Kostmann syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Kostmann syndrome. ORPHA:99749

Kostmann syndrome is a rare, severe, congenital neutropenia disorder characterized by a lack of mature neutrophils (absolute neutrophil counts less than 500 cells/mm3) associated with frequent, recurrent bacterial infections (e.g. otitis media, pneumonia, sinusitis, urinary tract infections, abscesses of skin and/or liver) and increased promyelocytes in the bone marrow. Periodontal disease, as well as neurological symptoms, such as cognitive impairment, severe neurodegeneration and epilepsy, have been reported in some patients.