

A Testing Conundrum

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In prenatal genetic counseling, we often assume that the services and technologies we provide for patients are helpful; that test “x” will lead to result “A” or “B”, and provide additional information. Very early in my career, however, I learned how technology can sometimes create a cloud of confusion for patients and professionals alike.

In one particularly difficult case, a 30-year-old woman and her husband were referred to me for genetic counseling because a choroid plexus cyst (a cyst in the brain) was seen during her routine 20-week ultrasound. This can be a benign finding, but it is sometimes associated with a chromosome abnormality such as Down Syndrome. Therefore the patient was offered amniocentesis as a way to learn more information about the significance of this finding. Like many, this couple struggled with the decision of whether or not to do the amniocentesis given the associated risk of miscarriage; however, they decided to proceed. As the patient’s husband put it, the amniocentesis could provide them with “peace of mind either way.”

Unfortunately, that black or white answer never arrived. Instead we had to struggle with gray results; an unusual chromosome rearrangement that was present in some cells, but not in all. This result fell into that “disclaimer clause” that accompanies each amniocentesis counseling session, and reads something like the amniocentesis results may reveal unexpected chromosomal changes that may require further investigation. I could rule out one of the more common chromosome abnormalities associated with this finding, but I could not tell them exactly what this result meant.

Testing of the parental bloods created further confusion when the patient was also found to have the same chromosome rearrangement in some of her cells. In other words, the mother was a mosaic for this chromosome finding. This result created a short-lived moment of relief. The patient is healthy and has the same findings; therefore, the baby should be healthy, right? That sense of relief was soon dampened by the reality that this case still carried the uncertainty associated with all mosaic results. You never know what percentage of cells in the body have normal chromosomes and what percentage have the rearrangement. There was no guarantee that this baby would be normal like the patient. All the reported cases of this particular chromosome rearrangement were found in individuals who had it in every cell, and as a result were mentally retarded or developmentally delayed. There were no published cases similar to this one, therefore we had nothing to go on.

Our discussion focused on what we knew, and what questions remained about the significance of the chromosome findings. More sophisticated testing could be done, but would more information help, or take them further into the unknown? As they struggled to make a decision the couple came to realize that with all testing options available, I could not provide the concrete answer that they wanted about the health of their baby. In the end they decided to pursue one more study, which was normal. Although reassuring, there was still no guarantee. Today, they have a healthy and developmentally normal 28-month-old daughter.

From my viewpoint, as their genetic counselor, this was an extremely challenging and frustrating case. It illustrates how even “routine” tests like ultrasound during pregnancy can result in unexpected and ambiguous results and start the patient down an information and technology road with no clear answers. Like my patients, I had become used to providing an “A” or “B” result. This case provided a reality check – sometimes the answer is not “A” or “B”, or even “C.” Sometimes the answer is “I don’t know”, which is the most difficult answer, for me as a genetic counselor, and even more so for my patients.

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