B-Thalassaemia: What We are Waiting For?

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B-Thalassaemia ia is a recessively inherited hemoglobin disorder with profound implications for individuals, families and health services[1]. Carrier couples have one iii four and two in four chance. in every pregnancy, of having a child with major and minor thalassaemia, respectively. If one of the parents is a carrier, then there is 50% chance of having a carrier child in each pregnancy. The estiniated birth of thalassaemia major in Pakistan is over 5000/year[2]. Also the prevalence of carriers is high 5,500,000 cases[3]. This is largely because of the fact that carriers are asymptomatic and family/community marriages are common in our society.

The only cure is bone marrow transplantation with a success rate of 50 to 80%[4]. The cost is above 100,000 dollars, The facility, when available in Pakistan, is expected to cost 20,000 dollars. Very few of us can afford this treatment. The alternative is regular blood transfusion and iron chelation. The acquisition of safe blood is an uphill task and cost of desferral is beyond the reach of niost families. The estimated average cost/child/year is 15,000 - 60,000 rupees. Complications are life long and troublesome. Life expectancy' in our country' is about 10—16 years in majority of the cases.

I Historically. thalassaemia major has been seen as a problem for Mediterranean populations. However, Government com in itment, concerted awareness campaigns and extensive funding has greatly reduced its birth prevalence in Mediterranean area[5]. For example in Cyprus there are now almost no new affected births[6].

B-Thalassaem ia is the niost common inherited disorder in Pakistan. Can we initiate prevention programs in our country? Government support would be needed to screen less than 20 years (about 50 million) population of the country and this would cost millions of dollars. The other option. which appears more feasible, is to start targeted prevention programs — starting from the family of affected child. The responsibility largely lies with NGO's who are providing treatment to thousands of thalassaemia niajor patients all over the country. The NGO's and few other relevant centers need to develop uniform strategies to control the propagation of thalassaernia. As a policy matter all the siblings, uncles, aunts and cousins of the affected child must be screened (Hb with absolute values). It would be better if all such centres avail the services of haematologists and have facility of basic screening test (CP analyzer). They should acquire the facility of Hb-electrophoresis or they should collaborate with standard labs to provide this test at nominal charges. The carriers he provided genetic counseling with insistence on no marriage with a carrier spouse. Couples at risk be stressed to go for prenatal diagnosis and termination of pregnancy (thalassaemia major fetus) for which strong medical and religious recommendations exist. Collaboration with Labs to provide prenatal diagnosis at reduced cost and family planning centres can give improved results. It is also important that affected family be asked to go for family planning if they already have or if they get one or two normal children.

Clearly, it is the responsibility of relevant centers and hematologists to make organized efforts for education / awareness of doctors, nurses and community at large. Establishing and supervising thalassaemia parents associations can be another step towards achieving the goals. It is also needed that pediatricians and gynaecologists are actively involved in targeted prevention programs. All standard labs be asked, through the platform of Pakistan association of Pathologists, to provide absolute values along with haemoglobin (no extra cost is involved). Meanwhile efforts be made at Government level for national prevention programs. Government

support be sought to have legislation for mandatory screening before marriage. Print and electronic media be utilized for widespread awareness. Schools, mosques. madarsas and local town/village committees be involved as almost 50% of the affected families are from rural areas. If would be worth while to incorporate thalassaem ia prevent ion programmes in primary healthy care and family planning programs in future.

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