

Fountain syndrome is an extremely rare genetic multisystem disorder that is characterized by intellectual disability; abnormal swelling of the cheeks and lips due to the excessive accumulation of body fluids under the skin (subcutaneous) of the face (edema); skeletal abnormalities; and/or deafness due to malformation of a structure (cochlea) within the inner ear. The exact underlying cause of Fountain syndrome is unknown. The disorder is believed to be inherited as an autosomal recessive trait. Fountain syndrome is an extremely rare inherited disorder that is believed to affect males and females in equal numbers. Only several cases have been reported in the medical literature. The exact incidence or prevalence of the disorder is unknown. Because cases may go undiagnosed or misdiagnosed, determining the true frequency of Fountain syndrome in the general population is difficult.