

Pancytopenia in a young girl with skin lesions

S.K. Bhavya, N.P. Prakash, T.M. Anoop, N. Rakul*

Department of Medical Oncology, Regional Cancer Centre, Thiruvananthapuram, India,
*corresponding author: email: rakulnambiar@yahoo.com

CASE REPORT

A 14-year-old-girl presented with complaints of being easily fatigued and bleeding gums of two weeks' duration. She had a history of seizures in childhood. Physical examination demonstrated papular lesions on the nose, and cheeks consistent with facial angiofibromas (*figure 1*) There were ash-leaf hypomelanotic macules on the limbs (*figure 2*). No oral or periungual fibromas were seen. Examination of the cardiovascular and respiratory system was unremarkable. Her blood investigations revealed a haemoglobin of 4.59 mmol/l, elevated leucocyte count $4.93 \times 10^9/l$ with 78% abnormal cells and thrombocytopenia

$1.6 \times 10^9/l$. Bone marrow showed immature cells having irregular nuclei with folding and invagination (*figure 3*). The immature cells were peroxidase positive. Magnetic resonance imaging of the brain revealed subependymal nodules in the lateral ventricle (*figure 4*).

WHAT IS THE HAEMATOLOGICAL CONDITION CAUSING PANCYTOPENIA AND WHAT IS THE SYNDROME IN THIS PATIENT?

See page 44 for the answer to this photo quiz.

Figure 1. Papular lesions on the nose, and cheeks consistent with facial angiofibromas



Figure 2. Ash-leaf hypomelanotic macules on limbs



Figure 3. Bone marrow showed immature cells having irregular nuclei with folding and invagination

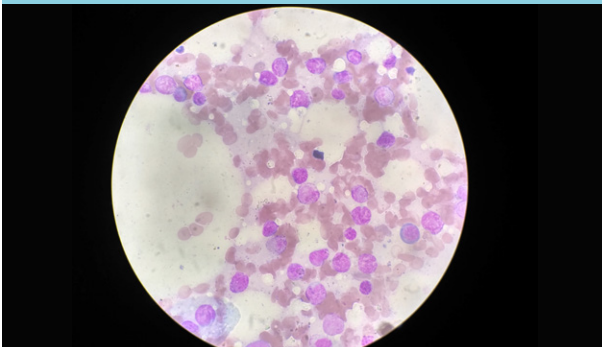


Figure 4. Magnetic resonance imaging of the brain demonstrated subependymal nodules in the lateral ventricle

