Chromosome 4, Trisomy 4p is a rare chromosomal disorder in which all or a portion of the short arm (p) of chromosome 4 appears three times (trisomy) rather than twice in cells of the body. Associated symptoms and physical findings may vary greatly in range and severity from case to case. Such variability may depend upon the specific length and location of the duplicated (trisomic) portion of chromosome 4p as well as other factors. However, many affected infants may have feeding and breathing difficulties, characteristic malformations of the head and facial (craniofacial) area, and abnormalities of the hands and feet. Additional features may include other skeletal defects, genital abnormalities in affected males, or heart (cardiac) defects. Trisomy 4p is also characterized by severe mental retardation.
Chromosome 4, Trisomy 4p appears to affect males and females in relatively equal numbers. Trisomy for the short arm of chromosome 4 was originally described in 1970 (Wilson MG) and delineated as a distinct clinical syndrome in 1977 (Gonzalez CH). More than 85 patients with the syndrome have been reported in the medical literature. Many chromosomal disorders may have features similar to those associated with Chromosome 4, Trisomy 4p. 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.)