Top Ten Things To Know
Cardiac Dysfunction in Beta Thalassemia Major

1. Beta-thalassemia major (TM) is a genetic condition where the beta globin chain of hemoglobin A (Hb) is severely reduced or absent. Erythropoiesis is ineffective, resulting in profound, life-threatening anemia from about 1-2 years of age.

2. TM is most common in areas of the world where the population is exposed to malaria (Asia, Middle-East, and Mediterranean Europe). The frequent hemoglobin genetic mutations in these populations that infer protection against malaria also result in mutations for TM.

3. Lifetime blood transfusions are required to prolong life, but total body iron increases, resulting in iron overload.

4. Heart failure (HF) is the most common cause of death in TM and primarily results from cardiac iron accumulation.

5. This consensus statement addresses the diagnosis and treatment of cardiac dysfunction in beta-thalassemia major, including how to measure cardiac iron and function, to identify and treat patients at high risk in order to prevent heart failure, and to diagnose and treat HF.

6. Treating HF in TM now includes iron chelation therapy. Agents discussed include deferoxamine, deferiprone, and deferasirox.

7. The use of chelation therapies for TM has moved the typical age of death by primarily cardiac causes from 10 years in the 1970’s to a median age of 35 years.

8. “Iron cardiomyopathy is the most common and feared complication of TM, but as it is caused by iron toxicity, it is reversible.”

9. Acute and chronic treatment of HF in TM is different for several reasons:
   - the patients are younger,
   - because this HF is a result of a toxic cardiomyopathy, the therapeutic goal is removal of the iron rather than treating the myocardial performance, and
   - additional co-morbidities may exist that require identification and specific treatment.

10. Many questions remain unanswered. There is a lack of high-quality studies in large populations, but trials are under way looking at new chelators and chelator combinations. Non-invasive ways to measure iron and strategies to prevent cardiac iron loading are other directions for research.

Pennell DJ, et al; on behalf of the American Heart Association Committee on Heart Failure and Transplantation of the Council on Clinical Cardiology and Council on Cardiovascular Radiology and Imaging. Cardiovascular Function and treatment in β-thalassemia major: a consensus statement from the American Heart Association. Circulation, 10.1161/CIR.0b013e31821c7c64.
http://circ.ahajournals.org/lookup/doi/10.1161/CIR.0b013e31829b2be6