

Trisomy 18 syndrome is a rare chromosomal disorder in which all or a critical region of chromosome 18 appears three times (trisomy) rather than twice in cells of the body. In some cases, the chromosomal abnormality may be present in only a percentage of cells, whereas other cells contain the normal chromosomal pair (mosaicism). Trisomy 18 syndrome was originally known as Edwards syndrome, after one of the investigators (JH Edwards) who initially recognized the condition as a distinct disease entity in 1960. The syndrome appears to affect females more frequently than males by a ratio of approximately three or four to one. Large population surveys indicate that it occurs in about one in 5,000 to 7,000 live births. The frequency of trisomy 18 syndrome increases with advancing age of the mother. Reports indicate that the mean maternal age is 32.5 years.