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Abstract

Abnormal Haemoglobins and Thalassaemia in Malta

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A new genetics programme for the understanding and control of haemoglobinopathies in the Maltese population was started last year in our department. The programme has the support of the University of Malta and the Malta Government Department of Health as well as the World Health Organisation (European Office) and of former colleagues in the Medical College of Georgia (Augusta, GA, U.S.A.). Past studies have shown that β-thalassaemia occurs frequently. In addition to the $^{A}\gamma^{T}$ polymorphism (Hb-F-Sardegna), there are two known γ globin variants, three α and four β globin variants which have been identified among Maltese people.

Laboratory resources include a core Laboratory of Biochemical Genetics for population testing and a separate Laboratory of Molecular Genetics for globin gene mapping, haplotyping and the identification of mutations through techniques based on the polymerase chain reaction. A new specialist clinic for improved co-ordination of long term health care and counselling of couples at risk has been established in the Medical School. Efforts to introduce appropriate educational and counselling tools that make informed consent meaningful, include the preparation and distribution of "information about" brochures, articles in the press, programmes on the media, and talks or lectures for public, professional and special interest audiences.

Heterozygotes, "couples at risk" and new patients are identified through Newborn Testing by isoelectric focussing (IEF) of cord blood cell lysates in our core laboratory. Newborn babies with decreased Hb A/Hb F ratio are recalled at six months of age. Additional testing employing standard protocols is done on pregnant ladies at the time of booking into the Obstetric service and on patients referred from other clinical services.

During 1989, 6,500 mothers and 5,000 newborn were tested with β-Thal resulting in 2.4% of the mothers. This value is lower than anticipated but is consistent with the number of homozygous patients known to us. A mild type of α^{+}-thalassaemia was found in two out of eighty newborn studied with gene mapping techniques. Neither Hb Bart's, nor Hb H have been identified. Hb F-Malta-I continues to be found in 1.4% of all newborn while two new β and one new α globin variants have been detected. A thorough evaluation of the testing programme in newborn is as yet premature.