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**Title: Transparency of Consumer Genetic Testing Services for ‘Health, Wellness and Lifestyle’: Analysis of Online Pre-Purchase Information for UK Consumers**

**Running Title: Transparency of DTC-GT Services UK**

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**Abstract**

The declining cost of DNA sequencing has been accompanied by a proliferation of companies selling “direct-to-consumer-genetic-testing” services (DTC-GT). Many of these are marketed online as tools for enabling citizens to make more informed decisions about their health, wellness and lifestyle. We assessed the ‘information for consumers’ provided

by these companies at the pre-purchase stage, which could influence initial decisions to part with money, data or tissue samples.

A scoping exercise revealed 65 DTC-GT companies advertising their services online to consumers in the United Kingdom (UK), of which 15 met our inclusion criteria. We benchmarked their consumer information against the good practice principles developed by the UK Human Genetics Commission (HGC).

No provider complied with all the HGC principles and overall levels of compliance varied considerably. While consent for testing was discussed by all but one company, information about data reuse for research or other purposes was often sparse and consent options limited or unclear. Most did not provide supplementary support services to help users better understand or cope with the implications of test results.

We provide recommendations for updating the pre-consumer transparency aspects of the HGC guidelines to ensure their fitness-for-purpose in this rapidly changing market. We also recommend improving coordination between relevant governance bodies to ensure minimum standards of transparency, quality and accountability. While DTC-GT has many potential benefits, close partnership between consumers, industry and government, along with interdisciplinary science input, are essential to ensure these innovations are used ethically and responsibly.

(242 words)

## Introduction

New DNA sequencing technology has spawned a growing market in genetic testing services sold directly to members of the public (1, 2), rather than being ordered by a clinician for medical reasons (3). These direct-to-consumer-genetic-tests (DTC-GT) are being marketed

online, promising purchasers insights about their ancestral past, present states e.g. being overweight, future risks ranging from premature balding to cancer, or even their own or their child's potential for intellectual or sporting achievement (4, 5). Health, wellness and lifestyle related services represent an important sub-sector of this market. While most are not strictly 'medical', many vendors of such services claim that these can empower consumers to make better health decisions by highlighting aspects of their genetic make-up that could affect their clinical risks or mediate the influence of lifestyle factors, medicines, nutritional choices or fitness regimes (6).

Although an exciting idea, DTC-GT comes with many uncertainties and challenges and given the psychosocial and privacy implications of genetic test results, it can represent a source of personal and societal risk. There is still limited evidence of the clinical utility of some of these tests, despite other types of personal and social value which purchasers may derive from them (3). Importantly, the extent to which consumers understand what they are buying, how they understand the results, what behaviours these trigger, and what will happen with their genetic information after the transaction, is still lacking (7, 8). Despite this uncertainty, governance of this rapidly diversifying sector has been hobbled by the multiplicity of bodies potentially involved in their oversight or regulation (Figure 1). For example, concerns about such products and services can span *in-vitro* medical device safety, consumer rights, fair advertising and data protection (9-12). To compound this, services sold online in one country may be underpinned by operations located in other regions with different regulatory jurisdictions, making it difficult for consumers to easily judge their rights for protection and recourse. Mindful of liability risks, many DTC-GT companies are marketing their services as "*information products*" rather than medical products, with the intent of them being "*for educational purposes only*" and stipulate that the test results are

not for diagnostic use. In the UK the DTC-GT industry is minimally regulated. Due to this lightweight governance, healthcare practitioners and professional bodies such as the Association for Molecular Pathology (13), the European Society for Human Genetics (14) and the American College of Medical Genetics and Genomics (15), have called for further guidelines and standards and have released their own (sometimes conflicting) guidance. In addition, consumer rights groups such as GeneWatch UK are becoming more vocal (16). Concerns have also been raised about the scientific integrity and clinical utility of some DTC-GT services, particularly those based on Genome Wide Association Studies (GWAS), which tend to have low discriminatory and predictive validity (17). The transparency of providers' activities has also been called into question relating to quality criteria and quality assurance (18). Although some guidelines cover scientific considerations, e.g. how to communicate the strength of evidence underpinning various genetic predictions (19), consistent global industry standards for transparency in the DTC-GT sector do not currently exist (11). Importantly, aside from scientific and clinical considerations, the ethical principles such as transparency, accountability and accessibility and choices and the quality of companies supporting information for customers is largely unchecked; a situation which has drawn attention from policymakers (20) and the biomedical industry itself (21).

In the United Kingdom companies are permitted to provide DTC-GT services to members of the public (22). Mindful of this, the UK government established a working group as part of the Human Genetics Commission (HGC), to advise on ethical practices within the industry. This included multidisciplinary stakeholders such as medical doctors, scientists, lawyers, professional organizations, government, and a company that was offering DTC-GT (deCODE Genetics) (23, 24). The HGC's 'Common Framework of Principles for DTC-GT' were released in 2010, and remain the only multi-stakeholder developed consensus guidelines focused on

best practice in DTC-GT in the UK. Significantly, the HGC's framework included specific recommendations relating to pre-purchase '*information for consumers*' which are particularly useful for benchmarking the transparency of DTC-GT companies, as is the focus of this study. Although other relevant guidelines have since been released by single stakeholder groups such as medical genetics professionals like the European Society of Human Genetics they do not cover issues of transparency or pre-purchase information in depth. For the purposes of our analysis we therefore selected the key HGC principles relating to '*information for consumers*' and sought to identify to what extent these recommended practices have been adopted by the sector, thereby assessing the responsiveness of the industry in adopting best practice standards (25).

## Methods

We searched for companies actively advertising to UK consumers for health, lifestyle and wellness related DTC-GT through Google UK searches, using the keywords "genetic testing", "health", "wellness" and "lifestyle". For the purposes of the study we defined active advertising in terms of companies' appearance in the first five pages of an online search on google.co.uk or appearing via "google ads". We acknowledge the variability in search engine results and so during the search process, we also identified a commercial website where DTC-GT companies pay to advertise and list their services to UK consumers (26), which we used to verify and supplement our findings. Of the companies identified by these searches, we selected only those offering DNA testing for health, lifestyle and wellness and excluded companies offering neonatal services or only performing ancestry or paternity testing, or not actively trading; for example, a telomere testing service that was described as "coming soon". Additionally, by default, we excluded private medical services where tests are

recommended by a clinician, focusing our attention only on those services that consumers can order directly online. The search took place between January 15<sup>th</sup> and June 30<sup>th</sup>, 2016.

To assess the transparency of information available to consumers at the pre-purchase stage, we extracted key documents from companies' websites for review. The target documents included Terms of Service (ToS), privacy statements, Frequently Asked Questions (FAQ) and informed consent documents, where available online. One company also had a "Code of Practice" statement, which we included. We worked on the principle that all the necessary key information should be presented openly to potential purchasers in these documents, without them having to search each website extensively for fragments of information or wait until after ordering the service.

We created an initial checklist of assessment questions consisting of 16 key criteria derived from the section of the HGC guidelines concerning "*Information for prospective consumers*". A pilot review assessed the applicability of the checklist to current DTC-GT provider practices (27). During this process, an additional 12 questions were added to the checklist, in most cases consisting of more detailed sub-questions associated with a broader item from the original HGC recommendations (see Supplementary Information SI Table 1 for details).

In the second phase, we moved to a full assessment, involving three evaluators; an expert in biomarkers and quality assessment (JH) a specialist in health information governance and data protection (RG) and two solicitors specializing in ethical practice (DD and SF, providing a single rating sheet). A training process for assessors took place to ensure common interpretation of the language and concepts used in the criteria and resolve ambiguities in advance of the task. The detailed assessment exercise took place between February and

June 2016. For each criterion, the available information was classified as “Addressed”, “Not Addressed” or “Not Applicable”. Inconsistencies were resolved through discussion and, where necessary, arbitrated by a fourth independent evaluator (CP).

## Results

65 companies were identified from the broad search. These companies offered a range of services within the broad categories of ancestry, paternity testing, neonatal testing, beauty (skincare) and ageing (telomere testing), as well as health, wellness and lifestyle. Out of these we selected those offering DNA testing in health, lifestyle and wellness (N=15), as per our inclusion criteria, meaning that 50 were eliminated from our review. The 15 remaining companies encompassed services prioritising physical fitness, nutrition and health-related wellness, lifestyle and phenotypic traits, in addition to those concerned with disease susceptibility, drug sensitivity, disease prevention and disease risk assessment (Table 1).

The 15 DTC-GT providers selected offered a diversity of services within the test categories presented in the Supplementary Information as defined in the HGC guidelines, ranging from pre-symptomatic tests, carrier testing, testing for disease susceptibility traits, pharmacogenetics, nutrigenetics and lifestyle/behavioural traits. Some providers focused on one or two types of service whereas others covered these categories. For example, only 3/15 services included carrier testing whilst 13/15 companies offered testing for nutrigenetic and lifestyle/behavioural traits testing (Table 1).

Only 10/15 eligible companies were registered in the UK. Others were registered elsewhere but were nevertheless attempting to engage with UK consumers. In some cases, it was difficult to identify the origins of the company based on the information provided on their



consumer-facing website; for example, language such as “principal place of business” was used ambiguously.

Not all companies provided similar documentation such as ToS and privacy statements, FAQ or informed consent templates. In some cases, these were referred to using different words or combined within the same document. For example, some companies embedded informed consent statements within their ToS or privacy statements, while others supplied separate detailed informed consent forms. Additionally, one company had a “Code of Practice” statement and one had a separate “Family Considerations” document as well as a separate “Biobank” consent form.

All reviewers remarked on both the variability of the reviewed documents’ format and content. Reviewers noted that, in some cases, the term ‘genetic data’ was not defined, there was little or no information about genetics or risks specific to genetics. Also, that the scope of testing performed was not always adequately explained and no assurances were given that further testing would not take place for purposes beyond those for which the test was purchased.

#### *Overall compliance with HGC principles*

As illustrated in Figure 3, none of the DTC-GT companies reviewed complied with every HGC recommendation. There did not appear to be any clear differences in the providers’ tendency to comply with the HGC criteria depending on whether the type of service emphasised health-related traits (e.g. carrier status, drug sensitivity, inherited conditions) or lifestyle and wellness factors (e.g. diet, weight-loss, nutrition and fitness related traits) and for this reason we have not presented a separated analysis.

Some criteria were widely met by the companies in the sample and others hardly at all. Table 2 summarises the most and least frequently adhered-with principles (the full list of criteria used are provided in the Supplementary Information). An important differentiating factor between DTC-GT providers, was the extent to which they were explicit about the ways in which they protected users' information, their intended future uses of customers' data and the options available for differential consent of withdrawal. These considerations are the focus of the section below.

### *Consent for genetic testing*

A key ethical principle specified by HGC is that patients should be required to give informed consent for genetic tests to be undertaken. Most companies (14/15) followed at least one of the following three themes. These themes were identified in the reviewer's discussion:

Theme A, the company explicitly stated in their ToS or Privacy Statement that informed consent was required before testing. 7/15 stated that customers would be asked to sign a separate informed consent document (for example after receiving the kit) if they wished to proceed with the test. Only 1 provided the informed consent document on their webpage, while 1 provided their informed consent document upon request (by email). Theme B the company mentioned consent within their ToS or privacy statements, indicating that if the consumer agreed with these then they would be assumed to have given informed consent for testing (5/15). This did not require ticking a separate box. For example, *"You acknowledge that by agreeing to the terms of this privacy statement when purchasing services on this site, you are giving us your explicit consent to process this sensitive personal data related to you."* Or *"By proceeding with any test you are formally providing your informed consent to undergo a specific test"*.

Theme C, the company included statements to say, *"we shall not process any data without*

*your consent*” but did not specify what the consent process involved (2/15).

One company (1/15) neither mentioned the requirement for informed consent nor provided an informed consent form, in the pre-purchase documentation we reviewed. Therefore, the rigour and transparency of the consent processes provided by companies were highly variable.

#### *Consent for reuse of genetic data*

Only 4 companies explicitly discussed the re-use of customers’ genetic data for research using specific and defined terminology, but they did so with varying levels of transparency. One company explicitly defined “aggregate data” as different from “individual level” genetic data and stated that it automatically shared “aggregate” genetic data for research if the service was purchased, but required a separate informed consent for the use of “individual level” genetic data in research. Another 3 companies stated that “anonymised” genetic data would be shared for research without defining what “anonymised” meant. Of these three, only one explicitly sought separate informed consent to share this data for research through an opt-in mechanism (as recommended by the HGC), while the other 2 automatically shared de-identified genetic data in research if the service was purchased. A fifth company simply claimed ownership of customers’ data, but gave no specific information about how this might be used. Some companies made no specific reference to secondary uses of the genetic data, making it impossible to judge their policies or practise in this regard. The variability in approach between companies was marked, those providing detailed information boosted confidence in their service offering and companies offering no information appeared weak in comparison.

#### *Withdrawal of consent/cancellation of service*

Only one company was explicit about the consequences of withdrawing from the testing

service, including what would happen to customers' genetic data upon account closure. This company's information clearly differentiated withdrawal from the testing service and withdrawal of consent for future uses of data in research, thereby allowing consumers to retain the testing service account even if they decided to withdraw from research. The remaining companies were much less clear on this point; some discussed the possibility of closing your account or if you no longer wanted to use the service, but were not clear on what would happen to the genetic data. Several (6/15) companies mentioned the right to cancel the purchase and offered a cooling off period, but did not discuss what would happen to any genetic data that may have been generated prior to cancellation. The distinction between the right to cancel a service (cooling off period) and the right to withdraw after participating in testing should be made clearer and both options should be available to the consumer.

#### *Client confidentiality and data protection*

Although many providers (9/15) referred to data protection law(s) applying to their service, only one company explicitly stated that it was registered with the UK Information Commissioner's Office as a data controller. Many gave general statements such as "*we shall not use or process your data without your consent*". Furthermore, as some companies spanned operations in several jurisdictions, a variety of laws governing uses of customers' tissue samples and genetic data were mentioned, with potential to confuse consumers as to their rights and confidentiality.

#### *Test accuracy and scope*

In the UK, and approximately 60 other countries, ISO15189 is the current standard for accreditation of laboratories that perform medical genetic tests, and ISO15189 states laboratories should participate in regular External Quality Assessment (EQA), to ensure the

accuracy of their test results. However, for companies offering DTC-GT for “information and educational purposes only” there is no requirement for ISO15189 accreditation (and thus participation in EQA), although this does not prevent them from voluntarily conforming with ISO standards and participating in EQA. A second quality indicator is the CE-mark, a well-known symbol designed to enable consumers to verify the quality and safety of products in the EU. The CE-mark is required for diagnostic kits under the EU *In Vitro* Diagnostic Medical Device regulation. In the UK, the CE-mark is given by the Medicines and Healthcare Products Regulatory Agency (MHRA), the body responsible for ensuring that medical diagnostic tests reach these standards in the UK. Some DTC-GT services do fall under the remit of the CE-mark, even if the DTC-GT service provided is informational rather than medical diagnostic testing. However, MHRA only reviews services related to testing for disease and disease risk, thus genetic testing for nutrition, lifestyle and behavioural traits - representing a large section of the current market – is not covered under this remit. Moreover, the assessment required for the CE-mark only covers the testing kit/ technology and not other aspects of the service such as the quality of consumer information, transparency or data protection. We analysed companies’ consumer pre-purchase information relating to these three points (ISO accreditation, participation in EQA and CE-marking). Relatively few companies mentioned the accreditation of the laboratories used, or indicated why this might be relevant. Two companies referred to ISO17025; a standard for accrediting laboratories’ which is not widely recognised as the standard for medical testing in the UK or EU; and one referenced Clinical Pathology Accreditation (CPA); which has been phased out since 2009. One referred to AABB accreditation (standards of transfusion medicine and cellular therapies to optimize patient and donor care and safety) which appeared irrelevant to the DTC-GT service, and one referred to the Care Quality Commission without specifying how

this applied to their service. Only one company mentioned ISO15189 accreditation of the laboratory used, which appeared to be specific to the BRCA1/BRCA2 testing service. This provider also mentioned participation in a recent, relevant, EQA scheme. One company referred to CLIA, the US-standard for clinical laboratory testing however CLIA does not require the same level of depth as ISO15189, the latter requiring more depth on quality management, the establishment of continual quality improvement, and true audit-based monitoring. No other companies mentioned participation in EQA schemes.

Whilst the MHRA recommends that consumers search for the CE-mark associated with DTG-GT kits when purchasing products (28), none of the companies mentioned CE-marking in their pre-purchase information. This is not to say that some companies did not have CE-marking but rather that this information was not provided within their pre-purchase information, despite its possible influence on consumers' decisions.

Although some companies referred to laboratory accreditation, as described above, all companies included disclaimers in their consumer information, stating that they offered no guarantee of the quality of their laboratory results. Most also stated that the tests were not for diagnostic use and were for information and educational purposes only. The lack of information of these quality indicators, or mention of accreditation that seemed irrelevant could lead the educated reader to a sense of distrust in the quality services offered. Greater efforts must be made to address these quality concerns in this field.

#### *Pre and Post-Test Genetic Counselling and Expert Advice*

The HGC recommends that genetic counselling should accompany any genetic tests "in the context of inherited or heritable disorders". Three out of the 15 services we reviewed included tests relating to carrier status of heritable disorders but only 2 offered post-test genetic counselling. In addition to inherited disorders, test results for health, lifestyle and

wellness factors can also be confusing for purchasers or may suggest lifestyle changes for which expert advice in the form of a face to face or telephone consultation with an expert may be beneficial. We extended this principle of a consultation to encompass all types of test and expert council (nutrition, fitness, drug sensitivity etc.), most companies (9/15) did not specify that customers would have an opportunity to speak to a counsellor or trained expert as part of their standard service. 4 companies offered an optional pre-test consultation and 6 a post-test consultation. Post-test consultation was offered as an optional service to purchase in half of these cases. Even those services that performed best in the overall assessment of best practice fell short on this criterion and, overall, there was no obvious trend for more counselling to be offered by companies meeting more HGC criteria. An open discussion needs to be had with the industry about minimum standards of diligence in conveying information back to consumers, that also extends beyond hereditary disease situations.

#### *Formal Complaints Process*

Across all the companies reviewed, there was a consistent lack of information for consumers regarding a formal complaints process. Only 3/15 companies provided some information about a complaints process and one of those stated that *“all decisions of the management are final”*. There was a lack of reference to any external body to which complaints could be made if the consumer was not satisfied, most likely because there is no overarching body currently in place that could fulfil this role.

#### Discussion

The HGC published its principles and recommendations in 2010, with a view to guiding companies active in or entering the DTC-GT space. This remains the only available set of guidelines that has been developed by representatives of all major stakeholders, including

industry members and regulatory bodies. While other guidelines exist, these typically represent the viewpoints of single organisations, such as those representing healthcare professionals.

The company that contributed to the HGC committee is no longer a provider of DTC-GT and none of the companies reviewed in this study participated in writing the HGC principles, indeed most did not exist at that time. This illustrates the growing disconnect between the sector offering services and the existing guidance, which is not fully addressed by newer academic guidelines developed by individual stakeholder groups (13,14,15).

Our analysis shows that none of the vendors actively targeting UK consumers comply fully with the HGC principles and the industry has not so far embraced the self-regulatory approach that was envisaged at the time these were developed. Adherence with individual HGC criteria is typically inadequate and varies widely. The information provided to prospective purchasers is, in many cases, lacking in detail or unclearly worded, and the stated ToS can be ambiguous. Even those companies that performed best overall in our assessment fell short on the provision of counselling or expert advice as part of their service or even as an add-on. One company stated its own best practice criteria ('code of practice') on its website, without any reference to these being grounded in any external guidance (29), suggesting that while the sector is beginning to recognize the need for ethical and quality standards there is still an absence of consensus on a single set of accepted criteria. We also identified aspects of the existing HGC guidance that require updating or extension to meet the needs of this rapidly evolving market; a greater emphasis on the *transparency* of vendors' ToS and privacy statements with respect to the products and services offered and the future uses of customers' data, distinction of *cancellation* of the service vs *withdrawal* from data (re)use, formal *complaints* processes and more explicit and specific



*consent* processes. Companies need to better address the language used to describe genetic data; indeed 7/15 companies did not even specify “genetic” or “DNA” when referring to data in their ToS and privacy statements but simply talked about “*your personal data*”.

Greater specification of the accuracy and reliability of tests used, and their ability to be used in medical decision making, alongside references to appropriate and current *quality* standards, such as laboratory accreditation, participation in proficiency testing and CE-marking (where applicable) are also needed to help consumers make informed purchasing decisions. A lack of counselling or an opportunity to discuss test results with an expert also represents an important weakness in this sector, given the *expertise* required to properly interpret test results and the fact that some results may lead to anxiety for consumers.

While a few companies offer a personalized consultation as part of their service, some either sell it as an extra service or make recommendations on how to obtain it, but many simply do not mention this as an option in their consumer-facing documents. These deficits in information and counselling may help to explain the results of recent research indicating that DTG-GT can leave consumers more confused about their health, rather than more enabled to manage it effectively (30). Nevertheless, the mere involvement of a physician is no guarantee of good practice; indeed, a recent review identified poor consent mechanisms in several companies whose services could only be ordered through a physician (31) and in our review the company with the worst overall rating for transparency had medical doctors available for post-testing counselling.

While we made efforts to identify all companies selling DTC-GT services to UK consumers in the areas of ‘health, wellness and lifestyle’, this review must be regarded as a snapshot of a rapidly evolving market that deserves ongoing monitoring and governance. At this time, no single government body or professional association fulfils this oversight role in the UK.

Our analysis focused only on the online documentation offered by vendors to potential customers at the pre-purchase stage, which may influence their decisions to part with money, tissue samples or personal data (Figure 3). We looked specifically at the documents relating to fair marketing such as quality of service, data protection, consumer safety, transparency and accountability. We acknowledge that vendors may provide additional information elsewhere, such as on separate protected websites available post-purchase or in paper documents provided alongside test kits, however our perspective was that key information should be available 'front-of-house' in an accessible and central form, even if repeated elsewhere, in order to support informed choice at the point of sale. Arguably, the transparency and usefulness of this information may also indicate companies' broader philosophies, experience, and management practices, although we leave this for others to unpack through a more in-depth analysis of purchased products and services.

Our emphasis on the HGC guidelines was driven by our focus, for the purposes of this study, on UK consumers and the UK market, given the time elapsed since the HGC was disbanded and our awareness of the complex and somewhat disjointed state of UK regulation, which spans multiple bodies and relies heavily on industry self-governance (Figure 4). This fragmentation has also been described in a recent systematic review of European guidelines, recommendations and position statements (12). Our assessment was based on a simple, actionable, yes/no checklist. We did not specifically examine the fitness-for-purpose of contract language relevant to the HGC principles and this would merit further study. Our interdisciplinary team nevertheless included experts in genetics, laboratory testing standards, eHealth, consumer rights law, information governance and bioethics. As our review included companies offering a range of health-related, wellness, fitness, testing services we were able to compare across sectors of the market, however no major

differences in HGC compliance between these types of company were discernible from our analysis, although we acknowledge the relatively small study size.

Our results are consistent with a recent review of the United States' DTC-GT market, which also revealed inconsistent provision of information regarding security protocols and tissue retention, along with significant gaps around transparency, readability and informed consent processes for the reuse of data and tissue samples in research, despite most companies posting privacy statements (31). In addition to DTC-GT companies transacting solely with consumers, those involving physicians can also exhibit poor practices. For example, in our study the DTC-GT company that performed worst on the HGC-derived transparency criterion had medical doctors both on the management board and providing in post-testing counselling as part of the service, whilst in the US study two out of the three companies failing to seek consent for research uses of customers' data or samples also explicitly required physicians to be involved in ordering the service (31).

## Conclusions and recommendations

Companies selling genetic tests directly to the public are proliferating in both number and diversity and are becoming increasingly popular with consumers seeking to understand or optimise their current and future health and wellbeing. Given the psychosocial and privacy implications of genetic test results, and their potential use by insurers, they also represent a source of personal and societal risk. Alongside the decreasing costs of genome sequencing the value of the DTC-GT sector has been projected to reach a \$340M by 2022 (32, 33), which may tempt some businesses to be less stringent with their offerings to reach lucrative markets of both direct citizen consumers and indirect consumers, such as commercial data brokers. However, companies which fail to innovate ethically and responsibly not only damage public trust but are also at commercial risk, if tougher penalties are being levied on

those that fail to comply with appropriate regulations (34). For this reason, standards for quality, transparency and accountability will prove increasingly essential, both for consumer protection and for the sustainability of this business sector.

Our study reveals that over the last 6 years since the HGC principles were released in the UK, self-regulation of DTC-GT in the UK has not led to consistent quality and transparency for consumers, with uptake of the HGC principles being inadequate and patchy. This leads us to question the efficacy of the current self-regulatory approach, which also reflects the absence of an integrated oversight body for DTC-GT in the UK, where relevant responsibilities are currently distributed across multiple agencies.

Although the HGC guidelines remain valuable for informing best practice they require updating to account for industry trends, including not only the proliferation of DTC-GT companies but also the diversification of their products and services, as well as emerging business practices such as the harvesting of customers' data for further research or re-sale, which threaten to compromise privacy and personal control. Given that DTC-GT are typically sold online, single-nation approaches are unlikely to be sufficient and global governance mechanisms with proactive oversight and regulatory alignment, are needed to protect consumers, maintain public trust and drive quality standards in the industry. This is a challenging prospect and our study demonstrates the difficulty of achieving this in one country alone. In this respect, it has been observed that numerous European policy reports on DTC-GT have been written but few have been implemented (12).

We recommend new efforts to agree minimum standards of transparency and quality, with appropriate mechanisms of review and certification of DTC-GT providers to increase the accountability of companies active in the DTC-GT market. Providing an independent trusted quality mark would benefit both consumers and healthcare professionals dealing with

questions from patients who have purchased or are thinking of purchasing DTC-GT services (35).

The results of our study have shone a light on inadequacy of the pre-purchase information currently being provided to UK consumers by DC-GT companies and its inconsistencies with the principles for good practice laid out by the HGC. Our suggestions for improving transparency and consumer support, decreasing risk and improving governance add to others in the literature and are compatible with several ongoing developments such as the EuroGenTest tool, which has been designed to aid professionals and consumers through choices about purchasing DTC-GT (36). While there is clearly a need to improve the transparency, quality and accountability of DTC-GT companies operating in the UK, in this global service market laboratories and business operations may be distributed across jurisdictions and greater international collaboration is also needed to agree consistent and actionable standards and mechanisms of governance.

While DTC-GT offers opportunities for consumers to better understand and manage their personal health and wellness, industry self-regulation has failed to ensure compliance with best practice and the transparency of the information available to guide consumers' purchasing decisions falls short on many criteria. Improving transparency, quality and accountability in this fast-evolving sector is essential to protect consumers and ensure responsible innovation and business practices. Achieving this will require partnership between citizens, industry and government, alongside interdisciplinary science input. As a first measure, we suggest revisiting and updating the 2010 HGC guidelines in the UK to take account of innovations and changes in this market and to create a specific set of criteria on transparency. We also recommend greater dialogue between the various stakeholders, including the companies, as well as increased coordination between regulatory bodies

concerned with consumer genetic testing, such as regulators of medical devices, consumer safety, data protection and fair marketing, to address their gaps and misalignments. In this global market, new initiatives to seek wider agreements on international standards for quality, transparency and certification are also called for.

### **Author contributions**

The study was conceived and designed by JH, in collaboration with CP. JH and JA undertook the pilot review to inform the criteria. JH, RG, DD and SF (acknowledged) conducted the full independent assessments. JH and CP drafted the manuscript, with input from RG, which was reviewed and approved by all authors.

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### **Conflicts of Interest**

The authors declare no conflicts of interest.

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### **Titles to legends and figures**

Figure 1. A schematic of the relevant governance bodies that touch upon aspects of DTC-GT services. Broken lines indicate bodies that could be involved in regulating DTC-GT but that currently do not play a dedicated role in regulating DTC-GT.

Figure 2. A heatmap of the companies' adherence with the items on the HGC-derived checklist, based on the information disclosed to consumers at the pre-purchase stage. Panel A indicates congruence with individual HGC criteria (indicated with \*) and additional criteria, and panel B indicates whether consultation with a specialist (genetic counsellor or trained expert) was provided as part of the service. The rows represent criteria and the columns the DTC-GT providers reviewed.

Figure 3. A schematic of the stages in the DTC-GT pathway, including relevant consumer issues, exit points and relevant governance bodies.

Table 1. Study flow chart. Note that some companies cover more than one area of testing services.

Table 2. Illustration of the most and least frequently adhered-with principles.

Supplementary information is available at European Journal of Human Genetics' website (Supplementary Information SI Table 1).

Table 1.

**Study flow chart**

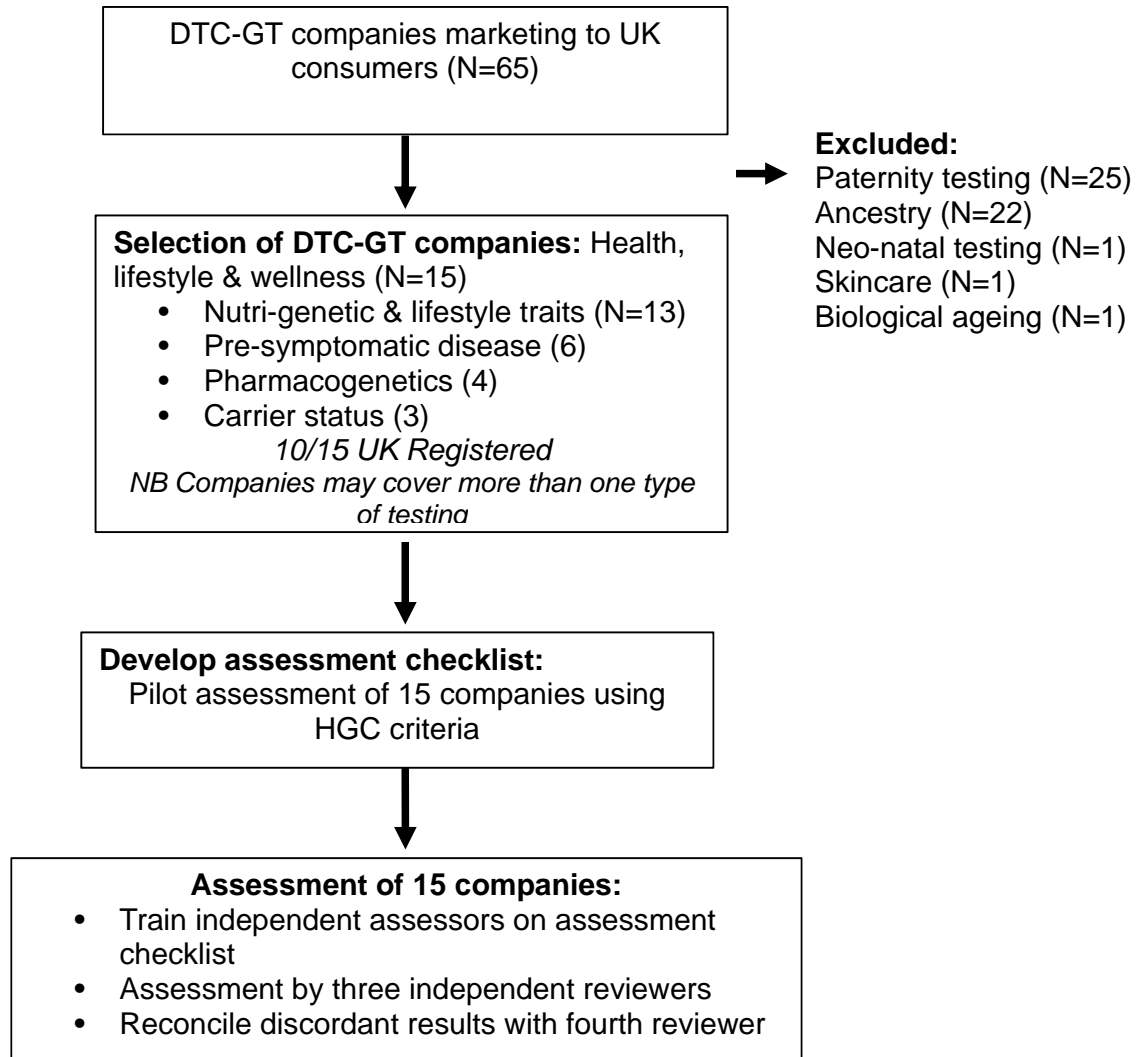


Table 2. Illustration of the most and least frequently adhered-with principles.

**Most frequently specified:**

- Purchaser must verify that tissue sample is their own (12/15) [Surreptitious sampling]
- That the company specified that informed consent is required for the testing service (12/15) [Specific Informed Consent Document] – *see below section on consent for details*
- Specified duration of storage of the tissue samples (10/15) [Duration of storage]
- Relevant data protection law applying to the service (9/15) [Data Protection]
- Information on safeguards implemented to ensure data security (9/15) [Data security measures]

*NB the above criteria are also criteria mandated by various regulatory requirements and are required for company compliance and reduction of company liability.*

**Least frequently specified:**

- No provider indicated the CE-mark on their webpage or mentioned its applicability to their service in the reviewed documents (0/15). [CE-marking]
- No provider had explicit language around what genetic data would be generated, including limitations and accuracy of tests and specified that no further testing outside that scope would be conducted (0/15). [Accuracy and scope of testing]
- Right to withdraw from the genetic testing service including specification of the consequences of withdrawal (e.g. data deletion, cost, access to stored information or future services) (1/15). [Right to withdraw from the service] (*One provider stated “if you wish to stop using your account, you can do so at any time, you do not need to inform us”*).
- Consent to purchase genetic testing service is separated from consent to re-use of personal data for research (1/11)\* [Service is independent of research]

*\*Not applicable to companies stating that they do not share data for secondary purposes and tissue samples used for testing are destroyed afterwards.*

# Patchwork Governance Ecosystem

## Who has oversight?



# A

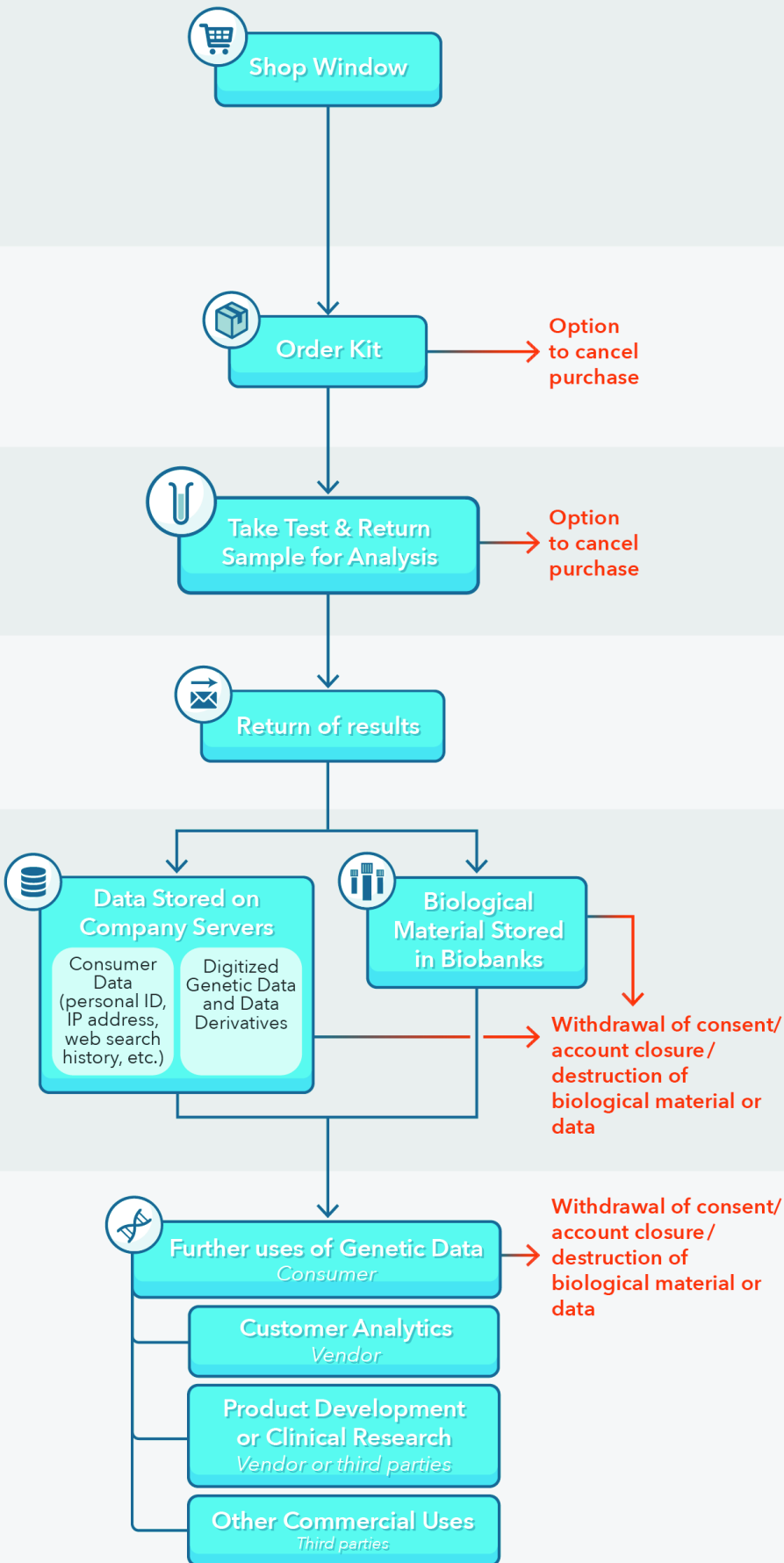
DTC-GT Providers	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
Surreptitious sampling*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed
Specified informed consent process (service)*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed
Requirement of informed consent for testing (service)*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed
Duration of storage*	Addressed	Addressed	Addressed	Not addressed	Addressed	Addressed	Addressed	Not addressed	Addressed	Addressed	Not addressed	Addressed	Not addressed	Addressed	Not addressed
Data protection	Addressed	Addressed	Not addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Not addressed
Data security measures*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Disclosure on data access*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed
Access by law*	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
T&C change*	Addressed	Not addressed	Addressed	Addressed	Not addressed	Addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed
Data location	Addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed
Tissue sample location	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed
Cancellation of purchase	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Addressed	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed
Statement of risks*	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Nature v.s. nurture*	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed
Separate informed consent for research (active assent)*	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not applicable	Not applicable	Not applicable	Not applicable	Not applicable	Not applicable
Impact on health insurance*	Addressed	Addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Data access	Not addressed	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not applicable	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Addressed
Ceasing trading*	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Genetic relationships*	Addressed	Addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Complaints process*	Not addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Updated test results*	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Consumer confidentiality	Addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Regulatory authority	Not addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Right to withdraw (from research)	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Scope and accuracy of testing*	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
Service is independent of research	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not applicable	Not applicable	Not applicable	Not applicable	Not applicable	Not applicable	Not applicable
Right to withdraw (from the service)	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed
CE marking	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed

# B

Pre-test consultation	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed
Post-test consultation	Not addressed	Addressed	Not addressed	Not addressed	Not addressed	Not addressed	Addressed	Not addressed	Addressed	Addressed	Not addressed	Not addressed	Addressed	Not addressed	Not addressed	Addressed



# Stages in the DTCGT Journey



## Consumer Information Needs

- Transparent and accurate marketing messages
- Frequently asked questions
- Professional association membership/best practice
- Accreditations and certifications

- Terms and conditions/ Terms of service agreement

- Informed consent for biological testing, storage, and data management
- Terms and conditions/ Terms of service agreement

- Counselling/further support of advice

- Differential consent options
- Informed consent for data sharing and data reuse
- Informed consent for biological material storage and/or reuse

- Transparency
- Interpretability
- Differential consent options
- Unintended uses
- Incidental or novel findings
- Access to innovation

## Ethical/Governance Considerations

- Advertising standards
- Consumer protection
- Professional body best practice recommendations

- Fair contract terms
- Consumer protection rights

- Consumer protection rights
- Regulations i.e. CE marking/EU IVD
- Laboratory accreditation and proficiency testing

- Access to appropriately trained experts for support

- Consumer protection
- Data protection and privacy rights
- Secure storage and transfer of data or biological material

- Fair insurance rights
- Consumer protection from unauthorized sharing or resale of personal data or biological materials
- Consumer rights to retain or manage personal information assets