## Open Peer Review on Qeios

## Roberts Syndrome

National Cancer Institute

## Source

National Cancer Institute. <u>Roberts Syndrome</u>. NCI Thesaurus. Code C126326.

A rare, autosomal recessive inherited syndrome caused by mutations in the ESCO2 gene. It is characterized by limb and facial abnormalities and slow growth. Intellectual impairment occurs in approximately half of the affected individuals.