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BLACK LIVES MATTER.

Individuals with Sickle Cell Disease and Sickle Cell Trait have LIVES that MATTER. They already suffer where we can't see. The color of their skin should not cause them more pain.

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Background: Sickle cell disease is a monogenic disease with early onset manifestations that begin in childhood and is characterized by high clinical heterogeneity, being influenced by genetic and environmental factors. Among disease modulators fetal hemoglobin and alpha thalassemia are among the most important. There is little data on these factors in sickle cell disease patients from Africa and none from Angola where the prevalence of the disease is very high. The aim of this study was to explore the possible association between alpha thalassemia, Fetal hemoglobin, hematological indices and clinical events in Angolan sickle cell disease Hydroxyurea-naïve pediatric patients.

Methods: This cross-sectional study is part of a large study in an Angolan sickle cell disease cohort conducted in the Hospital Pediátrico David Bernardino in Luanda and in Hospital Geral do Bengo in Caxito. Sampling was performed between April and August 2019. A total of 200 sickle cell disease children were included, after guardian informed consent, being 51,5% females. A venous blood sample was collected from each participant and used for hematological analyses, electrophoresis for diagnosis confirmation, Fetal hemoglobin quantification by HPLC (Bio-Rad variant II). DNA isolation was done by Qiagen blood mini kit, and the 3.7 kb alpha thalassemia deletion was studied by GAP-PCR. ANOVA, nonConference Abstract parametric tests and Chi-square tests were applied to compare the means, medians or frequencies between the three alpha thalassemia genotypes. P-value <0.05 was considered statistically significant.

Results: We observed a slight deviation from Hardy-Weinberg equilibrium regarding frequencies of 3.7 alpha thalassemia deletion with a decrease in homozygous without deletion and an increase in 3.7 heterozygous (55.0%) and homozygous (12.5%). When the frequency of deletion was compared between gender an increase in 3.7 alpha thalassemia deletion was observed in boys (17.5% vs 7.8%). Moreover, an increase in alpha thalassemia 3.7 deletion frequency was observed in children older than 5 years old (11.7% vs 13.00 %). This increase supports the hypothesis that the co-Inheritance of Alphathalassemia may improve survival of sickle cell disease patients. Further, alpha 3.7 homozygous had a significantly higher age of first manifestation (6 months vs 11 months), lower number of blood transfusions by year (0.48 vs 0.18), higher hemoglobin level (7.24 vs 7.78 g/dL), lower MCV (81.75 vs 62.73 fl), MCH (27.28 vs 20.25 pg) and number of reticulocytes (11.62 vs 6.46 103 /L). There were no differences in Fetal hemoglobin between the 3 genotypes. Moreover, the number of cases of stroke, osteomyelitis, splenomegaly, splenectomy and hepatomegaly were lower in the presence of the deletion. In sickle cell disease patients who are alpha thalassemia homozygous there was any event of stroke or osteomyelitis.

Conclusions: For the first time in an Angolan population, the results obtained reveal that alpha thalassemia deletion in sickle cell disease influences the hematological and clinical aspects and produces a mild phenotype. Sickle cell disease is well characterized in high-income countries but not in sub-Saharan Africa where it is most prevalent. These results were concordant with other studies performed in other areas and could be consequence of a reduction in hemolytic rate due to a lower HbS production and consequent lower cell sickling.