Isolated split hand-split foot malformation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Isolated split hand-split foot malformation. ORPHA:2440

Split hand-split foot malformation (SHFM) refers to a spectrum of genetically and clinically heterogenous terminal limb defect (see this term) characterized by hypoplasia/ absence of central rays of the hands and feet (that can occur in one to all four digits), median clefts of the hands and/ or feet, aplasia and syndactyly, with a wide range of severity ranging from malformed central finger/ toe to a lobster claw-like appearance of the hands and feet. SHFM can be an isolated malformation or can be a feature in various syndromes (ADULT syndrome, EEC syndrome; see these terms). SHFM usually follows an autosomal dominant pattern of inheritance with incomplete penetrance, but autosomal recessive and rarely X-linked inheritance have also been reported.