

Ear-patella-short stature syndrome (EPS), also known as Meier-Gorlin syndrome, is a rare genetic disorder characterized by small ears (microtia), absent or small knee caps (patellae), and short stature. Additional findings may include various skeletal abnormalities, early feeding difficulties, and poor weight gain. In addition, characteristic features of the head and face may be present including a small mouth (microstomia), with full lips, small circumference of the head (microcephaly), and/or underdevelopment (hypoplasia) of the upper (maxillary) and/or lower (mandibular) jaw bones (micrognathia). EPS is thought to be inherited as an autosomal recessive genetic disorder. Since EPS was first described in 1959 only about thirty-five cases have been reported in the medical literature (June, 2005). Researchers believe the disorder is under-diagnosed making it difficult to determine its true frequency in the general population. EPS is believed to affect males and females in equal numbers.