

Open Peer Review on Qeios

Townes-Brocks Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Townes-Brocks Syndrome</u>. NCI Thesaurus. Code C99085.

An autosomal dominant inherited syndrome caused by mutations in the SALL1 gene. Clinical manifestations include imperforate anus, malformations in the ears and hands, hearing loss, and kidney and heart abnormalities.

Qeios ID: 586GVM · https://doi.org/10.32388/586GVM