

Chromosome 7, Partial Monosomy 7p is a rare chromosomal disorder characterized by deletion (monosomy) of a portion of the short arm (p) of chromosome 7 (7p). Associated symptoms and findings may be variable and may depend on the specific size and location of the deleted segment of 7p. However, in many cases, there is early closure of the fibrous joints (cranial sutures) between certain bones of the skull (craniosynostosis), resulting in an abnormally shaped head. For example, depending on the specific sutures involved, the forehead may appear unusually "triangular shaped" (trigonocephaly) or the head may seem abnormally long and narrow with the top pointed or conical (turricephaly). Affected infants and children may also have additional malformations of the skull and facial (craniofacial) region. Such abnormalities may include an unusually small head (microcephaly), closely or widely set eyes (ocular hypotelorism or hypertelorism), downslanting eyelid folds (palpebral fissures), and/or other findings. Chromosome 7, Partial Monosomy 7p is a rare chromosomal disorder that appears to affect males and females in relatively equal numbers. More than 30 cases have been reported in the medical literature. Additional chromosomal disorders may be characterized by symptoms and findings similar to those associated with Chromosome 7, Partial Monosomy 7p. Chromosomal testing is necessary to confirm the specific chromosomal abnormality present. (For further information on such disorders, choose the name of the specific chromosomal disorder in question or use "chromosome" as your search term in the Rare Disease Database.)