

Patient: John Doe



Case No: P11-00351

DOB/Gender: xx/xx/xxxx (83 yrs.) - Male

SSN: xxx-xx-xxxx

MRN/ID: 123456

Provider: Jane Doe, MD

Account: Hematology Oncology Assoc

Phone: 800-123-4567 Fax: 800-123-4444

4 Science Park, New Haven, CT 06511  
Phone: 203-787-7888 Fax: 203-823-4882  
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Collected: xx/xx/xx

Received: xx/xx/xx

Reported: xx/xx/xx

Alert Status: Routine

Report Status: Final

Report Category: Neoplastic

**Clinical information:** Pancytopenia  
Received CBC, reported on 3/6/2016: WBC 0.5; RBC 4.36; HGB 12.7; HCT 39.0; MCV 89.4; MCH 29.0; MCHC 32.4; RDW 12.7; PLT 183; MPV 9.6; LYM 73.8%; MON 0.9%; NEU 16.8%; EOS 6.5%; BAS 1.7% (NP = not provided)

**Specimens received:** 2 Green-top tubes, 1 lavender-top tube

**COMPREHENSIVE DIAGNOSIS**

Professional Services Provided By  
**Yale SCHOOL OF MEDICINE**

**FINAL DIAGNOSIS:**

**Pancytopenia with abnormal clone detected by FISH (see comment)**

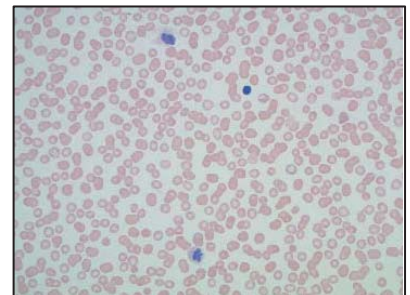
**COMMENT:**

The peripheral blood indices reveal pancytopenia with macrocytic anemia, confirmed on morphologic examination. Although there is no morphologic evidence of dyspoiesis or circulating blasts, the finding of deletion of chromosomes 7 and 20q by FISH are suspicious for a myeloid neoplasm, as these abnormalities are common findings in myelodysplastic syndrome and acute leukemia. A bone marrow core biopsy and aspirate smear is recommended as clinically indicated and feasible. Correlation with clinical and other laboratory findings (i.e. B12, folate, serum copper) is required for complete interpretation.

Electronically signed by: S. David Hudnall, MD, FCAP

**COMPONENT DIAGNOSES**

- Aspirate: Pancytopenia
- Flow Cytometry: Normal peripheral blood flow cytometry (see comment)
- FISH: **Abnormal clone with (1) Monosomy 7 in 9% of cells analyzed and (2) deletion 20q12 (D20S108) in 6% of cells analyzed with probes specific for recurrent abnormalities in Myelodysplastic Syndrome (MDS).**



**Peripheral Blood Smear**

**DIAGNOSIS:**

Peripheral blood: Pancytopenia

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**SMEAR REVIEW:**

The peripheral blood indices reveal a macrocytic anemia, leukopenia and mild thrombocytopenia, confirmed on morphologic examination. There is moderate anisocytosis with minimal poikilocytosis, and mild polychromasia. nRBCs are not identified. There are decreased numbers of white blood cells with normal differential, without atypical or dyspoietic forms. Circulating blasts are not identified. Platelets appear mildly decreased with normal granulation and appearance.

SAMPLE

**DIAGNOSIS:**

Peripheral blood: Normal peripheral blood flow cytometry (see comment)

**COMMENT:**

Diagnostic features of involvement by an acute leukemia or paroxysmal nocturnal hemoglobinuria are not seen by flow cytometry. If clinically indicated, a myelodysplastic syndrome can be best evaluated by morphologic and cytogenetic analysis of a bone marrow core and aspirate smear. Correlation with clinical and laboratory findings is advised.

Electronically Signed By: S. David Hudnall, MD, FCAP

**INTERPRETATION:**

Flow cytometric examination of the peripheral blood smear reveals no circulating blasts. No aberrant myeloid antigen expression is identified. The lymphocytes (24%) include 1% polyclonal B-cells, 90% mature T-cells with a slightly inverted CD4/CD8 ratio of 0.7, and 8% natural killer (NK) cells.

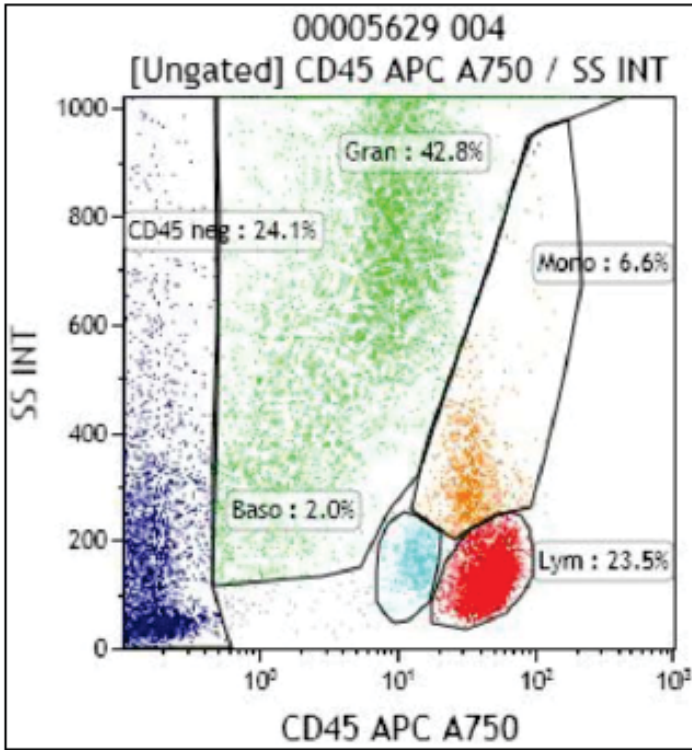
PNH analysis was performed on the leukocytes using CD14 and CD33 to differentiate the granulocytes and monocytes. Glycophorin (CD235a) was used to differentiate red blood cells (RBC). No lack of expression on CD55 or CD59 was observed on all cells analyzed as well as no lack of expression of FLAER on the leukocytes. No immunophenotypic evidence of PNH is observed.

**RESULT:**

**Analysis Time:** 12/11/11 13:47

**Viability:** 93% (Normal > 80%)

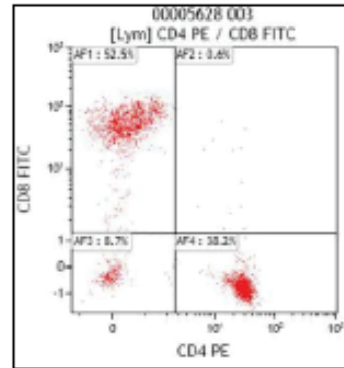
**Specimen:** PB, Lavender-top tube



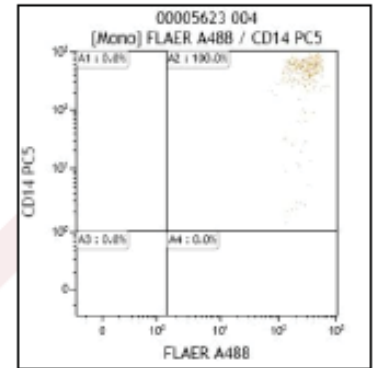
Flow Differential

Flow Cytometry Differential

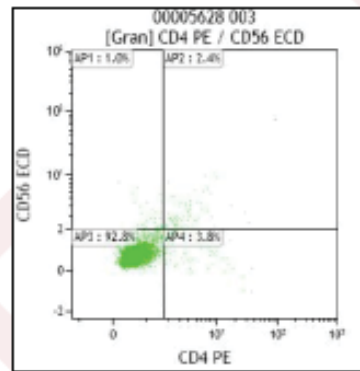
Lymphocytes:	24%
Monocytes:	7%
Granulocytes:	43%
Basophils:	2%
Non-lysed RBCs:	24%



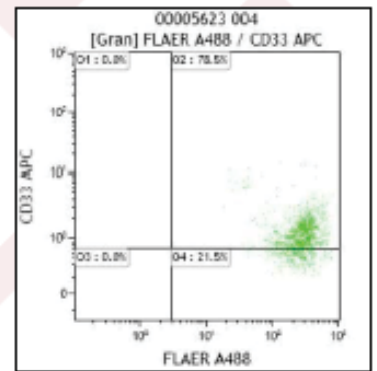
Inverted CD4/CD8 Ratio



FLAER+ Monocytes



CD56 Granulocytes



FLAER+ Granulocytes

**Granulocytes Gated Population (CD45 and side scatter)**

T/NK-cell	%	Myelo/Mono	%	Precursor	%	B-Cell	%
CD4	4	HLA-DR	2	CD117	2	CD10	71
CD56	1	CD13	94	CD34	3	CD19	44
		CD33	39				
		CD15	99				
		CD71	70				
		CD64	6				
		CD14	77				
		CD61	49				
		CD11b	99				
		CD16	92				
		CD55	96				
		CD59	99				
		FLAER	100				
<b>Other</b>	<b>%</b>						
CD45	100						

**Lymphocytes Gated Population (CD45 and side scatter)**

T/NK-cell	%	B-Cell	%	Other	%
CD8	53	sKappa	<1	CD45	100
CD4	38	sLambda	<1		
CD56	8	CD20	1		
CD2	96	CD38	<1		
CD3	90	CD10	<1		
CD5	90	CD19	1		
CD7	97				

**Monocytes Gated Population (CD45 and side scatter)**

Myelo/Mono	%	Other	%
CD55	100	CD45	100
CD59	97		
FLAER	100		

**Red Blood Cells Gated Population (CD45 and side scatter)**

Myelo/Mono	%	Red Blood Cells	%
CD55	99	CD235a	72
CD59	100		

Intensity: B = bright D = dim M = moderate

Disclaimer: The adequacy of staining is verified by the appropriate positive and negative controls. The reagents used for these assays are analyte specific reagents (ASR). Their performance characteristics have been validated by Precipio Diagnostics, LLC, New Haven, CT. They have not been reviewed by the FDA. The FDA has deemed that such approval is unwarranted. These assays are for clinical use and should not be viewed as experimental or "research use only".

**DIAGNOSIS:**

Abnormal clone with (1) Monosomy 7 in 9% of cells analyzed and (2) deletion 20q12 (D20S108) in 6% of cells analyzed with probes specific for recurrent abnormalities in Myelodysplastic Syndrome (MDS).

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**FISH "ISCN":**

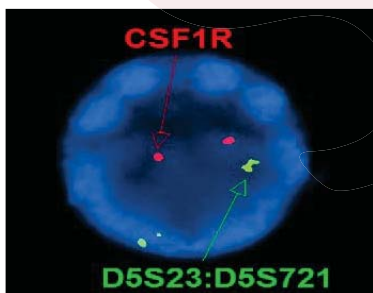
nuc ish  
 (RPN1,MECOMx2)[200],[CSF1Rx2,D5S23:D5S721x2][200],[D7S486x1,CEP7x1][18/200],[CEP8x2,D20S108x1][12/200],  
 (MLLx2)[200],[ETV6x2][200],[RARAx2][200],[Tel19p,TEL19q)x2[200]

**INTERPRETATION:**

Fluorescence in situ hybridization (FISH) with a panel of probes specific for detection of recurring chromosome abnormalities in MDS was performed on uncultured peripheral blood cells.

The regions/loci represented in these probe mixes were:

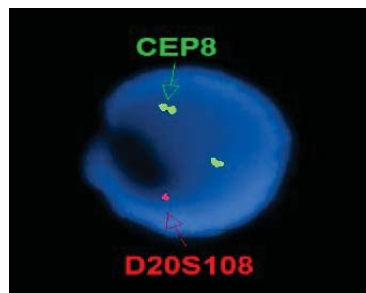
1. RPN1/MECOM dual color probes used to detect copy number/rearrangements of chromosome 3 and 3q21-q26 regions reveal a hybridization pattern within normal limits in 200 analyzed nuclei.
2. CSF1R (5q33~34) and D5S721:D5S23 (5p15.2), used to detect copy number abnormalities/deletion of chromosome 5, reveal a hybridization pattern within normal limits in 200 analyzed nuclei.
3. D7S486 (7q31) and a centromere probe to chromosome 7 (CEP 7), used to detect copy number abnormalities/deletion of chromosome 7, reveal an abnormal hybridization pattern consistent with monosomy 7 in 18 of 200 analyzed nuclei.
4. CEP8 (centromere probe to chromosome 8), used to detect copy number abnormalities of chromosome 8, reveals a hybridization pattern within normal limits in 200 analyzed nuclei.
5. MLL dual color break a part probe used to detect rearrangement/deletion at 11q23 region, reveals a hybridization pattern within normal limits in 200 analyzed nuclei.
6. RARA, used to detect rearrangement /deletion of 17q21.1 region, reveal a hybridization pattern within normal limits in 200 analyzed nuclei.
7. ETV6 dual color break a part probe, used to detect rearrangement/deletion at 12p13.2, reveals a hybridization pattern within normal limits in 200 analyzed nuclei.
8. Tel19p/19q used to detect copy number abnormalities of chromosome 19, reveal a hybridization pattern within normal limits in 200 analyzed nuclei.
9. D20S108 (20q12), used to detect deletion/copy number abnormalities of chromosome 20, reveals an abnormal hybridization pattern consistent with deletion 20q12 in 12 of 200 analyzed nuclei.



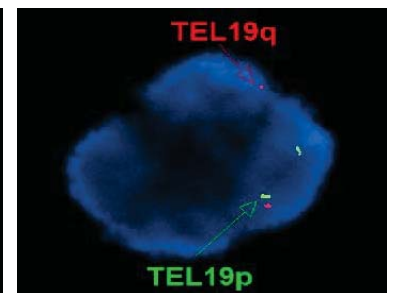
No 5q Deletion/Monosomy 5



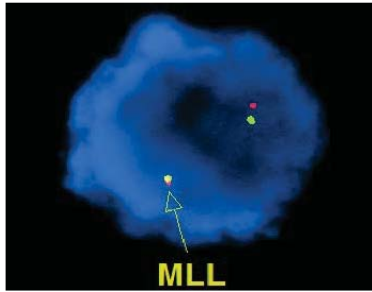
Monosomy 7



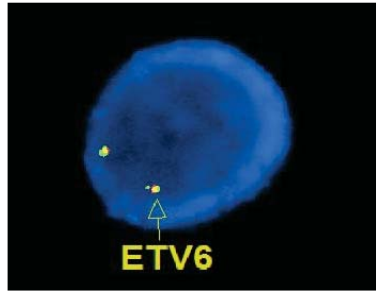
20q Deletion /No Trisomy 8



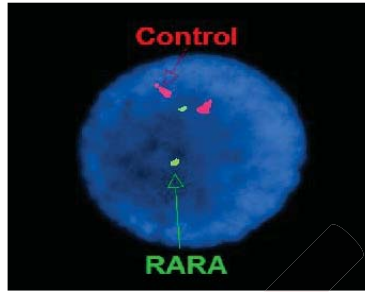
No Trisomy 19



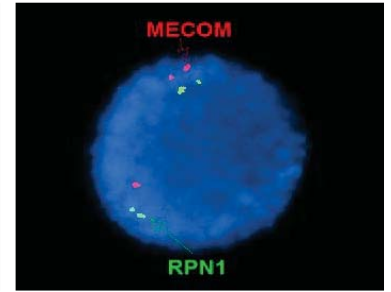
No MLL Rearrangement



No ETV6 Rearrangement



No RARA Rearrangement



Normal Chromosome 3

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