

"Chromosome 18q- syndrome (also known as Chromosome 18, Monosomy 18q) is a rare chromosomal disorder in which there is deletion of part of the long arm (q) of chromosome 18. Associated symptoms and findings may vary greatly in range and severity from case to case. However, characteristic features include short stature; mental retardation; poor muscle tone (hypotonia); malformations of the hands and feet; and abnormalities of the skull and facial (craniofacial) region, such as a small head (microcephaly), a ""carp-shaped"" mouth, deeply set eyes, prominent ears, and/or unusually flat, underdeveloped midfacial regions (midfacial hypoplasia). Some affected individuals may also have visual abnormalities, hearing impairment, genital malformations, structural heart defects, and/or other physical abnormalities. Chromosome 18q- syndrome usually appears to result from spontaneous (de novo) errors very early during embryonic development that occur for unknown reasons (sporadically). Chromosome 18q- syndrome appears to affect females more frequently than males by a ratio of approximately three to two. Since the disorder was originally reported in the medical literature in 1964, more than 80 cases have been recorded."