Chromosome Abnormalities
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Prenatal ultrasonography in the diagnosis of fetal chromosome abnormalities
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Chromosomally abnormal conceptuses result in a large number of children with birth defects, early pregnancy losses and stillbirths. Chromosome abnormalities occur in approximately 1 in 160 live births, most as a result of the common trisomies. At birth, the incidences are 1 in 800 for trisomy 21, 1 in 6,000 for trisomy 18 and 1 in 10,000 for trisomy 13. Not surprising, the prevalence in early pregnancy is even higher due to embryonic and fetal deaths that occur with advancing gestation. Over 50% of recognizable pregnancy losses are attributed to chromosome aneuploidy. Furthermore, 6-11% of all stillbirths and neonatal deaths are the result of chromosome abnormalities. In 2007, the American College of Obstetrics & Gynecology (ACOG) published new guidelines recommending that all women who sought prenatal care at a sufficiently early gestational age should be offered prenatal screening for aneuploidy, regardless of maternal age. Ultrasound in conjunction with various biochemical markers could be utilized in establishing a personalized, “adjusted” risk for aneuploidy in any given pregnancy. While not all birth defects are detectable by ultrasound, prenatal imaging is a powerful tool in the diagnosis of birth defects, including many chromosome abnormalities. It is this technology, in addition to adjunct techniques of MRI and echocardiography that will be the focus of this presentation.