

Trisomy 5p is a rare chromosomal disorder in which all or a portion of the short arm (p) of chromosome 5 (5p) appears three times (trisomy) rather than twice in cells of the body. Often the duplicated portion of 5p (trisomy) is due to a complex rearrangement involving other chromosomes. These individuals have a variable phenotype depending on which chromosome is involved, the size of the duplication and whether there is loss of material from the same or another chromosome. In observed cases, trisomy 5p has appeared to affect females slightly more often than males and affects all ethnic groups. More than 40 cases have been described since the original description by Lejeune in 1964. While the features for trisomy 5p appear consistent, the diagnosis cannot be made on clinical features alone. Many of the features seen in trisomy 5p can be seen in other genetic conditions or chromosomal abnormalities. (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.)