Editorial

Fetal screening; opportunities and challenges

Screening, in the medical field is defined as a program or strategy to survey population about a risk of health-threats by identifying at risk persons. Its goal is early detection for intervention and management and prevention of a disease and its complications. Therefore, a test or method of screening is necessary to have some certain characteristics as its sensitivity, specificity, repeatability, social acceptance by the people as well as feasibility, availability, case of interpretation and cost-effectiveness.

It is evident that the illness or the problem for which a continuous screening program is compiled, must be clinically significant, intervenable, preventable and treatable and its early detection must be possible. According to the citeria mentioned above, the screening program for assessing the fetal malformations and trisomic syndromes including trisomy of chromosome 21 (Down syndrome), trisomy of chromosome 18 (Edwards) syndrome , trisomy of chromosome 13 (Patu) syndrome, has been compiled and executed. Therefore, some opportunities, challenges, and common mistakes about the subject are considered in this article.

In the population screening programs, massive policy-making and cost-effectiveness calculations are critical issues. Without the accurate cost-effectiveness, cost-benefitness and cost utility analysis, health resources will be wasted and the health outcomes will not be achived. The sources are so important that the general policy program has also undergone changes. Giving an example in this case can be useful. After adding the fetal screening program for Down syndrome to the UK National Health Programs, the research indicated that a balance between the chromosomal abnormalities associated with maternal age and the established resources should exist. Hence, policymakers believed that the funds can cover pregnancies occuring over maternal age of 35 years and all pregnant mothers are not included. Therefore the age of 35 years old was selected as a borderline to perform the fetal screening thus the cut off for moms older than 35 years. Interestingly, the majority of screening consultants in Iran without the sufficient knowledge about the process, over emphasize on the age of older than 35 years. They unwantedly cause fear and presentiment and exert psychological pressure on mothers older than 35 years and their families. They even introduce the mothers with just a few days older than 35 years wih a high risk for fetal malformations.

Another mistake is related to the using novel technologies with very sensitive and expensive methods such as cell-free DNA(known as NIPT or NIFTY). Since Iranian people are welcoming to many modern methods, the health service provider centers and laboratories make advertisements encouraging people to use these methods without adequate explanations. Considering public benefit , most of them believe that cell-free DNA method must replace common biochemical marker screening methods for all pregnant women in the country, but this is not cost-benefit at the present time. On the other hand it should be mentioned that although cfDNA method have extremely high sensitivity, they are not diagnostic and have false positives for various reasons such as confined placental mosaicism (CPM) and so on. Therefore the right application and the proper on time use of the facilities with appropriate informed decision making besides, respecting people and providing new opportunities to health services for the Iranians especially in the tertiary centers, must be created.

The other notable points in this category are reporting some cases of diseases or syndromes brought up in different studies as an association with the results of screening tests (with lower sensitivity and specificity). However, they haven't been approved in international and national guidelines. It is necessary to know such reports which are not beneficiary for the patients. They also cause trouble for both physician and patient and cause anxiety, bewilderment imposing unnecessary expenses. This can also be problematic from the legal aspect. So the risk of overdiagnosis like misdiagnosis threatens screening methods and even diagnostic methods and both of them can cause harm.

The final point on this subject is the importance of the enrichment of the native data bank especially in population-based statistical methods. The accuracy of threshold and mean of median (MOM) defined in these methods whether for sonographic or laboratory (biochemical markers or cell-free DNA) work have high importance. Hence, without having correct threshold amounts (both are based on population and innate accuracy of the test), the accuracy of the results with acceptable false positive and false negative rates can not be certain.

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