

Risks in pregnancy- real and imagined

By Philip Buchanan, PhD, FACMG, and Laura Springstead, RNC, BSN

Kathy has had a lot of anxiety about becoming pregnant ever since she lost her first pregnancy last year. While she longs for a baby, her mind has been riddled with worry about what will happen, especially whether the baby will be okay this time. Kathy's husband, Jim, has been encouraging Kathy to try and become pregnant again. He is very supportive and doesn't understand why Kathy is so worried. Kathy's grief counselor encourages her to see a genetic counselor. The grief counselor thinks maybe Kathy imagines her risks to be higher than they really are because of her anxiety. Jim says, "It's as if all she can think of are the negative things that are possible. I'm the exact opposite. I keep thinking the most likely outcome will be a good one."

Kathy and Jim tell the counselor that before their miscarriage her doctor suggested she consider blood chromosome analysis and fragile X DNA carrier testing because her brother has mental retardation. She declined at that time, but since then all sorts of things have happened to change Kathy's perception of her risks. Kathy's parents were both killed in a tragic accident this year so Kathy and her sister have shared the responsibility of caring for their brother, John, ever since. John had lived with his parents his entire life and the change in lifestyle causes him significant distress. John's unhappiness makes his sisters wonder if a group home would suit him better so that he could be around friends and feel more independent. Once adjusted to the group home, he loves it there and seems more confident and content. During visits to his sisters' homes, he seems better than he's ever been.

While John has not been tested for fragile X, a common cause of mental retardation, workers at the group home have suggested it as a likely diagnosis. John hates having his blood drawn so Kathy now wants to pursue testing to see if she is a carrier for fragile X, and what her risks will be for having an affected child. Kathy explains her feelings to the genetic counselor, "I don't think I would end my pregnancy if anything was found to be wrong, but I think a pregnancy would be a lot easier on me if I knew whether or not I could have children with fragile X. If the baby does have it, well then.... I guess I'll just use the information to prepare myself." The counselor explains that the need for peace of mind is the reason many couples request prenatal diagnosis and assures her it is her choice either way. Amniocentesis will be offered to Kathy regardless of her family history because she will be 35 years old this year. Amniocentesis is offered to any woman age 35 or older, and also for a variety of other reasons, such as a family history of fragile X.

After the counselor discusses all the issues of fragile X carrier screening with the couple, Kathy opts for testing. A week later the results are given to her by her physician. Kathy is found to be a carrier for what is referred to as a premutation for fragile X syndrome. During a follow up session the counselor explains to Kathy and Jim that women with premutations typically do not have symptoms of fragile X. Premutation carriers are at risk for having a baby with fragile X, especially if the child is male. They also appear to be at increased risk for premature ovarian failure and early menopause. Because of this, the counselor discusses with them the importance of not waiting too long to start a family. Kathy tells the counselor her mom experienced early menopause, which began at age 38.

Kathy feels better knowing the real risks and now has a plan for future pregnancies. She expresses to Jim that even though she knows she is a carrier for fragile X, she also knows more now about the actual risks versus all of the imagined ones she had in her mind before. She realizes that before testing she had perceived her risks as much greater than she does now. Now Jim feels that she has a more realistic perception of what the risks really are, and Kathy feels comforted that a prenatal test can tell one way or the other about fragile X in their future babies. Kathy tells Jim, "I know no test can predict exactly how our babies will turn out. But it is nice to know a test is out there that can help clarify the risks we do have. It makes me feel better about giving this pregnancy idea a good try now, knowing that we may have a limited amount of time to have children." Jim replies silently with a twinkle in his eye that lets Kathy know he's ready to try again too.

GeneCare Medical Genetics Center, in Chapel Hill, offers genetic counseling and laboratory services, including fragile X screening and prenatal diagnostic testing. For more information, please call (919) 942-0021 or 1-800-277-4363. Or visit our web site at www.genecare.com.