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Case Report

Recurrent Macroscopic Hematuria and Abdominal Pain: Questions and Answers

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Abstract

A 6.5 yr old girl was admitted with a category of clinical signs and symptoms including recurrent gross hematuria, abdominal pain, and fever. After different examinations including genetic analysis, the disease was diagnosed as Familial Mediterranean fever (FMF). It is suggested to consider FMF as a rare cause of recurrent gross hematuria, which is responsive to colchicine treatment.

Keywords: Familial Mediterranean fever, Hematuria, Fever, Pain, Iran

Case history

A 6.5 yr old girl was admitted at Aliasghar Children's Hospital, Tehran, Iran in 2014 with a history of recurrent gross hematuria, abdominal pain and fever. Hematuria was independent to respiratory tract infections or gastrointestinal disorders. She had no history of edema and oliguria. Family history was unremarkable for any renal or systemic disorders. On admission, physical examination and vital signs were normal.

Laboratory examination showed normal complete blood count, serum biochemistry, coagulation and serologic tests including C3, C4, CH50, ANA, anti DNA, anti GBM antibody, CANCA and PANCA. Increased ESR (75ml/h) was the only abnormal hematologic test (Table 1).

Urinalysis showed isolated hematuria (RBC: 30-35/HPF) without proteinuria. Urine culture and schistosomiasis screening were negative. Measurement of urine crystals were in normal limits.

Urinary tract ultrasonography was normal. Nephrolithiasis and nutcracker syndrome were excluded by abdominal triphasic spiral CT scan.

Table 1: Laboratory findings of the patient

Variables	Results
WBC	6000
HgB (g/dl)	12
PLT	331000
ESR (mm/h)	75
PT	13
PTT	40
C3	82.3
C4	12.1
CH50	104
IgM	2.07
IgG	8.4
IgA	0.64
IgE	27.18
LDH	874
Urine RBC/HPF	30-35
Urine protein (mg/24h)	50
Urine calcium (mg/24h)	27
Urine oxalate (mg/24h)	0.32

Audiometry (including high frequency impedence), retinoscopy and endoscopy showed no significant abnormalities. Psychological consult was normal. Renal biopsy was normal with no immunofluorescence deposition. A diagnostic test was performed.

Questions

1- What is the diagnosis?
2- What is the diagnostic test?

Answers

Question 1: Familial Mediterranean fever
Question 2: Mutation analysis is the most specific diagnostic test in Familial Mediterranean fever, showed MEFV gene mutation. However, treatment response to colchicin is considered another diagnostic and therapeutic option.

Discussion

The patient was a young girl with unexplained recurrent macroscopic hematuria. Totally, macroscopic hematuria originates from glomerular (IgA nephropathy, Alports syndrome, benign familial hematuria, poststreptococcal glomerulonephritis (GN), membranoproliferative GN , systemic lupus erhytematosus, membranous nephropathy, rapidly progressive GN, Henoch schonlein purpura, and goodpasture disease), tubulointerstitial (acute pyelonephritis, acute interstitital nephritis, tuberculosis, hematologic diseases) and urinary tract disorders (bacterial or viral infections, nephrolithiasis, crystaluria, structural or congenital anomalies, polycystic kidney disease, trauma, tumors, exercise, and medications (1). Renal biopsy was not in favor of the above disorders.
The patient had a history of abdominal pain and occasional fever coincidental or independent to gross hematuria, which reminds to syndromes with recurrent fever including familial Mediterranean fever (FMF). Therefore, colchicin was started and the patient had a characteristic response to treatment. Fever and abdominal pain subsided in less than 3 days with no episode of gross hematuria during the follow up period. Screening for FMF with mutation analysis showed R202Q mutation in favor of FMF.

Familial Mediterranean fever is an autosomal recessive disorder, characterized by recurrent fever and polyserositis (peritonitis, pleuritis, pericarditis, and arthritis) (2). It is caused by more than 30 point mutations in the MEFV gene. Diagnosis confirms by clinical manifestations, and mutation analysis in 60% of patients (3). Transient increase of acute phase reactants (CRP, ESR, fibrinogen levels, platelets) may occur during FMF attacks, usually returns to normal or continues in 2/3 during asymptomatic period (2,3,4,).
Renal involvement usually occurs in the form of AA amyloidosis with proteinuria, nephrotic syndrome and chronic renal failure (2, 5). Other manifestations including transient or persistent microscopic hematuria and/or albuminuria, recurrent pyelonephritis, FMF-related GN, acute poststreptococcal GN, IgM nephropathy, IgA nephropathy, crescentic GN, focal proliferative GN, diffuse proliferative GN, focal glomerulitis, focal glomerulosclerosis, minimal change disease, membranoproliferative GN and chronic GN have been reported in FMF (3, 6, 7), with an appropriate prognosis and renal survival. Approximately 5% of patients with FMF have been reported with HSP and 1% with polyarteritis nodosa (2).
Our patient had FMF with a very uncommon manifestation of isolated recurrent macroscopic hematuria, irrespective to acute or chronic GN or vasculitis. Therefore, it is suggested to consider FMF as a rare cause of recurrent gross hematuria, which is responsive to colchicine treatment.

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References

1. Kevin E.C. Meyers, MBBCh (2004). Evaluation of hematuria in children. *Urol Clin N Am*, 31(3): 559–573
2. Girisgen I, Sonmez F, Koseoglu K, Erisen S, Yilmaz D (2012). Polyarteritis nodosa and Henoch-Schönlein purpura nephritis in a child

with familial Mediterranean fever: a case report. *Rheumatol Int,* 32 (2):529-33.
3. Rigante D, Federico G, Ferrara P, Maggiano N, Avallone L, Pugliese AL, Stabile A (2005). IgA nephropathy in an Italian child with familial Mediterranean fever. *Pediatr Nephrol,* 20 (11):1642-4.
4. Korkmaz C, Ozdogan H, Kasapçopur O, Yazici H (2002). Acute phase response in familial Mediterranean fever. *Ann Rheum Dis*, 61(1):79-81.
5. Tekin M, Yalçinkaya F, Tümer N, Cakar N, Koçak H, Ozkaya N, Gençgönül H (1999). Familial Mediterranean fever-renal involvement by diseases other than amyloid. *Nephrol Dial Transplant*, 14 (2): 475-9.
6. Akpolat T, Akpolat I, Karagoz F, Yilmaz E, Kandemir B, Ozen S (2004). Familial Mediterranean fever and glomerulonephritis and review of the literature. *Rheumatol Int*, 24 (1): 43-5.
7. Peleg H, Ben-Chetrit E (2013). The kidney in familial Mediterranean fever. *J Rheumatol*, 40 (12): 1948-50.