Abstract Title: Sickle Cell Anaemia: Kal, Aaj aur Kal

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Abstract: Sickle Cell Anaemia (SCA) is an original monogenic genetic disorder due to a universal point mutation in the beta-globin gene that replaces glutamic acid with valine leading to sickling of erythrocytes. This phenomenon is responsible for majority of clinical phenotypes of SCA, which include hemolytic anaemia, vaso-occlusive crisis, persistent pain, and many other complications. India happens to be one of the major capitals of SCA, and close to 17 states have people afflicted by this disease. The disease is known to be very severe across the world. However, it has variable phenotype. Especially in India, where it is said to be mild, largely due to high levels of fetal haemoglobin and co-inheritance of alpha-thalassemia. While it is known to be highly prevalent in tribal populations, the disease is also found in the common people as well. Despite high disease frequency and very severe morbid states, the data on Sickle Cell Anaemia is sketchy and only comes from specific pockets. Considering all these lacunae in mind, under the CSR sickle cell anaemia mission, a comprehensive program was conceived that included development of a program for screening of the population and identifying the hidden burden of the disease on the society, develop a screening cum confirmatory test, and understand the genetic and proteomic markers to predict the disease onset.

In this talk, I will present the successes from this program that are likely to be implemented in the reduction of Sickle Cell Anaemia burden as a part of National Sickle Cell Anaemia Mission. This will cover various aspects of basic research translating into a focused approach on handling this genetic disorder. It is also envisaged that the observations from can serve as a milestone and guide the direction for many other similar genetic disorders.