

The importance of family history risk assessment in the infertility setting

Amy Vance, MS, CGC, Bay Area Genetic Counseling, San Anselmo, CA

Christo Zouves, MD, Zouves Fertility Center, Daly City, CA

It is well-documented that three generation family histories are not taken as a routine part of physician visits. Although genetic counselors routinely obtain a pedigree, couples using assisted reproductive technology may meet with a genetic counselor only if they are considering prenatal diagnosis. The purpose of this study was to determine the number and type of risk factors identified in couples undergoing an infertility evaluation. Genetic counseling was provided for all patients (n=691) in one IVF center from January, 2003 until December, 2004. Three generation pedigrees were constructed, and risk assessment and appropriate testing was offered. Chart review was performed to categorize the risk factors. Of the 691 couples, 452 (65%) had an increased risk. Specifically, 23 individuals were carriers for cystic fibrosis, 5 for Tay Sachs, and 16 for a hemoglobinopathy. One hundred twelve had a family history of mental retardation, Down syndrome, or autism. Consanguinity was present in 5, maternal disease (epilepsy/lupus/diabetes) in 5, factor V leiden in 8, family history of early MI or stroke in 31. An increased risk for mental illness was seen in 79. An increased risk for cancer was identified in 75. A history of congenital heart defect in the patient or a first degree relative was identified in 29, chromosome anomaly (self or previous child) in 4, birth defect in 32, and 25 had a family history of a mendelian disorder. Family history risk assessment in an infertility setting allows couples to learn about their genetic risks prior to pregnancy. For some couples it provides information about the causes of their infertility. Armed with this knowledge, couples at risk of transmitting genetic conditions may choose to take full advantage of technology such as PGD. The standard model of providing these couples genetic counseling only at the time of prenatal diagnosis should be examined.