



Genetic Modifiers of Hemoglobinopathies: Recent Advances and Future Directions

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Deadline for manuscript submissions:
closed (30 June 2024)

Message from the Guest Editors

Hemoglobinopathies, including sickle cell disease and thalassemia syndromes, are the most common monogenic diseases worldwide. Although their primary cause is well established through known pathogenic variants in the two globin gene clusters, the diverse clinical manifestations and the varying severity of hemoglobinopathies are largely attributed to the influence of genetic modifiers. Hundreds of genetic modifiers of different hemoglobinopathy phenotypes have already been identified by association studies, and genome-wide association studies (GWAS) in particular have proven to be powerful tools for the evaluation of the effect of genetic loci on specific disease phenotypes. Established modifiers serve as therapeutic targets, and have already greatly contributed to existing insights in the molecular mechanisms of disease pathogenicity.

For this Special Issue, we aim to compile current knowledge and identify gaps related to the role of genetic modifiers for hemoglobinopathies and to highlight key priority areas for future collaborative research through INHERENT.





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