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# Sickle Cell Disease and Variation in the PAR4 Receptor

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### **Sickle Cell Disease and Variation in the PAR4 Receptor**

Sickle cell disease disproportionately affects African Americans in the U.S. Much can still be learned regarding determinants of frequency and severity of painful vaso-occlusive episodes in these patients. It has been reported that a variant in PAR4 (protease-activated receptor 4) has a unique distribution among African Americans. One variant (Thr120) is hyperactive, while the other (Ala120) is hypoactive. This receptor is present on platelets, vascular cells, and nociceptors. We wish ultimately to test the hypothesis that sickle cell patients with the hyperactive PAR4 receptor have greater pain severity. A genotype-phenotype correlation would have prognostic value. An adequately powered study to test this hypothesis would need to be multicenter. Therefore this is an ongoing pilot feasibility study to 1) Determine whether a sufficient number of sickle cell patients will consent to a focused genotype study; 2) Test if the current electronic health record (EHR) can be queried for an accurate depiction of sickle cell-related pain treatment; and 3) Collect single-center data on the genotype-phenotype correlation that can later be expanded to a multi-center study. 7/18 patients asked have consented to be in the study, the EHR in 5/7 enrolled has matched self-reported healthcare visits for vaso-occlusive episodes, and genetic studies are not being conducted until there are adequate numbers of samples. These in-progress results indicate patients will consent at an acceptable frequency and that the EHR is useful in objectively categorizing pain-severity phenotypes. Regardless of the date from the genetic component, preliminary results suggest a multi-center study could be productive.