Triploidy is a rare chromosomal abnormality. Triploidy is the presence of an additional set of chromosomes in the cell for a total of 69 chromosomes rather than the normal 46 chromosomes per cell. The extra set of chromosomes originates either from the father or the mother during fertilization. Infants with triploidy usually are miscarried early in the pregnancy. If the pregnancy continues to term, the infant dies within the first days of life. A few affected individuals have been reported to have survived to adulthood, but suffered from developmental delay, learning difficulties, seizures, hearing loss and other abnormalities. Those that survive have mosaic triploidy, meaning that some cells have the normal number of 46 chromosomes and other cells have 69 chromosomes per cell. Infants affected with complete triploidy suffer from growth restriction and multiple birth defects. Triploidy is the presence of a complete additional set of chromosomes. The triplication of the chromosomes is caused by the fertilization of an egg by two sperms, or the fertilization of an egg by a sperm that has an extra set of chromosomes or by the fertilization of an egg that has an extra set of chromosomes by a normal sperm. This disorder does not run in families and is not associated with maternal or paternal age. Triploidy accounts for 1-3 percent of all pregnancies. The condition occurs slightly more often in males than females; it is estimated that 2/3 of triploid pregnancies are male. The presence of multiple major malformations, low amniotic fluid and/or growth restriction on fetal ultrasound during pregnancy raises the suspicion of triploidy. The diagnosis can be made during pregnancy by chromosome analysis (karyotyping) of cells obtained by amniocentesis or chorionic villus sampling (CVS). The diagnosis can be confirmed after birth by chromosome analysis of tissue (skin) obtained from the affected infant. Triploidy cannot be diagnosed by chromosome microarray testing. The accuracy of non-invasive prenatal testing using cell-free fetal (cff) DNA in the diagnosis of triploidy is still being studied. Abnormal levels of specific maternal blood proteins such as alpha-fetoprotein, human chorionic gonadotropin, estriol and pregnancy-assisted plasma protein-A have been associated with an increased risk for triploidy.