Hematology-2013
OMICS Group Conferences
Accelerating Scientific Discovery
International Conference on
Hematology & Blood Disorders
September 23-25, 2013 Raleigh, USA

Emerging role of genetic studies in diagnosis of hemoglobin and red cell disorders

Mudra Kohli Kumar
University of South Florida, USA

Abstract

Abnormalities of hemoglobin may defy detection due to a relative inaccessibility of appropriate diagnostic testing alternatives, being electrophoretically silent or extreme instability rendering their presence to below detection level. In addition, vital information may be deemed to be lost if the patient receives red cell transfusion. The increasing ease of availability of DNA sequencing of globin genes has improved our ability to detect conditions that are presumed to be extremely rare. Genetic diagnosis may also be valuable in workup of gamma chain disorders, where the window for diagnosis by conventional means is relatively small. Confirmation of abnormality in the beta globin chain may be helpful to explore therapeutic interventions. We present 2 conditions whose diagnoses were made by pursuing genetic testing.

The first patient presented with cyanosis at birth. Preliminary studies suggested gamma chain abnormality, consistent with the improvement in clinical course as he grew older. Even though the level of fetal hemoglobin decreased to adult level, rendering it difficult to further characterize the abnormality, diagnosis of Hemoglobin F Cincinnati α2Gγ'241(C7)Phe→Ser was established by genetic sequencing of gamma chain (first ever description).

Genetic studies were pursued in mother and daughter with severe hemolytic anemia with DNA sequence analysis of the Beta globin gene identified Hemoglobin Manukau (HGVS:HBB:c.203T>G)-an extremely unstable hemoglobin, the second family in literature.

An astute clinician may benefit from being persistent and pursuing additional testing including molecular genetic characterization despite uninformative first line testing in cases where clinical suspicion remains high.

Biography

Mudra Kumar is an academic Pediatrician/Pediatric Hematologist with special interest in medical education. She has lived, studied and worked in three continents- Asia, Europe and America, giving her a unique and global perspective in education, practice and challenges in teaching, clinical practice, efficient utilization of resources and optimal delivery of healthcare Her special interests include red cell disorders and anemia: atypical presentations, uncommon and rare conditions. Apart from her involvement with the pediatric residency program, she also serves as Course Director and Clinical Integration Director for the second year medical students at USF, exploring novel and innovative directions in medical education.