Computable Phenotype for Identification of Patients with Sickle Cell Disease

The computable phenotype included 1) ICD-9 codes for sickle cell disease (sickle cell thalassemia 282.41, 282.42, HbSS 282.60, 282.61, 282.62, HbSC 282.63, 282.64, or "other sickle cell disease" 282.68, 282.69) in the problem list, any encounter diagnosis, or discharge diagnosis AND 2) two outpatient visits at least 30 days apart or one hospitalization in the database. The criteria of two outpatient visits was chosen to prevent the identification of patients misdiagnosed with sickle cell disease and potentially lost to follow up.

Queries were run using our i2b2 Clinical and Translational Research Informatics Data Warehouse. Our i2b2 data warehouse is primarily used for research and is a repository of patient data from the Epic EHR of our institution. The results of our retrospective queries were abstracted from the i2b2 database in the form of identified patient data, specifically medical record numbers.

Table 1. Summary of Computable Phenotype for the identification of Patients with Sickle Cell Disease

Inclusion Criteria	**Code**	**Description**
1. If a qualifying diagnosis of sickle cell disease (see ICD-9 codes and descriptions) has been made in the problem list, medical history, as a primary diagnosis at encounter, non-primary diagnosis at encounter, or as a discharge diagnosis	282.41	Sickle cell thalassemia without crisis
	282.42	Sickle cell thalassemia with crisis
	282.61	HbSS disease without crisis
	282.62	HbSS disease with crisis
	282.63	Sickle cell/HbC disease without crisis
	282.64	Sickle cell/HbC disease with crisis
	282.68	Other sickle cell disease without crisis
	282.69	Other sickle cell disease with crisis
2. AND two outpatient visits at least 30 days apart or one hospitalization in the electronic medical record	–	–
Exclusion Criteria		
1. If number of diagnoses for sickle cell trait diagnoses > qualifying sickle cell disease diagnoses	282.5	Sickle cell trait