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SCIENTIFIC LETTER

Hereditary Homozygous C3 Deficiency

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To the Editor: Primary C3 deficiency is a rare disease which manifests as early onset of recurrent bacterial infections; and sometimes with glomerulonephritis and autoimmune diseases [1]. Here we describe a girl who presented with recurrent infections from an early age along with rashes and arthralgia.

A 5-y-old girl, born of a 3rd degree consanguineous marriage, presented with a history of urinary tract infection, enteric fever, foot abscess and recurrent pneumonia from two and half years of age. She also had an episode of pyopneumothorax with *Escherichia coli* isolated from the drained pus. A history of erythematous rashes and arthralgia of the lower limbs was also present. Skin biopsy was suggestive of leucocytoclastic vasculitis. Anti dsDNA and Antinuclear antibody were negative. She was diagnosed as Henoch Schonlein Purpura and put on naproxen and oral steroids.

After six months of therapy, she came to us with multiple erythematous palpable rashes on both the lower limbs (Fig. 1). Systemic examinations were within normal limits. Investigations revealed hemoglobin: 8.4 g/dl; total white blood cell count 9.5 × 10^3/μL, Neutrophils 40% Lymphocytes 50% Monocytes 6% Eosinophils 4%; C-reactive protein 21 mg/L; platelets 5.84 × 10^3/μL; complement C3c <10 mg/dl (90–180); complement C4 15.4 mg/dl (10–40); immunoglobulin profile: within normal limits; pANCA, cANCA were negative. Noting this extremely low C3c levels, both parents and the younger sibling were screened. Father's complement C3c was 78.3 mg/dl; mother's 70.6 mg/dl and younger sister's 73 mg/dl. CH50 assay of the patient was 20% (69% – 139%). She was diagnosed as hereditary homozygous C3 deficiency, put on prophylactic oral penicillin and vaccinated against meningococcus, pneumococcus, typhoid and influenza. Oral hydroxychloroquine sulphate was started for the cutaneous vasculitic lesions, and the parents were counselled to seek medical advice at the earliest sign of any infection. On follow-up after six months, she remains asymptomatic except for on and off skin rashes.

Complement deficiencies form 1–6% of all primary immunodeficiency disorders [2]. Primary C3 deficiency is an autosomal recessive disorder, caused by homozygous or compound heterozygous mutation in the C3 gene (OMIM 120700) on chromosome 19p13 (OMIM 613799); resulting in recurrent infections with encapsulated bacteria that begin shortly after birth. Immune complex mediated diseases like systemic lupus erythematosus and membranoproliferative glomerulonephropathy have also been reported [1, 3].

The management includes prompt treatment of active infection and prevention of future infections. Patients should also be routinely immunized against encapsulated bacteria, if not already immunized. Replacement with regular plasma infusions, to replace the missing component, is generally not advocated [4, 5].

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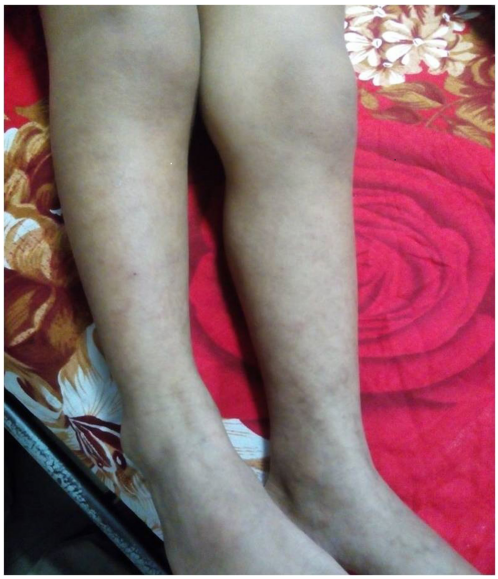

Fig. 1 Multiple erythematous palpable rashes on both lower limbs of the patient

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Compliance with Ethical Standards

Conflict of Interest None.

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