**Chromosome Abnormalities**
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**Syndrome Review I: Common Trisomies and Sex Chromosome Variations**
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Chromosome abnormalities affect about 1 in 200 liveborn infants and are estimated to account for at least 15% of birth defects in this group. Numerical chromosome abnormalities (also referred to as aneuploidies) are the most common type of chromosome disorder and include trisomies (having three copies instead of the normal pair) and monosomies (having a single copy of the chromosome). Common trisomies are trisomy 21, the chromosome constitution seen in 95% of infants with Down syndrome, trisomy 18 and trisomy 13. Clinical features and common birth defects seen in these conditions will be discussed. Trisomy 8 is rarely seen in liveborns and generally with mosaicism for a normal cell line. Trisomy 9 is also occasionally seen in a liveborn infant but is very rare (less than 1 in 100,000). The reason that trisomies for other autosomes (non-sex chromosomes; pairs 1-22) are not seen in infants is that these are typically lethal early in gestation, resulting in miscarriage. Monosomy for an entire chromosome is almost always lethal, with the exception of the sex chromosomes (the X and Y chromosomes or gonosomes). Monosomy X results in the condition known as Turner syndrome. Other numerical abnormalities of the sex chromosomes include an extra X chromosome (known as Triple X syndrome in the female and Klinefelter syndrome in the male) and additional Y chromosomes in the male. With the exception of Turner syndrome, sex chromosome variations are rarely associated with birth defects.

The mechanism that leads to a numerical chromosome error is known as nondisjunction, whereby two members of a chromosome pair do not divide properly during reproductive cell division called meiosis, resulting in an egg or sperm cell missing or with an extra chromosome. Although maternal age has long been a factor known to increase the likelihood of this event, errors in chromosome segregation can occur at many stages of cell division and only recently has there been discovery of genetic, epigenetic and environmental factors that may play a role in this.