Massive splenomegaly, an unexpected diagnosis

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Presenting illness

67 man
2 week history of left sided abdominal pain
History of only renal calculi
Seen at peripheral emergency department

Physical examination: NAD
? renal calculi
Pain resolved
Follow up with GP

Renal tract ultrasound
Referred to ED for review.
History and examination

- Night sweats last few months
- Belt girth reduced by 1 notch, appetite reduced
- No pruritis
- No frequent infections
- No medications or other medical history
- Family history unremarkable

- Tender spleen, 10cm below costal margin
- Lymph nodes:
  - Supraclavicular LN and inguinal LN
- Small petechiae over limbs
Investigations

- Hb 144 g/L
- WCC 6.0 x 10⁹/L
- Platelets 44 x 10⁹/L
- Film:
  - LDH 272 U/L (NR < 250)
  - Urate 0.32 mmol/L
  - Mild global LFT derangement, otherwise unremarkable Biochemistry
Bone marrow
Flow cytometry
Cerebrospinal fluid
Flow cytometry

• Expressing:
  • Dim CD45
  • CD4
  • CD56
  • CD123
  • CD33
  • HLA-DR
  • CD38
  • CD117

• Negative:
  • MPO
  • Monocytic: CD14, CD64/CD15
  • B and T lineage-specific markers
Cytogenetics

Test Requested: Cytogenetic analysis referred for "67 M with massive splenomegaly: lymphoma favoured over myelofibrosis."

Preparation: 24 hour synchronised and 72 hour unsynchronised cultures.
Band: G Banding. Resolution: 300bphs
Cells analysed: 16 Cells counted: 4 Cells scored: 0 Total cells: 20

Abnormal Karyotype Result:

Conclusion:
9/20 cells examined had a normal male karyotype: 46,XY.
11/20 cells were abnormal with a hypodiploid karyotype involving numerical and structural abnormalities:
Numerical changes:
- Loss of one copy of chromosomes 13 and 21 [-13, -21].
Structural rearrangements:
- Additional material of unknown origin on chromosomes 1p, 12p and 22p [add(1p), add(12p), add(21p)].

The abnormalities seen here are non-specific. However, they were only detected on the long term cultures, suggesting mature B-cell type abnormalities usually associated with lymphomas.

A complex karyotype such as seen here is usually associated with a less favourable outcome.

It may be beneficial to correlate this result with aspirate findings and other ancillary tests.
Histopathology
Diagnosis?

• Lymphoma
• AML
• ..something else..

• Blastic plasmacytoid dendritic cell neoplasm
Blastic Plasmacytoid Dendritic Cell Neoplasm

- Rare, aggressive, median age 67; M:F = 3:1
- Skin > bone marrow / peripheral blood > lymphadenopathy > leukaemia > CNS
- Diagnosis typically made on histology
- By flow cytometry:
  - Lack of lineage-specific antigens
  - Expresses: CD4, CD56, dim CD45RA
    - And a plasmacytoid dendritic cell associated antigen: CD123 (bright), BDCA-2/CD303, TCL1A, CD2AP, TCF4
    - Often: CD33, CD7, CD5, CD36, CD38
- Abnormal karyotype in two thirds; frequently complex
- Median survival 10-20 months; chemo-sensitive, relapse the rule.
Questions?