

From the Director's Desk



This year was a challenging year for the Institute. Our scientific and non scientific staff rose to the occasion and good research work which had been the hallmark of this Institute continued.

Several rare haemoglobin variants like Hb Agenogi (β 90 Glu \rightarrow Lys), Hb Hofu (β^{126} val – Glu) were found. A very large deletion in the β globin gene cluster (48.5 kb deletion) leading to HPFH-3 deletion was found in 7 cases. The efforts to unravel the molecular footprints in α and β globin gene cluster which leads to increased production of HbF on Hydroxymea therapy is continuing.

Demonstration of G6PD Namoru in South Indian tribes has added a new dimension to the history of human migration. This mutation which is a combination of a polymorphism in the background of African G6PD “A” mutation was found originally in the Island of Namoru. Finding the same mutation in South Indian tribes points to the development of this polymorphism in Indian soil as the African population was migrating to Australia via India in antiquity. A few unique mutations in Pyruvate Kinase gene were found during this year. The effect of this mutation on Pyruvate Kinase activity needs to be worked out in a structure activity relationship format.

We are refining every year our ability to detect foetal mutations for haemoglobinopathy from maternal blood. We expect that this non invasive method of prenatal diagnosis will become applicable in near future.

Interaction of severe factor VIII and IX deficiency in haemophilia patients with various coinherited thrombophilia genes had been of abiding interest of haemostasis department. This year in an extensive study there is a persuasive suggestion that co-inheritance of thrombophilia genes makes a severe factor VIII and IX deficiency clinically milder. A simple technique was developed to study the binding of vonWillebrand factor to recombinant factor VIII and using this technique it was demonstrated that approximately 10% of our vonWillebrand disease patients are of Normandy type.

Looking for mutations in factor IX gene in cases of factor IX deficiency has been very fruitful. One mutation Gly 60 Ser was found to be common among moderate and mild

factor IX deficiency patients from Gujarat. All the Gujarati patients having moderate or mild factor IX deficiency carry this mutation. A PCR based simple RFLP test has been developed in this Institute for easy recognition of this mutation. Mutation studies in factor X and factor VII have shown us some novel hitherto undescribed mutation in our patient. A genotype phenotype correlation is in progress.

FLT3 mutation causing tandem duplication has been found elsewhere to be associated with AML with poor prognosis. The study which is progressing in the Institute showed 14-15% of AML patients attending this Institute carry this mutation. Study on HLA B-27 and B7 related CREG's in seronegative spondarthritides is going on smoothly. A large number of patients have already been registered for the study. During this period 20 papers have been published in International Journals and 11 in National Journal.

Our scientists have taken active part in various national and international conferences. Dr. Vandana Pradhan, Technical Asst. was awarded Dr.S.S Mishra Memorial Award of NAMS, on 15th January 2006. She was also awarded with Traveling Fellowship Award of International Immunological Society to attend Advance Immunological course at AIIMS, New Delhi, 1-5 March 2006. This year Dr. Anish Nair and Dr. Manish Nema received their DM degree in Clinical Haematology in the first batch of the course jointly conducted by KEM Hospital and IIH. The Institute continued its service for haematological disorders. Prenatal diagnosis was given to more than 60 families at risk for haemophilia and more than 100 families at risk for β thalassemia major. Several of our staff members superannuated during the year.

Till now I have talked about the brighter side of the Institute i.e its achievements, but what are our challenges? Space has now become a big constraint for any further development of haematology programmes of this Institute. The MOU with KEM Hospital for building several floors above this building has yet to materialize, though we are making slow progress. Another area of worry is the diminishing manpower. Three of our officers position is vacant and needs to be filled up quickly. We also need much more scientific staff to carry out research work more efficiently and I hope in 11th plan the Institute will be granted a few more staff including Accounts and Administrative Officer to look after the Institutes administration and finance more efficiently.

It is the human resource which is the permanent asset of the Institute. In the penultimate year of Golden Jubilee Celebration of this Institute, I am proud to say that I have such an asset in this Institute which will make any Institute proud.