LETTER TO THE EDITOR

The future of Inherited Hemoglobin Disorders- Gene Therapy

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Dear Editor, Inherited haemoglobin disorders - thalassemia and sickle cell disorders - are common worldwide. Strategies to prevent the incidence of these majorly debilitating disorders include cost-effective screening at population level, carrier detection, and pre-marriage and prenatal counselling. According to World Health Organization (WHO), over 50,000 babies are born with severe beta-thalassemia major (BTM). The burden of more than 80% of these births fall on developing countries.[1]

Muslim countries like Pakistan, Saudi Arab, Lebanon, and Iran have come a long way from allowing only premarital screening (and disallowing marriage) to making prenatal diagnosis and abortion legal after religious reinterpretation on the grounds of "medical illness".[2] However, the situation is still not as controlled as it was expected to be. In Pakistan, the incidence of carrier state is 5-7% where around 5,000-9,000 babies are born with beta-thalassemia major each year[3]. BTM has devastating impacts on the day to day life, quality of life, and health outcomes. It is associated with major cardiac, endocrinologic, and infectious morbidity and mortality. All patients of BTM are dependent on periodic blood transfusions and regular iron chelation. A BTM patient is vulnerable to complications if left untreated and even when treated.[2] While this treatment has severe morbid effects on the quality of life as well as the life span, it is not at all easy financially as well. According to an Indian study, the average annual thalassemia treatment of one patient is USD 1135 (range: 629-2300 USD).[4] This translates to an average of 171,907 PKR (range: 95,268-348,358 PKR)

With research and advancements in the field of genetics, allogenic haematopoietic stem cell transplantation and gene therapy are established as curative approaches for BTM. These emerging techniques can efficiently reduce the necessity of regular blood transfusions and will also cut down the complications and mortality rate associated with it. BTM poses a substantial burden on the health care system of Pakistan. According to a report, Pakistan initiated stem cell therapy in 1995. Since then only 8 stem cell transplantation centres have been established in three main cities of the country. These centres performed a total of 1,851 haematopoietic stem cell transplantation (HSCT) in two years (2015 to 2017) with beta thalassemia being the number one indication (n=603). Still the number of patients treated are very few as compared to the existing burden of the disease.[5] Gene therapy is a very old technique with 90% survival rate, but its application is very limited due to its cost. One way of countering this hurdle is to introduce partnerships between developed and developing countries (north-south/south-south partnerships) as acknowledged in the WHO-TIF (Thalassemia International Federation) meeting held in 2007. Such partnerships have proven to be very cost effective in treatment of thalassemia.[6] Since Pakistan maintains strong economic ties with China, which is already emerging as a pioneer in this field of medicine, this partnership can be adopted by these two countries in curtailing the burden of BTM.[7]

Pakistan is already working to tackle various preventable diseases like RTIs, Diarrhoea and improving the overall sanitation situation, this increases the overall life expectancy of children which indirectly increases the survival rate of children suffering from haemoglobinopathies. Thus prolonging the already existing duration of treatment. Moreover, preventable diseases can be managed conservatively, Beta thalassemia requires a multidisciplinary approach for its management. Pakistan already has a high carrier rate, posing significant threat of transmission in the next generation. Therefore BTM requires equal consideration as other preventable diseases.[3]

Support from the government to develop health centres with appropriate efficient control programmes focusing on prevention of disease like in Iran and Sardinia can decrease the burden of BTM immensely.[3] Therefore, we believe that switching to newer approaches to the disease and prevention are the two only ways forward.

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