Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome. ORPHA:369861

Congenital sideroblastic anemia -B cell immunodeficiency- periodic fever-developmental delay syndrome is a form of constitutional sideroblastic anemia (see this term), characterized by severe microcytic anemia, B-cell lymphopenia , panhypogammaglobulinemia and variable neurodegeneration. The disease presents in infancy with recurrent febrile illnesses, gastrointestinal disturbances, developmental delay, seizures, ataxia and sensorineural deafness. Most patients require regular blood transfusion, iron chelation, and intravenous immunoglobulin (IVIG) replacement. Stem cell transplantation has been reported to be successful.