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
<table>
<thead>
<tr>
<th>Chromosome 8:</th>
<th>Not Detected</th>
<th>Chromosome 20:</th>
<th>Not Detected</th>
<th>5q-/-5/+5 tricolor:</th>
<th>DETECTED</th>
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</thead>
<tbody>
<tr>
<td>7q-/-7 tri:</td>
<td>Not Detected</td>
<td>KMT2A (MLL) (11q23)*:</td>
<td>Not Detected</td>
<td></td>
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</tr>
</tbody>
</table>

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