Stephanie Badini

Sir David Weatherall, an Oxford researcher-physician, was among the first to use the tools of molecular biology to understand thalassemia. He was in New York to receive the Lasker-Koshland Special Achievement Award for "50 years of international statesmanship in biomedical science."
http://www.nytimes.com/2010/10/12/science/12conv.html?_r=0

The alpha and beta thalassemias together with the sickle
cell disorders are by far the commonest diseases due to a single defective gene in the world population. They occur at their highest frequencies in the low and middle income countries of the tropical belt. About half of the severe forms of beta thalassemia are comprised of a condition called HbE beta thalassemia which occurs at an extremely high frequency throughout south and southeast Asia. Two major research programs relating to the thalassemias are being carried out with the support of the Anthony Cerami and Ann Dunne Foundation for World Health.

In a joint program between
Oxford University and Sri Lanka over 200 patients with HbE beta thalassemia have been followed carefully for the last 15 years. They show remarkable heterogeneity in their clinical course despite being due to similar mutations. At one end of the spectrum patients require lifelong transfusion for survival while at the other end patients grow and develop normally, albeit at a low hemoglobin level and require either occasional or no transfusion. The research program is directed at finding the genetic causes for this remarkable clinical diversity together with any possible environmental factors. About 40% of the variability has already been ascribed to a few genetic variants and currently studies are being carried out on the genomes of these patients in collaboration with the Sanger Center in Cambridge, UK to try to determine the other genes involved. An understanding of this remarkable clinical diversity is absolutely critical for the future management of these conditions in Asia.

The other research
which is being supported by the Foundation relates to the remarkable heterogeneity of the distribution of the different forms of thalassemia within relatively small geographical distances, an observation which makes it extremely difficult to determine the total burden of these diseases for the healthcare programs of affected countries. Recently a survey has been carried out in Sri Lanka of over 7,000 individuals, and even in this small island population there is quite remarkable variability in the distribution of carriers for these conditions in different parts of the country.
Currently the possible mechanisms for this remarkable phenomenon are being addressed. As well as providing a better understanding of the evolutionary biology of these diseases this work has important practical potential for allowing the true birth frequency of these conditions to be determined and hence for advising governments about the likely clinical load of these disorders and how they might be best controlled or prevented.