

Chromosome 3, Trisomy 3q2 is a rare chromosomal disorder in which a portion of the 3rd chromosome appears three times (trisomy) rather than twice in cells of the body. Associated symptoms and findings may be variable, depending upon the specific length and location of the duplicated (trisomic) portion of chromosome 3. However, many affected infants and children have developmental delays, mental retardation, and characteristic abnormalities of the head and facial (craniofacial) area, resulting in a distinctive facial appearance. Such craniofacial abnormalities may include a relatively short head (brachycephaly), widely spaced eyes (ocular hypertelorism), upwardly slanting eyelid folds (palpebral fissures), and a small nose with upturned nostrils (anteverted nares). Affected infants and children also tend to have long eyelashes; arched, bushy, well-defined eyebrows that grow together across the base of the nose (synophrys), an unusually low hairline on the forehead and the back of the neck; and generalized excessive hair growth (hirsutism). Chromosome 3, Trisomy 3q2 may also be characterized by eye (ocular) abnormalities, limb defects, structural heart malformations (congenital heart defects), or other physical features. Chromosome 3, Trisomy 3q2 appears to affect males and females in equal numbers. At least 50 cases have been reported in the medical literature. In some cases, the disorder may be suspected before birth (prenatally) based upon the results of specialized tests. (For further information, please see the "Standard Therapies" section of this report below.) The disorder is usually detected or confirmed at birth or during early infancy based upon characteristic symptoms and physical findings.