

Chromosome 4, Partial Trisomy Distal 4q is a rare chromosomal disorder in which a portion of the fourth chromosome appears three times (trisomy) rather than twice in cells of the body. Associated symptoms and findings may vary from case to case. However, common features include growth deficiency; mental retardation; distinctive malformations of the skull and facial (craniofacial) region, including an unusually small head (microcephaly), malformed ears, and a prominent nasal bridge; and/or defects of the hands and feet. In some cases, additional physical abnormalities may also be present, such as structural defects of the heart that are present at birth (congenital heart defects); genital abnormalities in affected males; urinary tract defects; and/or other findings. In most cases, the trisomy appears to result from a balanced chromosomal rearrangement in one of the parents; rarely, it is thought to arise from spontaneous (de novo) errors very early in embryonic development that occur for unknown reasons (sporadically). In observed cases, Chromosome 4, Partial Trisomy Distal 4q has appeared to affect males and females in relatively equal numbers. More than 60 cases have been reported in the medical literature.