AI 輔助翻譯與醫療參考提醒

解讀我的骨髓瘤檢驗報告：詳細版中英對照

閱讀前請先留意

本 PDF 由 AI 協助翻譯、整理與重排，目的是協助病友與家屬對照英文原文，理解多發性骨髓瘤相關檢驗資訊。

本資料僅供一般資訊參考，不能取代醫師診斷、治療建議、用藥調整或個別化醫療判斷。

不同醫院、不同實驗室的檢驗方法與參考區間可能不同；若檢驗數值偏高、偏低或有疑問，請以您的主治醫師說明為準。

若您打算分享本 PDF，請一併保留本提醒頁，避免下載者誤以為這是正式醫囑或個人化治療建議。

資料來源：Deciphering Myeloma Labs；本頁為繁體中文 AI 輔助翻譯與閱讀版重排。
網站：https://healthtreetw.org/posts/know-05/

DECIPHERING MY MYELOMA LABS
解讀我的骨髓瘤檢驗報告

Do you understand your myeloma diagnosis and your myeloma lab results?

您了解自己的骨髓瘤診斷與骨髓瘤檢驗結果嗎？

This guide attempts to simplify the complex process of understanding your myeloma markers and helps you track your treatment history. Based on the actual lab printouts you receive in the clinic, we have added color-coding to help you identify the most important myeloma markers.

本指南嘗試簡化理解骨髓瘤指標的複雜過程，並協助您追蹤治療歷史。根據您在門診收到的實際檢驗報告，我們加入顏色標示，幫助您辨識最重要的骨髓瘤指標。

Priority 優先關注	Secondary 次要關注	Not as important 較不重要

Special thanks to myeloma specialist Dr. Guido Tricot of the University of Iowa, pathologist Dr. Michael Misialek of Newton-Wellesley Hospital, Jen Higbee of Huntsman Cancer Institute, and Barbara Waagen for their contributions to this document.

特別感謝愛荷華大學骨髓瘤專家 Guido Tricot 醫師、Newton-Wellesley Hospital 病理醫師 Michael Misialek、Huntsman Cancer Institute 的 Jen Higbee，以及 Barbara Waagen 對本文件的貢獻。

Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established reference range.

請注意，所列參考範圍不一定在各實驗室之間完全一致。每個實驗室都必須建立自己的參考範圍。若檢驗結果超出既定參考範圍，異常結果必須標示為偏高、偏低或危急值。

Each section is linked to a HealthTree University Class. Feel free to click on the links in each section for further information.

每個章節都連結到 HealthTree University 課程。您可以點選各章節中的連結，了解更多資訊。

Do you have suggestions to make this document better? Send your comments to support@healthtree.org, and we will keep revising this document as we learn more.

您是否有讓本文件更完善的建議？請將意見寄至 support@healthtree.org；我們會隨著持續學習而修訂本文件。

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AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. INDEX - My Myeloma Summary; R-ISS; Mayo clinic risk stratification (mSMART); My Myeloma Labs.
   索引 - 我的骨髓瘤摘要；修訂版國際分期系統（R-ISS）；Mayo Clinic 多發性骨髓瘤風險分層（mSMART）；我的骨髓瘤檢驗。

2. Monoclonal proteins; SPEP; immunofixation; serum free light chains; UPEP; immunoglobulins; bone marrow biopsy.
   單株蛋白；血清蛋白電泳（SPEP）；免疫固定；血清游離輕鏈；尿蛋白電泳（UPEP）；免疫球蛋白；骨髓抽吸與切片。

3. Chemistry panels; complete blood count; coagulation; immunology/allergens; toxicology/drug levels; pulmonary function tests.
   血液生化檢查；全血球計數；凝血檢查；免疫/過敏原；毒物學/藥物濃度；肺功能檢查。

4. Imaging tests; genetics; cytogenetics/FISH; flow cytometry; NGS; MRD; GEP; SNP array; specific myeloma abnormalities.
   影像檢查；遺傳學；細胞遺傳學/FISH；流式細胞術；次世代定序（NGS）；微小殘留病灶（MRD）；基因表現分析（GEP）；SNP 陣列；骨髓瘤特定遺傳異常。

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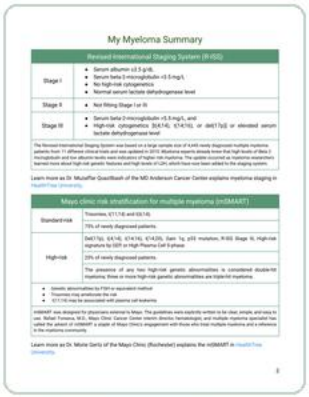

My Myeloma Summary

Revised-International Staging System (R-ISS)

Stage I	• Serum albumin ≥3.5 g/dL • Serum beta-2-microglobulin <3.5 mg/L • No high-risk cytogenetics • Normal serum lactate dehydrogenase level
Stage II	• Not fitting Stage I or III
Stage III	• Serum beta-2-microglobulin >5.5 mg/L, and • High-risk cytogenetics [t(4;14), t(14;16), or del(17p)] or elevated serum lactate dehydrogenase level

The Revised-International Staging System was based on a large sample size of 4,445 newly diagnosed multiple myeloma patients from 11 different clinical trials and was updated in 2015. Myeloma experts already know that high levels of Beta-2-microglobulin and low albumin levels were indicators of higher-risk myeloma. The update occurred as myeloma researchers learned more about high-risk genetic features and high levels of LDH, which have now been added to the staging system.

Learn more as Dr. Muzaffar Qazilbash of the MD Anderson Cancer Center explains myeloma staging in HealthTree University.

Mayo clinic risk stratification for multiple myeloma (mSMART)

Standard-risk	Trisomies, t(11;14) and t(6;14) 75% of newly diagnosed patients.
High-risk	Del(17p), t(4;14), t(14;16), t(14;20), Gain 1q, p53 mutation, R-ISS Stage III, High-risk signature by GEP or High Plasma Cell S-phase 25% of newly diagnosed patients. The presence of any two high-risk genetic abnormalities is considered double-hit myeloma, three or more high-risk genetic abnormalities are triple-hit myeloma.

• Genetic abnormalities by FISH or equivalent method
• Trisomies may ameliorate the risk
• IGH@ may be associated with plasma cell leukemia

mSMART was designed for physicians external to Mayo. The guidelines were explicitly written to be clear, simple, and easy to use. Rafael Fonseca, M.D., Mayo Clinic Cancer Center interim director, hematologist, and multiple myeloma specialist has called the advent of mSMART a staple of Mayo Clinic's engagement with those who treat multiple myeloma and a reference in the myeloma community.

Learn more as Dr. Morie Gertz of the Mayo Clinic (Rochester) explains the mSMART in HealthTree University.

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原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

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1. Revised-International Staging System (R-ISS): Stage I requires albumin >=3.5 g/dL, beta-2-microglobulin <3.5 mg/L, no high-risk cytogenetics, and normal LDH.
   修訂版國際分期系統（R-ISS）：第一期需符合白蛋白 >=3.5 g/dL、β2-微球蛋白 <3.5 mg/L、無高風險細胞遺傳異常，以及 LDH 正常。

2. Stage II means the findings do not fit Stage I or Stage III.
   第二期表示檢查結果不符合第一期或第三期。

3. Stage III includes beta-2-microglobulin >5.5 mg/L plus high-risk cytogenetics or elevated serum LDH.
   第三期包含 β2-微球蛋白 >5.5 mg/L，並合併高風險細胞遺傳異常或血清 LDH 升高。

4. R-ISS was updated in 2015 using 4,445 newly diagnosed patients; it added high-risk genetics and LDH to known albumin and beta-2-microglobulin risk markers.
   R-ISS 於 2015 年依據 4,445 位新診斷病人更新；除了既有的白蛋白與 β2-微球蛋白風險指標，也納入高風險遺傳特徵與 LDH。

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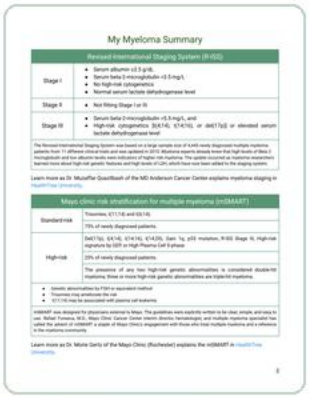

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5 mSMART standard-risk: trisomies, t(11;14), and t(6;14), about 75% of newly diagnosed patients.
mSMART 標準風險：三體、t(11;14) 與 t(6;14)，約佔新診斷病人的 75%。

6 mSMART high-risk: del(17p), t(4;14), t(14;16), t(14;20), gain 1q, p53 mutation, R-ISS Stage III, high-risk GEP signature, or high plasma-cell S-phase.
mSMART 高風險：del(17p)、t(4;14)、t(14;16)、t(14;20)、1q 增益、p53 突變、R-ISS 第三期、高風險 GEP 特徵，或漿細胞 S 期比例高。

7 Two high-risk genetic abnormalities are considered double-hit myeloma; three or more are triple-hit myeloma.
若有兩項高風險遺傳異常，稱為 double-hit 骨髓瘤；三項以上稱為 triple-hit 骨髓瘤。

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My Myeloma Labs

MONOCLONAL PROTEINS

Multiple myeloma is a blood cancer of white blood cells called plasma cells. Plasma cells come from the bone marrow and produce antibodies (also called immunoglobulins) that fight a wide variety of infections. In myeloma, one of these antibodies grows out of control in the bone marrow, crowding out the other antibodies and other components of the immune system, making too much of one type and reducing the ability for your body to create a broad spectrum of immunoglobulins to fight infections. This is called a monoclonal protein (also called M-protein or M-spike).

Serum protein electrophoresis (SPEP) is a test used to find any abnormal immunoglobulin. Then, another test, such as immunofixation or immunoelectrophoresis, is used to determine the exact type of abnormal antibody (IgG or some other type). This abnormal protein is known by several different names, including monoclonal immunoglobulin, M protein, M spike, and paraprotein.

Antibodies are made up of two parts: heavy chains and light chains. The types of monoclonal proteins are defined by these parts, although sometimes the heavy chain might be missing.

Videos explained by Myeloma specialists about this topic:

- What is the normal structure of an immunoglobulin (antibody)?
- What is a monoclonal protein (or M-spike) and how is it detected?
- What are the different types of myeloma?

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1. Multiple myeloma is a blood cancer of plasma cells, which arise from bone marrow and normally produce antibodies.
   多發性骨髓瘤是漿細胞的血液癌症；漿細胞源自骨髓，正常功能是製造抗體。

2. In myeloma, one antibody clone grows out of control, crowds out other immune components, and reduces the body's broad antibody defense.
   在骨髓瘤中，某一株抗體細胞不受控制地增生，排擠其他免疫成分，使身體產生多樣抗體以抵抗感染的能力下降。

3. This abnormal antibody is called a monoclonal protein, also known as M-protein or M-spike.
   這種異常抗體稱為單株蛋白，也稱 M 蛋白或 M 尖峰。

4. SPEP looks for abnormal immunoglobulin; immunofixation or immunoelectrophoresis then identifies the exact antibody type.
   SPEP 用來尋找異常免疫球蛋白；之後可用免疫固定或免疫電泳確認異常抗體的確切類型。

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5 Antibodies have heavy chains and light chains; monoclonal protein types are defined by these parts, though heavy chains may sometimes be absent.
抗體由重鏈與輕鏈組成；單株蛋白的類型由這些部分定義，但有時可能缺少重鏈。

6 Related videos explain normal immunoglobulin structure, monoclonal protein detection, and different types of myeloma.
相關影片說明正常免疫球蛋白結構、單株蛋白的偵測方式，以及不同骨髓瘤類型。

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1. SPEP separates serum proteins into albumin, alpha-1, alpha-2, beta, and gamma fractions, and can detect M-protein.
   SPEP 將血清蛋白分成白蛋白、α1、α2、β 與 γ 區段，並可偵測 M 蛋白。

2. Total protein measures the total protein amount in a blood sample.
   總蛋白測量血液樣本中的蛋白總量。

3. Albumin helps keep blood inside vessels and supports tissue growth/healing; low values may suggest malnutrition, kidney or liver disease, inflammation, or protein loss; high values may suggest dehydration.
   白蛋白有助於維持血液留在血管內，並支持組織生長與修復；低值可能提示營養不良、腎臟或肝臟疾病、發炎或蛋白流失；高值可能提示脫水。

4. Alpha-1 and alpha-2 changes may reflect severe liver disease, malnutrition, red blood cell breakdown, kidney disease, or acute/chronic inflammation.
   α1 與 α2 的變化可能反映嚴重肝病、營養不良、紅血球分解、腎臟疾病，或急性/慢性發炎。

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5 Beta globulins carry substances such as iron and help fight infection; abnormal values may be seen with malnutrition, liver fibrosis, high cholesterol, iron-deficiency anemia, MGUS, or multiple myeloma.

β 球蛋白協助運送鐵等物質並幫助抵抗感染；異常值可見於營養不良、肝纖維化、高膽固醇、缺鐵性貧血、MGUS 或多發性骨髓瘤。

6 Gamma proteins are antibodies; low values may indicate immune disorders, while high polyclonal or monoclonal patterns can point to inflammation, liver disease, MM, lymphoma, or Waldenstrom macroglobulinemia.

γ 蛋白即抗體；低值可能表示免疫疾病，高的多株或單株型態則可能指向發炎、肝病、多發性骨髓瘤、淋巴瘤或 Waldenstrom 巨球蛋白血症。

7 Paraprotein/M-spike is the monoclonal band; SPEP/UPEP measure amount, while immunofixation identifies type. Daratumumab can interfere with IgG kappa IFE interpretation.

副蛋白/M 尖峰是單株蛋白帶；SPEP/UPEP 測量其量，免疫固定確認其類型。Daratumumab 可能影響 IgG kappa 病人的 IFE 判讀。

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1. Serum free light chain test: kappa and lambda are the two light-chain types; the involved chain is followed in kappa or lambda myeloma.
   血清游離輕鏈檢查：kappa 與 lambda 是兩種輕鏈；若為 kappa 或 lambda 型骨髓瘤，會追蹤相對應的輕鏈數值。

2. As myeloma progresses, cells may produce more light chains than heavy chains; higher free light chains can indicate more aggressive disease.
   骨髓瘤進展時，細胞可能製造比重鏈更多的輕鏈；游離輕鏈越高，可能代表疾病越具侵襲性。

3. The kappa/lambda ratio reflects balance between light chains and can help indicate disease progression or remission.
   kappa/lambda 比值反映兩種輕鏈的平衡，可協助判斷疾病進展或緩解。

4. HealthTree classes cover antibody structure, light-chain reporting, measuring free light chains, and why the ratio matters.
   HealthTree 課程涵蓋抗體結構、輕鏈報告方式、游離輕鏈測量，以及為何比值重要。

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SERUM FREE LIGHT CHAIN TEST

LAB TESTED	REFERENCE RANGE	WHAT IT MEANS
KAPPA FREE LIGHT CHAINS	3.3 - 19.4 mg/L	There are two types of light chains kappa and lambda. If a patient has kappa myeloma, their doctor will watch for a rise in the kappa numbers. Likewise, the lambda number will be observed if a patient has lambda myeloma.
LAMBDA FREE LIGHT CHAINS	5.7 - 26.3 mg/L	When myeloma progresses, the myeloma cells start to produce more light chains than heavy chains. This can be measured by the Free Light Chain Assay test on a blood specimen. The higher the free light chains, the more aggressive the disease is. Therefore, the serum free light chain test is a better predictor of outcome than the amount of M-protein in the blood.
KAPPA/LAMBDA RATIO	0.26 to 1.65	The balance of kappa and lambda together is called the kappa/lambda ratio, which can also indicate a change in levels of diseases. "The ratio or proportion between the kappa and lambda light chains indicates an excess production of one chain over the other, and therefore can be used to indicate disease progression or remission," said Dr. Christina Gasparetto of Duke University.

HEALTHTREE UNIVERSITY CLASSES

- What is the normal structure of an immunoglobulin (antibody)?
- What are light chains, how are light chains reported, and why?
- How do you measure free light chains?
- Why is knowing your free light chain ratio important?

Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established normal range.

URINE PROTEIN ELECTROPHORESIS (URINE TEST)

The UPEP is generally performed on a single urine sample. Bence-Jones proteins will be detected if present. A routine urinalysis will not detect Bence-Jones proteins.

LAB TESTED	NORMAL RANGE (mg/day)	WHAT IT MEANS
URINE TOTAL PROTEIN (UPEP)	<226	Measures the total amount of protein in a urine sample of 24 hours.

5

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5 UPEP is usually performed on a urine sample; Bence-Jones proteins can be detected by UPEP but not by routine urinalysis.
UPEP 通常使用尿液樣本；Bence-Jones 蛋白可由 UPEP 偵測，但例行尿液分析通常偵測不到。

6 Urine total protein measures the total amount of protein in a 24-hour urine sample.
尿液總蛋白測量 24 小時尿液樣本中的蛋白總量。

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Urine albumin has similar meaning to serum albumin: low values may reflect malnutrition, kidney or liver disease, inflammation, or protein loss; high values may suggest dehydration.
   尿中白蛋白的意義與血清白蛋白相近：低值可能反映營養不良、腎臟或肝臟疾病、發炎或蛋白流失；高值可能提示脫水。

2. Urine globulin, alpha-1, alpha-2, and beta globulin patterns may help identify inflammation, liver disease, kidney disease, malnutrition, iron-deficiency anemia, MGUS, or MM.
   尿中球蛋白、α1、α2 與 β 球蛋白型態，可協助辨識發炎、肝病、腎病、營養不良、缺鐵性貧血、MGUS 或多發性骨髓瘤。

3. Urine paraprotein percent and urine M-protein describe monoclonal protein in urine, often called M-spike, reported as percent or mg/day.
   尿中副蛋白百分比與尿中 M 蛋白描述尿液中的單株蛋白，常稱 M 尖峰，可用百分比或 mg/day 表示。

4. Urine immunofixation is usually done on a 24-hour urine collection to measure Bence-Jones proteins and monitor treatment progress.
   尿液免疫固定通常以 24 小時尿液收集進行，用來測量 Bence-Jones 蛋白並監測治療進展。

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5 Urine free kappa/lambda light chains may detect myeloma earlier than UPEP, SPEP, or IFE, but those tests are still needed for the complete picture.
尿中游離 kappa/lambda 輕鏈可能比 UPEP、SPEP 或 IFE 更早偵測骨髓瘤，但仍需搭配其他檢查才能完整評估。

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中英重點對照

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1. Quantitative immunoglobulins measure IgG, IgA, IgM, IgD, and IgE levels in blood.
   定量免疫球蛋白測量血液中的 IgG、IgA、IgM、IgD 與 IgE 濃度。

2. In multiple myeloma, one immunoglobulin type overgrows and crowds out others, which can increase susceptibility to infections such as pneumonia.
   在多發性骨髓瘤中，某一類免疫球蛋白過度增生並排擠其他類型，可能增加肺炎等感染風險。

3. Elevated immunoglobulins unrelated to a person's myeloma may indicate current infection.
   若免疫球蛋白升高與個人的骨髓瘤無關，可能表示目前有感染。

4. Bone marrow aspiration and core biopsy show how many marrow cells are myeloma cells and whether the specimen quality is adequate.
   骨髓抽吸與核心切片可顯示骨髓中有多少細胞為骨髓瘤細胞，以及檢體品質是否足夠。

5. Immunohistochemistry uses antibodies that bind specific cell-surface molecules to help identify cell types.
   免疫組織化學使用會結合特定細胞表面分子的抗體，以協助辨識不同細胞類型。

6. Plasma-cell percentage and appearance on aspiration or biopsy help classify plasma-cell disorders and should be followed over time.
   抽吸或切片中的漿細胞百分比與外觀，有助於分類漿細胞疾病，並應隨時間追蹤。

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中英重點對照

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7 Rouleaux are stacked red blood cells and may occur with infection, multiple myeloma, Waldenstrom macroglobulinemia, and inflammatory or connective-tissue disorders.

紅血球錢串狀排列（rouleaux）是紅血球堆疊，可見於感染、多發性骨髓瘤、Waldenstrom 巨球蛋白血症，以及發炎或結締組織疾病。

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1. Some marrow report items, such as leukocyte number, platelet number, and many differential-count categories, are listed as not relevant for the myeloma assessment.
   骨髓報告中的某些項目，例如白血球數、血小板數與多項分類計數，原文列為與骨髓瘤評估不相關。

2. Circulating plasma cells are generally a poor prognostic factor.
   周邊循環漿細胞通常是不良預後因子。

3. Specimen quality indicates how representative the sample is; terms such as hemodiluted imply blood contamination.
   檢體品質表示樣本是否能代表骨髓狀態；例如 hemodiluted 表示可能受到血液污染。

4. Cellularity is reported as a percentage; look for normal, hypocellular, or hypercellular descriptions.
   細胞密度會以百分比報告；需注意正常、低細胞性或高細胞性的描述。

5. Spicules suggest the sample is from marrow with little or no blood contamination.
   骨髓小粒（spicules）的存在表示樣本來自骨髓，血液污染少或沒有污染。

6. Myeloid-to-erythroid ratio measures developing white-cell to red-cell lines.
   髓系與紅系比例（M:E ratio）衡量發育中白血球系與紅血球系的比例。

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7 Plasma-cell number and morphology are important; mature cells are generally a better prognostic feature than immature or atypical cells.
漿細胞數量與形態很重要；成熟漿細胞通常比未成熟或異型漿細胞代表較佳預後。

8 Core biopsy adequacy depends partly on size; bone trabeculae should be present to show the sample represents marrow.
核心切片是否足夠部分取決於大小；應可見骨小樑，代表樣本確實來自骨髓。

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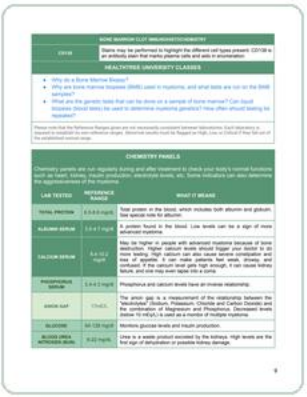

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中英重點對照

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1. CD138 staining highlights plasma cells and helps count them in bone marrow clot immunohistochemistry.
   CD138 染色可標示漿細胞，並協助在骨髓凝塊免疫組織化學中計數。

2. HealthTree classes discuss why bone marrow biopsies are done and what tests may be run on marrow samples.
   HealthTree 課程說明為何進行骨髓切片，以及骨髓樣本可進行哪些檢查。

3. Chemistry panels are repeated during and after treatment to monitor heart, kidney, insulin production, electrolytes, and other normal body functions.
   血液生化檢查會在治療期間與治療後反覆進行，以監測心臟、腎臟、胰島素產生、電解質與其他身體功能。

4. Total protein includes albumin and globulin. Low serum albumin can be a sign of more advanced myeloma.
   總蛋白包含白蛋白與球蛋白。血清白蛋白偏低可能是骨髓瘤較進展的徵象。

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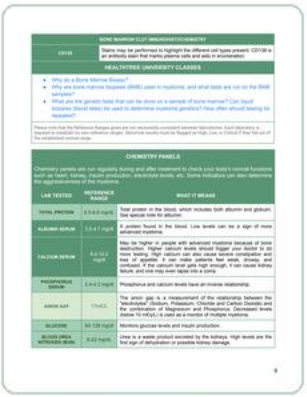

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5 Serum calcium may rise in advanced myeloma due to bone destruction; high calcium should prompt additional testing and can cause constipation, appetite loss, weakness, drowsiness, confusion, kidney failure, or coma.
晚期骨髓瘤可能因骨破壞造成血鈣升高；高血鈣應促使醫師進一步檢查，並可能造成便秘、食慾下降、虛弱、嗜睡、意識混亂、腎衰竭或昏迷。

6 Phosphorus and calcium have an inverse relationship; anion gap and glucose help monitor electrolytes and insulin production.
磷與鈣呈反向關係；陰離子間隙與血糖可協助監測電解質與胰島素產生。

7 BUN is a kidney-excreted waste product; high levels may be the first sign of dehydration or possible kidney damage.
BUN 是由腎臟排出的廢物；升高可能是脫水或潛在腎損傷的早期徵象。

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1. Creatinine is a kidney-excreted waste product; elevated levels may indicate poor hydration or possible kidney damage.
   肌酸酐是由腎臟排出的廢物；升高可能表示水分不足或可能的腎損傷。

2. Creatinine clearance is a gold-standard kidney-function measure comparing urine creatinine excretion with serum creatinine.
   肌酸酐清除率是腎功能的重要標準測量，會比較尿中肌酸酐排出量與血清肌酸酐。

3. Uric acid may be watched during treatment; elevation can suggest tumor lysis syndrome.
   尿酸在治療期間可能需要追蹤；升高可能提示腫瘤溶解症候群。

4. Alkaline phosphatase may be elevated in bone and liver disease.
   鹼性磷酸酶可能在骨骼或肝臟疾病中升高。

5. Beta-2-microglobulin helps assess MM severity; high levels suggest more aggressive disease, while decreases suggest good treatment response; it may also identify kidney damage.
   β2-微球蛋白有助於評估多發性骨髓瘤嚴重度；高值提示疾病較具侵襲性，下降則提示治療反應良好；也可能反映腎損傷。

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6 LDH is a nonspecific tissue-damage marker; elevated LDH in aggressive myeloma is associated with poor prognosis when no other cause explains it.
LDH 是非特異性的組織損傷指標；若無其他原因可解釋，侵襲性骨髓瘤中的 LDH 升高與較差預後相關。

7 Sodium, potassium, chloride, and carbon dioxide are electrolytes tied to acid-base balance; abnormal levels can affect the nervous system and heart.
鈉、鉀、氯與二氧化碳是與酸鹼平衡相關的電解質；異常可能影響神經系統與心臟。

8 Bilirubin and SGOT/AST help assess liver, muscle, red-cell, kidney, heart, pancreas, and treatment-related effects.
膽紅素與 SGOT/AST 可協助評估肝臟、肌肉、紅血球、腎臟、心臟、胰臟及治療相關影響。

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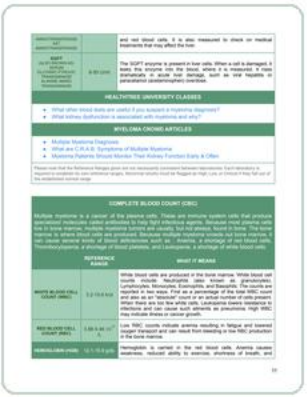

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1. SGPT/ALT is present in liver cells and rises when liver cells are damaged, including viral hepatitis or acetaminophen overdose.
   SGPT/ALT 存在於肝細胞中，肝細胞受損時會升高，例如病毒性肝炎或乙醯胺酚過量。

2. CBC section: plasma cells usually live in bone marrow, where blood cells are produced; myeloma can crowd out marrow and cause blood-cell deficiencies.
   全血球計數章節：漿細胞多位於骨髓，而骨髓也是血球生成處；骨髓瘤可能排擠骨髓並造成血球不足。

3. Possible deficiencies include anemia, thrombocytopenia, and leukopenia.
   可能出現的不足包含貧血、血小板低下與白血球低下。

4. White blood cell counts include neutrophils, lymphocytes, monocytes, eosinophils, and basophils, reported as percentages and absolute counts.
   白血球計數包含嗜中性球、淋巴球、單核球、嗜酸性球與嗜鹼性球，會以百分比與絕對數報告。

5. Too few white cells lowers infection resistance; high WBC may indicate illness or cancer growth.
   白血球過少會降低抗感染能力；白血球升高可能表示疾病或癌症增長。

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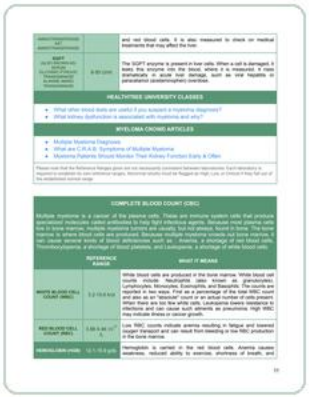

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6 Low RBC counts indicate anemia, causing fatigue and reduced oxygen transport, and may result from bleeding or low marrow production.
紅血球偏低表示貧血，會造成疲倦與氧氣運送下降，可能來自出血或骨髓製造不足。

7 Hemoglobin in red blood cells reflects anemia severity and may relate to weakness, reduced exercise ability, shortness of breath, and dizziness.
紅血球中的血紅素反映貧血嚴重度，可能與虛弱、運動耐受下降、呼吸急促與頭暈相關。

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1. Hematocrit measures the percentage of whole blood volume made up of red blood cells.
   血比容測量全血體積中紅血球所占的百分比。

2. MCV, MCH, and MCHC describe red-cell volume, hemoglobin per red cell, and hemoglobin concentration in packed red cells.
   MCV、MCH 與 MCHC 分別描述紅血球體積、單一紅血球血紅素量，以及濃縮紅血球中的血紅素濃度。

3. Platelets are produced in marrow and help clotting; low counts can make minor scrapes, cuts, or bruises cause serious bleeding.
   血小板由骨髓製造並協助凝血；數量低時，小擦傷、割傷或瘀青也可能造成嚴重出血。

4. Neutrophils are the most numerous white blood cells and fight infection by engulfing bacteria; low levels mean reduced ability to fight infection.
   嗜中性球是數量最多的白血球，會吞噬細菌以對抗感染；低值代表抗感染能力下降。

5. Lymphocytes include B cells, T cells, and natural killer cells; low levels also indicate reduced infection defense.
   淋巴球包含 B 細胞、T 細胞與自然殺手細胞；低值同樣表示抗感染防禦下降。

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6 Monocytes enter tissues, fight infection, ingest dead cells, and assist immune responses.
單核球會進入組織、對抗感染、吞噬死亡細胞並協助免疫反應。

7 Eosinophils may rise in allergic reactions and help fight some parasitic infections.
嗜酸性球可能在過敏反應中升高，並協助對抗某些寄生蟲感染。

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1. Basophils also participate in allergic reactions.
   嗜鹼性球也參與過敏反應。

2. Immature neutrophils or granulocytes may be released early from bone marrow.
   未成熟嗜中性球或顆粒球可能會提早從骨髓釋放。

3. Iron studies, ferritin, folate, TIBC, transferrin saturation, and vitamin B12 relate to red-cell production and disease monitoring.
   鐵相關檢查、鐵蛋白、葉酸、總鐵結合能力、轉鐵蛋白飽和度與維生素 B12，皆與紅血球生成及疾病監測有關。

4. TSH can be a contributing factor to anemia.
   甲狀腺刺激素（TSH）可能是貧血的促成因素。

5. HealthTree classes and Myeloma Crowd articles cover useful blood tests, plasma cell leukemia, and multiple myeloma diagnosis.
   HealthTree 課程與 Myeloma Crowd 文章涵蓋有用的血液檢查、漿細胞白血病與多發性骨髓瘤診斷。

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中英重點對照

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6 Reference ranges vary by laboratory; abnormal results should be flagged as high, low, or critical when outside that lab's range.
參考區間會因實驗室不同而異；超出該實驗室範圍時，異常結果應標示為高、低或危急值。

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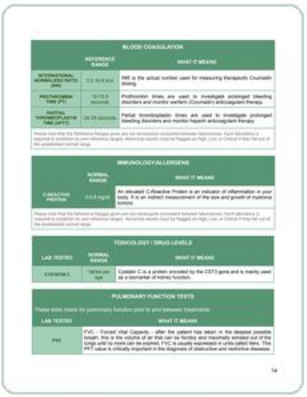

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Blood coagulation: INR is used to measure therapeutic Coumadin dosing.
   凝血檢查：INR 用於衡量 Coumadin 治療劑量。

2. Prothrombin time investigates prolonged bleeding disorders and monitors warfarin anticoagulant therapy.
   凝血酶原時間（PT）用於評估出血時間延長的疾病，並監測 warfarin 抗凝治療。

3. Partial thromboplastin time investigates prolonged bleeding disorders and monitors heparin anticoagulant therapy.
   部分凝血活酶時間（APTT）用於評估出血時間延長的疾病，並監測 heparin 抗凝治療。

4. C-reactive protein is an inflammation indicator and an indirect measure related to myeloma tumor size and growth.
   C 反應蛋白是體內發炎指標，也可作為與骨髓瘤腫瘤大小與生長相關的間接測量。

5. Cystatin C is encoded by the CST3 gene and is mainly used as a kidney-function biomarker.
   Cystatin C 由 CST3 基因編碼，主要作為腎功能生物標記。

6. Pulmonary function tests check lung function before and between treatments.
   肺功能檢查用於治療前與治療間評估肺功能。

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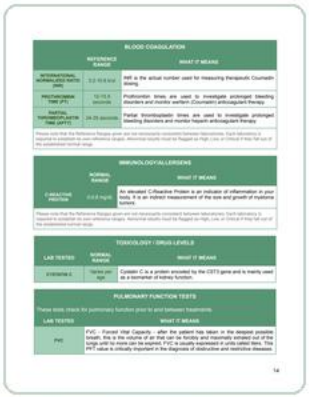

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7 FVC is forced vital capacity; FEV1 is forced expiratory volume in one second; DLCO measures diffusing capacity for carbon monoxide.
FVC 是用力肺活量；FEV1 是一秒用力呼氣量；DLCO 測量肺部對一氧化碳的擴散能力。

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1. Imaging tests are critical for myeloma diagnosis and detecting recurrence; not every test is done for every patient and tests may be alternated.
   影像檢查對骨髓瘤診斷與偵測復發很重要；並非每位病人都會做每一種檢查，檢查也可能輪替進行。

2. Skeletal survey/bone scan is a traditional X-ray approach to look for visible lytic lesions; lesion number can help describe myeloma stage.
   骨骼攝影/骨掃描是傳統 X 光方式，用於尋找可見溶骨性病灶；病灶數量可協助描述骨髓瘤分期。

3. CT or MRI may detect marrow abnormalities described as enhancement.
   CT 或 MRI 可能偵測骨髓異常，報告中可能描述為增強（enhancement）。

4. Whole-body low-dose CT is better at detecting bone defects and has prognostic significance, but gives more radiation than skeletal survey.
   全身低劑量電腦斷層較能偵測骨缺損且具預後意義，但輻射劑量高於骨骼攝影。

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5 PET scans can detect enlarged lymph nodes, liver or spleen involvement, and bone lesions; repeated studies can follow changes during and after treatment.
PET 可偵測淋巴結腫大、肝脾變化與骨病灶；重複檢查可追蹤治療期間與治療後的變化。

6 CT detects enlarged nodes, liver or spleen, and bone lesions, and can measure structures during and after treatment.
CT 可偵測淋巴結腫大、肝脾與骨病灶，並可在治療期間與治療後測量結構大小。

7 PET/CT merges functional PET with anatomic CT imaging and has prognostic significance, including in MRD assessment.
PET/CT 將 PET 的功能影像與 CT 的解剖影像合併，具有預後意義，也可用於 MRD 評估。

8 Whole-body diffusion-weighted MRI is highly sensitive for marrow imaging and may help detect, quantify, and guide therapy.
全身擴散加權 MRI 對骨髓影像非常敏感，可能協助偵測、量化疾病並引導治療。

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1. MRI is interpreted by a radiologist and used to monitor lesion changes.
   MRI 由放射科醫師判讀，用於監測病灶變化。

2. MRI can help rule out focal marrow lesions in suspected smoldering myeloma before true osteolytic disease appears.
   在疑似冒煙型骨髓瘤中，MRI 可在真正溶骨性疾病出現前協助排除局部骨髓病灶。

3. MRI is useful for extramedullary disease, suspected cord compression, or detailed imaging of a symptomatic area.
   MRI 對髓外疾病、疑似脊髓壓迫，或需要詳細評估的症狀部位很有幫助。

4. HealthTree imaging classes cover diagnosis imaging, repeat intervals, monitoring bone disease, lytic/focal lesions, extramedullary disease, plasmacytoma, and CT versus MRI.
   HealthTree 影像課程涵蓋診斷影像、重複檢查頻率、骨病監測、溶骨/局部病灶、髓外疾病、漿細胞瘤，以及 CT 與 MRI 的選擇。

5. Myeloma Crowd references include diagnosis and discussions of novel imaging in multiple myeloma.
   Myeloma Crowd 參考資料包含診斷與多發性骨髓瘤新型影像的討論。

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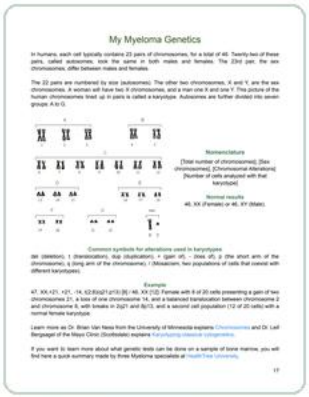

My Myeloma Genetics

In humans, each cell typically contains 23 pairs of chromosomes, for a total of 46. Twenty-two of these pairs, called autosomes, look the same in both males and females. The 23rd pair, the sex chromosomes, differ between males and females.

The 22 pairs are numbered by size (autosomes). The other two chromosomes, X and Y, are the sex chromosomes. A woman will have two X chromosomes, and a man one X and one Y. This picture of the human chromosomes lined up in pairs is called a karyotype. Autosomes are further divided into seven groups A to G.

Nomenclature

[Total number of chromosomes], [Sex chromosomes], [Chromosomal Alterations] [Number of cells analyzed with that karyotype]

Normal results

46, XX (Female) or 46, XY (Male).

Common symbols for alterations used in karyotypes

del (deletion), t (translocation), dup (duplication), + (gain of), - (loss of), p (the short arm of the chromosome), q (long arm of the chromosome), / (Mosaicism, two populations of cells that overall with different karyotypes).

Example

47, XX,+21, +21, -14, t(2;5)(q21;q13);[8] / 46, XX [12] Female with 8 of 20 cells presenting a gain of two chromosomes 21, a loss of one chromosome 14, and a balanced translocation between chromosome 2 and chromosome 5, with breaks in 2q21 and 5q13, and a second cell population (12 of 20 cells) with a normal female karyotype.

Learn more as Dr. Brian Van Ness from the University of Minnesota explains Chromosomes and Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explains Karyotyping molecular cytogenetics.

If you want to learn more about what genetic tests can be done on a sample of bone marrow, you will find here a quick summary made by three Myeloma specialists at Health Free University.

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1. My Myeloma Genetics: humans typically have 23 chromosome pairs, 46 chromosomes total.
   我的骨髓瘤遺傳學：人類通常有 23 對染色體，總共 46 條。

2. Twenty-two pairs are autosomes; the 23rd pair are sex chromosomes, which differ between males and females.
   其中 22 對為體染色體；第 23 對為性染色體，男女不同。

3. A paired chromosome picture is called a karyotype; autosomes are further divided into groups A to G.
   成對排列的染色體圖稱為核型；體染色體又分為 A 到 G 組。

4. Nomenclature includes total chromosome number, sex chromosomes, chromosomal alterations, and number of analyzed cells with that karyotype.
   命名法包含染色體總數、性染色體、染色體變化，以及具有該核型的分析細胞數。

5. Normal results are 46,XX for female or 46,XY for male.
   正常結果為女性 46,XX 或男性 46,XY。

6. Common symbols: del deletion, t translocation, dup duplication, + gain, - loss, p short arm, q long arm, and / mosaicism.
   常見符號：del 缺失、t 易位、dup 重複、+ 增加、- 缺失、p 短臂、q 長臂、/ 嵌合。

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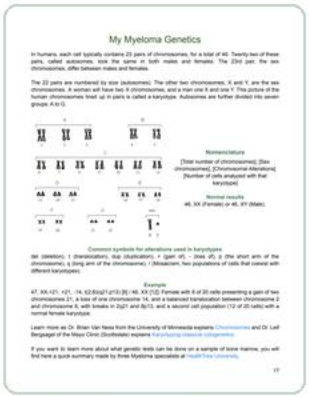

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Nomenclature

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Example

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7 The example describes two cell populations: one with multiple chromosome changes and one with a normal female karyotype.
範例描述兩群細胞：一群有多項染色體變化，另一群為正常女性核型。

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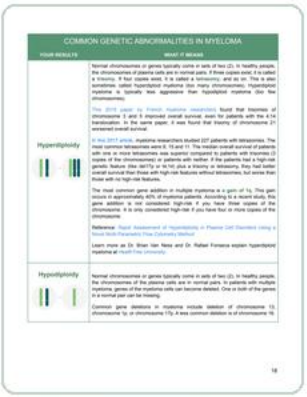

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1. Hyperdiploidy: normal chromosomes usually come in pairs; three copies are trisomy, four copies are tetrasomy.
   超二倍體：正常染色體通常成對；三份稱三體，四份稱四體。

2. Hyperdiploid myeloma, with too many chromosomes, is usually less aggressive than hypodiploid myeloma, with too few chromosomes.
   超二倍體骨髓瘤因染色體過多，通常比染色體過少的低二倍體骨髓瘤侵襲性低。

3. A 2015 French study found trisomy 3 and 5 improved overall survival, even with t(4;14), while trisomy 21 worsened overall survival.
   一篇 2015 年法國研究發現，即使有 t(4;14)，第 3 與第 5 染色體三體仍改善整體存活；第 21 染色體三體則使整體存活較差。

4. A 2017 study of tetrasomies found common tetrasomies 9, 15, and 11, with better median survival than trisomies or neither.
   一篇 2017 年四體研究發現常見四體為第 9、15 與 11 染色體，整體中位存活優於三體或兩者皆無者。

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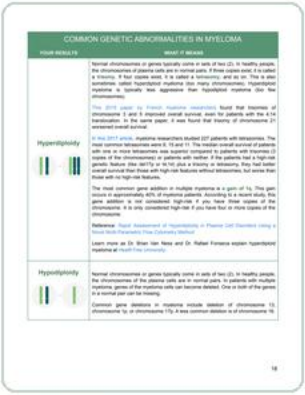

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5 Gain 1q is the most common gene addition in MM, about 40%; recent data suggest high risk mainly with four or more copies.
1q 增益是多發性骨髓瘤最常見的基因增加，約 40%；近期資料顯示主要在四份以上時才視為高風險。

6 Hypodiploidy means myeloma-cell genes can be deleted, including chromosome 13, 1p, 17p, and less commonly 16.
低二倍體表示骨髓瘤細胞基因可能缺失，常見包含第 13 染色體、1p、17p，較少見為第 16 染色體。

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Translocation

Normal chromosomes or genes typically come in sets of two (2). In healthy people, the chromosomes of plasma cells are in normal pairs. In patients with multiple myeloma, sometimes the chromosomes can break apart and swap pieces with parts of another chromosome. This is called a translocation. For example, in a t(4;14) translocation, part of chromosome 4 has swapped places with chromosome 14.

Common translocations include t(4;14), t(11;14), t(14;16), t(14;20). Less common translocations include t(6;14) and t(12;14). Translocations can be considered standard risk or high risk. Standard-risk include t(11;14) and t(6;14). High risk include t(4;14), t(14;16) and t(14;20).

Gene Mutation

A gene mutation is an alteration in the DNA sequence that makes up a gene. The gene is a protein inside a cell that helps the cell function properly by providing instructions. It can be inherited or acquired. The mutation can vary in size to involve a single gene or a group of genes on a chromosome. A mutation can cause the cell to malfunction and disrupt normal development or cause a medical condition.

HEALTHFREE UNIVERSITY CLASS

Learn more as Dr. Gareth Morgan from NYU Langone Health, Dr. Cesar Rodriguez Valdes of Wake Forest Baptist Health, and Dr. Brian Van Ness from the University of Minnesota explain what are the different types of chromosomal mutations (e.g. Translocations, Deletions, Insertions, Amplifications) in Health Free University.

CYTOGENETICS/FISH

YOUR RESULTS

WHAT IT MEANS

Cytogenetics
- Normal
- Abnormal

Gene additions
- Gain 1q21

Hypodiploid
- Present
- Monosomy

Hyperdiploid
- Near-tetraploid
- Near-triploid
- Hyperdiploid

Relevant deletions
- 1p
- 13q
- 17p
- 17p (TP53)

Translocations
- FGFR3/IGH(t(4;14))
- CCND1 t(11;14)
- CCND1/IGH (t(14;16))
- CCND3 MAF8 t(14;20)
- MUM1 (t(6;14))

Metaphase cytogenetics is a test that puts dividing myeloma cells in culture and identifies abnormalities while the cells are dividing. Because myeloma cells can't grow outside the bone marrow environment, only 30% of patients show abnormalities, which means they have more aggressive disease. For the 70% of patients whose myeloma cells don't show abnormalities during cell division, they have less aggressive disease. The advantage of this test is that it identifies the 30% of patients with more aggressive disease, but is not very informative in 70% of patients with less aggressive disease.

The FISH test allows you to look at certain "hot spots" or probes on the chromosomes and it allows you to find translocations of genes. The FISH test does not need actively dividing cells. This test is informative for all patients. The limitation is that you can only see what you're looking for, or there are only a certain number of probes. The FISH test evaluates the chromosomes in the normal

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1. Translocation means chromosome pieces break and swap places; for example, in t(4;14), chromosome 4 swaps with chromosome 14.
   易位表示染色體片段斷裂並交換位置；例如 t(4;14) 中，第 4 染色體片段與第 14 染色體交換。

2. Common translocations include t(4;14), t(11;14), t(14;16), and t(14;20); less common include t(6;14) and t(12;14).
   常見易位包含 t(4;14)、t(11;14)、t(14;16) 與 t(14;20)；較少見包含 t(6;14) 與 t(12;14)。

3. Standard-risk translocations include t(11;14) and t(6;14); high-risk include t(4;14), t(14;16), and t(14;20).
   標準風險易位包含 t(11;14) 與 t(6;14)；高風險包含 t(4;14)、t(14;16) 與 t(14;20)。

4. Gene mutation is a DNA-sequence alteration that may be inherited or acquired and can involve one gene or groups of genes.
   基因突變是 DNA 序列改變，可能遺傳或後天取得，可影響單一基因或一群基因。

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5 Cytogenetics/FISH results may list normal/abnormal cytogenetics, gain 1q21, trisomy, tetrasomy, hypodiploid categories, deletions, and named translocations.
細胞遺傳學/FISH 結果可能列出正常/異常、1q21 增益、三體、四體、低二倍體類別、缺失，以及具名易位。

6 Metaphase cytogenetics cultures dividing myeloma cells; because myeloma cells do not grow well outside marrow, only about 30% show abnormalities.
中期細胞遺傳學會培養分裂中的骨髓瘤細胞；因骨髓瘤細胞在骨髓外不易生長，約只有 30% 會顯示異常。

7 FISH examines specific chromosome hot spots or probes and does not require actively dividing cells, but only detects what the probes target.
FISH 檢查特定染色體熱點或探針，不需要活躍分裂的細胞，但只能偵測探針所針對的變化。

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1. FISH evaluates chromosomes in normal and myeloma cells from marrow; abnormalities may include too many, too few, or altered chromosomes.
   FISH 評估骨髓中正常細胞與骨髓瘤細胞的染色體；異常可能包括染色體過多、過少或其他改變。

2. FISH takes about 2-3 weeks and can use blood or marrow samples; specimen and test quality matter greatly.
   FISH 約需 2-3 週，可使用血液或骨髓樣本；檢體與檢驗品質非常重要。

3. Flow cytometry uses blood or marrow to evaluate myeloma cells and can detect low levels after high-dose therapy and transplantation.
   流式細胞術使用血液或骨髓評估骨髓瘤細胞，並可在高劑量治療與移植後偵測低量殘留。

4. Flow cytometry is used for MRD, does not need a baseline sample, and can reach sensitivity of 1 in 100,000 cells.
   流式細胞術可用於 MRD，不需要基準樣本，敏感度可達十萬分之一。

逐頁中英對照

第 21 頁（2/2） / Page 21

原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

5 It classifies cells by surface substances using laser detection; panels may range from 2 colors to 12 colors.
它以雷射偵測細胞表面物質來分類細胞；檢測組合可從 2 色到 12 色。

6 Next generation sequencing examines DNA nucleic acids to detect genomic alterations and unknown translocation patterns.
次世代定序檢查 DNA 核酸，可偵測基因體改變與未知易位型態。

7 Targeted sequencing panels usually test selected frequently mutated genes rather than the entire genome, making testing cheaper and faster.
標靶定序面板通常檢測常見突變基因，而非整個基因體，因此較便宜也較快。

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逐頁中英對照

第 22 頁（1/2）/ Page 22

原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. NGS can assess MRD, requires a baseline sample, and has sensitivity of 1 in 1 million cells.
   NGS 可評估 MRD，需要基準樣本，敏感度可達百萬分之一。

2. MRD means the small number of cancer cells that may remain during or after treatment and may eventually cause recurrence.
   MRD 指治療期間或治療後可能殘留的少量癌細胞，日後可能造成復發。

3. Residual cells usually cause no symptoms and require sensitive techniques to identify.
   殘留細胞通常不造成症狀，需要敏感技術才能辨識。

4. Monitoring MRD over treatment and remission provides important insight into disease status.
   在治療與緩解過程中監測 MRD，可提供疾病狀態的重要資訊。

5. Flow cytometry MRD is more accessible, does not need a baseline sample, and has sensitivity of 1 in 100,000 cells.
   流式細胞術 MRD 較容易取得，不需要基準樣本，敏感度為十萬分之一。

6. NGS MRD examines DNA, needs a baseline sample, and has higher sensitivity of 1 in 1 million cells.
   NGS MRD 檢查 DNA，需要基準樣本，敏感度較高，為百萬分之一。

逐頁中英對照

第 22 頁（2/2）/ Page 22

原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

7 MRD negativity means no myeloma cells were found in the tested sample, not that there are no myeloma cells anywhere in marrow.
MRD 陰性表示受測樣本中未找到骨髓瘤細胞，不代表整個骨髓完全沒有骨髓瘤細胞。

8 Sustained MRD negativity for over a year is associated with longer progression-free and overall survival.
MRD 陰性持續超過一年，與較長的無惡化存活與整體存活相關。

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HEALTHTREE UNIVERSITY CLASSES

GENE EXPRESSION PROFILE

WHAT IT MEANS

HEALTHTREE UNIVERSITY CLASS

SINGLE NUCLEOTIDE POLYMORPHISM (SNP) ARRAY

WHAT IT MEANS

HEALTHTREE UNIVERSITY CLASS

原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Gene expression profile reports a prognostic risk score, molecular subtype, and virtual karyotype unique to the disease.
   基因表現分析會報告預後風險分數、分子亞型與此疾病特有的虛擬核型。

2. These designations have been shown to predict overall survival, event-free survival, complete response duration, and post-relapse survival.
   這些分類已被證實可預測整體存活、無事件存活、完全反應持續時間與復發後存活。

3. MyPRS distinguishes high-risk and low-risk disease; the source lists 5-year OS probability as 38% for high-risk and 83% for low-risk.
   MyPRS 區分高風險與低風險疾病；原文列出高風險 5 年整體存活機率為 38%，低風險為 83%。

4. Molecular subtype assigns one of seven subtypes associated with genetic lesions, altered genes, and outcome variation.
   分子亞型會分配七種亞型之一，與遺傳病灶、改變基因及結果差異相關。

5. Virtual karyotype refers to cytogenetic/FISH results checking for high-risk chromosome abnormalities.
   虛擬核型指檢查高風險染色體異常的細胞遺傳學/FISH 結果。

逐頁中英對照

第 23 頁（2/2）/ Page 23

HEALTHTREE UNIVERSITY CLASSES

Learn more at HealthTree University, where we have a complete chapter dedicated to MRD testing. You can find here the first class in which several faculty doctors explain what MRD is and how useful it is.

GENE EXPRESSION PROFILE

WHAT IT MEANS

You receive a Prognostic Risk Score (GEP-70), Molecular Subtype, and Virtual Karyotype unique to your disease.

The Prognostic Risk score and the Molecular Subtype designations have been clinically shown to predict overall survival, event-free survival, complete response duration, and post-relapse survival.

Risk Score: MyPRS distinguishes between patients with high and low-risk disease.
High-Risk: Poor Prognosis. Probability of 5-year OS 38%
Low Risk: Good Prognosis. Probability of 5-year OS 83%

Molecular Subtype: You receive 1 of 7 molecular subtypes associated with your unique genetic lesions, altered genes and outcome variation

Virtual Karyotype: Your Cytogenetic (FISH) results checking for high-risk chromosome abnormalities.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Gareth Morgan of the NYU Langone Health and Dr. Brian Van Ness from the University of Minnesota explain Gene Expression Profile (GEP) in HealthTree University.

SINGLE NUCLEOTIDE POLYMORPHISM (SNP) ARRAY

WHAT IT MEANS

Single Nucleotide Polymorphism (SNP) Array is a genetic test currently not used in the clinical setting, that allows you to look for differences between cells, and is typically used as a way to estimate gains and losses of chromosomal material in the whole genome. It can be used for relevant mutations like Gain 1q21, or Deletions 17p/16p/13q/1p.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explains what is a SNP Array in HealthTree University.

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原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

6 SNP array is a genetic test not currently used clinically here; it can identify cell differences and estimate genome-wide chromosomal gains and losses.
SNP 陣列是此處所述目前未用於臨床情境的遺傳檢查；可辨識細胞差異並估計全基因體染色體增加與缺失。

7 SNP array may be used for mutations such as gain 1q21 or deletions 17p, 16p, 13q, and 1p.
SNP 陣列可用於 1q21 增益或 17p、16p、13q、1p 缺失等相關突變。

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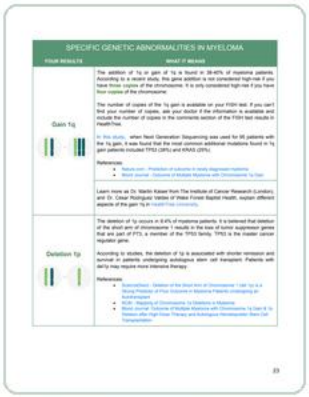

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Gain 1q means addition or gain of 1q and is found in about 38-40% of myeloma patients.
   1q 增益表示 1q 增加，約見於 38-40% 的骨髓瘤病人。

2. Recent study data cited in the source say gain 1q is not high-risk with three chromosome copies, but is high-risk with four copies.
   原文引用近期研究指出，若有三份染色體，1q 增益不被視為高風險；若有四份則被視為高風險。

3. The number of 1q copies may be available on FISH; if not found, ask the doctor whether it is available and record it in HealthTree comments.
   1q 拷貝數可能可在 FISH 檢查中取得；若找不到，可詢問醫師是否可取得，並記錄在 HealthTree 的 FISH 結果備註中。

4. In a 95-patient NGS study with gain 1q, common additional mutations included TP53 (38%) and KRAS (25%).
   在一項 95 位 1q 增益病人的 NGS 研究中，常見額外突變包含 TP53（38%）與 KRAS（25%）。

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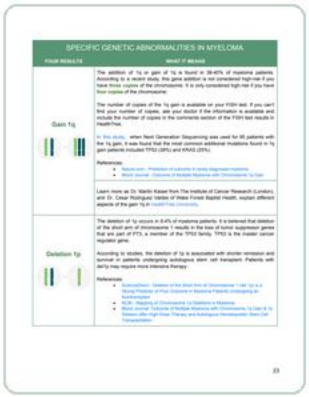

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

5 Deletion 1p occurs in about 8.4% of myeloma patients and may involve loss of tumor suppressor genes related to P73 and the TP53 family.
1p 缺失約見於 8.4% 的骨髓瘤病人，可能涉及 P73 與 TP53 家族相關腫瘤抑制基因流失。

6 Studies associate deletion 1p with shorter remission and survival after autologous stem cell transplant; patients may require more intensive therapy.
研究將 1p 缺失與自體幹細胞移植後較短緩解與存活相關聯；病人可能需要更密集治療。

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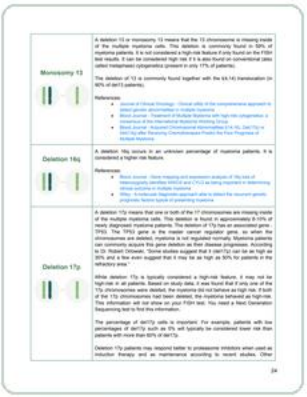

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Monosomy 13 or deletion 13 means chromosome 13 is missing inside myeloma cells and is found in about 59% of patients.
   第 13 染色體單體或缺失表示骨髓瘤細胞中缺少第 13 染色體，約見於 59% 病人。

2. Deletion 13 alone on FISH is not considered high-risk, but may be high-risk if also found on conventional/metaphase cytogenetics.
   若僅在 FISH 上看到第 13 染色體缺失，通常不視為高風險；若傳統/中期細胞遺傳學也發現，則可能視為高風險。

3. Deletion 13 is commonly found with t(4;14), reported in 90% of del13 patients.
   第 13 染色體缺失常與 t(4;14) 同時出現，原文稱見於 90% 的 del13 病人。

4. Deletion 16q occurs in an unknown percentage of patients and is considered a higher-risk feature.
   16q 缺失在病人中的比例未知，被視為較高風險特徵。

5. Deletion 17p means one or both chromosome 17 copies are missing and is found in about 8-10% of newly diagnosed patients.
   17p 缺失表示一份或兩份第 17 染色體缺失，約見於 8-10% 的新診斷病人。

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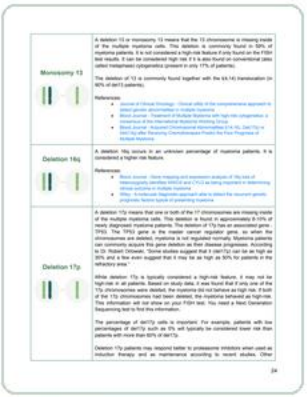

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

6 Deletion 17p involves TP53, the master cancer regulator gene; myeloma patients may acquire this deletion as disease progresses.
17p 缺失涉及 TP53 這個重要癌症調控基因；骨髓瘤病人可能隨疾病進展取得此缺失。

7 Risk may depend on whether one or both 17p copies are deleted and on the percentage of del17p cells; NGS is needed for copy-level information.
風險可能取決於缺失一份或兩份 17p，以及 del17p 細胞百分比；拷貝層級資訊需 NGS 才能得知。

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原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Deletion 17p patients may respond better to proteasome inhibitors when used for induction and maintenance, according to recent studies cited in the source.
   原文引用近期研究指出，17p 缺失病人在誘導與維持治療使用蛋白酶體抑制劑時，可能反應較好。

2. Other approaches may include three- or four-drug induction, one or two transplants, and double- or triple-agent maintenance.
   其他治療策略可能包含三藥或四藥誘導治療、一或兩次移植，以及雙藥或三藥維持治療。

3. The page lists multiple references on del17p, TP53, high-risk/double-hit myeloma, and targeted therapy discussions.
   本頁列出多項關於 del17p、TP53、高風險/double-hit 骨髓瘤與標靶治療討論的參考資料。

4. HealthTree classes cover what 17p deletion is, why it is high risk, percentage of affected cells, mutation plus deletion, and persistence after auto-ASCT.
   HealthTree 課程涵蓋什麼是 17p 缺失、為何屬高風險、受影響細胞百分比、突變合併缺失，以及自體移植後是否仍存在。

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原始頁面參考：位置與頁碼保留；下方重排為手機較易閱讀的中英對照。

中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

5 Translocation (6;14) occurs in about 1% of patients and swaps part of chromosome 6 with chromosome 14.
t(6;14) 易位約見於 1% 病人，表示第 6 染色體的一部分與第 14 染色體的一部分交換。

6 This affects Cyclin D3/CCND3, which can turn cells into cancer cells.
這會影響 Cyclin D3/CCND3，可能使細胞轉變為癌細胞。

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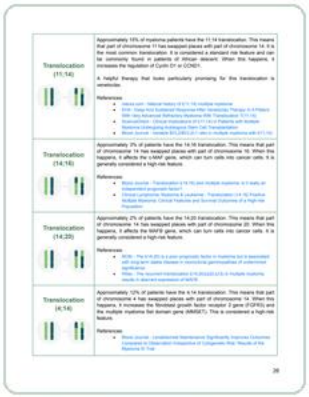

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. Translocation (11;14) occurs in about 15% of myeloma patients, is the most common translocation, and is generally standard-risk.
   t(11;14) 易位約見於 15% 的骨髓瘤病人，是最常見的易位，通常屬標準風險。

2. It may be common in patients of African descent and increases regulation of Cyclin D1/CCND1; venetoclax is noted as a promising therapy.
   此易位可能較常見於非洲裔病人，並增加 Cyclin D1/CCND1 調控；原文指出 venetoclax 是特別有前景的治療。

3. Translocation (14;16) occurs in about 3% of patients, affects c-MAF, and is generally considered high-risk.
   t(14;16) 約見於 3% 病人，會影響 c-MAF，通常被視為高風險。

4. Translocation (14;20) occurs in about 2% of patients, affects MAFB, and is generally considered high-risk.
   t(14;20) 約見於 2% 病人，會影響 MAFB，通常被視為高風險。

5. Translocation (4;14) occurs in about 12% of patients and swaps chromosome 4 with chromosome 14.
   t(4;14) 約見於 12% 病人，表示第 4 染色體與第 14 染色體片段交換。

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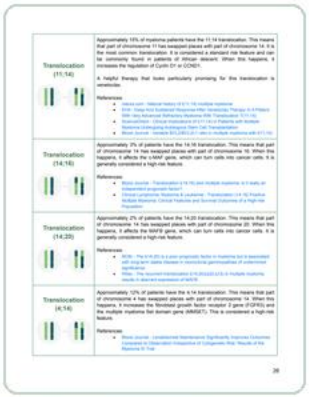

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

6 It increases FGFR3 and MMSET activity and is considered a high-risk feature.
此易位會增加 FGFR3 與 MMSET 活性，並被視為高風險特徵。

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中英重點對照

AI 輔助翻譯與重排，僅供資訊參考；醫療決策請諮詢主治醫師。

1. This page continues t(4;14) references, including MMSET activity and lenalidomide maintenance outcomes across cytogenetic risk groups.
   本頁延續 t(4;14) 參考資料，包含 MMSET 活性，以及不同細胞遺傳風險族群中 lenalidomide 維持治療的結果。

2. HealthTree University class: Dr. Fotis Asimakopoulos explains the importance of chromosome 14 and its relationship to high-risk myeloma.
   HealthTree University 課程：Fotis Asimakopoulos 醫師說明第 14 染色體的重要性及其與高風險骨髓瘤的關係。

3. The source also points readers to Myeloma Crowd for more on translocation 4;14 and high-risk myeloma.
   原文也引導讀者到 Myeloma Crowd 了解更多 t(4;14) 與高風險骨髓瘤資訊。

DECIPHERING MY MYELOMA LABS

Do you understand your myeloma diagnosis and your myeloma lab results?

This guide attempts to simplify the complex process of understanding your myeloma markers and helps you track your treatment history. Based on the actual lab printouts you receive in the clinic, we've added color-coding to help you identify the most important myeloma markers.

Priority	Secondary	Not as important

Special Thanks to myeloma specialist Dr. Guido Tricot of the University of Iowa, pathologist Dr. Michael Misialek of Newton-Wellesley Hospital, Jen Higbee of Huntsman Cancer Institute, and Barbara Waagen for their contributions to this document.

Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established reference range.

Each section is linked to a HealthTree University Class. Feel free to click on the links in each section for further information.

Do you have suggestions to make this document better? Send your comments to support@healthtree.org, and we will keep revising this document as we learn more.

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My Myeloma Summary

Revised-International Staging System (R-ISS)	
Stage I	• Serum albumin ≥3.5 g/dL • Serum beta-2-microglobulin <3.5 mg/L • No high-risk cytogenetics • Normal serum lactate dehydrogenase level
Stage II	• Not fitting Stage I or III
Stage III	• Serum beta-2-microglobulin >5.5 mg/L, and • High-risk cytogenetics [t(4;14), t(14;16), or del(17p)] or elevated serum lactate dehydrogenase level
The Revised International Staging System was based on a large sample size of 4,445 newly diagnosed multiple myeloma patients from 11 different clinical trials and was updated in 2015. Myeloma experts already knew that high levels of Beta 2 microglobulin and low albumin levels were indicators of higher risk myeloma. The update occurred as myeloma researchers learned more about high-risk genetic features and high levels of LDH, which have now been added to the staging system.	

Learn more as Dr. Muzaffar Quazilbash of the MD Anderson Cancer Center explains myeloma staging in HealthTree University.

Mayo clinic risk stratification for multiple myeloma (mSMART)	
Standard-risk	Trisomies, t(11;14) and t(6;14).
	75% of newly diagnosed patients.
High-risk	Del(17p), t(4;14), t(14;16), t(14;20), Gain 1q, p53 mutation, R-ISS Stage III, High-risk signature by GEP, or High Plasma Cell S-phase.
	25% of newly diagnosed patients.
	The presence of any two high-risk genetic abnormalities is considered double-hit myeloma; three or more high-risk ganetic abnormalities are triple-hit myeloma.
• Genetic abnormalities by FISH or equivalent method • Trisomies may ameliorate the risk • t(11;14) may be associated with plasma cell leukemia	
mSMART was designed for physicians external to Mayo. The guidelines were explicitly written to be clear, simple, and easy to use. Rafael Fonseca, M.D., Mayo Clinic Cancer Center interim director, hematologist, and multiple myeloma specialist has called the advent of mSMART a staple of Mayo Clinic's engagement with those who treat multiple myeloma and a reference in the myeloma community.	

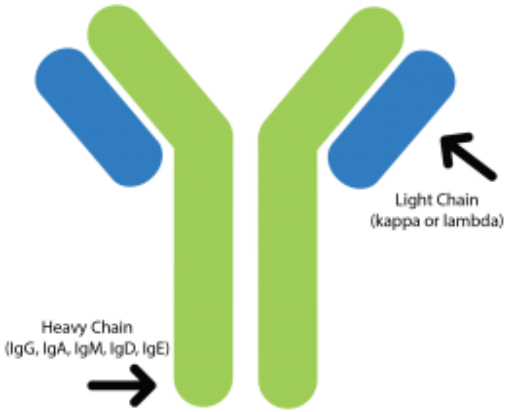
Learn more as Dr. Morie Gertz of the Mayo Clinic (Rochester) explains the mSMART in HealthTree University.

My Myeloma Labs

MONOCLONAL PROTEINS

Multiple myeloma is a blood cancer of white blood cells called plasma cells. Plasma cells come from the bone marrow and produce antibodies (also called immunoglobulins) that fight a wide variety of infections. In myeloma, one of these antibodies grows out of control in the bone marrow, crowding out the other antibodies and other components of the immune system, making too much of one type and reducing the ability for your body to create a broad spectrum of immunoglobulins to fight infections. This is called a monoclonal protein (also called M-protein or M-spike).

Serum protein electrophoresis (SPEP) is a test used to find any abnormal immunoglobulin. Then, another test, such as immunofixation or immunoelectrophoresis, is used to determine the exact type of abnormal antibody (IgG or some other type). This abnormal protein is known by several different names, including monoclonal immunoglobulin, M protein, M spike, and paraprotein.



Antibodies are made up of two parts: heavy chains and light chains. The types of monoclonal proteins are defined by these parts, although sometimes the heavy chain might be missing.

Videos explained by Myeloma specialists about this topic:

- What is the normal structure of an immunoglobulin (antibody)?
- What is a monoclonal protein (or M-spike) and how is it detected?
- What are the different types of myeloma?

SERUM PROTEIN ELECTROPHORESIS (BLOOD TEST)

The SPEP is broken down into five categories: Albumin, Alpha-1, Alpha-2, Beta, and Gamma (see below). In particular, this test can detect the presence of "M protein," another name for the large number of abnormal monoclonal antibodies being produced.

LAB TESTED	REFERENCE RANGE (g/dL)	WHAT IT MEANS
TOTAL PROTEIN	6.0 - 8.3	Measures the total amount of protein in a blood sample.
ALBUMIN	3.75 - 8.3	Albumin proteins keep the blood from leaking out of blood vessels and are important for tissue growth/healing. Low values can indicate malnutrition, kidney or liver disease, inflammation, and protein-losing problems. High values can indicate dehydration.
ALPHA 1	0.19 - 0.46	Low values can indicate severe liver disease, and High values can indicate acute/chronic inflammation.
ALPHA 2	0.48 - 1.05	Low values can indicate malnutrition, severe liver disease, or red blood cell disintegration. High values can indicate kidney disease or acute/chronic inflammation.
BETA	0.48 - 1.0	Beta globulin proteins help carry substances, such as iron, through the bloodstream and help fight infection. Low values can indicate malnutrition or fibrous tissue in the liver, and High values can indicate hypercholesterolemia, iron deficiency anemia, MGUS, or MM.
GAMMA	0.62 - 1.51	These proteins are also called antibodies. They help prevent and fight infection. Low=immune disorder, High/Polyclonal = inflammatory diseases like arthritis, lupus, liver problems, High/Monoclonal = MM, lymphoma, Waldenstrom's macroglobulinemia.
PARAPROTEIN	0 or Negative	Paraproteins form a narrow band or "spike" when they are all the same protein. They are also referred to as "**M proteins**." (M=monoclonal). This is the M-spike number. SPEP and UPEP tell us how much monoclonal protein there is, but not the type.
IMMUNOFIXATION	Negative	Identifies the type of immunoglobulin protein(s) presenting monoclonal bands on a protein electrophoresis pattern; typically, immunofixation determines the presence of a heavy chain (IgG, IgM, IgA) and a light chain (kappa or lambda).
DARATUMUMAB	IgG Kappa	Daratumumab (Darzalex) can influence the results of IFE if a patient has IgG Kappa myeloma and is being evaluated for a complete response. The patient may be in a complete response, but a tiny band of IgG Kappa will show up on the test.

HEALTHTREE UNIVERSITY CLASS

- What is a Serum Protein Electrophoresis (SPEP)?

Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established normal range.

SERUM FREE LIGHT CHAIN TEST

LAB TESTED	REFERENCE RANGE	WHAT IT MEANS
KAPPA FREE LIGHT CHAINS	3.3 - 19.4 mg/L	There are two types of light chains: kappa and lambda. If a patient has kappa myeloma, their doctor will watch for a rise in the kappa numbers. Likewise, the lambda number will be observed if a patient has lambda myeloma. When myeloma progresses, the myeloma cells start to produce more light chains than heavy chains. This can be measured by the Free Light Chain Assay test on a blood specimen. The higher the free light chains, the more aggressive the disease is. Therefore, the serum-free light chain test is a better predictor of outcome than the amount of M-protein in the blood.
LAMBDA FREE LIGHT CHAINS	5.7 - 26.3 mg/L	
KAPPA:LAMBDA RATIO	0.26 to 1.65	This balance of kappa and lambda together is called the kappa/lambda ratio, which can also indicate a change in levels of disease. "The ratio or proportion between the kappa and lambda light chains indicates an excess production of one chain over the other, and therefore can be used to indicate disease progression or remission," said Dr. Christina Gasparetto of Duke University.

HEALTHTREE UNIVERSITY CLASSES

- What is the normal structure of an immunoglobulin (antibody)?
- What are light chains, how are light chains reported, and why?
- How do you measure free light chains?
- Why is knowing your free light chain ratio important?

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URINE PROTEIN ELECTROPHORESIS (URINE TEST)

The UPEP is generally performed on a single urine sample. Bence-Jones proteins will be detected if present. A routine urinalysis will not detect Bence-Jones proteins.

LAB TESTED	NORMAL RANGE (mg/day)	WHAT IT MEANS
URINE TOTAL PROTEIN (UPEP)	<229	Measures the total amount of protein in a urine sample of 24 hours.

ALBUMIN IN URINE	33 - 50	Albumin proteins keep the blood from leaking out of blood vessels and are essential for tissue growth/healing. Low values can indicate malnutrition, kidney or liver disease, inflammation, and protein-losing problems. High values can indicate dehydration.
ALBUMIN % URINE	%	
GLOBULIN URINE	50 - 66	The other major protein in the urine, along with albumin.
ALPHA 1 % URINE	%	Low values can indicate severe liver disease. High values can indicate acute/chronic inflammation.
ALPHA 2 % URINE	%	Low values can indicate malnutrition, severe liver disease, or red blood cell disintegration. High values can indicate kidney disease or acute/chronic inflammation.
BETA GLOBULIN % URINE	%	Beta globulin proteins help carry substances, such as iron, through the bloodstream and help fight infection. Low values can indicate malnutrition or fibrous tissue in the liver. High values can indicate hypercholesterolemia, iron-deficient anemia, MGUS orMM
PARAPROTEIN % URINE	0%	The percentage of a monoclonal protein, if present, in the urine.
URINE M-PROTEIN	0 or Negative	This protein is the monoclonal antibody detected in the urine. It is usually called M-Spike. It may be measured in percentage or mg/day.
IMMUNOFIXATION	Negative	This test is generally performed on a 24-hour collection of urine and will measure exact amounts of Bence-Jones proteins present and is used to monitor treatment progress. The higher the level, the more tumor growth.
URINE FREE KAPPA LIGHT CHAINS	<0.9 mg/dL	The serum-free test is tested in the urine. This test can test for free light chains but not whole immunoglobulins. Myeloma can be detected earlier than with UPEP, SPEP, or IFE, but these tests are needed for the complete picture.
URINE FREE LAMBDA LIGHT CHAINS	<0.7 mg/dL	
URINE FREE KAPPA/LAMBDA RATIO	0.7-6.2	

HEALTHTREE UNIVERSITY CLASS

- What is urine protein electrophoresis (UPEP), and is it still used?

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IMMUNOGLOBULINS (HEAVY CHAINS BLOOD TEST)

LAB TESTED	NORMAL RANGE (mg/dL)	WHAT IT MEANS
IgG	768 - 1632	This test measures the blood levels of the different antibodies (also called immunoglobulins). There are several different types of antibodies in the blood. Each type of antibody fights a different type of infection. The levels of these immunoglobulins are measured to see if any are abnormally high or low. In multiple myeloma, one type of immunoglobulin has an overgrowth that crowds out the other immunoglobulins. Thus you may be susceptible to certain kinds of infections, like pneumonia. Elevated immunoglobulins that are not related to a person's myeloma can indicate current infection.
IgA	68 - 378	
IgM	60 - 263	
IgD	< or =10	
IgE	< or =214	

HEALTHTREE UNIVERSITY CLASSES

- What does the quantitative immunoglobulin test measure?
- What is hyperviscosity? How is it treated?

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BONE MARROW ASPIRATION AND CORE BIOPSY

The bone marrow biopsy tests will show how many cells in the bone marrow are myeloma cells and will also indicate the quality of the specimen that was taken.

LAB TESTED	WHAT IT MEANS
IMMUNOHISTOCHEMISTRY	Part of the biopsy is treated with antibodies that attach to specific molecules on the cell surface, and help to identify different types of cells
PLASMA CELLS ON ASPIRATION	The percentage of plasma cells and their appearance will be reported. This number is important in classifying the type of plasma cell disorder and should be followed overtime.
PLASMA CELLS ON BIOPSY	The percentage of plasma cells and their appearance will be reported. This number is important in classifying the type of plasma cell disorder and should be followed overtime.
CD56 POSITIVE ON IMMUNOFIXATION	Not relevant for the myeloma assessment.
ROULEAUX	Rouleaux are stacks or aggregations of red blood cells (RBCs) that form because of the unique discoid shape of the cells in vertebrates. Conditions that cause rouleaux formation include infections, multiple myeloma, Waldenström's macroglobulinemia, inflammatory and connective tissue disorders.
LEUKOCYTE NUMBER	Not relevant for the myeloma assessment.

LEUKOCYTE MORPHOLOGY	Not relevant for the myeloma assessment.
CIRCULATING PLASMA CELLS	Circulating plasma cells are generally a poor prognostic factor.
PLATELET NUMBER	Not relevant for the myeloma assessment.
PLATELET MORPHOLOGY	Not relevant for the myeloma assessment.
ASPIRATION DIFFERENTIAL COUNT (300 CELLS)	
ERYTHROIDS	Not relevant for the myeloma assessment.
PLASMA CELLS	Circulating plasma cells are usually a poor prognostic factor.
MYELOBLASTS	Not relevant for the myeloma assessment.
MYELOIDS	Not relevant for the myeloma assessment.
M:E RATIO	Not relevant for the myeloma assessment.
BONE MARROW ASPIRATE OR CORE BIOPSY	
SPECIMEN QUALITY	An indication of how representative the specimen is of the marrow. Look for terms such as “hemodiluted,” which implies blood contamination.
CELLULARITY	A measure of how cellular the marrow is. It will be reported as a percentage. Look for terms normal, hypo or hypercellular.
SPICULES	A measurement of quality. The presence of spicules means the sample is from the marrow with little or no blood contamination.
MYELOID TO ERYTHROID RATIO	A measure of developing white to red cells.
TRILINEAGE HEMATOPOIESIS	Refers to whether there is a normal development of the white cells, red cells, and platelets.
HEMATOPOIETIC MATURATION	
MEGAKARYOCYTE MORPHOLOGY	Appearance of the platelet precursor cells.
PLASMA CELL NUMBER	The percentage will be a significant determinant of classifying the type of disorder. A typical percentage of plasma cells is <5%
PLASMA CELL MORPHOLOGY	Look for terms such as “mature,” “immature,” or “atypical.” “Mature” is generally a better prognostic feature than the others.
CORE BIOPSY SPECIMEN QUALITY	Size of biopsy is important for assessing adequacy. It should be over 1cm.
BONE TRABECULAE	Should be present, which implies the sample is truly representative of the marrow.
CELLULARITY	Reported as a percentage of cells to fat in the marrow. Look for terms like normal, hypo or hypercellular.

BONE MARROW CLOT IMMUNOHISTOCHEMISTRY	
CD138	Stains may be performed to highlight the different cell types present. CD138 is an antibody stain that marks plasma cells and aids in enumeration.

HEALTHTREE UNIVERSITY CLASSES

- Why do a Bone Marrow Biopsy?
- Why are bone marrow biopsies (BMB) used in myeloma, and what tests are run on the BMB samples?
- What are the genetic tests that can be done on a sample of bone marrow? Can liquid biopsies (blood tests) be used to determine myeloma genetics? How often should testing be repeated?

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CHEMISTRY PANELS

Chemistry panels are run regularly during and after treatment to check your body's normal functions such as heart, kidney, insulin production, electrolyte levels, etc. Some indicators can also determine the aggressiveness of the myeloma.

LAB TESTED	REFERENCE RANGE	WHAT IT MEANS
TOTAL PROTEIN	6.5-8.6 mg/dL	Total protein in the blood, which includes both albumin and globulin. See special note for albumin.
ALBUMIN SERUM	3.5-4.7 mg/dl	A protein found in the blood. Low levels can be a sign of more advanced myeloma.
CALCIUM SERUM	8.4-10.2 mg/dl	May be higher in people with advanced myeloma because of bone destruction. Higher calcium levels should trigger your doctor to do more testing. High calcium can also cause severe constipation and loss of appetite. It can make patients feel weak, drowsy, and confused. If the calcium level gets high enough, it can cause kidney failure, and one may even lapse into a coma.
PHOSPHORUS SERUM	2.4-4.3 mg/dl	Phosphorus and calcium levels have an inverse relationship.
ANION GAP	17mE/L	The anion gap is a measurement of the relationship between the "electrolytes" (Sodium, Potassium, Chloride and Carbon Dioxide) and the combination of Magnesium and Phosphorus. Decreased levels (below 10 mEq/L) is used as a monitor of multiple myeloma.
GLUCOSE	64-128 mg/dl	Monitors glucose levels and insulin production.
BLOOD UREA NITROGEN (BUN)	6-22 mg/dL	Urea is a waste product excreted by the kidneys. High levels are the first sign of dehydration or possible kidney damage.

CREATININE SERUM	52-1.08 mg/dl	Creatinine is a waste product excreted by the kidneys. Elevated levels indicate poor hydration or possible kidney damage.
CREATININE CLEARANCE	>60 mL/min	Creatinine clearance is the gold standard measurement for kidney function. This measures urine excretion of creatinine against serum creatinine. If serum creatinine is elevated, creatinine clearance is low. As the kidneys start to fail, they lose the ability to dispose of excess salt, fluid, and body waste products, leading to symptoms such as weakness and leg swelling.
URIC ACID SERUM	2.5-7.0 mg/dl	Uric acid levels are watched during treatment. An elevated level can indicate tumor lysis syndrome.
ALKALINE PHOSPHATASE	38-126 IU/l	Alkaline Phosphatase levels are elevated in bone and liver disease.
BETA 2 MICROGLOBULIN, SERUM	1.21-2.70 mcg/mL	Tests for the severity of MM. This is another protein produced by the malignant cells. High levels indicate more aggressive disease. Decrease shows good treatment response. It can also identify kidney damage.
LACTATE DEHYDROGENASE (LDH), SERUM	100 - 253 or 300 - 600 or u/L	LD blood level is a nonspecific marker for the presence of tissue damage somewhere in the body. It cannot be used to identify the underlying cause or location of the cellular damage. LDH levels are elevated in aggressive myeloma and are associated with poor prognosis if there is no explanation for its increase other than myeloma. Each laboratory has a different range for what's normal. Your lab report should show the range that your lab uses for each test. LDH labs with different reference ranges cannot be compared to each other. The normal range is just a guide. Your doctor will look at your results based on your age, health, and other factors. A value that isn't in the normal range may still be normal for you.
SODIUM SERUM	136-144 mEq/l	Sodium, Potassium, Chloride and Carbon Dioxide are also known as "electrolytes" and are all linked to acid/base balance which is a delicate system maintained by lungs and kidneys which eliminate excessive amounts of each one to maintain this critical pH balance. The most important effects are the neurological and cardiac effects of elevated or decreased levels.
POTASSIUM SERUM	3.3-5.0 mEq/l	
CHLORIDE SERUM	98-107 mEq/l	
CARBON DIOXIDE	20-29 mM	A measure of acid-base status.
MAGNESIUM SERUM	1.6-2.3 mEq/l	Magnesium and calcium levels are linked and need to be kept in balance.
BILIRUBIN TOTAL	0.2-1.3 mg/dl	Total Bilirubin includes both direct and indirect bilirubin. Total bilirubin is a byproduct of hemoglobin and/or red cell destruction.
BILIRUBIN, INDIRECT	0.1-1.0 mg/dl	
BILIRUBIN, DIRECT	0-0.4 mg/dl	
SGOT (ALSO KNOWN AS) SERUM GLUTAMIC - OXALOACETIC TRANSAMINASE ASPARTATE	15-40 U/ml	The SGOT test measures an enzyme found in the liver, muscles (including the heart), and red blood cells. It is released into the blood when cells that contain it are damaged. The SGOT level is measured to check the function of your liver, kidneys, heart, pancreas, muscles,

AMINOTRANSFERASE AST AMINOTRANSFERASE		and red blood cells. It is also measured to check on medical treatments that may affect the liver.
SGPT (ALSO KNOWN AS) SERUM GLUTAMIC-PYRUVIC TRANSAMINASE ALANINE AMINO TRANSAMINASE	8-50 U/ml	The SGPT enzyme is present in liver cells. When a cell is damaged, it leaks this enzyme into the blood, where it is measured. It rises dramatically in acute liver damage, such as viral hepatitis or paracetamol (acetaminophen) overdose.

HEALTHTREE UNIVERSITY CLASSES

- What other blood tests are useful if you suspect a myeloma diagnosis?
- What kidney dysfunction is associated with myeloma and why?

MYELOMA CROWD ARTICLES

- Multiple Myeloma Diagnosis
- What are C.R.A.B. Symptoms of Multiple Myeloma
- Myeloma Patients Should Monitor Their Kidney Function Early & Often

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COMPLETE BLOOD COUNT (CBC)

Multiple myeloma is a cancer of the plasma cells. These are immune system cells that produce specialized molecules called antibodies to help fight infectious agents. Because most plasma cells live in bone marrow, multiple myeloma tumors are usually, but not always, found in bone. The bone marrow is where blood cells are produced. Because multiple myeloma crowds out bone marrow, it can cause several kinds of blood deficiencies such as : Anemia, a shortage of red blood cells, Thrombocytopenia, a shortage of blood platelets, and Leukopenia, a shortage of white blood cells.

	REFERENCE RANGE	WHAT IT MEANS
WHITE BLOOD CELL COUNT (WBC)	3.2-10.6 k/ul	White blood cells are produced in the bone marrow. White blood cell counts include: Neutrophils (also known as granulocytes), Lymphocytes, Monocytes, Eosinophils, and Basophils. The counts are reported in two ways. First as a percentage of the total WBC count and also as an "absolute" count or an actual number of cells present. When there are too few white cells, Leukopenia lowers resistance to infections and can cause such ailments as pneumonia. High WBC may indicate illness or cancer growth.
RED BLOOD CELL COUNT (RBC)	3.88-5.46 10^{12} /L	Low RBC counts indicate anemia resulting in fatigue and lowered oxygen transport and can result from bleeding or low RBC production in the bone marrow.
HEMOGLOBIN (HGB)	12.1-15.9 g/dL	Hemoglobin is carried in the red blood cells. Anemia causes weakness, reduced ability to exercise, shortness of breath, and

		dizziness. If The hemoglobin is less than 7.5 G/DL, many facilities will transfuse unless the patient is symptomatic. If it is less than 9%, Aranesp injections are sometimes used to stimulate red blood cell production.
HEMATOCRIT (HCT)	34.3-46.6 %	Hematocrit measures the percentage of whole blood volume made up of red blood cells. This measurement depends on the number of red blood cells and the size of red blood cells.
MEAN CORPUSCULAR VOLUME (MCV)	77.8-94 fL	MCV measures the average red blood cell volume
MEAN CORPUSCULAR HGB (MCH)	26.5-32.6 pg	MCH is a measure of the hemoglobin content in the average red cell.
MEAN CORPUSCULAR HGB CONCENTRATION (MCHC)	32.7-36.9	MCHC is the average hemoglobin concentration in a given volume of packed red cells.
RED CELL DISTRIBUTION WIDTH (RDW)	10.8-14.1%	
PLATELET COUNT	150 - 440 k/ul	Platelets are produced in the bone marrow, and their primary function is to induce coagulation at points of injury. When blood platelet counts are low (thrombocytopenia), minor scrapes, cuts, or bruises may cause serious bleeding.
MEAN PLATELET VOLUME	5.9-9.8 fl	Platelets are microscopic cells. This test measures their size and volume.
NEUTROPHIL / GRANULOCYTE ABSOLUTE COUNT (ANC) or (AGC)	1.3-7 k/ul	Neutrophils (granulocytes) are the most numerous of the white blood cells. They are the "soldiers" that fight infections. They engulf infectious particles (bacteria)in your body. Low levels indicate the inability to fight infection.
NEUTROPHIL OR GRANULOCYTE PERCENT	44-76 %	
LYMPHOCYTE ABSOLUTE COUNT	0.8-3.1 k/ul	Lymphocytes are the essential cell type in the body's immune system. There are three major types of lymphocytes: B lymphocytes that produce antibodies; T lymphocytes that have several functions and assist in antibody production; and natural killer (NK) cells that can attack virus-infected cells or tumor cells. Low levels indicate the inability to fight infection.
LYMPHOCYTE PERCENT	14.7-42.6 %	
MONOCYTE ABSOLUTE COUNT	0.2-0.7 k/ul	Monocytes also help to fight infections. When they enter the tissues, they fight infection, ingest dead cells and assist in immune responses.
MONOCYTE PERCENT	4-8.9 %	
EOSINOPHIL ABSOLUTE COUNT	0-0.4 k/ul	Eosinophils are elevated in allergic reactions and help to fight certain parasitic infections.

EOSINOPHIL PERCENT	0-6 %	
BASOPHIL ABSOLUTE COUNT	0-0.1 k/ul	Basophils also participate in allergic reactions.
BASOPHIL PERCENT	0.0-1.7 %	
IMMATURE GRANULOCYTE ABSOLUTE COUNT (MD)	-	Immature neutrophils or granulocytes cells may be released prematurely from the bone marrow.
IRON	60-150 µg/dl	These tests are all related to your body's red cell production status and help the physician monitor your disease because red cells transport iron.
IRON BINDING CAPACITY	35-150 mcg/dL	
IRON SATURATION	14-50%	
FERRITIN	11-336 mcg/L	
FOLATE	>4.0 mcg/L	
TOTAL IRON BINDING CAPACITY (TIBC)	250-400 mcg/dL	
TRANSFERRIN SATURATION		
VITAMIN B12	180-914 ng/L	
THYROID STIMULATING HORMONE (TSH)	0.3-4.2 mIU/L	Can be a contributing factor to anemia.

HEALTHTREE UNIVERSITY CLASSES

- What other blood tests are useful if you suspect a myeloma diagnosis?
- What is plasma cell leukemia?
- Primary PCL vs. secondary PCL

MYELOMA CROWD ARTICLES

- Multiple Myeloma Diagnosis

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BLOOD COAGULATION		
	REFERENCE RANGE	WHAT IT MEANS
INTERNATIONAL NORMALIZED RATIO (INR)	3.2-10.6 k/ul	INR is the actual number used for measuring therapeutic Coumadin dosing.
PROTHROMBIN TIME (PT)	12-15.5 seconds	Prothrombin times are used to investigate prolonged bleeding disorders and monitor warfarin (Coumadin) anticoagulant therapy.
PARTIAL THROMBOPLASTIN TIME (APTT)	24-35 seconds	Partial thromboplastin times are used to investigate prolonged bleeding disorders and monitor heparin anticoagulant therapy
Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established normal range.		

IMMUNOLOGY/ALLERGENS		
	NORMAL RANGE	WHAT IT MEANS
C-REACTIVE PROTEIN	0-0.8 mg/dL	An elevated C-Reactive Protein is an indicator of inflammation in your body. It is an indirect measurement of the size and growth of myeloma tumors.
Please note that the Reference Ranges given are not necessarily consistent between laboratories. Each laboratory is required to establish its own reference ranges. Abnormal results must be flagged as High, Low, or Critical if they fall out of the established normal range.		

TOXICOLOGY / DRUG LEVELS		
LAB TESTED	NORMAL RANGE	WHAT IT MEANS
CYSTATIN C	Varies per age	Cystatin C is a protein encoded by the CST3 gene and is mainly used as a biomarker of kidney function.

PULMONARY FUNCTION TESTS	
These tests check for pulmonary function prior to and between treatments.	
LAB TESTED	WHAT IT MEANS
FVC	FVC - Forced Vital Capacity - after the patient has taken in the deepest possible breath, this is the volume of air that can be forcibly and maximally exhaled out of the lungs until no more can be expired. FVC is usually expressed in units called liters. This PFT value is critically important in the diagnosis of obstructive and restrictive diseases.

FEV1	Forced Expiratory Volume in One Second - this is the volume of air that can be forcibly exhaled from the lungs in the first second of a forced expiratory maneuver. It is expressed as liters. This PFT value is critically important in the diagnosis of obstructive and restrictive diseases.
DLCO / DSBHB	Diffusing capacity of the lungs for carbon monoxide (DLCO).

MY IMAGING TESTS

Imaging tests are critical for a myeloma diagnosis as well as detection of recurring myeloma. It is important to know that not every imaging test will be performed on every patient and many times the imaging tests are alternated.

IMAGING TEST	WHAT IT MEANS
SKELETAL SURVEY / BONE SCAN	This is a traditional X-ray that looks for the number of visible lytic lesions. The number of lytic lesions found can help describe the stage of myeloma.
BONE MARROW CHARACTERISTICS	Sometimes CT or MRI can detect marrow abnormalities, which would be reported as “enhancement.”
WHOLE BODY LOW DOSE COMPUTED TOMOGRAPHY	The more advanced Whole-Body Low Dose Computed Tomography (WBLDCT) is superior in detecting bone defects, and it has prognostic significance. However, it has the disadvantage of a much larger radiation dose than a skeletal survey. Like the skeletal survey, CT displays mostly bone destruction, but it can also be useful for assessing bone stability. The International Myeloma Working Group (IMWG) includes lytic lesions detected by WBLDCT as justification for treatment in myeloma patients, even those who are not otherwise symptomatic.
POSITIVE EMISSION TOMOGRAPHY (PET) SCAN	A PET scan can detect enlarged lymph nodes, liver or spleen, and bone lesions. The test is repeated to measure the size of these and other structures during and after treatment. A report is generated by a radiologist with an interpretation for your physician. PET is helpful for evaluating focal lesions after therapy.
FOCAL/LYTIC LESIONS ON PET	Particular notes of focal/osteolytic lesions progression on PET scans.
COMPUTED TOMOGRAPHY (CT) SCAN	A CT Scan can detect enlarged lymph nodes, liver or spleen, and bone lesions. A CT scan can be used to measure the size of these and other structures during and after treatment. A report is generated by a Radiologist with an interpretation for your physician.
FOCAL/LYTIC LESIONS ON CT	Particular notes of focal/osteolytic lesions progression on CT scans.
PET/CT	When PET and CT are combined, sequential images are acquired and merged into a single superimposed image. This means that the functional imaging of PET can be aligned with the anatomic imaging of CT scanning. Importantly, PET/CT has prognostic significance and is considered a factor in assessing measurable or minimal residual disease status (MRD).
WHOLE BODY DIFFUSION WEIGHTED IMAGING	Diffusion-Weighted Magnetic Resonance Imaging, also known as Whole Body Diffusion-Weighted Imaging (WB DWI), is an advance in imaging that will likely have an increasing role in detecting and quantifying disease as well as guiding therapy. WB DWI is currently the most sensitive technique for bone marrow imaging.

MAGNETIC RESONANCE IMAGING (MRI)	Magnetic Resonance Imaging results are reported by a Radiologist with an interpretation for your physician. This test is used to monitor changes in lesions. MRI scans are helpful in patients with suspected SMM to rule out focal bone marrow lesions that can be seen before true osteolytic disease occurs. Helpful in assessing extramedullary disease, suspected cord compression, or when detailed imaging of a specific symptomatic area is needed.

HEALTHTREE UNIVERSITY CLASSES

- What imaging is used at diagnosis to detect myeloma bone disease?
- How often should imaging be repeated?
- How is myeloma bone disease monitored?
- What are lytic and focal bone lesions and how common are they?
- What is extramedullary disease?
- What is a plasmacytoma?
- Do you need a CT scan or is an MRI enough?

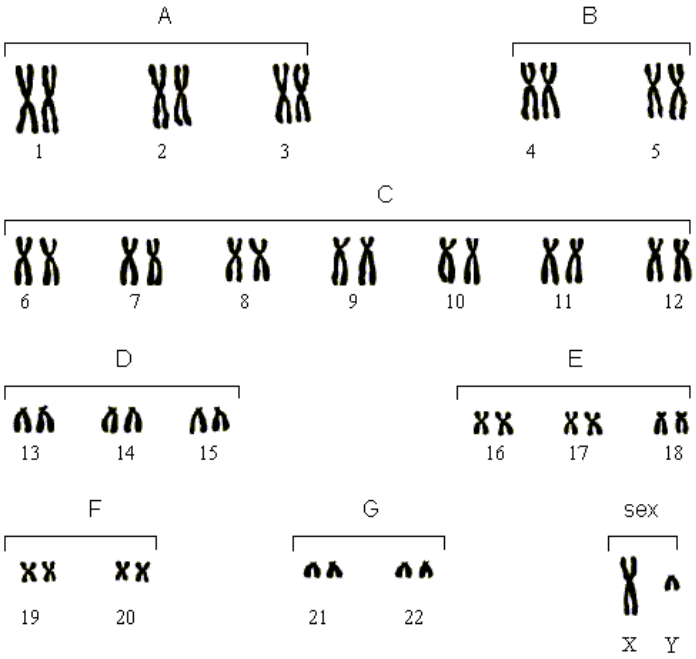
MYELOMA CROWD ARTICLES

- Multiple Myeloma Diagnosis
- Dr. Jens Hillengass from Roswell Park Comprehensive Cancer Center presented Novel Imaging in Multiple Myeloma.
- Myeloma Crowd Round Table with Drs. Jens Hillengass and Ola: Myeloma Imaging

My Myeloma Genetics

In humans, each cell typically contains 23 pairs of chromosomes, for a total of 46. Twenty-two of these pairs, called autosomes, look the same in both males and females. The 23rd pair, the sex chromosomes, differ between males and females.

The 22 pairs are numbered by size (autosomes). The other two chromosomes, X and Y, are the sex chromosomes. A woman will have two X chromosomes, and a man one X and one Y. This picture of the human chromosomes lined up in pairs is called a karyotype. Autosomes are further divided into seven groups: A to G.



Nomenclature

[Total number of chromosomes], [Sex chromosomes], [Chromosomal Alterations] [Number of cells analyzed with that karyotype]

Normal results

46, XX (Female) or 46, XY (Male).

Common symbols for alterations used in karyotypes

del (deletion), t (translocation), dup (duplication), + (gain of), - (loss of), p (the short arm of the chromosome), q (long arm of the chromosome), / (Mosaicism, two populations of cells that coexist with different karyotypes).

Example

47, XX,+21, +21, -14, t(2;8)(q21;p13) [8] / 46, XX [12]: Female with 8 of 20 cells presenting a gain of two chromosomes 21, a loss of one chromosome 14, and a balanced translocation between chromosome 2 and chromosome 8, with breaks in 2q21 and 8p13, and a second cell population (12 of 20 cells) with a normal female karyotype.

Learn more as Dr. Brian Van Ness from the University of Minnesota explains Chromosomes and Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explains Karyotyping-classical cytogenetics.

If you want to learn more about what genetic tests can be done on a sample of bone marrow, you will find here a quick summary made by three Myeloma specialists at HealthTree University.

COMMON GENETIC ABNORMALITIES IN MYELOMA

YOUR RESULTS	WHAT IT MEANS
Hyperdiploidy 	Normal chromosomes or genes typically come in sets of two (2). In healthy people, the chromosomes of plasma cells are in normal pairs. If three copies exist, it is called a **trisomy**. If four copies exist, it is called a **tetrasomy**, and so on. This is also sometimes called hyperdiploid myeloma (too many chromosomes). Hyperdiploid myeloma is typically less aggressive than hypodiploid myeloma (too few chromosomes). This 2015 paper by French myeloma researchers found that trisomies of chromosome 3 and 5 improved overall survival, even for patients with the 4;14 translocation. In the same paper, it was found that trisomy of chromosome 21 worsened overall survival. In this 2017 article, myeloma researchers studied 227 patients with tetrasomies. The most common tetrasomies were 9, 15 and 11. The median overall survival of patients with one or more tetrasomies was superior compared to patients with trisomies (3 copies of the chromosomes) or patients with neither. If the patients had a high-risk genetic feature (like del17p or t4;14) plus a trisomy or tetrasomy, they had better overall survival than those with high-risk features without tetrasomies, but worse than those with no high-risk features. The most common gene addition in multiple myeloma is **a gain of 1q**. This gain occurs in approximately 40% of myeloma patients. According to a recent study, this gene addition is not considered high-risk if you have three copies of the chromosome. It is only considered high-risk if you have four or more copies of the chromosome. Reference: Rapid Assessment of Hyperdiploidy in Plasma Cell Disorders Using a Novel Multi-Parametric Flow Cytometry Method. Learn more as Dr. Brian Van Ness and Dr. Rafael Fonseca explain hyperdiploid myeloma at HealthTree University.
Hypodiploidy 	Normal chromosomes or genes typically come in sets of two (2). In healthy people, the chromosomes of the plasma cells are in normal pairs. In patients with multiple myeloma, genes of the myeloma cells can become deleted. One or both of the genes in a normal pair can be missing. Common gene deletions in myeloma include deletion of chromosome 13, chromosome 1p, or chromosome 17p. A less common deletion is of chromosome 16.

Translocation 	Normal chromosomes or genes typically come in sets of two (2). In healthy people, the chromosomes of plasma cells are in normal pairs. In patients with multiple myeloma, sometimes the chromosomes can break apart and swap places with parts of another chromosome. This is called a translocation. For example, in a 4;14 translocation, part of chromosome 4 has swapped places with chromosome 14. Common translocations include t(4;14), t(11;14), t(14;16), t(14;20). Less common translocations include t(6;14) and t(12;14). Translocations can be considered standard risk or high risk. Standard risk include t(11;14) and t(6;14). High risk include t(4;14), t(14;16) and t(14;20).
Gene Mutation	A gene mutation is an alteration in the DNA sequence that makes up a gene. The gene is a protein inside a cell that helps the cell function properly by providing instructions. It can be inherited or acquired. The mutation can vary in size to involve a single gene or a group of genes on a chromosome. A mutation can cause the cell to malfunction and disrupt normal development or cause a medical condition.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Gareth Morgan from NYU Langone Health, Dr. Cesar Rodriguez Valdes of Wake Forest Baptist Health, and Dr. Brian Van Ness from the University of Minnesota explain what are the different types of chromosomal mutations (e.g. Translocations, Deletions, Insertions, Amplifications) in HealthTree University.

CYTOGENETICS/FISH

YOUR RESULTS	WHAT IT MEANS
Cytogenetics ❑ Normal ❑ Abnormal **Gene additions** ❑ Gain 1q21 **Hyperdiploid** ❑ Trisomy ❑ Tetrasomy **Hypodiploid** ❑ Near-tetraploid ❑ Pseudodiploid ❑ Hypodiploid **Relevant deletions** ❑ 1p ❑ 13q ❑ 16p ❑ 17p (TP53) **Translocations** ❑ FGFR3/MMSET t(4;14) ❑ CCND1 t(11;14) ❑ CCDN2 C-MAF t(14;16) ❑ CCND2 MAFB t(14;20) ❑ MUM1 t(6;14)	**Metaphase cytogenetics** is a test that puts dividing myeloma cells in culture and identifies abnormalities while the cells are dividing. Because myeloma cells can't grow outside the bone marrow environment, only 30% of patients show abnormalities, which means they have more aggressive disease. For the 70% of patients whose myeloma cells don't show abnormalities during cell division, they have less aggressive disease. The advantage of this test is that it identifies the 30% of patients with more aggressive disease, but is not very informative in 70% of patients with less aggressive disease. **The FISH test** allows you to look at certain "hot spots" or probes on the chromosomes and it allows you to find translocations of genes. The FISH test does not need actively dividing cells. This test is informative for all patients. The limitation is that you can only see what you're looking for, or there are only a certain number of probes. The FISH test evaluates the chromosomes in the normal

Pathway Activation/Inactivation ❑ Rb pathway inactivation (P16/INK4a, P18/INK4c, RB1) ❑ PTEN inactivation ❑ P53 inactivation (17p13) **Other Mutations** ❑ C-MYC abnormalities ❑ RAS mutations (K-RAS and N-RAS)	and myeloma cells in the bone marrow. Some may have too many chromosomes, too few chromosomes or other chromosome abnormalities. This test takes approximately 2-3 weeks. It can be used on regular blood or bone marrow samples. The quality of the specimen and of the test matters greatly.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Gareth Morgan of the NYU Langone Health and Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explain FISH testing in HealthTree University.

FLOW CYTOMETRY

YOUR RESULTS	WHAT IT MEANS
❑ Cytoplasmic Kappa ❑ Cytoplasmic Lambda ❑ CD 138 ❑ CD 117 ❑ CD 81 ❑ CD 56 ❑ CD 45 ❑ CD 38 ❑ CD 20 ❑ CD 19	**Flow cytometry** is a test used on both blood samples and bone marrow samples to evaluate for the presence of myeloma cells and can detect low levels of myeloma plasma cells after high-dose therapy and transplantation. It is used as the method of choice to assess minimal residual disease (MRD), it doesn't need a baseline sample, and its sensitivity is 1 in 100,000 cells. It looks in more detail than the immunohistochemistry tests and studies the light chains. This test is used to classify cells according to substances present on their surfaces. Cells are passed in front of a laser beam which causes them to give off light. Groups of cells can be separated and counted. Flow cytometry sensitivity tests can range from 2 color tests (less sensitive) up to 12 color tests (more sensitive). This test helps to identify "markers" on the cell's surface and may give us targets for the use of monoclonal antibodies

NEXT GENERATION SEQUENCING

YOUR RESULTS	WHAT IT MEANS
❑ ATM ❑ BCAR3 ❑ BRAF ❑ DIS3 ❑ FAM46C ❑ FGFR3 ❑ IDH2 ❑ KRAS ❑ NRAS ❑ TP53	**Next Generation Sequencing (NGS)** examines the individual nucleic acids that make up the DNA, this allows the detection of all types of genomic alterations, and unknow translocation patterns. Usually, a targeted sequencing panel is used to test a select number of frequently mutated genes instead of the entire genome, as this is a cheaper and faster approach. The different mutated sequences will be compared with a normal sequence, the frequency of any given change not seen in a healthy person will be reported and categorized. It is used as a method to

❑ TRAF3	assess minimal residual disease (MRD), it needs a baseline sample for assessment, and its sensitivity is 1 in 1 million cells.

HEALTHTREE UNIVERSITY CLASSES

Learn more as Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explains the difference between genetic testing such as FISH and Next Generation Sequencing in HealthTree University. Also you'll find a comparison between MRD by Flow cytometry, and MRD by NGS, explained by Myeloma Specialists in HealthTree University.

MINIMAL RESIDUAL DISEASE (MRD) TESTING

YOUR RESULTS	WHAT IT MEANS
By flow cytometry /100,000 analyzed cells **By NGS** /1 million analyzed cells	**Measurable (or minimal) residual disease (MRD)** refers to the small number of cancer cells that can remain in a patient's body during and after treatment and may eventually cause recurrence of the disease. These residual cells typically cause no physical signs or symptoms and are present at such low levels that more refined and sensitive techniques are required to identify them. Monitoring MRD at various points throughout the course of treatment and remission provides important insight into the status of a patient's disease. **Flow cytometry** is a test used on both blood samples and bone marrow samples to evaluate for the presence of myeloma cells and can detect low levels of myeloma plasma cells after high-dose therapy and transplantation. It is used as the method of choice to assess minimal residual disease (MRD) due to its higher accessibility, it doesn't need a baseline sample, and its sensitivity is 1 in 100,000 cells. **Next Generation Sequencing (NGS)** examines the individual nucleic acids that make up the DNA. This is used as a method to assess minimal residual disease (MRD), it needs a baseline sample for assessment, and it has a higher sensitivity of 1 in 1 million cells. **MRD negativity** does not mean there are no myeloma cells in the bone marrow. It means that there are no myeloma cells in the tested sample. Patients with Sustained MRD Negativity (Remain MRD- for over a year) are more likely to have a longer progression-free survival (PFS) and overall survival(OS). If your institution does not do MRD testing, you can send a bone marrow sample to a lab that does. Mayo Clinic is one of the labs accepted by Medicare.

HEALTHTREE UNIVERSITY CLASSES

Learn more at HealthTree University, where we have a complete chapter dedicated to MRD testing. You can find here the first class in which several faculty doctors explain what MRD is and how useful it is.

GENE EXPRESSION PROFILE

WHAT IT MEANS

You receive a **Prognostic Risk Score** (GEP-70), **Molecular Subtype**, and **Virtual Karyotype** unique to your disease.

The Prognostic Risk score and the Molecular Subtype designations have been clinically shown to predict overall survival, event-free survival, complete response duration, and post-relapse survival.

Risk Score: MyPRS distinguishes between patients with high and low-risk disease.
High-Risk: Poor Prognosis, Probability of 5-year OS 38%
Low Risk: Good Prognosis, Probability of 5 year OS 83%

Molecular Subtype: You receive 1 of 7 molecular subtypes associated with your unique genetic lesions, altered genes and outcome variation.

Virtual Karyotype: Your Cytogenetic (FISH) results checking for high-risk chromosome abnormalities.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Gareth Morgan of the NYU Langone Health and Dr. Brian Van Ness from the University of Minnesota explain Gene Expression Profile (GEP) in HealthTree University.

SINGLE NUCLEOTIDE POLYMORPHISM (SNP) ARRAY

WHAT IT MEANS

Single Nucleotide Polymorphism (SNP) Array is a genetic test currently not used in the clinical setting, that allows you to look for differences between cells, and is typically used as a way to estimate gains and losses of chromosomal material in the whole genome. It can be used for relevant mutations like Gain 1q21, or Deletions 17p/16p/13q/1p.

HEALTHTREE UNIVERSITY CLASS

Learn more as Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explains what is a SNP Array in HealthTree University.

SPECIFIC GENETIC ABNORMALITIES IN MYELOMA

YOUR RESULTS	WHAT IT MEANS
Gain 1q 	The addition of 1q or gain of 1q is found in 38-40% of myeloma patients. According to a recent study, this gene addition is not considered high-risk if you have **three copies** of the chromosome. It is only considered high-risk if you have **four copies** of the chromosome. The number of copies of the 1q gain is available on your FISH test. If you can't find your number of copies, ask your doctor if the information is available and include the number of copies in the comments section of the FISH test results in HealthTree. In this study, when Next Generation Sequencing was used for 95 patients with the 1q gain, it was found that the most common additional mutations found in 1q gain patients included TP53 (38%) and KRAS (25%). References: • Nature.com - Prediction of outcome in newly diagnosed myeloma • Blood Journal - Outcome of Multiple Myeloma with Chromosome 1q Gain
	Learn more as Dr. Martin Kaiser from The Institute of Cancer Research (London), and Dr. Cesar Rodriguez Valdes of Wake Forest Baptist Health, explain different aspects of the gain 1q in HealthTree University.
Deletion 1p 	The deletion of 1p occurs in 8.4% of myeloma patients. It is believed that deletion of the short arm of chromosome 1 results in the loss of tumor suppressor genes that are part of P73, a member of the TP53 family. TP53 is the master cancer regulator gene. According to studies, the deletion of 1p is associated with shorter remission and survival in patients undergoing autologous stem cell transplant. Patients with del1p may require more intensive therapy. References: • ScienceDirect - Deletion of the Short Arm of Chromosome 1 (del 1p) is a Strong Predictor of Poor Outcome in Myeloma Patients Undergoing an Autotransplant • NCBI - Mapping of Chromosome 1p Deletions in Myeloma • Blood Journal: Outcome of Multiple Myeloma with Chromosome 1q Gain & 1p Deletion after High-Dose Therapy and Autologous Hematopoietic Stem Cell Transplantation

Monosomy 13 	A deletion 13 or monosomy 13 means that the 13 chromosome is missing inside of the multiple myeloma cells. This deletion is commonly found in 59% of myeloma patients. It is not considered a high-risk feature if only found on the FISH test results. It can be considered high risk if it is also found on conventional (also called metaphase) cytogenetics (present in only 17% of patients). The deletion of 13 is commonly found together with the t(4;14) translocation (in 90% of del13 patients). References: • Journal of Clinical Oncology - Clinical utility of the comprehensive approach to detect genetic abnormalities in multiple myeloma • Blood Journal - Treatment of Multiple Myeloma with high-risk cytogenetics: a consensus of the International Myeloma Working Group • Blood Journal - Acquired Chromosomal Abnormalities t(14;16), Del(17p) or Del(13q) after Receiving Chemotherapies Predict the Poor Prognosis of Multiple Myeloma
Deletion 16q 	A deletion 16q occurs in an unknown percentage of myeloma patients. It is considered a higher risk feature. References: • Blood Journal - Gene mapping and expression analysis of 16q loss of heterozygosity identifies WWOX and CYLD as being important in determining clinical outcome in multiple myeloma • Wiley - A molecular diagnostic approach able to detect the recurrent genetic prognostic factors typical of presenting myeloma
Deletion 17p 	A deletion 17p means that one or both of the 17 chromosomes are missing inside of the multiple myeloma cells. This deletion is found in approximately 8-10% of newly diagnosed myeloma patients. The deletion of 17p has an associated gene - TP53. The TP53 gene is the master cancer regulator gene, so when the chromosomes are deleted, myeloma is not regulated normally. Myeloma patients can commonly acquire this gene deletion as their disease progresses. According to Dr. Robert Orlowski, “Some studies suggest that it (del17p) can be as high as 30% and a few even suggest that it may be as high as 50% for patients in the refractory area.” While deletion 17p is typically considered a high-risk feature, it may not be high-risk in all patients. Based on study data, it was found that if only one of the 17p chromosomes were deleted, the myeloma did not behave as high risk. If both of the 17p chromosomes had been deleted, the myeloma behaved as high-risk. This information will not show on your FISH test. You need a Next Generation Sequencing test to find this information. The percentage of del17p cells is important. For example, patients with low percentages of del17p such as 5% will typically be considered lower risk than patients with more than 60% of del17p. Deletion 17p patients may respond better to proteasome inhibitors when used as induction therapy and as maintenance according to recent studies. Other

	treatment approaches may include three or four-drug induction therapies, one or two transplants, and maintenance therapies with double or triple agents as opposed to a single maintenance therapy. References: • Journal Of Clinical Oncology - Mutational Spectrum, Copy Number Changes, and Outcome: Results of a Sequencing Study of Patients With Newly Diagnosed Myeloma • NCBI - The level of deletion 17p and bi-allelic inactivation of TP53 has a significant impact on clinical outcome in multiple myeloma • MyelomaCrowd - Why is del17p in multiple myeloma so aggressive? • Sue Armstrong - p53: The Gene that Cracked the Cancer Code • NCBI - Treatment of multiple myeloma with high-risk cytogenetics: a consensus of the International Myeloma Working Group • nature.com - A high-risk, Double-Hit, group of newly diagnosed myeloma identified by genomic analysis • MyelomaCrowd - ASH 2018: High Risk and "Double Hit" Myeloma • MyelomaCrowd - MCRI Full Show: Overcoming Deletion 17p Using a Specifically Targeted Therapy with Dr. Robert Orlowski, MD, PhD, MD Anderson
	Learn more as Dr. Fotis Asimakopoulos from Moores Cancer Center (UCSD), Dr. Cesar Rodriguez Valdes of Wake Forest Baptist Health, Dr. Donna Reece from Princess Margaret Cancer Centre, and Dr. Leif Bergsagel of the Mayo Clinic (Scottsdale) explain different aspects of deletion 17p in HealthTree University: • What is a 17p deletion? Why is Del of 17p considered high risk? • Can only a percentage of your myeloma cells possess 17p deletion? How is that percentage determined? • What does it mean if you have a mutation in chromosome 17 and a deletion? HowNext-Generation? How does this affect risk? • Will Del17p and other chromosomal abnormalities go away after an auto ASCT? If not why may it not be detected?
Translocation (6;14) 	Approximately 1% of patients have the 6;14 translocation. This means that part of chromosome 6 has swapped places with part of chromosome 14. When this happens, it affects the Cyclin D3 gene or CCND3, which can turn cells into cancer cells. References: NCBI - Cyclin D3 at 6p21 is dysregulated by recurrent chromosomal translocations to immunoglobulin loci in multiple myeloma

Translocation (11;14) 	Approximately 15% of myeloma patients have the 11;14 translocation. This means that part of chromosome 11 has swapped places with part of chromosome 14. It is the most common translocation. It is considered a standard risk feature and can be commonly found in patients of African descent. When this happens, it increases the regulation of Cyclin D1 or CCND1. A helpful therapy that looks particularly promising for this translocation is venetoclax. References: • nature.com - Natural history of t(11;14) multiple myeloma • EHA - Deep And Sustained Response After Venetoclax Therapy In A Patient With Very Advanced Refractory Myeloma With Translocation T(11;14) • ScienceDirect - Clinical Implications of t(11;14) in Patients with Multiple Myeloma Undergoing Autologous Stem Cell Transplantation • Blood Journal - Variable BCL2/BCL2L1 ratio in multiple myeloma with t(11;14)
Translocation (14;16) 	Approximately 3% of patients have the 14;16 translocation. This means that part of chromosome 14 has swapped places with part of chromosome 16. When this happens, it affects the c-MAF gene, which can turn cells into cancer cells. It is generally considered a high-risk feature. References: • Blood Journal - Translocation t(14;16) and multiple myeloma: is it really an independent prognostic factor? • Clinical Lymphoma, Myeloma & Leukemia - Translocation (14;16) Positive Multiple Myeloma: Clinical Features and Survival Outcomes of a High-risk Population
Translocation (14;20) 	Approximately 2% of patients have the 14;20 translocation. This means that part of chromosome 14 has swapped places with part of chromosome 20. When this happens, it affects the MAFB gene, which can turn cells into cancer cells. It is generally considered a high-risk feature. References: • NCBI - The t(14;20) is a poor prognostic factor in myeloma but is associated with long-term stable disease in monoclonal gammopathies of undermined significance. • Wiley - The recurrent translocation t(14;20)(q32;q12) in multiple myeloma results in aberrant expression of MAFB
Translocation (4;14) 	Approximately 12% of patients have the 4;14 translocation. This means that part of chromosome 4 has swapped places with part of chromosome 14. When this happens, it increases the fibroblast growth factor receptor 3 gene (FGFR3) and the multiple myeloma Set domain gene (MMSET). This is considered a high-risk feature. References: • Blood Journal - Lenalidomide Maintenance Significantly Improves Outcomes Compared to Observation Irrespective of Cytogenetic Risk: Results of the Myeloma XI Trial

	• Blood Journal - MMSET I Acts As an Oncoprotein and Regulates GLO1 Expression in T(4;14) Multiple Myeloma Cells
HEALTHTREE UNIVERSITY CLASS	
Learn more as Dr. Fotis Asimakopoulos from Moores Cancer Center (UCSD) explains the importance of **Chromosome 14** and its relation to high-risk myeloma in HealthTree University.	
Learn more about **translocation 4;14** and its relation to high-risk myeloma in Myeloma Crowd.	

AI 輔助翻譯與醫療參考提醒

解讀我的骨髓瘤檢驗報告：詳細版中英對照

閱讀後再次提醒

本 PDF 由 AI 協助翻譯、整理與重排，目的是協助病友與家屬對照英文原文，理解多發性骨髓瘤相關檢驗資訊。

本資料僅供一般資訊參考，不能取代醫師診斷、治療建議、用藥調整或個別化醫療判斷。

不同醫院、不同實驗室的檢驗方法與參考區間可能不同；若檢驗數值偏高、偏低或有疑問，請以您的主治醫師說明為準。

感謝您閱讀。本資料適合帶著問題與醫療團隊討論；請勿只依據本 PDF 自行判斷病情或調整治療。

資料來源：Deciphering Myeloma Labs；本頁為繁體中文 AI 輔助翻譯與閱讀版重排。
網站：https://healthtreetw.org/posts/know-05/