Congenital Amegakaryocytic Thrombocytopenia

Alternative Names
CAT
CAMT

WHO International Classification of Diseases
Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism

OMIM Number
604498

Mode of Inheritance
Autosomal recessive

Gene Map Locus
1p34

Description
Congenital amegakaryocytic thrombocytopenia (CAMT) is a rare disease characterized by a severe hypomegakaryocytic thrombocytopenia (absence of megakaryocytes in the bone marrow) during the first years of life that develops into a pancytopenia in later childhood, suggesting a general defect in hematopoiesis. Because of the severity of this disease, it is usually recognized shortly after birth. Currently, patients with congenital amegakaryocytic thrombocytopenia can only be cured by bone marrow transplantation.

Molecular Genetics
Congenital amegakaryocytic thrombocytopenia is caused by mutations that affect the major platelet growth factor receptor and usually worsens over time until no cells are made in the bone marrow (aplastic anemia). In addition, the disease can also be caused by mutations in myeloproliferative leukemia virus oncogene (MPL).

Epidemiology in the Arab World

Saudi Arabia
Al-Ahmari et al. (2004) performed stem cell transplantation (SCT) from HLA-matched related donors in five patients with confirmed congenital amegakaryocytic thrombocytopenia (CAT). The median age at SCT was 3.2 years (range, 0.4-5 years). Conditioning regimen consisted of busulfan (BU) and cyclophosphamide (CY). Antithymocyte globulin (ATG) was also given; graft-versus-host disease (GVHD) prophylaxis was with cyclosporine and methotrexate. Four of the patients engrafted survived and became transfusion independent with a median follow up time of 30 months (range, 16-45 months). One patient failed to engraft and underwent a second SCT four months later but died of respiratory failure.

United Arab Emirates
Revesz et al. (1993) reviewed the occurrence of rare congenital forms of aplastic anemias in the United Arab Emirates and concluded that amegakaryocytic thrombocytopenia occurs exceptionally rarely in the population.

References

Contributors
Ghazi O. Tadmouri: 9.5.2005