Open Peer Review on Qeios

X-Linked Cornelia De Lange Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>X-Linked Cornelia De Lange Syndrome</u>. NCI Thesaurus. Code C75485.

An X-linked inherited form of Cornelia De Lange syndrome caused by mutations in the SMC1A gene mapped to chromosome Xp11.22. Patients have a milder form of the syndrome compared to patients with the NIPBL gene mutation.