

occult blood. Screening test for Factor XIII levels was normal and platelet aggregation studies showed normal aggregation with ADP, AA, Collagen and Ristocetin.

It was explained to the parents that there was a discrepancy between their child's clinical profile and her investigation reports. In due course of time, the child revealed to the clinician and the parents that she had been applying her mother's liquid vermilion to fake bleeding. Thus, she was diagnosed as a case of Munchausen Syndrome. She was referred for psychiatric treatment, which the parents refused.

Munchausen Syndrome is an extreme form of factitious disorders, wherein the sufferer simulates illness, to assume patient role [1]. It is characterized by multiple outpatient visits or hospitalizations [2]. Typically, sufferers lie deliberately and may consume drugs like insulin, vitamins, warfarin to produce adverse effects like hypoglycemia, and bleeding symptoms [3]. It is uncommon in children and should be suspected if there is a discrepancy between symptoms and signs/investigation results.

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MEENU PANDEY AND *ASHU SAWHNEY
Jaypee Hospital, Sector 128, NOIDA, UP, India.
*dr. ashusawhney@gmail.com



FIG. 1 Patient showing factitious bleeding from the right eye.

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Identification of PML/RAR α Fusion Gene by RT-PCR Acute Promyelocytic Leukaemia

The genetic hallmark of acute promyelocytic leukemia (APL) is the balanced reciprocal chromosomal translocation of (15;17) leading to a fusion of the promyelocytic leukemia (PML) gene on chromosome 15 and the retinoic acid receptor- α (RAR α) gene on chromosome 17 [1]. Identification of PML/RAR α fusion gene by reverse transcriptase-polymerase chain reaction (RT-PCR) without evidence of t(15;17) both on conventional karyotype and fluorescence *in situ* hybridization (FISH) is rare [2,3].

An 8-year-old girl was admitted with the complaints of fever and generalized weakness for last 1½ month, and epistaxis and bleeding from gums for last 20 days. She had

moderate pallor and hepatosplenomegaly. Routine blood examination revealed hemoglobin 7.4 g/dL, total white cell count $9.8 \times 10^9/L$ and platelet count was $40 \times 10^9/L$. Peripheral blood smear showed 35% neoplastic promyelocytes with cytoplasmic hypergranulation. Coagulation profile revealed normal prothrombin time and activated partial thromboplastin time, but serum fibrinogen level was low. Bone marrow aspiration showed a hypercellular marrow with 66% neoplastic promyelocytes and presence of multiple Auer rods (faggot cells).

Cytochemical staining with myeloperoxidase was strongly positive. Child was diagnosed morphologically as AML-M3 (hypergranular variant). Immunophenotyping analysis of the bone marrow cells was consistent with APL. Conventional cytogenetic analysis showed 46, XY, isochromosome (17q). FISH analysis was negative for t(15;17). RT-PCR was positive for PML-RAR α fusion transcripts. She had completed both induction and consolidation phase of all-trans retinoic acid (ATRA) based chemotherapy regimen. Child responded dramatically and went into molecular remission.

Conventional cytogenetic analysis can identify reciprocal chromosomal translocation t(15;17) in upto 90% of cases with APL. The remaining 10% cases lacking t(15;17) stay associated with the cryptic insertion of the PML/RAR α fusion gene [4,5]. FISH analysis and RT-PCR are the valuable tools to identify the PML/RAR α hybrid transcript in a cytogenetically negative APL patient. The routinely used dual-colored break apart probes that are used in FISH are not sensitive enough to hybridize with such small cryptic insertions and therefore do not produce a signal as in our case. However, these small cryptic insertions can be amplified and detected by RT-PCR. These RT-PCR positive cases for hybrid PML/RAR α transcript classify a new cytogenetic subgroup of APL.

We suggest that RT-PCR should be performed at baseline to detect this small subset of t(15;17) negative APL cases, with cryptic or masked insertions.

RADHESHYAM PURKAIT AND *TUPHAN KANTI DOLAI

*Department of Paediatric Medicine and *Haematology, NRS
Medical College & Hospital,
Kolkata, WB, India.
radheshyampurkait@gmail.com*

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Vitamin D Deficiency: An Uncommon Cause of Quadripareisis

Vitamin D deficiency can present with neuromuscular symptoms at all ages from floppiness in infancy, delayed motor milestones in toddlers and acquired proximal muscle weakness in adolescents and young adults. Proximal limb myopathy associated with rickets is well known but truncal weakness is rare [1].

A 6-year-old girl presented with progressive weakness of body for six months and inability to get up from bed for two months. She was a vegetarian and had an aversion to milk and milk products. She received vigorous physiotherapy from a local hospital. At presentation, child had stable vitals and had pallor. Her weight was 17.5 Kg, The muscle bulk was normal, and power was MRC scale 3/5 at shoulder and hip joints and 4/5 at distal joints. Truncal and neck muscles were severely involved. Deep tendon reflexes were elicitable. There were no signs of sensory involvement, meningeal irritation, cerebral dysfunction or cranial nerve involvement. Wrist joints

were widened. X-ray knee joint showed cupping and fraying of lower end of femur, upper end of tibia and fibula, and fracture of upper part of both fibulae; severe osteopenia was present. Serum calcium was 7.0 mg/dL, phosphorus 3.1 mg/dL and alkaline phosphatase 2375 IU/L. Serum 25(OH) vitamin D levels were 5 ng/mL (normal 40-60 ng/mL) and Parathormone level was 70 pg/mL (normal 13-66 pg/mL). Serum electrolytes, renal and liver function tests, thyroid function test and anti-tissue transglutaminase antibody levels were within normal limits. Electromyography (EMG) could not be performed as patient was not cooperative; Nerve-conduction in lower limbs was done which was normal. She was given intramuscular injection of 600,000 IU of vitamin D and started on calcium supplements (200mg/kg/day). Within a week child started getting up from bed, and next week she was able to walk without support.

Vitamin D probably exerts its actions on muscles through two pathways. The genomic pathway affects calcium uptake, phosphate transport and phospholipid metabolism as well as myoblast differentiation and division involving *de novo* protein synthesis. Non-genomic pathway affects calcium transport and contractility apparatus [2].

Vitamin D deficiency is highly prevalent in Indian