## Open Peer Review on Qeios

## MASA Syndrome

National Cancer Institute

## Source

National Cancer Institute. <u>MASA Syndrome</u>. NCI Thesaurus. Code C129930.

An X-linked recessive condition caused by mutation(s) in the L1CAM gene, encoding neural cell adhesion molecule L1. It is characterized by mental retardation, aphasia, shuffling gait and adducted thumbs.