65, 15.3%). Overall, 364 patients (85.6%) were ≥18 years of age. An estimated total exposure of 839 patient-years was observed in 412 patients who received ≥1 month DFP BID treatment. In total, 1250 AEs and 437 SAEs were reported. All reports of agranulocytosis (2 events) and neutropenia (7 events) recovered. None of the 33 fatal outcomes reported were assessed as possibly or probably related to DFP treatment.

Conclusion: DFP BID is well-tolerated in real-world clinical practice in patients with thalassemia, SCD, or other anemias, with no new safety concerns identified compared to DFP TID. Limitations associated with real-world data collection must be acknowledged.

Title: Exposure-response of daily versus twice daily deferasirox dosing in patients with transfusion dependent thalassemia

Abstract Category: Iron overload and management

Authors: Johnny Li[1], Janel Long-Boyle[1,2], Felicity Usac[2], Anne Rishon[2], Ashutosh Lal[2], and Beth Apsel Winger[1,2]

[1] Arranged in order of appearance in Conference Programme

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 17.15 – 17.30

Abstract

Background: Deferasirox is routinely used in transfusion-dependent thalassemia (TDT) to treat iron overload. However, suboptimal response and adverse reactions are common. There is high inter-individual variability in plasma concentrations of deferasirox and low exposure is associated with inadequate chelation. Although once daily dosing is standard for deferasirox, patients may transition to splitting the daily dose into twice daily dosing in hopes of improving efficacy and mitigating toxicity. However, the impact of daily versus twice daily dosing on plasma drug levels or outcomes is unknown.

Method: This was a single center observational pharmacokinetic (PK) study. Blood samples were collected prior to four scheduled transfusion visits. Samples were analyzed for steady state plasma deferasirox concentrations by a validated high performance liquid chromatography assay. Laboratory studies, deferasirox dosing information, and time of last deferasirox administration were collected at each study visit. Deferasirox plasma concentrations were dose-normalized and summarized for patients who had daily versus twice daily dosing.

Results: Fifty patients with TDT enrolled in the study. Median age was 31 years old and there were 13 (26%) pediatric patients (median 10 years old, range 6-17) and 37 (74%) adult patients (median 36 years old, range 21-63). Diagnoses included -thalassemia major (n=33), -thalassemia intermedia (n=1), HbE -thalassemia (n=14), and HbH Constant Spring (n=2). Initial analysis of 181 blood samples showed patients on once daily dosing (n=27) had significantly lower dose-normalized median plasma deferasirox concentrations (2.0 g/ml, range 0.02-8.6 g/ml) than those on twice daily dosing (n=22, 3.3 g/ml, range 0.01-17 g/ml, Students t-test p <0.001, CI 0.93 - 1.8).

Conclusion: Patients on once daily dosing of deferasirox had 40% lower drug exposure at steady state compared to patients on twice daily dosing. Further analysis is ongoing to determine the impact of these exposure differences on efficacy and toxicity outcomes.

Title: Weaker Bones Relative to Body Size in Patients with Thalassemia Compared to Healthy Controls

Abstract Category: Bone Health

Authors: Ellen B. Fung, PhD RD CCD, Vanessa Yingling, PhD

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 17.30 – 17.45

Abstract

Background: Up to 60% of adults with thalassemia have low bone mass and increased fracture risk that is independent of short stature. There is a paucity of data focused on how bone adapts to body size, or bone size (robusticity). Our objective was to determine if patients with Thal have weaker bones relative to body size compared to healthy controls.

Method: Bone strength, architecture and density were assessed using peripheral quantitative computed tomography in the non-dominant leg of 45 transfusion dependent Thal (TDT: 20 ± 9 yrs, 22 Male) and 34 controls (18 ± 6 yrs, 15 Male). Bone strength and robustness were plotted against relative body size (body weight * tibial length) for both trabecular and cortical regions of the tibia. Based on the regression residuals, functionality was determined, and two groups were formed, those considered to have Strong Bones for body size (SB: positive quadrant) and those with Weaker Bones for body size (WB: negative quadrant). Student's t-test determined differences between groups.

Results: There were no differences in body weight, tibial length or cortical density between SB and WB groups. However, tibias that were robust relative to body size (SB) were 31% stronger in the cortical bone and 34% stronger in the trabecular bone compared to WB group. Cortical area was 19% greater, total area 21% greater and bone diameter 45% greater than the WB group (all p<0.01). Overall, 56% of controls were categorized in the SB group compared to only 27% of TDT (p=0.012).

Conclusion: Despite the similarity in body weight between groups, 73% of Thal patients had under-adapted bone strength for body size. Exercise, which has the benefit of expanding not only total bone area but improving bone density, should be considered in TDT to improve overall bone strength relative to size and resistance to fracture.

Title: Activism-based commentary on communication breakdown regarding transfusion policy at MUHC

Abstract Category: Psycho-social Issues

Authors: Scala, Julia[1], Pavate, Veeresh[2]

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 17.45 – 18.00

Abstract

Background: In Spring 2025, the MUHC abruptly revoked its washed red blood cell policy for thalassaemia patients without prior notice or explanation. This breach of trust ignited fears of allergic and immunogenic reactions, undermined psychological safety in long-term care, and galvanized the "Fighting for Blood" advocacy movement.

Method: This qualitative study synthesizes three complementary approaches—critical policy analysis of MUHC

transfusion guidelines and public communications; narrative review of patient-authored open letters, petitions, and media coverage; and reflexive autoethnography drawn from the author's embedded experience within Montreal's thalassaemia advocacy networks.

Results: Data from existing texts and patient testimonials, analyzed through a psycho social lens, showed that indirect notification of the policy reversal sparked intense anxiety, a sense of betrayal, and even consideration of out of province care. In turn, the "Fighting for Blood" movement emerged patients published letters, issued press releases to compel MUHC leadership to respond.

Conclusion: The "Fighting for Blood" movement shows that unannounced policy changes shatter trust and leave patients feeling vulnerable. Transfusion services must provide advance notice (e.g., a month) and clear, patient-centered explanations to safeguard both health and trust.

Title: Decreased lean mass is associated with reduced muscle function in patients with Thalassemia

Abstract Category: Lifestyle Issues

Authors: Ellen B. Fung, PhD RD, Juliet Hagar, Emily Van Horn, MS, Raquel Manzo, Mohit Prasad, Viviana Preciado, Gabriel Querubin, Elena Suarez, Jose Zaragoza, Ashutosh Lal, MD, Vanessa Yingling, PhD

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 18.00 -18.15

Abstract

Background: Patients with thalassemia (Thal) have been shown to have abnormally high body fat for body weight, correlated with reduced bone mineral density (BMD). However, the relationship of body composition to muscle function and exercise capacity has not been explored.

Method: Dual Energy X-ray Absorptiometry was used to determine BMD, lean and fat mass. Upper and lower body muscle function and power were assessed using published methods for bilateral maximal hand grip strength (HGS) and vertical jump (VJ), while the 6-minute walk test (6MWT) was used as a functional marker of endurance. Data were compared to published norms for equally aged individuals without Thal.

Results: 18 Thal subjects [13 Male, 27.5±8.6 years (Mean±SD), 89% transfused] were assessed prior to an exercise intervention. Elevated percent body fat was observed in 78% of the subjects, although BMI was high (>25 Kg/m2) in only 28%. Five subjects (28%) also had elevated visceral adipose tissue (VAT >100). Total body lean mass (Z-score: -1.4 ± 0.7) and spine BMD (Z-score: -2.2±1.1) were reduced relative to age and gender reference data. HGS in females and males was 51% and 63% of expected, respectively. VJ power in females and males was 63% and 72% of expected, while 6MWT distance was 81% of expected for each gender. Total body lean mass was highly correlated with vertical jump power ($r=0.9$, $p<0.001$), grip strength ($r=0.72$, $p=0.002$), and spine BMD ($r=0.54$, $p=0.037$), while 6MWT was inversely related to age ($r=-0.85$, $p=0.047$) but not to lean mass.

Conclusion: These data illustrate that young adults with Thal have increased fat mass with normal BMI and reduced

lean mass for body weight. We observed a strong correlation between reduced lean mass and decreased muscle function. Exercise interventions to increase lean mass and muscle function may also improve bone health, a persistent problem in thalassemia.

Title: The Greek-American diaspora: a patient population in need of 2 increased β-thalassemia carrier status education and screening

Abstract Category: Epidemiology

Authors: Rhea Sullivan[1], Bridget A. Rafferty[1,2], Eugene J. Lengerich[3], Daniel J. McKeone[1], and Zissis Chroneos[1,4]

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 18.15 – 18.30

Abstract

Background: β-thalassemia is a heritable hemoglobinopathy with high prevalence in people of Mediterranean descent. There is lack of quantifiable data to assess healthcare and public health awareness and burden of β-thalassemia critical to prevention, control, management, and standard of care of patients with β-thalassemia-related disorders in the United States despite decades of immigration from affected countries. In this study, we report the development of an educational survey in collaboration with a local Greek American community in Central Pennsylvania. We hypothesized that older first-generation participants (45 years and older) who immigrated from countries with established screening programs of β-thalassemia would have better knowledge and awareness of the disease than those born in the United States.

Method: Our survey, which was administered electronically via REDCap in both Greek and English, was designed to assess knowledge and perceptions of the disease, as well as disease symptoms prevalence, inheritance, pathogenesis, treatment, diagnosis, and known carrier status. Fisher's exact and Chi-square tests were used to test for categorical associations. All analyses were performed using the R statistical software packages. All tests were two-sided using a statistical significance level 0.05.

Results: Our results show that although age was associated with the ability to identify terms related to β-thalassemia, it was not associated with increased understanding of the disease. Unfamiliarity with the disease in younger participants (13 – 44 years) was prominent; over 60% of young participants did not recognize the term "β-thalassemia" or related terms. Furthermore, 37% of young adults had not heard of the disease through any medical professional, formal education, or word of mouth.

Conclusion: Our pilot study indicates that there is high interest to increase awareness and gain deeper understanding of β-thalassemia. Our findings support a wider effort to improve quality and delivery of care for β-thalassemia in at-risk communities in the United States.

Title: Improvement of Symptomatic Anemia with Luspatercept in a patient having Heterozygous STEPA3/TSAP6 mutation and Beta-Thalassemia trait

Abstract Category: New Advances in Treatment

Authors: Akhila Arcot Vadivelan[1], Thomas Coates[2,3]

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 18.30 – 18.45

Abstract

Background: Classically "Dominant Thalassemia (DT)" has been thought of as isolated beta thalassemia trait (BTT), with a Hb < 10, a big spleen with the implication that BTT mutation is severe. We now know DT is usually a multiple heterozygous state, most commonly due to increased alpha globin genes. STEAP3 IS A FERRIREDUCTASE THAT REDUCES FE3+ TO FE2+ IN THE ENDOSOMES, WHICH IS IMPORTANT FOR MOBILIZING IRON TO THE ERYTHROID PRECURSORS. STEAP3 mutation leads to microcytic, hypochromic anemia in patients like that of BTT.

Method: A 27-year-old female of Northern European descent with a lifetime history of symptomatic anemia (Hb is ~ 6.9 g/dL). Mild splenomegaly noted on exam. The father and patient's 2 sisters have dominant thalassemia. Labs showed, Hb 5.9 g/dL, RBC 2.65 x 106 /microliter, MCV 79.2, MCH 22.3 pg, MCHC 28.1 %, serum iron 356 mcg/dL, transferrin saturation 70%, TIBC 509 mcg/dL and ferritin 220 ng/mL. Peripheral blood smear showed RBC cytoskeleton abnormalities with pronounced basophilic stippling and abnormal tear drop shaped red cells. MRI showed mild hepatic siderosis with a liver iron concentration between 2.1- 5.1 mg/g, no pancreatic or cardiac iron. The red cell enzymes genetic panel was normal. The severity of anemia and significant phenotypic variability among family members raised the suspicion for a co-existing pathology. Focused exome sequencing revealed a heterozygous STEAP3 mutation (Chr2:119,247,805, NM_182915.3: c.649C>T, p.Arg217Cys) besides a heterozygous mutation on HBB(Chr11:5,227,020, NM_000518.5: c.2T>G,p.Met1?).

The patient was started on luspatercept every 3 weeks as part of a Phase 2 clinical trial of luspatercept in non-transfusion dependent thalassemia (NCT03342404).

Results: Significant rise in Hb in the range of 8.5- 10.5 g/dL and improvement in symptoms was noted.

Conclusion: Co-existence of heterozygous HBB and STEAP3 gene mutations can cause clinically significant microcytic, hypochromic anemia as in DT. Luspatercept can be used to treat the same.

Title: Hematopoietic Stem Cell Tranplantation (HCT) for symptomatic non-transfusion dependant Congenital Dyserythropoietic Anemia II (CDAII)

Abstract Category: Haematopoietic Stem Cell Transplantation (HSCT)

Authors: Mamatha Mandava[1], Elizabeth Yang[3], Sameeya Winston[1], Kelly Lyons[1], Robert Nickel[1,2], Anant Vatsayan[1]

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Presentation on 11 July 2025 in Hall 'Regency A/B', at 18.45 – 19.00

Abstract

Background: Congenital dyserythropoietic anemia type II (CDA II) is a rare genetic disorder featuring ineffective erythropoiesis, hemolytic anemia, and secondary iron overload with significant clinical variability. While supportive care manages most cases, hematopoietic stem cell transplantation (HCT) provides the only curative treatment, eliminating anemia and iron-related complications. The optimal conditioning approach remains a subject of investigation.

Method: Case report analysis.

Results: A 12-year-old Hispanic patient with CDA II (compound heterozygous SEC23B variants: Ex2 c.40C>T(R14W) and Ex9 c649C>T(R217X)) was diagnosed at age 4. His condition featured normocytic anemia with baseline hemoglobin of 7-8 g/dL, reticulocytopenia, illness-related nadirs of 3 g/dL requiring transfusion, and declining growth from ~30th to <3rd percentile.

The patient underwent 10/10 HLA-matched sibling donor (MSD) HCT with reduced intensity conditioning (RIC) combining anti-thymocyte globulin, fludarabine, cyclophosphamide, and 2Gy total body irradiation. Post-transplant monitoring revealed declining donor myeloid chimerism (86% on day 30 to 27% by day 270), necessitating a second transplant from the same donor.

The second transplant employed modified RIC with rATG, fludarabine, increased cyclophosphamide, and an alpha-beta T-cell depleted graft. This approach achieved full donor myeloid chimerism (>95%) by day 60, maintained through 18-month follow-up. The patient remains asymptomatic with improved growth to the 10th percentile.

Conclusion: Published literature shows MSD HCT for CDA II patients yields better survival rates (85%) than unrelated donor transplants (60%). Our case underscores potential graft failure with RIC protocols and emphasizes the importance of optimizing conditioning regimens. Although supportive care effectively manages most patients, this case demonstrates that HCT should be considered beyond traditional non-malignant hematologic disease indications, particularly when disease progression occurs, or conventional therapies cannot maintain normal growth and development.

POSTER ABSTRACTS[2]

Title: Implementation of red cell genotyping in children with beta thalassaemia in Turkey

Abstract Category: Blood Transfusion

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Abstract

Background: Blood group serology may be inadequate for patients receiving transfusions. Red cell genotyping (RCG) in Turkey 2013 demonstrated 51% of transfused patients with phenotype/genotype discrepancy. Many children have arrived from Syria since 2011. However, RCG has not become a routine diagnostic method in Turkey. We aim to compare RCG with serological methods and evaluate possible clinical consequences in children with β-thalassaemia major.

Method: All patients with β-thalassaemia major followed at the main outpatient clinic in Ankara between January and September 2024 were studied. Transfusion data and EDTA-anticoagulated peripheral whole blood were collected. HEA BeadChip DNA array was used.

Results: We collated the demographics of 51 consecutive patients. No parameter differed between the 2 nationalities. Patient age was not associated with liver function tests, ferritin, echocardiography or MRI findings.

We analysed 6 antigens of the Rh and Kell systems and found 25 patients (49%) with at least 1 phenotype/genotype discrepancy. Mixed-field reaction in serological typing occurred in 6 and alloantibodies in 3 patients (5.8%). Transfusions, weight or height percentiles, ferritin, liver function tests, echocardiography and MRI did not differ between patients with or without phenotype/genotype discrepancy.

Cardiac iron overload occurred in 3 Turkish and 2 Syrian patients and was not associated with phenotype/genotype discrepancy(p=0.63). However, hepatic iron overload, assessed by R2* MRI, occurred in 21 patients with a phenotype/genotype discrepancy and 15 patients without a discrepancy(84% vs. 58%, p=0.048).

Conclusion: The phenotype/genotype discrepancy rate was 49%, unchanged since a similar study in 2013. Because RCG has not been adopted, no improvement of care in this regard has occurred for thalassaemia patients in Turkey more than 10 years. Despite our extended antigen matching policy, the alloimmunisation rate was 5.8%. Further studies are needed to determine whether the transfusion of genotypically matched red cells could lead to a decrease in transfusion frequency, help prevent iron overload and organ damage.

Title: ~~Case Report: Unexpected complication: femoral head~~ necrosis in a pediatric beta-thalassemia major patient

Abstract Category: Bone Health

Authors: Alessandra Cristina Oliveira Borges[1], Paula Cella Giacometto[1], Bruna Oliveira Borges[2]

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Abstract

Background: Beta-thalassemia major is a hereditary hematologic disorder characterized by ineffective erythropoiesis and chronic transfusion dependency. As survival rates improve, long-term complications—including musculoskeletal disorders such as avascular necrosis (AVN)—are increasingly recognized. Although AVN is more commonly associated with sickle cell disease, its occurrence in thalassemia patients is underreported and may be linked to iron overload, chronic anemia, and corticosteroid use.

Method: We describe the case of a 13-year-old male diagnosed with beta-thalassemia major since infancy and receiving regular red blood cell transfusions every three weeks. The patient presented with progressive limping, in the absence of pain, fever, or trauma. A physical exam revealed reduced range of motion in the right hip. Magnetic resonance imaging (MRI) was performed to evaluate joint integrity.

Results: MRI findings indicated right hip synovitis and degeneration of the femoral head, suggestive of avascular necrosis. Despite a surgical recommendation for joint decompression or arthroplasty, the multidisciplinary team opted for conservative treatment, including intensified physiotherapy and an increased transfusion frequency to maintain hemoglobin above 10 g/dL. Iron chelation therapy had previously been discontinued. Follow-up MRI after 18 months showed complete resolution of necrotic changes, and the patient reported full functional recovery without the need for surgery.

Conclusion: Although rare, AVN can occur in patients with beta-thalassemia major and may present without pain. Early recognition and conservative treatment may prevent irreversible joint damage. This case highlights the importance of maintaining adequate hemoglobin levels and monitoring for orthopedic complications in transfusion-dependent thalassemia patients.

Keywords: Thalassemia major, avascular necrosis, pediatric, transfusion.

Title: Evaluation of a microchip electrophoresis platform for hemoglobinopathy screening in a premarital setting in Turkey: a comparative study with HPLC

Abstract Category: Diagnosis

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Abstract

Background: Hemoglobinopathies are the most common autosomal recessive monogenic disorders globally, encompassing thalassemia syndromes and structural hemoglobin abnormal variants. Current diagnostic approaches are effective but costly and require skilled personnel and sophisticated infrastructure, limiting their utility in low-resource settings. Gazelle®, a paper-based microchip electrophoresis platform, offers a portable, affordable, and rapid point-of-care (POC) alternative for identifying common hemoglobin variants.

Method: This study evaluated the performance of the Gazelle® microchip electrophoresis platform compared to HPLC for premarital screening of hemoglobinopathies and confirmed with beta-globin gene sequencing. Couples presenting to the Hemoglobinopathy Diagnosis Thalassemia Center of Mediterranean Blood Diseases Foundation (MBDF) for premarital thalassemia screening were enrolled. The samples were tested on Gazelle® and HPLC at the Antalya Genetic Diseases Assessment Center. Beta-globin gene sequencing was conducted on all samples with suspected variants.

Results: A total of 616 individuals participated in the study. A total of sixteen traits (3.2%) were identified as 14 beta thalassemia trait, one Hb S, and one Hb D trait were identified among the healthy group (n=516).

As a control group, 100 carrier individuals (81 beta-thalassemia and 19 sickle cell) were included. Analyses for comparing the devices were performed using IBM SPSS Version 29 software. The Kappa Analysis method was used to measure the diagnostic sufficiency of the two devices. Both devices were found to be 100% consistent in diagnosis.

Conclusion: Gazelle® demonstrated diagnostic equivalence to HPLC in identifying and quantifying hemoglobin variants, including Hb A, Hb S, Hb F, and Hb A2, enabling accurate detection of β-thalassemia carriers. Its rapid time-to-result (<8 minutes), digital connectivity, portability, and affordability positions it as a valuable tool for expanding access to hemoglobinopathy screening.

Title: Case Report: Pregnancy in a patient with beta-thalassemia major

Abstract Category: Fertility & Pregnancy

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Abstract

Background: Beta-thalassemia major is a transfusion-dependent hemoglobinopathy associated with multiple complications, including iron overload. Pregnancy in these patients is uncommon and requires careful planning and management due to transfusion burden and chelation therapy interruption.

Method: We report the case of a 29-year-old white female diagnosed with beta-thalassemia major at 8 days of life, who initiated regular red blood cell transfusions every 21 days. She underwent cholecystectomy in 2009 and splenectomy in 2015. In 2020, transfusion reactions led to the use of washed red blood cells. Her iron burden was managed with various chelation regimens over the years. In 2022, while on deferiprone, her serum ferritin was 403 ng/mL. Chelation was

discontinued in December 2022 for conception planning, and pregnancy was confirmed in April 2023, approximately 90 days later.

Management and Outcome: Prenatal care focused on maintaining hemoglobin levels >10 g/dL, with transfusion frequency increased to every 15 days. At six weeks of gestation, the patient experienced mild vaginal bleeding, managed successfully with progesterone. No chelation therapy was used during pregnancy. Delivery occurred at 38 weeks via elective cesarean section. Moderate intraoperative vaginal bleeding was controlled without transfusion. The newborn weighed 3,170 g and had Apgar scores of 9 and 9 at one and five minutes, respectively.

Conclusion: This case demonstrates that a well-controlled iron burden prior to conception, along with close transfusion monitoring, can result in a successful pregnancy and delivery in a patient with beta-thalassemia major. It reinforces the importance of individualized care and timing of chelation therapy in women of reproductive age with transfusion-dependent anemias.

Keywords: Beta-thalassemia major; Pregnancy; Iron chelation; Transfusion.

Title: Improving apheresis collection of autologous hematopoietic progenitor cells for beta thalassemia gene therapy candidates

Abstract Category: Gene Regulation & Therapy

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Abstract

Background: Gene modification is a promising therapy for patients with transfusion dependent Beta Thalassemia. To successfully meet the dose needed for engraftment, sufficient autologous hematopoietic progenitor cells (HPC (A)) must be collected. However, there are challenges and barriers to ensure adequate collection. We review collection data and discuss improvements our Apheresis Collection Facility has incorporated to improve HPC(A) apheresis collections over time.

Method: We retrospectively reviewed data for 10 HPC(A) collections for four pediatric Beta Thalassemia patients occurring between 2023-2025. All collections were completed using the continuous mononuclear cell collection (CMNC) procedure on the Spectra Optia apheresis system at a single facility. Anticoagulation settings, collection efficiency, collection preference, number of cells collected, and product hematocrit were evaluated in comparison to success or failure of meeting gene manufacturing goals.

Results: We determined that lowering the inlet: AC ratio and consistently targeting darker collection color with the collection preference (CP) tool improved collection efficiency (CE). Patients anticoagulated with an AC: ratio of 8:1 had increased CE and a greater likelihood of meeting the collection goal compared to those anticoagulated at 10:1 or higher. Additionally, targeting a darker color using the CP tool best captured the cells of the buffy coat and increased the

hematocrit (Hct) of our sample, which also increased our CE. In response, our facility has standardized AC: inlet ratio settings and developed job aids, optimized lighting sources, and incorporated second opinion checks to consistently determine appropriate collection preference color.

Conclusion: This study highlights improvements to our apheresis collection protocol that have improved HPC(A) collections for our pediatric Beta Thalassemia patients. Incorporating job aids and accounting for differences in color perception has enabled us to more consistently determine collection preference. These practices show promise as we continue to improve collection efficiency in our apheresis collections over time.

Title: Outcomes of allogeneic transplantation for transfusion-dependent anaemias in Hong Kong

Abstract Category: Haematopoietic Stem Cell Transplantation (HSCT)

Authors: CHAN Wilson Yau-Ki[1], LEE Pamela Pui-Wah[1,2], LEUNG Wing Hang[2,3], CHEUK Daniel Ka-Leung[1,2]

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Abstract

Background: Allogeneic haematopoietic stem cell transplantation (HSCT) had been the standard curative treatment for transfusion-dependent thalassemias (TDT) and some other transfusion-dependent anaemias (TDAs).

Method: All patients aged 18 years or below with TDAs including TDTs and other transfusion-dependent haemoglobinopathies or enzymopathies but excluding aplastic anaemias and other inherited marrow failure syndromes, who underwent allogeneic HSCT in Hong Kong from 1 January 1992 till 31 March 2019 in QMH or from 1 April 2019 to 31 July 2024 in HKCH were included.

Results: Total 107 allogeneic HSCTs were performed in 95 patients including 42 boys and 53 girls . All patients were Chinese. 78 patients (82.1%) suffered from beta-thalassemia major, while the remaining 17 patients had other TDAs. Median age at first HSCT was 8.4 years (range 0.8-17.95 years) and two-thirds had HSCT done before the age of 9 years. Same transplant protocol on conditioning and supportive medications was generally employed for all patients with same donor type and stem cell source. Neutrophil (>0.5x10^9/L) and platelet (>20x10^9/L) engrafted at a median of 15 (9-26) and 19 (9-158) days respectively. Total 12 patients underwent 2 HSCTs due to graft failure/rejection, with graft failure/rejection rate of 11.2%. Total 21 patients (22%) and 16 patients (16.8%) had grade III-IV acute and chronic graft-versus-host disease (GVHD) respectively. Hepatic sinusoidal obstruction syndrome (SOS) occurred in 12 patients (12.6%). Transplant-related mortality was 8.4% with 9 transplant-related deaths due to severe GVHD (n = 3), infective complications (n = 3), severe hepatic SOS (n = 1) and others (idiopathic interstitial pneumonitis = 1, intracerebral haemorrhage = 1). Overall survival and thalassemia-free survival were 87.4% and 83.2% respectively.

Conclusion: In summary, local paediatric HSCT center demonstrated comparable outcome in HSCT for patients with TDAs. Regional, national or international collaboration is advocated to further improve HSCT outcomes.

Title: Hematopoietic Stem Cell Tranplantation (HSCT) for symptomatic non-transfusion dependant thalassemia

Abstract Category: Haematopoietic Stem Cell Transplantation (HSCT)

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Abstract

Background: Hematopoietic stem cell transplantation (HSCT) and gene therapy are recognized curative treatments for thalassemia syndromes, including hemoglobin E/β-thalassemia. However, due to significant associated risks and the generally milder course of non-transfusion dependent thalassemia (NTDT) compared to transfusion-dependent thalassemia (TDT), HSCT is rarely considered for NTDT patients.

Method: Case report of 11-year-old Thai child with non-transfused Hemoglobin E/β0 thalassemia.

Results: The patient initially maintained stable hemoglobin levels of 9 g/dL, which decreased to ~8 g/dL by age 3. Hydroxyurea (HU) therapy was initiated to prevent extramedullary hematopoiesis and further hemoglobin decline. Despite initial improvement to 10 g/dL with HU, her hemoglobin subsequently decreased to 8 g/dL, accompanied by growth challenges (height and weight below the 2nd percentile). A temporary discontinuation of HU resulted in hemoglobin dropping to 6.6 g/dL, requiring resumption of therapy, but she progressively lost response with hemoglobin declining to ~7 g/dL.

Given the availability of an unaffected HLA-identical sibling donor, the patient underwent 8/8 HLA-matched sibling donor transplantation with a reduced intensity conditioning regimen (HU, Alemtuzumab, Fludarabine, Melphalan, Thiotepa). Post-transplant complications included acute gastrointestinal GVHD and transplant-associated thrombotic microangiopathy, both successfully treated.

Following engraftment, the patient achieved and maintained 100% donor chimerism through two years post-HSCT. Her hemoglobin levels stabilized between 12-14 g/dL, with normal hemoglobin electrophoresis patterns at day 60 and the last follow-up of 2 years (Hgb A: 97.1%; Hgb F: 0.3%; Hgb A2: 2.6%). The patient remains asymptomatic and is gaining growth curve percentiles.

Conclusion: While HSCT is typically not recommended for NTDT hemoglobin E thalassemia patients due to procedure risks outweighing benefits, this case demonstrates that transplantation can be appropriate when conventional treatments prove inadequate. Though most NTDT patients can be effectively managed with supportive care, HSCT becomes viable when the disease progresses in severity or when standard therapies fail to support normal growth and development.

Title: Effective iron overload assessment is crucial for transfusion-dependent thalassemia patients from the age of two

Abstract Category: Iron overload and management

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Abstract

Background: Our objective is to demonstrate that very young children with thalassemia dependent-transfusion (TDT) can present significant liver iron concentration (LIC) and cardiac iron overload (IOL).

Method: Retrospective study (2005-2022) analyzed the first MRI data of patients aged ≤11 years (y) with TDT at two Brazilian centers. T2* images were acquired using a 1.5T scanner with a rapid single breath-holding sequence comprising multiple echo times for heart/liver imaging. The full examination, including T2* mapping and left ventricular function assessment, was completed in under 10minutes. Statistical analyses included median values, unpaired T-tests, and Fisher's exact test.

Results: Data was collected from 23 patients (≤3y: 2; 4–5y: 4; 6–7y: 8; 8–11y: 9). Iron chelation therapy (ICT) began at a median of 29 months with deferoxamine, deferasirox, or deferiprone chosen based on age and drug availability. Lack of an infusion pump in Brazil limited the access to deferoxamine. Median age at first MRI was 7y, mean LIC (mg/g dry weight) was highest in children ≤3y (12,5), without significant differences between groups (4–5y: 9.8; 6–7y: 9.4; 8–11y: 10.2). Liver IOL was observed as 2y and 10months. Median cardiac IOL was normal but declined with age. A 7-y-old patient had severe cardiac IOL. Adherence to ICT was adequate in 56.5% of patients; non-adherence (medication shortages, adverse events, treatment refusal) was associated with cardiac IOL ($p<0.01$). Adjusting ICT based on MRI findings led to the reversal of IOL.

Conclusion: MRI should be considered for children over 2y, particularly for liver assessment, and especially with low ICT adherence. Poor ICT adherence directly impacts cardiac IOL. Given the technical MRI challenges in children over 2y, during appointments an educational plan about the MRI process (machine, sounds, scanning environment), and the need to remain still for at least 10 minutes should be implemented.

Title: Importance of recognizing chelation toxicity in thalassemia patients

Abstract Category: Iron overload and management

Authors: Jeanette Rosenberg-Line DNP and Aimee Foord DO

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Abstract

Background: Transfusion dependent thalassemia major patients require iron chelation to prevent organ damage. Deferasirox, a common drug of choice for pediatric chelation, is generally well tolerated. Although rare in general, and even rarer in the pediatric population, it is important to note that deferasirox carries a black box warning regarding hepatic toxicity.

Method: Chart review of a patient presenting to ER with nonspecific GI symptoms of nausea, abdominal pain with worsening jaundice.

Results: 16-year-old with hemoglobin E/beta (null) thalassemia presented to ER with nausea and abdominal pain in between his routine transfusions. His labs were significant for a total bilirubin of 22mg/dL with elevated direct bilirubin of 14mg/dL and transaminitis with AST 282u/L and ALT 344 u/L, coagulopathy of PT 17sec, and aPTT 37sec. His home medications included folic acid, vitamin d, and deferasirox at 18mg/kg/day. Work up included negative hepatitis panel, GI pcr, serum viral studies, and autoimmune hepatitis panel. Positive findings included abdominal ultrasound concerning for cholecystitis, however HIDA scan was negative for cholecystitis with a new concern of biliary tract obstruction. MRCP done and negative for obstruction or dilatation. His chelation was discontinued, symptoms improved, and he was discharged, with continued scheduled transfusions and routine monitoring of his labs.

Conclusion: Due to ongoing hemolysis, thalassemia patients carry risks of GI complications such as cholecystitis, choledocholithiasis causing biliary obstruction, as well as common complications that can cause hyperbilirubinemia and transaminitis including viral infections. Our case highlights that chelation toxicity should also be highly considered if work-up is otherwise unrevealing. This patient with no other known liver injury showcases the rare but possible risk of hepatic impairment that can occur in pediatric patients with deferasirox. Recognition of this toxicity is vital to stop further liver damage and the possible progression to liver failure.

Title: REDSCORE

Abstract Category: Miscellaneous

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Abstract

Objective: REDSCORE is an additional value that represents the percentage expression of information derived from a mathematical calculation. This calculation aims to illustrate the combined behaviour of three quantifiable variables in the red blood cell formula: RBC count, haemoglobin (HGB), and mean corpuscular volume (MCV). Its primary goal is to enable users to quickly infer a diagnostic impression based on these data at any given moment, facilitating clinical follow-up regarding red blood cell formula changes. Its greatest utility lies in monitoring patients with haemoglobin levels above 8.5 g/dL, as individuals with lower values require a more urgent clinical approach and personalized attention in a shorter time frame. REDSCORE helps detect trends and assess the relevance of variations in one or more parameters for individuals who are not haematology specialists, including patients. For physicians, it provides a different perspective and an additional data point to aid in diagnostic interpretation of red blood cell formulas and stratifies risk levels.

Methodology: A retrospective analysis of patient data was conducted to propose a formula that evaluates discrete yet clinically significant changes over time.

Results: It works as shown below:

Table 1: Shows the REDSCORE variations in different observations for one person

Observation	1	2	3	4	5	6	7
RBC (106/µL)	4.9	5.0	5.0	5.3	3.9	4.8	4.6
HGB (g/dl)	11.6	11.8	14.5	14.7	8.5	9.7	10.1
MCV (fL)	71.7	72.4	82.3	81.3	71.0	67.0	69.4
Redscore (%)	**74.1**	**76.0**	**90.8**	**94.0**	**51.5**	**63.5**	**64.2**

Conclusion: The percentage-based REDSCORE is sensitive to subtle changes in any of the evaluated parameters, which may often be imperceptible but can have a significant impact on an individual's health status and treatment course.

Title: Healthcare Professionals in the Care of Patients with Thalassemia

Abstract Category: Miscellaneous

Authors: Janaína Rosenburg Gioseffi, MSc[1], Nina Victória Menezes Melo[1], Fernanda Cristina dos Santos Simão[1], Eduardo Fróes[1], Talita Garrido de Araújo[1], Fábio Augusto Fedozzi[1], Catherine Moura, MD, MSc[1], Sandra Loggetto, MD[2]

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Abstract

Background: Care for thalassemia within Brazil's public healthcare system (SUS) or the private sector still faces numerous challenges, even in wealthy and significant states such as Paraná.

Objective: The aim of this study was to understand the perspective of healthcare professionals at Hemepar (Paraná's Hemocenter, Curitiba/Paraná, Brazil) regarding the challenges in providing care for thalassemia, including infrastructure, access to diagnostic tests, and specialized referrals.

Methodology: Interviews were conducted with six professionals from various areas (management, clinical care, pharmacy, hematology, and social services), using a semi-structured script with open-ended questions.

Results: Hemepar currently treats around 25 patients with thalassemia. The main difficulties reported by the professionals included the lack of specialized diagnostic tests (such as T2* magnetic resonance imaging, echocardiograms, etc.), shortage of infusion pumps for desferoxamine administration, lack of human resources, and the need for staff training. Patients' access to a multidisciplinary team was found to be limited and dependent on the healthcare referral system (public or private). Moreover, referrals to specialists, which occur through primary care and state-level regulation, often receive no response. Most professionals also reported a lack of specific training for personalized care and noted that few educational materials on thalassemia are distributed.

Conclusion: The professionals' accounts reveal structural and care-related barriers along the patients' journey, significantly impacting the quality of care. The absence of continuing education and the burden of excessive workloads further hinder the implementation of comprehensive care. An ineffective referral and regulation system exacerbates the barriers to access.

Title: Psychosocial and Socioeconomic Aspects of People with Beta Thalassemia in Brazil: A National Survey

Abstract Category: Psycho-social issues

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Abstract

Background: Thalassemia impacts multiple dimensions of people's lives, from childhood to young adults in the workforce. Our objective was to identify the psychosocial and socioeconomic information of people with Beta Thalassemia in Brazil, through a survey conducted with patients from different regions of the country, seeking to understand how these aspects relate to living with the disease.

Method: This was an observational, quantitative study, consisting of a self-administered questionnaire completed by patients who participated in a focus group in Curitiba. The questionnaire was pre-structured and addressed psychosocial and socioeconomic issues.

Results: A total of 58 people with thalassemia from various regions of Brazil participated in the survey, with a notable representation from the states of São Paulo (n=20) and Rio de Janeiro (n=11), reflecting the presence of reference centers for treatment and more organized patient associations in these areas. The majority had Thalassemia Major (74%), with 14% having Thalassemia Intermedia and 12% Thalassemia Minor. Most participants were female and young adults (29% between 30 and 39 years old, male and female combined), an economically active age group. Regarding affective relationships, half of the respondents (50%) lived with a spouse or partner. Considering the impact of thalassemia on professional life, 24% of respondents had taken temporary leave from work due to the disease, revealing the influence of thalassemia on work stability and continuity. In terms of family life, most people with thalassemia do not have children (72%), which may be related to concerns about health, fertility, or socioeconomic factors. Quality of life was a concern for 93% of participants, signaling a significant impact of thalassemia on daily life, even though the experience with the disease was not predominantly negative (34% rated it as good, and 33% as normal). Only 22% received psychological support. The majority (60%) receive treatment through the Unified Health System (SUS), while 26% use private health insurance for transfusions, and 14% combine both services.

Discussion and Conclusion: The higher number of people with Thalassemia Major highlights the need for healthcare, emotional support, and public policies tailored to this more vulnerable population. As young adults, they are in active work life and require policies that ensure a balance between work and treatment. Despite the possible limitations imposed by the health condition, these individuals overcome challenges and achieve success in establishing emotional bonds. There is an unmet demand for emotional and psychological support, as few have access to this support, and many are concerned about their quality of life. The priority of treatment within the SUS system reinforces the importance of maintaining and strengthening public policies for this group of individuals.

Title: "Fighting for blood": psycho-social impact on Montreal's thalassaemia community - a qualitative overview

Abstract Category: Psycho-social issues

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Abstract

Background: Persons with Thalassaemia rely on frequent periodic lifelong blood transfusions which are the needs of people living with thalassemia to survive. For years, the McGill University Health Centre (MUHC) in Montreal provided washed red blood cells to minimize transfusion reactions. Recently, however, this policy was suddenly reversed—without warning, consultation, or clear explanation. Patients are now receiving unwashed blood, a change that has triggered profound ethical and psycho-social concerns. For a population already managing a chronic, life-threatening condition, this sudden shift in care has undermined trust in the healthcare system and significantly affected their sense of safety, stability, and well-being.

Method/Approach: This study draws upon the lived experiences of patients affected by the policy change, using patient interviews, activism, advocacy initiatives as primary sources, and qualitative methods.

Results/Findings: The abrupt discontinuation of washed red blood cells has triggered significant fear, emotional distress, and erosion of trust within the thalassaemia community. This lack of transparency has exacerbated psychological distress, leaving patients feeling abandoned, unheard, and increasingly marginalized. Repeated attempts to obtain clarification from the MUHC, Héma-Québec, and the Ministry of Health have been met with institutional silence and a deflection of accountability. This research will disclose further insights into our findings in future.

Conclusion: This situation highlights the serious psycho-social consequences of policy decisions made without patient consultation. Emotional trauma, loss of agency, and institutional betrayal threaten not only individual well-being but also the trust required for sustainable healthcare relationships. Ethical and transparent communication is essential when implementing changes that directly affect patients' lives. The Montreal thalassaemia community's experience serves as a powerful case for greater attention to the psychological and social dimensions of care — especially for populations historically marginalized in health systems.

Keywords: Thalassemia, Blood, Psychosocial, Quality of Life, Ethics, Advocacy

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Title: Focus Group with People Living with Thalassemia

Abstract Category: Quality of Life

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Abstract

Background: Thalassemia care within different healthcare

systems reveals structural and service-related challenges that directly affect the quality of care. At Hemepar, in Curitiba (Paraná), individuals living with thalassemia shared their experiences and the limitations they face in their care.

Objective: The aim of this research was to give voice to the experiences and perceptions of patients with thalassemia regarding their care journey, access to treatment, social and emotional impacts, and future expectations.

Methods: A focus group was conducted with nine patients (including minors accompanied by family members), in a roundtable format, moderated by a representative from ABRASTA (Brazilian Thalassemia Association), following a structured discussion guide.

Results: Some participants reported delays in early diagnosis due to healthcare professionals' lack of knowledge about the disease, as well as difficulties accessing exams, medication, and transportation—often requiring legal action or formal complaints to ensure their rights. Participants also described experiences of prejudice, bullying, and emotional challenges during childhood and adolescence. Feelings of low self-esteem and fear about the future were common. The word most frequently used to describe the disease was "overcoming." Many reported a lack of adequate information about thalassemia in public health services.

Discussion and Conclusion: The experiences shared reveal that the impact of thalassemia goes beyond biomedical aspects. The disease directly influences daily life, social relationships, education, and employment. Actively listening to patients highlighted the importance of supportive policies, psychological assistance, and effective communication. Despite the challenges, many remain hopeful and engaged..

Title: Post-Splenectomy Hemorrhage in Transfusion-Dependent Thalassemia: A Tale of Two Siblings

Abstract Category: Thrombosis

Authors: Henna Butt, MD, MHSc & Jennifer Webb, MD, MSCE

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Abstract

Background: Patients with transfusion-dependent thalassemia (TDT) have a higher baseline risk of thrombosis due to a prothrombotic state, with an incidence ranging from 1.6% to 15%. Splenectomy, often required to reduce transfusion needs in cases of hypersplenism or splenic sequestration, further increases thrombotic risk, warranting consideration of anticoagulation. We report two TDT siblings who experienced post-splenectomy bleeding complications.

Methods: This is a case report.

Results: A 20-year-old male with TDT, iron overload, and splenic sequestration, underwent splenectomy. He recovered well but developed a subdural hemorrhage four months later after a fall while on rivaroxaban and aspirin for thrombocytosis. Anticoagulation with rivaroxaban had been extended beyond four weeks due to a language barrier. Coagulation studies showed elevated PT, INR, PTT, low Factor II, and partial correction on mixing studies—suggestive of acquired coagulopathy.

His 21-year-old sister with TDT, iron overload, and hypersplenism underwent open splenectomy and liver

biopsy two months later. Preoperative bruising led to a workup revealing prolonged PTT, transiently low factors (II, IV, VIII, X), and thrombocytopenia due to hypersplenism. Platelet aggregation studies showed abnormal clot formation, decreased clot strength by thromboelastography. Platelet mapping showed significant AA inhibition and normal range ADP inhibition Liver disease from iron overload and hepatitis C was suspected. She received both FFP and platelets prior to surgery. On postoperative day 1, she experienced massive hemorrhage (Hgb dropped to 4.9 g/dL). She required reoperation for 3L hemoperitoneum; bleeding source was not identified. A personalized gene panel for bleeding and platelet disorders via GeneDx including 122 genes was negative. Liver biopsy showed LIC of 28,027 mcg/g and stage 2 fibrosis.

Conclusion: These cases highlight the complexity of managing TDT patients post-splenectomy. Individualized anticoagulation plans, thorough preoperative assessment, and monitoring for coagulopathy—particularly in the context of liver dysfunction—are critical to reduce bleeding risk.

Title: Territory-wide retrospective review on outcomes of allogeneic transplantation for haemoglobin Bart's hydrops fetalis syndrome in Hong Kong

Abstract Category: α-thalassaemia syndromes

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Abstract

Background: Haemoglobin Bart's hydrops fetalis syndrome (BHFS) was once considered a fatal condition universally. Availability of antenatal screening and intrauterine transfusions as well as advanced neonatal intensive care enable long-term survival of fetuses suffering from BHFS even beyond adulthood. Hong Kong being a prevalent region for thalassaemia has 14 long-term BFHS survivors till date, which contributed to around one-fifth of globally reported cases in the literature.

Methods: All BHFS patients who underwent allogeneic HSCT in Hong Kong from 1 January 1996 till 31 December 2024 were included. Basic demographic data, perinatal history, transplant details, long-term outcomes, and morbidities were reviewed.

Results: Total 8 allogeneic HSCTs (7 postnatal HSCTs and 1 intrauterine HSCT) were performed in 7 patients (3 males and 4 females) at a median age of 22 months (range from 24 weeks of gestation to 114 months), which include one 8/8 matched-sibling bone marrow transplant, one 5/6 matched-sibling cord blood transplant with HLA-DR antigenic mismatch, two 12/12 matched-unrelated peripheral blood stem cell transplant (PBSCT), two haploidentical PBSCTs with TCRαβ/CD45RA depletion from maternal donor and one intrauterine HSCT at 24 weeks of gestation followed by haploidentical PBSCTs with TCRαβ/CD62L depletion from paternal donor. Neutrophil and platelet (>20 x 109/L) engrafted at median of 13 days (range 8-26 days) and 17 days (range 10-38 days) respectively. The transplant course was uneventful without major infective complications, hepatic sinusoidal obstruction or other organ toxicities. All remained transfusion-independent with full donor chimerism achieved with a median follow-up duration of 10 years (range 14 months to 25 years). Immune reconstitution was satisfactory. There were no features of severe grade III-IV

acute GVHD or chronic GVHD.

Conclusion: To conclude, local data demonstrated favorable outcome of allogeneic HSCT for BHFS patients. Non-directive approach in antenatal counseling and international collaboration is recommended.

Title: α3.7 deletion and discordant phenotype: relevance of -α3.7 fusion gene sequencing in the diagnosis of α thalassemia

Abstract Category: α-thalassaemia syndromes

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Abstract

Background: Alpha thalassemia is characterized by decreased synthesis of alpha-globin chains. Clinical severity depends on the number of genes affected, with phenotypes ranging from asymptomatic to lethal.

The -α3.7 deletion is the most common. While most carriers of this deletion are clinically and hematologically normal, it should be noted that the -α3.7 fusion gene can acquire mutations that reduce or eliminate its expression, leading to a more severe phenotype.

Typically, the molecular study of alpha thalassemia begins with the evaluation of the most common deletions In heterozygous (ht) for -α3.7 deletion which present a more severe clinical phenotype, sequencing of the -α3.7 fusion gene should be considered.

Methods: To describe the hematological and molecular characteristics of ht patients (pts) for the -α3.7 deletion with a discordant phenotype.

Materials and Methods: Ten pts from five unrelated families who consulted for persistent microcytosis and hypochromia were studied.

The most common large deletions (-α3.7, -α4.2, --MED, -α20.5) were assessed by GAP-PCR. Point mutations and small insertions/deletions in the -α3.7 fusion gene were evaluated by Sanger sequencing.

Results: All pts presented with microcytosis and hypochromia, hemoglobin electrophoresis and iron profile were normal in all cases.

All pts had the -α3.7 deletion in heterozygosity as the only alteration found after GAP-PCR screening. Molecular analysis of the -α3.7 fusion gene was expanded.

All 10 pts presented a point mutation within the fusion gene:

9 had the c.-2_-3del variant, and 1 pt had the c.49A>T; p.(Lys17Ter) variant.

Conclusion: It is important to carefully assess heterozygosity for the -α3.7 deletion detected by GAP-PCR or MLPA screening, particularly when the genotype does not correlate with the observed phenotype.

Recognizing and investigating more complex genotypes is essential to provide an accurate molecular diagnosis and appropriate genetic counseling aimed at preventing severe forms of alpha thalassemia.vvv

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