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Cyclic or Congenital Neutropenia: Phenotypic Variations of Blood Neutrophil Counts in Patients with the Same ELA2 Mutation and Same Paternal Haplotype: Evidence for Phenotype Determination by Modifying Genes

Talia Pindyck
University of Massachusetts Medical School

Talia Pindyck, Class of 2008
Pediatrics Department

Cyclic or Congenital Neutropenia: Phenotypic Variations of Blood Neutrophil Counts in Patients with the same ELA2 Mutation and Same Paternal Haplotype: Evidence for Phenotype Determination by Modifying Genes

Talia N. Pindyck, BA[1]; Laurence A. Boxer, MD[2], David C. Dale, MD[3];
Audrey Anna Bolyard, RN[3]; Andrew A.G. Aprikyan, PhD[3]; Peter E. Newburger, MD[1]

[1] Department of Pediatrics, University of Massachusetts Medical School, Worcester, MA;
[2] Department of Pediatrics, University of Michigan Medical School, Ann Arbor, MI;
[3] Department of Medicine, University of Washington Medical School, Seattle, WA

Background: Cyclic neutropenia (CN) and severe congenital neutropenia (SCN) are disorders of neutrophil production that differ in severity and their responsiveness to treatment. Most cases of CN and many of SCN are due to mutations of the ELA2 gene.

Objectives: To determine the role of modifying genes in the ELA2 mutation-associated neutropenia phenotype.

Methods: Case Report

Results: We report on the progeny of a common donor used by an *in vitro* fertilization program whose affected offspring all had the same S97L ELA2 mutation.
The child presented was diagnosed as CN; seven other offspring were diagnosed as SCN.
Conclusion: This case suggests a role for modifying genes in determining the clinical phenotype of neutropenia due to ELA2 gene mutation. It also suggests that CN and SCN represent phenotypes on a disease spectrum.