Thrombo-Guide
vom Labor zur Diagnose

Hematological Systemic Diseases

Description

Hematologic systemic diseases affect the hematopoietic bone marrow. This leads to a disruption of hematopoiesis and usually to alterations in all cell lines (erythrocytes, platelets, and leukocytes). Malignant hematologic diseases are characterized by the uncontrolled proliferation of a mutated cell clone and the displacement of other cell lines. In the differential blood count and bone marrow analyses, the malignant cells are usually detectable. Due to increased cell turnover, LDH and uric acid are also frequently elevated in the serum. Examples include acute and chronic leukemias (myeloid or lymphoid), lymphomas (various subtypes), myelodysplastic syndromes, myelofibrosis, and multiple myeloma.
Non-malignant hematologic diseases sometimes affect only a single cell line and are not always clearly diagnosable. Examples include aplastic anemia (mild or severe form), paroxysmal nocturnal hemoglobinuria (PNH), or various forms of anemia.

Diagnostics

A presumptive diagnosis can be made based on the complete blood count (including reticulocyte count and differential blood count). Leukocyte typing (via flow cytometry – FACS) may also be necessary. A bone marrow examination serves to confirm the diagnosis. Usually, a posterior superior iliac crest biopsy is performed under local anesthesia. A skin incision is made over the posterior superior iliac spine, and a bone sample of approx. 2 cm in length is taken using a special biopsy needle (Jamshidi needle). This sample is fixed in Schaffer's solution, then decalcified in the laboratory, embedded, and cut into thin sections using a microtome. These preparations can be stained and microscopically evaluated, or analyzed for staining behavior after the addition of antibody solutions or chemical reagents. Molecular methods can also be applied to these specimens.
Additionally, liquid bone marrow is aspirated via the biopsy needle. A drop is smeared onto glass slides, stained in the laboratory, and examined using cytochemical, immunological, or molecular methods. Cellular immunophenotyping (FACS), cytogenetic, and molecular investigations—frequently utilizing polymerase chain reaction (PCR) and sequencing—are performed on liquid, anticoagulated bone marrow. An example video showing the conduction of a bone marrow biopsy can be found at:
https://youtu.be/3RgGmErO50g?si=yOBP9iV0Mnbi7qJ4

Therapeutic Options

Depending on the disease, specific therapy may include cytostatic agents, other immunosuppressants, antibodies, biologicals, or transfusions. Details can be found at:
https://www.onkopedia.com/de/onkopedia/guidelines#section1

Monitoring

Regular monitoring of the complete blood count as well as disease-specific markers.

In case of questions or ambiguities, contact a specialized hematology department.

References

Thomas L, Labor und Diagnose, 2023, Release 5: https://www.labor-und-diagnose.de/index.html
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Onkopedia: https://www.onkopedia.com/de/onkopedia/guidelines#section1