NEO GENOMICS
866.776.5907, option 3

COMPASS®

Client 1630
Sample Hematopathology/Oncology Facility
999 Elm Street, QA
Mars, FL
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CC 1

Patient Name: **Doe, Jane**
Patient DOB / Sex: **01/01/1950 / F**
Specimen Type: **Bone Marrow**
Body Site: **Bone Marrow**
Specimen ID: **12345**
MRN:
Reason for Referral: **Thrombocytopenia**

Ordering Physician(s): **Pathologist Sample**
Treating Physician(s): **Sample Oncologist, M.D.**
Accession / CaseNo: **418056 / GPS19-000724**
Collection Date: **10/15/2019**
Received Date: **10/16/2019 10:44:00 AM CDT**
Report Date: **10/17/2019 12:15:49 PM EST**

Clinical History:

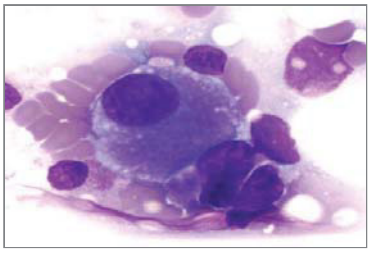
69-year-old female with a history of thrombocytopenia. Accompanying CBC report, dated 10/14/2019, indicates WBC 5.0 K/uL, RBC 3.27 M/uL, Hgb 9.7 g/dL, HCT 32.5%, MCV 99.4 fL, MCH 29.7 pg, MCHC 29.8 g/dL, RDW 14.5%, platelets 89 K/uL with a differential count of granulocytes 70.7%, lymphocytes 20.1%, monocytes 6.6%, eosinophils 1.2%, basophils 1.4%.

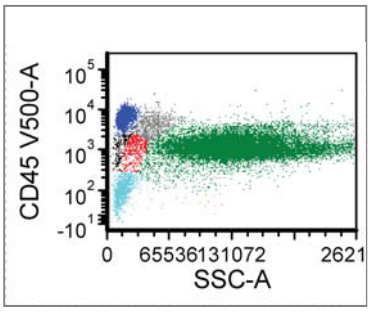
Final Diagnosis:

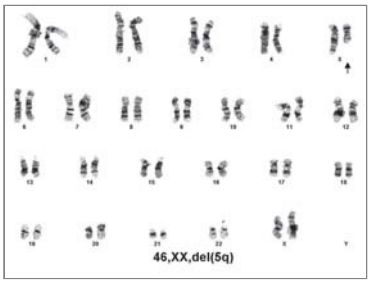
MYELODYSPLASTIC SYNDROME WITH ISOLATED DEL(5Q) AND TP53 MUTATION

Comprehensive Assessment

The bone marrow shows prominent dysmegakaryopoiesis, with no increase in blasts. Additional studies show an isolated del(5q) by cytogenetics and a TP53 mutation by the NextGen Sequencing. The CBC shows thrombocytopenia as well as anemia. The findings are diagnostic of a myelodysplastic syndrome with isolated del(5q). While this subtype of MDS is typically associated with a favorable prognosis and response to lenalidomide, the concomitant presence of a TP53 mutation is associated with an increased risk of leukemia transformation and a poorer response to lenalidomide (Jadersten M, et al. J Clin Oncol 2011;29:1971-9). Revised International Prognostic Scoring System (IPSS-R) for myelodysplastic syndrome: Cumulative score of 2.5, as determined by 1 point for good cytogenetics, 0 points for <=2% blasts, 1 point for 8-<10 g/dL Hgb, 0.5 points for 50-<100 K/uL platelets, and 0 points for =>0.8 K/uL ANC (Greenberg PL, et al. Blood 2012;120:2454-65).







Morphology

Bone marrow aspirate smears and core biopsy:
Hypercellular bone marrow with dysmegakaryopoiesis and no increase in blasts, consistent with a myelodysplastic syndrome

Flow Cytometry

No definitive immunophenotypic evidence of a monoclonal B-cell, aberrant T-cell, or increased blast population

Cytogenetics

46,XX,del(5)(q13q33)[9]/46,XY[11] ABNORMAL FEMALE KARYOTYPE
Chromosome analysis shows a female karyotype with one ABNORMAL clone(s) showing a deletion of 5q (9/20 cells).

FISH

Chromosome 8:	Not Detected	Chromosome 20:	Not Detected	**5q-/-5/+5 tricolor:**	**DETECTED**
7q-/-7 tri:	Not Detected	KMT2A (MLL) (11q23)*:	Not Detected		

FISH analysis for 5p/5q, 7p11/7q31, chromosome 8, KMT2A (MLL) and 20q: ABNORMAL results with 5q-

NeoTYPE™ Myeloid Disorders Profile

TP53; p. E285K **DETECTED**