

Chromosome 8, Monosomy 8p is a rare chromosomal disorder characterized by deletion (monosomy) of a portion of the eighth chromosome. Associated symptoms and findings may vary greatly in range and severity from case to case. However, common features include growth deficiency; mental retardation; malformations of the skull and facial (craniofacial) region, such as a small head (microcephaly) and vertical skin folds that may cover the eyes' inner corners (epicanthal folds); heart (cardiac) abnormalities; and/or genital defects in affected males. Additional craniofacial features may also be present that tend to become less apparent with age, such as a short, broad nose; a low, wide nasal bridge; and/or a small jaw (micrognathia). In most cases, Chromosome 8, Monosomy 8p appears to result from spontaneous (de novo) errors very early in embryonic development that occur for unknown reasons. Chromosome 8, Monosomy 8p appears to affect males and females in relatively equal numbers. Since the disorder was originally described in 1973, over 20 cases have been reported in the medical literature.