THALASSAEMIA AND HB VARIANTS:  
FROM THE LABORATORY TO THE COMMUNITY  

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Molecular characterisation of genetic disorders in a multi-racial population can be  
challenging due to the many combinations of mutations. Prenatal diagnosis is even  
more overwhelming as mutations in the parents need to be identified and the  
genotype of the foetus rapidly confirmed in affected families. It is estimated that  
7% of the world’s population are carriers of haemoglobin disorders. Thalassaemia is  
a public health problem in Malaysia where 3.5 % of the population are carriers of β–  
thalassaemia, particularly in the Malays and Chinese. Alpha-thalassaemia in the  
Malaysian Chinese results in the fatal condition, Hb Bart’s hydrops foetalis. The  
situation is further compounded by the:

1. Heterogeneous mutations present in Malaysia’s multi-racial population.
2. High frequency of compound heterozygosity (different mutations) in one  
   family.
3. Presence of clinical phenotypes that do not match with haematology results.
4. Association of thalassaemias with haemoglobin variants that produce  
different haematological results from the otherwise clear-cut thalassaemia  
disorders.

Molecular characterisation is the only method to confirm the thalassaemias and  
haemoglobin variants. Identification of mutations will allow prenatal diagnosis to  
be offered to couples at risk, who can then with genetic counselling make informed  
decisions with regards to affected pregnancies. Specific protocols for prenatal  
diagnosis have been established for each of the Malay, Chinese and Orang Asli  
groups. The protocols were developed to allow rapid analyses with high sensitivity  
and specificity. The protocols were carefully optimised to ensure simplicity such  
that laboratories with minimal molecular equipment and trained staff can accurately  
carry out prenatal diagnosis.

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