## Open Peer Review on Qeios

## Pancytopenia-developmental delay syndrome

## INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.* <u>*Pancytopenia-developmental delay syndrome.*</u> ORPHA:401764

Pancytopenia-developmental delay syndrome is a rare, genetic, hematologic disorder characterized by progressive trilineage bone marrow failure (with hypocellularity), developmental delay with learning disabilities, and microcephaly. Mild facial dysmorphism and hypotonia have also been reported.