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BRIEF COMMUNICATION

Benefits and pitfalls of prenatal screening in a twin pregnancy

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Prenatal diagnostic testing for aneuploidy is recommended for gravidas of advanced maternal age (AMA) [1]. Screening tests are available when diagnostic testing is declined. Table 1 compares current screening options; as can be seen, fetal aneuploidy screening for AMA women with multiple gestations has inherent limitations. The following case report illustrates some difficulties of aneuploidy screening for AMA women with multiples.

A Caucasian couple was referred for genetic counseling for AMA in a dichorionic twin pregnancy at 13 weeks. Consanguinity was denied between the patient, a 38-year old G4P0030 of English, Welsh, and Estonian ancestry and her husband, a 43-year old of French Canadian and Irish ancestry. An age-related Down syndrome risk (DSR) of 1 in 97 was calculated [2]. First trimester sonographic nuchal translucency (NT) screening produced a DSR of 1 in 179 for Twin A and 1 in 164 for Twin B. Combined first trimester screening (FTS) revised the DSR to 1 in 1071. Both the patient and her husband were identified as cystic fibrosis (CF) carriers for the delta F508 mutation; Tay Sachs screening was declined. Second trimester sonogram detected isolated echogenic bowel in Twin B; risks of CF, trisomy,

Table 1 Comparison of first and second trimester screening in singleton vs. twin pregnancies for the detection of Down syndrome> [1,3]

Screening method	Detection rate for singletons	False positive rate	Detection rate for twins	False positive rate
First trimester screening	~90%	5%	~80%	5%
Second trimester maternal serum screening	67—76%	5%	~45%	5%
Ultrasound screening	50—90%	17—27%	50—90%	17—27%

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and infection were discussed. Amniocentesis was declined, and the pregnancy continued uneventfully. Following delivery, both twins were found to be homozygous for the CF mutation. Furthermore, Twin B's karyotype was trisomy 21.

It is imperative to recognize that genetic disorders are independent events. Although rare, two separate genetic conditions can occur simultaneously. When discussing genetic testing in a twin pregnancy, the options are not as straightforward as for a singleton pregnancy. These limitations must be communicated as well as the possibility that the various screening tests may present conflicting information, necessitating diagnostic testing [3].

Biochemical serum markers in twin pregnancies may produce confusing results. Abnormal protein levels produced by one fetus may be obscured by those produced by the other fetus(es) [3]. In our case, the reassuring revised FTS arose from apparently normal serum results due to the twin gestation.

As FTS becomes more popular, screening in multiples should become more dependable.

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