

Oculocerebral Syndrome with Hypopigmentation is an extremely rare inherited disorder characterized by the lack of normal color (hypopigmentation) of the skin and hair and abnormalities of the central nervous system that affect the eyes and certain parts of the brain (oculocerebral). Physical findings at birth include unusually light skin color and silvery-gray hair. Abnormal findings associated with the central nervous system may include abnormal smallness of one or both eyes (microphthalmia); clouding (opacities) of the front, clear portion of the eye through which light passes (cornea); and/or rapid, involuntary eye movements (nystagmus). Additional symptoms that may develop during infancy include involuntary muscle contractions, associated loss of muscle function (spastic paraplegia), developmental delays, and/or mental retardation. Oculocerebral Syndrome with Hypopigmentation is believed to be inherited as an autosomal recessive genetic trait.