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Editorial

Should Screening for Genetic Diseases be Integrated in Primary Health Care?

Screening for genetic diseases is a controversial service in primary health care (PHC). While screening would save lives and may reduce the costs of care in certain manageable diseases, the cost of screening itself and unavailability of treatment for other diseases may make screening unacceptable for other diseases or conditions. Integration of these screening programs to PHC is yet another controversial aspect. Introducing these into the service package of PHC should be accompanied with coverage of the full range of service from screening to confirmed diagnosis and should include referral, treatment and counseling. Otherwise introduction of screening without these further steps may only increase fear and may not have any effect on health indicators.

The introduction of screening for beta Thalassemia major (BTM)in Iran is a good case study to focus on how PHC could be involved in a sophisticated screening service at national level. The disease was once very common with a high burden. Considering this high burden BTM was considered as a priority in health system and the national program for control of thalassemia was started on 1992 as an integral part of primary health care (PHC) system [1]. The program on that time consisted of screening of couples before registration of marriage contract and genetic counseling to at risk couples. On 1997 the legal and clerical permission for abortion of affected fetus with BTM major were approved. This promoted the development of the national net work for prevention, screening, counseling and prenatal diagnosis (PND) further [2].

There were three stages of implementation of national program for control of BTM: screening of all couples at the time of marriage to detect carriers of gene and affording counseling and PND to at risk couples, extending the program to parents of known cases of BTM who were in reproductive age, screening of other couples aged less than 40 years who married before the start of national screening program.

The two initial stages now has gained wide acceptance with high coverage but the final stage could not reach such accomplishment.

Recent reports show efficacy of program in reducing the incidence of new cases by more than 20 folds [3].

Despite this great successes there are still births of infants with BTM nationwide. The cause of failure of program in preventing these new cases is mainly failure of stage three of the national program described above to cover the unscreened married couples. Other causes were late referral for PND, and errors in the in the healthcare [3].

This program is now active for more than two decades. Integration of this program to the existent PHC system and collaboration of other sectors specially notary public in advising for mandatory pre marriage screening were among key factors of success of this program.

This model could be considered in controlling of other genetic diseases as well. We have already started sharing experience in PHC with the most needy population in states with the hope of reducing disparities in health [4].

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