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Editorial

Prevention of Beta Thalassemia in Pakistan

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Beta Thalassemia is the most common genetic disorder with an autosomal recessive inheritance pattern that results from reduced synthesis of β Globin chain. About 3% of the world's population carries the genes for Beta Thalassemia and it is estimated that every year about 60000 thalassemic babies are born all over the world.[1] Carrier rate in Pakistan ranges between 5-8%, thus there are about 9.8 million carriers of the total population and around 5000 children are diagnosed each year with beta thalassemia in Pakistan.[2] Consanguinity is the main factor leading to high prevalence in Pakistan. There are 25,000 children registered with thalassemia federation of Pakistan however the actual figure is much higher which may be around one lac as a lot many are living in villages that are not registered with any thalassemia center.

This genetic disease presents with pallor and failure to thrive in first year of life, requiring regular blood transfusions throughout the life and resulting in iron overload and metabolic complications. Moreover this lifelong transfusion therapy along with iron chelation therapy puts an enormous financial burden on parents, society and blood banks. Bone marrow transplant is the only curative approach but it is unaffordable by majority and thus most of patients are getting sub-optimal treatment. According to WHO if for any disease, the birth rate of the affected infant exceeds 0.1/1000 an effective screening programme should be initiated. So for thalassemics too without preventive measures (to limit affected births), it could not be possible to offer optimal treatment to thalassemics. Thalassemia is a preventable disease but there is no national screening program in Pakistan, only a few NGOs are running the programmes in their own capacity.

The main preventive strategies for thalassemia include appropriate information of the disease and importance of screening through awareness programmes, screening and counselling of target families, screening of general population, premarital and prenatal screening. Public awareness about the disease in Pakistan is very poor. Even the parents of thalassemic children residing in Pakistan know very little about the disease. By far the most effective way for prevention of thalassemia is mass education through media (print/electronic), seminars, symposia, education and training of general practitioners, obstetricians, paediatricians, social workers etc. Moreover knowledge about the disease may be incorporated in the curriculum. In Australia and Canada education about genetic tests and screening is provided at school level. The national screening programme in school children of 14-18 years has been found very promising in Montreal and other Western countries, with dramatic fall in newly diagnosed cases of thalassemia. Ideally screening is recommended for whole population, but in countries with limited economic resources and where consanguineous marriages are common; extended family screening may be an acceptable strategy to start with a national prevention programme. Extended family screening means screening of all the family members of an index case and in Pakistan this concept was first given by Suhaib Ahmad. Carrier rate in these families vary between 31- 68% as compared to 5% in general population.[3,4]

Likewise premarital and prenatal screening have significantly reduced the prevalence of β Thalassemia in various countries as Italy, Greece, Cyprus, Iran, and Saudi Arabia with significant reduction in births of affected children. In Pakistan however labelling someone even thahssemia trait may be associated with various social problems as for example rendering an individual unfit as a suitable marriage partner. The stigma associated with being a carrier can only be reduced significantly through greater awareness and public education. Prenatal screening is available in Pakistan but is underutilized mainly due to lack of awareness, poor access and high cost. Premarital screening instead of prenatal screening and target family screening may be a better strategy for prevention of thalassemia. In our country knowledge about thalassemia screening and diagnosis is inadequate. In Arab countries and even in Iran a significant reduction in new cases of thalassemia has been reported as they have made screening for thalassemia mandatory before marriage and at antenatal visit. It is thus recommended that it should be included as baseline antenatal tests during the first trimester. If one of the partners is a known a known thalassemic, the other should be screened for thalassemia. Moreover sometimes concomitant iron deficiency may mask thalassemia trait and thus it is suggested that either Hb electrophoresis is repeated after correction of iron deficiency or PCR may be advised if early diagnosis is required.

Blood CP is the first and an important laboratory investigation in the identification of thalassemia carriers.[5] Therefore red cell parameters are usually assessed before going for further investigations In countries like Pakistan no

case of microcytic/hypochromic anemia with a high index of suspicion for beta thalassemia trait should be declared as normal on Hb-electrophoresis alone. In such cases, the possibility of co-existing iron deficiency should be ruled out by serum iron profile determination. Moreover in view of presence of silent mutations in our population (though rare), all the cases with suspicion of thalassemia trait should be subjected to PCR if Hb electrophoresis does not reveal raised HbA2 levels (seen in thalassemia trait).

If the couple is found to be carrier for thalassemia, the female should undergo chronic villous sampling (and then PCR) at 10-12 week of gestation to determine the status of fetus as in every pregnancy there is 25% chance that the baby will be thalassemia major, 50% thalassemia trait and 25% chance that baby will be normal. If baby is found to be a case of thalassemia major then termination of pregnancy should be offered to the couple, as this is now been practiced with permission by religious scholars. But this is still a big dilemma that in our country even many thalassemic families are not aware of it and do not opt for prenatal diagnosis (PND) of thalassemia in each pregnancy. PND was introduced in Pakistan in 1994 [6] and it is available in big cities like Karachi, Lahore, Rawalpindi and Multan. About 5000-7000 are born every year and total number of PND s performed in these 16 years is <3000. This indicates that 80-90% of carrier couples are not availing this facility. [7] Factors identified for this low utilization of services were lack of awareness, poor access, delay in seeking advice, religious beliefs and high cost [8]

Thalassemia is a major public health problem, though it is a preventable disease many of our neighbouring countries have been successful reduced the number of new cases by running effective screening programmes and have significantly reduced the number of affected children but we are far behind. In Pakistan various Non-Government organizations and societies are working for this noble issue under the umbrella of Pakistan Thalassemia Federation but at National level no such serious efforts have been observed. It is thus strongly recommended that efficient and well-organized National screening programmers should be started addressing this national issue, with an emphasis on screening in schools, colleges universities, screening before marriage, screening at first antenatal visit and chorionic villous sampling in known thalassemic couples and termination of pregnancy. Above all there is a need to educate general public and medical practitioners to create awareness about the preventive aspects of thalassemia both in urban and rural population in order to reduce the burden of disease in Pakistan, as all these strategies have shown promising results in other countries.

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