ARTIGOS PRODUÇÃO CIENTÍFICA 2026

1 - Genomic ancestry, F8 variants, and immune tolerance in hemophilia A patients with inhibitors: exome sequencing insights.

Sant'Anna H, Tou R, Faria-Costa L, Duarte J, Miwa B, De Souza RP, Camelo RM, Chaves DG, Lorenzato CS, Anegawa TH, De Oliveira AG, Ferreira CB, Carvalho LEM, Franco VKB, Cerqueira MH, Roberti MDRF, De Araujo Callado FMR, Etto LY, De Cerqueira MAF, De Souza Pinto IS, Garcia AA, Tan DM, Neves DCF, Dias MM, Zuccherato LW, Tarazona-Santos E, Rezende SM.

Haematologica. 2026 Feb 1;111(2):687-692.

doi: 10.3324/haematol.2024.287232. Epub 2025 May 15.

PMID: 40371890

Resumo não disponível.

2 - Adherence to Clotting Factor Prophylaxis in Adolescent and Adult Males With Haemophilia.

Beserra NM, Camelo RM, Sacramento AOR, Nascimento ES, Machado RPG, Gonçalves Machado CM, Dantas SMC, Rocha SGMD, Carvalho LEM, Lemes RPG.

Haemophilia. 2026 May-Jun;32(3):739-747.

doi: 10.1111/hae.70244. Epub 2026 Mar 23.

PMID: 41869884 Free PMC article.

Resumo:

Introduction: Clotting factor prophylaxis remains the most prescribed standard-of-care treatment for people with haemophilia (PwH). Prophylaxis prevents bleeds, joint damage, and improves quality of life (QoL). Its success depends on treatment adherence, consisting of socioeconomic, behavioural, and disease- or therapy-related factors. Aim: To identify determinants of clotting factor prophylaxis adherence among PwH followed at a Hemophilia Treatment Center (HTC) in Brazil. Methods: A cross-sectional study was conducted at a Brazilian HTC in June-October/2022. Eligible participants were PwH A or B without inhibitors and on clotting factor prophylaxis, aged ≥ 14 years. Demographic, clinical, and therapeutic data were collected from records and interviews. Health-related QoL was assessed by the SF-36 questionnaire. Adherence was assessed using the Brazilian validated Validated Hemophilia Regimen Treatment Adherence Scale-Prophylaxis (VERITAS-Pro)

questionnaire.Results: Among 78 PwH (median age 30 years), 70 (90%) had severe haemophilia, 46 (59%) reported joint impairment, and 67 (86%) infused 3 times/week. Median SF-36 score was 70.3 (interquartile range [IQR] 57.3-83.3). Median VERITAS-Pro score was 44.0 (IQR 35.0-50.0), resulting in 70 (90%) adherent PwH. SF-36 score was not correlated with VERITAS-Pro. Clotting factor prophylaxis adherence was associated with having haemophilia A ($p = 0.012$) and infusing factor >2 times weekly ($p = 0.008$), and inversely correlated with prophylaxis duration ($r = 0.317$ [weak clinical correlation]; $p = 0.005$), and directly correlated with SF-36 domain "Mental health" ($r = -0.304$ [weak clinical correlation]; $p = 0.007$).Conclusion: The main factors determining adherence to clotting factor prophylaxis were having haemophilia A, receiving factor infusions more than twice weekly, having a shorter duration of prophylaxis, and good mental health.

3 - Functional capacity in sickle cell disease: A pilot study with 1-minute sit-to-stand test.

Thomaz MB, Suassuna LF, Fabri JC, Araújo IO, Almeida JC, Rodrigues DOW.

Hematol Transfus Cell Ther. 2026 Apr-Jun;48(2):106230.

doi: 10.1016/j.htct.2025.106230. Epub 2026 Jan 10.

PMID: 41520508 Free PMC article.

Resumo:

Background: Sickle cell disease, the most prevalent monogenic recessive genetic disorder in the world, is characterized by two main pathogenic mechanisms: vaso-occlusion and hemolysis. These characteristics lead to reduced tolerance to physical exertion and, consequently, a reduced functional capacity which can be assessed using the one-minute sit-to-stand test. Complications from sickle cell disease result in poor quality of life, increased absenteeism from school and work, and impaired social interaction. Method: Between January 2023 and April 2024, a pilot cross-sectional study was conducted with sickle cell disease patients aged from 18 to 60 years. The one-minute sit-to-stand test, Borg's perceived exertion scale, and the SF-36 quality of life questionnaire were utilized. Patients were monitored during the test. The sample was dichotomized based on test performance and SF-36 scores. Furthermore, clinical and demographic variables were analyzed. Main results: Fifty-eight individuals participated in the final analysis. The mean age was 29.84 ± 11.20 years; 55.1 % were men, and 79.3 % identified themselves as Black or mixed race. The most prevalent genotype was hemoglobin SS (67.2 %), and 77.5 % were taking Hydroxyurea. The group with a better performance in the one-minute sit-to-stand test showed better quality of life as assessed using the SF-36 questionnaire. Conclusion: Functional capacity is a significant factor in the autonomy and quality of life of patients with sickle cell disease. The one-minute sit-to-stand test is a low-cost and easily applicable test,

which can contribute to the assessment of functional capacity in the routine follow-up of these patients.

4 - Dengue, chikungunya and Zika virus surveillance in blood donors in Brazil, 2019-2021.

Grebe E, Buccheri R, Montalvo L, Kashima S, Miranda C, Milani P, Stone M, Livezey K, Capuani L, Alencar CS, Amorim L, Loureiro P, Ribeiro M, da Costa AG, Mendrone A Jr, Busch MP, Sabino EC, Custer B; Recipient Epidemiology and Donor Evaluation Study-IV-Pediatric (REDS-IV-P)

Brazil.Vox Sang. 2026 Feb;121(2):196-201.

doi: 10.1111/vox.70150. Epub 2025 Dec 17.

PMID: 41407329 Free PMC article.

Resumo:

Background and objectives: Arbovirus infections are a major public health concern in Brazil and an ongoing blood safety concern. Periodic outbreaks of such infections are common in the general population. This study aimed to establish the rates of dengue, chikungunya and Zika virus (DENV, CHIKV, ZIKV) RNAemia among blood donors at six public blood centres during the 2019-2020 and 2020-2021 outbreak seasons. Materials and methods: Residual minipool samples from nucleic acid testing (NAT) screening for human immunodeficiency virus, hepatitis B virus and hepatitis C virus were further pooled to form pools of 18 donation samples and tested using a Grifols research-use-only triplex transcription-mediated amplification assay for DENV, CHIKV and ZIKV RNA to establish the rates of RNAemia and infection incidence. We used these rates and estimated the durations of RNAemia, juxtaposed with public health reporting of cases. Results: A total of 5,616 minipools representing 101,088 donations were tested. During both outbreak seasons, the highest rates of DENV RNAemia were observed in Ribeirão Preto, at 122/100,000 donations (95% confidence interval [CI]: 66-224) and 100/100,000 (95% CI: 52-189), respectively, with DENV RNAemia also detected in São Paulo, Recife and Manaus in the 2019/2020 season and in the latter two during the 2020/2021 season. CHIKV RNAemia was detected in Recife and ZIKV RNAemia in Manaus during the 2020/2021 season. The estimated numbers of DENV, CHIKV and ZIKV RNAemic components released for transfusion over the study period were 338, 22 and 6, respectively. Conclusion: Surveillance for arbovirus RNAemia in blood donors is a useful adjunct to public health surveillance, particularly when surveillance systems are under strain, and has implications for transfusion safety.

5 - Laboratory and genetic characteristic associated with gallbladder-related outcomes in sickle cell disease in Brazil: results from the REDS-III multicenter cohort study.

Belisário AR, Ozahata MC, Moura ICG, Miranda C, Carneiro-Proietti AB, Sabino EC, Ferraz A, Máximo C, Flor-Park MV, de Oliveira Werneck Rodrigues D, Mota RA, Custer B, Kelly S, Dinardo CL;

REDS-III Brazil SCD Cohort study and the TOPMed consortium.Ann Hematol. 2026; Feb 16;105(4):115.

doi: 10.1007/s00277-026-06800-z.

PMID: 41692868 Free PMC article.

Resumo:

Sickle cell disease (SCD) is a hereditary disorder characterized by HBB variants, leading to chronic hemolytic anemia and vaso-occlusion. Hepatobiliary complications, including cholelithiasis, are common but underreported. This study investigated the rates and risk factors for cholelithiasis, cholecystitis, and cholecystectomy in a large Brazilian SCD cohort. Data from 2,778 individuals across six referral centers in the REDS-III Brazilian SCD cohort were analyzed. Clinical, laboratory, and genetic data were obtained retrospectively at enrollment and prospectively during follow-up. Gallbladder-related outcomes were assessed through medical records and imaging. Whole-genome sequencing was performed via the TOPMed program. Genome-wide association analyses used logistic mixed models adjusted for age, sex, genotype, and the first 10 principal components. Cholelithiasis, cholecystitis, and cholecystectomy occurred in 35.9%, 25.1%, and 10.6% of participants, respectively. Indirect bilirubin was consistently associated with all outcomes, while associations with other laboratory variables varied by genotype. Genetic analyses confirmed associations between UGT1A1 variants and bilirubin levels and identified genome-wide associations with cholecystectomy. Novel loci, including FER1L6, LRFN5, and SDK2, were also implicated. These findings indicate a high burden of gallbladder-related disease in Brazilian individuals with SCD and highlight both established and novel genetic pathways that may inform risk stratification and preventive strategies.

6 - Corrigendum to 'Deleterious variants cluster in the A3 domain of factor VIII in people with severe hemophilia A and inhibitors': [Research and Practice in Thrombosis and Haemostasis Volume 9, Issue 6, August 2025, 103006].

Zuccherato LW, Souza RP, Camelo RM, Santana MAP, Dias MM, Jardim LL, de Oliveira AG, Lorenzato CS, Cerqueira MH, Franco VKB, Ferreira CB, de Albuquerque Ribeiro R, Etto LY, Roberti MDRF, Callado FMRA, de Cerqueira MAF, Pinto ISS, Garcia AA, Anegawa TH, Fontes Neves DC, Chaves DG, Rezende SM; HEMFIL and the Brazilian Immune Tolerance (BrazIT) Study.Res

Pract Thromb Haemost. 2026 Feb 6;10(1):103343.

doi: 10.1016/j.rpth.2026.103343. eCollection 2026 Jan.

PMID: 41704800 Free PMC article.

Resumo não disponível.

7 - J Pediatr Nurs. 2026 Jun 15:90:161-167.

Animated educational video for self-care in school-aged children with sickle cell anemia: Development and validation

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PMID: 42296706

DOI: 10.1016/j.pedn.2026.06.005

Resumo:

This study was conducted to develop and validate an animated educational video focused on self-care for school-aged children with sickle cell anemia METHODS: This is a methodological study performed in a specialized hematology center in Brazil conducted in sequential stages: identification of informational needs through interviews with children; theoretical foundation; script and storyboard development; content validation by experts; video production; evaluation by the target audience; and final video adaptation. Content validity was analyzed using the content validity index (CVI) RESULTS: The video presented high content validity indices, with an overall CVI of 0.99 in the expert evaluation and 1.00 in the target audience evaluation. The animation used accessible language and visual elements compatible with child development, favoring understanding of the disease and self-care practices CONCLUSION: The educational video proved to be a viable, methodologically rigorous, and appropriate educational technology to support health education and develop self-care in school-aged children with sickle cell anemia IMPLICATIONS FOR PRACTICE: Technology can be used by pediatric nurses and multidisciplinary teams to standardize

guidelines, strengthen child-centered communication, and support management of sickle cell anemia in outpatient, hospital, and school settings.