

Chromosome 6, Partial Trisomy 6q is an extremely rare chromosomal disorder in which a portion of the 6th chromosome (6q) is present three times (trisomy) rather than twice in cells of the body. Associated symptoms and findings may vary in range and severity from case to case. However, many affected infants and children have slow physical development (growth retardation); mental retardation; malformations of the skull and facial (craniofacial) region; an unusually short, webbed neck; abnormal bending (flexion) or extension of certain joints in fixed postures (joint contractures); and/or other physical abnormalities. In most cases, Chromosome 6, Partial Trisomy 6q has been the result of a balanced translocation in one of the parents. Chromosome 6, Partial Trisomy 6q is an extremely rare chromosomal disorder that appears to affect males and females equally. Approximately 30 cases have been reported in the medical literature. Additional chromosomal disorders may have features similar to those potentially associated with Chromosome 6, Partial Trisomy 6q. 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.)