

"Setleis syndrome is an extremely rare inherited disorder that belongs to a group of diseases known as ectodermal dysplasias. Ectodermal dysplasias typically affect the hair, teeth, nails, and/or skin. Setleis syndrome is characterized by distinctive abnormalities of the facial area that may be apparent at birth (congenital). Most affected infants have multiple, scar-like, circular depressions on both temples (bitemporal). These marks closely resemble those made when forceps are used to assist delivery. In addition, affected infants may have puffy, wrinkled skin around the eyes (periorbital) and/or abnormalities of the eyelashes, eyebrows, and eyelids. Infants with Setleis syndrome may be missing eyelashes on both the upper and lower lids, or they may have multiple rows of lashes on the upper lids but none on the lower lids. In addition, in some cases, the bridge of the nose may appear flat, while the tip may appear unusually rounded (bulbous). Affected infants often have loose, excessive (redundant) skin, particularly in the area of the nose and the chin. Due to such facial abnormalities, infants with Setleis syndrome may have an aged and/or "leonine" (lion-like) appearance. The range and severity of symptoms may vary from case to case. Most cases of Setleis syndrome are thought to be inherited as an autosomal recessive genetic trait. Setleis syndrome is an extremely rare inherited disorder that, in theory, affects males and females in equal numbers. Approximately 20 cases have been reported in the medical literature. The majority of these cases have occurred in individuals from Puerto Rico. Setleis syndrome is usually diagnosed shortly after birth based upon a thorough clinical evaluation and identification of characteristic features, such as distinctive scar-like, circular depressions on both temples; an aged and/or "leonine" facial appearance; and characteristic abnormalities of the eyelashes, eyebrows, and eyelids. It is possible that microscopic examination of small samples of skin tissue (biopsy) from the temples may reveal abnormal thinning of the outer layer of the skin (epidermis) and absence of certain specialized structures normally located within the inner layer of the skin (e.g., sweat glands, sebaceous glands, hair follicles)."