Evidence-based Case: Refusal Of Triple-marker Screening Testing

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Abstract

Triple-marker screening testing is a widely accepted and utilized prenatal test. Evidence-based case study investigation provided a way for practitioners to examine the evidence supporting the use of this prenatal test. From this investigation, practitioners can apply the information in their practice settings.

INTRODUCTION

The following evidence-based case explored the use of triple-screening marker testing in the antepartum woman. For this example, initials ensure client confidentiality.

CASE STUDY

CLIENT PRESENTATION

SL is a 21-year-old, gravida 3, para 2, Mexican-American Catholic in her 15th week of pregnancy. SL has no family history of birth defects, Down's Syndrome, or spina bifida. SL refuses to have a triple-marker screening drawn at this visit. Her rational was: "If my baby has a defect I couldn't have an abortion."

CLINICAL QUESTION

In this HMO, standard practice is for all pregnant women to receive a triple-marker screening test between 15-18 weeks of pregnancy. Ultrasounds are not routinely performed, and are only indicated when abnormal triple-marker screening values are reported. Amniocentesis is offered instead of triple-marker screening to women over 35 years. The question is how to council this woman?

EVIDENCE SUPPORTING USE OF TRIPLE-MARKER SCREENING TESTS

Multiple-marker screening for both fetal Down's Syndrome and neural tube defects (spina-bifida) includes maternal serum alpha-fetoprotein (MSAFP), unconjugated estriol (uE3), and human chorionic gonadotropin (hCG) (1). The purpose of the multiple-marker screening test is to optimize diagnosis and minimize unnecessary diagnostic procedures. MSAFP is used for screening only, and not for diagnosis. Lowered MSAFP levels in second-trimester was shown to be independent of maternal age for pregnancies affected with fetal trisomy 21 (1). For those pregnant women younger than 35, a lowered MSAFP was found in 25% of Down's Syndrome fetuses and a reduction in uE3 concentration in 27% of Down's Syndrome fetuses (1). Others have reported elevated hCG levels in pregnancies affected with fetal Down's Syndrome (2). Used in combination, all three markers decrease the risk of false-positives. Canick and Knight report that for a pregnant 20-year-old the rate of detection of fetal Down's syndrome with the triple screening marker is 40.4% with a 2.4% rate of false-positives (1, 2).

Smith and Hau pointed out that an ultrasound is expensive, time consuming to read, and subject to the variability of the analyzer, and the standard and resolution of the equipment (3). Wilkins-Haug explained use of triple-marker screening as both inexpensive and automated when compared to the use of ultra sound (4).

An elevated MSAFP level is associated with increased risk of neural tube defects. The most common forms of neural tube malformations include anencephaly, spina bifida, and myelomeningocele (4-6). Early identification of neural tube defects permits altering delivery care. It is safer for a baby with neural tube defect to have a cesarean delivery, as a vaginal delivery might cause damage to the external membranous sac (6).

EVIDENCE NOT SUPPORTING USE OF TRIPLE-MARKER SCREENING TESTS

Other information supports not using the triple-marker-screening test. Covington reported triple-marker test results

may be altered in women with primary placental defects, pre-eclampsia, or hypertension (5). Weiss stated this variability leads to the controversy of triple-marker screening and to the accuracy of the results, in the percent of "false-positives (6)." "False-positives" are not the sole problem, but the follow-up tests do carry risks. A follow-up amniocentesis carries a 1-2% rate of fetal loss (6). Other authors reported other elevated levels in triple-marker screening for women with missed abortion, incorrectly reported maternal weight, undetected diabetes, and multiple gestation (4, 5). Overestimation of gestational age is a common reason for an initial positive result with triple markers. Overestimation results in spuriously low MSAFP and uE3 and high hCG, which is associated with an increased risk. Lower levels of MSAFP and uE3 have been reported in fetal demise and fetal growth restriction. These risks have been shown independent of one another (4, 5).

WHO DECLINES ANTENATAL SERUM SCREENING?

Researchers have examined the characteristics of women who refuse prenatal genetic screening. For one group of 595 women in California, researchers described no significant association between number of previous pregnancies, number of spontaneous miscarriages, or number of live births with a woman's decision to accept or decline MSAFP (7). These researchers did find an association between ethnicity and test refusal. Spanish-speaking Latinas were almost twice as likely to refuse testing and women who had never terminated a pregnancy were also more likely to refuse testing (7).

The effects of religion and religiosity for the same sample were complex. Women raised Catholic or Protestant were equally likely to refuse testing. Both Catholic and Protestant European-American women who scored high on "observant of church teaching on reproduction," which included agreement with religious teachings on birth control, abortion, when life begins, and extra marital sex, were significantly more likely to decline MSAFP testing (7).

The last significant effect found by these researchers described a combined effect of ethnicity and acculturation in the case of Spanish-speaking Latinas (7). Those women scoring less on acculturation on the Marin Short Acculturation Scale were significantly more likely to refuse MSAFP testing (7).

Other researchers, analyzing 886 responses from the 1996 Louisville Metropolitan Survey, indicated that women who had the following characteristics were less likely to give a "no" response to genetic screening: currently married, politically conservative, had less accepting attitude toward abortion, lower income, a high score on a chance health locus of control, and a strong identification with their religion (8). Favorable beliefs toward abortion were strongly associated with a positive attitude toward seeking prenatal genetic testing (8).

GENETIC TESTING, ETHICAL ISSUES, AND NURSING IMPLICATIONS

Genetic testing raises a number of ethical issues. The cloning of the sheep Dolly and the new technologies available soon which allow for altering of DNA will change people's childbearing choices. Genetic testing today is about predictive and future risk. In genetic testing information is not provided regarding severity, duration, or impact of the genetic condition just about the risk or probability of a genetic problem occurring (9, 10). Additionally, gene detection has the potential for the undesired outcomes of stigmatization and of discrimination (11).

Clients may misunderstand or may be distressed with the findings from their genetic tests (5, 13). Covington and colleagues described the anxiety experienced by families who have received borderline results and described the discrepancies in views, values, and understanding experienced by family members on hearing the same information (5). Evans and colleagues (1988) found younger clients with abnormal maternal serum alpha-fetoprotein levels required more sensitivity in counseling than did advanced maternal age clients (13).

Prior to or after genetic screening, an advanced practice nurse (APN) can provide the client with appropriate information regarding the meanings associated with MSAFP screening risk factor probabilities. This information can help reduce client anxiety (11, 12). Another time when an APN can counsel clients regarding genetic testing is before conception. At this time possible risk factors, diagnostic evaluations, and the need for genetic testing can be explored (12). A family history of Down's Syndrome, a genetic disorder, or spina bifida may affect a client's receptivity for genetic counseling (5, 11).

APPLICATION OF EVIDENCE TO SL

SL was told that if the triple-marker screening was abnormal, it did not necessarily mean she had to have further testing, or that she had to terminate her pregnancy. The APN explained to SL that a positive probability for this test would

indicate she had a greater risk of having a child with a neural tube defect. The importance of the test results was that her delivery could be changed to a cesarean, providing a safer delivery for her child. SL was also told she would be supported in whichever decision she chose to make. She chose to have the test.

CONCLUSION

Evidence-based case study investigation is an excellent way for practitioners to examine current practice in light of a literature review. Through this practice investigation, clinical experts, journal research, and web-based resources were reviewed and findings applied to an actual client situation. This format could be implemented in numerous settings to promote synthesis and application of theoretical knowledge into clinical practice.

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