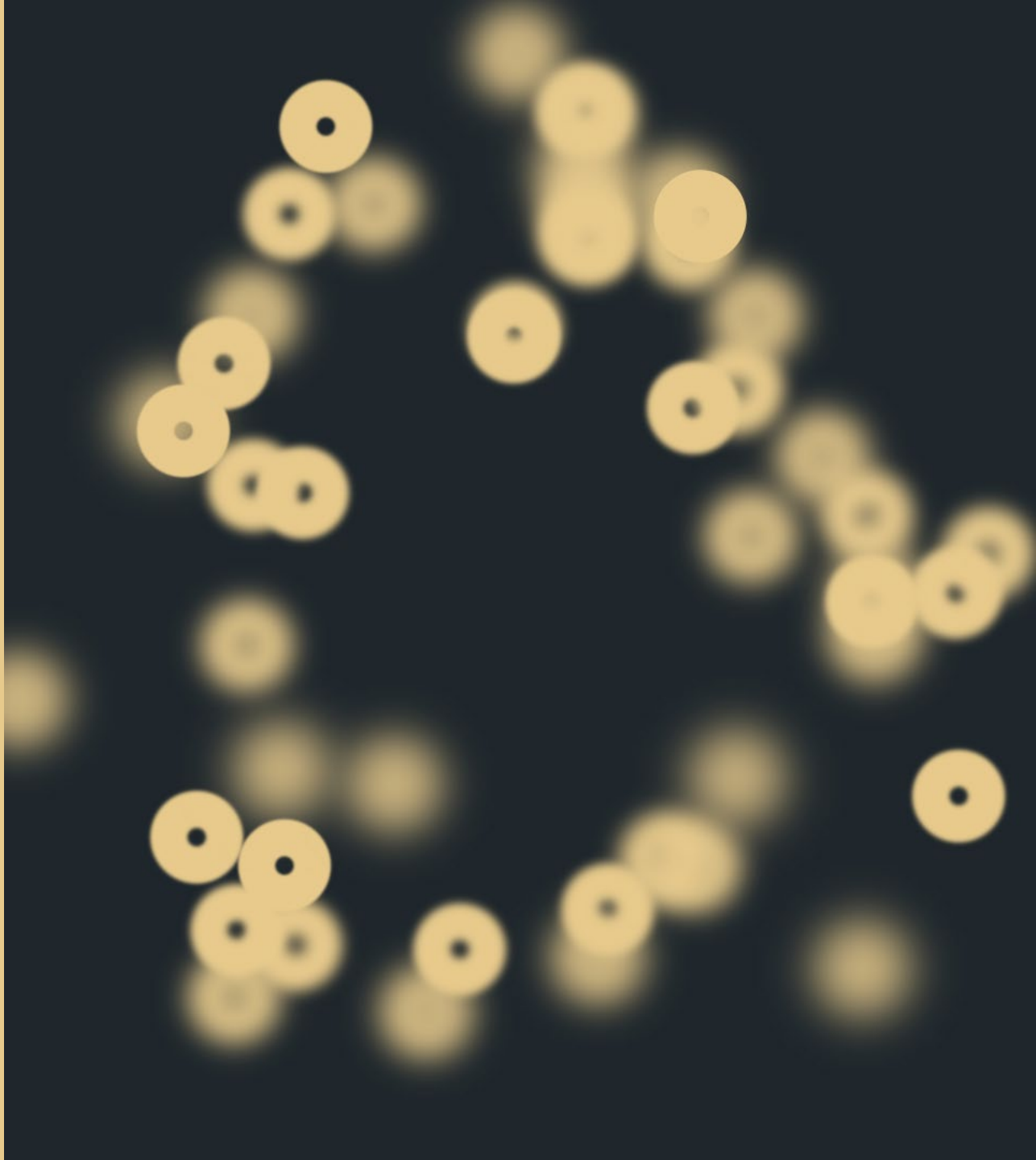
eha

EHA-SWG Scientific Meeting on Recent Advances in the Pathogenesis and Treatment of Secondary Acute Myeloid Leukemias

Berlin, Germany
April 25-26, 2025



Swing Between Two Lineages.

The Exceptional Case of Primary Myelofibrosis Transformation to Acute Lymphoblastic Leukemia and Subsequent Relapse of Acute Myeloid Leukemia.

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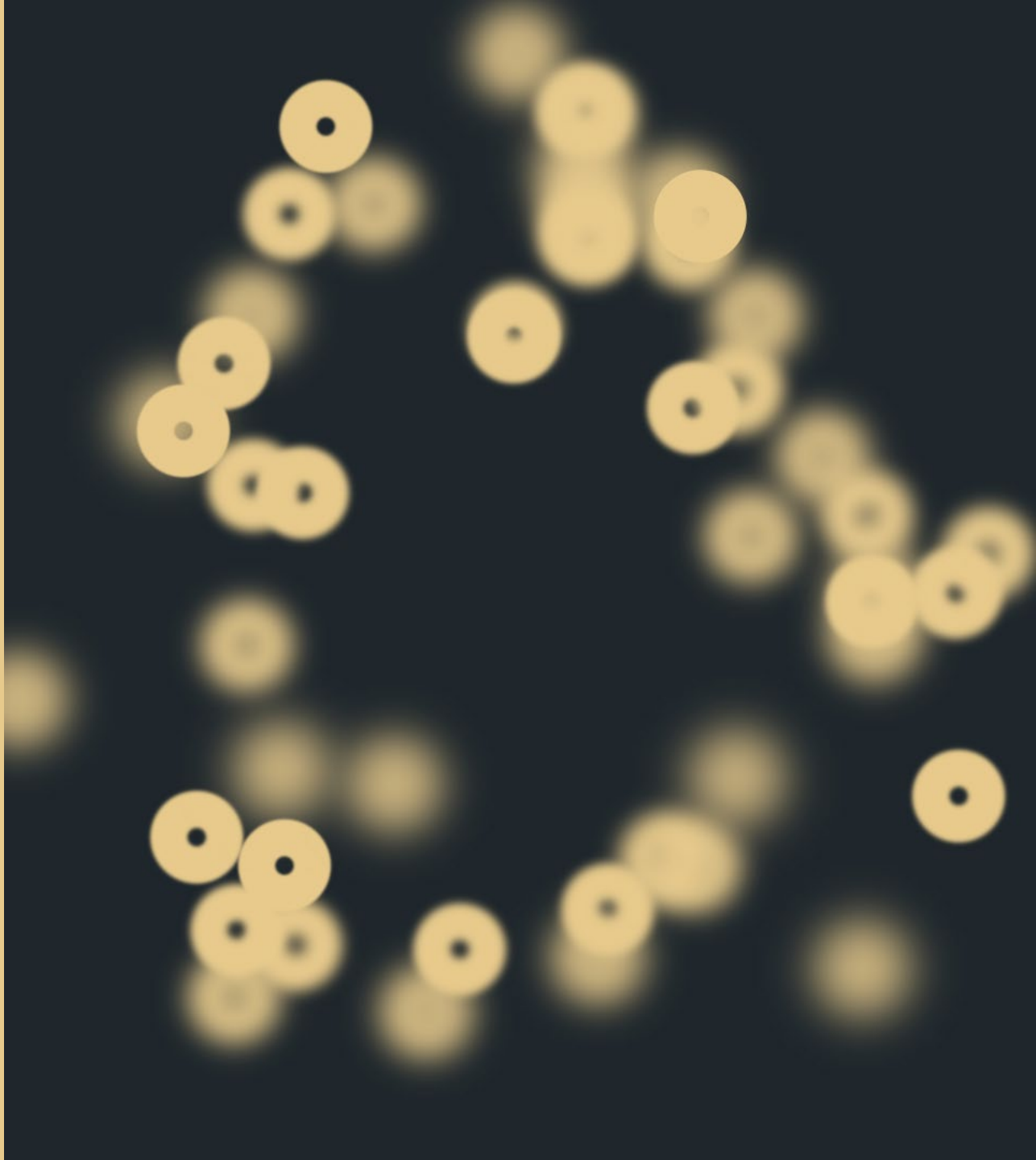
25.04.2025



Contents

CONFLICT OF INTERESTS

There are no conflicts of interest to declare.

INTRODUCTION

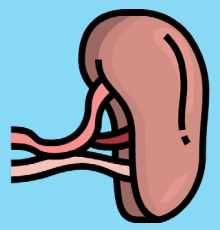
- Primary myelofibrosis carries a 9-12% risk of transformation to acute leukemia.
- The vast majority develop acute myeloid leukemia (AML), while progression of MPNs to acute lymphoblastic leukemia (ALL) is episodic.
- Only 21 cases of MPN progression to ALL have been reported, but the subsequent relapse to AML has not yet been published.

CASE REPORT - diagnosis

- A 50-year-old male patient was admitted to the hospital due to anemia, thrombocytopenia, and presented symptoms.
- The bone marrow biopsy revealed features of fibrosis.
- Status of genetic mutations: JAK V617F (-), CARL (-), MPL (-).
- Final diagnosis: **Triple Negative Primary Myelofibrosis.**
- First-line treatment: ruxolitinib 15 mg twice a day.

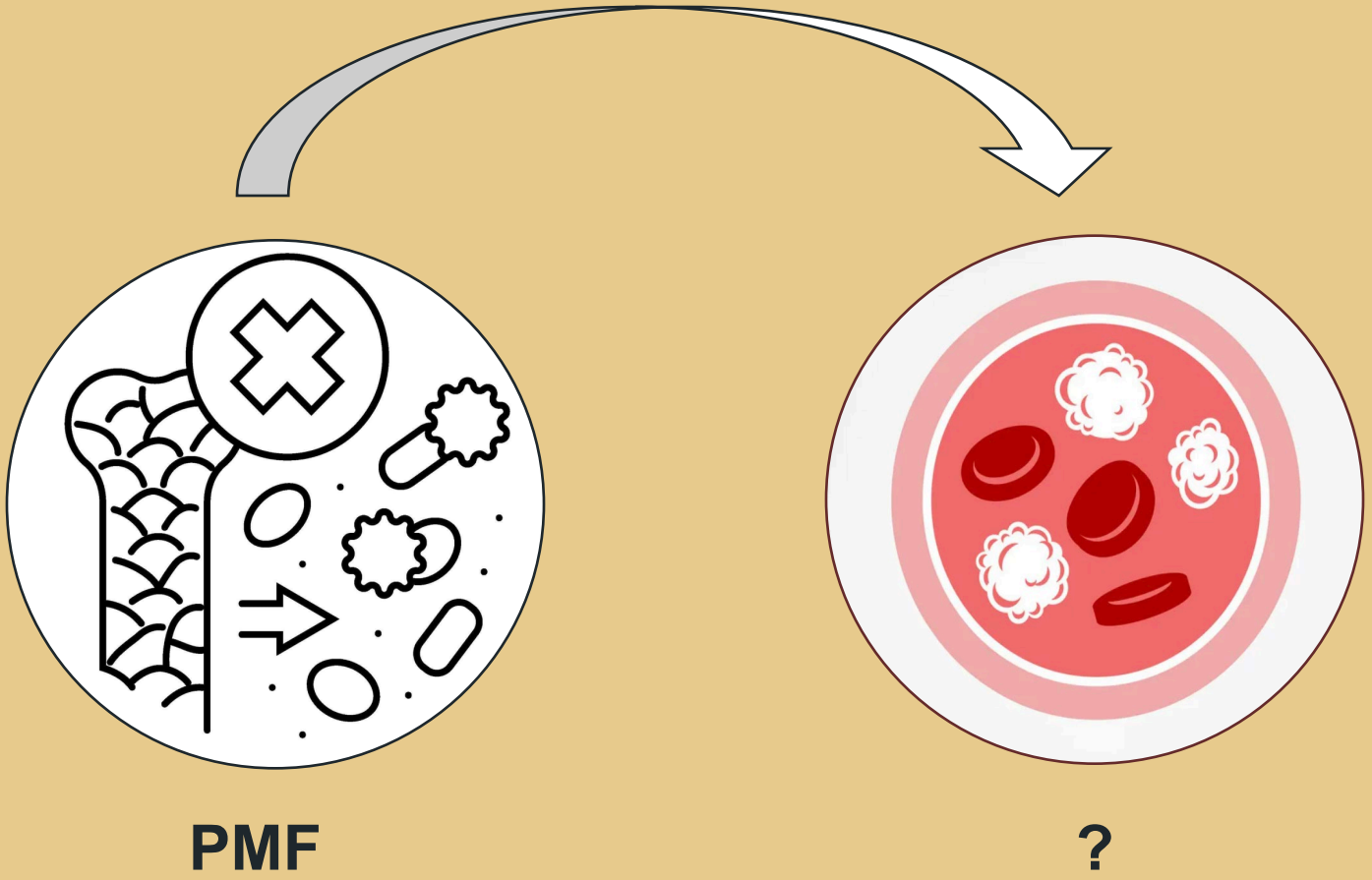
Blood tests results:
- Moderate anemia: 9 g/dl
- Moderate thrombocytopenia: 102 x 10*9/L

Symptoms:
- Weight loss (8 kg in 3 months)
- Fatigue
- Night sweats



CT scan results:
- Splenomegaly (20 cm)

TRANSFORMATION



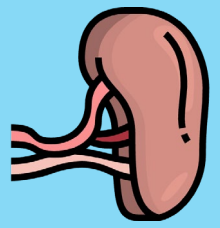
Swing Between Two Lineages.
PMF Transformation to ALL and Subsequent Relapse of AML

CASE REPORT - transformation to ALL

- Three months later, the patient was admitted to the hospital due to suspicion of transformation to acute leukemia.
- Bone marrow immunophenotyping results: 68% of lymphoblasts: CD19+, CD38+, cyCD22+, CD22+, CD10+, TdT+, CD20+/-, HLA-DR+, cyCD79a+, CD9+, CD81+, CD24+, CD52-, CD34-.
- **Diagnosis:** ALL Ph(-).
- **Induction therapy:** hyperCVAD.
- **Treatment response**: complete remission.
- Successful unrelated donor alloHSCT following non-myeloablative conditioning with fludarabine and total body irradiation.

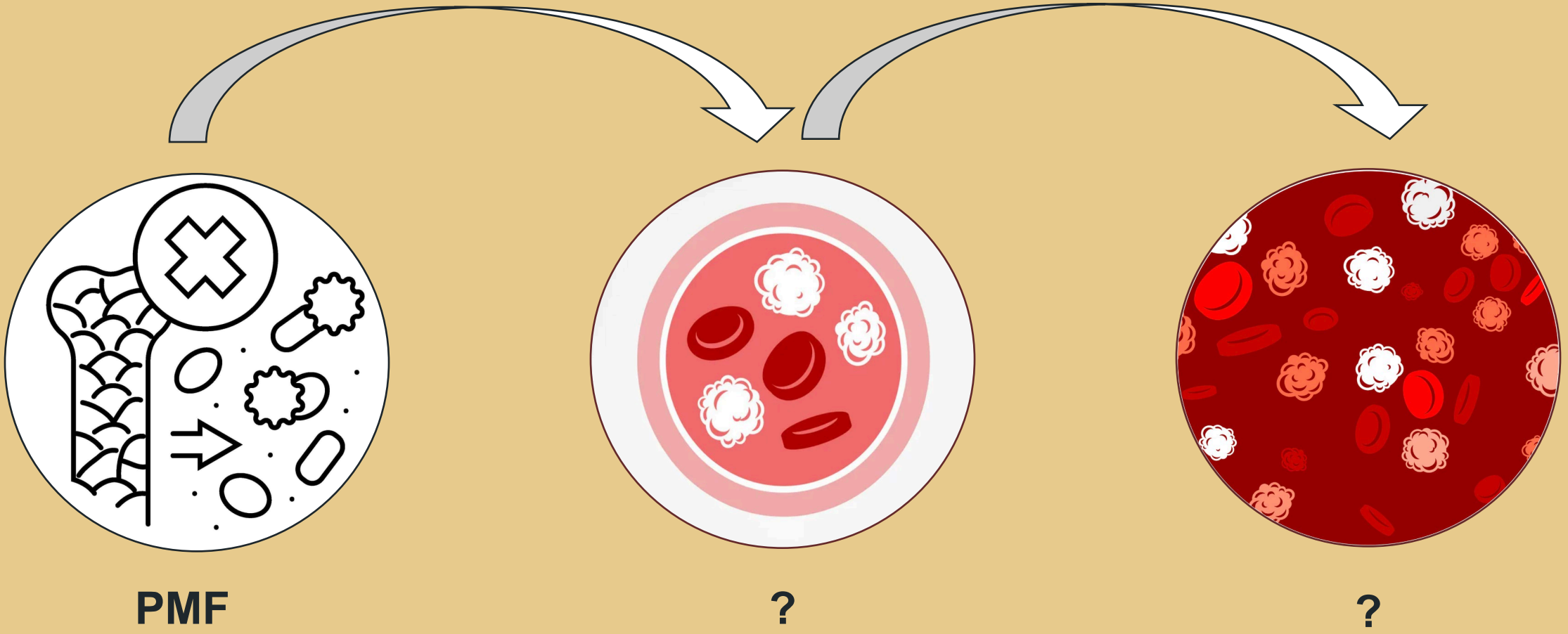
Blood tests results:
- Leukocytosis: 34,6 x 10*9/L
- Severe anemia: 6,7 g/dl
- Severe thrombocytopenia: 28 x 10*9/L

Symptoms:
- Fevers for a week
- Fatigue



CT scan results:
- No reduction of splenomegaly (20 cm)

RELAPSE



Swing Between Two Lineages.
PMF Transformation to ALL and Subsequent Relapse of AML

CASE REPORT - the relapse of AML

- At the 30th day post-alloHSCT, still no regeneration of thrombopoiesis or erythropoiesis was observed, along with increasing leukocytosis.
- Bone marrow immunophenotyping results: 21% blasts, CD13+/-, CD38+, CD33+, CD117+, MPO-, CD14-). No signal of the previous blast phenotype.
- **Diagnosis:** AML
- **Treatment:** palliative care

Blood tests results:
- Leukocytosis: 14,8 x 10*9/L
- Severe anemia: 6,2 g/
- Severe thrombocytopenia: 12 x 10*9/L

DISCUSSION

Our hypothesis:

- The alternative impairment of the JAK/STAT pathway may involve both myeloid and lymphoid lineages. It was reported that therapy with JAK1/2 inhibitors may increase the risk of the development of aggressive lymphomas.
- Lack of myeloid immunophenotype information at the time of ALL - a possible omission of mixed phenotype acute leukemia.
- Lineage switch has been reported for ALL, especially for BCR::ABL and KMT2A rearrangement carriers. Although the patient was diagnosed with ALL Ph negative, the lack of further NGS examination complicates the analysis of this case.
- Eventually, B-ALL might develop separately from myeloid disorders.

Thank you for your attention!