Split Hand Foot Malformation (SHFM) is a rare limb malformation occurring in 1/8500 - 1/25000 newborns. It occurs with variable expressivity.

Genotypes

- **SHFM type 1 - 7q21**: DLX5, DLX6, DSS1, Autosomal Dominant (reported in 1 family)
- **SHFM type 2 - Xq26**: FGF13, X-linked Recessive (reported in 1 family)
- **SHFM type 3 - 10q24**: HOX11, FGF8, Autosomal Dominant (20% of reported cases)
- **SHFM type 4 - 3q27**: TP63, Autosomal Dominant (10 - 16% reported cases)
- **SHFM type 5 - 2q13**: HOXD13, Autosomal Dominant
- **SHFM type 6 - 12q13**: WNT10B, Autosomal Recessive (reported in 3 families, 1 sporadic case)

Phenotype

Features include underdevelopment or absence of digits as well as fusions.

Genetic cause can be identified in 50% of cases. The risk of recurrence in offspring is 30-50%.

Diagnosis includes genetic counseling, karyotype, cytogenetic and molecular testing, human genome scan and Whole Exome Sequencing (WES).

Resources

- American college of medical genetics
- National Society of genetic counselors
- Genetic and Rare Diseases information center (GARD)

References

https://rarediseases.info.nih.gov/diseases/6319/split-hand-foot-malformation

Example