Roberts-SC Phocomelia Syndrome

National Cancer Institute

Source

National Cancer Institute. Roberts-SC Phocomelia Syndrome. NCI Thesaurus. Code C4681.

A rare genetic syndrome with an autosomal recessive pattern of inheritance. It is caused by a mutation in the ESCO2 gene. Clinical signs at birth include multiple limb and facial abnormalities. It is considered to be a mild variant of Roberts syndrome.