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Sankaran - Boston-Brazil Collaborative Study of Sickle Cell Disease (Boston-Brazil_SCD)

Updated 02/14/2018

Introductory slides from 7/28/16 [1]

Fetal hemoglobin is an important modifier of clinical severity in sickle cell disease. Increased production of fetal hemoglobin can ameliorate the severity of this disease. Genome-wide association studies have provided important insight into the regulation of fetal hemoglobin. Indeed, such studies from our group and others have helped to identify the key regulator of hemoglobin switching, BCL11A. In addition, a variety of hematologic parameters also serve as important modifiers of clinical severity. This collaborative project aims to examine common, low frequency, and rare variation associated with fetal hemoglobin and other hematologic traits in a cohort of ~950 patients from Brazil with sickle cell disease. We plan to use comprehensive whole genome sequencing data to survey all of these types of variation using both single variant and gene burden analyses (as well as using other aggregation methods). We additionally plan to assess associations with clinical severity in this cohort alone, as well as in conjunction with other related studies.

Source URL (modified on 02/14/2018 - 6:57pm):https://topmed.nhlbi.nih.gov/group/boston-brazil_scd
Links
[1] https://topmed.nhlbi.nih.gov/system/files/documents/Sankaran_TOPMedMtg_7-27-16_v2.pdf