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

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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.

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

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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.