Robinow Syndrome

National Cancer Institute

Source

National Cancer Institute. Robinow Syndrome. NCI Thesaurus. Code C85048.

A rare autosomal recessive or dominant inherited disorder. The autosomal recessive form is caused by mutations in the ROR2 gene. There is no causative mutation identified for the autosomal dominant form. It is manifested with short limbs, abnormal facial features, underdeveloped genitalia, and wedge-shaped vertebrae.