

Split hand/split foot malformation (SHFM) is a genetic disorder characterized by the complete or partial absence of some fingers or toes, often combined with clefts in the hands or feet. There may also be the appearance of webbing between fingers or toes (syndactyly). This may give the hands and/or feet a claw-like appearance. Split hand/split foot malformation affects males and females equally. Frequency is estimated at one out of 18,000 newborns. The diagnosis of SHFM is based on physical features present at birth. X-rays may provide additional information about the skeletal anomalies. Molecular genetic testing by DNA analysis is available for SHFM4, caused by a mutation in the TP63 gene. Mutations in the TP63 gene are responsible for only about 10% of all cases of SHFM.