

Chromosome 22 Ring is a rare disorder characterized by abnormalities of the 22nd chromosome. Associated symptoms and findings may be extremely variable from case to case. However, the disorder is typically associated with moderate to severe mental retardation. Some affected individuals may also have relatively mild, nonspecific physical (i.e., dysplastic) features, whereas others may have more distinctive, potentially severe physical abnormalities. According to reports in the medical literature, common findings include diminished muscle tone (hypotonia) and motor incoordination; an unsteady manner of walking (gait); pronounced verbal delays; and/or certain malformations of the skull and facial (craniofacial) region. Such craniofacial abnormalities may include an unusually small head (microcephaly); vertical skin folds that may cover the eyes' inner corners (epicanthal folds); unusually large ears; and/or other malformations. Chromosome 22 Ring is usually caused by spontaneous or "de novo" errors very early in the development of the embryo that appear to occur randomly for unknown reasons. Based upon observed cases, Chromosome 22 Ring has appeared to affect females more frequently than males. Over 50 cases have been reported in the medical literature.