North Thames
Genomic Medicine Service

Bitesize basics – How genomic testing is changing in primary care

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Go to our **WEBSITE** for up-to-date genetic testing information.

This is one segment of an eight-part bitesize basics learning series for Primary Care, focusing on ordering CF carrier testing:

1. How genetic testing is changing in Primary Care
2. What is CF carrier testing?
3. Should I order a CF carrier test?
4. Is ethnicity important in CF carrier testing?
5. Consenting for CF carrier testing
6. How to order a CF carrier test?
7. What do I do with a CF carrier test result?
8. Genetics quiz – CF carrier testing

Genetic testing & Primary Care

Advances are being made in the field of genetic medicine. They are having an impact in primary care.

As the gateway to the NHS, primary care practitioners are vital for:

- The early identification of genetic issues,
- Clinical management of genetic conditions,
- Communicating genetic information.

Which genetic tests can be ordered in primary care?

At the time of publishing, three genetic tests can NOW be ordered in primary care:

- **R185 Cystic fibrosis carrier testing**
- **R191 Alpha-1-antitrypsin deficiency**
- **R195 Iron overload – hereditary haemochromatosis testing**

Eligibility criteria and who can order genetic tests in England are given in the NHS National Genomic Test Directory.

https://www.england.nhs.uk/publication/national-genomic-test-directories/

Information is correct as of 27 May 2024. Refer to the National Genomic Test Directory for the most current list of available tests, as this information is subject to change.

8 ways genetics is impacting Primary Care practices

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1. Personalised medicine

Genetic testing can provide valuable insights into an individual's genetic makeup, allowing healthcare providers to tailor treatment and preventive strategies to a patient's unique genetic profile.

This can lead to more effective and personalised healthcare.

2. Disease Risk Assessment

Genetic testing can help identify an individual's susceptibility to certain diseases or conditions, such as hereditary cancers (e.g., *BRCA* mutations), cardiovascular diseases, and neurodegenerative disorders.

This information can guide early interventions and preventive measures.

3. Medication Response

Some genetic tests can predict how a patient may respond to certain medications, helping healthcare providers choose the most appropriate drugs and dosages while minimising adverse effects.

4. Family History Assessment

Genetic testing can complement family history information by providing concrete genetic data that may not be evident from clinical history alone.

This can be particularly useful in cases where family history is incomplete or unknown.

5. Carrier Screening

Genetic testing can identify carriers of recessive genetic disorders, such as cystic fibrosis or sickle cell anaemia, enabling informed family planning decisions and early intervention for affected offspring.

6. Prenatal Screening

Genetic testing can be used for prenatal screening to detect chromosomal abnormalities or specific genetic conditions in the developing fetus.

This information can guide decision-making during pregnancy.

7. Risk Reduction and Prevention

By identifying genetic risk factors, patients can take proactive steps to reduce their risk of developing certain diseases. For example, individuals at high risk for familial hypercholesterolemia can adopt lifestyle changes and receive early treatment to prevent cardiovascular problems.

8. Research and Clinical Trials

Genetic data collected through primary care can contribute to research on the genetics of diseases and facilitate patient participation in clinical trials based on their genetic profiles.

Contacts and information

Contact us, email: nt-gmsa@gosh.nhs.uk

Visit our website: https://norththamesgenomics.nhs.uk

For a more comprehensive understanding of the information provided in this bitesize resource, please download from our website the *CF Carrier Testing Toolkit*

Coming soon! Our *CF Carrier Testing* module video.

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