National Genomic Test Directory haematological oncology (HaemOnc) eligibility criteria

国家基因组检测目录 血液肿瘤血液肿瘤(HaemOnc)检测资格标准检测资格标准

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1. Test package: Acute Myeloid Leukaemia TP450

Acute Myeloid Leukaemia Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

NB some M codes are just listed as GTas these tests are aligned to the simplification process

WGS tests

GT726 / M80.1 WGS Germline & Tumour - AML
GT24 / M80.57 WGS Tumour First - AML
GT802 / M80.58 WGS Follow-up Germline - AML

GT517 / M90.1 WGS Germline & Tumour - BPDCN
GT879 / M90.6 WGS Tumour First - BPDCN
GT1221 / M90.7 WGS Follow-up Germline - BPDCN

All patients (of any age i.e. paediatric, TYA and adult) with a morphological diagnosis of acute myeloid leukaemia (AML) are eligible for whole genome sequencing (WGS), primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / (RELAPSE/REFRACTORY)

Tumour and germline samples can be submitted simultaneously (i.e M80.1) or separately i.e. tumour first (M80.57) followed by a germline when/if available (M80.58). At any presentation point either M80.1 or M80.57 +/- M80.58 is permissible, however once a non-contaminated germline has been submitted (either as part of M80.1 or M80.58) this can be bioinformatically re-used as a germline if any subsequent tumour samples are submitted for WGS.

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

Targeted tests

GT533 / M80.18 (*FLT3* ITD)

1.检测套餐：急性髓系白血病TP450

急性髓系白血病检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

NB部分M代码仅作为GTas列出因为这些检测已对齐至简化流程流程

WGS 检测

GT726 / M80.1 WGS 胚系与肿瘤 - AML GT24 / M80.57 WGS 肿瘤优先 - AML

GT802 / M80.58 WGS 随访胚系 - AML

GT517 / M90.1 WGS 胚系与肿瘤 - BPDCN GT879 / M90.6 WGS 肿瘤优先 - BPDCN

GT1221 / M90.7 WGS随访胚系-BPDCN

所有年龄(即儿童、青少年和青年及成人)经形态学诊断为急性髓系白血病(AML)的患者均有资格接受全基因组测序(WGS)，主要在诊断时进行。若诊断时未进行 WGS，在复发或对某线治疗难治时进行该检测可能合适。在特殊情况下，若有高概率发生显著克隆演化，可能指示重复 WGS。诊断/(复发/难治)

肿瘤和胚系样本可同时提交(即 M80.1)或分别提交，即先提交肿瘤样本(M80.57)，随后在可获得时提交胚系样本(M80.58)。在任何就诊节点，允许 M80.1 或 M80.57 +/- M80.58，但一旦提交了未受污染的胚系样本(作为 M80.1 或 M80.58 的一部分)，若随后提交任何肿瘤样本进行 WGS，该胚系样本可通过生物信息学方式重新用作胚系。

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

靶向检测

GT533 / M80.18（FLT3 ITD）

GT916 / M80.21 (*FLT3* TKD)
GT921 / M80.22 (*NPM1*)
GT494 / M80.23 (*IDH1*)
GT649 *BCR::ABL1*-multiplex

- For patients with a morphological diagnosis of AML at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria [*NPM1*]. DIAGNOSIS
- For patients with a morphological diagnosis of acute myeloid leukaemia with concerns it had evolved from background chronic myeloid leukaemia. DIAGNOSIS
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification for allogeneic haematopoietic stem cell transplant if appropriate [*NPM1, FLT3* ITD]. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors) [*NPM1, FLT3* ITD*, FLT3* TKD*, IDH1*]. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML receiving/received targeted inhibitor treatment where assessment of disease response at a molecular level may influence the subsequent treatment strategy [*FLT3* ITD*, FLT3* TKD*, IDH1*]. (NB the detection limits of any assay used for this testing indication should be considered when determining how informative such testing is).

Small variant / copy number detection via panel

GT253 / M80.2 (NGS small variant panel)
GT629 / M80.53 (NGS copy number variant panel – to be delivered in conjunction with the NGS small variant panel. NB It is envisaged that any copy number variant [CNV] panel will have a SNP backbone or similar, therefore allowing detection of CNVs across the genome as opposed to be limited to the target regions in the test directory)

- For patients with a morphological diagnosis of AML at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification for allogeneic haematopoietic stem cell transplant if appropriate. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory to inform treatment (may include conventional

GT916 / M80.21（FLT3 TKD）
GT921 / M80.22（NPM1）
GT494 / M80.23（IDH1）
GT649 BCR::ABL1-多重检测

- 对于诊断时形态学诊断为 AML 的患者，以根据 WHO 2022 或 ICC 标准更好地完善诊断[*NPM1*]。诊断
- 对于形态学诊断为急性髓系白血病、但担心其由背景慢性髓性白血病演变而来的患者。诊断
- 对于诊断/复发/难治就诊时形态学诊断为 AML 的患者，用于预后意义，包括适当时用于异基因造血干细胞移植的风险分层[*NPM1, FLT3* ITD]。诊断 / 复发/难治
- 对于诊断/复发/难治时形态学诊断为 AML 的患者，用于指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)[*NPM1, FLT3* ITD，*FLT3* TKD，*IDH1*]。诊断 / 复发/难治
- 对于形态学诊断为 AML、正在接受/已接受靶向抑制剂治疗的患者，其中分子水平的疾病反应评估可能影响后续治疗策略[*FLT3* ITD，*FLT3* TKD，*IDH1*]。(NB 在确定此类检测的信息价值时，应考虑用于该检测指征的任何检测方法的检测限)。

通过 panel 检测小变异/拷贝数

GT253 / M80.2(NGS小变异panel)
GT629 / M80.53(NGS 拷贝数变异检测板 - 与 NGS 小变异检测板联合提供。NB 预计任何拷贝数变异[CNV]检测板都将具有 SNP 骨架或类似结构，因此允许检测全基因组范围内的 CNV，而非仅限于检测目录中的目标区域)

- 对于诊断时形态学诊断为 AML 的患者，以根据 WHO 2022 或 ICC 标准更好地完善诊断。诊断
- 对于诊断/复发/难治就诊时形态学诊断为 AML 的患者，用于预后意义，包括适当时用于异基因造血干细胞移植的风险分层。诊断 / 复发/难治
- 对于诊断/复发/难治时形态学诊断为 AML 的患者，用于指导治疗(可能包括常规

chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELAPSE/REFRACTORY

- For patients with a morphological diagnosis of AML receiving/received curative intent treatment where assessment of disease response at a molecular level may influence the subsequent treatment strategy (NB to use an NGS assay for this purpose it must be of suitable sensitivity/sequencing depth to be informative and it should be noted that a standard depth NGS panel is not a formal measurable residual disease [MRD] assay). It is expected that using this test for this indication will be the exception rather than the norm. MONITORING DURING / POST-TREATMENT

化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

- 对于经形态学诊断为 AML 并正在/已经接受治愈性意图治疗的患者，若在分子水平评估疾病反应可能影响后续治疗策略(注意：为此目的使用 NGS 检测时，其灵敏度/测序深度必须适合以提供信息，并应注意标准深度的 NGS 检测板并非正式的可测量残留病灶[MRD]检测)。预计将此检测用于该适应证将是例外而非常规。治疗期间/治疗后监测

Global copy number +/- structural variant via cytogenetics

GT1096 / M80.3 (Karyotype)
GT17 (SNP array) – used as a salvage test in the event of insufficient metaphases being available for karyotype analysis.

- For patients with a morphological diagnosis of AML at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. NB SNP array would typically be used in the event of karyotype failure rather than as a first-line diagnostic test. DIAGNOSIS
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification for allogeneic haematopoietic stem cell transplant if appropriate. NB SNP array would typically be used in the event of karyotype failure rather than as a first-line diagnostic test. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens or targeted inhibitors). NB SNP array would typically be used in the event of karyotype failure rather than as a first-line diagnostic test. DIAGNOSIS / RELAPSE/REFRACTORY

通过细胞遗传学检测全局拷贝数 +/- 结构变异

GT1096 / M80.3(核型)
GT17(SNP 阵列)- 在可用于核型分析的中期分裂相不足时用作补救检测。

- 对于诊断时形态学诊断为 AML 的患者，以根据 WHO 2022 或 ICC 标准更好地完善诊断。NB SNP 阵列通常会在核型分析失败时使用，而非作为一线诊断检测。诊断
- 对于诊断/复发/难治就诊时形态学诊断为 AML 的患者，用于预后意义，包括适当时用于异基因造血干细胞移植的风险分层。NB SNP 阵列通常会在核型分析失败时使用，而非作为一线诊断检测。诊断 / 复发/难治
- 对于诊断/复发/难治时形态学诊断为 AML 的患者，用于指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。NB SNP 阵列通常会在核型分析失败时使用，而非作为一线诊断检测。诊断 / 复发/难治

Targeted copy number +/- structural variant via FISH / RT-PCR

M80.5, M80.25- M80.40, M80.54 (Individual FISH tests)
Replaced by GT101, GT1033, GT1064, GT112, GT116, GT1294, GT113, GT14, GT157, GT180, GT258, GT275, GT289, GT61, GT656, GT700, GT716, GT74 (see excel spreadsheet for individual test mapping)
M80.7, M80.41-M80.52 (Individual RT-PCR tests) non-quantitative
Replaced by GT351 (see excel spreadsheet for individual test mapping)

- For patients with a morphological diagnosis of AML at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. The specific identify of

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

M80.5、M80.25- M80.40、M80.54(单项FISH检测)
由 GT101、GT1033、GT1064、GT112、GT116、GT1294、GT113、GT14、GT157、GT180、GT258、GT275、GT289、GT61、GT656、GT700、GT716、GT74 替代(各检测对应关系见 excel 表格)M80.7、M80.41-M80.52(单项 RT-PCR 检测)非定量由 GT351 替代(各检测对应关系见 excel 表格)

- 用于初诊时形态学诊断为 AML 的患者，以根据 WHO 2022 或 ICC 标准更好地完善诊断。具体的

FISH / RT-PCR targets should correlate with national guidelines (e.g. BSH Good Practice Paper: https://doi.org/10.1111/bjh.18516) and will reflect local testing strategy in order to meet nationally mandated genomic turn-around-times (TATs). It is expected that FISH / RT-PCR testing will not duplicate other structural / copy number variant testing unless required to meet rapid TATs. DIAGNOSIS

- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification for allogeneic haematopoietic stem cell transplant if appropriate. The specific identify of FISH / RT-PCR targets should correlate with national guidelines (e.g. BSH Good Practice Paper: https://doi.org/10.1111/bjh.18516) and will reflect local testing strategy in order to meet nationally mandated genomic turn-around-times (TATs). It is expected that FISH / RT-PCR testing will not duplicate other structural / copy number variant testing unless required to meet rapid TATs. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). The specific identify of FISH / RT-PCR targets should correlate with national guidelines (e.g. BSH Good Practice Paper: https://doi.org/10.1111/bjh.18516) and will reflect local testing strategy in order to meet nationally mandated genomic turn-around-times (TATs). It is expected that FISH / RT-PCR testing will not duplicate other structural / copy number variant testing unless required to meet rapid TATs. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML receiving/received treatment where assessment of disease response at this level of sensitivity (i.e. detection by FISH – see below for formal RT-qPCR MRD testing) may influence the subsequent treatment strategy. MONITORING DURING / POST-TREATMENT

Structural variant detection via NGS panel

GT185 / M80.8 (NGS Structural variant panel)

- For patients with a morphological diagnosis of AML at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as karyotype and NGS small variant panel if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of

FISH / RT-PCR 靶点应与国家指南(例如 BSH 良好实践文件：https://doi.org/10.1111/bjh.18516)相一致，并应反映本地检测策略，以满足国家规定的基因组周转时间(TATs)。预期 FISH / RT-PCR 检测不会重复其他结构 / 拷贝数变异检测，除非为满足快速 TATs 所需。诊断

- 用于初诊 / 复发 / 难治时形态学诊断为 AML 的患者，以提供预后意义，包括适当时进行异基因造血干细胞移植的风险分层。FISH / RT-PCR 靶点的具体身份应与国家指南(例如 BSH 良好实践

文件：https://doi.org/10.1111/bjh.18516)相一致，并应反映本地检测策略，以满足国家规定的基因组周转时间(TATs)。预期 FISH / RT-PCR 检测不会重复其他结构 / 拷贝数变异检测，除非为满足快速 TATs 所需。诊断 / 复发 / 难治

- 用于初诊 / 复发 / 难治时形态学诊断为 AML 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。FISH / RT-PCR 靶点的具体身份应与国家指南(例如 BSH 良好实践

文件：https://doi.org/10.1111/bjh.18516)相一致，并应反映本地检测策略，以满足国家规定的基因组周转时间(TATs)。预期 FISH / RT-PCR 检测不会重复其他结构 / 拷贝数变异检测，除非为满足快速 TATs 所需。诊断 / 复发 / 难治

- 用于形态学诊断为 AML 且正在接受 / 已接受治疗的患者，在此灵敏度水平上评估疾病缓解(即通过 FISH 检测--正式的 RT-qPCR MRD 检测见下文)可能影响后续治疗策略。治疗期间 / 治疗后监测

通过NGS panel进行结构变异检测

GT185 / M80.8(NGS结构变异检测组合)

- 对于诊断时经形态学诊断为 AML 的患者，以根据 WHO 2022 或 ICC 标准更好地细化诊断。预计 NGS 结构变异检测板最通常在其他诊断(如核型和 NGS 小变异检测板)之后使用，前提是未识别出可能的驱动基因组改变。已确认不同实验室可能有略微不同的检测策略，这意味着该检测在路径中可能更频繁或更早使用。其局限性

a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology. DIAGNOSIS

- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification for allogeneic haematopoietic stem cell transplant if appropriate. It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as karyotype and NGS small variant panel if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as karyotype and NGS small variant panel if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology. DIAGNOSIS / RELAPSE/REFRACTORY
- For patients with a morphological diagnosis of AML at diagnosis for whom a curative approach is being pursued to identify an appropriate target for formal measurable residual disease (MRD) monitoring. It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as karyotype and NGS small variant panel if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology. DIAGNOSIS

Resistance mutation testing

GT303 / M80.15 (*BCR::ABL1* TKD variant testing)

- For patients with a morphological diagnosis of AML known to have a *BCR::ABL1* rearrangement whose response to a targeted inhibitor is such that there is clinical suspicion that there may be a resistance variant within the *ABL1* tyrosine kinase domain. NB this assay should have a minimum variant allele frequency level of detection of 5% (i.e. 5% of

应认识到靶向 RNA 检测板，使得原癌基因的失调不太可能通过此方法检测到。诊断

- 对于诊断时/复发/难治表现时经形态学诊断为 AML 的患者，用于预后意义，包括适当时用于异基因造血干细胞移植的风险分层。预计 NGS 结构变异检测板最通常在其他诊断(如核型和 NGS 小变异检测板)之后使用，前提是未识别出可能的驱动基因组改变。已确认不同实验室可能有略微不同的检测策略，这意味着该检测在路径中可能更频繁或更早使用。应认识到靶向 RNA 检测板的局限性，即此方法不太可能检测到原癌基因的失调。诊断/复发/难治

- 对于诊断时/复发/难治时经形态学诊断为 AML 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。预计 NGS 结构变异检测板最通常在其他诊断(如核型和 NGS 小变异检测板)之后使用，前提是未识别出可能的驱动基因组改变。已确认不同实验室可能有略微不同的检测策略，这意味着该检测在路径中可能更频繁或更早使用。应认识到靶向 RNA 检测板的局限性，即此方法不太可能检测到原癌基因的失调。诊断/复发/难治

- 对于诊断时形态学诊断为 AML 且正在接受治愈性治疗的患者，以确定用于正式可测量残留病灶(MRD)监测的合适靶标。预计 NGS 结构变异 panel 通常会在其他诊断(如核型分析和 NGS 小变异 panel)之后使用，前提是尚未识别出可能的驱动性基因组改变。需认识到不同实验室的检测策略可能略有差异，这意味着该检测在路径中可能被更频繁或更早地使用。应认识到靶向 RNA panel 的局限性，即原癌基因的失调不太可能通过该方法检出。诊断

耐药突变检测

GT303 / M80.15(*BCR::ABL1* TKD 变异检测)

- 用于形态学诊断为 AML 且已知具有*BCR::ABL1*重排的患者，其对靶向抑制剂的反应使得临床怀疑*ABL1*酪氨酸激酶结构域内可能存在耐药变异。注意：该检测的最低变异等位基因频率检测水平应为 5%(即 5% 的

the *BCR::ABL1* transcripts need to bear the variant). MONITORING DURING TREATMENT

Measurable residual disease testing

GT1062 / M80.9 (*NPM1* MRD)
GT1228 / M80.56 (Rare *NPM1* transcript MRD)
GT475 / M80.10 (*PML::RARA* MRD)
GT343 / M80.11 (*RUNX1::RUNX1T1* MRD)
GT870 / M80.12 (*CBFB::MYH11* MRD)
GT31 / M80.13 (*BCR::ABL1* MRD)
GT673 / M80.55 (Rare *BCR::ABL1* MRD)
GT703 / M80.14 (Other MRD)

- For patients with a morphological diagnosis of AML, known to have the specific described genomic aberration of the assay, receiving/received curative treatment where assessment of disease response at a measurable residual disease [MRD] level may influence the subsequent treatment strategy. The frequency of such monitoring should follow any available national guidelines or otherwise be concentrated at points where knowledge of the transcript level has the potential to influence treatment strategy. MONITORING DURING / POST-TREATMENT

These eligibility criteria pertain only to acquired genomic abnormalities. Information regarding eligibility for germline testing in the context of AML (including predictive testing of potential carriers) is available in the rare disease test directory.

*BCR::ABL1*转录本需要携带该变异)。治疗期间监测

可测量残留病检测

GT1062 / M80.9（NPM1 MRD）
GT1228 / M80.56(罕见*NPM1*转录本 MRD)GT475 / M80.10(*PML::RARA* MRD)
GT343 / M80.11（RUNX1::RUNX1T1 微小残留病）GT870 / M80.12（CBFB::MYH11 微小残留病）GT31 / M80.13（BCR::ABL1 微小残留病）
GT673 / M80.55(罕见*BCR::ABL1* MRD) GT703 / M80.14(其他 MRD)

- 对于形态学诊断为 AML、已知具有该检测所描述的特定基因组异常、正在接受/已接受治愈性治疗的患者，当在可测量残留病灶(MRD)水平评估疾病反应可能影响后续治疗策略时。此类监测的频率应遵循任何可用的国家指南，否则应集中在了解转录本水平有可能影响治疗策略的时间点。治疗期间/治疗后监测

这些检测资格标准仅适用于获得性基因组异常。关于 AML 背景下胚系检测资格的信息(包括对潜在携带者的预测性检测)可在罕见病检测目录中获取。

2. Test package: Myelodysplastic Syndromes TP202

Myelodysplastic syndrome (including myelodysplastic / myeloproliferative neoplasms & hypoplastic MDS/ aplastic anaemia differential) Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT1312 / M82.6 WGS Germline & Tumour - MDS
GT1439 / M82.22 WGS Tumour First - MDS
GT1023 / M82.23 WGS Follow-up Germline - MDS

GT1382 / M83.4 WGS Germline & Tumour - Aplastic Anaemia
GT1340 / M83.5 WGS Tumour First - Aplastic Anaemia
GT304 / M83.6 WGS Follow-up Germline - Aplastic Anaemia

GT1238 / M88.2 WGS Germline & Tumour - JMML
GT702 / M88.11 WGS Tumour First - JMML
GT1114 / M88.12 WGS Follow-up Germline - JMML

GT265 / M224.4 WGS Germline & Tumour - MDS/MPN
GT889 / M224.41 WGS Tumour First - MDS/MPN
GT128 / M224.42 WGS Follow-up Germline - MDS/MPN

NB Myelodysplasia (MDS), Juvenile myelomonocytic leukaemia (JMML), Myelodysplastic/myeloproliferative neoplasms (MDS/MPN) will be referred to under the umbrella term of dysplastic neoplasm in the remainder of this document.

All paediatric and TYA patients with a confirmed or suspected diagnosis of a dysplastic neoplasm OR aplastic anaemia are eligible for WGS primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / (REFRACTORY)

Tumour and germline samples can be submitted simultaneously (i.e M82.6 / M83.4 / M88.2 / M224.4) or separately i.e. tumour first (M82.22 / M83.5 /

2.检测套餐：骨髓增生异常综合征TP202

骨髓增生异常综合征(包括骨髓增生异常 / 骨髓增殖性肿瘤及低增生性 MDS / 再生障碍性贫血鉴别)检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT1312 / M82.6 WGS 胚系与肿瘤 - MDS GT1439 / M82.22 WGS 肿瘤优先 - MDS
GT1023 / M82.23 WGS 随访胚系 - MDS

GT1382 / M83.4 WGS 胚系与肿瘤 - 再生障碍性贫血 GT1340 / M83.5 WGS 肿瘤优先 - 再生障碍性贫血
GT304 / M83.6 WGS随访生殖系--再生障碍性贫血

GT1238 / M88.2 WGS 胚系与肿瘤 - JMML GT702 / M88.11 WGS 肿瘤优先 - JMML
GT1114 / M88.12 WGS 随访胚系 - JMML

GT265 / M224.4 WGS 胚系与肿瘤 - MDS/MPN GT889 / M224.41 WGS 肿瘤优先 - MDS/MPN
GT128 / M224.42 WGS随访胚系--MDS/MPN

注意：骨髓增生异常(MDS)、幼年型粒单核细胞白血病(JMML)、骨髓增生异常 / 骨髓增殖性肿瘤(MDS/MPN)在本文档剩余部分将统称为发育异常性肿瘤。

所有确诊或疑似发育异常性肿瘤或再生障碍性贫血的儿童和 TYA 患者主要在一线诊断时有资格接受 WGS。如果诊断时未进行 WGS，在一线治疗难治时进行该检测可能是合适的。在特殊情况下，如果极有可能发生显著的克隆演化，则可能指示重复 WGS。诊断 /(难治)

肿瘤和胚系样本可同时提交(即 M82.6 / M83.4 / M88.2 / M224.4)或分别提交，即肿瘤优先(M82.22 / M83.5 /

M88.11 / M224.41) followed by a germline when/if available (M82.23 / M83.6 / M88.12 / M224.42). At any presentation point either M82.6 or M82.22 +/- M82.23, M83.4 or M83.5 +/- M83.6, M88.2 or M88.11 +/- M88.12, M224.4 or M224.41 +/- M224.42 is permissible, however once a non-contaminated germline has been submitted (either as part of M82.6 / M83.4 / M88.2 / M224.4 or M82.23 / M83.6 / M88.12 / M224.42) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

For adult patients with a confirmed or suspected diagnosis of a dysplastic neoplasm or aplastic anaemia, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Small variant / copy number detection via panel

GT1297 / M82.1 (NGS small variant panel – it is envisaged that the choice of gene footprint will ensure all relevant variants including putative resistance mutations are included).
GT397 / M82.16 (NGS copy number variant panel – to be delivered in conjunction with the NGS small variant panel. NB It is envisaged that any copy number variant [CNV] panel will have a SNP backbone or similar, allowing detection of CNVs across the genome as opposed to limited to the target regions in the test directory).

- For patients with a suspected diagnosis of a dysplastic neoplasm or aplastic anaemia where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia where demonstration of a specific genomic abnormality would allow access to a targeted treatment (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have undergone a change in their disease phenotype

M88.11 / M224.41)，随后在可用时 / 如果可用时提交胚系(M82.23 / M83.6 / M88.12 / M224.42)。在任何就诊点，M82.6 或 M82.22 +/- M82.23、M83.4 或 M83.5 +/- M83.6、M88.2 或 M88.11 +/- M88.12、M224.4 或 M224.41 +/- M224.42 均允许，然而一旦提交了未受污染的胚系(作为 M82.6 / M83.4 / M88.2 / M224.4 或 M82.23 / M83.6 / M88.12 / M224.42 的一部分)，该胚系可通过生物信息学方式重新用于所有后续肿瘤样本。

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

对于确诊或疑似发育异常性肿瘤或再生障碍性贫血的成人患者，WGS 可在适应症 M235 下获得，即已穷尽标准诊疗检测和 / 或治疗，前提是符合该检测资格标准。样本可同时提交(M235.1)或分别提交，即肿瘤优先(M235.2)，随后在可用时 / 如果可用时提交胚系(M235.3)。

通过 panel 检测小变异/拷贝数

GT1297 / M82.1(NGS 小变异检测板--预期基因覆盖范围的选择将确保包含所有相关变异，包括推定耐药突变)。

GT397 / M82.16(NGS 拷贝数变异检测板--与 NGS 小变异检测板联合提供。注意：预期任何拷贝数变异[CNV]检测板将具有 SNP 骨架或类似结构，允许检测全基因组范围内的 CNV，而非仅限于检测目录中的靶区域)。

- 用于疑似发育异常性肿瘤或再生障碍性贫血的患者，其中克隆标志物的证明将有助于诊断和 WHO 2022 或 ICC 分类。诊断
- 用于已知诊断为发育异常性肿瘤或再生障碍性贫血的患者，以协助预后判断，从而指导患者管理。诊断
- 用于已知诊断为发育异常性肿瘤或再生障碍性贫血的患者，其中特定基因组异常的证明将允许获得靶向治疗(注意：应预期基因组确定的治疗将在特定异常的证明(或不证明)后可用)。诊断、治疗前、治疗变更前
- 用于已知诊断为发育异常性肿瘤或再生障碍性贫血且疾病表型已发生变化的患者

(e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION

- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia receiving treatment where assessment of the disease response at a molecular level may influence treatment strategy (e.g. knowledge of molecular response prior to an allogeneic haematopoietic stem cell transplant). MONITORING DURING / POST-TREATMENT
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Global copy number +/- structural variant via cytogenetics

GT1375 / M82.2 (Karyotype)
GT 635 (SNP array) – it is envisaged this will be used in line with the national SNP array guidelines i.e. particularly used as a salvage test in the event of insufficient metaphases for karyotype analysis. If this test is used in place of karyotyping, clarifications of the limitations of the technique, particularly around the inability to detect balanced translocations, should be clearly stated on the report.

- For patients with a suspected diagnosis of a dysplastic neoplasm or aplastic anaemia where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Targeted copy number +/- structural variant via FISH

M82.4, M82.7-M82.9, M82.19-20, M82.22 (Individual FISH tests)

(例如原始细胞增多、血细胞减少加重)，其中克隆演化的证明可能影响治疗策略。进展

- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、正在接受治疗且分子水平的疾病反应评估可能影响治疗策略的患者(例如在异基因造血干细胞移植前了解分子学反应)。治疗期间/治疗后监测

- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，以监测HSCT后是否存在基因组异常，特别是在供者嵌合率<100%的情况下,以便为供者淋巴细胞输注策略提供依据。移植后监测

通过细胞遗传学检测全局拷贝数 +/- 结构变异

GT1375 / M82.2(核型)
GT 635(SNP array)--预计将按照国家SNP array指南使用，即特别用作核型分析中期分裂相不足时的挽救性检测。若以本检测替代核型分析，应在报告中明确说明该技术的局限性，特别是无法检测平衡易位。

- 用于疑似发育异常性肿瘤或再生障碍性贫血的患者，其中克隆标志物的证明将有助于诊断和 WHO 2022 或 ICC 分类。诊断

- 用于已知诊断为发育异常性肿瘤或再生障碍性贫血的患者，以协助预后判断，从而指导患者管理。诊断

- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、疾病表型已发生变化(例如原始细胞增多、血细胞减少加重)的患者，若证实克隆演化可能影响治疗策略。进展

- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，以监测HSCT后是否存在基因组异常，特别是在供者嵌合率<100%的情况下,以便为供者淋巴细胞输注策略提供依据。移植后监测

通过 FISH 进行靶向拷贝数 +/- 结构变异检测

M82.4、M82.7-M82.9、M82.19-20、M82.22(单项FISH检测)

Replaced by GT1124, GT1248, GT172, GT198, GT595, GT939, GT1302

- For patients with a suspected diagnosis of a dysplastic neoplasm or aplastic anaemia where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a dysplastic neoplasm or aplastic anaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

被 GT1124、GT1248、GT172、GT198、GT595、GT939、GT1302 替代

- 用于疑似发育异常性肿瘤或再生障碍性贫血的患者，其中克隆标志物的证明将有助于诊断和 WHO 2022 或 ICC 分类。诊断
- 用于已知诊断为发育异常性肿瘤或再生障碍性贫血的患者，以协助预后判断，从而指导患者管理。诊断
- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、疾病表型已发生变化(例如原始细胞增多、血细胞减少加重)的患者，若证实克隆演化可能影响治疗策略。进展
- 对于已知诊断为发育异常性肿瘤或再生障碍性贫血、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，以监测HSCT后是否存在基因组异常，特别是在供者嵌合率<100%的情况下,以便为供者淋巴细胞输注策略提供依据。移植后监测

3. Test package: Myeloproliferative Neoplasm TP228

Myeloproliferative Neoplasms Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT552 / M85.13 WGS Germline & Tumour - MPN
GT543 / M85.37 WGS Tumour First - MPN
GT1236 / M85.38 WGS Follow-up Germline - MPN

GT1063 / M86.3 WGS Germline & Tumour - Systemic Mastocytosis
GT12 / M86.4 WGS Tumour First - Systemic Mastocytosis
GT1029 / M86.5 WGS Follow-up Germline - Systemic Mastocytosis

All paediatric and TYA patients with a confirmed or suspected diagnosis of a myeloproliferative neoplasm are eligible for WGS, primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / (REFRACTORY)

Tumour and germline samples can be submitted simultaneously (i.e M85.13 or M86.3) or separately i.e. tumour first (M85.37 or M86.4) followed by a germline when/if available (M85.38 or M86.5). At any presentation point either M85.13 / M86.3 or M85.37 +/- M85.38 or M86.4 +/- M86.5 is permissible, however once a non-contaminated germline has been submitted (either as part of M85.13 / M86.3 or M85.38 / M86.5) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

For adult patients with a confirmed or suspected diagnosis of a myeloproliferative neoplasm, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

3.检测套餐：骨髓增殖性肿瘤TP228

骨髓增殖性肿瘤检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT552 / M85.13 WGS 胚系与肿瘤 - MPN GT543 / M85.37 WGS 肿瘤优先 - MPN
GT1236 / M85.38 WGS随访胚系-MPN

GT1063 / M86.3 WGS 胚系与肿瘤 - 系统性肥大细胞增多症 GT12 / M86.4 WGS 肿瘤先行 - 系统性肥大细胞增多症
GT1029 / M86.5 WGS 随访胚系 - 系统性肥大细胞增多症

所有确诊或疑似诊断为骨髓增殖性肿瘤的儿童和TYA患者均有资格接受WGS，主要在诊断时进行。若诊断时未进行WGS，在一线治疗难治时进行本检测可能是合适的。在特殊情况下，若极有可能已发生显著的克隆演化，可能提示需重复WGS。诊断/(难治)

肿瘤和胚系样本可同时提交(即M85.13或M86.3)或分别提交，即肿瘤先行(M85.37或M86.4)，随后在可获得时提交胚系样本(M85.38或M86.5)。在任何就诊节点，允许M85.13 / M86.3或M85.37 +/- M85.38或M86.4 +/- M86.5，但一旦提交了未受污染的胚系样本(作为M85.13 / M86.3或M85.38 / M86.5的一部分)，该样本可通过生物信息学方式复用于所有后续肿瘤样本的胚系对照。

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

对于确诊或疑似诊断为骨髓增殖性肿瘤的成人患者，WGS可在适应症M235下获得，即标准诊疗检测和/或治疗已用尽，前提是符合该检测资格标准。样本可同时提交(M235.1)或分别提交，即肿瘤先行(M235.2)，随后在可获得时提交胚系样本(M235.3)。

Targeted tests

GT986 / M85.14 (*JAK2* V617F)

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm when either stand-alone *JAK2* V617F testing is appropriate (e.g. isolated polycythaemia) OR the small targeted NGS panel M85.1 (MPN multi-targeted NGS small variant panel) has failed, there is insufficient DNA to attempt this approach, or there is ambiguity in the panel result. DIAGNOSIS
- It is recognised that there is a small population of *JAK2* V617F-mutated patients who are receiving / have received disease modifying treatment where knowledge of the response of the *JAK2* V617F-mutated clone may assist in further management; in these circumstances infrequent monitoring (or repeat testing in response to a change in clinical phenotype) may be appropriate. MONITORING DURING / POST-TREATMENT

GT965 / M85.15 (*JAK2* exon 12)

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm when the small targeted NGS panel M85.1 (MPN multi-targeted NGS small variant panel) has failed, there is insufficient DNA to attempt this approach or there is ambiguity in the panel result. DIAGNOSIS

GT1042 / M85.16 (*CALR* exon 9)

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm when the small targeted NGS panel M85.1 (MPN multi-targeted NGS small variant panel) has failed, there is insufficient DNA to attempt this approach or there is ambiguity in the panel result. DIAGNOSIS

GT326 / M85.17 (*MPL* exon 10)

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm when the small targeted NGS panel M85.1 (MPN multi-targeted NGS small variant panel) has failed, there is insufficient DNA to attempt this approach or there si ambiguity in the panel result. DIAGNOSIS

GT649 / M85.11 (*BCR::ABL1* multiplex) *NB Please note chronic myeloid leukaemia has its own separate clinical indication (M84)*

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm where testing to date has not demonstrated a driver mutation OR the phenotype is such that there is reason to believe there is dual pathology. DIAGNOSIS

靶向检测

GT986 / M85.14（JAK2 V617F）

- 对于可能/疑似诊断为骨髓增殖性肿瘤的患者，当单独*JAK2* V617F检测是合适的(例如孤立性红细胞增多症)或小靶向NGS检测板M85.1(MPN多靶标NGS小变异检测板)失败、DNA不足无法尝试该方法，或检测板结果存在歧义时。诊断
- 已知存在一小部分*JAK2* V617F突变患者正在接受/已接受疾病修饰治疗，了解*JAK2* V617F突变克隆的反应可能有助于进一步管理；在这些情况下，不频繁的监测(或针对临床表型变化进行重复检测)可能是合适的。治疗期间/治疗后监测

GT965 / M85.15(*JAK2*外显子 12)

- 对于可能/疑似诊断为骨髓增殖性肿瘤的患者，当小靶向NGS检测板M85.1(MPN多靶标NGS小变异检测板)失败、DNA不足无法尝试该方法或检测板结果存在歧义时。诊断

GT1042 / M85.16(*CALR*外显子 9)

- 对于可能/疑似诊断为骨髓增殖性肿瘤的患者，当小靶向NGS检测板M85.1(MPN多靶标NGS小变异检测板)失败、DNA不足无法尝试该方法或检测板结果存在歧义时。诊断

GT326 / M85.17(*MPL*外显子 10)

- 对于可能/疑似诊断为骨髓增殖性肿瘤的患者，当小靶向NGS检测板M85.1(MPN多靶标NGS小变异检测板)失败、DNA不足无法尝试该方法或检测板结果存在歧义时。诊断

GT649 / M85.11(*BCR::ABL1*多重)*NB*请注意慢性髓性白血病有其单独的临床适应症*(M84)*

- 对于可能/疑似诊断为骨髓增殖性肿瘤、迄今检测未显示驱动突变，或表型提示有理由认为存在双重病理的患者。诊断

GT246 / M86.2 (high sensitivity *KIT* D816)

- For patients with a possible / suspected diagnosis of systemic mastocytosis. DIAGNOSIS

Small variant / copy number detection via panel

GT717 / M85.1 (MPN multi-target NGS limited panel small variant)

- For patients with a possible / suspected diagnosis of a myeloproliferative neoplasm. DIAGNOSIS

GT819 / M85.2 (Multi-target NGS panel)
GT736 / M85.36 (Multi-target NGS panel CNV)

- For patients with a known / suspected diagnosis of a myeloproliferative neoplasm where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm where demonstration of a specific genomic abnormality would allow access to a targeted treatment (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT
- For patients with a known diagnosis of a myeloproliferative neoplasm who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a myeloproliferative neoplasm receiving treatment where assessment of the disease response at a molecular level may influence treatment strategy (e.g. knowledge of molecular response prior to an allogeneic haematopoietic stem cell transplant). MONITORING DURING / POST-TREATMENT
- For patients with a known diagnosis of a myeloproliferative neoplasm who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Global copy number +/- structural variant via cytogenetics

GT1233 / M85.3 (Karyotype)
GT100 (SNP array)

GT246 / M86.2(高灵敏度*KIT* D816)

- 适用于可能/疑似诊断为系统性肥大细胞增多症的患者。诊断

通过 panel 检测小变异/拷贝数

GT717 / M85.1(MPN多靶点NGS有限panel小变异)

- 适用于可能/疑似诊断为骨髓增殖性肿瘤的患者。诊断

GT819 / M85.2(多靶点NGS panel)
GT736 / M85.36(多靶点NGS panel CNV)

- 适用于已知/疑似诊断为骨髓增殖性肿瘤、且证实克隆性标志物有助于诊断及 WHO 2022 或 ICC 分类的患者。诊断
- 适用于已知诊断为骨髓增殖性肿瘤、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为骨髓增殖性肿瘤、且证实特定基因组异常可获得靶向治疗机会的患者(注：应预期在证实(或不证实)特定异常后，相应的基因组指导治疗是可及的)。诊断、治疗前、治疗变更前
- 适用于已知诊断为骨髓增殖性肿瘤、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展
- 适用于已知诊断为骨髓增殖性肿瘤、正在接受治疗、且分子水平疾病反应评估可能影响治疗策略的患者(如异基因造血干细胞移植前了解分子学反应)。治疗期间/治疗后监测
- 适用于已知诊断为骨髓增殖性肿瘤、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过细胞遗传学检测全局拷贝数 +/- 结构变异 GT1233 / M85.3(核型)GT100(SNP 芯片)

- For patients with a suspected diagnosis of a myeloproliferative neoplasm where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a myeloproliferative neoplasm who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Targeted copy number +/- structural variant via FISH / RT-PCR

GT1211 / M85.5 (Other FISH)
GT763 / M85.24 (*BCR::ABL1* FISH)
GT152 / M85.31 (*ABL1* FISH)
GT699 / M85.7 (*FLIP1L1-PDGFRA* FISH)
GT373 / M85.25 (*PDGFRA* FISH)
GT1282 / M85.9 (*ETV6::PDGFRB* FISH)
GT1355 / M85.26 (*PDGFRB* FISH)
GT 147 / M85.27 (*FGFR1* FISH)
GT818 / M85.10 (*PCM1::JAK2* FISH)
GT13 / M85.28 (*JAK2* FISH)
GT172 / M85.29 (*MECOM* FISH)
GT1248 / M85.19 (Chr8 FISH)
GT198 / M85.20 (Chr7 FISH)
GT1224 / M85.21 (Chr5 FISH)
GT355 / M85.22 (Chr17 FISH)
GT614 / M85.12 (RT-PCR Other)

- For patients with a suspected diagnosis of a myeloproliferative neoplasm where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm to assist with prognostication which will inform patient management. DIAGNOSIS

- 适用于疑似诊断为骨髓增殖性肿瘤、且证实克隆性标志物有助于诊断及 WHO 2022 或 ICC 分类的患者。诊断
- 适用于已知诊断为骨髓增殖性肿瘤、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为骨髓增殖性肿瘤、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展
- 适用于已知诊断为骨髓增殖性肿瘤、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

GT1211 / M85.5(其他 FISH)
GT763 / M85.24（BCR::ABL1 荧光原位杂交）GT152 / M85.31（ABL1 荧光原位杂交）
GT699 / M85.7 (FLIP1L1-PDGFRA FISH) GT373 / M85.25 (PDGFRA FISH) GT1282 / M85.9 (ETV6::PDGFRB FISH) GT1355 / M85.26 (PDGFRB FISH) GT 147 / M85.27 (FGFR1 FISH) GT818 / M85.10 (PCM1::JAK2 FISH) GT13 / M85.28 (JAK2 FISH) GT172 / M85.29 (MECOM FISH) GT1248 / M85.19 (8号染色体 FISH) GT198 / M85.20 (7号染色体 FISH) GT1224 / M85.21 (5号染色体 FISH) GT355 / M85.22 (17号染色体 FISH) GT614 / M85.12 (RT-PCR 其他)

- 适用于疑似诊断为骨髓增殖性肿瘤、且证实克隆性标志物有助于诊断及 WHO 2022 或 ICC 分类的患者。诊断
- 适用于已知诊断为骨髓增殖性肿瘤、且有助于预后判断从而指导患者管理的患者。诊断

- For patients with a known diagnosis of a myeloproliferative neoplasm who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a myeloproliferative neoplasm who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Structural variant detection via NGS panel

GT819 / M85.35 (Multi-target NGS panel – structural variant)

- For patients with a suspected diagnosis of a myeloproliferative neoplasm where demonstration of a clonal marker will assist in the diagnosis and WHO 2022 or ICC classification. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of a myeloproliferative neoplasm who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a myeloproliferative neoplasm who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Measurable residual disease testing

GT662 / M85.8 (MRD *FIP1L1::PDGFRA*)

- For patients with a known *FIP1L1::PDGFRA* fusion who are receiving disease modifying treatment where there is an expectation of a reduction in the clone size containing the *FIP1L1::PDGFRA* fusion and knowledge of this change will assist in decisions around ongoing / further management. MONITORING DURING TREATMENT

GT1497/M85.39 (MRD *JAK2 V617F* High sensitivity at least 0.1%)

- For patients following therapy where confirmation of complete molecular response is desirable. MONITORING POST TREATMENT

- 适用于已知诊断为骨髓增殖性肿瘤、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展

- 适用于已知诊断为骨髓增殖性肿瘤、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过NGS panel进行结构变异检测

GT819 / M85.35(多靶点NGS panel--结构变异)

- 适用于疑似诊断为骨髓增殖性肿瘤、且证实克隆性标志物有助于诊断及 WHO 2022 或 ICC 分类的患者。诊断

- 适用于已知诊断为骨髓增殖性肿瘤、且有助于预后判断从而指导患者管理的患者。诊断

- 适用于已知诊断为骨髓增殖性肿瘤、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展

- 适用于已知诊断为骨髓增殖性肿瘤、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

可测量残留病检测

GT662 / M85.8(MRD FIP1L1::PDGFRA)

- 适用于已知存在*FIP1L1::PDGFRA*融合、正在接受疾病修饰治疗、预期含*FIP1L1::PDGFRA*融合的克隆大小将会缩小、且了解这一变化有助于决定持续/进一步管理的患者。治疗期间监测

GT1497/M85.39(MRDJAK2V617F高灵敏度至少 0.1%)

- 适用于治疗后希望确认完全分子学反应的患者。治疗后监测

4. Test package: Chronic Myeloid Leukaemia TP374

Chronic Myeloid Leukaemia Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT538 / M84.11 WGS Germline & Tumour - CML
GT1373 / M84.24 WGS Tumour First - CML
GT1357 / M84.25 WGS Follow-up Germline - CML

All paediatric and TYA patients with a confirmed or suspected diagnosis of a chronic myeloid leukaemia are eligible for whole genome sequencing (WGS), primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / (RELAPSE/REFRACTORY)

Tumour and germline samples can be submitted simultaneously (i.e M85.11) or separately i.e. tumour first (M84.24) followed by a germline when/if available (M84.25). At any presentation point either M84.11 or M84.24 +/- M84.25 is permissible, however once a non-contaminated germline has been submitted (either as part of M84.11 or M84.25) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

For adult patients with a confirmed or suspected diagnosis of chronic myeloid leukaemia, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Targeted tests

GT649 / M84.1 (*BCR::ABL1* multiplex)

4.检测套餐：慢性髓系白血病TP374

慢性髓性白血病检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT538 / M84.11 WGS 胚系与肿瘤 - CML GT1373 /
M84.24 WGS 肿瘤优先 - CML
GT1357 / M84.25 WGS 随访胚系 - CML

所有确诊或疑似诊断为慢性髓性白血病的儿童及 TYA 患者均有资格接受全基因组测序(WGS)，主要在诊断时进行。若诊断时未进行 WGS，在复发或对某一线治疗难治时进行该检测可能是合适的。在特殊情况下，若极有可能已发生显著的克隆演化，则可能有必要重复 WGS。诊断/(复发/难治)

肿瘤和胚系样本可同时提交(即 M85.11)或分别提交，即先提交肿瘤样本(M84.24)，随后在可获得时/若可获得再提交胚系样本(M84.25)。在任何就诊节点，M84.11 或 M84.24 +/- M84.25 均允许，但一旦提交了未受污染的胚系样本(作为 M84.11 或 M84.25 的一部分)，该样本可通过生物信息学方式复用于所有后续肿瘤样本的胚系对照。

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

对于确诊或疑似诊断为慢性髓性白血病的成人患者，WGS 可在适应症 M235 下获得，即已穷尽标准诊疗检测和/或治疗，前提是符合该检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时/若可获得再提交胚系样本(M235.3)。

靶向检测

GT649 / M84.1(*BCR::ABL1*多重)

- For patients with a possible / suspected diagnosis of chronic myeloid leukaemia. DIAGNOSIS

- 适用于可能/疑似诊断为慢性髓性白血病的患者。诊断

Small variant / copy number detection via panel

GT1426 Multi-target NGS panel
GT1414 Multi-target NGS panel CNV

- For patients with a known diagnosis of a chronic myeloid leukaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of chronic myeloid leukaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION

通过 panel 检测小变异/拷贝数

GT1426 多靶点 NGS 检测组合 GT1414 多靶点 NGS 检测组合 CNV

- 适用于已知诊断为慢性髓性白血病、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展

Global copy number +/- structural variant via cytogenetics

GT1096 / M84.4 Karyotype
GT1265 SNP array

- For patients with a suspected diagnosis of chronic myeloid leukaemia where demonstration of *BCR::ABL1* will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of a chronic myeloid leukaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of chronic myeloid leukaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a chronic myeloid leukaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

通过细胞遗传学检测全局拷贝数 +/- 结构变异 GT1096 / M84.4 核型
GT1265 SNP 芯片

- 适用于疑似诊断为慢性髓性白血病、且证实*BCR::ABL1*有助于诊断的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展
- 适用于已知诊断为慢性髓性白血病、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

Targeted copy number +/- structural variant via FISH / RT-PCR

GT1193 / M84.6 (FISH other)
GT1371 / M84.3 (*BCR::ABL1* FISH)
GT1034 / M84.12 (Chr8 FISH)
GT71 / M84.13 (Chr19 FISH)
GT73 / M84.14 (Chr7 FISH)
GT1424 / M84.15 (Chr5 FISH)
GT1345 / M84.16 (Chr17 FISH)

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

GT1193 / M84.6(FISH 其他)
GT1371 / M84.3(*BCR::ABL1* FISH)GT1034 / M84.12(8 号染色体 FISH)GT71 / M84.13(19 号染色体 FISH)GT73 / M84.14(7 号染色体 FISH)GT1424 / M84.15(5 号染色体 FISH)GT1345 / M84.16(17 号染色体 FISH)

GT690 / M84.19 (*MECOM* FISH)
GT478 / M84.20 (*KMT2A* FISH)

- For patients with a suspected diagnosis of chronic myeloid leukaemia where demonstration of *BCR::ABL1* will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of a chronic myeloid leukaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of chronic myeloid leukaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a chronic myeloid leukaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Structural variant detection via NGS panel

GT99 / M84.22 (Multi-target NGS structural variant panel)

- For patients with a suspected diagnosis of chronic myeloid leukaemia where demonstration of *BCR::ABL1* will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of a chronic myeloid leukaemia to assist with prognostication which will inform patient management. DIAGNOSIS
- For patients with a known diagnosis of chronic myeloid leukaemia who have undergone a change in their disease phenotype (e.g. increased blasts, worsening cytopenias), where demonstration of clonal evolution may influence the treatment strategy. PROGRESSION
- For patients with a known diagnosis of a chronic myeloid leukaemia who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Drug Resistance Mutation testing

GT303 / M84.8 (*BCR::ABL1* TKD NGS)

- For patients with a known diagnosis of chronic myeloid leukaemia receiving a tyrosine kinase inhibitor who have failed to achieve an optimal response,

GT690 / M84.19(MECOM FISH)GT478 / M84.20(KMT2A FISH)

- 适用于疑似诊断为慢性髓性白血病、且证实*BCR::ABL1*有助于诊断的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展
- 适用于已知诊断为慢性髓性白血病、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过NGS panel进行结构变异检测

GT99 / M84.22(多靶点NGS结构变异检测组合)

- 适用于疑似诊断为慢性髓性白血病、且证实*BCR::ABL1*有助于诊断的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且有助于预后判断从而指导患者管理的患者。诊断
- 适用于已知诊断为慢性髓性白血病、且疾病表型已发生变化(如原始细胞增多、血细胞减少加重)、证实克隆演化可能影响治疗策略的患者。进展
- 适用于已知诊断为慢性髓性白血病、已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测 HSCT 后基因组异常的存在，特别是在供者嵌合率<100% 的情况下,以指导供者淋巴细胞输注策略。移植后监测

耐药突变检测

GT303 / M84.8 (BCR::ABL1 TKD NGS)

- 适用于已知诊断为慢性髓性白血病、正在接受酪氨酸激酶抑制剂治疗、未能达到最佳反应、

or have lost a previous response to treatment. The level of detection of this assay needs to be at least 5% (i.e. will detected variants within the *BCR::ABL1* TKD transcript with an allele frequency of at least 5%). PROGRESSION / RELAPSE / REFRACTORY

- In the event a variant is detected in *the BCR::ABL1* TKD, monitoring for the persistence of this variant is permissible. MONITORING

Measurable Residual Disease testing

GT31 / M84.2 (*BCR::ABL1* RT-qPCR p190 / p210)
GT673 / M84.23 (*BCR::ABL1* MRD rare)

- For patients with a known diagnosis of chronic myeloid leukaemia to monitor response to tyrosine kinase inhibitors. Frequency of monitoring should be as per ELN guidelines. DIAGNOSIS / PROGRESSION / RELAPSE / REFRACTORY / MONITORING

或已丧失既往治疗反应的患者。该检测的检测水平需至少为 5%(即检测*BCR::ABL1* TKD 转录本中等位基因频率至少为 5% 的变异)。进展/复发/难治

- 若在*BCR::ABL1* TKD 中检测到变异，允许对该变异的持续存在进行监测。监测

可测量残留病灶检测

GT31 / M84.2（BCR::ABL1 RT-qPCR p190 / p210）
GT673 / M84.23（BCR::ABL1 MRD 罕见）

- 对于已知诊断为慢性髓性白血病的患者，监测其对酪氨酸激酶抑制剂的反应。监测频率应按照 ELN 指南。诊断 / 进展 / 复发 / 难治 / 监测

5. Test package: Acute Lymphoblastic Leukaemia - B cell TP241

Acute Lymphoblastic Leukaemia – B cell Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT467 / M91.1 WGS Germline & Tumour - B-ALL
GT781 / M91.82 WGS Tumour First - B-ALL
GT1050 / M91.83 WGS Follow-up Germline - B-ALL

GT670 / M89.1 WGS Germline & Tumour – Acute Leukaemia Other
GT254 / M89.108 WGS Tumour First - Acute Leukaemia Other
GT825 / M89.109 WGS Follow-up Germline - Acute Leukaemia Other

All patients (of any age i.e. paediatric, TYA and adult) with an immunophenotypic diagnosis of acute lymphoblastic leukaemia (ALL) are eligible for WGS primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / (RELAPSE/REFRACTORY)

Tumour and germline samples can be submitted simultaneously (i.e. M91.1) or separately i.e. tumour first (M91.82) followed by a germline when/if available (M91.83). At any presentation point either M91.1 or M91.82 +/- M91.83 is permissible, however once a non-contaminated germline has been submitted (either as part of M91.1 or M91.83) this can be bioinformatically re-used as a germline for all subsequent tumour samples. Currently WGS is performed as an addition to any eligible to any standard of care.

Targeted tests

GT649 / M91.8 (BCR::ABL1 multiplex)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria which will have

5.检测套餐：急性淋巴细胞白血病-B细胞TP241

急性淋巴细胞白血病 - B细胞检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT467 / M91.1 WGS 胚系与肿瘤 - B-ALL GT781 / M91.82 WGS 肿瘤先行 - B-ALL
GT1050 / M91.83 WGS随访生殖系 - B-ALL

GT670 / M89.1 WGS 胚系与肿瘤 - 其他急性白血病 GT254 / M89.108 WGS 肿瘤先行 - 其他急性白血病
GT825 / M89.109 WGS随访胚系 - 其他急性白血病

所有(任何年龄，即儿童、TYA 和成人)经免疫表型诊断为急性淋巴细胞白血病(ALL)的患者均有资格主要在诊断时接受 WGS。如果诊断时未进行 WGS，在复发或对某一线治疗难治时进行该检测可能是合适的。在特殊情况下，如果极有可能已发生显著的克隆演化，则可能指示重复 WGS。诊断 /(复发/难治)

肿瘤和胚系样本可同时提交(即 M91.1)，也可分别提交，即先提交肿瘤样本(M91.82)，随后在可获得时提交胚系样本(M91.83)。在任何就诊时间点，允许提交 M91.1 或 M91.82 +/- M91.83，然而一旦提交了未受污染的胚系样本(作为 M91.1 或 M91.83 的一部分)，该样本可通过生物信息学方式重新用作所有后续肿瘤样本的胚系对照。目前 WGS 作为任何符合条件标准治疗的附加检测进行。

靶向检测

GT649 / M91.8(BCR::ABL1 多重检测)

• 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断，这将具有

prognostic implications and also direct treatment with a tyrosine kinase inhibitor. DIAGNOSIS

Small variant / copy number detection via panel

GT849 / M91.15 (NGS small variant panel – it is envisaged that the choice of gene footprint will ensure all relevant variants including putative resistance mutations are included).
GT350 / M91.78 (NGS Copy Number panel)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with a morphological diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

Global copy number +/- structural variant via cytogenetics

GT1096 / M91.2 (Karyotype) Other diagnostic modalities such as a combination of SNP array and FISH testing can replace karyotyping providing required TATs are met.
GT295 (SNP array) SNP array is seen as a frontline test for ALL and can be performed in preference to karyotyping providing the required TATs are met. The SNP array should be at such a resolution such that it can detect small disruptive deletions (i.e. partial exon) in those genes deemed prognostic in the ALLTogether1 trial copy number abnormality (CNA) profile.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

预后意义，并指导使用酪氨酸激酶抑制剂治疗。诊断

通过 panel 检测小变异/拷贝数

GT849 / M91.15(NGS 小变异检测板 - 预期基因覆盖范围的选择将确保纳入所有相关变异，包括推定的耐药突变)。

GT350 / M91.78(NGS拷贝数panel)

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治
- 对于诊断时/复发/难治时经形态学诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

通过细胞遗传学检测全局拷贝数 +/- 结构变异

GT1096 / M91.2(核型)其他诊断方法如 SNP array 和 FISH 检测的组合可替代核型分析，前提是满足所需的 TAT。GT295(SNP array)SNP array 被视为 ALL 的一线检测，在满足所需 TAT 的前提下可优先于核型分析进行。SNP array 的分辨率应足以检测 ALLTogether1 试验拷贝数异常(CNA)谱中具有预后意义的基因中的小破坏性缺失(即部分外显子)。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治
- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

Targeted copy number +/- structural variant via FISH / RT-PCR

M91.4, M91.10, M91.24, M91.26, M91.44, M91.53-M91.62, M91.84-89 (Individual FISH tests)
Replaced by GT1052, GT1089, GT1163 / GT1180 / GT1191 / GT1300 / GT1308 / GT1309 / GT160 / GT200 / GT296 / GT301 / GT315 / GT483 / GT512 / GT546 / GT548 / GT56 / GT7 / GT723 / GT729 / GT756 / GT844 / GT85 / GT874
M91.6, M91.63 (Individual RT-PCR tests) non-quantitative
GT577 / GT739
It is expected that FISH / RT-PCR testing will not duplicate other structural / copy number variant testing unless required to meet rapid TATs.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL receiving/received treatment where assessment of disease response at a cytogenetic level may influence the subsequent treatment strategy. MONITORING DURING / POST-TREATMENT

Structural variant detection via NGS panel

GT329 / M91.7 (NGS Structural variant panel)

It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as SNP array, FISH, and (karyotype) if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

M91.4、M91.10、M91.24、M91.26、M91.44、M91.53-M91.62、M91.84-89(单项 FISH 检测)

已由 GT1052、GT1089、GT1163 / GT1180 / GT1191 / GT1300 / GT1308 / GT1309 / GT160 / GT200 / GT296 / GT301 / GT315 / GT483 / GT512 / GT546 / GT548 / GT56 / GT7 / GT723 / GT729 / GT756 / GT844 / GT85 / GT874 替代

M91.6, M91.63(单项RT-PCR检测)非定量 GT577 / GT739

预期 FISH / RT-PCR 检测不会与其他结构变异/拷贝数变异检测重复，除非需要满足快速 TAT。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义。诊断 / 复发/难治
- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治
- 对于正在接受/已接受治疗的经免疫表型诊断为 ALL 的患者，在细胞遗传学水平评估疾病反应可能影响后续治疗策略。治疗期间 / 治疗后监测

通过NGS panel进行结构变异检测

GT329 / M91.7(NGS 结构变异检测组合)

预计 NGS 结构变异 panel 最通常在其他诊断如 SNP array、FISH 和(核型)之后使用，前提是未发现可能的驱动基因组改变。已知不同实验室的检测策略可能略有差异，这意味着该检测在路径中可能被更频繁或更早地使用。应认识到靶向 RNA panel 的局限性，即原癌基因的失调不太可能通过该方法检测到。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断

- For patients with immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL at diagnosis at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

Drug Resistance Mutation testing

GT303 / M91.11 (BCR::ABL1 TKD variant testing)

- For patients with an immunophenotypic diagnosis of ALL known to have a BCR::ABL1 rearrangement whose response to a targeted inhibitor is such that there is clinical suspicion that there may be a resistance variant within the ABL1 tyrosine kinase domain. NB this assay should have a minimum variant allele frequency level of detection of 5%. MONITORING DURING TREATMENT

Measurable residual disease testing

GT31 / M91.9 (BCR::ABL1)
GT673 / M91.79 (Rare transcript BCR::ABL1 MRD)
GT214 / M91.14 (other MRD)
GT738 / M91.22 (Ig target identification for MRD)
GT1247 / M91.23 (Ig target quantification for MRD)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to identify Ig rearrangements suitable for use as measurable residual disease (MRD) targets (Ig target identification for MRD). DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL known to have the specific described genomic aberration of the assay, receiving/received curative treatment where assessment of disease response at a measurable residual disease [MRD] level may influence the subsequent treatment strategy. (Ig target quantification for MRD). MONITORING DURING / POST-TREATMENT

Pharmacogenomic targets

GT1185 / M91.80 (TPMT)
GT694 / M91.81 (NUDT15)

- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治
- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

耐药突变检测

GT303 / M91.11(BCR::ABL1 TKD变异检测)

- 对于已知具有 BCR::ABL1 重排的经免疫表型诊断为 ALL 的患者，其对靶向抑制剂的反应使得临床怀疑 ABL1 酪氨酸激酶结构域内可能存在耐药变异。注意本检测的最低变异等位基因频率检测限应为 5%。治疗期间监测

可测量残留病检测

GT31 / M91.9（BCR::ABL1）
GT673 / M91.79(罕见转录本 BCR::ABL1 MRD)GT214 / M91.14(其他 MRD)GT738 / M91.22(用于 MRD 的 Ig 靶标鉴定)GT1247 / M91.23(用于 MRD 的 Ig 靶标定量)

- 对于诊断时经免疫表型诊断为 ALL 的患者，以鉴定适合用作可测量残留病灶(MRD)靶标的 Ig 重排(用于 MRD 的 Ig 靶标鉴定)。诊断
- 对于已知具有该检测所描述的特定基因组异常的经免疫表型诊断为 ALL 的患者，正在接受/已接受治愈性治疗，在可测量残留病灶[MRD]水平评估疾病反应可能影响后续治疗策略。(用于 MRD 的 Ig 靶标定量)。治疗期间 / 治疗后监测

药物基因组学靶点

GT1185 / M91.80（TPMT）
GT694 / M91.81（NUDT15）

- For patients with an immunophenotypic diagnosis of ALL who are planned to receive a purine analogue as part of their treatment. DIAGNOSIS

6. Test package: Acute Lymphoblastic Leukaemia - T cell TP34

Acute Lymphoblastic Leukaemia - T cell Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT183 / M91.1 WGS Germline & Tumour - T-ALL
GT687 / M91.82 WGS Tumour First - T-ALL
GT225 / M91.83 WGS Follow-up Germline - T-ALL

All patients (of any age i.e. paediatric, TYA and adult) with an immunophenotypic diagnosis of acute lymphoblastic leukaemia (ALL) are eligible for WGS at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS / RELASPE/REFRACTORY

Tumour and germline samples can be submitted simultaneously (i.e. M91.1) or separately i.e. tumour first (M91.82) followed by a germline when/if available (M91.83). At any presentation point either M91.1 or M91.82 +/- M91.83 is permissible, however once a non-contaminated germline has been submitted (either as part of M91.1 or M91.83) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an addition to any eligible to any standard of care.

Targeted tests

GT649 / M91.8 (BCR::ABL1 multiplex)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria which will have

• 对于经免疫表型诊断为 ALL 且计划接受嘌呤类似物作为其治疗一部分的患者。诊断

6. 检测套餐：急性淋巴细胞白血病-T细胞TP34

急性淋巴细胞白血病 - T 细胞检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT183 / M91.1 WGS 胚系与肿瘤 - T-ALL GT687 / M91.82 WGS 肿瘤先行 - T-ALL GT225 / M91.83 WGS 随访胚系 - T-ALL

所有(任何年龄，即儿童、TYA 和成人)经免疫表型诊断为急性淋巴细胞白血病(ALL)的患者均有资格在诊断时接受 WGS。如果诊断时未进行 WGS，在复发或对某一线治疗难治时进行该检测可能是合适的。在特殊情况下，如果极有可能已发生显著的克隆演化，则可能指示重复 WGS。诊断 / 复发/难治

肿瘤和胚系样本可同时提交(即 M91.1)或分别提交，即肿瘤先行(M91.82)，随后在可获得时/如果可获得则提交胚系(M91.83)。在任何就诊时间点，M91.1 或 M91.82 +/- M91.83 均可，但一旦提交了未受污染的胚系(作为 M91.1 或 M91.83 的一部分)，该样本可通过生物信息学方式重新用作所有后续肿瘤样本的胚系。

目前 WGS 作为任何符合条件标准治疗的补充进行。

靶向检测

GT649 / M91.8(BCR::ABL1 多重检测)

• 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断，这将具有

prognostic implications and also direct treatment with a tyrosine kinase inhibitor. DIAGNOSIS

Small variant / copy number detection via panel

GT849 / M91.15 (NGS small variant panel – it is envisaged that the choice of gene footprint will ensure all relevant variants including putative resistance mutations are included).
GT350 / M91.78 (NGS Copy Number panel)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with a morphological diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

Global copy number +/- structural variant via cytogenetics

GT1096 / M91.2 (Karyotype) Other diagnostic modalities such as a combination of SNP array and FISH testing can replace karyotyping providing required TATs are met.
GT527 (SNP array) SNP array is seen as a frontline test for ALL and can be performed in preference to karyotyping providing the required TATs are met. The SNP array should be at such a resolution such that it can detect small disruptive deletions (i.e. partial exon) in those genes deemed prognostic in the ALLTogether1 trial copy number abnormality (CNA) profile.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

Targeted copy number +/- structural variant via FISH / RT-PCR

预后意义，并指导使用酪氨酸激酶抑制剂治疗。诊断

通过 panel 检测小变异/拷贝数

GT849 / M91.15(NGS 小变异检测板 - 预期基因覆盖范围的选择将确保纳入所有相关变异，包括推定的耐药突变)。

GT350 / M91.78(NGS拷贝数panel)

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治
- 对于诊断时/复发/难治时经形态学诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

通过细胞遗传学检测全局拷贝数 +/- 结构变异

GT1096 / M91.2(核型)其他诊断方法如 SNP array 和 FISH 检测的组合可替代核型分析，前提是满足所需的 TAT。GT527(SNP array)SNP array 被视为 ALL 的一线检测，在满足所需 TAT 的前提下可优先于核型分析进行。SNP array 的分辨率应足以检测 ALLTogether1 试验拷贝数异常(CNA)谱中具有预后意义的基因中的小破坏性缺失(即部分外显子)。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治
- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

M91.4, M91.10, M91.25, M91.39-M91.59, M91.61-62, M91.84, M91.88-M91.89 (Individual FISH tests)
Replaced by GT10, GT1089, GT1163, GT1180, GT1191, GT1300, GT1308, GT1309, GT200, GT239, GT296, GT301, GT512, GT844, GT85
M91.6 (Individual RT-PCR tests) non-quantitative
Replaced by GT739

It is expected that FISH / RT-PCR testing will not duplicate other structural / copy number variant testing unless required to meet rapid TATs.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications. DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY
- For patients with an immunophenotypic diagnosis of ALL receiving/received treatment where assessment of disease response at a cytogenetic level may influence the subsequent treatment strategy. MONITORING DURING / POST-TREATMENT

Structural variant detection via NGS panel

GT329 / M91.7 (NGS Structural variant panel)

It is envisaged that NGS structural variant panels will most typically be used following other diagnostics such as SNP array, FISH, and (karyotype) if no likely driver genomic alteration has been identified. It is acknowledged that different laboratories may have slightly varying testing strategies which means this assay is deployed more frequently or earlier in the pathway. The limitations of a targeted RNA panel should be recognised such that deregulation of a proto-oncogene is unlikely to be detected by this methodology.

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to better refine the diagnosis as per WHO 2022 or ICC criteria. DIAGNOSIS
- For patients with immunophenotypic diagnosis of ALL at diagnosis / relapse/refractory presentation for prognostic implications including risk stratification DIAGNOSIS / RELASPE/REFRACTORY

M91.4、M91.10、M91.25、M91.39-M91.59、M91.61-62、M91.84、M91.88-M91.89(单项 FISH 检测)

由 GT10、GT1089、GT1163、GT1180、GT1191、GT1300、GT1308、GT1309、GT200、GT239、GT296、GT301、GT512、GT844、GT85 M91.6(单项 RT-PCR 检测)非定量替代由 GT739 替代

预期 FISH / RT-PCR 检测不会与其他结构变异/拷贝数变异检测重复，除非需要满足快速 TAT。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义。诊断 / 复发/难治
- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治
- 对于正在接受/已接受治疗的经免疫表型诊断为 ALL 的患者，在细胞遗传学水平评估疾病反应可能影响后续治疗策略。治疗期间 / 治疗后监测

通过NGS panel进行结构变异检测

GT329 / M91.7(NGS 结构变异检测组合)

预计 NGS 结构变异 panel 最通常在其他诊断如 SNP array、FISH 和(核型)之后使用，前提是未发现可能的驱动基因组改变。已知不同实验室的检测策略可能略有差异，这意味着该检测在路径中可能被更频繁或更早地使用。应认识到靶向 RNA panel 的局限性，即原癌基因的失调不太可能通过该方法检测到。

- 对于诊断时经免疫表型诊断为 ALL 的患者，以按照 WHO 2022 或 ICC 标准更好地细化诊断。诊断
- 对于诊断时/复发/难治表现时经免疫表型诊断为 ALL 的患者，用于预后意义包括风险分层。诊断 / 复发/难治

- For patients with an immunophenotypic diagnosis of ALL at diagnosis at diagnosis / relapse/refractory to inform treatment (may include conventional chemotherapy regimens, non-chemotherapy regimens, or targeted inhibitors). DIAGNOSIS / RELASPE/REFRACTORY

Drug Resistance Mutation testing

GT303 / M91.11 (BCR::ABL1 TKD variant testing)

- For patients with an immunophenotypic diagnosis of ALL known to have a BCR::ABL1 rearrangement whose response to a targeted inhibitor is such that there is clinical suspicion that there may be a resistance variant within the ABL1 tyrosine kinase domain. NB this assay should have a minimum variant allele frequency level of detection of 5%. MONITORING DURING TREATMENT

Measurable residual disease testing

GT31 / M91.9 (BCR::ABL1)
GT673 / M91.79 (Rare transcript BCR::ABL1 MRD)
GT985 / M91.14 (other MRD)
GT332 / M91.22 (TCR target identification for MRD)
GT1149 / M91.23 (TCR target quantification for MRD)

- For patients with an immunophenotypic diagnosis of ALL at diagnosis to identify TCR rearrangements suitable for use as measurable residual disease (MRD) targets (TCR target identification for MRD). DIAGNOSIS
- For patients with an immunophenotypic diagnosis of ALL known to have the specific described genomic aberration of the assay, receiving/received curative treatment where assessment of disease response at a measurable residual disease [MRD] level may influence the subsequent treatment strategy. (TCR target quantification for MRD). MONITORING DURING / POST-TREATMENT

Pharmacogenomic targets

GT1185 / M91.80 (TPMT)
GT694 / M91.81 (NUDT15)

- For patients with an immunophenotypic diagnosis of ALL who are planned to receive a purine analogue as part of their treatment. DIAGNOSIS

- 对于诊断时/复发/难治时经免疫表型诊断为 ALL 的患者，以指导治疗(可能包括常规化疗方案、非化疗方案或靶向抑制剂)。诊断 / 复发/难治

耐药突变检测

GT303 / M91.11(BCR::ABL1 TKD变异检测)

- 对于已知具有 BCR::ABL1 重排的经免疫表型诊断为 ALL 的患者，其对靶向抑制剂的反应使得临床怀疑 ABL1 酪氨酸激酶结构域内可能存在耐药变异。注意本检测的最低变异等位基因频率检测限应为 5%。治疗期间监测

可测量残留病检测

GT31 / M91.9 (BCR::ABL1)
GT673 / M91.79(罕见转录本 BCR::ABL1 MRD) GT985 / M91.14(其他 MRD) GT332 / M91.22(用于 MRD 的 TCR 靶标鉴定) GT1149 / M91.23(用于 MRD 的 TCR 靶标定量)

- 用于诊断时免疫表型诊断为 ALL 的患者，以鉴定适合用作可测量残留病灶(MRD)靶标的 TCR 重排(用于 MRD 的 TCR 靶标鉴定)。诊断

- 用于免疫表型诊断为 ALL 且已知具有该检测所描述的特定基因组异常的患者，正在接受/已接受治愈性治疗，且在可测量残留病灶[MRD]水平评估疾病缓解可能影响后续治疗策略。(用于 MRD 的 TCR 靶标定量)。治疗期间/治疗后监测

药物基因组学靶点

GT1185 / M91.80 (TPMT)
GT694 / M91.81 (NUDT15)

- 对于经免疫表型诊断为 ALL 且计划接受嘌呤类似物作为其治疗一部分的患者。诊断

7. Test package: Chronic Lymphocytic Leukaemia TP533

Chronic Lymphocytic Leukaemia Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

For adult patients with a confirmed or suspected diagnosis of a chronic lymphocytic leukaemia, WGS is available under
the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can
be submitted simultaneously (M235.1) or separately i.e. tumour
first (M235.2) followed by a germline when/if available (M235.3).

Small variant / copy number detection via panel

GT1209 / M94.1 (Multi-target NGS panel small variant) [*TP53, BTK, PLCG2, BCL2*]
GT929 / M94.2 (Multi-target NGS panel copy number variant) [*TP53, ATM, DLEU2/7, RB1*, trisomy 12]

TP53 small variants and copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia who have not previously been identified to have disruption of the *TP53* gene who are about to embark on treatment. This testing should be repeated before each line of treatment providing no previous disruption of this gene has been identified. PRE-TREATMENT
BTK/PLCG2 small variants for patients with a known diagnosis of chronic lymphocytic leukaemia who have not responded to a BTK inhibitor or have lost a previous response to a BTK inhibitor where an alternative BTK inhibitor is being considered as part of treatment. PRIOR TO CHANGE IN TREATMENT
BCL2 small variants for patients with a known diagnosis of chronic lymphocytic leukaemia who have not responded to a BCL2 inhibitor or have lost a previous response to a BCL2inhibitor where an alternative BCL2 inhibitor is being considered as part of treatment. PRIOR TO CHANGE IN TREATMENT
ATM, DLEU2/7, RB1, trisomy 12 copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia, where the clinician has indicated knowledge of these copy number abnormalities will inform management of the patient. DIAGNOSIS

Global copy number +/- structural variant via cytogenetics

GT935 (SNP array for *TP53, ATM, DLEU2/7, RB1*, trisomy 12)

TP53 copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia who have not previously been identified to have disruption of the *TP53* gene who are about to embark on treatment. This

7.检测套餐：慢性淋巴细胞白血病TP533

慢性淋巴细胞白血病检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

对于确诊或疑似诊断为慢性淋巴细胞白血病的成年患者，WGS 可在适应症 M235 下使用，即已穷尽标准治疗检测和/或治疗，前提是符合该检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时/若可获得提交胚系样本(M235.3)。

通过 panel 检测小变异/拷贝数

GT1209 / M94.1(多靶标 NGS 检测板小变异)[*TP53*、*BTK*、*PLCG2*、*BCL2*]

GT929 / M94.2(多靶标 NGS 检测板拷贝数变异)[*TP53*、*ATM*、*DLEU2/7*、*RB1*、12 三体]

*TP53*小变异和拷贝数变异，用于已知诊断为慢性淋巴细胞白血病且既往未发现*TP53*基因破坏、即将开始治疗的患者。只要既往未发现该基因破坏，应在每线治疗前重复该检测。治疗前

*BTK/PLCG2*小变异，用于已知诊断为慢性淋巴细胞白血病、对 BTK 抑制剂无应答或既往对 BTK 抑制剂应答丧失、且正在考虑将替代 BTK 抑制剂作为治疗一部分的患者。治疗变更前

*BCL2*小变异，用于已知诊断为慢性淋巴细胞白血病、对 BCL2 抑制剂无应答或既往对 BCL2 抑制剂应答丧失、且正在考虑将替代 BCL2 抑制剂作为治疗一部分的患者。治疗变更前

ATM、*DLEU2/7*、*RB1*、12 三体拷贝数变异，用于已知诊断为慢性淋巴细胞白血病、且临床医生已表明了解这些拷贝数异常将指导患者管理的患者。诊断

通过细胞遗传学检测全局拷贝数 +/- 结构变异 GT935(用于*TP53*、*ATM*、*DLEU2/7*、*RB1*、12 三体的 SNP 阵列)

*TP53*拷贝数变异，用于已知诊断为慢性淋巴细胞白血病且既往未发现*TP53*基因破坏、即将开始治疗的患者。该

testing should be repeated before each line of treatment providing no previous disruption of this gene has been identified. PRE-TREATMENT *ATM, DLEU2/7, RB1,* trisomy 12 copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia, where the clinician has indicated knowledge of these copy number abnormalities will inform management of the patient. DIAGNOSIS

Targeted copy number +/- structural variant via FISH

GT923 / M94.4 *TP53* FISH
GT231 / M94.8 11q FISH
GT338 / M94.9 13q FISH
GT1281 / M94.10 chr12 FISH
GT926 Other FISH targets

TP53 copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia who have not previously been identified to have disruption of the *TP53* gene who are about to embark on treatment. This testing should be repeated before each line of treatment providing no previous disruption of this gene has been identified. PRE-TREATMENT *ATM, RB1,* trisomy 12 copy number variants for patients with a known diagnosis of chronic lymphocytic leukaemia, where the clinician has indicated knowledge of these copy number abnormalities will inform management of the patient. DIAGNOSIS

Structural variant detection

GT551 / M94.5 (IgHV somatic hypermutation multiplex sequencing)
GT1181 / M94.6 (IgHV somatic hypermutation NGS)

Patients with a known diagnosis of chronic lymphocytic leukaemia who are about to embark on their first-line of treatment. NB this only need to be undertaken once. PRE-TREATMENT

在每线治疗前应重复检测，前提是未发现该基因既往存在破坏。治疗前*ATM*、*DLEU2/7*、*RB1*、12号染色体三体拷贝数变异，适用于已确诊慢性淋巴细胞白血病的患者，且临床医生已表明了解这些拷贝数异常将有助于患者管理。诊断

通过 FISH 进行靶向拷贝数 +/- 结构变异检测

GT923 / M94.4 TP53 FISH GT231 / M94.8 11q FISH GT338 / M94.9 13q FISH GT1281 / M94.10 chr12 FISH GT926 其他FISH靶标

*TP53*拷贝数变异，适用于已知诊断为慢性淋巴细胞白血病且既往未发现*TP53*基因破坏、即将开始治疗的患者。在未发现该基因既往破坏的情况下，应在每线治疗前重复此检测。治疗前*ATM*、*RB1*、12三体拷贝数变异，适用于已知诊断为慢性淋巴细胞白血病的患者，且临床医生已表明了解这些拷贝数异常将指导患者管理。诊断

结构变异检测

GT551 / M94.5(IgHV体细胞超突变多重测序) GT1181 / M94.6(IgHV体细胞超突变NGS)

已确诊慢性淋巴细胞白血病且即将开始一线治疗的患者。注意此项仅需进行一次。治疗前

8. Test package: Myeloma TP126

Myeloma (and other diseases with paraproteins) Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

For adult patients with a confirmed or suspected diagnosis of a plasma cell neoplasm, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Small variant / copy number detection via panel

GT429 / M92.1 (NGS small variant panel)
GT75 / M92.13 (NGS copy number variant panel)

Patients with a confirmed diagnosis of a plasma cell neoplasm where demonstration of high-risk genomic aberration(s) will alter patient management (may relate to intensity of chemotherapy, choice of single or tandem autologous HSCT, intensity and duration of maintenance, follow-up regimen) (17p deletion / 1p loss / 1q gain / chr5 gain / chr9 gain / chr 15 gain) DIAGNOSIS / PRE-TREATMENT (until a high-risk genomic aberration is demonstrated)

- Patients with a confirmed diagnosis of plasma cell myeloma where demonstration of a *BRAF, KRAS* or *NRAS* variant may allow access to a specific targeted inhibitor (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS / PRE-TREATMENT

Global copy number +/- structural variant via cytogenetics

GT178 (SNP array for TP53 / 1p / 1q / chr5 / chr9 / chr 15)
Patients with a confirmed diagnosis of a plasma cell neoplasm where demonstration of high-risk genomic aberration(s) will alter patient management (may relate to intensity of chemotherapy, choice of single or tandem autologous HSCT, intensity and duration of maintenance, follow-up regimen) (17p deletion / 1p loss / 1q gain / chr5 gain / chr9 gain / chr 15 gain) DIAGNOSIS / PRE-TREATMENT (until a high-risk genomic aberration is demonstrated)

Targeted copy number +/- structural variant via FISH

GT40 / M92.2 *IGH::FGFR3* FISH
GT890 / M92.3 *IGH::CCND3* FISH

8.检测项目包：骨髓瘤TP126

骨髓瘤(及其他伴有副蛋白的疾病)检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

对于确诊或疑似浆细胞肿瘤的成年患者，WGS可在适应症M235下提供，即标准诊疗检测和/或治疗已耗尽，前提是符合本检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时提交胚系样本(M235.3)。

通过panel检测小变异/拷贝数 GT429 / M92.1(NGS小变异panel)
GT75 / M92.13(NGS 拷贝数变异检测组合)

已确诊浆细胞肿瘤的患者，若证实高风险基因组异常可改变患者管理(可能涉及化疗强度、单次或串联自体HSCT的选择、维持治疗的强度与持续时间、随访方案)(17p缺失/1p丢失/1q获得/5号染色体获得/9号染色体获得/15号染色体获得)诊断/治疗前(直至证实高风险基因组异常)

- 已确诊浆细胞骨髓瘤的患者，若证实存在*BRAF*、*KRAS*或*NRAS*变异可能使其获得特定靶向抑制剂治疗(注意：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可及)。诊断/治疗前

通过细胞遗传学检测全局拷贝数 +/- 结构变异

GT178(TP53 / 1p / 1q / chr5 / chr9 / chr 15 的SNP阵列)
已确诊浆细胞肿瘤的患者，若证实高风险基因组异常可改变患者管理(可能涉及化疗强度、单次或串联自体HSCT的选择、维持治疗的强度与持续时间、随访方案)(17p缺失/1p丢失/1q获得/5号染色体获得/9号染色体获得/15号染色体获得)诊断/治疗前(直至证实高风险基因组异常)

通过FISH检测靶向拷贝数+/-结构变异 GT40 / M92.2 *IGH::FGFR3* FISH GT890 / M92.3 *IGH::CCND3* FISH

GT187 / M92.4 *IGH::CCND1* FISH
GT549 / M92.5 *IGH::MAF* FISH
GT746 / M92.6 *IGH::MAFB* FISH
GT725 / M92.8 *IGH* FISH
GT1277 / M92.9 Hyperdiploidy copy number FISH
GT855 / M92.10 & M92.11 *CDKN2C* (del1p) & *CKSC1* (gain1q) FISH
GT383 / M92.12 *TP53* (del17p) FISH
GT632 / M92.14 *MYC* rearrangement FISH
GT565 Other FISH Targets

- Patients with a confirmed diagnosis of a plasma cell neoplasm where demonstration of high-risk genomic aberration(s) will alter patient management (may relate to intensity of chemotherapy, choice of single or tandem autologous HSCT, intensity and duration of maintenance, follow-up regimen) (*IGH::FGFR3, IGH::MAF, IGH::MAFB,*
CDKN2C (del1p), *CKSC1* (gain1q), *TP53* (del17p), *MYC*). DIAGNOSIS / PRE-TREATMENT (until a high-risk genomic aberration is demonstrated)
- Patients with a confirmed diagnosis of plasma cell myeloma where demonstration of *IGH::CCND1* will allow access to venetoclax (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS / PRE-TREATMENT

Structural variant detection

GT1274 / M92.7 (NGS structural variant panel – delivered using DNA)

- Patients with a confirmed diagnosis of a plasma cell myeloma where demonstration of high-risk genomic aberration(s) will alter patient management (may relate to intensity of chemotherapy, choice of single or tandem autologous HSCT, intensity and duration of maintenance, follow-up regimen) (*IGH::FGFR3, IGH::MAF, IGH::MAFB, MYC*). DIAGNOSIS / PRE-TREATMENT (until a high-risk genomic aberration is demonstrated)
- Patients with a confirmed diagnosis of plasma cell myeloma where demonstration of *IGH::CCND1* will allow access to venetoclax (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS / PRE-TREATMENT

GT187 / M92.4 IGH::CCND1 荧光原位杂交 GT549 / M92.5 IGH::MAF 荧光原位杂交 GT746 / M92.6 IGH::MAFB 荧光原位杂交 GT725 / M92.8 IGH 荧光原位杂交
GT1277 / M92.9 超二倍体拷贝数 FISH
GT855 / M92.10 & M92.11 CDKN2C（del1p）& CKSC1（gain1q）FISH GT383 / M92.12 TP53（del17p）FISH
GT632 / M92.14 *MYC*重排 FISH GT565 其他 FISH 靶标

• 已确诊浆细胞肿瘤的患者，若证实高风险基因组异常可改变患者管理(可能涉及化疗强度、单次或串联自体HSCT的选择、维持治疗的强度与持续时间、随访方案)(*IGH::FGFR3*、*IGH::MAF*、*IGH::MAFB*、

CDKN2C(del1p)、*CKSC1*(gain1q)、*TP53*(del17p)、*MYC*)。诊断/治疗前(直至证实高风险基因组异常)• 确诊浆细胞骨髓瘤的患者，其中证实*IGH::CCND1*将允许获得venetoclax(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。诊断/治疗前

结构变异检测

GT1274 / M92.7(NGS结构变异检测组合--使用DNA进行检测)

• 确诊浆细胞骨髓瘤的患者，其中证实高风险基因组异常将改变患者管理(可能涉及化疗强度、单次或串联自体HSCT的选择、维持治疗的强度和持续时间、随访方案)(*IGH::FGFR3*、*IGH::MAF*、*IGH::MAFB*、*MYC*)。诊断/治疗前(直至证实高风险基因组异常)• 确诊浆细胞骨髓瘤的患者，其中证实*IGH::CCND1*将允许获得venetoclax(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。诊断/治疗前

9. Test package: Mature B Cell Neoplasms TP377

Mature B cell Neoplasms(excluding CLL) Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT1344 / M107.7 WGS Germline & Tumour - Marginal Zone Lymphoma
GT1389 / M107.9 WGS Tumour First - Marginal Zone Lymphoma
GT1365 / M107.10 WGS Follow-up Germline - Marginal Zone Lymphoma

GT1347 / M99.8 WGS Germline & Tumour - High Grade Lymphoma
GT1393 / M99.10 WGS Tumour First - High Grade Lymphoma
GT1351 / M99.11 WGS Follow-up Germline - High Grade Lymphoma

GT1350 / M96.7 WGS Germline & Tumour - Burkitt Lymphoma
GT1349 / M96.9 WGS Tumour First - Burkitt Lymphoma
GT1388 / M96.10 WGS Follow-up Germline - Burkitt Lymphoma

GT1356 / M93.3 & M95.7 WGS Germline & Tumour - Low Grade Lymphoma
GT1377 / M93.4 & M95.10 WGS Tumour First - Low Grade Lymphoma
GT1366 / M93.5 & M95.11 WGS Follow-up Germline - Low Grade Lymphoma

GT1378 / M101.4 & M182.3 WGS Germline & Tumour - ALK Positive Large B Cell Lymphoma
GT1360 / M101.6 & M182.5 WGS Tumour First - ALK Positive Large B Cell Lymphoma
GT1383 / M101.7 & M182.6 WGS Follow-up Germline - ALK Positive Large B Cell Lymphoma

GT1339 / M97.2 WGS Germline & Tumour - B Cell Lymphoma with 11q Aberration
GT1390 / M97.4 WGS Tumour First - B Cell Lymphoma with 11q Aberration
GT1363 / M97.5 WGS Follow-up Germline - B Cell Lymphoma with 11q Aberration

GT1341 / M98.2 WGS Germline & Tumour - B Cell Lymphoma with IRF4 Rearrangement
GT1392 / M98.4 WGS Tumour First - B Cell Lymphoma with IRF4 Rearrangement
GT1367 / M98.5 WGS Follow-up Germline - B Cell Lymphoma with IRF4 Rearrangement

GT1342 / M110.2 WGS Germline & Tumour - Follicular Lymphoma
GT1379 / M110.3 WGS Tumour First - Follicular Lymphoma

9.检测套餐：成熟B细胞肿瘤TP377

成熟B细胞肿瘤(不包括CLL)检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT1344 / M107.7 WGS胚系与肿瘤 - 边缘区淋巴瘤 GT1389 / M107.9 WGS肿瘤先行 - 边缘区淋巴瘤
GT1365 / M107.10 WGS随访胚系--边缘区淋巴瘤

GT1347 / M99.8 WGS胚系与肿瘤 - 高级别淋巴瘤 GT1393 / M99.10 WGS肿瘤先行 - 高级别淋巴瘤
GT1351 / M99.11 WGS随访胚系--高级别淋巴瘤

GT1350 / M96.7 WGS胚系与肿瘤 - Burkitt淋巴瘤 GT1349 / M96.9 WGS肿瘤先行 - Burkitt淋巴瘤
GT1388 / M96.10 WGS 随访胚系 - 伯基特淋巴瘤

GT1356 / M93.3 & M95.7 WGS胚系与肿瘤 - 低级别淋巴瘤 GT1377 / M93.4 & M95.10 WGS肿瘤先行 - 低级别淋巴瘤
GT1366 / M93.5 & M95.11 WGS 随访胚系 - 低级别淋巴瘤

GT1378 / M101.4 & M182.3 WGS胚系与肿瘤 - ALK阳性大B细胞淋巴瘤

GT1360 / M101.6 & M182.5 WGS肿瘤优先--ALK阳性大B细胞淋巴瘤

GT1383 / M101.7 & M182.6 WGS随访胚系 - ALK阳性大B细胞淋巴瘤

GT1339 / M97.2 WGS 胚系与肿瘤 - 伴11q异常的B细胞淋巴瘤

GT1390 / M97.4 WGS肿瘤先行--伴11q异常的B细胞淋巴瘤

GT1363 / M97.5 WGS随访生殖系--伴11q异常的B细胞淋巴瘤

GT1341 / M98.2 WGS 胚系与肿瘤 - 伴IRF4重排的B细胞淋巴瘤

GT1392 / M98.4 WGS肿瘤优先--伴IRF4重排的B细胞淋巴瘤

GT1367 / M98.5 WGS随访胚系--伴IRF4重排的B细胞淋巴瘤

GT1342 / M110.2 WGS胚系与肿瘤 - 滤泡性淋巴瘤 GT1379 / M110.3 WGS肿瘤先行 - 滤泡性淋巴瘤

GT1343 / M110.4 WGS Follow-up Germline - Follicular Lymphoma

GT1381 / M100.4 WGS Germline & Tumour - Primary Mediastinal B Cell Lymphoma
GT1391 / M100.7 WGS Tumour First - Primary Mediastinal B Cell Lymphoma
GT1387 / M100.8 WGS Follow-up Germline - Primary Mediastinal B Cell Lymphoma

All paediatric and TYA patients with a confirmed or suspected diagnosis of a mature B cell neoplasm are eligible for WGS primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have
occurred. DIAGNOSIS/RELASPE/REFRACTORY

Tumour and germline samples can be submitted simultaneously (i.e M) or separately i.e. tumour first (M) followed by a germline when/if available (M). At any presentation point either M or M +/- M is permissible, however once a non-contaminated germline has been submitted (either as part of M or M) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

For adult patients with a confirmed or suspected diagnosis of a mature B cell neoplasm, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1)
or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Targeted tests

GT272 / M95.12, M104.2, M105.2 & M106.1 (*MYD88* L265P)
GT321 / M108.2 (*BRAF* V600)
GT1337 (*CD79B* Y196)
GT1253 / M95.1, M108.5, M109.1, M181.4 (*IgH*V somatic hypermutation testing multiplex seq)
GT1380 / M95.2, M108.6, M109.2, M181.5 (*IgH*V somatic hypermutation testing NGS)

- *MYD88* L265P: for patients with a suspected diagnosis of lymphoplasmacytic lymphoma OR large B cell lymphoma in an immune privileged (intra-ocular / CNS / testicular) where knowledge of the presence or absence of this variant will assist in diagnosis including that of IgM MGUS. DIAGNOSIS

GT1343 / M110.4 WGS 随访胚系 - 滤泡性淋巴瘤

GT1381 / M100.4 WGS 胚系与肿瘤 - 原发纵隔B细胞淋巴瘤

GT1391 / M100.7 WGS肿瘤优先--原发纵隔B细胞淋巴瘤

GT1387 / M100.8 WGS 随访胚系 - 原发性纵隔B细胞淋巴瘤

所有确诊或疑似成熟B细胞肿瘤的儿童和TYA患者主要可在诊断时进行WGS。若诊断时未进行WGS，在复发或对某线治疗难治时进行本检测可能合适。在特殊情况下，若极有可能已发生显著克隆演化，可考虑重复WGS。诊断/复发/难治

肿瘤和胚系样本可同时提交(即M)或分别提交，即先提交肿瘤样本(M)，随后在可获得时提交胚系样本(M)。在任何就诊时间点，允许M或M +/- M，但一旦提交了未受污染的胚系样本(作为M或M的一部分)，该样本可通过生物信息学方法在所有后续肿瘤样本中重复用作胚系。

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

对于确诊或疑似成熟B细胞肿瘤的成年患者，WGS可在适应症M235下提供，即标准诊疗检测和/或治疗已耗尽，前提是符合本检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时提交胚系样本(M235.3)。

靶向检测

GT272 / M95.12、M104.2、M105.2 & M106.1（MYD88 L265P）
GT321 / M108.2（BRAF V600）GT1337（CD79B Y196）

GT1253 / M95.1、M108.5、M109.1、M181.4(*IgH* V体细胞超突变检测多重测序)

GT1380 / M95.2、M108.6、M109.2、M181.5(*IgH* V体细胞超突变检测NGS)

- *MYD88* L265P：适用于疑似淋巴浆细胞淋巴瘤或免疫豁免部位(眼内/中枢神经系统/睾丸)大B细胞淋巴瘤的患者，了解该变异存在与否将有助于诊断，包括IgM MGUS的诊断。诊断

- *MYD88* L265P: for patients with a diagnosis of a mature B cell neoplasm where knowledge of the presence or absence of this variant will assist in the treatment choice. PRE-TREATMENT
- *BRAF* V600: for patients with a suspected diagnosis of hairy cell leukaemia where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS
- *BRAF* V600: for patients whose clinical presentation would render them eligible for treatment with a BRAF-inhibitor in the event they had this variant (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT
- *CD79B* Y196: for patients with a suspected diagnosis of a large B cell lymphoma in an immune privileged (intra-ocular / CNS / testicular) where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS
- *IgH*V hypermutation testing: for patients with hairy cell leukaemia where knowledge of hypermutation will assist with management. DIAGNOSIS / PRE-TREATMENT

Small variant / copy number detection via panel (includes Resistance mutation testing)

It is envisaged that small variants and copy number variants will be delivered on a single panel.

GT267 / M93.6 (*BTK, PLCG2, RAS, MAP2K1, BCL2*), M95.4 (*EZH2*), M95.6 (*BTK2, PLCG2*), M102.5 (*TP53*), M103.4 (*CARD11, CREBBP, EZH2, ARID1A, EP300, MEF2B, FOXO1*), M104.1 (*MYD88, CXCR4*), M105.1 (*MYD88, CXCR4*), M108.1 (*BRAF*), M181.1 (*MAP2K1*), M110.1 (*MAP2K1*) (NGS small variant panel)
GT306 / M95.9 (*EZH2*) & M100.5 (*CD274/PDCD1LG2, REL*) (NGS copy number panel)

- *MYD88*: for patients with a suspected diagnosis of lymphoplasmacytic lymphoma OR large B cell lymphoma in an immune privileged (intra-ocular / CNS / testicular) where knowledge of the presence or absence of this variant will assist in diagnosis including that of IgM MGUS. DIAGNOSIS
- *CD79B*: for patients with a suspected diagnosis of a large B cell lymphoma in an immune privileged (intra-ocular / CNS / testicular) where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS
- *BRAF*: for patients with a suspected diagnosis of hairy cell leukaemia where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS
- *MAP2K1*: for patients with a suspected diagnosis of variant hairy cell leukaemia where knowledge of the presence or absence of this variant will assist in diagnosis.
- *CARD11, CREBBP, EZH2, ARID1A, EP300, MEF2B, FOXO1*: for patients with a known diagnosis of follicular lymphoma where knowledge

• *MYD88* L265P：适用于确诊成熟B细胞肿瘤的患者，了解该变异存在与否将有助于治疗选择。治疗前

• *BRAF* V600：适用于疑似毛细胞白血病的患者，了解该变异存在与否将有助于诊断。诊断

• *BRAF* V600：适用于临床表现使其在具有该变异时有资格接受BRAF抑制剂治疗的患者(注意：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可及)。治疗前

• *CD79B* Y196：适用于疑似免疫豁免部位(眼内/中枢神经系统/睾丸)大B细胞淋巴瘤的患者，了解该变异存在与否将有助于诊断。诊断

• *IgH* V 超突变检测：用于毛细胞白血病患者，了解超突变状态有助于指导管理。诊断/治疗前

通过 panel 检测小变异/拷贝数(包括耐药突变检测)

预期小变异和拷贝数变异将在单一 panel 上交付。

GT267 / M93.6（BTK、PLCG2、RAS、MAP2K1、BCL2）、M95.4（EZH2）、M95.6（BTK2、PLCG2）、M102.5（TP53）、M103.4（CARD11、CREBBP、EZH2、ARID1A、EP300、MEF2B、FOXO1）、M104.1（MYD88、CXCR4）、M105.1（MYD88、CXCR4）、M108.1（BRAF）、M181.1（MAP2K1）、M110.1（MAP2K1）（NGS小变异panel）GT306 / M95.9（EZH2）& M100.5（CD274/PDCD1LG2、REL）（NGS拷贝数panel）

• *MYD88*：适用于疑似诊断为淋巴浆细胞淋巴瘤或免疫豁免部位(眼内/中枢神经系统/睾丸)大B细胞淋巴瘤的患者，了解该变异的存在或缺失将有助于诊断，包括IgM MGUS的诊断。诊断 • *CD79B*：适用于疑似诊断为免疫豁免部位(眼内/中枢神经系统/睾丸)大B细胞淋巴瘤的患者，了解该变异的存在或缺失将有助于诊断。诊断

• *BRAF*：用于疑似毛细胞白血病患者，了解该变异的存在或缺失有助于诊断。诊断

• *MAP2K1*：用于疑似变异型毛细胞白血病患者，了解该变异的存在或缺失有助于诊断。

• *CARD11*、*CREBBP*、*EZH2*、*ARID1A*、*EP300*、*MEF2B*、*FOXO1*：用于已知诊断为滤泡性淋巴瘤的患者，了解

of the presence or absence of variants in these genes will assist with prognostication and treatment choice. DIAGNOSIS / PRE-TREATMENT

- *TP53*: for patients with a known diagnosis of mantle cell lymphoma where knowledge of the presence or absence of variants in these genes will assist with prognostication and treatment choice. DIAGNOSIS / PRE-TREATMENT
- *MYD88, CXCR4*: for patients with a diagnosis of a mature B cell neoplasm (predominantly lymphoplasmacytic lymphoma) where knowledge of the presence or absence of this variant will assist in the treatment choice. PRE-TREATMENT
- *BRAF*: for patients whose clinical presentation would render them eligible for treatment with a BRAF-inhibitor in the event they had an activating variant in this gene (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT
- *MAP2K1*: for patients whose clinical presentation would render them eligible for treatment with a MEK-inhibitor in the event they had an activating variant in this gene (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT
- *EZH2*: for patients whose clinical presentation would render them eligible for treatment with a EZH2-inhibitor in the event they had an activating variant in this gene (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT
- *BTK, PLCG2, RAS, MAP2K1, BCL2*: for patients whose clinical presentation would render them eligible for treatment with a specific inhibitor where knowledge of variants in one or more of these genes would assist in the treatment decision OR for patients already receiving a specific inhibitor who have not responded or are no longer responding as expected and where knowledge of variants in one or more of these genes may explain the lack of / loss of response (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT / RELAPSE / REFRACTORY
- *EZH2* deletion: for patients whose clinical presentation would render them eligible for treatment with a EZH2-inhibitor in the event they a deletion of this gene (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT
- *CD274/PDCD1LG2, REL* copy number abnormality: for patients with a suspected diagnosis of primary mediastinal large B cell lymphoma where knowledge of the presence or absence of these copy number abnormalities will assist with diagnosis. DIAGNOSIS

这些基因中变异的存在或缺失有助于预后判断和治疗选择。诊断/治疗前 • *TP53*：用于已知诊断为套细胞淋巴瘤的患者，了解这些基因中变异的存在或缺失有助于预后判断和治疗选择。诊断/治疗前

• *MYD88*、*CXCR4*：用于诊断为成熟 B 细胞肿瘤(主要为淋巴浆细胞淋巴瘤)的患者，了解该变异的存在或缺失有助于治疗选择。治疗前

• *BRAF*：适用于临床表现使其在该基因存在激活变异时有资格接受BRAF抑制剂治疗的患者(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。治疗前 • *MAP2K1*：适用于临床表现使其在该基因存在激活变异时有资格接受MEK抑制剂治疗的患者(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。治疗前 • *EZH2*：适用于临床表现使其在该基因存在激活变异时有资格接受EZH2抑制剂治疗的患者(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。治疗前 • *BTK*、*PLCG2*、*RAS*、*MAP2K1*、*BCL2*：适用于临床表现使其有资格接受特定抑制剂治疗、且了解这些基因中一个或多个的变异将有助于治疗决策的患者，或已接受特定抑制剂但未按预期应答或不再按预期应答、且了解这些基因中一个或多个的变异可能解释缺乏应答/应答丧失的患者(注：应预期在证实(或不证实)特定异常后，基因组决定的治疗将可获得)。治疗前/复发/难治

• *EZH2*缺失：适用于临床表现使其在存在该基因缺失时有资格接受 EZH2 抑制剂治疗的患者(注意：应预期基因组确定的治疗将在特定异常得到证实(或未证实)后可用)。治疗前 • *CD274/PDCD1LG2*、*REL*拷贝数异常：适用于疑似原发性纵隔大 B 细胞淋巴瘤诊断的患者，了解这些拷贝数异常的存在与否将有助于诊断。诊断

Global copy number +/- structural variant via cytogenetics

GT918 / M225.5 (karyotype)
GT444 / M97.1 (SNP array for 11q abnormalities)

通过细胞遗传学检测全局拷贝数 +/- 结构变异 GT918 / M225.5(核型)

GT444 / M97.1(用于11q异常的SNP阵列)

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- Karyotype: to be considered in exceptional circumstances where other testing has failed to assist with diagnosis. DIAGNOSIS
- SNP array for 11q abnormalities: for patients with a suspected diagnosis of a 'Burkitt-type' lymphoma in whom a *MYC* rearrangement has not been detected. DIAGNOSIS

Targeted copy number +/- structural variant via FISH / RT-PCR

GT852 / M95.3 (*IGH*) SV
GT414 / M95.5 (*EZH2*) CN
GT1394 / M96.1 & M99.1 (*MYC*) SV
GT576 / M96.2 & M99.2 (*IGH::MYC*)
GT664 / M96.3 & M99.3 (*IGK::MYC*)
GT312 / M96.4 & M99.4 (*IGL::MYC*)
GT613 / M96.5, M99.5 & M103.2 (*BCL2*) SV
GT646 / M99.6 & M103.1 (*IGH::BCL2*) SV
GT1285 / M96.6 / M99.7 / M103.3 (*BCL6*) SV
GT479 / M97.1 (11q) CN
GT369 / M98.1 (*IRF4*) SV
GT1404 / M100.1 & M100.2 (*CD274/PDCD1LG2*) SV & CN
GT1348 (*CIITA*) SV
GT534 / M100.3 (*REL*) CN
GT1354 / M101.1 (*ALK*) SV
GT1407 / M102.1 (*IGH::CCND1*) SV
GT868 / M102.2 (*CCND1*) SV
GT637 / M102.3 (*CCND2*) SV
GT1007 / M107.6 (*FOXP1*) SV
GT120 / M107.1, M107.3 & M107.4 (*MALT1*) SV
GT665 Other FISH Targets
GT779 / M107.2 & M107.5 (*BCL10*) SV

- *MYC / BCL2 / BCL6 / IGH / IGK / IGL*: for patients with a suspected diagnosis of a large B cell lymphoma OR Burkitt lymphoma where demonstration of the presence or absence of rearrangements of some of these genes will assist with diagnosis. It is recognised that labs will have specific algorithms as per which of these probes and in which order they are used in specific clinical / morphological / immunohistochemical scenarios. DIAGNOSIS
- *ALK* – for patients with a suspected diagnosis of a large B cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an *ALK*-rearrangement. DIAGNOSIS
- *IRF4* - for patients with a suspected diagnosis of a large B cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an IRF4-rearrangement. DIAGNOSIS
- 11q - for patients with a suspected diagnosis of a 'Burkitt-type' lymphoma in whom a MYC rearrangement has not been detected. DIAGNOSIS
- *CD274/PDCD1LG2, CIITA, REL* - for patients with a suspected diagnosis of a primary mediastinal B cell lymphoma where knowledge of

- 核型：在特殊情况下考虑，当其他检测未能协助诊断时。诊断

- 用于 11q 异常的 SNP array：用于疑似"伯基特型"淋巴瘤且未检测到*MYC*重排的患者。诊断

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

GT852 / M95.3（IGH）结构变异
GT414 / M95.5（EZH2）拷贝数

GT1394 / M96.1 & M99.1（MYC）结构变
异 GT576 / M96.2 & M99.2（IGH::MYC）
GT664 / M96.3 & M99.3（IGK::MYC）
GT312 / M96.4 & M99.4（IGL::MYC）

GT613 / M96.5、M99.5 & M103.2（BCL2）结构变异
GT646 / M99.6 & M103.1（IGH::BCL2）结构变异
GT1285 / M96.6 / M99.7 / M103.3（BCL6）结构变异
GT479 / M97.1（11q）拷贝数 GT369 /
M98.1（IRF4）结构变异

GT1404 / M100.1 & M100.2（CD274/PDCD1LG2）结构变异 & 拷贝
数 GT1348（CIITA）结构变异 GT534 / M100.3（REL）拷贝数
GT1354 / M101.1（ALK）结构变异

GT1407 / M102.1（IGH::CCND1）结构
变异 GT868 / M102.2（CCND1）结构变
异 GT637 / M102.3（CCND2）结构变异
GT1007 / M107.6（FOXP1）结构变异

GT120 / M107.1、M107.3 & M107.4（MALT1）结构变
异 GT665 其他FISH靶标 GT779 / M107.2 &
M107.5（BCL10）结构变异

- *MYC / BCL2 / BCL6 / IGH / IGK / IGL*：用于疑似大 B 细胞淋巴瘤或伯基特淋巴瘤患者，证明其中某些基因重排的存在或缺失有助于诊断。已认识到各实验室将根据具体临床/形态学/免疫组化情形制定特定的算法，决定使用哪些探针及其顺序。诊断

- *ALK* - 用于疑似大 B 细胞淋巴瘤患者，其免疫组化提示可能存在*ALK*-重排。诊断

- *IRF4* - 用于疑似大 B 细胞淋巴瘤患者，其免疫组化提示可能存在 IRF4 重排。诊断
- 11q - 用于疑似"伯基特型"淋巴瘤且未检测到 MYC 重排的患者。诊断

- *CD274/PDCD1LG2*、*CIITA*、*REL* - 用于疑似原发性纵隔 B 细胞淋巴瘤患者，了解

the presence or absence of these copy number abnormalities will assist with diagnosis. DIAGNOSIS

- *IGH::BCL2* – for patients with a suspected diagnosis of follicular lymphoma where demonstration of the presence or absence of this translocation will assist with diagnosis. DIAGNOSIS
- *BCL6* – for patients with a suspected diagnosis of follicular lymphoma who have not been shown to have an *IGH::BCL2* rearrangement where demonstration of the presence or absence of this translocation will assist with diagnosis. DIAGNOSIS
- *IGH::CCND1 / CCND1 / CCND2* – for patients with a suspected diagnosis of mantle cell lymphoma; it is envisaged that a rearrangement of *CCND1* will be sought first and only if not detected will *CCND2* be tested, if this also negative and the clinical suspicion for a diagnosis of mantle cell lymphoma remains high, the possibility to test for rearrangements of *CCND3* should be explored via WGS or custom panel. DIAGNOSIS
- *MALT1 / BCL10 / (BIRC3::MALT1 / IGH::BCL10 / IGH::MALT1) / FOXP1* - for patients with a suspected diagnosis of extra-nodal marginal lymphoma of mucosa-associated lymphoid tissue where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS
- *EZH2* - for patients whose clinical presentation would render them eligible for treatment with a EZH2-inhibitor in the event they had a deletion of this gene (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). PRE-TREATMENT

Structural variant detection via NGS panel

Given many clinically significant gene rearrangements in lymphoma involve the juxtaposition of the regulatory regions of highly expressed genes in B or T cells next to a gene relevant for cell proliferation or survival, there is often no resulting conventional ‘fusion’ protein (rather the rearrangement results in deregulation and over-expression). If no fusion protein results from a rearrangement, an RNA panel is unlikely to be useful in its detection, rather if an NGS panel approach is to be pursued this will likely require the use of DNA as the substrate.

GT795 NGS DNA based panel - structural variants
M95.8 (*IGH / IGK / IGL*)
M96.8 / M99.9 (*IGH / IGK / IGL / MYC / BCL2 / BCL6*)
M98.3 (*IRF4*)
M100.6 (*CD274 / CIITA*)
M102.4 (*IGH::CCDN1, CCND1, CCND2, CCND3*)
M103.5 (*IGH::BCL2, BCL2, BCL6*)
M107.8 (*IGH::MALT1, IGH::BCL10, FOXP1, BIRC3::MALT1*)
M101.5 (*CLTC::ALK, ALK::NPM1, ALK*)

GT50 NGS RNA based panel - structural variants
M100.6 (*CD274 / CIITA*)
M101.5 (*CLTC::ALK, ALK::NPM1, ALK*)

这些拷贝数异常的存在或缺失有助于诊断。诊断

• *IGH::BCL2* - 用于疑似滤泡性淋巴瘤患者，证明该易位的存在或缺失有助于诊断。诊断

• *BCL6* - 用于疑似滤泡性淋巴瘤且未显示有*IGH::BCL2*重排的患者，证明该易位的存在或缺失有助于诊断。诊断

• *IGH::CCND1 / CCND1 / CCND2* - 用于疑似套细胞淋巴瘤患者；预期首先寻找 CCND1 重排，仅在未检测到时才检测 CCND2，若后者亦为阴性且套细胞淋巴瘤的临床怀疑仍然很高，应通过 WGS 或定制 panel 探索检测*CCND3*重排的可能性。诊断

• *MALT1 / BCL10 / (BIRC3::MALT1 / IGH::BCL10 / IGH::MALT1) / FOXP1* - 适用于疑似黏膜相关淋巴组织结外边缘区淋巴瘤诊断的患者，了解该变异的存在与否将有助于诊断。诊断 • *EZH2* - 适用于临床表现使其在存在该基因缺失时有资格接受 EZH2 抑制剂治疗的患者(注意：应预期基因组确定的治疗将在特定异常得到证实(或未证实)后可用)。治疗前

通过NGS panel进行结构变异检测

鉴于淋巴瘤中许多具有临床意义的基因重排涉及将 B 细胞或 T 细胞中高表达基因的调控区域与与细胞增殖或存活相关的基因并置，通常不会产生常规的“融合”蛋白(而是重排导致失调和过表达)。如果重排不产生融合蛋白，RNA 检测板不太可能有助于其检测，相反，如果采用 NGS 检测板方法，这很可能需要使用 DNA 作为底物。

GT795 基于NGS DNA的panel - 结构变异 M95.8（IGH / IGK / IGL）M96.8 / M99.9（IGH / IGK / IGL / MYC / BCL2 / BCL6）M98.3（IRF4）M100.6（CD274 / CIITA）

M102.4（IGH::CCDN1、CCND1、CCND2、CCND3）
M103.5（IGH::BCL2、BCL2、BCL6）
M107.8（IGH::MALT1、IGH::BCL10、FOXP1、BIRC3::MALT1）
M101.5（CLTC::ALK、ALK::NPM1、ALK）

GT50 基于 NGS RNA 的 panel - 结构变异 M100.6(
CD274 / CIITA)

- *MYC / BCL2 / BCL6 / IGH / IGK / IGL*: for patients with a suspected diagnosis of a large B cell lymphoma OR Burkitt lymphoma where demonstration of the presence or absence of rearrangements of some of these genes will assist with diagnosis. It is recognised that labs will have specific algorithms as per which of these probes and in which order they are used in specific clinical / morphological / immunohistochemical scenarios. DIAGNOSIS
- *ALK* – for patients with a suspected diagnosis of a large B cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an ALK-rearrangement. DIAGNOSIS
- *IRF4* - for patients with a suspected diagnosis of a large B cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an IRF4-rearrangement. DIAGNOSIS
- *CD274/PDCD1LG2, CIITA* - for patients with a suspected diagnosis of a primary mediastinal B cell lymphoma where knowledge of the presence or absence of these copy number abnormalities will assist with diagnosis. DIAGNOSIS
- *IGH::BCL2* – for patients with a suspected diagnosis of follicular lymphoma where demonstration of the presence or absence of this translocation will assist with diagnosis. DIAGNOSIS
- *BCL6* – for patients with a suspected diagnosis of follicular lymphoma who have not been shown to have an *IGH::BCL2* rearrangement where demonstration of the presence or absence of this translocation will assist with diagnosis. DIAGNOSIS
- *IGH::CCND1 / CCND1 / CCND2* – for patients with a suspected diagnosis of mantle cell lymphoma; it is envisaged that a rearrangement of *CCND1* will be sought first and only if not detected will *CCND2* be tested, if this also negative and the clinical suspicion for a diagnosis of mantle cell lymphoma remains high, the possibility to test for rearrangements of *CCND3* should be explored via WGS or custom panel. DIAGNOSIS
- *MALT1 / BCL10 / (BIRC3::MALT1 / IGH::BCL10 / IGH::MALT1) / FOXP1* - for patients with a suspected diagnosis of extra-nodal marginal lymphoma of mucosa-associated lymphoid tissue where knowledge of the presence or absence of this variant will assist in diagnosis. DIAGNOSIS

• *MYC / BCL2 / BCL6 / IGH / IGK / IGL*：用于疑似大 B 细胞淋巴瘤或伯基特淋巴瘤患者，证明其中某些基因重排的存在或缺失有助于诊断。已认识到各实验室将根据具体临床/形态学/免疫组化情形制定特定的算法，决定使用哪些探针及其顺序。诊断

• *ALK* - 用于疑似大 B 细胞淋巴瘤患者，其免疫组化提示可能存在 ALK 重排。诊断

• *IRF4* - 用于疑似诊断为弥漫大B细胞淋巴瘤且免疫组化结果提示可能存在IRF4重排的患者。诊断

• *CD274/PDCD1LG2*、*CIITA* - 用于疑似诊断原发性纵隔B细胞淋巴瘤的患者，了解这些拷贝数异常的存在与否将有助于诊断。诊断

• *IGH::BCL2* - 用于疑似滤泡性淋巴瘤患者，证明该易位的存在或缺失有助于诊断。诊断

• *BCL6* - 用于疑似滤泡性淋巴瘤且未显示有*IGH::BCL2*重排的患者，证明该易位的存在或缺失有助于诊断。诊断

• *IGH::CCND1 / CCND1 / CCND2* - 用于疑似套细胞淋巴瘤患者；预期首先寻找 CCND1 重排，仅在未检测到时才检测 CCND2，若后者亦为阴性且套细胞淋巴瘤的临床怀疑仍然很高，应通过 WGS 或定制 panel 探索检测*CCND3*重排的可能性。诊断

• *MALT1 / BCL10 / (BIRC3::MALT1 / IGH::BCL10 / IGH::MALT1) / FOXP1* - 用于疑似诊断黏膜相关淋巴组织结外边缘区淋巴瘤的患者，了解该变异的存在与否将有助于诊断。诊断

10. Test package: Mature T Cell Neoplasms TP58

Mature T cell and NK cell Neoplasms Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT1352 / M116.3 WGS Germline & Tumour - Hepatosplenic T Cell Lymphoma
GT1361 / M116.5 WGS Tumour First - Hepatosplenic T Cell Lymphoma
GT1364 / M116.6 WGS Follow-up Germline - Hepatosplenic T Cell Lymphoma

GT1386 / M111.5 WGS Germline & Tumour - T Cell NHL
GT1358 / M111.7 WGS Tumour First - T Cell NHL
GT1353 / M111.8 WGS Follow-up Germline - T Cell NHL

GT1369 / M112.5 WGS Germline & Tumour - ALK Negative Anaplastic Large Cell Lymphoma
GT1362 / M112.7 WGS Tumour First - ALK Negative Anaplastic Large Cell Lymphoma
GT1359 / M112.8 WGS Follow-up Germline - ALK Negative Anaplastic Large Cell Lymphoma

GT1376 / M115.2 WGS Germline & Tumour - NK Cell/Gamma-Delta T Cell Lymphoma
GT1384 / M115.3 WGS Tumour First - NK Cell/Gamma-Delta T Cell Lymphoma
GT1372 / M115.4 WGS Follow-up Germline - NK Cell/Gamma-Delta T Cell Lymphoma

All paediatric and TYA patients with a confirmed or suspected diagnosis of a mature T or NK cell neoplasm are eligible for WGS primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS/RELASPE/REFRACTORY

Tumour and germline samples can be submitted simultaneously (i.e M) or separately i.e. tumour first (M) followed by a germline when/if available (M). At any presentation point either M or M +/- M is permissible, however once a non-contaminated germline has been submitted (either as part of M or M) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

10.检测套餐：成熟T细胞肿瘤TP58

成熟 T 细胞和 NK 细胞肿瘤检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT1352 / M116.3 WGS 胚系与肿瘤 - 肝脾T细胞淋巴瘤

GT1361 / M116.5 WGS 肿瘤先测 - 肝脾T细胞淋巴瘤 GT1364 / M116.6 WGS 随访胚系 - 肝脾T细胞淋巴瘤

GT1386 / M111.5 WGS 胚系与肿瘤 - T细胞NHL GT1358 / M111.7 WGS 肿瘤先测 - T细胞NHL
GT1353 / M111.8 WGS 随访胚系 - T 细胞 NHL

GT1369 / M112.5 WGS 胚系与肿瘤 - ALK阴性间变性大细胞淋巴瘤

GT1362 / M112.7 WGS Tumour First - ALK阴性间变性大细胞淋巴瘤

GT1359 / M112.8 WGS 随访胚系 - ALK阴性间变性大细胞淋巴瘤

GT1376 / M115.2 WGS 胚系与肿瘤 - NK细胞/γδT细胞淋巴瘤

GT1384 / M115.3 WGS 肿瘤优先 - NK细胞/γδT细胞淋巴瘤

GT1372 / M115.4 WGS随访生殖系--NK细胞/γδT细胞淋巴瘤

所有确诊或疑似诊断为成熟T细胞或NK细胞肿瘤的儿童和TYA患者主要在原发诊断时符合WGS检测资格。若诊断时未进行WGS，在复发或对某一线治疗难治时进行该检测可能是合适的。在特殊情况下，若存在高度可能已发生显著克隆演化，可考虑重复WGS。诊断/复发/难治

肿瘤和胚系样本可同时提交(即M)或分别提交，即先提交肿瘤样本(M)，随后在可获得时提交胚系样本(M)。在任何就诊时间点，允许M或M +/- M，但一旦提交了未受污染的胚系样本(作为M或M的一部分)，该样本可通过生物信息学方法在所有后续肿瘤样本中重复用作胚系。

Currently WGS is performed as an **addition** to any standard of care testing that an individual is eligible for.

For adult patients with a confirmed or suspected diagnosis of a mature B cell neoplasm, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Small variant detection via panel

GT1279 / M111.1 (*RHOA, DNMT3A, IDH2, TET2, PLCG1, CARD11, IRF4, POT1, VAV1, FYN*), M114.1, M115.1 & M116.1 (*STAT3, STAT5b*)

- *RHOA, DNMT3A, IDH2, TET2, PLCG1, CARD11, IRF4, POT1, VAV1, FYN* – for patients with a suspected diagnosis of a mature T cell neoplasm where knowledge of the presence or absence of variants in these genes will assist in the differentiation from reactive looking T cells. DIAGNOSIS
- *STAT3, STAT5b* – for patients with a suspected diagnosis of T-cell large granular lymphocyte leukaemia, NK-cell large granular lymphocyte leukaemia or hepatosplenic T cell lymphoma where knowledge of the presence or absence of variants in these genes will assist with diagnosis. DIAGNOSIS

Global copy number +/- structural variant via cytogenetics

GT918 / M225.5 (karyotype)

- Karyotype: to be considered in exceptional circumstances where other testing has failed to assist with diagnosis. DIAGNOSIS

Targeted copy number +/- structural variant via FISH / RT-PCR

GT1146 / M111.4 (*TRA, TRB, TRG*)
GT256 / M112.3 (*IRF4/DUSP22*)
GT733 / M112.4 (*TP63*)
GT1385 / M182.1 (*ALK::NPM1*)
GT1416 / M182.2 (*ALK*)
GT59 / M113.1 (*TCL1A*)
GT1422 / M113.2 (chromosome 8)
GT345 / M116.2 (chromosome 7)
GT1338 Other FISH Targets

- *TRA, TRB, TRG* - for patients with a suspected diagnosis of a mature T cell neoplasm where knowledge of the presence or absence of rearrangements of these genes will assist in the differentiation from reactive looking T cells. DIAGNOSIS

目前 WGS 作为个体有资格接受的任何标准诊疗检测的**补充**进行。

对于确诊或疑似成熟B细胞肿瘤的成年患者，WGS可在适应症M235下提供，即标准诊疗检测和/或治疗已耗尽，前提是符合本检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时提交胚系样本(M235.3)。

通过 panel 检测小变异

GT1279 / M111.1（RHOA、DNMT3A、IDH2、TET2、PLCG1、CARD11、IRF4、POT1、VAV1、FYN）、M114.1、M115.1 & M116.1（STAT3、STAT5b）

- *RHOA*、*DNMT3A*、*IDH2*、*TET2*、*PLCG1*、*CARD11*、*IRF4*、*POT1*、*VAV1*、*FYN* - 适用于疑似成熟 T 细胞肿瘤诊断的患者，了解这些基因中变异的存在与否将有助于与反应性外观 T 细胞相鉴别。诊断
- *STAT3*、*STAT5b* - 适用于疑似 T 细胞大颗粒淋巴细胞白血病、NK 细胞大颗粒淋巴细胞白血病或肝脾 T 细胞淋巴瘤诊断的患者，了解这些基因中变异的存在与否将有助于诊断。诊断

通过细胞遗传学检测全局拷贝数 +/- 结构变异 GT918 / M225.5(核型)

- 核型：在特殊情况下考虑，当其他检测未能协助诊断时。诊断

通过 FISH / RT-PCR 进行靶向拷贝数 +/- 结构变异检测

GT1146 / M111.4 (TRA、TRB、TRG)
GT256 / M112.3 (IRF4/DUSP22) GT733 /
M112.4 (TP63) GT1385 / M182.1
(ALK::NPM1) GT1416 / M182.2 (ALK)
GT59 / M113.1 (TCL1A) GT1422 /
M113.2 (8号染色体) GT345 / M116.2 (7号
染色体) GT1338 其他FISH靶标

- *TRA*、*TRB*、*TRG* - 用于疑似诊断成熟T细胞肿瘤的患者，了解这些基因重排的存在与否将有助于与反应性T细胞进行鉴别。诊断

- *IRF4/DUSP22, TP63* – for patients with a known diagnosis of *ALK*-negative anaplastic large cell lymphoma where knowledge of the presence or absence of rearrangements of these genes will assist in prognostication and treatment decisions. DIAGNOSIS, PRE-TREATMENT
- *ALK, ALK::NPM1* - for patients with a suspected diagnosis of *ALK*-positive anaplastic large cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an *ALK* rearrangement. DIAGNOSIS
- *TCL1A* – for patients with a suspected diagnosis of T-cell prolymphocytic leukaemia where knowledge of the presence or absence of a rearrangement of this gene will assist with diagnosis. DIAGNOSIS
- Trisomy 8 – for patients with a suspected diagnosis of T-cell prolymphocytic leukaemia where knowledge of the presence or absence of this copy number abnormality will assist with diagnosis. DIAGNOSIS
- Iso7q – for patients with a suspected diagnosis of hepatosplenic T cell lymphoma where knowledge of the presence or absence of this copy number abnormality will assist with diagnosis. DIAGNOSIS

Structural variant detection via NGS panel

GT821 NGS DNA based panel - structural variants
M111.6 (*TRA, TRB, TRG*)
M112.6 (*IRF4/DUSP22, TP63*)
M113.3 (*TCL1A*)

GT1153 NGS RNA based panel - structural variants
M112.6 (*TP63*)
M182.4 (*ALK::NPM1, ALK*)

- *TRA, TRB, TRG* - for patients with a suspected diagnosis of a mature T cell neoplasm where knowledge of the presence or absence of rearrangements of these genes will assist in the differentiation from reactive looking T cells. DIAGNOSIS
- *IRF4/DUSP22, TP63* – for patients with a known diagnosis of *ALK*-negative anaplastic large cell lymphoma where knowledge of the presence or absence of rearrangements of these genes will assist in prognostication and treatment decisions. DIAGNOSIS, PRE-TREATMENT
- *ALK, ALK::NPM1* - for patients with a suspected diagnosis of *ALK*-positive anaplastic large cell lymphoma whose immunohistochemistry is such that there is suspicion they may have an *ALK* rearrangement. DIAGNOSIS
- *TCL1A* – for patients with a suspected diagnosis of T-cell prolymphocytic leukaemia where knowledge of the presence or absence of a rearrangement of this gene will assist with diagnosis.
- Trisomy 8 – for patients with a suspected diagnosis of T-cell prolymphocytic leukaemia where knowledge of the presence or absence of this copy number abnormality will assist with diagnosis. DIAGNOSIS

• *IRF4/DUSP22*、*TP63* - 用于已知诊断为*ALK*阴性间变性大细胞淋巴瘤的患者，了解这些基因重排的存在与否将有助于预后判断和治疗决策。诊断、治疗前

• *ALK*、*ALK::NPM1* - 用于疑似诊断*ALK*阳性间变性大细胞淋巴瘤且免疫组化结果提示可能存在*ALK*重排的患者。诊断

• *TCL1A* - 适用于疑似 T 细胞幼淋巴细胞白血病诊断的患者，了解该基因重排的存在与否将有助于诊断。诊断 • 8 三体 - 适用于疑似 T 细胞幼淋巴细胞白血病诊断的患者，了解该拷贝数异常的存在与否将有助于诊断。诊断 • Iso7q - 适用于疑似肝脾 T 细胞淋巴瘤诊断的患者，了解该拷贝数异常的存在与否将有助于诊断。诊断

通过NGS panel进行结构变异检测

GT821 NGS DNA基础panel - 结构变异 M111.6(*TRA*、*TRB*、*TRG*) M112.6(*IRF4/DUSP22*、*TP63*) M113.3(*TCL1A*)

GT1153 基于NGS RNA的panel--结构变异 M112.6(*TP63*)

• *TRA*、*TRB*、*TRG* - 用于疑似诊断成熟T细胞肿瘤的患者，了解这些基因重排的存在与否将有助于与反应性T细胞进行鉴别。诊断

• *IRF4/DUSP22*、*TP63* - 用于已知诊断为*ALK*阴性间变性大细胞淋巴瘤的患者，了解这些基因重排的存在与否将有助于预后判断和治疗决策。诊断、治疗前

• *ALK*、*ALK::NPM1* - 用于疑似诊断*ALK*阳性间变性大细胞淋巴瘤且免疫组化结果提示可能存在*ALK*重排的患者。诊断

• *TCL1A* - 用于疑似诊断T细胞幼淋巴细胞白血病的患者，了解该基因重排的存在与否将有助于诊断。

• 8三体 - 用于疑似诊断T细胞幼淋巴细胞白血病的患者，了解该拷贝数异常的存在与否将有助于诊断。诊断

- iso7q – for patients with a suspected diagnosis of hepatosplenic T cell lymphoma where knowledge of the presence or absence of this copy number abnormality will assist with diagnosis. DIAGNOSIS

- iso7q - 用于疑似诊断肝脾T细胞淋巴瘤的患者，了解该拷贝数异常的存在与否将有助于诊断。诊断

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11. Test package: Histiocytosis TP182

Histiocytosis Testing Criteria

The following testing criteria describe the genomic tests that may be used during the analysis of a patient's samples to refine diagnosis, prognosis and treatment decisions. Not all tests are necessary for every patient and will be selected on the basis of the patient's clinical presentation.

WGS tests

GT593 / M117.16 WGS Germline & Tumour - Histiocytosis
GT35 / M117.17 WGS Tumour First - Histiocytosis
GT836 / M117.18 WGS Follow-up Germline - Histiocytosis

All paediatric and TYA patients with a confirmed or suspected diagnosis of histiocytosis are eligible for WGS primarily at diagnosis. If WGS has not occurred at diagnosis, undertaking this test at relapse or if refractory to a line of treatment may be appropriate. In exceptional circumstances, repeat WGS may be indicated if there is a high probability that significant clonal evolution is likely to have occurred. DIAGNOSIS/RELASPE/REFRACTORY

Tumour and germline samples can be submitted simultaneously (i.e M117.16) or separately i.e. tumour first (M117.17) followed by a germline when/if available (M117.18). At any presentation point either M117.16 or M117.17 +/- M117.18 is permissible, however once a non-contaminated germline has been submitted (either as part of M117.16 or M117.18) this can be bioinformatically re-used as a germline for all subsequent tumour samples.

Currently WGS is performed as an addition to any eligible to any standard of care.

For adult patients with a confirmed or suspected diagnosis of histiocytosis, WGS is available under the indication M235 i.e. Exhausted standard of care testing and or treatment, providing they meet this eligibility criteria. Samples can be submitted simultaneously (M235.1) or separately i.e. tumour first (M235.2) followed by a germline when/if available (M235.3).

Targeted tests

GT69 / M117.19 High sensitivity *BRAF* V600E

- For patients with a suspected diagnosis of histiocytosis where demonstration of the *BRAF* p.(V600E) variant will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of histiocytosis where demonstration of a specific genomic abnormality would allow access to a

11. 检测套餐：组织细胞增生症TP182

组织细胞增生症检测标准

以下检测标准描述了在分析患者样本以完善诊断、预后和治疗决策时可能使用的基因组检测。并非每位患者都需要所有检测，将根据患者的临床表现进行选择。

WGS 检测

GT593 / M117.16 WGS 胚系与肿瘤 - 组织细胞增生症 GT35 / M117.17 WGS 肿瘤先测 - 组织细胞增生症 GT836 / M117.18 WGS 随访胚系 - 组织细胞增生症

所有确诊或疑似诊断为组织细胞增生症的儿童和TYA患者主要在原发诊断时符合WGS检测资格。若诊断时未进行WGS，在复发或对某一线治疗难治时进行该检测可能是合适的。在特殊情况下，若存在高度可能已发生显著克隆演化，可考虑重复WGS。诊断/复发/难治

肿瘤和胚系样本可同时提交(即M117.16)或分别提交，即先提交肿瘤样本(M117.17)，随后在可获得时提交胚系样本(M117.18)。在任何就诊时间点，M117.16或M117.17 +/- M117.18均可接受，但一旦提交了未受污染的胚系样本(作为M117.16或M117.18的一部分)，该样本可通过生物信息学方法复用于所有后续肿瘤样本的胚系对照。

目前 WGS 作为任何符合条件标准治疗的补充进行。

对于确诊或疑似诊断为组织细胞增生症的成年患者，WGS可在M235适应证下使用，即已耗尽标准诊疗检测和/或治疗，前提是符合该检测资格标准。样本可同时提交(M235.1)或分别提交，即先提交肿瘤样本(M235.2)，随后在可获得时提交胚系样本(M235.3)。

靶向检测

GT69 / M117.19 高灵敏度*BRAF* V600E

- 用于疑似诊断组织细胞增生症的患者，证明*BRAF* p.(V600E)变异将有助于诊断。诊断
- 用于已知诊断为组织细胞增生症的患者，证明特定基因组异常将允许获得

targeted treatment (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT

- For patients with a known diagnosis of histiocytosis receiving treatment where assessment of the disease response at a molecular level may influence treatment strategy (e.g. knowledge of molecular response prior to an allogeneic haematopoietic stem cell transplant). MONITORING DURING / POST-TREATMENT
- For patients with a known diagnosis of histiocytosis who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT
-

Small variant via panel

GT892 / M117.1 (NGS small variant panel)

- For patients with a suspected diagnosis of histiocytosis where demonstration of a clonal marker will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of histiocytosis where demonstration of a specific genomic abnormality would allow access to a targeted treatment (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT
- For patients with a known diagnosis of histiocytosis receiving treatment where assessment of the disease response at a molecular level may influence treatment strategy (e.g. knowledge of molecular response prior to an allogeneic haematopoietic stem cell transplant). MONITORING DURING / POST-TREATMENT
- For patients with a known diagnosis of histiocytosis who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of mutations post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Targeted structural variant via FISH

GT579 / M117.3 (*ALK* rearrangement FISH)
GT426 / M117.7 (*BRAF* rearrangement FISH)
GT456 / M117.12 (*NTRK1* rearrangement FISH)

- For patients with a suspected diagnosis of histiocytosis where demonstration of a clonal marker will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of histiocytosis where demonstration of a specific genomic abnormality would allow access to a targeted treatment (NB there should be an expectation that

靶向治疗(注：应预期在证明(或不证明)特定异常后，基因组所确定的治疗将可及)。诊断、治疗前、治疗变更前

• 用于已知诊断为组织细胞增生症且正在接受治疗的患者，在分子水平评估疾病缓解可能影响治疗策略(例如在异基因造血干细胞移植前了解分子缓解情况)。治疗期间/治疗后监测

• 用于已知诊断为组织细胞增生症且已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，在HSCT后监测基因组异常的存在，特别是在供者嵌合率<100%的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过 panel 检测小变异

GT892 / M117.1(NGS小变异panel)

• 对于疑似组织细胞增生症诊断的患者，若证实克隆性标志物有助于诊断。诊断 • 对于已知组织细胞增生症诊断的患者，若证实特定基因组异常可获得靶向治疗机会(注：应预期基因组确定的治疗将在证实(或不证实)特定异常后可用)。诊断、治疗前、治疗变更前

• 用于已知诊断为组织细胞增生症且正在接受治疗的患者，在分子水平评估疾病缓解可能影响治疗策略(例如在异基因造血干细胞移植前了解分子缓解情况)。治疗期间/治疗后监测

• 用于已知诊断为组织细胞增生症且已接受治愈性意图治疗(即异基因造血干细胞移植[HSCT])的患者，在HSCT后监测突变的存在，特别是在供者嵌合率<100%的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过 FISH 检测靶向结构变异

GT579 / M117.3(*ALK*重排FISH) GT426 / M117.7(
*BRAF*重排FISH) GT456 / M117.12(*NTRK1*重排FISH)

• 对于疑似组织细胞增生症诊断的患者，若证实克隆性标志物有助于诊断。诊断 • 对于已确诊组织细胞增生症的患者，若证实特定基因组异常可获得靶向治疗机会(注：应预期

the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT

- For patients with a known diagnosis of histiocytosis receiving treatment where assessment of the disease response at a molecular level may influence treatment strategy (e.g. knowledge of molecular response prior to an allogeneic haematopoietic stem cell transplant). MONITORING DURING / POST-TREATMENT
- For patients with a known diagnosis of histiocytosis who have received curative intent treatment (i.e. allogeneic haematopoietic stem cell transplant [HSCT]) to monitor for presence of genomic abnormalities post-HSCT, particularly in the context of <100% donor chimerism in order to inform donor lymphocyte infusion strategy. MONITORING POST-TRANSPLANT

Structural variant detection via NGS panel

GT914 / M117.2 (Multi-target NGS panel – structural variant)

- For patients with a suspected diagnosis of histiocytosis where demonstration of a clonal marker will assist in the diagnosis. DIAGNOSIS
- For patients with a known diagnosis of histiocytosis where demonstration of a specific genomic abnormality would allow access to a targeted treatment (NB there should be an expectation that the genomically-determined treatment will be available in response to the demonstration (or not) of a specific abnormality). DIAGNOSIS, PRE-TREATMENT, PRIOR TO CHANGE IN TREATMENT

基因组确定的治疗将在特定异常得到证实(或未证实)后提供。诊断、治疗前、治疗变更前

• 用于已知诊断为组织细胞增生症且正在接受治疗的患者，在分子水平评估疾病缓解可能影响治疗策略(例如在异基因造血干细胞移植前了解分子缓解情况)。治疗期间/治疗后监测

• 对于已知诊断为组织细胞增生症、已接受治愈性意向治疗(即异基因造血干细胞移植[HSCT])的患者，用于监测HSCT后基因组异常的存在，特别是在供者嵌合率<100%的情况下,以指导供者淋巴细胞输注策略。移植后监测

通过NGS panel进行结构变异检测

GT914 / M117.2(多靶点NGS panel--结构变异)

• 对于疑似诊断为组织细胞增生症的患者，证实克隆性标志物将有助于诊断。诊断

• 对于已知诊断为组织细胞增生症的患者，证实特定基因组异常将使其能够获得靶向治疗(注意：应预期基因组确定的治疗将在特定异常得到证实(或未证实)后提供)。诊断、治疗前、治疗变更前

12. Test package: Chimerism Testing TP401

Chimerism Testing

GT1368 / M118.1 STR Chimerism testing post-allogeneic haematopoietic stem cell transplant

- Individuals who have undergone an allogeneic haematopoietic stem cell transplant where determination of the level of engraftment i.e. chimerism of the bone marrow / peripheral blood, would be informative in patient management. In the absence of nationally agreed guidelines, specific cell lineage(s), sample type and timing of testing should follow locally agreed protocols. INITIAL TESTING OF HOST & DONOR PRE-TRANSPLANT / MONITORING

GT1370 / M118.2 XY chromosome FISH post-allogeneic haematopoietic stem cell transplant

- Individuals who have undergone a sex-mismatched allogeneic haematopoietic stem cell transplant where determination of the level of engraftment i.e. chimerism of the bone marrow / peripheral blood, would be informative in patient management. In the absence of nationally agreed guidelines, specific cell lineage(s), sample type and timing of testing should follow locally agreed protocols. MONITORING

GT1335 / M242.1 STR Chimerism testing post-allogeneic solid organ transplant

- Individuals who have undergone an allogeneic solid organ transplant where there is a clinical concern that there may be passenger lymphocyte syndrome i.e. evidence of lymphocytes from the donor in the host. It should be noted that this testing is not implemented routinely but rather on request from the treating clinicians. INITIAL TESTING OF HOST & DONOR PRE-TRANSPLANT (accepting that there is often not a suitable donor sample for comparative testing)/ MONITORING

GT1336 / M242.2 XY chromosome FISH post-allogeneic solid organ transplant

- Individuals who have undergone a sex-mismatched allogeneic solid organ transplant where there is a clinical concern that there may be passenger lymphocyte syndrome i.e. evidence of lymphocytes from the donor in the host. It should be noted that this testing is not implemented routinely but rather on request from the treating clinicians. MONITORING

12. 检测套餐：嵌合体检测TP401

嵌合体检测

GT1368 / M118.1 STR 异基因造血干细胞移植后嵌合状态检测

- 已接受异基因造血干细胞移植的个体，确定植入水平即骨髓/外周血嵌合状态对患者管理具有指导意义。在缺乏国家共识指南的情况下，特定细胞谱系、样本类型和检测时机应遵循当地共识方案。移植前受者与供者初始检测/监测

GT1370 / M118.2 XY染色体FISH 异基因造血干细胞移植后

- 已接受性别不匹配异基因造血干细胞移植的个体，确定植入水平即骨髓/外周血嵌合状态对患者管理具有指导意义。在缺乏国家共识指南的情况下，特定细胞谱系、样本类型和检测时机应遵循当地共识方案。监测

GT1335 / M242.1 异基因实体器官移植后 STR 嵌合体检测

- 已接受异基因实体器官移植的个体，临床担心可能存在过客淋巴细胞综合征，即宿主中存在来自供者的淋巴细胞证据。应注意该检测非常规实施，而是应治疗临床医生的要求进行。移植前受者与供者初始检测(承认通常没有合适的供者样本进行对比检测)/监测

GT1336 / M242.2 XY染色体FISH检测用于同种异体实体器官移植后

- 已接受性别不匹配异基因实体器官移植的个体，临床担心可能存在过客淋巴细胞综合征，即宿主中存在来自供者的淋巴细胞证据。应注意该检测非常规实施，而是应治疗临床医生的要求进行。监测

13. Test package: Clonality Testing TP62

Clonality Testing

GT1423 / M225.1 B-cell clonality testing multiplex sequencing
GT1408 / M225.2 B-cell clonality testing NGS

- Individuals where there is a suspicion of a clonal B cell malignancy or where otherwise demonstrating the presence or absence of clonal B cells will aid in patient management. DIAGNOSIS

GT39 / M225.3 & M111.2 T-cell clonality testing multiplex sequencing
GT508 / M225.4 & M111.3 T-cell clonality testing NGS

- Individuals where there is a suspicion of a clonal T cell malignancy or where otherwise demonstrating the presence or absence of clonal T cells will aid in patient management. DIAGNOSIS

There are certain clinical presentations e.g. T cell rich B cell lymphoma where undertaking both B and T clonality is appropriate.

13. 检测套餐：克隆性检测TP62

克隆性检测

GT1423 / M225.1 B细胞克隆性检测多重测序 GT1408 / M225.2 B细胞克隆性检测 NGS

- 怀疑克隆性B细胞恶性肿瘤的个体，或证实克隆性B细胞存在或不存在将有助于患者管理的个体。诊断

GT39 / M225.3 & M111.2 T细胞克隆性检测多重测序 GT508 / M225.4 & M111.3 T细胞克隆性检测 NGS

- 怀疑克隆性T细胞恶性肿瘤的个体，或证实克隆性T细胞存在或不存在将有助于患者管理的个体。诊断

某些临床表现，例如T细胞丰富型B细胞淋巴瘤，同时进行B细胞和T细胞克隆性检测是适当的。

14. Test package: HaemOnc Tumours - Exhausted SOC TP245

Proven or suspected haematological tumour exhausted all standard of care testing and treatment.

GT104 / M235.1 WGS Germline & Tumour - exhausted SOC
GT88 / M235.2 WGS Tumour First - exhausted SOC
GT1076 / M235.3 WGS Follow-up Germline - exhausted SOC

- All patients of any age who do not have another WGS option in the test directory who have a proven or suspected haematological tumour and have exhausted all standard of care testing and treatment are in theory eligible under this category. However, there should be a specific question that it is hoped WGS will answer and a realistic prospect that the data currently returned as part of a standard analysis could do this. Patients who are potentially eligible under this criteria should be discussed with the relevant GLH haematological malignancies medical lead or their designee in order to seek their agreement that WGS is an appropriate testing choice based on the clinical question. All patients sequenced under this indication should have data collected post-analysis as to whether the testing had an impact on patient care. DIAGNOSIS / RELAPSE/REFRACTORY

14.检测套餐：血液肿瘤肿瘤-已穷尽SOCTP245

确诊或疑似血液肿瘤已穷尽所有标准诊疗检测和治疗。

GT104 / M235.1 WGS 胚系与肿瘤 - 已穷尽SOC GT88 / M235.2 WGS 肿瘤优先 - 已穷尽SOC
GT1076 / M235.3 WGS随访生殖系--SOC已用尽

- 理论上，任何年龄的患者，只要在检测目录中没有其他 WGS 选项，且已确诊或疑似患有血液肿瘤并已用尽所有标准诊疗检测和治疗，均符合本类别的纳入条件。然而，应当存在一个希望 WGS 能够回答的具体问题，并且目前作为标准分析一部分返回的数据有现实可能性回答该问题。可能符合本标准的患者应与相关 GLH 血液恶性肿瘤医学负责人或其指定人员讨论，以征得其同意：基于临床问题，WGS 是合适的检测选择。根据本适应证进行测序的所有患者，均应在分析后收集数据，以了解该检测是否对患者诊疗产生了影响。诊断 / 复发/难治

How this file was made

This is a complete Chinese translation of NHS England's National Genomic Test Directory, haematological oncology (HaemOnc) eligibility criteria. Tables, captions, headers, footers and numbering all keep their original positions. Nothing has been cut.

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