
Start-up Resource – NHLBI CureSCi CDE Recommendations

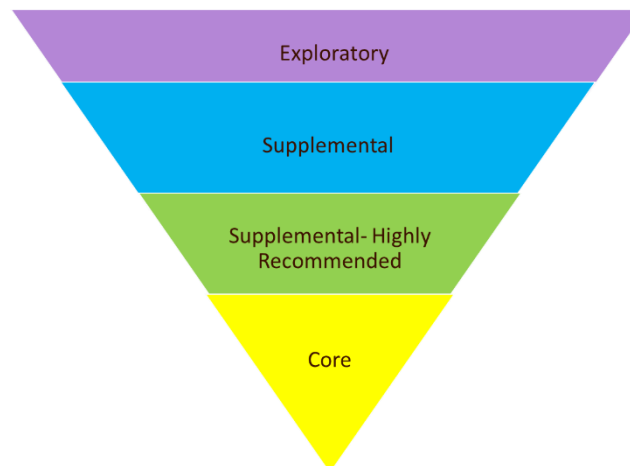
Each CDE or instrument could be classified according to the definitions below:

Core: Data element that collects essential/required information based on the current clinical research best practices. If these elements are absent/lacking, it may impact the utility of the domain. These have been used previously in SCD studies and are required should the study design involve this domain as an important target outcome. Validated measures were used when available.

Supplemental - Highly Recommended: Data element that is important for study design or protocol but not required for collection.

Supplemental: Data element which is commonly collected in clinical research studies. Use depends upon the study design, protocol or type of research involved.

Exploratory: Data elements that require further development but address gaps in data reporting and/or could substitute for an existing element after validation. These type of data elements show promise but require validation before they are ready for standard use in clinical research studies.



Summary of Core Recommendations: Sickle Cell Disease Genetic Studies CDEs

NIH Resources: *The NHLBI encourages researchers to use these NIH developed materials for NHBLI-sponsored research, when appropriate. Utilization of these resources will enable greater consistency for NHBLI-sponsored research studies. Some of these tools are free of charge.*

NIH Toolbox

Patient-Reported Outcomes Measurement Information System (PROMIS)

Adult Sickle Cell Quality of Life Measurement Information System (ASCQ-Me)

PhenX Toolkit (consensus measures for Phenotypes and eXposures)

The following is an overview of the domains/subgroup areas with Core CDEs. There are 240 Core CDEs and additional Core from Patient-Reported Outcomes and for Cardiac MRI or Echocardiogram (one should be used with preference to Cardiac MRI).

Physical Examination and Medical History	Sickle Cell Related Acute Painful Episodes	Genetics and Assays	Gene Product CRF
	Laboratory Tests		Immune Reconstitution
	Baseline Abnormal Hematopoiesis		Drug Product CRF
	Medical History	Monitoring Side Effects	Genetic Diagnostic Testing
	Transfusion History		Genotoxicity Form
	Endocrine Infertility Bone		Iron Overload
	Priapism		Endocrine, Fertility and Bone
	Acute Anemia		Serious Adverse Events
	Chronic Anemia		Mortality and Survival
	Chronic Pain		Numeric Rating Scale (NRS)
	Acute Spleen		ASCQ-Me Pain Impact
	Behavioral History		ASCQ-Me Pain Episodes
	Physical Exam		PedsQL Pain Impact SCD module (children)
	Vital Signs and Blood Gases	PedsQL Pain and Hurt, SCD modules	
	Cardiopulmonary, Renal and Cerebrovascular	Acute Chest Syndrome	Outcomes
Echocardiogram		PROMIS Fatigue/Pediatric Fatigue	
Cardiac MRI		PedsQL Multidimensional Fatigue Scale	
CNS Assessments		PROMIS 10 Global Health	
Six-Minute Walk Test		PROMIS 7+2 Global Health	
Pulmonary Function Test			
Renal Function Assessments			



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

This is the complete list of Core CDEs:

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Participant Characteristics/Demographics	Demographics	Date of birth	C00007
Participant Characteristics/Demographics	Demographics	Gender	C00035
Participant Characteristics/Demographics	Demographics	Ethnicity	C00020
Participant Characteristics/Demographics	Demographics	Race	C00030
Participant Characteristics/General Health History	Baseline Abnormal Hematopoiesis	Was bone marrow aspirate/biopsy done prior to initiation of conditioning regimen for curative therapy?	C60625
Participant Characteristics/General Health History	Behavioral History	Current tobacco use?	C00710
Participant Characteristics/General Health History	Behavioral History	Past tobacco use?	C00711
Participant Characteristics/General Health History	Behavioral History	Age started tobacco use	C00703
Participant Characteristics/General Health History	Behavioral History	Age stopped tobacco use	C00701
Participant Characteristics/General Health History	Behavioral History	Type(s) of tobacco used	C00719
Participant Characteristics/General Health History	Behavioral History	Other, specify	C18770
Participant Characteristics/General Health History	Behavioral History	Have you ever used an electronic vaporizer such as an e-cigarette?	C60111
Participant Characteristics/General Health History	Behavioral History	During the last 30 days, on how many days (if any) have you used an electronic vaporizer such as an e-cigarette?	C60112
Participant Characteristics/General Health History	Behavioral History	Average number of cigarettes smoked per day	C00709
Participant Characteristics/General Health History	Behavioral History Short Form	If Current or Former smoker: Year Started:	C60615
Participant Characteristics/General Health History	Behavioral History Short Form	Maximum packs/day	C60616
Participant Characteristics/General Health History	Behavioral History Short Form	Year Stopped (if Former smoker):	C60617
Participant Characteristics/General Health History	Medical History Core	Organ System	C60194



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Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Participant Characteristics/General Health History	Medical History Core	Other muscle/bone/joint condition, specify	C60195
Participant Characteristics/General Health History	Medical History Core	Other heart condition, specify:	C60196
Participant Characteristics/General Health History	Medical History Core	Other Kidney/Urinary/Genital condition, specify:	C60197
Participant Characteristics/General Health History	Medical History Core	Other Liver condition, specify:	C60198
Participant Characteristics/General Health History	Medical History Core	Other Spleen condition, specify:	C60199
Participant Characteristics/General Health History	Medical History Core	Other Lung Disease, specify:	C60200
Participant Characteristics/General Health History	Medical History Core	Other Neurological Problems, specify:	C60201
Participant Characteristics/General Health History	Medical History Core	Other Blood Problems Besides Sickle Cell, specify:	C60202
Participant Characteristics/General Health History	Medical History Core	Other Infections, specify:	C60203
Participant Characteristics/General Health History	Medical History Core	Specify VTE (Venous Thromboembolism)	C60204
Participant Characteristics/General Health History	Medical History Core	Has iron overload ever been assessed by liver biopsy?	C60207
Participant Characteristics/General Health History	Medical History Core	Cancer, describe	C60205
Participant Characteristics/General Health History	Medical History Core	Other Diseases/Ailments not listed, specify:	C60206
Participant Characteristics/General Health History	Medical History Core	Past History	C60208
Participant Characteristics/General Health History	Medical History Core	Year(s) of Diagnosis	C60209
Participant Characteristics/General Health History	Medical History Core	Ongoing?	C60210
Participant Characteristics/General Health History	Medical History Core	Comments	C18027
Participant Characteristics/General Health History	Medical History Core	Influenza vaccine?	C08096
Participant Characteristics/General Health History	Medical History Core	Most recent date	C60211
Participant Characteristics/General Health History	Medical History Core	Psychological/Mental Health	C60212
Participant Characteristics/General Health History	Medical History Core	Year of onset	C60213
Participant Characteristics/General Health History	Medical History Core	Still present?	C60214
Participant Characteristics/General Health History	Transfusion History	Ever transfused	C60574
Participant Characteristics/General Health History	Transfusion History	RBC alloantibodies (which ones?)	C60575



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Participant Characteristics/General Health History	Transfusion History	Estimated number of lifetime transfusions	C60576
Participant Characteristics/Social History	Social Status	Years of education	C00015
Disease or Treatment Related Events/Anemia	Acute Anemia	Hemoglobin Nadir	C60092
Disease or Treatment Related Events/Anemia	Acute Anemia	Reticulocyte Count Value	C60093
Disease or Treatment Related Events/Anemia	Acute Anemia	Reticulocyte Count Units	C60094
Disease or Treatment Related Events/Anemia	Acute Anemia	Platelet Count Value	C59968
Disease or Treatment Related Events/Anemia	Acute Anemia	Platelet Count Units	C60095
Disease or Treatment Related Events/Anemia	Acute Anemia	LDH Value	C60096
Disease or Treatment Related Events/Anemia	Acute Anemia	LDH Units	C60097
Disease or Treatment Related Events/Anemia	Acute Anemia	On HU?	C60098
Disease or Treatment Related Events/Anemia	Acute Anemia	Hydroxyurea doses held?	C60099
Disease or Treatment Related Events/Anemia	Acute Anemia	Hydroxyurea how long? (weeks, months)	C60100
Disease or Treatment Related Events/Anemia	Acute Anemia	Hydroxyurea how long? (weeks, months)	C60349
Disease or Treatment Related Events/Anemia	Acute Anemia	Cause	C60101
Disease or Treatment Related Events/Anemia	Acute Anemia	If Infection – Organism if known	C60102
Disease or Treatment Related Events/Anemia	Acute Anemia	If Infection – viral titers	C60103
Disease or Treatment Related Events/Anemia	Acute Anemia	Transfusion required?	C60104
Disease or Treatment Related Events/Anemia	Acute Anemia	Exchange	C60105
Disease or Treatment Related Events/Anemia	Acute Anemia	Simple	C60106
Disease or Treatment Related Events/Anemia	Acute Anemia	Simple (how much?)	C60107
Disease or Treatment Related Events/Anemia	Acute Anemia	Simple (how much?)	C60348
Disease or Treatment Related Events/Anemia	Acute Anemia	Associated conditions – e.g., acute chest	C60108
Disease or Treatment Related Events/Anemia	Acute Anemia	Other	C60109
Disease or Treatment Related Events/Anemia	Acute Anemia	Change from baseline	C60110
Disease or Treatment Related Events/Anemia	Chronic Anemia	Change from baseline	C60110
Disease or Treatment Related Events/Anemia	Chronic Anemia	Was an erythropoietin (EPO) level obtained?	C60134



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Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Anemia	Chronic Anemia	EPO level: ___ ___ ___ IU/L	C60135
Disease or Treatment Related Events/Anemia	Chronic Anemia	Date sample drawn	C60136
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Avascular necrosis at/before cure	C60577
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, where	C60578
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, stage (1-4) of Lesion 1	C60579
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, stage (1-4) of Lesion 2	C60580
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, stage Unknown/Unavailable	C60581
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If Yes, did you receive growth hormone?	C60582
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Hypogonadotropic hypogonadism: Age at diagnosis	C60583
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Fertility Preservation	C60584
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If no, why (pt may decline to answer)	C60585
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If Yes, how paid for (check all that apply)	C60586
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If insurance, copay	C60587
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If Yes, taking hydroxyurea at fertility preservation?	C60588
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If HU has been stopped, please indicate for how long?	C60589



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Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, what is banked	C60590
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, storage cost for cryopreserved tissue/gametes (per year):	C60591
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Storage cost paid by	C60592
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Ovarian tissue preserved	C60593
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If oocytes preserved	C60594
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, how many?	C60595
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If embryos preserved	C60596
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, how many?	C60597
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If semen analysis performed, results:	C60598
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Sperm concentration	C60599
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Sperm motility:	C60600
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Sperm morphology	C60601
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Sperm preserved	C60602
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Are you continuing to preserve your oocytes/ ovarian tissue /embryos?	C60603



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Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If yes, what is current cost of preservation	C60604
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	If no, why	C60605
Disease or Treatment Related Events/Fertility and Bone	Endocrine, Infertility and Bone Health	Have you been pregnant since your last study visit?	C60606
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Acute Chest Syndrome	C60155
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Temperature (highest on day of diagnosis)	C60160
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Temperature (highest on day of diagnosis)	C60161
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Heart rate (highest on day of diagnosis)	C60162
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Respiratory rate	C01535
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	Systolic blood pressure on day of diagnosis	C60163
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	SpO2 (O2 saturation) in room air (FiO2 = 0.21) decreased by 2% or more from baseline oximetry	C60164
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	O2 Saturation Value:	C60165
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	PaO2 < 60 mmHg	C60166
Disease or Treatment Related Events/Pain	Acute Chest Syndrome	PaO2 Value:	C60167
Disease or Treatment Related Events/Pain	Chronic Pain	Has the patient had at least 15 days/month with pain for at least 6 months?	C60613
Disease or Treatment Related Events/Pain	Chronic Pain	Does the patient display at least 1 of the following signs	C60614
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	In the last 12 months, has participant used an After Hours/Urgent Care Clinic/day hospital/infusion clinic or Emergency Room due to vaso-occlusive crisis (sickle cell related acute pain episode)?	C60082



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Number of inpatient admissions for vasoocclusive crisis or VOC (sickle cell related acute pain episode) in last 12 months	C60083
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Dates	C60084
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Length of stay each	C60085
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Number of treat and release day hospital/ infusion room visits for VOC/ 12 months	C60086
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Number of treat and release ED visits for VOC/ 12 months	C60087
Disease or Treatment Related Events/Pain	Sickle Cell Related Acute Painful Episodes	Total Acute care utilization/ 12 months	C60088
Disease or Treatment Related Events/Priapism	Priapism Core	How long has patient reported erection?	C60607
Disease or Treatment Related Events/Priapism	Priapism Core	Percent erect:	C60608
Disease or Treatment Related Events/Priapism	Priapism Core	Intervention provided	C60609
Disease or Treatment Related Events/Priapism	Priapism Core	How long did episode last until complete resolution or discharge home?	C60610
Disease or Treatment Related Events/Priapism	Priapism Core	Was the patient prescribed medications for prevention?	C60611
Disease or Treatment Related Events/Priapism	Priapism Core	If yes	C60612
Disease or Treatment Related Events/Renal	Renal Function Assessments	Serum Creatinine	C58735
Disease or Treatment Related Events/Renal	Renal Function Assessments	Serum Cystatin C	C60332
Disease or Treatment Related Events/Renal	Renal Function Assessments	Estimated GFR	C60333



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Renal	Renal Function Assessments	Urine Albumin	C59206
Disease or Treatment Related Events/Renal	Renal Function Assessments	Urine Creatinine	C59201
Disease or Treatment Related Events/Renal	Renal Function Assessments	Urine Creatinine	C59202
Disease or Treatment Related Events/Renal	Renal Function Assessments	Urine Albumin/Creatinine	C59013
Disease or Treatment Related Events/Renal	Renal Function Assessments	Daily Urine Output	C60343
Disease or Treatment Related Events/Renal	Renal Function Assessments	Systolic Blood Pressure	C01565
Disease or Treatment Related Events/Renal	Renal Function Assessments	Diastolic Blood Pressure	C01507
Disease or Treatment Related Events/Renal	Renal Function Assessments	24 hour diastolic blood pressure	C01502
Disease or Treatment Related Events/Spleen	Acute Spleen	Acute splenomegaly	C60113
Disease or Treatment Related Events/Spleen	Acute Spleen	Acute splenomegaly Date and Time (m m /d d/ y y y y)	C60114
Disease or Treatment Related Events/Spleen	Acute Spleen	Spleen size by PE	C60115
Disease or Treatment Related Events/Spleen	Acute Spleen	Spleen size by PE (cm below left costal margin)	C60116
Disease or Treatment Related Events/Spleen	Acute Spleen	Hypersplenism	C60117
Disease or Treatment Related Events/Spleen	Acute Spleen	Splenic sequestration episodes - 0,1,2, etc	C60118
Disease or Treatment Related Events/Spleen	Acute Spleen	Abdominal pain	C60119
Disease or Treatment Related Events/Spleen	Acute Spleen	Date: (m m /d d/ y y y y)	C60120
Disease or Treatment Related Events/Spleen	Acute Spleen	Hemoglobin nadir	C60121
Disease or Treatment Related Events/Spleen	Acute Spleen	Platelet count nadir	C60122



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Disease or Treatment Related Events/Spleen	Acute Spleen	PRBC transfusion	C60123
Disease or Treatment Related Events/Spleen	Acute Spleen	Hemodynamic instability	C60124
Disease or Treatment Related Events/Spleen	Acute Spleen	Imaging done	C60125
Disease or Treatment Related Events/Spleen	Acute Spleen	Spleen size	C60126
Disease or Treatment Related Events/Spleen	Acute Spleen	Surgical Splenectomy	C60127
Disease or Treatment Related Events/Spleen	Acute Spleen	Surgical Splenectomy Date	C60128
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Was a cardiac MRI completed?	C60215
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Give date	C60216
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Date of birth	C00007
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Sex	C00035
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Height	C01522
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Weight	C01541
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Body surface area (BSA) in square meters (m ²)	C56157
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Systolic blood pressure (SBP)	C01565
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Diastolic blood pressure (DBP)	C01507
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Heart rate at baseline	C01521
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Type of scanner	C60217
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Strength of magnet	C02495
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Contrast	C10570
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV ejection fraction (LVEF)	C60230
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-diastolic volume (LVEDV)	C60231
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-diastolic volume (LVEDV) Indexed to BSA	C60232
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-systolic volume (LVESV)	C60233
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-systolic volume (LVESV) Indexed to BSA	C60234
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV stroke volume (LVSV)	C60235
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV stroke volume (LVSV) Indexed to BSA	C60247



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Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Cardiac output (CO)	C14539
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-diastolic Mass	C60236
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LV end-diastolic Mass Indexed to BSA	C60238
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Left Atrium (LA) diameter	C60237
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LA Volume	C60239
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	LA Volume Indexed to BSA	C60240
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	RV ejection fraction (RVEF)	C60242
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	RV end-diastolic volume (RVEDV)	C60243
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	RV end-diastolic volume (RVEDV) Indexed to BSA	C60244
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	RV end-systolic volume (RVESV)	C60245
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	RV end-systolic volume (RVESV) Indexed to BSA	C60246
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Iron Analysis T2* Heart	C60289
Assessments and Examinations/Imaging Diagnostics	Cardiac MRI	Iron analysis T2* Liver	C60290
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Participant ID number	C19247
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Was an echocardiogram completed?	C10723
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Body surface area (BSA)	C56157
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Heart rate (1st frame of echo)	C01521
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Left ventricle (LV) Septum	C60292
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Left ventricle (LV) Posterior wall	C60293
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-diastolic dimension (LVEDD)	C60294
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-systolic dimension (LVESD)	C60295
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-diastolic volume (LVEDV)	C12896
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-diastolic volume (LVEDV) Indexed to BSA	C12898
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-systolic volume (LVESV)	C12900
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LV end-systolic volume (LVESV) Indexed to BSA	C12902
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Left ventricle (LV) EF Simpson's	C10740



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Left ventricle (LV) EF visual estimate (if Simpson's not possible)	C60297
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Fractional shortening (pediatric only)	C60298
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LVOT diameter	C60300
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LVOT Velocity Time Integral (VTI)	C60302
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LA volume (LAV)	C60307
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	LA volume (LAV) Indexed to BSA	C60308
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	RV basal diameter	C60309
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Tricuspid Annular Plane Systolic Excursion (TAPSE)	C60311
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	RA volume (RAV)	C60313
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	RA volume (RAV) Indexed to BSA	C60314
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	IVC diameter	C60315
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Estimated RA pressure	C60316
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Tricuspid regurgitation peak velocity	C60317
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Estimated RV systolic pressure	C12909
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Mitral inflow peak E velocity	C60318
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Septal e' velocity	C60320
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Lateral e' velocity	C60321
Assessments and Examinations/Imaging Diagnostics	Echocardiogram	Tricuspid regurgitation	C10743
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Method Used	C60434
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Specify	C60435
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results HgS %	C60436



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Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results HgC %	C60437
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results HgF %	C60438
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results HgA %	C60439
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results HgA2 %	C60440
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results Genetic Modified A Variant %	C60441
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Results Other %	C60442
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Specify	C60443
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Genetic Diagnostic Testing	Thalassemia Testing Results	C60444
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Visit Date	C17409
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Sample collected	C60142
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Reason not done	C60143
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Collection Date	C60144
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Collection Time	C60350
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Accession Number	C60145



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Antiglobulin Test, Direct	C60146
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Antiglobulin Test, Indirect	C60147
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	ABO Blood Group	C60148
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Sample collected	C60149
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	If 'Blood gas sample collect indicator' = No, Reason not done	C60150
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Collection Date and Time	C01701
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Laboratory test name	C01705
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Result	C01706
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Unit	C01711
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Comparison to Reference Range	C60151
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Clinically Significant	C60152
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Method of Test	C60153
Assessments and Examinations/Laboratory Tests and Biospecimens/Biomarkers	Laboratory Tests	Specify other hemoglobin	C60154
Assessments and Examinations/Physical Examinations	Physical Exam	Date of Exam	C01010



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Assessments and Examinations/Physical Examinations	Physical Exam	Was a physical exam performed on the participant/subject	C01015
Assessments and Examinations/Physical Examinations	Physical Exam	Other, specify	C18666
Assessments and Examinations/Physical Examinations	Physical Exam	Body System	C00312
Assessments and Examinations/Physical Examinations	Physical Exam	Result	C01012
Assessments and Examinations/Physical Examinations	Physical Exam	Describe Abnormality or Comment if Body System is Not Examined	C01013
Assessments and Examinations/Vital Signs and Other Body Measures	Vital Signs and Blood Gases	Heart rate beats per minute	C01521
Assessments and Examinations/Vital Signs and Other Body Measures	Vital Signs and Blood Gases	Blood pressure	C01565
Assessments and Examinations/Vital Signs and Other Body Measures	Vital Signs and Blood Gases	Blood pressure	C01507
Assessments and Examinations/Vital Signs and Other Body Measures	Vital Signs and Blood Gases	Oxygen saturation (%)	C01554
Assessments and Examinations/Vital Signs and Other Body Measures	Vital Signs and Blood Gases	Respiratory rate in breaths per minute	C01535
Safety Data/Adverse Events/Toxicities	Iron Overload	Was a liver MRI performed?	C60626
Safety Data/Adverse Events/Toxicities	Iron Overload	If Yes, what was the liver iron content (LIC)?	C60627
Safety Data/Adverse Events/Toxicities	Iron Overload	Known, by	C60628
Safety Data/Adverse Events/Toxicities	Iron Overload	Date of most recent liver biopsy	C58755
Safety Data/Adverse Events/Toxicities	Iron Overload	Iron Result	C60629
Safety Data/Adverse Events/Toxicities	Iron Overload	% Serum transferrin (% saturation) Result	C60630
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VISIT [X] CRF (Time since Infusion):	C60631



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Date sample drawn	C60392
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Current CBC	C60632
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Current Hemoglobin analysis	C60633
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Tissue monitoring site:	C60634
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Tissue monitoring site:	C60635
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Cells sorted?	C60636
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Immunophenotype	C60637
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Blood VCN	C60481
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	CD3 depleted?	C60482
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VCN in Specific Lineages: CD3+	C60483
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VCN in Specific Lineages: CD19+	C60484
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VCN in Specific Lineages: CD15+	C60485
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VCN in Specific Lineages: GPA+	C60486
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	VCN in Specific Lineages: CD14+	C60487
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percentage of Blood Cells Positive for Integrating Vector: PBMC	C60488
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percentage of Blood Cells Positive for Integrating Vector: BFU-E	C60489
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone Marrow VCN: BMMC	C60490
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone Marrow VCN: CD34+	C60491
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone Marrow VCN: CFC	C60492
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percentage of Bone Marrow Cells Positive for Integrating Vector: BMMC	C60493
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percentage of Bone Marrow Cells Positive for Integrating Vector: CD34+	C60494
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percentage of Bone Marrow Cells Positive for Integrating Vector: CFC	C60495



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Blood: CD3 depleted?	C60638
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion	C60639
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target indels:	C60640
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target indels:	C60641
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion in Specific Blood Lineages: CD3+	C60519
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion in Specific Blood Lineages: CD19+	C60520
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion in Specific Blood Lineages: CD15+	C60521
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion in Specific Blood Lineages: GPA+	C60522
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Percent conversion in Specific Blood Lineages: CD14+	C60523
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target Indels in Specific Blood Lineages: CD3+	C60524
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target Indels in Specific Blood Lineages: CD19+	C60525
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target Indels in Specific Blood Lineages: CD15+	C60526
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target Indels in Specific Blood Lineages: GPA+	C60527
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	On-target Indels in Specific Blood Lineages: CD14+	C60528
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target Indels in Specific Blood Lineages: CD3+	C60642
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target Indels in Specific Blood Lineages: CD19+	C60643



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target Indels in Specific Blood Lineages: CD15+	C60644
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target Indels in Specific Blood Lineages: GPA+	C60645
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Off-target Indels in Specific Blood Lineages: CD14+	C60646
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	BMMC Percent Conversion	C60529
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	BMMC On-Target Indels:	C60532
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	BMMC Off-Target Indels:	C60647
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: CD34+ Percent Conversion	C60530
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: On-target Indels:	C60533
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: CD34+ Off-target Indels:	C60648
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: CFC percent conversion:	C60531
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: CFC On-target Indels:	C60534
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Bone marrow: CFC Off-target Indels:	C60649
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Method	C60650
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Reporter gene assay: Specify:	C60651
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	Is the on-target indel frequency associated with an adverse outcome? (e.g. thalassemia phenotype)	C60652
Outcomes and End Points/Genetics/Assays	Gene Product Follow-Up	If yes, describe	C60653
Outcomes and End Points/Genetics/Assays	Immune Reconstitution Core	IgA:(QIGA)	C60622
Outcomes and End Points/Genetics/Assays	Immune Reconstitution Core	IgG:(QIGG)	C60623
Outcomes and End Points/Genetics/Assays	Immune Reconstitution Core	IgM:(QIGM)	C60624



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Domain/Subdomain	Case Report Form Name	CDE Long Name	CDE ID
Outcomes and End Points/Mortality/Survival	Death Form	Event:	C02203
Outcomes and End Points/Mortality/Survival	Death Form	Date last known alive:	C59945
Outcomes and End Points/Mortality/Survival	Death Form	Date of death:	C58800
Outcomes and End Points/Mortality/Survival	Death Form	Primary cause of death:	C04800
Outcomes and End Points/Mortality/Survival	Death Form	Secondary cause(s) of death:	C59947
Outcomes and End Points/Mortality/Survival	Death Form	Source of cause of death (select all that apply):	C59948
Outcomes and End Points/Mortality/Survival	Death Form	Was the death related to, or a complication of, gene therapy for sickle cell disease?	C59949
Treatment/Intervention Data/Therapies	Drug Product CRF	Purity: Proportion of CD34+ cells (of CD45+)	
Treatment/Intervention Data/Therapies	Drug Product CRF	Identity: Confirm/quantify production of globin chain of interest	
Treatment/Intervention Data/Therapies	Drug Product CRF	VCN, INDELS or Correction	
Treatment/Intervention Data/Therapies	Drug Product CRF	off-target editing frequency	
Treatment/Intervention Data/Therapies	Drug Product CRF	CFC Assay	
Treatment/Intervention Data/Therapies	Drug Product CRF	Potency Assay: Additional drug product potency assay to measure the "intended biological effect" as to be determined by sponsor based on MOA of product	

Genetics/Assays References:



**Summary of Core Recommendations:
Sickle Cell Disease Genetic Studies CDEs**

Immune Reconstitution	Immunoglobulins Panels: serum IgG, IgA, IgM level	Balandya E, et al 2016
Gene Product CRF	VCN, INDELS or percentage of correction in peripheral blood	Bulk VCN/Editing/Correction in a whole blood sample
Gene Product CRF	Laboratory Procedure Hemoglobin Genetic Modified A Variant Result Percentage Value	https://www.aphl.org/aboutAPHL/publications/Documents/NBS_HemoglobinopathyTesting_122015.pdf
Gene Product CRF	Proportion of transduced, edited or corrected cells in peripheral blood: Single cell assessment of transduction/editing/correction	
Drug Product CRF	Purity: Proportion of CD34+ cells (of CD45+)	https://www.fda.gov/media/113760/download
Drug Product CRF	Identity: Confirm/quantify production of globin chain of interest	https://www.fda.gov/media/113760/download
Drug Product CRF	VCN, INDELS or Correction	https://www.fda.gov/media/113760/download
Drug Product CRF	off-target editing frequency	https://www.fda.gov/media/113760/download
Drug Product CRF	CFC Assay	https://www.fda.gov/media/113760/download
Drug Product CRF	Potency Assay: Additional drug product potency assay to measure the "intended biological effect" as to be determined by sponsor based on MOA of product	https://www.fda.gov/media/79856/download
Laboratory Tests	The method used for hemoglobin analysis.	https://www.aphl.org/aboutAPHL/publications/Documents/NBS_HemoglobinopathyTesting_122015.pdf

CURE SICKLE CELL.

Summary of Core Recommendations: Sickle Cell Disease Genetic Studies CDEs

Additional Core Items of Note	Questions Text	
Genotoxicity Form	Was bone marrow aspirate/biopsy done prior to infusion?	Yes/No
Genotoxicity Form	Date	
Genotoxicity Form	Is there evidence of Premalignant/Malignant Hematopoiesis	Yes/No
Genotoxicity Form	Was a genetic mutational panel performed?	Yes/no
Baseline Abnormal Hematopoiesis	Was bone marrow aspirate/biopsy done prior to initiation of conditioning regimen for curative therapy ?	Yes/no

Summary of Core Recommendations: Sickle Cell Disease Genetic Studies CDEs

Core Instrument Recommendations:

Cardiac MRI Or Echocardiogram
6- Minute Walk
Pulmonary Function Test
CNS Assessments (Baseline and Follow-Up MRI/MRA, TCD)
Numeric Rating Scale (NRS)
ASCQ-Me Pain Impact
ASCQ-Me Pain Episodes
PedsQL Pain Impact SCD module (children)
PedsQL Pain and Hurt, SCD modules
PROMIS Pain Interference; Adult and Pediatric modules
PROMIS Fatigue/Pediatric Fatigue
PedsQL Multidimensional Fatigue Scale
PROMIS 10 Global Health
PROMIS 7+2 Global Health

For the complete list of CureSCi CDE recommendations, please see the [CureSCi CDE website](#).