

Chromosome 10, monosomy 10p is a rare chromosomal disorder in which the end (distal) portion of the short arm (p) of chromosome 10 is missing (deleted or monosomic). The range and severity of symptoms and findings may be variable, depending upon the exact size or location of the deletion on chromosome 10p. However, associated features often include severe intellectual disability; growth delays after birth (postnatal growth retardation); distinctive malformations of the skull and facial (craniofacial) region; a short neck; and/or structural defects of the heart that are present at birth (congenital heart defects). Several cases have also been reported in which affected individuals have some features of DiGeorge syndrome (DGS). DGS is a congenital disorder characterized by underdevelopment or absence of the thymus and parathyroid glands, potentially causing abnormalities of the immune system, deficient production of parathyroid hormone (hypoparathyroidism), a heart defect, and associated findings. In many cases, chromosome 10, monosomy 10p appears to occur spontaneously (de novo) for unknown reasons. Chromosome 10, monosomy 10p is typically evident at birth (congenital). Since the disorder was originally reported in the medical literature in 1970, over 46 cases have been recorded. In such observed cases, males appear to be more frequently affected than females.