

Lenz Microphthalmia syndrome is an extremely rare inherited disorder characterized by abnormal smallness of one or both eyes (unilateral or bilateral microphthalmos) and/or droopy eyelids (blepharoptosis), resulting in visual impairment. In rare cases, affected infants may exhibit complete absence of the eyes (anophthalmia). Most affected infants also exhibit developmental delay and mental retardation, ranging from mild to severe. Additional physical abnormalities are often associated with this disorder such as an unusually small head (microcephaly) and/or malformations of the teeth, ears, and/or fingers and/or toes (digits). The range and severity of findings may vary from case to case. Lenz microphthalmia syndrome is an extremely rare inherited disorder that is fully expressed in males only and is apparent at birth. However, females who carry a single copy of the disease gene (heterozygous carriers) may exhibit some milder symptoms associated with the disorder. Approximately 12 affected males with the fully expressed disorder have been reported. The disorder was first described by Lenz in 1955.