Zygosity Testing

Clinical Utility:
This testing is used to determine if multiple pregnancies consist of monozygotic or polyzygotic gestations and to determine gender. This may be useful in determining the risk to a fetus for potentially having an X-linked disorder or the need for a fetus to undergo genetic testing. This analysis may also be performed postnatally to determine if two individuals are monozygotic or dizygotic twins. It may be also used for specimen identification purposes.

Test Methods:
DNA is “fingerprinted” using a series of markers selected by the forensic community and also used for medical applications such as detecting chimerism after bone marrow transplantation. The markers are 11 simple tandem repeats (STRs), either tetranucleotide or pentanucleotide repeats, that are highly polymorphic. In addition, the test identifies the presence of X and Y chromosomes by detecting the X and Y versions of the AMEL gene. All markers are amplified by multiplex PCR using fluorescently labeled primers. The size of each amplicon is determined by capillary electrophoresis and indicates the number of repeats in each allele (typically 6-20) except for AMEL.