Updating the National Genomic Test Directory

Consultation document

Contents

Purpose

1. This document sets out the proposed approach to updating the National Genomic Test Directory.

2. Stakeholders are invited to review the proposed approach and provide feedback to NHS England and NHS Improvement. Feedback is particularly invited on the following key topics:

    1. Appropriate involvement of patients and the public throughout the process;

    2. The proposed timelines for submission of applications; and

    3. Any potential impact the proposal may have on health equalities

3. Feedback on the approach should be submitted through the consultation survey via the consultation hub.

4. Feedback received via the consultation will be evaluated by NHS England and NHS Improvement and used to inform any changes required to the approach to updating the National Genomic Test Directory and the final process which will be implemented.

Context

1. The NHS Genomic Medicine Service (GMS) was established in October 2018 with the aim of providing consistent and equitable care to the country's 55 million population.

2. A central feature of the NHS GMS is the National Genomic Test Directory (Test Directory), which identifies the full range of genomic tests – from whole genome sequencing (WGS) to tests for single genes and molecular markers – that are commissioned by the NHS in England. The Test Directory sets out the technology by which tests are available and the patients who will be eligible to access a test.

3. Seven NHS Genomic Laboratory Hubs (GLHs) are responsible for delivering the testing outlined in the Test Directory across England.

4. The first version of the Test Directory was published in October 2018. It was developed from the existing provision of genomic testing in the NHS and was subject to extensive review by national clinical and scientific experts, existing genetic laboratory staff, patient and public representatives and organisations, and organisations including the British Society for Genetic Medicine and the Joint Committee for Genomics in Medicine.

5. An update to the Test Directory was published in March 2019 to simplify the format and make minor updates to testing criteria. A further minor update has been made this year to ensure the Test Directory reflects new priorities and new technologies and to make further minor updates as appropriate. Further information on this year's update is included in annex 1.

6. From the 2021/22 update to the Test Directory NHS England and NHS Improvement, supported by a Genomics Clinical Reference Group (CRG) and genomic test evaluation working groups wishes to introduce a more structured process that allows for a wider group of stakeholders to contribute to. It is proposed to update the Test Directory on an annual basis following a robust and evidence-based process. Through this process NHS England and NHS Improvement will continue to consider the potential costs and benefits of

expanding or altering the genomic testing which it commissions, ensuring that this testing is available for all patients for whom it would be of clinical benefit.

7. It is proposed to introduce the approved approach to updating the Test Directory immediately following the conclusion of this public consultation. NHS England and NHS Improvement will therefore follow the approach outlined in this document to update the Test Directory for 2021/22, however will receive applications between September and November 2020 instead of the usual timescales presented.

8. From April 2021 onwards, NHS England and NHS Improvement will follow the full approach and annual timescales to inform the update to the Test Directory to be published in April 2022.

9. The annual update for the Test Directory will achieve the following three aims:

    1. To systematically review all available genomic testing by condition to inform the definitive repertoire of tests commissioned by the NHS in England and ensure that the best value is achieved;

    2. To support ongoing evaluation of new tests and technologies to enable access to the most effective and affordable technology now and in the future, including replacing tests where appropriate; and

    3. To assess clinical utility of genomic tests and to understand the implications of the genomic testing on the end to end patient pathway.

10. This document sets out:

    1. The clinical oversight of updates to the Test Directory;

    2. The proposed process for updating the Test Directory in response to in-year policy decisions (e.g. medicines approvals);

    3. The proposed annual process for evaluating new genomic tests and applications to amend the Test Directory;

    4. The governance arrangements to sign off updated versions of the Test Directory; and

    5. How the process for updating the Test Directory will be reviewed.

Clinical Oversight

Genomics Clinical Reference Group

1. Updates to the Test Directory will be overseen by the Genomics Clinical Reference Group (CRG).

2. The Genomics CRG has been convened to support implementation of the NHS GMS. Through its professional, patient and public representation, the Genomics CRG carries out the following functions:

    1. Advising on clinical policy and strategy for genomics, including implementation of Long Term Plan commitments and future developments of the NHS GMS;

    2. Overseeing a clear and transparent process for annual review of the Test Directory; and

    3. Supporting activities to raise awareness and embedding genomics across all clinical specialties.

3. The Genomics CRG meets three times per year and review applications that are received on an ongoing basis. The CRG will make recommendations toto NHS England and NHS Improvement's Genomics Programme Board on updates to the Test Directory once per year in line with the annual process.

Genomic test evaluation working groups

4. To provide clinical and scientific support to the Test Directory updates, NHS England and NHS Improvement has recruited to three test evaluation working groups. The working groups will report into the Genomics CRG and will cover rare disease, cancer and pharmacogenomics.

5. Membership of the genomic test evaluation working groups will include scientists, clinicians, health economists, and patient and public representatives.

In year updates to the Test Directory

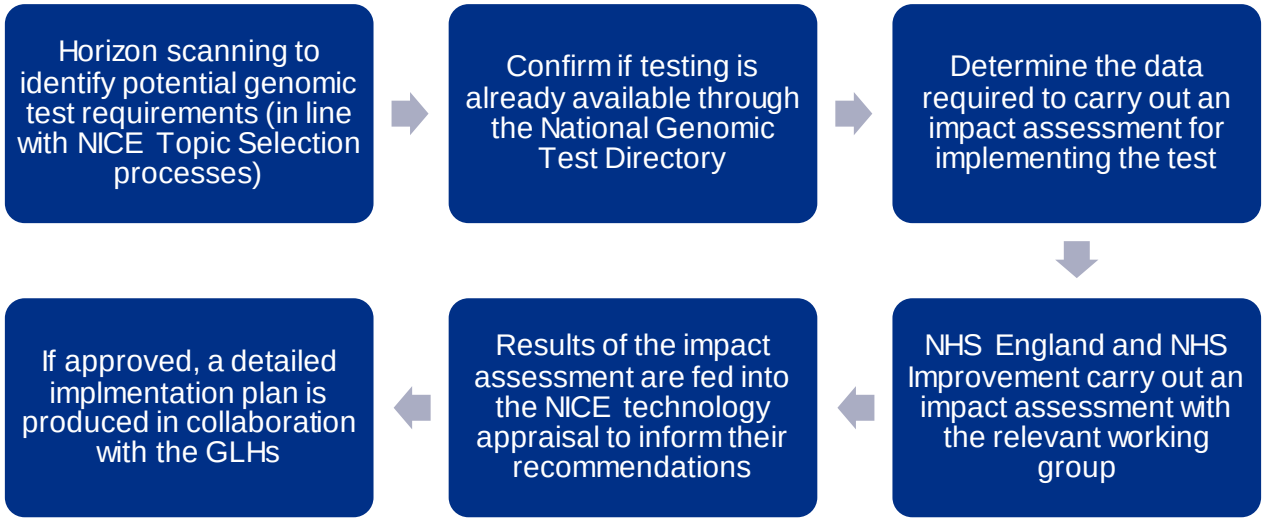
Funding decisions

1. Updates to the National Genomic Test Directory may be mandated in year by policy decisions, such as medicine approvals by the National Institute for Health and Care Excellence (NICE), where a genomic test forms part of the access criteria for a drug, or urgent policy statements issued by NHS England and NHS Improvement Specialised Commissioning, where NHS England and NHS Improvement needs to urgently implement a commissioning policy involving genomic tests. Updates may also be required as a result of the publication of NICE Diagnostics Assessment Programme (DAP) guidance.

2. Drugs and other treatments recommended by NICE through its technology appraisal programmes must be funded by the NHS through the 'funding requirement'. In most cases, when the funding requirement is applied the NHS makes the treatment available within 90 days.

3. NHS England and NHS Improvement will work closely with NICE to ensure the NHS is prepared for potential implementation of new genomic tests, and corresponding updates to the National Genomic Test Directory.

Process for informing NICE Technology Appraisals



4. NHS England and NHS Improvement are involved in the NICE Topic Selection process to identify medicines which may proceed through the NICE technology appraisal process and include a genomic test as part of the patient pathway. Where relevant medicines are identified, the impact of implementing the genomic testing element of the patient pathway will be included as part of the technology appraisal.

5. In the first instance, NHS England and NHS Improvement will confirm whether the required genomic test is already listed within the Test Directory and therefore commissioned within England.

6. If the genomic test is already commissioned, NHS England and NHS Improvement will work with the relevant test evaluation working group and the GLHs to determine the impact of any increase in activity which may occur as a result of the medicine being recommended.

7. Where the relevant genomic testing is not commissioned, NHS England and NHS Improvement will work with the genomic test evaluation working groups and the GLHs to determine the impact of implementing the testing if the medicine is recommended by NICE. Patient and Public Voice (PPV) representatives on the Genomics CRG and test evaluation working groups will ensure that the patient view is captured at this stage.

8. The outcome of the NHS England and NHS Improvement impact assessment should be submitted as per the requirements of the NICE technology appraisal guidance development process. If the impact assessment identifies that implementation within 90 days would be unachievable, an extension to the funding requirement may be requested. In these cases, NHS England and NHS Improvement will set out how long the extension is requested for and detail plans for implementation during the extended period for agreement by NICE.

9. Following the NICE recommendation of the medicine for commissioning, NHS England and NHS Improvement will work closely with GLHs to develop and follow a detailed implementation plan to ensure that testing is available within the appropriate time scale.

NICE Diagnostics Assessment Programme Guidance

10. The NICE Diagnostics Assessment Programme (DAP) advise on the formulation of NICE's guidance on diagnostic technologies and matters related to the evaluation of diagnostic technologies. The DAP develop recommendations for the NHS in accordance with NICE's published methods and processes for developing guidance on diagnostic technologies.

11. DAP guidance is developed via the diagnostics assessment process[1], involving professional and lay specialist committee members with expert knowledge of the subject under consideration. As part of the diagnostics assessment process, all guidance undergoes robust evidence review and stakeholder consultation.

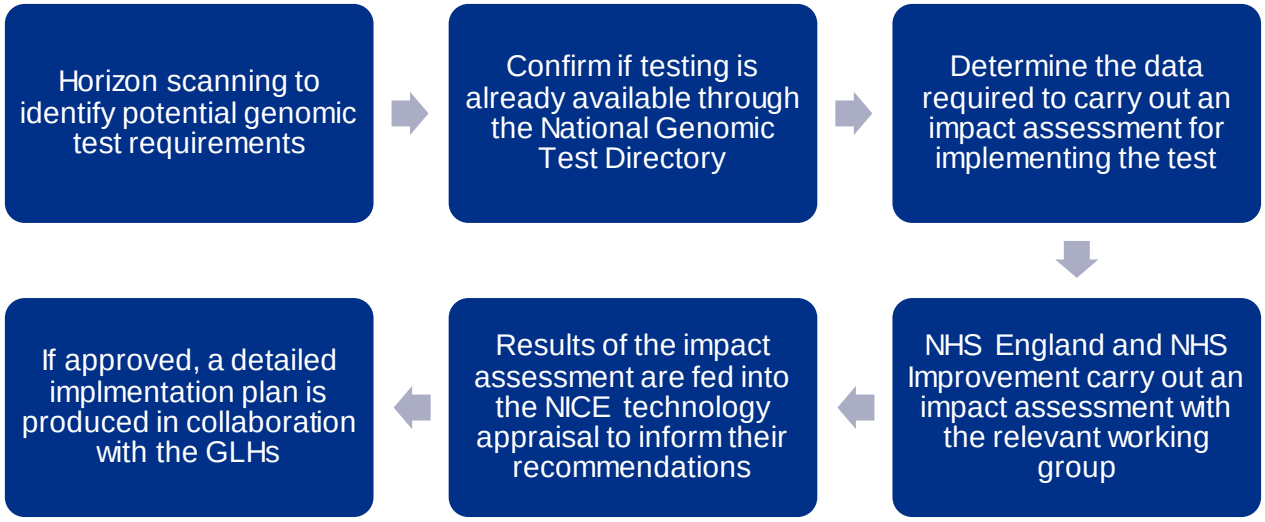
12. NHS England and NHS Improvement will work with the NICE Diagnostics programme to ensure that relevant DAC guidance involving recommendations for genomics is further reviewed and impact assessed as part of the process for updates to the Test Directory.

13. NHS England and NHS Improvement will not duplicate the robust clinical and scientific evidence review undertaken by the DAC but will review and impact assess published DAC guidance in relation to the operational, workforce and

[1] https://www.nice.org.uk/about/what-we-do/our-programmes/nice-guidance/nice-diagnostics-guidance

financial implications of implementing the proposed amendment to the Test Directory as outlined in the process for updates to the Test Directory.

Process for NHS England and NHS Improvement urgent policy statements



14. In circumstances when NHS England and NHS Improvement needs to urgently implement a commissioning policy, a policy statement or urgent policy statement will be developed. Policy statements will go through the standard development process for all policies, including evidence review, impact analysis and decision by the Clinical Priorities Advisory Group (CPAG).

15. For any policies which involve or require genomic testing, the NHS England and NHS Improvement Genomics Unit will be notified of the policy.

16. In the first instance, the Genomics Unit will confirm whether the required genomic test is already listed within the Test Directory and therefore commissioned within England.

17. If the genomic test is already commissioned, NHS England and NHS Improvement will work with the relevant test evaluation working group and the GLHs to determine the impact of any increase in activity which may occur as a result of the introduction of the policy.

18. Where the relevant genomic testing is not commissioned, NHS England and NHS Improvement will work with the genomic test evaluation working groups and the GLHs to determine the impact of implementing the testing. Patient and Public Voice (PPV) representatives on the Genomics CRG and test evaluation working groups will ensure that the patient view is captured at this stage. The outcome of the NHS England and NHS Improvement impact assessment will inform the NHS England and NHS Improvement policy implementation.

19. Following policy approval, NHS England and NHS Improvement will work closely with GLHs to develop and follow a detailed implementation plan to ensure that testing is available within the appropriate time scale.

Annual updates to the Test Directory

Annual test evaluation

1. Any proposed amendments to the Test Directory which are not mandated by a policy decision will go through an annual evaluation process to ensure that NHS commissioning of genomic tests in England is supported by the most up-to-date scientific, clinical and economic evidence.

2. The test evaluation process will be overseen by the Genomics CRG and supported by three genomic test evaluation working groups for cancer, rare disease and pharmacogenomics.

3. The formal test evaluation process will be triggered upon receipt of an application to amend the Test Directory. Applications may seek to:

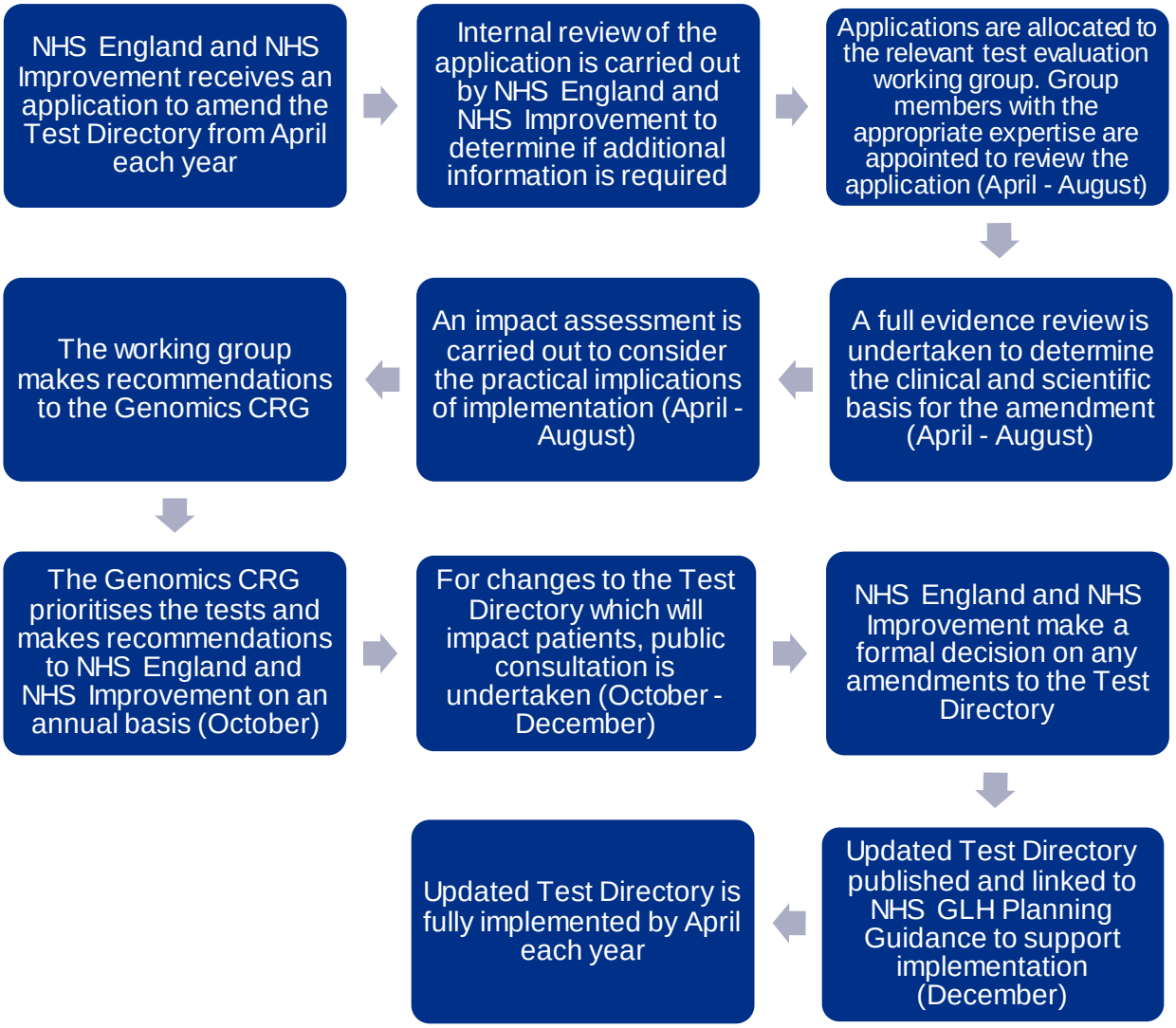
    1. Add new clinical indications to the Test Directory;
    2. Amend the eligibility criteria, constituent tests or technologies for existing clinical indications;
    3. Amend the content of gene panels
    4. Decommission a test where it is obsolete or no longer supported by the clinical scientific or economic evidence
    5. Move a clinical indication to whole genome sequencing (once this technology becomes more widely available)

4. The Test Directory will be updated on an annual basis. The annual update to the Test Directory will provide stability and clarity for the NHS GMS in implementing genomic testing and will be linked to the appropriate annual funding and commissioning processes.

5. Applications for amendments to the Test Directory must be submitted to the NHS England and NHS Improvement Genomics Unit. The window for

submitting applications will be from January to April for amendments coming into effect by April of the following year. This allows for sufficient time to comprehensively evaluate the proposed updates and ensure stakeholders are involved and engaged in decision making.

Process for annual updates



6. The NHS England and NHS Improvement Genomics Unit will carry out initial review of all applications and will request further information or clarifications where necessary. The application will then be passed to the relevant genomic test evaluation working group who will carry out a full evidence review to determine the clinical and scientific basis for the proposed amendment.

7. The genomic test evaluation working group will carry out a full review of the literature to assess the scientific and clinical basis for the amendment. If the working group supports the proposed amendment at this stage, they will proceed to impact assessment.

8. An impact assessment will be carried out in collaboration with the GLHs to consider the operational, workforce and financial implications of implementing the proposed amendment to the Test Directory, including (but not limited to):

    1. Clinical utility and analytical validity;
    2. Cost effectiveness; and
    3. Workforce required to deliver testing.

9. Evidence reviews and impact assessments will take place between April and August each year.

10. An assessment will be carried out of the impact of proposed changes to the Test Directory on patients and the public in accordance with NHS England and NHS Improvement's legal duties to involve the public in decisions around healthcare commissioning. We will follow the principles and processes outlined in the Framework for patient and public participation in specialised commissioning[2]. Where a proposed amendment is identified to have a significant impact on patients, early engagement will be carried out with relevant patient groups to ensure that their views are taken into consideration in advance of recommendations being produced.

11. Based on the evidence review, impact assessment and early patient and public engagement, the test evaluation working group will make recommendations to the Genomics CRG to support or not to support the proposed amendment to the Test Directory.

12. The proposed amendments will be collated by NHS England and NHS Improvement and a formal decision through the appropriate governance will be made on the level of patient and public consultation which will be required to support prioritisation. Where amendments are limited to technical updates, a formal consultation may not be required. However, where the proposed

[2] www.england.nhs.uk/wp-content/uploads/2017/01/specialised-participation-frmwrk.pdf

amendments are likely to have a significant impact on patients, NHS England and NHS Improvement may consider a 30, 60, or 90-day public consultation.

13. The Genomics CRG will receive the outputs from any public consultation, along with the recommendations from the test evaluation working groups. After confirming that all processes have been followed correctly, the Genomics CRG will prioritise the proposals to develop recommendations on how the budget for updating the Test Directory should be spent.

14. The process for prioritising amendments to the Test Directory will be developed in collaboration with the Genomics CRG and will be based on the following principles:

    1. Proposed amendments are assigned a score by the test evaluation working groups based on assessment across several domains (see annex 2 for further details of the test evaluation group evaluation and scoring), including:

        (1) Evidence of patient benefit and clinical utility, including if the amendment meets an unmet clinical need;

        (2) Strength of scientific and clinical evidence supporting the amendment;

        (3) Financial implication of implementation (cost saving, cost neutral, cost pressure, level of investment required);

        (4) GLH resource required to implement the amendment and technical feasibility of implementation;

        (5) Confidence that benefits to patients and clinical pathways would be realised upon implementation; and

        (6) Alignment to an NHS England and NHS Improvement clinical priority area

    2. Scoring allows proposed amendments to be compared and prioritised; and

    3. Clear and transparent process for allocating NHS spending. Funding is will be allocated based on scoring, starting from highest scoring amendments.

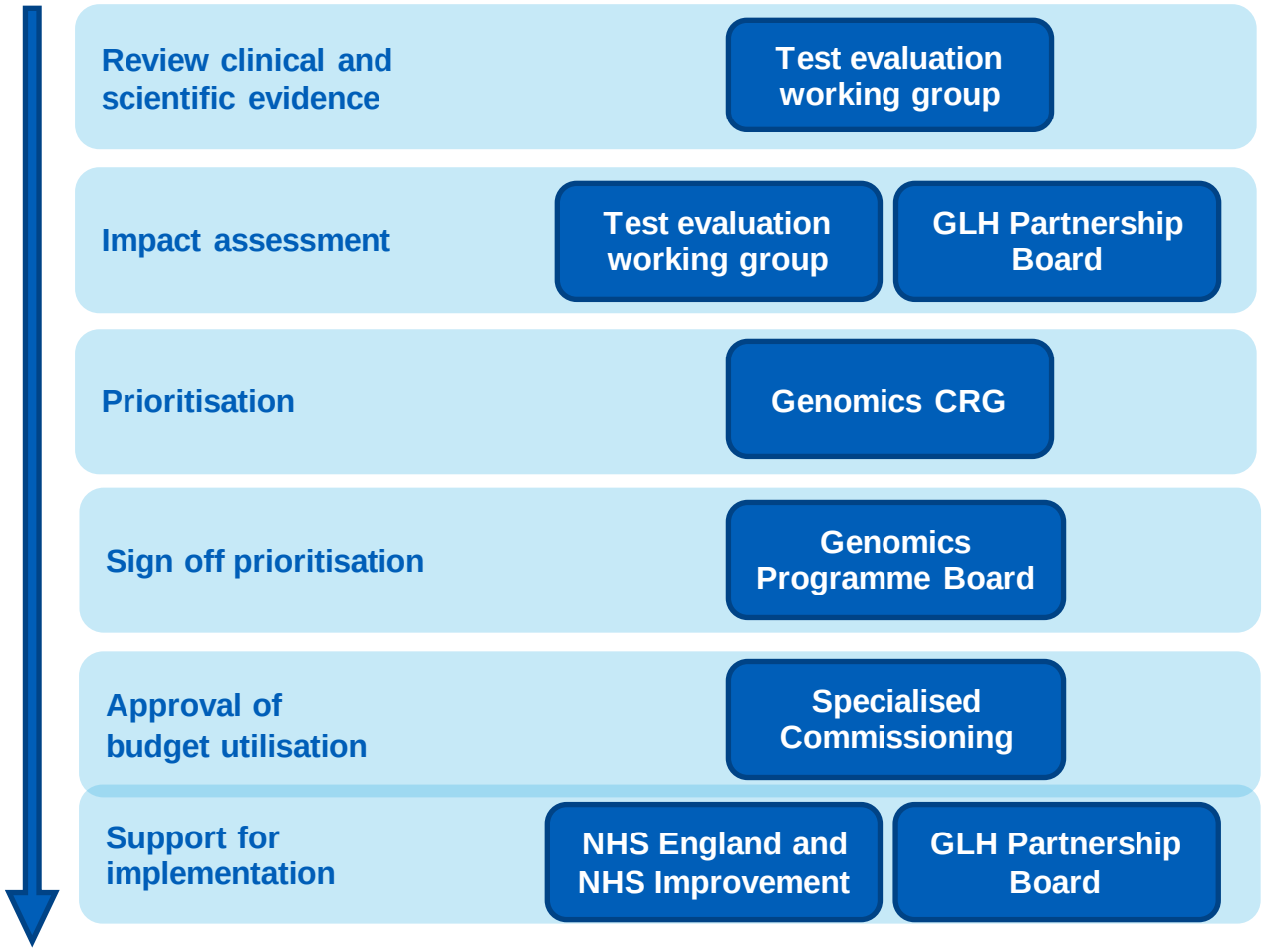
15. The Genomics CRG will make recommendations to the Genomics Programme Board in November each year.

Governance and funding

1. NHS England and NHS improvement will set aside a portion of its budget each year to pay for implementation of new tests or implementing other changes to the Test Directory. The total funding envelope for updating the Test Directory will be determined on an annual basis taking into account the spending from the previous and other spending commitments within the year. The funding envelope will be allocated to the proposed amendments based on scoring, starting from highest scoring amendments.

2. Genomics CRG will prioritise tests and make recommendations to the Genomics Programme Board who confirm that the correct process has been followed and confirm their support for the Genomic CRG's recommendations.

3. Approval to utilise the Test Directory updates budget will be acquired through annual Specialised Commissioning planning processes.



Future development of the Test Directory

1. The Genomics CRG and genomic test evaluation working groups will assess the operation of the approach to updating the Test Directory. Particular areas for review will include:

    1. Whether the annual process was successful in supporting development of an update version of the Test Directory;

    2. Reflections on the time allocated for each stage of the review process and whether this was appropriate or should be revised; and

    3. Whether any aspects of the review process should be amended to ensure that it runs smoothly.

2. Any potential changes to the annual review process will be considered by the Genomics CRG in collaboration with NHS England and NHS Improvement.

3. The Genomics CRG and genomic test evaluation working groups will continue to assess the success of the annual review process each year to ensure that the process continues to support development of a Test Directory which reflects the most recent scientific, clinical and economic evidence.

Annex 1 - Updates to the National Genomic Test Directory for 2020/21

1. The National Genomic Test Directory (the Test Directory) sets out the genomic tests which are commissioned by NHS England and NHS Improvement, the technology platform by which the testing will be delivered, and the patients who will be eligible to access to a test. The current version of the Test Directory was published on the NHS website in March 2019.

2. Following the consultation outlined by this document, NHS England and NHS Improvement will introduce a clear and transparent process for annual review of the Test Directory during to underpin the Test Directory update for April 2021 and beyond.

3. Whilst work is underway to establish this longer term process, and due to the impacts of the COVID-19 response, NHS England and NHS Improvement have worked with the test evaluation working groups and Genomics CRG through an interim process to make a number of amendments to the Test Directory for implementation during 2020/21, to ensure the Test Directory reflects new priorities and new technologies. These amendments are either linked to wider policy workstreams or require limited resources to implement.

4. Amendments to this version of the Test Directory have been collated through two routes:

    1. a review of the content of large cancer panels; and

    2. a review of proposed amendments which have been identified and reported to NHS England and NHS Improvement since first publication of the Test Directory.

5. For cancer, the proposed amendments were developed through formal review of large cancer panels and evidence collation for proposed changes to gene targets.

6. For rare and inherited disease, proposed amendments which were reported to NHS England and NHS Improvement from stakeholders including GLHs, NHS Genomic Medicine Centres (GMCs), specialist test working groups, and individuals were assessed to identify those which could be implemented with relatively low impact to GLHs during this financial year.

7. Additional updates linked to wider policy work streams were prioritised for inclusion in this version of the Test Directory, including the addition of *NTRK* gene fusion testing and *DPYD* testing.

8. The proposed amendments for cancer and rare and inherited disease were reviewed by the genomic test evaluation working groups in May 2020. Recommendations from the genomic test evaluation working groups were made to the Genomics Clinical Reference Group (CRG) in June 2020, who reviewed and endorsed the proposed amendments to the Genomics Programme Board.

9. The Genomics Programme Board approved the updates to the Test Directory in June 2020. The updated Test Directory is expected to be published shortly.

Annex 2 - Evaluation and scoring of amendments to the Test Directory

Evaluation and scoring of amendments to existing clinical indications in the Test Directory

Test Evaluation for an Amendment to an Existing Clinical Indication - Scoring
The following scoring system is in place for Test Evaluation Working Group member evaluations:
SCORING (1-5):
5 - strong evidence and case for implementation provided
4 - relatively strong evidence and case for implementation provided
3 - moderate evidence and case for implementation provided
2 - relatively weak evidence and case for implementation provided
1 - weak evidence and case for implementation provided

OUTCOMES (max. score 30):	Score
Weak case for implementation	<15
Moderate case for implementation	15 to 20
Strong case for implementation	>20

Evaluation Criteria		
Clinical Utility	**Score (1-5)**	**Comments**
Positive impact on clinical outcomes and benefit to patient	--select--	
Minimal impact on existing testing pathways	--select--	
Minimal impact on existing clinical pathways	--select--	
Low impact on existing activity figures/testing volumes	--select--	
Low financial impact	--select--	
Limited laboratory operational impact	--select--	
Total Score	0	
Outcomes	**Test Evaluation Group Member Response**	
Change accepted?	--select--	
Recommended for discussion at Test Evaluation Group?	--select--	
Reason for discussion at Test Evaluation Group (e.g. new area not currently included in the Test Directory, emerging scientific evidence)		
If appropriate, please note any legal, ethical or social implications of this application you would like the Test Evaluation Group to be aware of		
Other comments		

Evaluation and scoring of new clinical indications proposed to be added to the Test Directory

Test Evaluation for a New Clinical Indication - Scoring
The following scoring system is in place for Test Evaluation Working Group member evaluations:
SCORING (1-5):
5 - strong evidence and case for implementation provided
4 - relatively strong evidence and case for implementation provided
3 - moderate evidence and case for implementation provided
2 - relatively weak evidence and case for implementation provided
1 - weak evidence and case for implementation provided

OUTCOMES (max. score 45):	Score
Weak case for implementation	0 to 20
Moderate case for implementation	21 to 30
Strong case for implementation	>31

Evaluation Criteria		
Clinical Utility	**Score (1-5)**	**Comments**
Evidence of clinical utility and patient benefit	--select--	
Evidence of unmet diagnostic need	--select--	
Strength of scientific evidence base	--select--	
Health Economic Case	**Score (1-5)**	**Comments**
Low level of additional investment required	--select--	
Cost effectiveness	--select--	
Evidence of appropriate diagnostic yield	--select--	
NHS Implementation	**Score (1-5)**	**Comments**
Aligns to an NHS England and NHS Improvement clinical priority area	--select--	
Practicality of implementation in the GMS	--select--	
Technical feasibility	--select--	
Overall Score:	0	
Test method	**Y/N**	**Comments**
Do you have any concerns over the test method proposed in the application? If yes, please provide details	--select--	
Are there opportunities for the generation of further evidence to support commissioning decisions? (e.g. WGS for high cost drugs)	--select--	
Evaluation Outcomes	**Test Evaluation Group Member Response**	
Recommended for GMS implementation?	--select--	
Proposed eligibility criteria accepted?	--select--	
Recommended for discussion at Test Evaluation Group?	--select--	
Reason for discussion at Test Evaluation Group		
If appropriate, please note any legal, ethical or social implications of this application you would like the Test Evaluation Group to be aware of		
Other comments		

Please note: *the scoring provided as part of this review is indicative to guide the Test Evaluation Working Group and Genomics CRG decision only. Scoring alone will not mean an automatic accept/rejection of any applications.*