

[Open Peer Review on Qeios](#)

Distal 18q Deletion Syndrome

National Cancer Institute

Source

National Cancer Institute. *Distal 18q Deletion Syndrome*. NCI Thesaurus. Code C130986.

A contiguous gene deletion syndrome involving deletion of the distal portion of the long arm of chromosome 18. The clinically heterogenous condition is characterized by some or all of the following: growth hormone deficiency with resulting short stature; hand, foot, skull, facial, and genital anomalies; hypotonia; and developmental delay.