BSGM The British Society for Genetic Medicine

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ROYAL PHARMACEUTICAL SOCIETY

BRITISH PHARMACOLOGICAL SOCIETY

UK Clinical Pharmacy Association

Direct-to-consumer genomic testing

Position statement

September 2025

Developed by the DTC-GT Working Group and the pharmacogenetic subgroup

About this statement

This position statement on direct-to-consumer genomic testing (DTC-GT) is intended to support healthcare professionals working in primary care and community settings.

Citation for this document

British Society for Genetic Medicine, Royal College of General Practitioners, Royal Pharmaceutical Society, British Pharmacological Society, UK Clinical Pharmacy Association. *Direct-to-consumer genomic testing: Position statement.* September 2025. London: BSGM, 2025.

Review date: 2028

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Acknowledgements

This updated statement was developed by the DTC-GT Working Group and the pharmacogenetic subgroup. These included patient representatives (PPI), patient group representatives, and members of the British Society for Genetic Medicine (BSGM), the Royal College of General Practitioners (RCGP), the Royal Pharmaceutical Society (RPS), the British Pharmacological Society (BPS), the UK Clinical Pharmacy Association (UKCPA) and academic experts.

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We are grateful for comments and clarifications provided by the UK Cancer Genetics Group (CGG) and the Prenatal Genomics Group (PGG) and for the administrative support provided by Salim Ahmed.

Contents

Introduction

This position statement is primarily aimed at healthcare professionals working in primary care and community settings. It covers direct-to-consumer genomic* testing (DTC-GT), which can describe any genomic test that has been sought and paid for outside routine NHS care, whether purchased online or via a commercial provider. Companies that offer DTC-GT are expanding their activities in the UK and globally. Alongside this, people may consider DTC-GT because of potential issues in accessing genomic testing through local NHS services. In addition, DTC-GT can offer people more choice, enhanced accessibility and greater privacy.

Patients may ask healthcare professionals in an NHS setting, or those delivering services on behalf of the NHS, to interpret the results of DTC-GT. There is considerable variation in the quality and validity of DTC-GT and healthcare professionals should exercise caution if asked to interpret such results or to use them as part of clinical decision making.

People with a personal or family history of a health condition should be aware that DTC-GT is not a substitute for a broader assessment by a healthcare professional. Individuals remain entitled to NHS care, such as family history and risk assessment, healthy lifestyle advice, and/or referral to specialist care, regardless of their DTC-GT result.[1]

Note that terms underlined in grey text are clickable links to a glossary at the end of the document, which displays the linked term at the top of the page.

* Genomic testing is used here as an umbrella term and includes genetic testing and pharmacogenetic testing.

Key recommendations

- Healthcare professionals should not take at face value, or attempt to interpret, reports from non-accredited laboratories or third-party interpretation services.†
- Patients presenting with DTC-GT results to healthcare professionals should be offered the same NHS care as those who have not had DTC-GT.
- DTC-GT results should not affect access to treatment(s) that are not commissioned for that indication within the NHS.

We also note the recommendations in the House of Commons Science and Technology Committee's 2021 report on direct-to-consumer genomic testing,[2] in particular:

> The Government should require manufacturers of direct-to-consumer genomic tests to have the performance of their tests assessed by an external body prior to placing their products on the UK market. (Paragraph 41)

> The Government should extend the scope of the performance requirements on direct-to-consumer genomic tests to explicitly cover clinical performance as well as analytical performance. (Paragraph 47)

> In addition to pre-market validation of direct-to-consumer tests, the Government should consider requiring companies offering such tests to regularly update the evidence submitted to the external validation body, and for that body to review this, for example on an annual basis. (Paragraph 53)

> The Government should consider extending the definition of products covered by the regulation of genomic tests to include software and other services offering analysis and interpretation of genomic test results obtained from third parties. (Paragraph 104)

† There are a range of DTC tests and products, and these may be accredited or approved for market through various means. For example, products might have a UKCA or CE mark. Laboratories may be accredited to standards set by the International Organisation for Standardization (ISO), by organisations such as the United Kingdom Accreditation Service, member organisations of European Accreditation, the College of American Pathologists and the United States Accreditation Board.

Additional recommendations for DTC-GT results for single gene conditions or predispositions

> Where a patient has a personal or family history indicating the need for genomic testing, they should be referred via appropriate pathways regardless of their DTC-GT result.

> If a patient has had DTC-GT and is reported to have a disease-predisposing variant in a gene for which NHS testing is offered (e.g. *BRCA1/2*), or both members of a couple are found to be carriers for a genetic condition, healthcare professionals should contact their local regional NHS genetics clinic to discuss whether a referral is appropriate.

> Where a DTC-GT has been performed in an accredited laboratory[†] with robust external quality assurance,[‡] advice should be sought from regional genetics services to enable robust clinical interpretation of findings.

> If a patient wants to discuss a DTC-GT result for which NHS genomic testing is not offered (e.g. paternity tests or ancestry information), they should be signposted back to the commercial DTC provider.

Additional recommendations for DTC-GT pharmacogenetic testing results

> Where a DTC-GT has been performed in an accredited laboratory[†] with robust external quality assurance,[‡] pharmacogenetic results may be used to support a decision regarding choice of medicine for an individual patient.

> Particular caution about pharmacogenetic results should be exercised in the absence of clinical guidance concerning the relevant gene-drug pair.

> Consensus pharmacogenetic prescribing guidance should be agreed upon for the UK.

Additional recommendations for DTC-GT tumour sample/circulating tumour DNA (ctDNA) results

> ctDNA tests should not yet be used as a cancer screening test for asymptomatic people.

[†] There are a range of DTC tests and products, and these may be accredited or approved for market through various means. For example, products might have a UKCA or CE mark. Laboratories may be accredited to standards set by the International Organisation for Standardization (ISO), by organisations such as the United Kingdom Accreditation Service, member organisations of European Accreditation, the College of American Pathologists and the United States Accreditation Board.
[‡] Provided by, for example, UK NEQAS.

Standards of professional practice and resources supporting these recommendations

- Prescribers should ensure that the choice of medicine is in accordance with locally agreed formularies, pathways and guidelines.
- Where appropriate, local procedures for non-formulary, off-label or unlicensed use of medicines should be followed.
- Information may be available from trusted sources such as MHRA Drug Safety alerts, Summaries of Product Characteristics (SmPCs) and the British National Formulary (BNF).
- The Clinical Pharmacogenetics Implementation Consortium (CPIC)[3] and Dutch Pharmacogenetic Working Group (DPWG)[4] both provide peer-reviewed prescribing guidelines, which are used internationally.

Background

The analytical validity, clinical validity and clinical utility of DTC-GT may be lower than consumers perceive it to be and lower than would be acceptable within the NHS. There is also a significant risk of a false positive result or a false negative result.

People who undergo DTC-GT can receive raw (uninterpreted) DTC-GT data. They may want to find out more by submitting the data to a third-party genomic interpretation (TPI) service. However, many TPI services provide false positive results and misinterpret variants.[5–7]

Differences between patients' and healthcare professionals' understanding of the accuracy, validity and value of health risk information derived from DTC-GT and TPI services can have a detrimental effect on the relationship between the consumer-as-patient and healthcare professionals.[8]

The types of DTC-GT available include:

1. **Variants giving information regarding single gene conditions or predispositions**: e.g. variants in the breast and ovarian cancer predisposition genes (*BRCA1* and *BRCA2*). Interpreting how/if genomic variants impact a person's health or risk of developing a condition requires contextual information such as clinical signs, symptoms and/or family history. DTC-GT can produce false positive or false negative results.

   False positive results: most companies in operation in 2025 use single nucleotide polymorphism (SNP) array technology for DTC-GT. These are highly likely to identify rare variants incorrectly.[9,10] Most rare variants identified as disease causing will be false positive, i.e. will suggest that a person has a high risk when they do not. False positive results could lead to:

   - people being needlessly distressed
   - people considering unnecessary interventions such as risk-reducing surgery
   - unnecessary referrals placing inappropriate demands on stretched NHS services and resources.

   False negative results: most genes have many different disease-predisposing variants within them. Single nucleotide polymorphism (SNP) array technology generally has a low false negative rate; but many individually rare variants are not included and will therefore be missed.[10] False negative results mean that people are likely to be inappropriately reassured about their risks, which could result in:

   - people being less likely to seek guidance from NHS healthcare professionals even when there is a need to because of a strong family history or concerning symptoms
   - people deciding they do not need to start/continue with regular physical checkups and screening.

2. **Pharmacogenetic variants**: these give information about an individual's predicted response to a medicine, including the likelihood of treatment being effective and the risk of an adverse drug reaction. Most pharmacogenetic testing is performed using a small

panel of genes and looks for variants known to impact the response to medicines someone is given. This means that false positive results may be less likely than for some other types of DTC-GT. But, if the gene panel does not cover certain genes or certain variants, then a false negative result is possible; this is particularly important when considering the common but complex structural variation in some pharmacogenes, e.g. *CYP2D6.* The interpretation of pharmacogenetic test results is complicated because it can vary depending on the clinical situation and the medicine being prescribed. Where the prescribing guidance is taken from and how it is interpreted also needs to be considered.

3. **Testing of circulating tumour DNA (ctDNA)**: there is evidence that ctDNA testing has analytical validity, clinical validity and clinical utility in the context of some cancer diagnoses.[11–13] When this is the case, the results can affect standard treatment options and/or early assessment of the effectiveness of treatment.

   ctDNA tests may become useful as a screening/early detection test for cancer. This is being evaluated in clinical trials (eg Inherited Cancer Early Diagnosis Study[14]) and in specific clinical contexts (eg NHS pilots faster lung cancer diagnosis with blood test[15]). In the meantime, people with no signs or symptoms may seek DTC-GT ctDNA testing because of concerns about their risk of developing cancer. However, ctDNA testing does not yet have high analytical or clinical validity or clinical utility as a screening/early detection test in asymptomatic people.[16–18]

4. **Polygenic scores (PGS)**: common conditions, e.g. some forms of heart disease and diabetes, are influenced by genomic, environmental and other factors. Genomic variants can individually have a small effect on the risk of developing a common condition. When these variants occur together, they may account for an important part of the genomic risk. PGS measure the contribution of these multiple variants to risk. The predictive (or reassurance) value of PGS is limited because they do not take account of the effects of the environmental and other non-genomic factors that contribute to most common conditions.[19]

5. **Non-invasive prenatal testing (NIPT)**: is a technique used to screen a fetus for genetic conditions and variations, i.e. NIPT is not a diagnostic test. In the UK, NIPT is available as a second-tier screening test for pregnant women who receive a higher chance result from the NHS combined or quadruple test. NIPT is usually undertaken from around 10 weeks of pregnancy to screen for trisomy 13 (Patau syndrome), trisomy 18 (Edwards' syndrome) and trisomy 21 (Down syndrome). If the result indicates there is a higher chance of one of these conditions, then a woman can consider having an invasive diagnostic test.

   Some companies offer screening for other conditions as part of NIPT, including sex chromosome anomalies, chromosomal (micro)deletions and (micro)duplications, rare autosomal trisomies and for common conditions with a genetic component (see section about polygenic scores). NIPT can screen with a high accuracy for trisomy 13, trisomy 18 and trisomy 21, but there is limited information about the performance of NIPT when used to screen fetuses for these other conditions. Due to a low background prevalence of such conditions and confined placental mosaicism, false positive results are often

high.[20,21] This can lead to women/couples to seek unnecessary invasive diagnostic tests and to increased anxiety. In addition, some conditions for which screening is offered (e.g. cancer susceptibility) have an onset in adulthood, and environmental and other non-genomic factors make a significant contribution to the chance of developing these conditions. As with other types of DTC-GT, there can be false negative results, which can inappropriately reassure women/couples.

6. **Non-invasive screening for single gene conditions**: is a technique to screen for anomalies such as fetal single-nucleotide variants in individual genes. In the NHS, a diagnostic invasive prenatal test may be offered to look for these when anomalies are seen during an ultrasound scan in pregnancy. The decision to offer this is based on the scan findings indicating an underlying single gene condition, followed by a multidisciplinary review, and when making a molecular diagnosis may influence parental decision-making, management of the pregnancy or management of the baby in the neonatal period.

   Some companies offer non-invasive screening for single gene conditions outside of these criteria (e.g. in the absence of specific findings during an ultrasound scan). But there are minimal validation and follow-up data in low-risk pregnancies and both false positive results and false negative results are possible.[22] This can also lead to women/couples seeking unnecessary invasive diagnostic tests and to increased anxiety.

7. **Expanded carrier screening (ECS):** this test is used to identify couples at risk of having children affected by autosomal recessive conditions and X-linked conditions; multiple conditions are tested for at the same time.

   In the NHS, carrier testing for an autosomal recessive condition is offered in specific circumstances to partners of people who are affected themselves or who are known carriers (NHS testing criteria for rare and inherited disease[23]). Carrier testing for an X-linked condition is offered to women with a close family history of the condition.

   Some companies offer ECS in the absence of a family history to individuals planning a pregnancy, to those undergoing fertility treatment and to gamete donors. There is considerable variation in the number of genes offered for screening by different providers, and the selection criteria for deciding which genes to include in a screening panel is not always clear.[24] One of the selection criteria often referred to is the seriousness/severity of the condition.[25,26] However, the assessment of whether a condition is serious/severe is subjective, and perceptions about this can change, e.g. as treatments become available.[24] Some companies also offer tests for conditions with variable expressivity, reduced penetrance or conditions with an onset in adulthood.

Conclusions

Patients presenting with DTC-GT results should be offered standard NHS care, including family history and risk assessment, with onward referral and testing along standard NHS pathways and protocols. Genomic services are generally able to provide advice when it is unclear if a particular patient meets existing referral criteria. There are significant NHS costs in confirming (or refuting) DTC-GT results; these are not warranted unless there are clinical indications for testing.

Explanation of terms

Adverse drug reaction	unintended harm experienced by a patient as a result of medication.
Analytical validity	how well a test accurately and reliably detects if a specific genetic variant is present or absent.
Chromosomal (micro)deletion	the loss of genetic material from a chromosome. When part of a chromosome is deleted, the genes in that part will also be deleted. The effect of the deletion depends on which genes/parts of genes have been deleted.
Chromosomal (micro)duplication	the duplication of genetic material in a chromosome. When part of a chromosome is duplicated, the genes in that part will also be duplicated. The effect of the deletion depends on which genes/parts of genes have been duplicated.
Circulating tumour DNA (ctDNA)	as a tumour grows, its cells die and are replaced; dead cells are broken down and the contents, including DNA, are released into the bloodstream.
Clinical utility	how well a test improves patient outcomes and informs clinical decision-making.
Clinical validity	how well a test predicts a certain state of health.
Confined placental mosaicism	means that the cells in the placenta have too many/too few chromosomes but the cells in the fetus have the usual number of chromosomes.
DNA	deoxyribonucleic acid – the molecule that contains/encodes genetic information.
False positive result	indicates that a person has an increased/high risk when they do not.
False negative result	indicates that a person has a reduced/low risk when they do not.
Genetics	the study of genes and their impact on health.
Genomics	the study of the whole genome (both genes and non-coding regions).
Pharmacogenes	genes that affect how a person responds to medicines. These genes are often involved in the absorption, distribution, metabolism and excretion of drugs.
Prevalence	a measure of the frequency of a condition in the population at a particular point in time.
Rare autosomal trisomies	trisomy means having three copies of a chromosome instead of two. Rare autosomal trisomies involve three copies of a chromosome except for chromosomes 13, 18, 21, X and Y.
Reduced penetrance	some people have clear signs and symptoms due to a specific variant, others with the same variant do not have any signs or symptoms.

Sex chromosome anomalies	occur when a whole sex chromosome is missing or when there is an extra sex chromosome; there can also be deletions or duplications of sex chromosomes.
Single nucleotide polymorphism (SNP) arrays	a test used to identify variants at thousands of locations across the genome, originally developed to find common variants.
Variable expressivity	many conditions present differently in different people in the same family. This means that some people can have mild symptoms, while others can have significant symptoms.
Variants	each person has around 5 million genetic variants. Most genetic variants are harmless changes, and only a small proportion are disease-causing. Single gene conditions / predispositions are usually caused by variants that are very rare in the population (seen in <1 in 10,000 people).

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