Research Proposal

Running Title: Prevalence and clinical patterns of hemoglobinopathies diagnosis in Indian population by using CE-HPLC method : A pilot study

Background of research area :

The hemoglobinopathies comprise inherited disorders of the structure or synthesis of hemoglobin. They are the commonest single gene disorders in the world. It is estimated that about 450,000 infants with hemoglobinopathies are born in the world. Almost 85.9% of these hemoglobinopathies are sickle cell disease (SCD). Births of children with β-thalassemia alone or combined with hemoglobin (Hb) E are about 44, 000 (9.7%) per year, whereas infants affected with Hb Barts and HbH disease are about 20,000 (4.4%). Approximately 80% of the annual births of babies with these disorders occur in low- or middle-income countries. These disorders originated in populations in tropical Africa, Asia, and the Mediterranean region and have spread via migration throughout the world. They are, therefore, of concern in all countries

Literature of review

The prevention of inherited haemoglobinopathies has been internationally recognized as a health priority for several decades (Cousens et al. 2010). Both voluntary programmes to identify carriers in high school students (Mitchell et al. 1996; Amato et al. 2014) and mandatory premarital testing Bozkurt 2007; Loukopoulos 2011; Alswaidi et al. 2012) have been successfully employed, with the reduction in affected births achieved via an informed decision made by carriers either regarding their choice of partner or early prenatal testing and selective termination of pregnancy (Giordano 2009; Miri et al. 2013).

The current population of India is estimated to be 1.37 billion (PRB 2018), equivalent to 18% of the global total and despite rapid urbanisation some 67% of people in India continue to live in rural areas. In terms of health indicators, the rural population significantly lags their urban counterparts, as reflected by the rural infant mortality rate of 38/1000 live births compared with 23/1000 in towns and cities (RGI 2017). Further, differences within the country, both in terms of the prevalence and causes of diseases across the 29 states, are highlighted in the recent comprehensive report on the burden of disease at the individual state level. The report also confirms a significant overall shift from communicable to non-communicable diseases, with congenital defects listed among the 20 commonest causes of disease listed (India State-Level Disease Burden Initiative Collaborators 2017). The genetic diversity of the Indian population, with large numbers of endogamous ethnic, geographical, religious and social groupings each with extended, unbroken genealogical histories, adds to the complexity of the prevalence and burden of genetic disorders (Basu et al. 2016; Nakatsuka et al. 2017), as observed in regional and caste-specific analyses of mutation data on haemoglobinopathies (Sinha et al. 2009; Black et al. 2010; Trehan et al. 2015).

My Aim and Objectives: (Hemoglobinopathies in India—Clinical and Laboratory Aspects**)**

1. To study the different clinical patterns of hemoglobinopathies diagnosed by high-performance liquid chromatography (HPLC) in India population.

2. This pilot study is aimed to find the prevalence of hemoglobinopathies in India and identify the change in the demographic profile.

Data collection and laboratory Analytical Methods:

1. Sysmex XN9000 Hematology 6 part analyzer 2. Biorad variant II (CE HPLC) 3. Sebia CE

Study design & Statistical resource :

Expected outcome and research recommendation:

The Government of India is presently engaged in the implementation of a prevention and control programme for two major forms of haemoglobinopathies, thalassaemia major and sickle cell disease, with guidelines for their prevention and management formulated under the National Health Mission. Based on projections for the population up to the year 2026, the annual blood requirement for treatment will increase to 9.24 million units, together with an 86% increase in budgetary requirements which then would account for over 19% of the current National Health Budget. To avert a public health crisis there is an urgent need to fully implement the prevention programme for haemoglobinopathies.

From the above projections and the assessment of available resources, it is evident that effective implementation of well-designed and efficiently implemented prevention strategies is urgently required. In India, screening for the detection of carriers in an annual cohort of adolescents in schools and/or of all pregnant women is the most feasible prevention strategy available for reducing the birth of children affected with thalassaemia. Besides improving genetic literacy in the population, an important side benefit of such screening programmes would be the capacity to derive accurate, representative data on the incidence of the major haemoglobinopathies at state and community levels which, in turn, would facilitate the compilation of comprehensive disease registries.

Reference:

Cousens NE, Gaff CL, Metcalfe SA, Delatycki MB (2010) Carrier screening for Beta-thalassaemia: a review of international practice. Eur J Hum Genet 18:1077–1083

Mitchell JJ, Capua A, Clow C, Scriver CR (1996) Twenty-year outcome analysis of genetic screening programs for Tay-Sachs and β-thalassemia disease carriers in high schools. Am J Hum Genet 59:793–798

Giordano PC (2009) Prospective and retrospective primary prevention of hemoglobinopathies in multiethnic societies. Clin Biochem 42:

Sujata Sinha etal (2020). Haemoglobinopathies in India: estimates of blood requirement and treatment costs for the decade 2017–2026, J Community 40 Genet, 11:39–45