

Chromosome 18 Ring is a rare disorder in which there is loss (deletion) of genetic material from one or both ends of the 18th chromosome and joining of the chromosomal ends to form a ring. Associated symptoms and findings may vary greatly in range and severity from case to case, depending upon the amount and location of lost genetic material and other factors. A ring may also be formed without the loss of any genetic material. However, many individuals with the disorder are affected by mental retardation; low muscle tone (hypotonia); growth retardation; repeated infections during the first years of life; and/or malformations of the skull and facial (craniofacial) region. Such craniofacial features often include an unusually small head (microcephaly), widely spaced eyes (ocular hypertelorism), and/or vertical skin folds that cover the eyes' inner corners (epicanthal folds). Chromosome 18 Ring is usually caused by spontaneous (de novo) errors very early in the development of the embryo that appear to occur randomly for unknown reasons (sporadically). Since Chromosome 18 Ring was originally described, more than 70 cases have been reported in the medical literature. In these observed cases, females appear to be affected slightly more often than males. The disorder is often detected at birth or during prenatal testing.