Int. J. Curr. Res. Med. Sci. (2018). 4(2): 134-137

International Journal of Current Research in Medical Sciences

ISSN: 2454-5716

P-ISJN: A4372-3064, E -ISJN: A4372-3061

www.ijcrims.com

Review Article

Volume 4, Issue 2 -2018

DOI: http://dx.doi.org/10.22192/ijcrms.2018.04.02.020

The first trimester screening -A Review

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Abstract

The protocols that are most frequently used at present *include* nuchal translucency measurement by ultrasound and measuring two of the analytes existing in maternal serum. This screening is conducted in weeks 11-14.In the most common screening protocol, NT measurement and serum level of hCG and PAPP-A are combined with each other. By using this protocol, the diagnosis rate of Down syndrome in large scale prospective trial will be 79-87% (with the false positive rate of 5%). In comparison to week 13, if it is reduced, the diagnosis rate of trisomy 18 will be like Down syndrome, and the false positive rate will be only 0.5%.Quad test is the most frequently applied screening test of the second trimester for aneuploidy diagnosis. In general, if women do not start pregnancy cares until the second trimester, or if screening tests of the first trimester are not accessible, Quad test will be conducted as a single test. As it will be discussed later, if Quad test and screening tests of the first trimester are combined, the diagnosis rate of aneuploidy will be higher than the current rate.

Keywords: screening, first trimester

Introduction

The first trimester screening

The protocols that are most frequently used at present include nuchal translucency measurement by ultrasound and measuring two of the analytes existing in maternal serum. This screening is conducted in weeks 11-14 (1).

Nuchal translucency (NT)

Nuchal translucency includes the largest thickness of the subcutaneous translucency space that is located within skin and the soft tissue covering the cervical vertebrae. Nuchal translucency is measured on Sagittal plane when the crown-rump length is 38-84 millimeters.

The specific criteria of NT measurement have been listed in tables 3-10. NT value is expressed as a multiple of specific median for the gestational age and a similar median of serum markers used in the aneuploidy screening. Increased NT thickness is not considered an abnormality; however, it indicates an increased risk. Almost one thirds of the fetuses who has increased nuchal translucency will have a chromosomal abnormality half of which is Down syndrome (2).

Table 1. Some screening strategies of Down syndrome and their diagnosis rates

Diagnosis rate[a] (percent)	Analytes	Strategy
79-87	NT, PAPP-A, hCG, or free β-hCG	Screening in the first trimester
64-70	Only NT	NT
61-70	MSAFP, hCG, or free β-hCG, uE3	Triple test
74-81	MSAFP, hCG, or free β-hCG, uE3, inh	Quadral test (Quad)
94-96	The screening of the first trimester and Quad test (the results will be kept until the Quad test is completed)	Integrated screening
90-95	The screening of the first trimester and Quad test • After the screening of the first trimester, as many as 1% of the individuals are recommended to conduct the diagnostic test. • In 99% of the cases, the Quad test is conducted (the results will be kept until the Quad test is completed).	Step-by-step sequential screening
88-94	The screening of the first trimester and Quad test • After the screening of the first trimester, as many as 1% of the individuals are recommended to conduct the diagnostic test. • In 15% of the cases, the Quad test is conducted (the results will be kept until the Quad test is completed). • After the screening of the first trimester, in 84% of the cases, no other test is conducted and no analysis is measured (**massively parallel genomic sequencing)**	Conditional sequential screening
98		Fetal extracellular DNA measurement (in high-risk pregnancies)

a. Based on positive screening rate of 5%

Free β-hCG: the free subcategory of Hcg: human chorionic gonadotropin;inh: *dimeric alpha inhibin;*

MSAFP: maternal serum alpha-fetoprotein; NT: nuchal translucency; pregnancy-associated plasma protein A; Ue3: *unconjugated estriol (7).*

As it is indicated in table 1, as a single marker, NT identifies as many as 64-70% of fetuses suffering from Down syndrome; the false positive rate of this method is 5% and its maximum sensitivity is in week 11 (Malone, 2005 b). The risk associated with increased nuchal translucency is independent of the risk associated with serum analytes, and combining NT with the values of serum analytes will result in a significant improvement of aneuploidy diagnosis (3). Thus, NT is generally used as a single marker only in multiple pregnancies in which serum screening is not accessible or precise enough. The only exception is when NT value increases for 3 to 4 millimeters; it is unlikely that applying the measurements of serum analytes confirms that aneuploidy likelihood is normal. Thus, aggressive tests need to be recommended (4).

Increased NT is also associated with other aneuploidies, genetic syndromes, and congenital defects especially fetal cardiac abnormalities (5). For this very reason, if NT value is 3.5 millimeters and more, in addition to fetal karyotyping, the patient needs to undergo purposeful ultrasound (with or without fetal echocardiography)(6).

For the aneuploidy diagnosis to be precise and accurate, NT imaging and measurement need to be conducted with high accuracy.

Serum analytes

Two analytes that are used for aneuploidy screening in the first trimester include human chorionic gonadotropin (both in the complete form and as Free β-hCG) and pregnancy-associated plasma protein A (PAPP-A) in women having a fetus suffering from a Down syndrome. The free BhCG level is higher in the first trimester and is around 2.0 MoM. Moreover, PAPP-A is lower and is around 0.5 MoM.In 18 and 13 trisomies, the level of both analytes will reduce (8).If the gestational age is diagnosed correctly, by applying these serum markers (without using NT measurement), the diagnosis rate of the fetuses suffering from Down syndrome will be 67% and the false positive rate will be 5% (9). If these analytes of the first trimester are combined with one or two of the following, aneuploidy diagnosis will significantly increase: 1. NT measurement by using ultrasound; 2. The analytes of the second trimester. The latter form is referred to as serum combined screening.

In twin pregnancies, the serum level of free β-hCG and PAPP-A are approximately doubled in comparison to single pregnancies (10). Even by using special diagrams, the results of screening in normal di-chorionic twins tend to be normal. Thus, aneuploidy diagnosis rate is at least 15% less (11).

Combined screening test of the first trimester

In the most common screening protocol, NT measurement and serum level of hCG and PAPP-A are combined with each other. By using this protocol, the diagnosis rate of Down syndrome in large scale prospective trial will be 79-87% (with the false positive rate of 5%). In comparison to week 13, if it is reduced, the diagnosis rate of trisomy 18 will be like Down syndrome, and the false positive rate will be only 0.5% (12).

In Down syndrome, a fourth marker i.e.dimeric alpha inhibin will increase. The average value of this marker is 1.8 MoM (13). By adding dimeric inhibin to the other three markers, quadral test of Quad will be obtained in which the diagnosis rate is 80% for trisomy 21 and its false positive rate is 5% .Just like the screening of the first trimester, the diagnosis rate of aneuploidy is slightly lower in women who were younger than 35 years old at the time of delivery. However, *the diagnosis rate of* aneuploidy is slightly higher in women who were older than 35 years old at the time of delivery. If serum screening of the second trimester is applied in twin pregnancies, the diagnosis rate of aneuploidy will be significantly lower (14).

Quad test is the most frequently applied screening test of the second trimester for aneuploidy diagnosis.In general, if women do not start pregnancy cares until the second trimester, or if screening tests of the first trimester are not accessible, Quad test will be conducted as a single test. As it will be discussed later, if Quad test and screening tests of the first trimester are combined, the diagnosis rate of aneuploidy will be higher than the current rate (15).

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How to cite this article:
Maryam Nakhaee Moghadam, Marzie Davoodi, Raziyeh Behzadmehr. (2018). The first trimester screening -A Review. Int. J. Curr. Res. Med. Sci. 4(2): 134-137.
DOI:http://dx.doi.org/10.22192/ijcrms.2018.04.02.020