Dyskeratosis Congenita

A 5 year old boy presented with history of progressively increasingly pallor and abnormality of nails for one year. On examination, the child had severe pallor with petechiae and purpura. There was dystrophy of nails in all the fingers and toes (***Fig.*1**), lacey reticulated pigmentation over chest (***Fig.*2**) and oral leukoplakia. There was no hepatosplenomegaly. Hemoglobin (Hb) was 2.0 g/dL, total leucocyte count 2450/cmm with a differential count of N19L80M1 and platelet count of 16,000/µL. General blood picture, bone marrow aspiration and biopsy were suggestive of hypoplastic anemia. In view of the typical physical and hematologic findings a diagnosis of Dyskeratosis congenita was made. The child was started on androgens.

Dyskeratosis congenita (DC) (also known as Zinssor – cole – Engmak syndrome) is an inherited bone marrow failure syndrome which presents with bone marrow failure and diagnostic triad of lacey reticulated pigmentation, dysplastic nails and oral leukoplakia. The diagnosis is often made in adulthood and most of the physical findings appear with increasing age. Treatment is initiated when Hb is <8g/dL, platelets <30,000/µl and absolute neutrophil count <500/µL. Androgen therapy may improve but not cure aplastic anemia. Stem cell transplantation is recommended if there is an HLA-matched sibling donor. There is an increased incidence of carcinomas particularly of the head and neck.

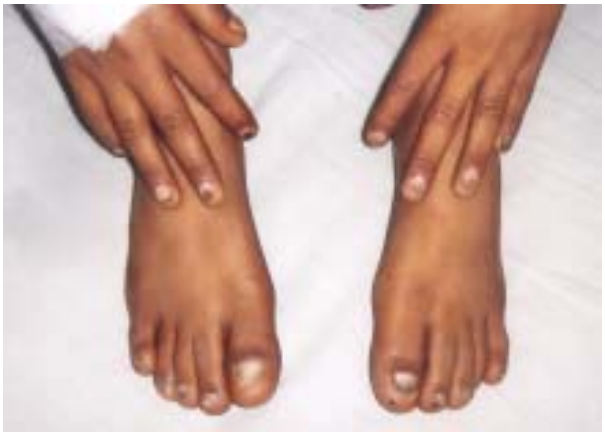

FIG. 1 *Dysplasia of nails.*

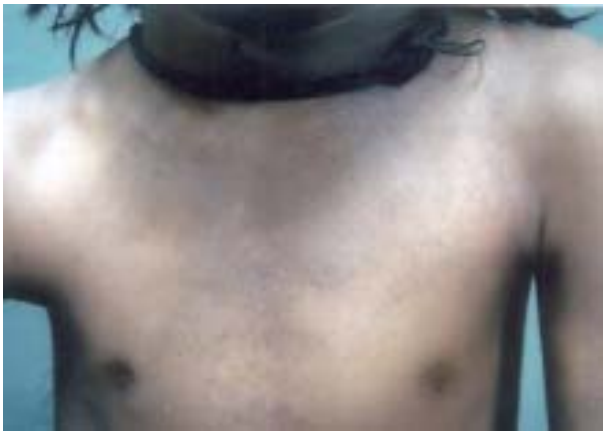

FIG. 2 *Lacey reticulated pigmentation.*

V Gupta,
BD Bhatia,
Department of Pediatrics,
Institute of Medical Sciences,
Banaras Hindu University,
Varanasi 221 005, India.
E-mail: vineetaguptabhu@gmail.com

BOOK REVIEWS

Manual of Pediatric Therapeutics
GRAEF JW, WOLFSDORF JI, GREENES DS
7th Edition. New Delhi: Lippincott Williams & Wilkins: 2008. Pages: 716.
Price: Rs. 495/-.

This is a very useful book for all categories of pediatricians, be it a naïve medical student stepping into a realm of pediatrics, practicing pediatrician or a pediatric consultant dealing with complex child health issues. It can rightly be termed a ready reckoner for management of not so familiar diseases. Lucid and crisp presentation, the updates and the precise format are its endearing features. 14 years wait for the new