

Yunis-Varon syndrome is a rare genetic multisystem disorder with defects affecting mostly the skeletal system, the nervous system, and ectodermal tissue (hair and teeth). It is characterized by large fontanelles, clavicular hypoplasia, characteristic facial features and/or abnormalities of fingers and toes. Characteristic features may include microcephaly, ear abnormalities, anteverted nares, midfacial hypoplasia, tented upper lip and small jaw (micrognathia), sparse or absent eyebrows and/or eyelashes. Abnormalities of the fingers and toes may include absence (aplasia) or underdevelopment (hypoplasia) of the fingers and toes. Most infants with this disorder experience severe feeding problems and respiratory difficulties. Some of these difficulties are related to the severe involvement of the central nervous system, with malformations of the brain affecting half of the children and hypotonia (low tone) affecting all of them. In addition, affected infants may have heart defects (e.g., abnormal enlargement of the heart muscle [hypertrophic cardiomyopathy]). Frequently, feeding problems, respiratory difficulties, and/or heart defects may result in life-threatening complications during infancy. Yunis-Varon syndrome is inherited as an autosomal recessive condition. Yunis-Varon syndrome is an extremely rare inherited disorder that affects males and females in equal numbers. 25 cases from 19 families have been reported since the disorder's initial description in the medical literature in 1980.