

Weill Marchesani syndrome is a rare genetic disorder of connective tissue characterized by abnormalities of the lens of the eye, short stature, an unusually short, broad head (brachycephaly) and joint stiffness. The eye (ocular) abnormalities can include small round lenses (microspherophakia), abnormal position of the lens (ectopia lentis) nearsightedness (myopia) resulting from the abnormal shape of the eye and lens and eye disease that damages the optic nerve (glaucoma) that can lead to blindness. Heart defects are present in some affected individuals. Weill Marchesani syndrome follows autosomal recessive or autosomal dominant inheritance. Weill Marchesani syndrome is a very rare disorder. The prevalence has been estimated to be approximately 1 in 100,000.