

Nance-Horan syndrome is a rare genetic disorder that may be evident at birth (congenital). It is primarily characterized by abnormalities of the teeth and clouding of the lens of the eyes (congenital cataracts), resulting in poor vision. Additional eye (ocular) abnormalities are also often present, such as unusual smallness of the front, clear portion of the eye through which light passes (microcornea) and involuntary, rapid, rhythmic eye movements (nystagmus). In some affected individuals, the disorder may also be associated with additional physical abnormalities and/or intellectual impairment. The range and severity of symptoms may vary greatly from one person to another, including among affected members of the same family. As discussed above, Nance-Horan syndrome is usually fully expressed in males only, although females who carry a single copy of the disease gene (heterozygotes) may develop certain features. The disorder is present at birth (congenital). Nance-Horan syndrome was described in 1974 by two research teams independent of each other. Fewer than 50 families (kindreds) have been described in the medical literature. The exact incidence of the disorder is unknown. Researchers believe that affected individuals may go unrecognized or misdiagnosed, making it difficult to determine the true frequency of Nance-Horan syndrome in the general population.