Letter to Editor

Prospects of non-invasive prenatal diagnosis of ß-thalassaemia in Pakistan

Madam, ß-thalassaemia is a major health problem in Pakistan with approximately 5000 children being diagnosed each year in the country. To reduce the growing burden of the disease, provision of invasive prenatal diagnostic procedure to at risk couples, followed by termination of pregnancy is the only way to avoid the birth of affected infants. Most Pakistani couples at risk of having a child with ß-thalassaemia-major are in favor of termination of pregnancy for the condition, particularly if prenatal diagnosis and termination of pregnancy are offered in the first trimester. Unfortunately, the prenatal diagnostic procedure (Chorionic Villus Biopsy) in Pakistan is limited by its association with significant risk of foetal miscarriage of up to 7%.[1] As each time an at-risk couple expects a viable pregnancy, unfortunately, they remain victims of chance. Over and above this burden, the possibility of loosing an unaffected foetus through invasive procedure is hard to bear. Secondly the cost of processing of the sample which is estimated to be approximately Rs 17,160 (US $286), is beyond the reach of a common man and finally the availability of prenatal diagnostic service in only five centers across Pakistan.

Clearly, an alternative means to diagnose ß-thalassaemia in the first trimester is much needed. Chueng et al.[2] and Di Naro et al.[3] demonstrated the feasibility of prenatally diagnosing ß-thalassaemia non-invasively, by a novel approach of enriching fetal zeta globin containing nucleated red blood cells from maternal blood. In 1997, Lo and his group from Chinese University of Hong Kong made a major breakthrough by observing DNA of foetal origin in maternal circulation, normally forming 1-3% of all DNA in maternal serum or plasma. Recently, Lo and his group,[4] and Hahn and his team from University of Basel, Switzerland[5] have shown the proof-of-principle studies of diagnosing ß-thalassaemia non-invasively, by isolating cell-free foetal DNA from maternal plasma.

Although the results are encouraging, it could take a while before such a novel method is brought into clinical practice from the bench side. Collaboration between the scientists of the developed and developing country like Pakistan is the need of the hour and could be beneficial in bringing the technology closer to a clinical application. However, implementation of the technology will, of course, be dependent on the development of accurate and technically robust test. Other factors such as cost-effectiveness of the test, its likely integration into the existing prenatal care and the use of simple equipment are crucial to the acceptability of this novel technology in a developing country like Pakistan. Bringing this innovative non-invasive approach to the bedside could eliminate the risk of foetal loss and provide an additional option for high-risk couples in future.

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