Sickle cell anemia (SCA) is an inherited blood disorder with a broad range of complications, including vaso-occlusion and hemolytic anemia. SCA patients present arginine deficiency that contributes to a lower nitric oxide (NO) bioactivity. The amino acid citrulline increases arginine levels and promotes NO production.

We studied the association between hematological and biochemical parameters with genetic variants from eNOS gene, in 26 pediatric SCA patients. Effects of oral citrulline supplementation in SCA were also considered.

Results from this study show a significant statistical association between some parameters and genetic variants: high levels of neutrophils were associated with the eNOS4a allele and an increased reticulocyte count and high serum lactate dehydrogenase levels were associated with both the rs2070744_TT and the rs1799983_GG genotypes at eNOS gene. A symptomatic improvement was observed in patients with citrulline supplementation.

Our results reinforce the importance of NO bioactivity in SCA. We presume that NO, and its precursors such as citrulline, might be used as therapy to improve the quality of life of SCA patients.