

"Filippi syndrome is an extremely rare genetic disorder that may be apparent at birth (congenital). The disorder is characterized by an unusual facial appearance, abnormalities of the fingers and toes, and mild to severe mental retardation. Primary physical findings include growth delays, webbing or fusion (syndactyly) of certain fingers and toes, inward deviation or bending (clinodactyly) of the fifth fingers ("pinkies") and microcephaly, condition that indicates that the head circumference is smaller than would be expected for an infant's age and sex. Filippi syndrome is transmitted as an autosomal recessive trait. The diagnosis of Filippi syndrome may be made at birth or during early infancy based upon a thorough clinical evaluation and characteristic physical findings. Specialized testing, such as certain advanced imaging techniques, may also be conducted to detect or characterize particular findings that may be associated with the disorder."