

Thalassaemia - Hopes Amidst Despairs

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Thalassaemia is a genetic disease with a huge burden. In Pakistan every year approximately eight to ten thousands children are born with beta thalassaemia major and carrier frequency varies from 5 to 9%. World over effective measures can be witnessed to tackle this issue. Once a problem is discerned out then how to tackle it? Thalassaemia can be taken as a model disease where all the strata of society behaved in an upfront manner. From prevention to treatment and cure thalassaemia opens numerous vistas and avenues. It can be taken as one of the pioneer disease where specific genetic defects are delineated in an explicit manner, upto an extent that now even in Pakistan its genetic make-up is established in every race. It can be taken as a genetic disease where diagnostic algorithms were developed successfully, very early.

Thalassaemia is the disease in which preventive measures and strategies can be seen to be placed. Genetic counseling, carrier screening, extended family screening all are being advocated and efforts are there to educate the people. Legislations through assemblies are in pipeline for mandatory screening for thalassaemia carrier status, before marriage. At government level, Punjab Thalassaemia Prevention Programme (PTPP) has a complete strategy in whole of the province. PTPP has regional offices in all divisional headquarters. It is being strengthened by field officers, gynaecologists and pathologists. It is providing free thalassaemia screening services to premarital couples and families in which there is already a patient of thalassaemia (Extended family screening). Thalassaemia is one of the diseases in which prenatal diagnosis was introduced. It has a high success rate in thalassaemia prevention. PTPP offers free prenatal diagnosis services to effected couples. The centre in Rawalpindi, located in Holy family Hospital affiliated with Rawalpindi Medical University, is not just giving services areas adjacent to Rawalpindi, but people from KPK, Azad Kashmir, Gilgit Baltistan and Afghanistan are also catered at this centre.

Non government organizations (NGOs) play an important role in uplifting of human society. In health sector, NGOs working for thalassaemia can be taken as a roll model. In almost all the big cities philanthropists are going hand in hand for thalassaemia prevention and management. These NGOs have a broad spectrum of activities, i.e., awareness, seminars, screening, blood camps, blood transfusion services, iron chelation therapy and many others. Working of these NGOs is a case study that how community behaves once a problem is unearthed.

In our setup blood donation, in majority of cases, is directed-donations, where people donate blood for the relatives or people known to them. Voluntary blood donation is farless. But, what so ever voluntary blood donation is there primarily it is for thalassaemics. Blood donation camps at different educational institutions is a big respite for thalassaemics and is reflecting the response of our youth.

At the management side iron chelation therapy is a part and parcel of thalassaemia management, as it is required to remove too much toxic iron, which get accumulated in the body of these patients. Medical science witnessed a giant leap in the regard, from cumbersome eight to ten hours injectable iron chelation to oral dispersible tablets. It can be considered as one of the milestones in medical history. Bone marrow transplantation (BMT) is the only curative option for thalassaemia. Thalassaemia is the pioneer disease where BMT has promising results, with maximum, success in overall survival and disease free survival. In Pakistan different centers are providing this service, with a partial support from Pakistan Bait- Ul- Mal. Recently Children Hospital Lahore and Daw Medical University Karachi started this service free of cost, in government sector. All the aforementioned aspects and efforts can be taken as an evidence that despite all odds, even our country and society, has a potential and resilience to circumvent and control untoward happenings, which have a huge burden on our health system. It can assumed that choices reflect our hopes against fears