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Cytogenetic analysis in various tissues of pregnancy loss135

To the Editor:

We have noticed that our success rate and abnormality rate for products of conceptions (POCs) have increased over the past several years. We recently surveyed five years of data. Although our numbers are small, we did obtain results similar to those recently reported by Menasha et al.,[1] in that these findings reflect a change in the type of specimens we receive and/or our preparation of these tissues.

Cytogenetic reports on 214 samples received during the period from January, 2000 through December, 2004 were categorized as fetal tissue (often umbilical cord), chorionic villi, or unspecified POC (unable to distinguish origin of tissue). Approximately 5% of samples were contaminated when received and almost 25% failed to grow. The average success rate varied by tissue with fetal tissue being the lowest at 60.6% and villi the highest at 95.2%. The overall abnormality rate was 50% for villi and about 30% for fetal tissues and unspecified POC.

Samples received during the first trimester were more often from villi and unspecified POC, while fetal tissues were from second and third trimester pregnancy losses. The majority of growth failures were from fetal tissues, many due to intrauterine fetal demise.

Abnormal karyotypes from fetal tissue were mainly from second and third trimester, and consisted of potentially viable conditions, whereas the majority of abnormal karyotypes from unspecified POC and villi in the first trimester were nonviable abnormalities. The trisomic conditions identified during first trimester included 3, 6, 9, 10, 12, 14, 15, 16, and 22. We also identified two cases with multiple aneuploidy; trisomies 3 and 15, and trisomies 2, 8, 17 and 20, neither of which have been previously reported.[1]

Our findings agree with Menasha et al.[1] that the type of tissue received by the cytogenetics laboratory is important for success of growth and karyotyping, and hence, the information that can be provided to couples. Any methodology that optimizes the viability of specimens analyzed in the cytogenetics laboratory would increase the amount of clinically useful information provided to clinicians, thereby increasing quality of care for the patient. Therefore, clinicians should be encouraged to submit chorionic villi whenever possible.

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Reference

1. Menasha J, Levy B, Hirschhorn K, Kardon NB. Incidence and spectrum of chromosome abnormalities in spontaneous abortions: new insights from a 12-year study. *Genet Med* 2005;7:251–263.