

Chromosome 15, Distal Trisomy 15q is an extremely rare chromosomal disorder in which the end (distal) portion of the long arm (q) of the 15th chromosome (15q) appears three times (trisomy) rather than twice in cells of the body. The disorder is characterized by growth delays before and/or after birth (prenatal and/or postnatal growth retardation); mental retardation; and/or distinctive malformations of the head and facial (craniofacial) area. Additional abnormalities typically include an unusually short neck; malformations of the fingers and/or toes; abnormal sideways curvature of the spine (scoliosis) and/or other skeletal malformations; genital abnormalities, particularly in affected males; and/or, in some cases, heart (cardiac) defects. The range and severity of symptoms and physical findings may vary from case to case, depending upon the length and location of the duplicated portion of chromosome 15q. In most cases, Chromosome 15, Distal Trisomy 15q is due to a chromosomal balanced translocation in one of the parents. Chromosome 15, Distal Trisomy 15q is an extremely rare chromosomal disorder that is thought to affect males approximately twice as often as females. Since the disorder was originally described in the medical literature in 1974 (A. Fujimoto), more than 30 cases have been reported in the literature. The majority of symptoms and physical features associated with the disorder are apparent at birth. Other chromosomal disorders may be characterized by prenatal and postnatal growth retardation, mental retardation, distinctive craniofacial abnormalities, genital malformations, congenital heart defects, and/or other abnormalities similar to those potentially associated with Chromosome 15, Distal Trisomy 15q. 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.)