

Ablepharon-Macrostomia Syndrome (AMS) is an extremely rare inherited disorder characterized by various physical abnormalities affecting the head and facial (craniofacial) area, the skin, the fingers, and the genitals. In addition, affected individuals may have malformations of the nipples and the abdominal wall. Infants and children with AMS may also experience delays in language development and, in some cases, mental retardation.

Ablepharon-Macrostomia Syndrome (AMS) is an extremely rare genetic disorder that is apparent at birth (congenital). The disorder was originally described in 1977 (McCarthy GT) in two unrelated male children. A few additional isolated cases have since been recorded in the medical literature. In addition, investigators have described familial AMS in the sister of a previously reported affected female whose father has more minor features of the syndrome.