## Open Peer Review on Qeios

## Coffin-Lowry Syndrome

National Cancer Institute

## Source

National Cancer Institute. <u>Coffin-Lowry Syndrome</u>. NCI Thesaurus. Code C84643.

An inherited syndrome caused by mutations in the RPS6KA3 gene. It is characterized by mental retardation, developmental delay, microcephaly, short stature and kyphoscoliosis.