Hemoglobinopathies, including α- and β-thalassemia, are the most common genetic disorders worldwide.

Carrier Screening can identify individuals who are at risk of having affected offspring, but to be clinically effective, it must have both high sensitivity and specificity.

Current ACOG guidelines recommend traditional methods for hemoglobinopathy screening, i.e., a combination of complete blood counts and hemoglobin electrophoresis.

However, the most common pathogenic hemoglobinopathy variant worldwide, α3.7, is not detected by these traditional screening methods.

Recent work by our group has demonstrated the superior sensitivity of next generation sequencing (NGS) over traditional screening methods for hemoglobinopathy variant detection (>99% detection rate), including the detection of α3.7.

In this post, we present support for the superiority of NGS and illustrate the undue burden caused by misidentification of hemoglobin variants through traditional screening methods.