Congenital neutropenia-myelofibrosis-nephromegaly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital neutropenia-myelofibrosis-nephromegaly syndrome. ORPHA:369852

Congenital neutropenia-myelofibrosis-nephromegaly syndrome is rare, genetic, primary immunodeficiency disorder characterized by severe congenital neutropenia, bone marrow fibrosis and neutrophil dysfunction which is refractory to granulocyte colony-stimulating factor, manifesting with life-threatening infections and/or deep-seated abscesses, hepato-/splenomegaly, thrombocytopenia, hypergammaglobulinemia, anemia with reticulocytosis and nephromegaly. Other reported features include osteosclerosis and neurological abnormalities (e.g. developmental delay, cortical blindness, hearing loss, thin corpus callosum or dysrhythima on EEG).