

Chromosome 14 Ring is a rare disorder that is characterized by abnormalities of the 14th chromosome. Affected infants and children typically have delays in the acquisition of skills that require the coordination of physical and mental activities (psychomotor delays), mental retardation, growth delays, and episodes of uncontrolled electrical activity in the brain (seizures). The disorder is also characterized by distinctive abnormalities of the head and facial (craniofacial) area. Such abnormalities may include an unusually small head (microcephaly) with a high forehead; an elongated face; widely spaced eyes (ocular hypertelorism); a thin upper lip; a flat nasal bridge with a prominent nasal tip; and large, low-set ears. Chromosome 14 Ring is an extremely rare chromosomal disorder that has appeared to affect males slightly more often than females. There have been over 40 cases reported in the medical literature. Additional chromosomal disorders may have features similar to those associated with Chromosome 14 Ring. 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.)