

Chromosome 13, Partial Monosomy 13q is a rare chromosomal disorder in which a portion of the long arm (q) of chromosome 13 is missing (deleted or monosomic). The range and severity of symptoms may vary greatly, depending upon the exact size and location of the deletion on 13q. Chromosome 13, Partial Monosomy 13q is usually apparent at birth and may be characterized by low birth weight, malformations of the head and facial (craniofacial) area, abnormalities of the eyes, defects of the hands and/or feet, genital malformations in affected males, and/or additional physical abnormalities. Affected infants and children may also exhibit delays in the acquisition of skills requiring the coordination of mental and muscular activity (psychomotor retardation) as well as varying degrees of intellectual disability. In the majority of cases, Chromosome 13, Partial Monosomy 13q appears to occur randomly, for no apparent reason (sporadic). Chromosome 13, Partial Monosomy 13q appears to affect females slightly more frequently than males. Although rare, deletions involving chromosome 13q are among the most commonly observed monosomies. Since the disorder was originally reported in 1963, more than 125 cases have been recorded in the medical literature.