Indy Hematology Review 2022

Challenging Cases Presentation

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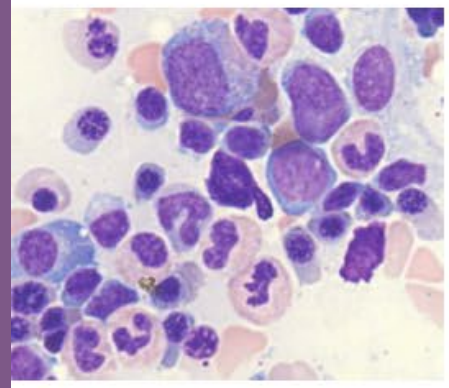
Challenging Cases Presentation: Indy Hematology Review® 2022

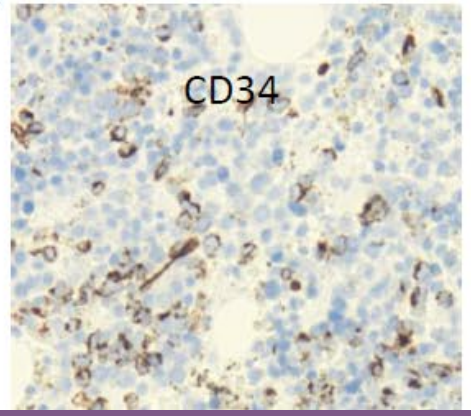
PATIENT CASE #1: MDS with del (20q)

- 79-year-old female with a history of MDS diagnosed in March 2008 following her presentation with anemia and thrombocytopenia and a bone marrow biopsy which revealed hypercellularity with 50% trilineage hematopoiesis, increased erythrocytosis, and abnormal cytogenetics with a 20q deletion.
- Initial therapy: Erythropoietin with disease control.
- In January 2022, the patient presented with progressive cytopenias: WBC: 4, Hg: 7.8, Plts133K, and ANC1200.
- Repeat BM biopsy: Myelodysplastic syndrome with excess blasts type 2 with 15-19% myeloblasts, CD34 positive, and an abnormal karyotype with interstitial deletion of 20q without other clonal cytogenetic abnormalities observed.
- NGS: mutation of U2AF1 p.(S34F), RUNX1. Notable negativity were IDH1 and IDH2mutations and as a result of which decitabine instituted.
- May 2022: A follow up BM biopsy: 5-9% myeloblasts with low level deletion of chromosome 20q , findings consistent with persistent myelodysplastic syndrome with excess blasts type 1.

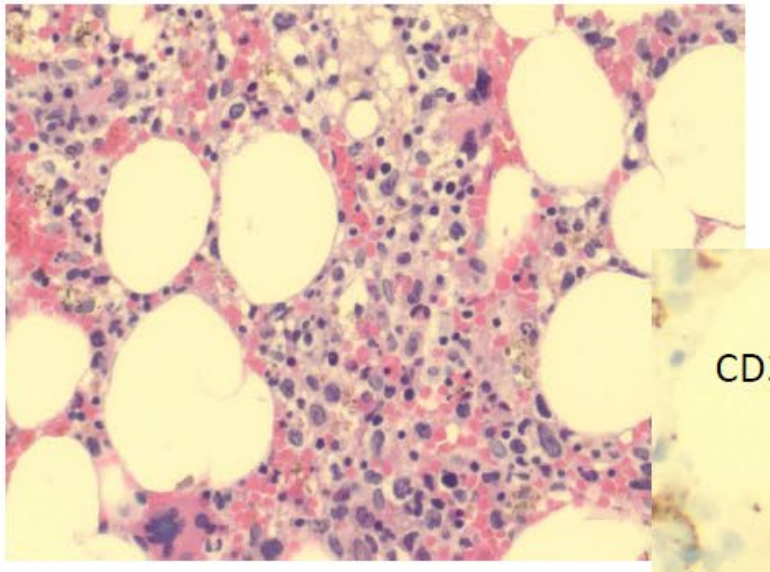
Case#1

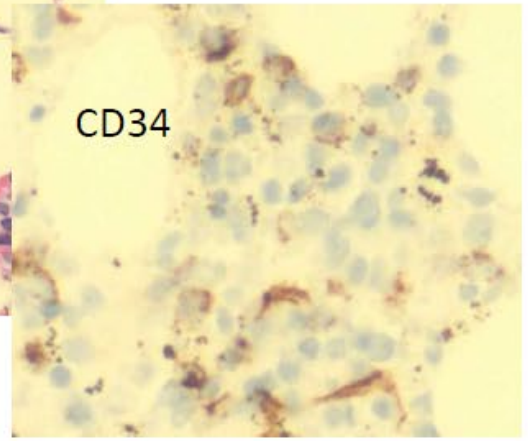
- 2008:
  - MDS with del(20q)
- 01/2022
  - MDS-EB-2; 15-19% blasts
  - Cytogenetics: del(20q)
  - NGS: U2Af1, RUNX1





- 05/2022
  - MDS-EB-1; 5-9% blasts
  - Cytogenetics: del(20q)





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PATIENT CASE #2: IDH-2 Mutant MDS

- An 81-year-old man with a history of MGUS diagnosed in 2010 presents in December 2021 with pancytopenia: WBC: 2.3, hg 9.8, platelet count 68K, and ANC 920.
- Bone marrow biopsy: markedly hypercellularity with left shifted granulocytic maturation and increased CD34+ blasts of ~ 5-9% and trilineage dysplasia with rare plasma cells and rare lymphoid aggregates and trace iron stores.
- Flow cytometry to be CD34+, CD117+, HLA-DR+, CD33+, CD13+, and cMPO+ with NTdT-.
- Cytogenetics revealed a 46XY normal male karyotype
- NGS revealed mutations of SRSF2, AXL1, and IDH2 and negative for FLT3, MPL1, and IDH1.
- Diagnosis: Myelodysplastic syndrome.

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CASE #3: Chronic Phase CML in MMR4 since 2015

- 68-year old female with a history of chronic myelogenous leukemia which was apparently diagnosed in August 2006 following her presentation with leukocytosis
- BM biopsy: Hypercellular with scattered small hyperlobulated megakaryocytes and increased reticuloendothelial iron stores, findings suspicious for myeloproliferative disorder, resulting in further workup including cytogenetics which showed the absence of Philadelphia chromosome: 19 of 22 metaphases with atypical banding pattern containing an additional copy of chromosome 19.
- FISH revealed a chromosomal translocation of BCR/abl. In addition, 180 of 200 nuclei with a deletion of a derivative of chromosome 9 associated with a poor prognosis CML.
- Diagnosis: Chronic myelogenous leukemia, initially treated with imatinib, tolerated poorly, resulting in multiple dose modifications with significant toxicities, resulting in discontinuation.
- Subsequently therapy: Dasatinib discontinued for pericardial and pleural effusion now being treated with bosutinib @ 200mg/day since March 2015 in MMR4.

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PATIENT CASE #4; CORE BINDING PROTEIN AML

- 39-year young female, noticed anemia on evaluation in the ER for abdominal pain,
- 6/22/2022: Hgb 8.5, MCV 75, PLT 146, blasts 1%, ANC 300
- 6/29/2022: Hgb 8.0, PLT 148, ANC 140, PB FCM: Revealed 13% blasts, underwent BMBX
- AML CFBP-MYH11, PTPN11.

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Case#4

- 07/2022
  - AML
  - Cytogenetics: pending
  - NGS: CBFB-MYH11, PTPN11