

Case description

An obstetrician from Hyderabad sent a sample of aborted tissue (missed abortion of primi) for karyotyping. The sample was collected in normal saline. Male partner attended genetic counseling session.

Genetic Counseling

Karyotyping test requires dividing cells. Missed abortion samples usually lack viable cells and mostly fail to grow in culture. Moreover POC is usually contaminated with bacteria resulting in culture failure. Genetic counselor therefore recommended POC Aneuploidy test by FISH which identifies aneuploidy of 8 chromosomes on non dividing cells of the products. The turnaround time of the test is (4-6 days) compared to karyotyping (2-3 wks). Although the test does not examine all 46 chromosomes, 80% of all chromosomal defects observed in pregnancy loss are covered.

FISH Aneuploidy Test on products

FISH Aneuploidy test was done for 9, 13,16,18, 21, 22, X,Y chromosomes on non dividing cells of the aborted material. Report was ready in 4 days. Trisomy 16 was identified. Rest of the chromosomes were normal. Trisomy 16 is one of the common findings in pregnancy losses and recurrence risk is low. Patients were reassured. No further testing was recommended.

