

Chromosome 4, Monosomy Distal 4q is a rare chromosomal disorder in which there is deletion (monosomy) of a portion of the 4th chromosome. Associated symptoms and findings may be variable, depending upon the specific length and location of the deleted portion of chromosome 4. However, characteristic features include growth deficiency after birth (postnatal growth retardation), varying degrees of mental retardation, malformations of the skull and facial (craniofacial) region, structural heart defects, abnormalities of the hands and feet, and/or other physical findings. Chromosome 4, Monosomy Distal 4q usually appears to result from spontaneous (de novo) errors very early during embryonic development that occur for unknown reasons (sporadically). Chromosome 4, Monosomy Distal 4q appears to affect males and females in relatively equal numbers. Partial deletion of chromosome 4q was originally reported in a child in 1967. Chromosome 4, Monosomy Distal 4q was proposed as a distinct chromosomal syndrome with characteristic symptoms and findings in 1979. More than 30 patients with the syndrome have been reported in the medical literature. Additional chromosomal disorders may be characterized by symptoms and findings similar to those associated with Chromosome 4, Monosomy Distal 4q. 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.)