

Cost Effective Innovative Screening Device for Sick Cell Disease (SCD)

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Received Date: September 10, 2018; **Published Date:** September 24, 2018

Abbreviations: SCD: Sick Cell Disease; HPLC: High Performance Liquid Chromatography; Hb: Haemoglobin; POC; Point of Care

Commentary

Sickle cell disease (SCD) is the most prevalent inherited blood condition worldwide resulting from single DNA mutation within the beta globin gene [1,2]. The disease is highly prevalent in areas of sub-Saharan Africa, Saudi Arabia, India, and Mediterranean countries such as Turkey, Greece, Italy, South East Asia and eastern Brazil. Due to high population movements, there is gene flow from high allele frequency areas to Europe, and parts of America [3]. This has led to sizeable sickle cell populations emerging in previously unaffected areas of the world. Hence, it is a global health concern and a major economic burden both for the developed and developing countries alike. Even after over a century of its first clinical reporting in the US in 1910 and its identification as genetic inheritance due to a point mutation in 1949 we are still battling to bring down the score and better management modalities [4,5]. WHO has declared SCD as a public health priority [6]. It has been estimated that over more than 14 million individuals worldwide has been afflicted with SCD, and its magnitude is more intense in economically disadvantaged populations. Six million children are born with SCD or sickle cell trait every year.

Up to 90% of newborns with SCD will die of this disease if undiagnosed.

SCD has a high prevalence in India, especially in the central and western regions, and poses a considerable health burden [6,7]. With effective interventions, several infectious diseases are being brought under control but genetic diseases are assuming a proportionately greater importance. Sickle cell disease contributes significantly to morbidity and mortality world over and India is no exception. This disease in India affects the tribal community more than the non tribal population [8]. According to the Census of India in 2011, the tribal population of India is 8.6 per cent of the total population which is about 67.8 million people. Hence, the need of the hour is to necessitate for a well established innovative screening programmes, a nationwide reporting system or registries to minimize the occurrence of SCD [9-11].

The aetiology of SCD can be briefed like this: The single amino acid substitution in the beta chain wherein glutamic acid is replaced for valine at position 6, thereby producing abnormal haemoglobin called Hb S, instead of normal haemoglobin Hb A. On deoxygenation, sickle haemoglobin undergoes a change in conformation that promotes intracellular polymerization, which leads to an alteration of the normal biconcave erythrocyte disc into the distinctive and pathological crescent shape.

The resulting hemolytic anaemia manifests as recurrent vasoocclusion and organ damage. The varied clinical symptoms results in more life threatening consequences leading to high mortality and morbidity [12].

Having such an aggressive manifestation with just one genetic point mutation, all efforts are being channelized towards addressing the disease. There are various well established technologies available to detect SCD that are considered as gold standards but incidentally most of it is all lab based. Current laboratory-based screening platforms, although widely available are not feasible for operation at the point of care (POC) in the resource-limited settings and in the developing world due to high infrastructure and operational costs [13]. Also, there is a need for skilled operators. Moreover, typical turnaround time for screening test results in low-resource environment is too long, sometimes takes weeks together. Some of the high end diagnostic assays which are routinely used for diagnosis of SCD includes: High performance liquid chromatography (HPLC), haemoglobin (hb) electrophoresis, Isoelectric focussing, Sick cell Scan, heme chip to name a few [14]. All the above mentioned tests are considered as gold standards from the point of view of SCD diagnosis in spite of some of its pit falls. HPLC is very expensive in terms of cost, requires a well skilled technician and can be performed only in batches. Same is for the case of haemoglobin (hb) electrophoresis also. Sick cell Scan is also very expensive in terms of costs and limits of detection are compromised. Having briefed on some of the ongoing available tests for diagnosis of SCD, the need of the hour is an affordable and non invasive screening device at point of care, in the remote settings, especially in the tribal area where the incidence is very high. Any confirmation if required can be further ascertained using the gold standard as mentioned above. Hence there is an urgent unmet need for a better innovative screening tools and technologies. The scientific community should have a disruptive outlook towards combating this disease, more particularly in terms of early detection, and specially the neonates.

Better screening tools here defines, the technology should be user friendly, portable and compact and can be used at point of care (POC). It should be cost effective, easy to interpret without any manual intervention and more particularly should not involve any biological waste i.e., it should be non invasive with high degree of sensitive and specific detection capabilities for sickle cell disease. We at ChroGene Aarogyam in collaboration with biomedical team in Osmania University Hyderabad, India are working towards bringing out such a device which can help towards addressing the pressing societal issues because of its high prevalence rate in tribal region in our country.

Further we would like to extend this technology to other countries where the prevalence is very high. Hence, the technology that we intend to introduce is just not focussed towards addressing only the domestic concern but the global prevalence at large. The Indian Patent application with the title "NON -INVASIVE POINT OF CARE DIAGNOSTICS FOR SICKLE CELL DISEASE" has already been filed and has been published recently in Indian Patent journal.

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