Chromosome Analysis for Blood and Bone Marrow Cancers

Introduction

Chromosome analysis of bone marrow is addressed by this guideline.

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan’s procedure code list for management requirements.

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<th>Procedures addressed by this guideline</th>
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What are chromosome abnormalities in cancer

Introduction

A chromosome abnormality is any difference in the structure, arrangement, or amount of genetic material packaged into the chromosomes. Chromosome abnormalities have been identified in many types of cancer, including leukemias, lymphomas, and solid tumors.

Chromosome abnormalities

Chromosome abnormalities can include

- deletions
- duplications
- balanced or unbalanced rearrangements, and
- gain or loss of whole or partial chromosomes.

Some chromosome abnormalities are characteristic of certain types of malignancy, and can be used to classify a type or subtype of cancer. For example, the “Philadelphia
“chromosome” is defined by a common translocation between chromosomes 9 and 22, and indicates chronic myelogenous leukemia in most cases.¹

**Disease monitoring and treatment response**

These abnormalities can play a key role in the development, diagnosis, and monitoring of cancer.¹

The cytogenetics of a cancer can also change over time or in response to treatment. Therefore, chromosome analysis can be used to monitor disease progression and treatment response.¹

**Test information**

**Introduction**

Chromosome analysis is routinely performed on bone marrow for the diagnosis and monitoring of leukemia, lymphoma, and other hematological disorders. Lymph node is preferred for analysis of lymphoma, as bone marrow is not always involved.

**Chromosome analysis**

Chromosome analysis (karyotyping) requires arresting cell division at metaphase when the chromosomes can be seen microscopically, and staining to visualize the banding patterns.²

Chromosome analysis identifies any differences from normal that can be seen under the microscope. This includes all of the following:

- entire missing or extra chromosomes
- deletions or duplications within a chromosome that are large enough to be seen by microscope, and
- rearrangements including translocations and inversions.

**Guidelines and evidence**

**Introduction**

This section includes relevant guidelines and evidence pertaining to chromosome analysis of bone marrow.

**National Comprehensive Cancer Network**

The National Comprehensive Cancer Network (NCCN) considers chromosome analysis of a bone marrow biopsy to be routine standard of care in the evaluation of
acute myeloid leukemia (AML), chronic myelogenous leukemia (CML), multiple myeloma (MM), myelodysplastic syndromes (MDS), and Burkitt's lymphoma (BL).³

**American College of Medical Genetics**

The American College of Medical Genetics (ACMG, 2016) provides technical standards and guidelines for chromosome analysis in acquired chromosomal abnormalities of blood and bone marrow:⁴

- Bone marrow is the preferred specimen for cytogenetic analysis of hematopoetic neoplasms.
- “Cytogenetic analyses of hematological neoplasms are performed to detect and characterize clonal chromosomal abnormalities that have important diagnostic, prognostic, and therapeutic implications.”
- “Furthermore, cytogenetic analysis can provide crucial information regarding specific genetically defined subtypes of these neoplasms that have targeted therapies.”
- “At time of relapse, cytogenetic analysis can be used to confirm recurrence of the original neoplasm, detect clonal disease evolution, or uncover a new unrelated neoplastic process.”
- Acute Myeloid Leukemia: “G-banded chromosome analysis should preferably be performed first. However, interphase FISH analysis for KMT2A (MALL) gene rearrangement is highly recommended on all diagnostic AML samples because these abnormalities are often cryptic and have a pronounced prognostic impact.”
- Acute Lymphocytic Leukemia: “B-lineage ALL is more frequent, accounting for 85% of pediatric ALL and 75% of adult ALL.¹ - In pediatric/young adult B-lineage ALL, G-banded chromosome analysis should be performed simultaneously with interphase FISH.”

**Criteria**

Chromosome analysis on bone marrow is considered medically necessary when performed in the evaluation of leukemia, lymphoma, and other hematological disorders.

**References**

**Introduction**

These references are cited in this guideline.


4. American College of Medical Genetics. Section E6.1-6.4 of the ACMG technical standards and guidelines: chromosome studies of neoplastic blood and bone marrow – acquired chromosomal abnormalities. 2016. Available at https://pdfs.semanticscholar.org/34bc/c7ce2ff76bf7637f8c5b9892f823cbd0f68e.pdf