



**Wessex**  
Cancer Alliance

# Genomics update

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# Report to the Wessex Cancer Alliance Board

|   |   |                 |                     |                    |
|---|---|-----------------|---------------------|--------------------|
| <b>Title:</b>   | Genomics update   |                 |                     |                    |
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| <b>Date:</b>  | 22/09/2021  |                 |                     |                    |
| <b>Purpose</b>  | <b>Assurance or reassurance</b><br>Y  | <b>Approval</b> | <b>Ratification</b> | <b>Information</b> |
| <b>Summary of paper:</b>  | <p>This briefing sets out an overview of the progress of genomics services across Wessex against the ambitions outlined in the NHS Long Term Plan and operationalised in the Cancer Plan for Wessex 2019-2024.</p> <p>Our ambitions for 2021/22 are as follows:</p> <ol style="list-style-type: none"> <li>1. Roll out streamlined Lynch syndrome testing across all Wessex through for both colorectal and endometrial cancer pathways</li> <li>2. Whole genome sequencing, working with GMSA to ensure equitable access. Initial focus on children with cancer and patients of any age with sarcoma and leukaemia</li> <li>3. BRCA mainstreaming – embedding ovarian testing pathway and mainstreaming breast pathway in at least two local trusts</li> </ol> |                 |                     |                    |
| <b>Implications: (Clinical, Organisational, Governance, Legal?)</b> | This briefing has clinical implications as all patients should have access to genomic testing where appropriate   |                 |                     |                    |
| <b>Key risks and mitigations:</b>                                   | <p>Genomics may not be a priority for already overstretched frontline clinicians</p> <p>Pressures on laboratory staff with increase in testing and no concomitant increase in funding/capacity.</p> <p>Need to agree an effective and efficient way to resource additional testing.</p>   |                 |                     |                    |
| <b>Summary: Conclusion and/or recommendation</b>                    | <ul style="list-style-type: none"> <li>• Increase uptake of whole genome service, including additional indications</li> <li>• Continue to work with GMSA to deliver projects, particularly implementation of universal Lynch syndrome testing and pathology transformation</li> <li>• Establish laboratory and clinical strategy and workforce plan for Wessex genomics service</li> </ul>  |                 |                     |                    |



# GMSA: National strategic priorities (cancer)



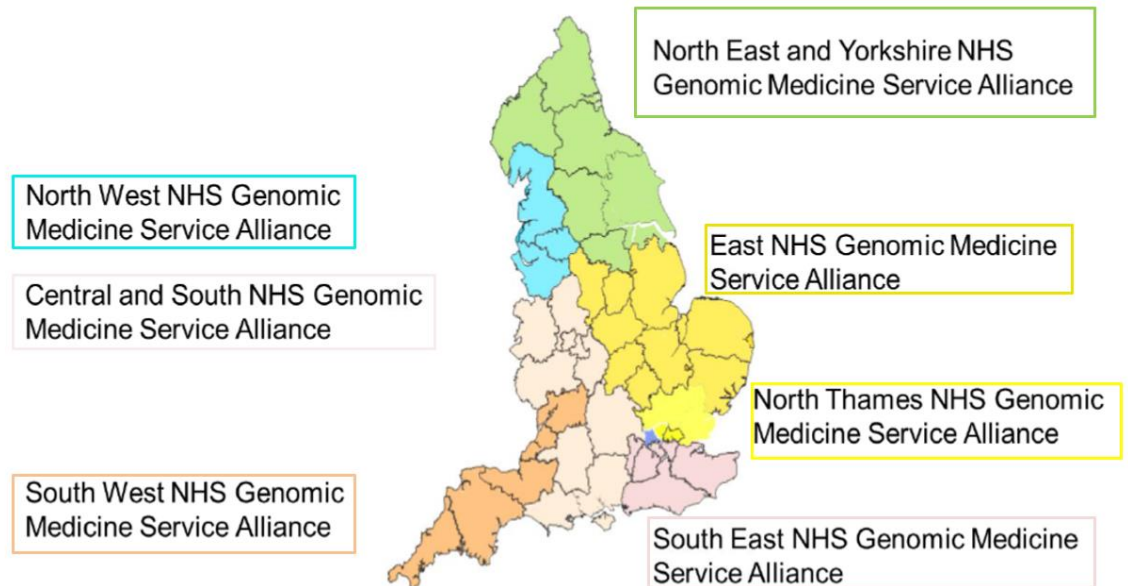
- **Pharmacogenomics:** Equitable and timely access to *DPYD*\* genotyping for prediction of fluoropyrimidine chemotherapy toxicity as an exemplar service
- **Lynch syndrome pathways:** Embedding testing for all patients newly diagnosed with colorectal and endometrial cancer
- **Nursing and midwifery genomics education** and development
- **Cellular pathology transformation** to facilitate genomic testing
- **100,000 genome project legacy work** and establishing **clinical whole genome sequencing (WGS) service**

\*DPYD = dihydropyrimidine dehydrogenase, an enzyme critical to the metabolism of certain chemotherapy drugs

# Ambitions delivered through...

- 7 Genomic Laboratory Hubs (GLHs) contracted by NHS E/I to coordinate a network of sub-contracted local genomic laboratories
- 7 Genomic Medicine Service Alliances to drive necessary changes to clinical services

- Lab network delivers standard repertoire of tests\* from:
  - Rare disease test directory
  - Cancer test directory
- Tests commissioned through specific laboratories
- NHS funding to GLHs for everything in test directories including WGS
- At least annual updates to genomic test directories



\*<https://www.england.nhs.uk/publication/national-genomic-test-directories/>



## National and local ambitions for genomics



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NHS Long Term Plan: *“to extend the use of molecular diagnostics and, over the next ten years, routinely offer genomic testing to all people with cancer for whom it would be of clinical benefit and expand participation in research.”*

### Wessex ambitions 21/22:

1. Roll out streamlined Lynch syndrome testing across all Wessex for both colorectal and endometrial cancer pathways
2. Whole genome sequencing, working with GMSA to ensure equitable access. Initial focus on children with cancer and patients of any age with sarcoma and leukaemia
3. BRCA mainstreaming – embedding ovarian testing pathway and mainstreaming breast pathway in at least two local trusts



# 1. Lynch Syndrome

## Lynch Syndrome:

- Under-diagnosed, inherited cancer predisposition syndrome
- NICE diagnostic algorithm to increase detection by testing tumour samples from all patients with a new diagnosis of colorectal cancer (2017) and endometrial cancer (2020)
- Requires sequence of immunohistochemistry and molecular tests



# Lynch Syndrome

- Good Wessex participation rates in national survey of colorectal and endometrial MDT members to determine baseline testing position
- Plan to take forward info in discussions with trusts to achieve universal testing coverage
- NHSE funding to support testing received and in process of distributing equal share to regional cellular pathology departments

| Trust | Colorectal response | Endometrial response | Universal LS screening colorectal | Universal LS screening endometrial |
|-------|---------------------|----------------------|-----------------------------------|------------------------------------|
| HHFT  | Y                   | Y                    | Y                                 | Y                                  |
| UHS   | Y                   | Y                    | Y                                 | Y                                  |
| QAH   | Y                   | N                    | Y                                 | ?                                  |
| IOW   | Y                   | Y                    | Y                                 | N                                  |
| UHD   | N                   | Y                    | ?                                 | N                                  |
| DCH   | N                   | Y                    | ?                                 | Y                                  |

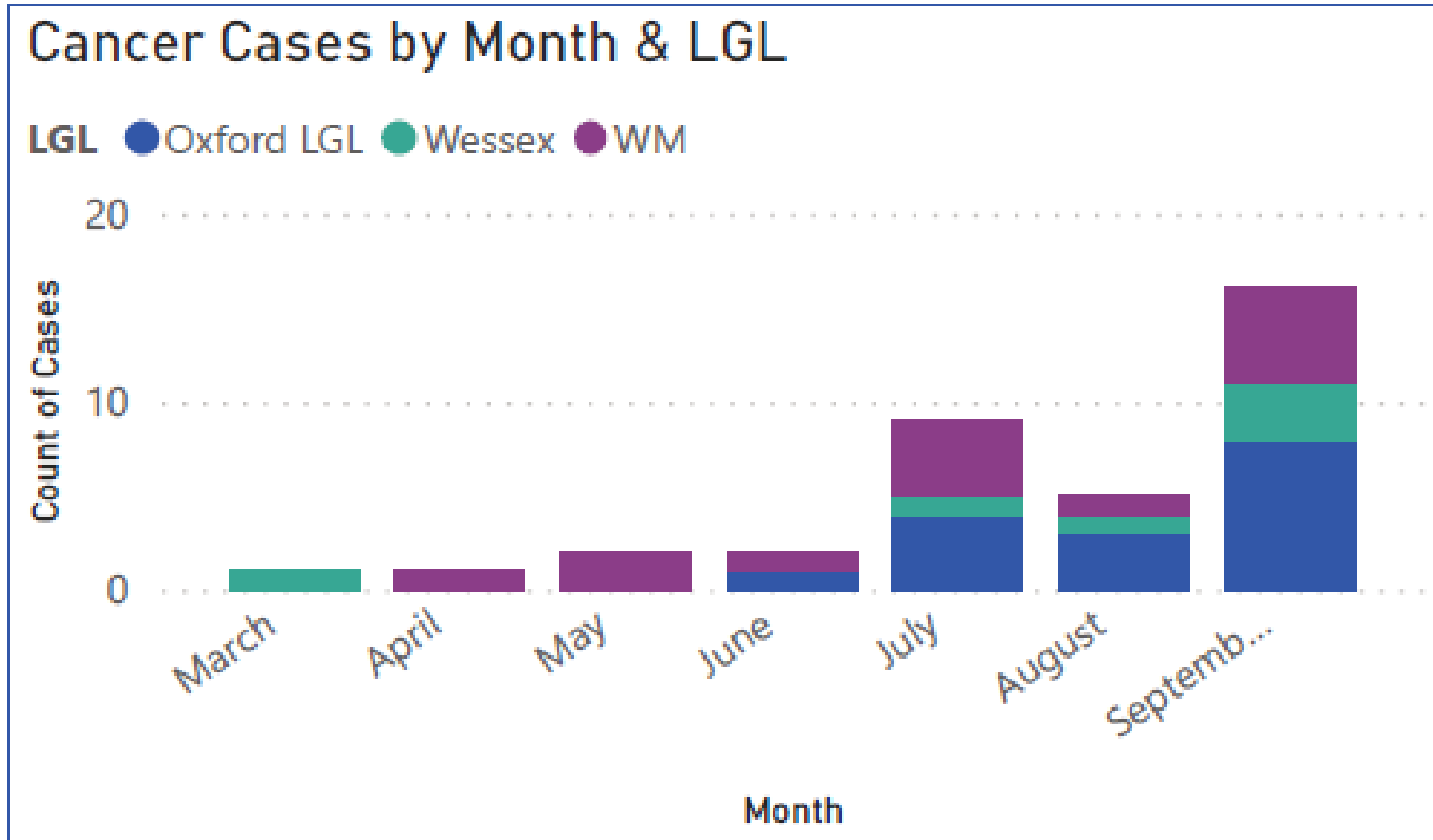


## 2. Whole genome sequencing

- Strategic priority for Alliance, particularly paediatric cancer due to provision of information about the cause of the cancer and how to treat it, potentially improving outcomes for patients with targeted treatments
- Pathways and processes for paediatric cancer, sarcoma and leukaemia have been established at UHS with some regional discussions involving other trusts
- Jan 2021 Live clinical testing started at Salisbury and Southampton
- First leukaemia sample submitted for whole genome sequencing on 19/02/21
- Four cases completed and reviewed at Wessex genomic tumour advisory board to date



# Progress: whole genome sequencing





### 3. **BRCA** mainstreaming

- Mutations in the *BRCA1/2* genes result in an increased chance of developing breast and ovarian cancer
- *BRCA1/2* tumour/ germline mutation analysis can also inform PARP inhibitor therapy\* eligibility in ovarian, breast and prostate cancer
- Mainstreaming is where the clinical team who know the patient can counsel, consent and give results directly to patients
- Ovarian pathway rolled out through Wessex and service evaluation underway
- Breast pathway in the process of being rolled out
- Collaboration with University of Southampton Macmillan Survivorship Research Group on evaluation of online BRCA decision aid

\* A type of targeted therapy



# Workforce and strategy



- Feb 21 new genomic tumour advisory board coordinator role with pump prime funding of £30K from Wessex Cancer Alliance
- 2x1 year posts to support Lynch syndrome pathway development across Wessex: B7 coordinator and B4 pathway coordinator; B7 appointed (start Jan 2022)
- GMSA project manager in post for Wessex
- Pump prime funding for whole genome sequencing from NHSE via GLH/GMSA being used to support staffing resource
- Oct 21 Wessex Genomics Strategy Steering group reinstated
- Nov 21 Business case for unified Wessex genomics laboratory service



## Training and education



- Ongoing ovarian mainstreaming training with healthcare professionals; HEE competency framework included in training and webinar being finalised
- Breast mainstreaming training for healthcare professionals (UHS, Salisbury, HHFT, IoW, Guernsey clinicians so far)
- Lynch training for colorectal clinical nurse specialists has been rolled out



## Next steps

- Increase uptake of whole genome service now processes in place
- Facilitate additional WGS indications expected to go live in November 2021:
  - change in paediatric age range to incorporate whole TYA population; increase from 19 to 25 years at diagnosis
  - any patient with a solid or haematological tumour where standard of care testing/ treatment options have been exhausted
  - all patients with central nervous system tumours
  - pilots in patients with high-grade serous ovarian carcinoma and triple negative breast cancer
- Continue to support GMSA projects, particularly implementation of universal Lynch syndrome testing and pathology transformation
- Establish laboratory and clinical strategy and workforce plan for Wessex genomics service