

"Penta X Syndrome is a rare chromosomal disorder that affects females. Females normally have two X chromosomes. However, in those with Penta X Syndrome, there are three additional (or a total of five) X chromosomes in the nuclei of body cells (pentasomy X). The condition is typically characterized by moderate to severe mental retardation, short stature, malformations of the skull and facial (craniofacial) region, and/or other physical abnormalities. Characteristic craniofacial malformations may include upslanting eyelid folds (palpebral fissures), a flat nasal bridge, malformed ears, a short neck with a low hairline, and/or other findings. Penta X Syndrome may also be characterized by abnormal deviation (clinodactyly) or permanent flexion (camptodactyly) of the ""pinkies"" or fifth fingers; heart and/or kidney defects; deficient development of the ovaries and uterus; and/or other physical findings. The disorder results from errors during the division of reproductive cells in one of the parents. Penta X Syndrome is a rare chromosomal disorder that affects only females. Since the syndrome was originally described in 1963 (Kesaree N), over 20 cases have been reported in the medical literature. Some females with Penta X Syndrome were originally thought to be affected by Down Syndrome due to the presence of certain features sometimes associated with the latter disorder. (For further information, please see the "Related Disorders" section of this report below.) Penta X Syndrome is diagnosed based upon thorough clinical examination; detection of characteristic physical findings; and chromosomal analysis that confirms the presence of three extra X chromosomes in body cells. In some instances, the abnormality may be detected before birth (prenatally) based on chromosomal analysis following certain procedures, such as amniocentesis or chorionic villus sampling (CVS). During amniocentesis, a sample of fluid that surrounds the developing fetus is removed and analyzed, while CVS involves the removal of tissue samples from a portion of the placenta."