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Case Report

CML Masquerading as Infective Endocarditis

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History and Examination

23 years young male patient, who is a known case of rheumatic heart disease with prosthetic AV valve, on chronic anticoagulation therapy, came with history of fever on and off since 1 month. Physical examination revealed hepatomegaly with approximately 16 cm splenomegaly with systolic murmur over mitral area. Splinter hemorrhages were seen over finger nails and no other stigmata of endocarditis were found. Other systems were unremarkable. All vitals were stable.

So, provisional diagnosis of Infective Endocarditis was made and patient was started on iv antibiotics.

Investigation and Stay in Hospital

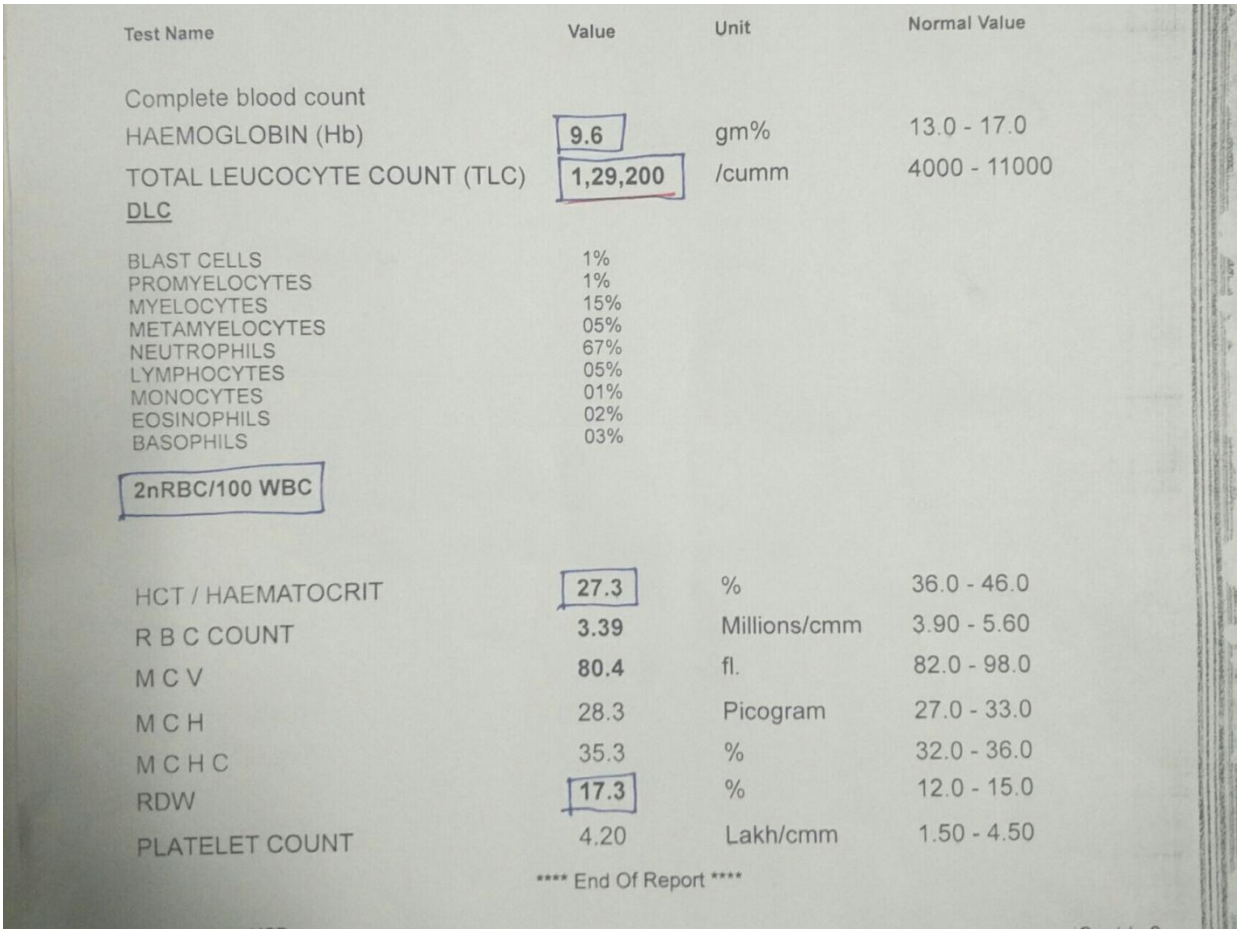
CBC reports were astonishing as it revealed high TLC counts of about 1,29,000 per cubic mm. DLC were reported as follows – blast cell -1%, promyelocytes – 1%, Myelocytes – 15%, Metamyelocytes – 5%, Neutrophil – 67%, Lymphocyte – 5%, Monocytes – 1 %, Eosinophil -2% and basophil – 3%. Hemoglobin 9.6 gm%. Peripheral smear reports revealed Chronic Myeloproliferative leukemia. USG abdomen and pelvis revealed severe splenomegaly of 22.9cm and mild hepatomegaly. Echocardiography revealed prosthetic AV valve with eccentric LVH with moderate MS with MR with no evidence of Infective endocarditis. All other reports were unremarkable including blood culture and sensitivity reports. Urine R/M showed microscopic hematuria.

In view of above mentioned reports; BCR-ABL gene rearrangement (PCR qualitative test) was done and was positive and type of translocation was major.

A Final Diagnosis of Chronic Myeloproliferative Leukemia with RHD was made and patient was started on Tab Imatinib. After 2 days of therapy fever resolved and spleen size regressed to about 16 cm.

Patient was symptomatically better and was discharged and regularly monitored on follow up. Follow up reports after one week revealed much better TLC counts of 48,000 per cubic mm and spleen size regressed to about 8 cm.

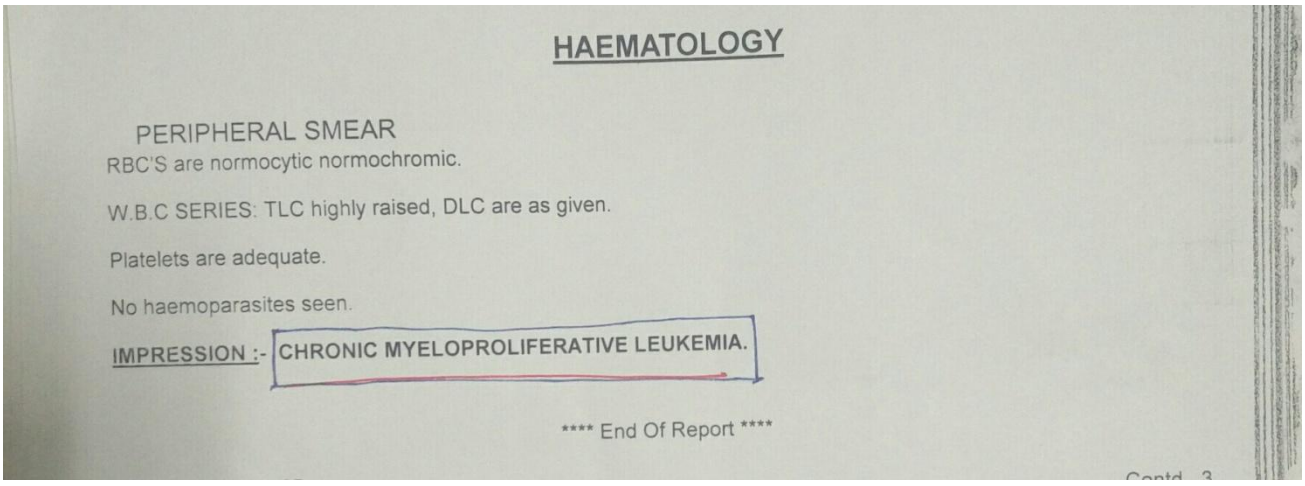
Complete Blood Count Reports during admission of the Patient



Test Name	Value	Unit	Normal Value
Complete blood count			
HAEMOGLOBIN (Hb)	9.6	gm%	13.0 - 17.0
TOTAL LEUCOCYTE COUNT (TLC)	1,29,200	/cumm	4000 - 11000
DLC			
BLAST CELLS	1%		
PROMYELOCYTES	1%		
MYELOCYTES	15%		
METAMYELOCYTES	05%		
NEUTROPHILS	67%		
LYMPHOCYTES	05%		
MONOCYTES	01%		
EOSINOPHILS	02%		
BASOPHILS	03%		
2nRBC/100 WBC			
HCT / HAEMATOCRIT	27.3	%	36.0 - 46.0
R B C COUNT	3.39	Millions/cmm	3.90 - 5.60
M C V	80.4	fl.	82.0 - 98.0
M C H	28.3	Picogram	27.0 - 33.0
M C H C	35.3	%	32.0 - 36.0
RDW	17.3	%	12.0 - 15.0
PLATELET COUNT	4.20	Lakh/cmm	1.50 - 4.50

**** End Of Report ****

Peripheral Smear Reports



HAEMATOLOGY

PERIPHERAL SMEAR

RBC'S are normocytic normochromic.

W.B.C SERIES: TLC highly raised, DLC are as given.

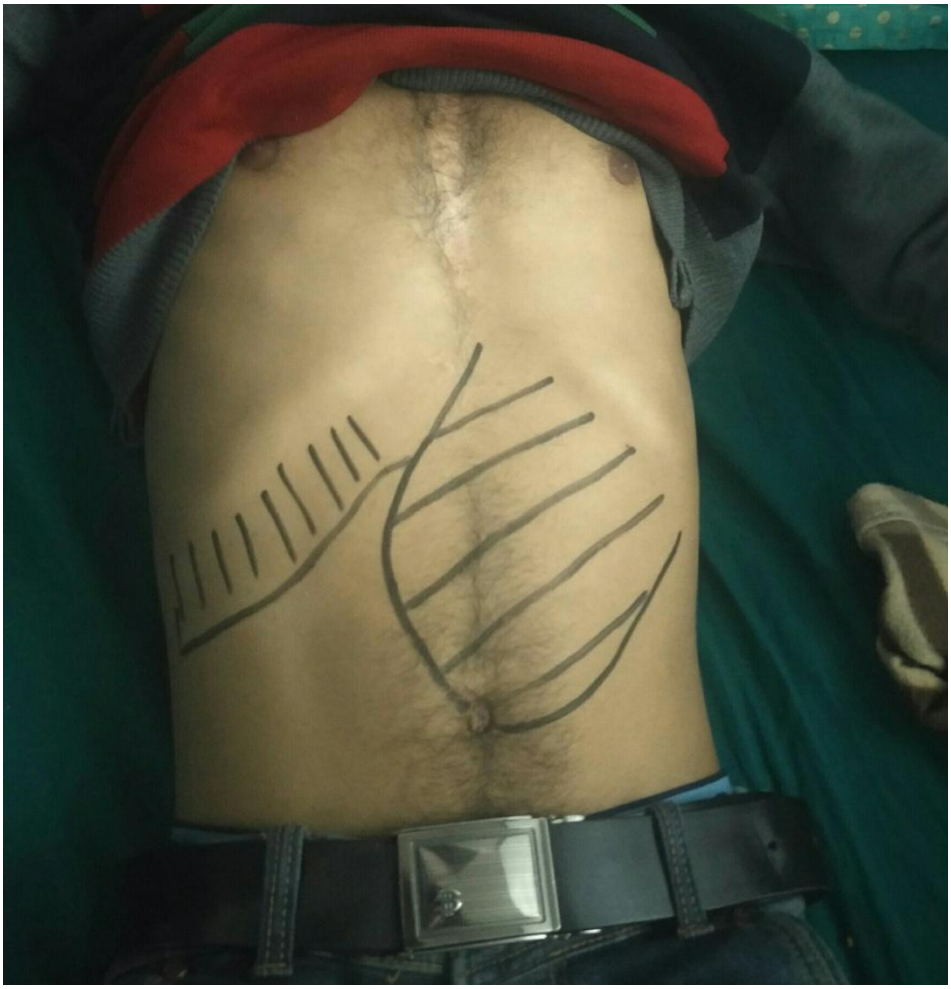
Platelets are adequate.

No haemoparasites seen.

IMPRESSION :- CHRONIC MYELOPROLIFERATIVE LEUKEMIA.

**** End Of Report ****

Massive Splenomegaly with Hepatomegaly on Examination of the Patient



BCR-ABL Gene Rearrangement, PCR Qualitative Reports

Test Name	Results	Units	Bio. Ref
BCR-ABL GENE REARRANGEMENT, PCR QUALITATIVE (Real Time PCR)			
BCR-ABL gene rearrangement	**Positive**		
Type of Translocation	Major		

Note

1. Sensitivity of the assay is 0.01% when copies of ABL detected is 100,000
2. Limit of detection is 10 copies of BCR-ABL fusion gene transcripts per PCR
3. This is an in-house developed assay designed as per EAC (Europe Against Cancer) protocol
4. This test detects Major (M) gene rearrangements namely- e13a2 & e14a2 and Minor (m) gene arrangement e1a2. This test does not detect micro gene rearrangement e19a2
5. Test conducted on Whole blood / Bone Marrow

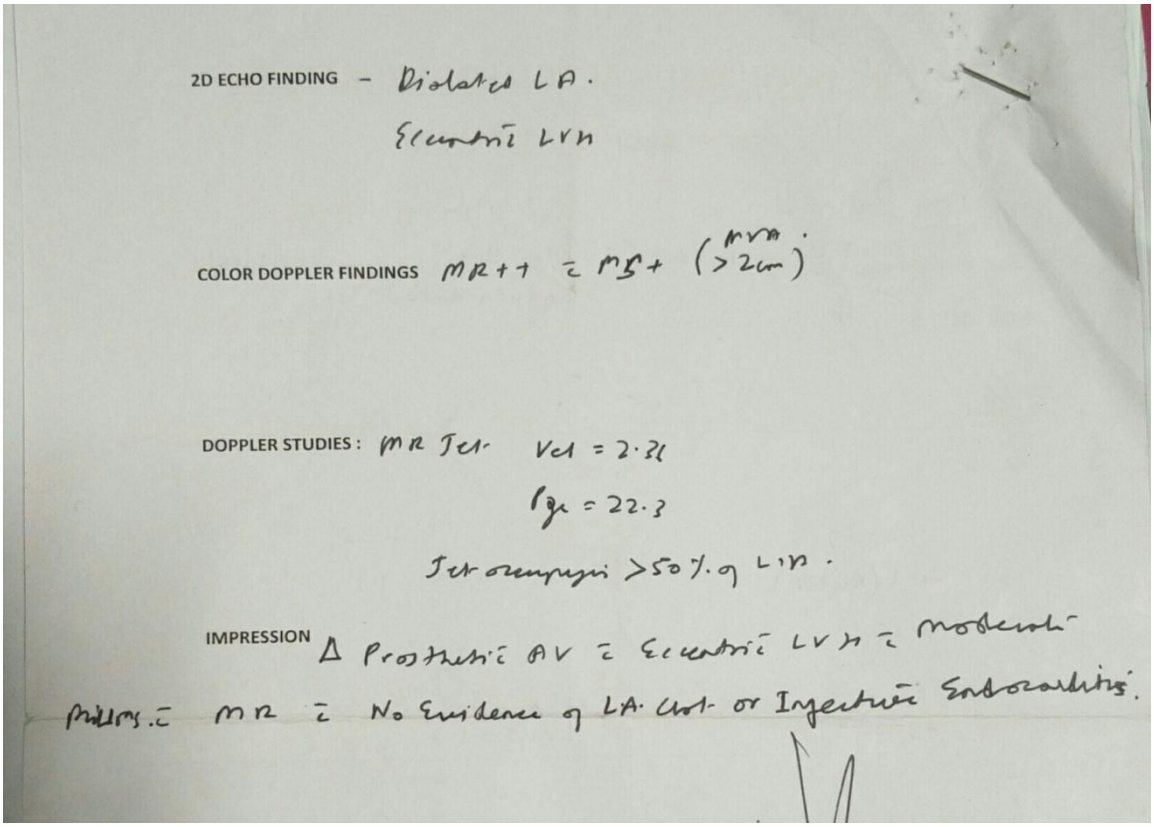
Comments

Chronic Myeloid Leukemia (CML) is the commonest myeloproliferative neoplasm and possibly the commonest adult leukemia in India. This clonal stem cell disorder is characterized by a proliferation of myeloid cells at all stages of differentiation and the t(9:22) (q34:q11) leading to formation of BCR-ABL fusion gene. Cytogenetic and molecular studies are vital for the diagnosis of CML by using detection procedures for Philadelphia chromosome. The abnormality is present in over 95% patients of CML while remainder 5% have complex or variant translocations involving additional chromosomes. Major gene rearrangements are detected in CML while minor gene rearrangement may be detected in ALL.

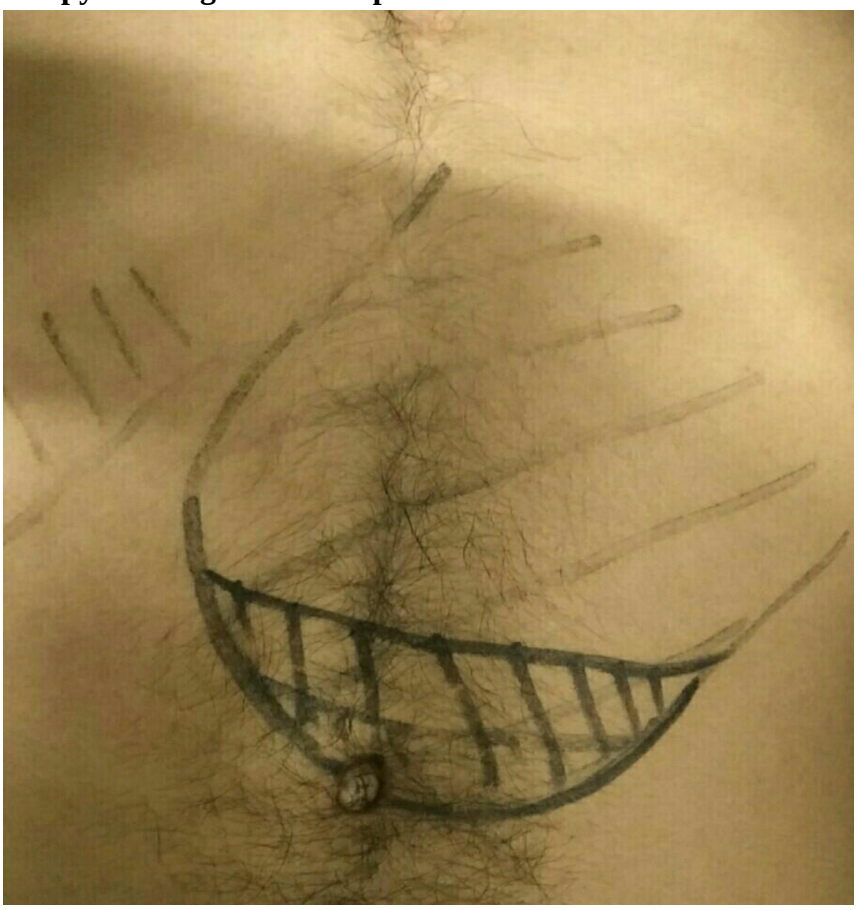
Uses

- To detect & monitor therapy in CML patients.
- As a prognostic marker in ALL patients. Presence of BCR-ABL gene rearrangement is associated with poor prognosis.

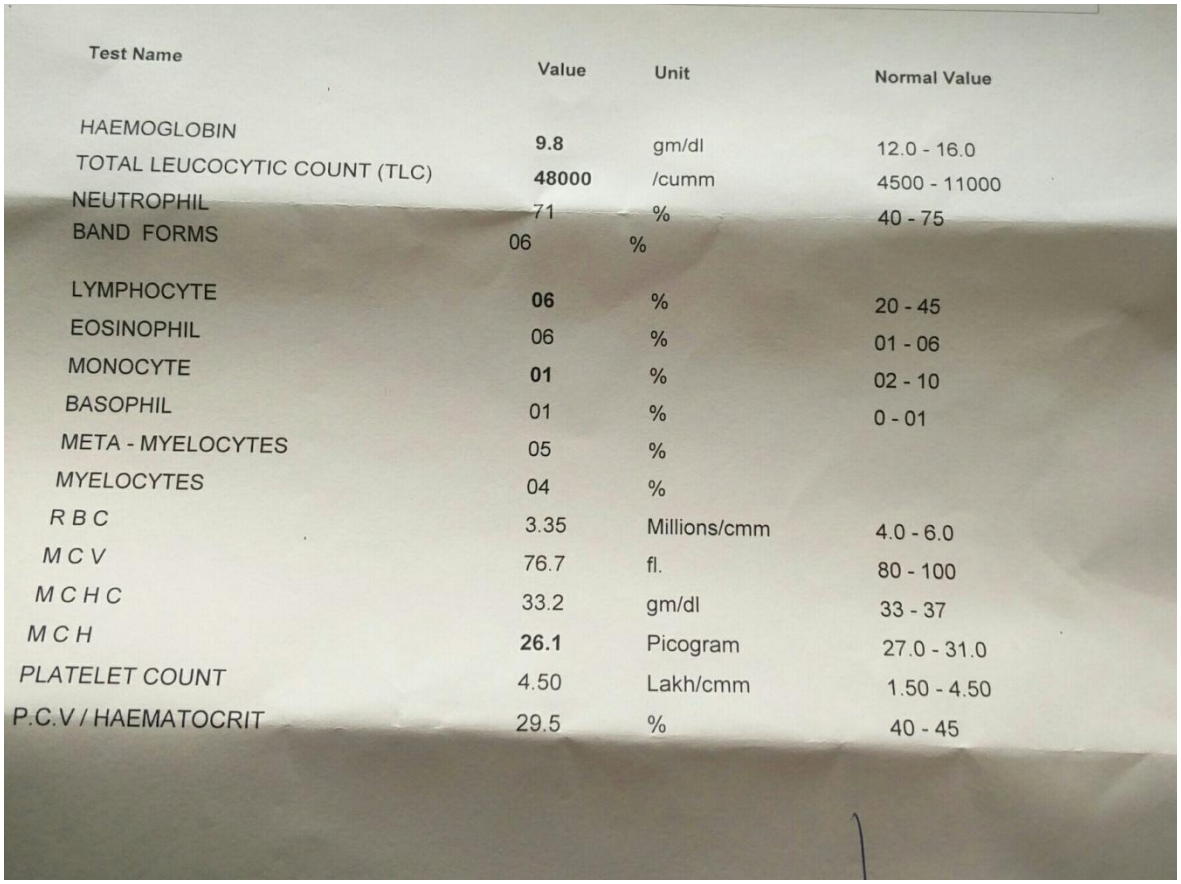
2D ECHO Reports



Response to Imatinib Therapy and Regression of Spleen Size



Follow Up Complete Blood Count Reports



Test Name	Value	Unit	Normal Value
HAEMOGLOBIN	9.8	gm/dl	12.0 - 16.0
TOTAL LEUCOCYTIC COUNT (TLC)	48000	/cumm	4500 - 11000
NEUTROPHIL	71	%	40 - 75
BAND FORMS	06	%	
LYMPHOCYTE	06	%	20 - 45
EOSINOPHIL	06	%	01 - 06
MONOCYTE	01	%	02 - 10
BASOPHIL	01	%	0 - 01
META - MYELOCYTES	05	%	
MYELOCYTES	04	%	
R B C	3.35	Millions/cmm	4.0 - 6.0
M C V	76.7	fl.	80 - 100
M C H C	33.2	gm/dl	33 - 37
M C H	26.1	Picogram	27.0 - 31.0
PLATELET COUNT	4.50	Lakh/cmm	1.50 - 4.50
P.C.V / HAEMATOCRIT	29.5	%	40 - 45

Conclusion

As the patient was a known case of RHD with Prosthetic aortic valve with hepato-splenomegaly, with splinter hemorrhages and microscopic hematuria there was definite possibility of Infective endocarditis which seemed more likely. But sometimes less compelling diagnosis like CML in this case was the root problem. Hence this emphasizes the role of differential diagnosis in better management of patients.