

Simpson dysmorphia syndrome types 1 and 2 are two forms of a rare, X-linked recessive, inherited disorder characterized by unusually large fetuses (prenatal overgrowth) and unusually large babies (postnatal overgrowth). In addition, affected individuals have characteristic facial features, more than two nipples (super-numerary nipples), and multisystemic malformations that may vary from child to child. Chief among these are cardiac malformations, mild to moderate mental retardation, cleft palate, and more than the five fingers and/or toes (polydactyly). Simpson dysmorphia syndrome is apparent at birth (congenital). The syndrome has been diagnosed only in males, although the carrier females may display symptoms to varying degrees. Between 10 and 20 affected families with SDYS1 have been recorded in the medical literature. The variant SDYS2 has been recorded in 1 family.