

A Longitudinal Cohort Study of Patients with Thalassemia in the Thalassemia Clinical Research Network

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Synopsis:

The aim of this study is to describe the prevalence and incidence of complications related to thalassemia among participants.

SECONDARY AIMS:

1. To describe genotypic and phenotypic variation among participants;
2. To assess the prevalence, development, and treatment of the following complications: low bone mineral density, cardiac disease, pulmonary hypertension, growth defects, hypogonadotropic hypogonadism, and other endocrine defects;
3. To assess body iron burden and its relationship to prevalence and development of complications of thalassemia;
4. To assess fertility and pregnancy outcomes;
5. To estimate mortality rates and predictors of mortality risk;
6. To assess relationships among adherence, quality of life, and complications of thalassemia;
7. To determine the availability of potential participants for other Thalassemia Clinical Research Network studies.

The purpose of the Thalassemia Clinical Research Network (TCRN) Thalassemia Longitudinal Cohort (TLC) is to describe the prevalence and incidence of complications related to thalassemia among participants. The TLC will collect routine clinical care data at baseline and annually for patients with thalassemia. Standardizing clinical evaluations among sites and patients will accelerate the improvement of care for patients with thalassemia. Detailed genotypic and phenotypic characterization of the cohort and maintenance of a plasma bank will support conduct of ancillary substudies investigating novel hypotheses. Finally, the TLC will facilitate recruitment into other TCRN protocols.

The TCRN is a cooperative network of clinical sites, their affiliated satellites, a coordinating center, and NHLBI formed to examine the blood disorder thalassemia. The TCRN was established to conduct multi-center trials and to initiate studies of existing and future therapies for the treatment of thalassemia. To support this mandate, the TCRN established a registry of over 800 patients with thalassemia followed at participating sites. The utility of the Registry is limited because data were collected only at the time of enrollment (2001-present) and thus do not permit studies of change over time.

The TLC will build on the Registry by adding longitudinal follow-up through annual collection of clinical and laboratory data. Participants in the original Registry and new patients will be eligible to participate in the TLC. This is a minimal risk, non-interventional study. From the standpoint of the TCRN network sites and the supervisory regulatory committees, this new protocol may be thought of as functionally equivalent to the Registry (approved in the first five year period of the TCRN) with the following changes.

1. The survey instruments are updated to 2006 care from 2001. Thus, T2* MRI for cardiac iron assessment, R2 MRI (Ferriscan) for liver iron assessment, and oral iron chelator deferasirox are added, essentially as amendments to existing Registry documents.
2. The survey will be repeated annually, rather than a cross-sectional, one-time assessment.
3. Quality of Life instruments, and other potential ancillary studies will be possible with the longitudinal cohort.
4. The requirement for annual follow up as an entry criterion, and for ongoing data review, requires new informed consent.