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Method of Risk Assessment Affects Acceptance Rate of Amniocentesis

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Risk assessment for aneuploidy is an integral part of modern obstetrics. However, patients might choose or reject an amniocentesis, based, not on the risk, but on the screening method used. We examined records of patients referred for evaluation of aneuploidy risk from January 1999 to December 2000. Patients found to have a risk above 1/250 were offered amniocentesis. Out of 555 patients found at risk, 336 (60.5%) accepted. Acceptance rate varied depending on risk factor identified: advanced maternal age 178/290 (61.4%), abnormal serum

screening 107/198 (54%), sonographic markers 35/48 (72.9%) or a previous child aneuploidy 16/19 (84.2%). The risk assessment method used influences significantly the amniocentesis acceptance rate and may affect sensitivity of testing.

Key words. Amniocentesis, Aneuploidy, Down syndrome, Alpha fetoprotein, Serum screening, Triple marker screening, Advanced maternal age, Sonographic markers.

Prenatal diagnosis of congenital anomalies is an integral part of modern obstetric management. Although diagnostic sensitivity of conditions such as neural tube defects may be as high as 100% with sonography (1), chromosomal anomalies, particularly trisomy 21, can only be detected in 40 to 60% of cases (2,3). Risk assessment is, thus, the mainstay of aneuploidy screening. Evaluation of multiple risk factors such as maternal age, history, biochemical screening results and second trimester ultrasonographic findings are used to establish individual risks for chromosomal anomalies. An estimated risk of 1/200-1/250, similar to that of a 35 year old mother, continues to be the most common parameter used to recommend an amniocentesis (4). However, patients may choose or decline a genetic amniocentesis, based, not necessarily on the calculated risk but, rather on the method used for risk assessment (sonography, biochemical screening, etc). This can create a potential bias in favor of some methods and may affect sensitivity of testing. This study was designed to evaluate if the method of risk assessment influences significantly the acceptance rate of amniocentesis in our Hispanic population.

Materials and Methods

We examined the records of all patients referred to our University Hospital high-risk prenatal clinic for estimation of risk of aneuploidy from January 1999 to December 2000. Patients found to have a risk of Down's syndrome greater than 1/250 were divided into one of 4 groups depending on the risk factor detected:

1. Abnormal serum screening (either a low alpha fetoprotein or triple marker screening).
2. Advanced maternal age (35 or older at time of delivery).
3. A marker identified on second trimester sonographic examination (echogenic intracardiac foci, short femur or humerus, echogenic bowel, nuchal thickening, or pyelectasis).
4. A previous child born with a chromosomal anomaly. Patients with more than one risk factor or a congenital anomaly found on sonography were excluded from analysis. Acceptance rates of amniocentesis were calculated for each group and statistical comparison of proportions was performed. The results of the amniocentesis or incidence of chromosomal anomalies were not part of this evaluation and were not considered.

Results

A total of 555 patients were identified to have a single risk factor with an associated Down's syndrome probability

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of 1/250 or more. These patients were evaluated, counseled and an amniocentesis offered. Of these, 336 patients (60.5%) accepted amniocentesis. Patients were then divided into groups according to the risk factor identified and the amniocentesis acceptance rates for each group were evaluated. Results are shown in table I.

Table I. Amniocentesis acceptance rates for each group studied*

Group	Patients	Accepted amniocentesis
Abnormal serum screening	198	107 (54%)
Maternal age 35 or older	290	178 (61.4%)
Detection of sonographic markers	48	35 (72.9%)
Previous child born with a chromosomal anomaly	19	16 (84.2%)
All groups	555	336 (60%)

*Comparison between groups showed these differences to be statistically significant (p < 0.007).

Of the 198 patients with an abnormal serum screening, 107 (54%) chose to undergo amniocentesis. In the group where maternal age of 35 or older was an indication, 178 of 290 (61.4%) elected amniocentesis. Of patients with identifiable sonographic markers, 35/48 (72.9%) elected amniocentesis. Finally, in the group consisting of patients with a previous child born with a chromosomal anomaly, 16 of 19 (84.2%) elected amniocentesis.

Discussion

In order for a Down's syndrome screening program to be successful, it must not only detect a group of patients that are at risk, but these patients would need to undergo amniocentesis as a diagnostic test. A low amniocentesis acceptance rate in the population would reduce the detection rate, increase the cost per detected patient and significantly reduce cost-effectiveness of the screening method. Thus, the method of risk assessment must be accompanied by a high amniocentesis acceptance rate. This study shows significant differences in our population between acceptance rates depending on the method used. Age and serum screening showed the lowest acceptance rates while sonographic identification of markers and a previous child with a chromosomal anomaly showed the highest. It is easy to understand why a patient with a previous child born with a chromosomal anomaly would choose diagnostic testing in her next pregnancy. However, the fact that patients in the sonographic marker group chose amniocentesis more frequently than patients in the advanced maternal age or serum screening group suggests that they may see these risk assessments differently. They may look at sonography as a more direct (and thus more accurate) method of evaluation while age estimation of

risk and serum screening may be seen as more indirect (and thus less accurate) methods. The results suggest that they are being influenced by the method of risk assessment more than the calculated risk itself.

A sonographic identification of renal pyelectasis in a fetus may only increase the risk of Down's syndrome minimally (5) while an abnormal triple marker serum screening may increase this risk by a factor of 4 (6). However, patients appear to be more inclined to choose an amniocentesis after a sonographic marker such as this one is found than after an abnormal serum screening that may reflect a higher risk of Down's syndrome.

This tendency creates a bias in favor of some methods and against others affecting the sensitivity of screening and must be taken into account when counseling these patients. Second trimester sonographic evaluations for identification of markers may be a better method of risk assessment in our Hispanic population since it is associated to a higher amniocentesis acceptance rate. Even if the method has a lower detection rate than triple marker screening, the fact that 72.9% of patients identified as at risk by this method accept an amniocentesis should make it more cost-effective than serum screening which is associated to only a 54% acceptance rate. The added benefits of sonographic evaluation in detecting other anomalies, dating pregnancy, identifying viability and multiple pregnancy, evaluating growth, amniotic fluid, and placental disorders would make this method even more attractive to health systems with severely limited funds such as those of most Hispanic populations.

Resumen

Evaluar el riesgo para desórdenes cromosómicos es parte integral de la obstetricia moderna. No obstante, las pacientes podrían estar aceptando o rechazando una amniocentesis basándose en el método de cernimiento y no en el riesgo. Examinamos todos los récords referidos para evaluar el riesgo de desórdenes cromosómicos de enero 1999 a diciembre 2000. A las pacientes con un riesgo de 1/250 o más se les ofreció una amniocentesis. De los 555 pacientes identificados con riesgo, 336 (60.5%) aceptaron una amniocentesis. La respuesta de las pacientes varió dependiendo del factor de riesgo identificado: edad maternal avanzada 178/290 (61.4%), prueba de sangre anormal 107/198 (54%), marcadores sonográficos 35/48 (72.9%) o tener un hijo con un desorden cromosómico 16/19 (84.2%). El método de cernimiento para riesgo de desórdenes cromosómicos significativamente influenció la decisión de las pacientes de aceptar o rechazar una amniocentesis. Esto a su vez, podría afectar la sensibilidad del estudio.

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