

Chromosome 9, Partial Monosomy 9p is a rare chromosomal disorder in which there is deletion (monosomy) of a portion of the 9th chromosome. Characteristic symptoms and findings include mental retardation; distinctive malformations of the skull and facial (craniofacial) region, such as an abnormally shaped forehead (i.e., trigonocephaly), upwardly slanting eyelid folds (palpebral fissures), and unusually flat midfacial regions (midfacial hypoplasia); structural malformations of the heart (congenital heart defects); genital defects in affected males and females; and/or additional physical abnormalities. In most cases, Chromosome 9, Partial Monosomy 9p appears to result from spontaneous (de novo) errors very early in embryonic development that occur for unknown reasons (sporadically). In observed cases, Chromosome 9, Partial Monosomy 9p has appeared to affect females more frequently than males. Since the disorder was originally described, more than 100 cases have been reported in the medical literature.