Pathways, Processes and Protections: Australia's Clinical and Direct-to-Consumer Genetic Testing Spaces

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Abstract

Health-related genetic testing (clinical genetic testing), once available exclusively within the medical space, is now available directly to consumers within the commercial space (direct-to-consumer genetic testing). This paradigm shift from *medica*l to *consumer* has presented challenges to regulators, healthcare professionals and individuals. This article reports on mapping and modelling of pathways, processes and protections afforded to Australians whether they obtain genetic tests and results from their medical professionals or directly from commercial players. What emerged was a complex web of protections available to those obtaining health-related genetic testing in Australia's medical space. These protections, however, have time and cost implications, resulting in the potential for protracted translation of genetic discoveries into tests and treatments. In contrast, those obtaining health-related genetic testing in Australia's commercial space or online rely on the protections afforded all consumers in all marketplace transactions. Modelling also revealed the potential for these two, initially bifurcated pathways to merge, either by business model selected or individual choice. Individuals pursuing commercial options obtain personal genetic information, which they can self-interpreted and, if they choose, share with family, medical professionals and online. Sharing online in commercial websites results in increasingly larger databases available for monetisation. While this article focuses on health-related genetic testing, it gives insight into what will undoubtedly be continued incursions into the medical sphere by commercial players.

Keywords:

Clinical genetic testing, direct-to-consumer genetic testing, in vitro diagnostic medical devices, Therapeutic Goods Administration

Introduction

Traditionally, health-related genetic testing has been the exclusive purview of medical professionals, with access tightly controlled and results expertly interpreted and actioned (referred to herein as clinical genetic testing or CGT). Likewise, the regulation of genetic testing has traditionally been closely aligned with its clinical locus. This all began to change in the mid-1990s, most notably when the Icelandic government entered into an agreement with the company DeCode Genetics to establish a health sector database to map disease-related genes.[1] By the early 2000s, a number of other companies had emerged, offering health and ancestry-related genetic services directly to consumers.[2] The most prominent amongst these was 23andme, a California-based company.[3]

With the advent of this new form of health-related direct-to-consumer genetic testing (DTCGT), individuals seeking genetic information, whether to satiate curiosity, in response to familial or personal health-related concerns, or because tests are not available via CGT, can now simply 'spit in a tube', wherever and however they choose. For a comparatively small fee they can receive what the industry represents as a 'treasure chest' of genetic information. DTCGT's proponents exhort the benefits of opening the treasure chest, revealing genetic information that can be used by individuals in health and lifestyle decision-making.[4] DTCGT's critics urge caution, suggesting the treasure chest may in fact be Pandora's Box, with the potential to generate more harm than good.[5] Whomever is ultimately proven correct, DTCGT represents a paradigm shift from *medical* to *commercial* and ultimately *medical* to *consumer*, with new players entering the genetic testing space, presenting challenges to regulators, healthcare professionals and individuals.[6]

A solid understanding of the regulatory space, its key players, and gatekeepers is fundamental to analysis of any sector.[7] This is particularly important in the contested and rapidly evolving genetic testing space. It is well recognised that genetic testing raises profound ethical, legal and social implications.[8] The flow of genetic testing services from the clinic to the company is adding to these ethical, legal and social challenges. Although Australia is not a major player in commercial DTCGT, a

[1] R Chadwick, "The Icelandic Database—Do Modern Times Need Modern Sagas?" (1999) 319 *British Medical Journal* 234.
[2] Nuffield Council on Bioethics, *Medical Profiling and Online Medicine: The Ethics of Personalised Healthcare in a Consumer Age* (2010); P Borry, MC Cornel, HC Howard, "Where Are You Going, Where Have You Been: A Recent History of The Direct-To-Consumer Genetic Testing Market" (2010) 1 *Journal of Community Genetics* 101; SM Hartz, "My Experience with Direct to Consumer Genetic Testing" (2015) 5 *Narrative Inquiry in Bioethics* 208.
[3] https://www.23andme.com/en-int/
[4] D Magnus, MK Cho and R Cook-Deegan, "Direct-to-Consumer Genetic Tests: Beyond Medical Regulation?" (2009) 1 *Genome Medicine* Article 17; T Caulfield et al, "Direct-to-Consumer Genetic Testing: Good, Bad or Benign?" (2009) *Clinical Genetics* 77: 101; HC Howard and P Borry, "Direct-to-Consumer Genetic Testing: More Questions than Benefits? " (2008) 5 *Personalized Medicine* 317.
[5] Ibid.
[6] A Maguire and W Burke, "Health System Implications of Direct-to-Consumer Personal Genome Testing" (2011) 14 *Public Health Genomics* 53; S Hogarth, G Javitt and D Melzer, "The Current Landscape for Direct-to-Consumer Genetic Testing: Legal, Ethical, and Policy Issues" (2008) 9 *Annual Review of Genomics and Human Genetics* 161.
[7] See, for example, S Taylor-Alexander et al, "Beyond Regulatory Compression: Confronting the Liminal Spaces in Health Research Regulation' (2016) 8(2) *Law, Innovation and Technology* 149.
[8] K Brothers and M Rothstein, "Ethical, Legal, and Social Implications of Incorporating Personalized Medicine into Healthcare" (2015) 11 *Personalized Medicine* 43.

detailed understanding of the regulatory space is as important here as it is in other jurisdictions. Indeed, it could be argued that *because* there are limited DTCGT offerings within Australia, it becomes all the more important to understand the extent and limits of our regulatory competence. There is little doubt that Australian consumers are already actively engaging with DTCGT providers, whether in Australia or in other jurisdictions.[9]

The next part of this article describes the different pathways open to individuals seeking health-related genetic information via CGT and DTCGT. The article then reports the results of a mapping and modelling exercise aimed at identifying the key players and gatekeepers in Australia's CGT and DTCGT spaces.[10] This analysis is then used as a lens through which to view the consequences of these different encounters for individuals. This analysis illustrates the regulatory protections available to individuals who engage with CGT as patients and with DTCGT as consumers within Australia are fundamentally different. And once Australian tissue samples leave Australian shores, so too does the jurisdictional competence to regulate how they are used.

Pathways to Health-Related Genetic Testing

Individuals seeking health-related genetic information in response to symptoms or family history have traditionally had one pathway for access to CGT – through healthcare practitioners within each country's healthcare system. When individuals engage with health-related genetic testing through this pathway, they enter the medical sphere as a *patient*, afforded all rights, responsibilities and protections this designation entails.[11] Traditionally, access to CGT has only been provided for testing deemed 'clinically relevant' by qualified healthcare providers.[12] In Australia, clinically relevant services are those generally accepted by the medical profession as necessary for appropriate patient treatment.[13] Healthcare professionals view CGT through the lens of clinical utility, undertaking CGT only if tests have predictive value and/or can inform proven medical care options.

The emergence of DTCGT has provided an alternative pathway, allowing individuals to access health-related genetic information 'at the click of a mouse'. Individuals engaging with health-related genetic testing through this pathway enter the commercial sphere as a *consumer*, again afforded all

[9] Note, however, that survey evidence suggests that there is somewhat less interest in use of DTCGT services in Australia when compared with the US: J Charbonneau et al, 'Public Reactions to Direct-to-consumer Genetic Health Tests: A Comparison across the US, UK, Japan and Australia' (2020) 28 *European Journal of Human Genetics* 339.

[10] This article focuses only on health-related genetic testing, excluding companies exclusively providing genetic tests for ancestry, paternity, sporting prowess, etc.

[11] A *patient* is a person receiving medical care from a licensed healthcare practitioner, including diagnosis and providing treatment. See <https://medical-dictionary.thefreedictionary.com>, <https//www.medicinenet.com>.

[12] The increasing availability of whole genome sequencing and whole exome sequencing in the CGT space will, however, have implications for this traditional model. See D Chalmers, et al, "Personalised Medicine in the Genome Era" (2013) 20 *Journal of Law and Medicine* 577.

[13] Medicare Benefits Schedule, Department of Health, Australian Government. <http://www.health.gov.au/internet/hta/publishing.nsf/content/mbs-1>. See also Medical Board of Australia, *Good Medical Practice: A Code of Conduct for Doctors in Australia* March 2014 <https://www.medicalboard.gov.au/codes-guidelines-policies/code-of-conduct.aspx>.

the rights, responsibilities and protections this designation entails.[14] While two separate pathways exist, individuals are not precluded from pursuing both, depending on their particular motivations and situations.

CGT operates wholly within Australia's healthcare system, a complex system with combined federal and state and territory responsibility. The Commonwealth government takes a leadership role in policy making and research. State and territory governments bear primary responsibility for the delivery and management of public sector health services and maintain direct relationships with health care providers. Healthcare is costly and expected to continue rising, with funding provided primarily from federal, state and territorial government subsidisation, with the remainder from private health insurance, personal contributions and other non-governmental sources.[15] Consistent with Australia's view of healthcare as public good and therefore a public responsibility, since 1984 Medicare has provided *universal* access to public hospitals, registered primary healthcare professionals, required tests and drugs for citizens, permanent residents and those from countries with reciprocal health agreements.[16] Medicare is funded through compulsory levies on taxable income, administered federally through the Department of Human Services and has three key components − hospital, medical and pharmaceutical. Subsidisation for eligible *clinically relevant services* is detailed in the Medicare Benefits Schedule (MBS) and eligible pharmaceuticals in the Pharmaceutical Benefit Scheme (PBS).

The Commonwealth has actively encouraged development of a strong private health insurance sector, enacting a regulatory framework to encourage uptake by Australian citizens.[17] Private health insurance is regulated by the *Private Insurance Act 2007* (Cth) and provides subsidised access to private hospitals including choice of doctor, and allied healthcare such as dental if individuals so choose. Private health insurance is community-rated, meaning premiums and benefits are based on specific cover selected not criteria such as health status – individuals do not have to declare health status or undergo medical examinations prior to or during coverage periods.[18] Private insurance companies themselves decide on specific coverage packages and fees, waiting periods and reimbursement levels. Fees increase annually with the overall industry percentage increase requiring government approval. To encourage uptake, the Commonwealth government provides income and age-tested rebates on premiums and imposes additional surcharges on non-privately insured high-income earners.[19]

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[14] In Australia, a *consumer* is person acquiring goods or services of a kind ordinarily acquired for personal, domestic or household use or consumption under the value of $40,000: Schedule 2, s3 *Competition and Consumer Act 2010* (Cth).

[15] Australian Government, Australian Institute of Health and Welfare, *Health Expenditure Australia 2016 – 17* (2018) <https://www.aihw.gov.au/reports/health-welfare-expenditure/health-expenditure-australia-2016-17/formats>; Australian Institute of Health and Welfare, *Australia's Health 2018: In brief* (2018) Cat. no AUS201, Canberra, AIHW, 39.

[16] *Health Insurance Act 1973* Part I s3 for definition of 'registered primary healthcare professional'.

[17] See F McDonald and S Duckett, "Regulation, Private Health Insurance and the Australian Health System" (2017) 11(1) *McGill Journal of Law and Health* S31.

[18] *Private Insurance Act 2007* (Cth) s55-5(2).

[19] See *Private Insurance Act 2007* (Cth) Divisions 2 and 3. Life, income protection and disability insurance however are risk-rated, requiring declaration of health status, including known genetic conditions for determination of coverage and rates.

In contrast to CGT, DTCGT operates within each country's commercial space as well as the online environment. Companies typically conduct marketing activities and return results online, consistent with established e-commerce models where traditional intermediaries such as retailers are eliminated and transactions are completed online. As Internet penetration has increased, consumers have demonstrated increasing acceptance of e-commerce. While a country's offline commercial space clearly falls within their jurisdiction, the online environment presents particular challenges as jurisdiction and applicable legislation is determined by country of registration, denoted by URL extension. DTCGT companies have also capitalised on the 'tectonic shift in the ways in which patients consume health and medical information …'[20] with increasing use of online health-related information and self-diagnosis tools, with those searching excessively now referred to as 'cyberchondriacs'.[21] Moving genetic testing into the online environment market is undoubtedly redefining healthcare roles, making consumers active rather than passive participants in healthcare decision-making. The DTCGT industry's commodification and monetisation focus has imbued genetic data with economic value – making it simply too valuable to ignore.

Mapping and Modelling of the CGT and DTCGT Sectors

For the purpose of this study, a mapping and modelling exercise was undertaken of the regulatory spaces for both CGT and DTCGT sectors in Australia. In the context of CGT, the modelling was based on mapping of applicable legislation, regulations and industry standards. Discussions were also conducted with a consenting primary care physician, to verify processes. Modelling of the DTCGT regulatory space, in contrast, relied on methodology developed by the US Genetics & Policy Center. The Center undertook a mapping exercise in 2009 and 2011 of DTCGT companies offering testing for at least one medical condition, pharmacogenomic and/or nutrigenomic test to US consumers either onshore or online.[22] In 2013-2014, this mapping exercise was replicated and extended for the Australian market for the purpose of this study. The aim was not to produce a comprehensive list but rather to investigate the different options available for Australian consumers wishing to purchase DTCGT services.[23] Discussions were also conducted with a consenting adult who had purchased DTCGT tests from both 23andMe, based in the US, and EasyDNA, based in Australia, again for the purpose of verifying the analysis.

Figure 1 charts the flow of DNA samples, results, and advice for CGT offered in Australia's healthcare system. Tests are ordered by healthcare professionals if deemed warranted by them, with payment either coming from Medicare, state or territory funding (as is most common), or patient contributions. DNA sample provision is either done in clinic or in an accredited laboratory, ensuring patient identification verification, proper labelling of samples, and sterile environments. DNA

[20] B Hesse et al, "Trust and Sources of Health Information" (2005) 165 *Archives of Intern Medicine* 2618, 2618.
[21] See E Doherty-Torstrick, K Walton and B Fallon, "Cyberchondria: Parsing Health Anxiety from Online Behaviour" (2016) 57 *Psychosomatics* 390; E McElroy and M Shevlin, "The Development and Initial Validation of the Cyberchondria Severity Scale" (2014) 28 *Journal of Anxiety Disorders* 259.
[22] Genetics and Public Policy Center, "GPPC Releases Updated List of DTC Genetic Testing Companies" (2011). It should be noted, however, this link is no longer operational. US company and test list, methodology employed, and key words used in Google searches are available from the authors.
[23] For a comprehensive listing of DTC companies and the services they provide spanning the years 2011 − 2018 see A Phillips, "Data on Direct-to-Consumer Genetic Testing and DNA Testing Companies", 19th February 2018, DOI: 10.5281/zenodo.117922.

samples are provided to accredited laboratories that return results to originating healthcare professionals. Results interpretation is conducted by healthcare professionals and interventions developed, with the assistance of genetic professionals if required. Healthcare professionals *then* discuss results, advice and possible interventions with patients, calling on genetics specialists if required. Medical or pharmaceutical interventions consented to by patients are then arranged, involving medical specialists as required.

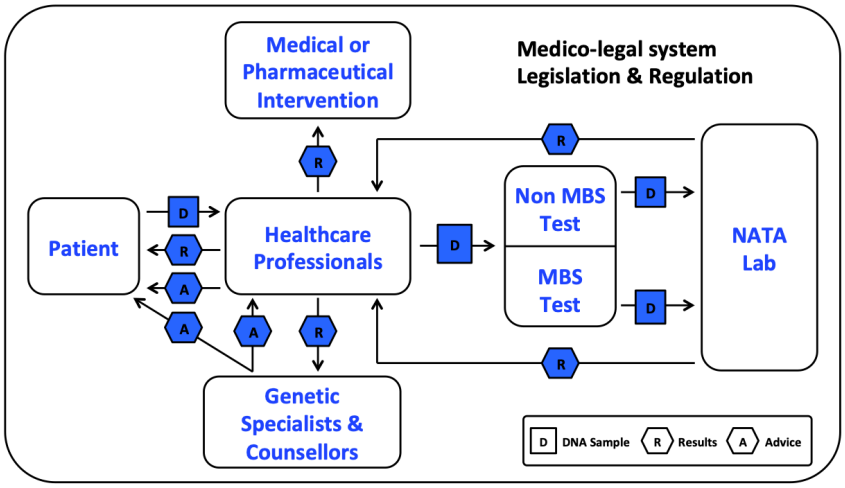

Figure 1: CGT in Australia's healthcare system

The DTCGT sector operates in a fundamentally different way, as illustrated in Figure 2. While most DTCGT companies employ standard corporate e-commerce models, others operate either fully or partially through multi-channel options including online shopping sites (e.g. eBay), 'over the counter' retailers (e.g. pharmacies), 'affiliates' (e.g. wellness centres), and 'accredited' allied professionals (e.g. nutritionists).[24] As the sector has matured, companies have begun offering whole genome and exome sequencing, as well as providing raw data files in addition to results reports. Figure 2 charts the flow of test kits, DNA samples, results, and advice in the standard DTCGT e-commerce model. The DTCGT e-commerce model operates totally within the commercial sphere, subject to applicable legislation, regulation and each country's legal system.

[24] In 2015 UK retail pharmacy chain Superdrug became the first to sell test kits in its 600 outlets.

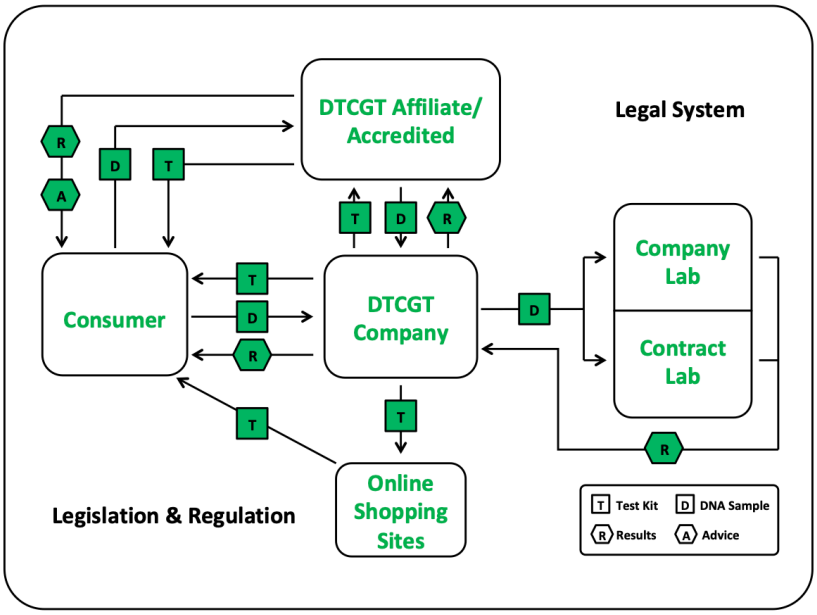

Figure 2: DTCGT: E-commerce model

The online shopping sites identified in Figure 2 function simply as distributors of test kits, extending DTCGT's reach to international consumers. The role of affiliates and accredited providers varies, with some returning and providing analysis of results and others functioning simply as test kit providers or submitting DNA samples. Either way, these intermediaries interject personal selling into the process and 'muddy the waters', making it difficult to determine when and with whom contractual relationships are formed. It is also significant these intermediaries are the only ones who potentially could provide consumers with advice e.g. results interpretation and intervention suggestions. This aspect is notably absent with DTCGT companies themselves who make it clear results are provided for 'research, education and information only'.

Figures 1 and 2 illustrate that CGT and DTCGT operate in two separate spaces, each with its own particular sets of processes and protections. Significantly, however, further analysis of the DTCGT space and flow-on effects illustrates that there is the potential for engagement between the commercial and medical sphere, suggesting the initial paradigm shift (medical to consumer) may in reality be a paradigm merge (consumer/medical). Two forms of DTCGT engagement with healthcare professionals were identified in this study: one selected by companies as their business model, and the other by consumers as their choice. These are presented in Figure 3, with aspects occurring in the commercial space in green and those in the medical space in blue.

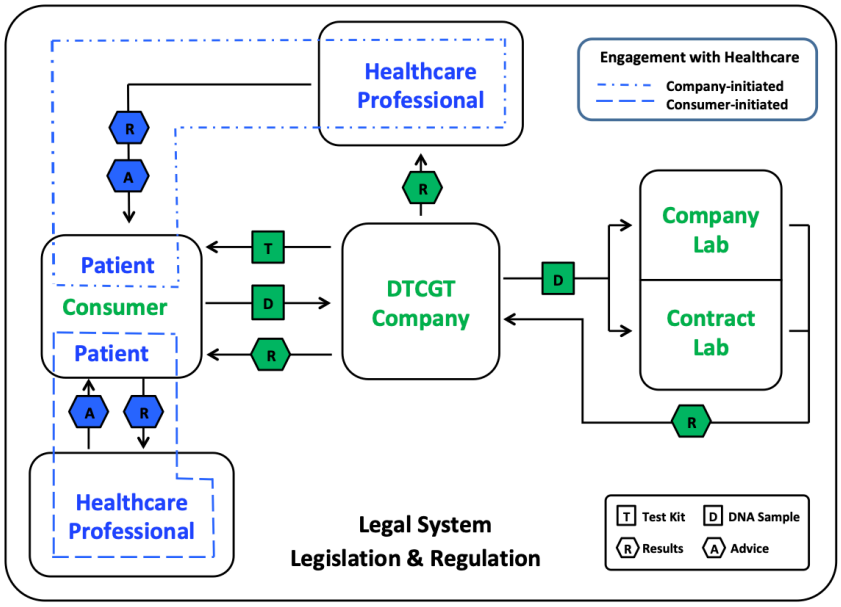

Figure 3: Company and consumer-initiated engagement with healthcare professionals

Where engagement with healthcare professionals is company-initiated, marketing is conducted directly with consumers but test ordering and/or return of results is via the individual's healthcare professionals or via corporate doctors. Some companies bypass involvement of individuals' personal physicians, instead offering to select doctors and obtain sign-off once consumers have purchased tests. Consumers have limited, if any, interaction with 'corporate' doctors, and it is unclear in what instances these 'paid for service' doctors would reject sign-off. As such, with company-initiated interaction, *consumers* are 'required' to engage with physicians if they wish to receive the results they paid for, and physicians are 'required' to provide *patients* with results from tests they may be unfamiliar with, or question their validity and utility. It should be noted, however, that while this business model appears to be gaining some traction in the US, most DTCGTs have adopted more traditional e-commerce models, eschewing direct engagement with healthcare professionals.[25]

As such, engagement with healthcare is more typically a consumer-initiated interaction, with individuals taking DTCGT results to physicians for confirmation, interpretation assistance, intervention recommendations or psychosocial support, again passing the burden to the healthcare system. Coupled with the DTCGT disclaimer of results being for 'research, education and information only' is usually the recommendation to 'seek professional medical advice', in essence prompting interaction with the healthcare system and removing any potential accusations of practising medicine without a licence.[26] Post-engagement confirmation testing and medical or pharmaceutical interventions, however, remain the exclusive purview of the medical sphere. Once this occurs, either as a result of company-initiated or consumer-initiated engagement, the *consumer* changes their role to that of *patient*.

[25] I Swetlitz, "Genetic Tests Ordered by Doctors Race to Market, while 'Direct-To-Consumer Tests Hinge on FDA Approval" *Stat* 16 March 2019 <https://www.statnews.com/2018/03/16/genetic-tests-fda-regulations/>.
[26] See A McGuire and W Burke, "Health System Implications of Direct-To-Consumer Personal Genome Testing" (2011) 14 *Public Health Genomics* 53; C Marietta and A McGuire, "Direct-To-Consumer Genetic Testing: Is it the Practice of Medicine?' (2009) 37 *Journal of Law and Medical Ethics* 369.

There is one other flow-on effect of consumers receiving DTCGT results or raw genetic data that needs to be considered – whether, how, and with whom they choose to share that data. Options are increasingly becoming available for voluntarily sharing genetic/health/medical/treatment details and receiving interpretation information and psychosocial support through online communities such as PatientsLikeMe.com and CureTogether.com. Those with raw data files can also use sites such as Promethease.com or Livewello.com to further interpret data for a larger range of health-related results, with both sites offering direct upload of files from major DTCGT companies. It should be noted that these sites are not simply passive sharing devices. For instance, 23andme has corporate ownership of CureTogether.com, and PatientsLikeMe.com on-sells contributor data to pharmaceutical companies.[27] As consumer contributions increase in online communities, these sites, like DTCGT companies, will amass extremely valuable databases of both genotype and phenotype data. Further, it has now became standard practice for DTCGT companies to routinely and actively seek consumer agreement to allow research to be conducted on submitted samples and genetic results.

Figure 4 illustrates what consumers might voluntarily do with their results and raw data files, including sharing with family, online communities, online interpretation sites or with doctors. Consumer also have the option of seeking additional disease-related information from DTCGT or general health websites. What is of note is that only engagement with healthcare professionals is within the medical sphere (if consumers chose to voluntarily share). All other forms of sharing remain firmly within commercial (online sharing) or personal (sharing with family) spheres.

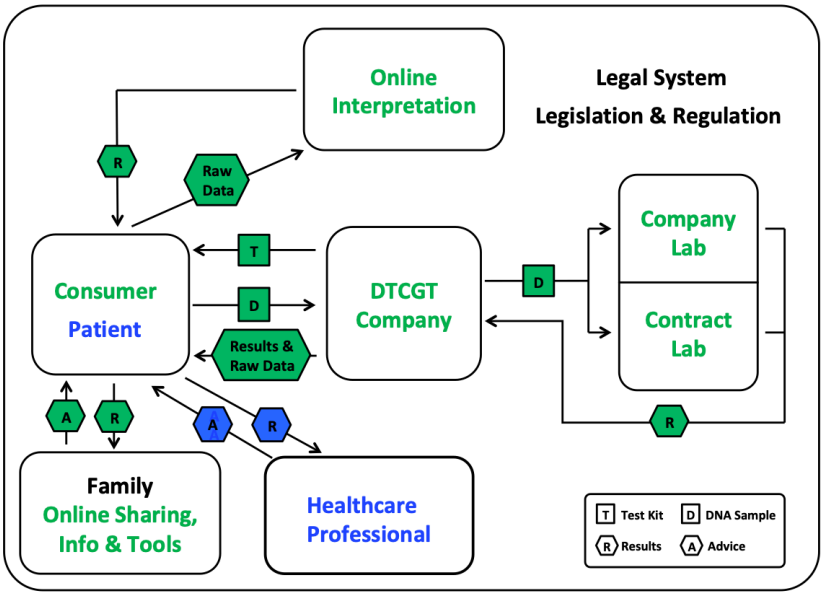

Figure 4: DTCGT's flow-on effect

Protections for CGT *Patients*

The medico-legal system within which CGT operates is extremely complex and the general protections afforded all patients, such as those offered under the Australian Charter of Healthcare

[27] See <http://news.patientslikeme.com/faq-item/faq/how-does-patientslikeme-make-money>.

Rights, are assumed adequate to protect CGT patients.[28] Regulatory theorists, however, have long recognised there is more to regulation than laws and other regulatory instruments.[29] Modelling of the CGT space identified high levels of professional and government oversight, with gatekeepers functioning as de facto regulators. Each gatekeeper provides specific protection for CGT patients.

Figure 5 illustrates three key categories of independent yet co-dependant gatekeepers: medical, financial and quality. Medical gatekeepers interact directly with patients while the other two groups operate behind the scenes. While the system may be a complex web with stringent and often duplicated regulation or oversight in some areas and gaps in others, at its core it seeks to protect CGT patients in their interactions with healthcare practitioners and ensures proper handling of sensitive genetic samples and resultant data. It aims to ensure that all CGT is valid, and all laboratory results provided for use in patient management are accurate, consistent and adhere to accepted domestic and international standards. While in many ways CGT is simply another form of medical testing, results have familial implications, requiring medical gatekeepers to always consider broader ethical and social implications of testing.

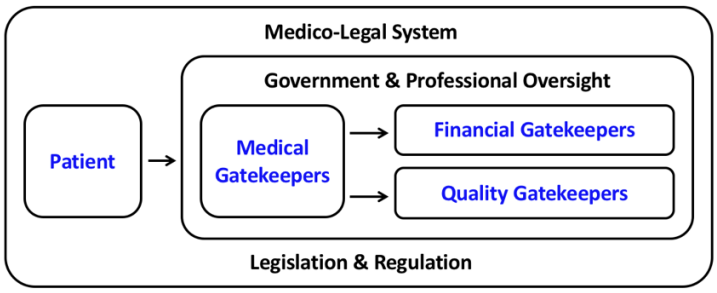

Figure 5: Gatekeepers in CGT

Medical gatekeepers: Doctors and genetic specialists

From the patient perspective, their first encounters are with medical gatekeepers. The *Health Insurance Act 1973* (Cth) only provides funding for eligible healthcare services (s10) rendered by registered healthcare practitioners (s3), with the Australian Health Practitioner Regulation Registry (AHPRA) providing the national registration and accreditation scheme. Australian patients can be assured only those who are who are 'suitably trained and qualified to practice in a competent and ethical manner are registered'.[30] The Medical Board of Australia (MBA), Australian Medical Association (AMA) and National Health and Medical Research Council (NHMRC) collectively develop the professional codes healthcare professionals must adhere to in private practice and medical research, with the need for ongoing registration ensuring compliance.[31] Patients with complaints or

[28] The Charter applies to all health settings and was endorsed by all Australian health ministers in 2008. Rights include, for example, the right to be informed about services, treatment options and costs. <https://www.safetyandquality.gov.au/wp-content/uploads/2012/01/Charter-PDf.pdf>.
[29] See J Braithwaite, "The Essence of Responsive Regulation" (2011) 44 *UBC Law Review* 475.
[30] The AHPRA is a statutory body under the *Health Practitioner Regulation National Law* (the National Law), in effect since 2010. It is an offence to falsely use professional titles or claim registration. See Division 10 Subsection 1 s113(1). Includes pharmacists, nurses and psychologists.
[31] See professional codes of ethics at <http://www.ama.com.au> and CGT guidance at NHMRC *Medical Genetic Testing Information for Health Professionals* (April 2010) <https://www.nhmrc.gov.au/guidelines/publications/e99>.

concerns both pre and post CGT have recourse through the AHPRA and MBA complaint procedures or through state and territory ombudsmen.[32]

These medical gatekeepers also determine if counselling pre or post-testing is required. The Human Genetics Society of Australasia (HGSA) certifies genetic counsellors, again developing specific codes, guidelines and educational requirements.[33] Access to genetic specialists is by general practitioner (GP) referral in most states and territories, although in certain instances individuals can self-refer. According to the NHMRC, 'the degree of counselling required depends on the level of uncertainty regarding the clinical implications of the test result, the potential implications for the patient, and the further implications for the patient's family.'[34]

Patients must give informed consent for CGT and resulting medical, pharmaceutical or counselling intervention, with such testing and intervention occurring in situations where individuals are duty bound to keep information confidential.[35] Access to test results and medical records is highly restricted.[36] The designation 'patient' enlivens an established duty of care relationship in common law between patient and healthcare professional.[37] Medical negligence occurs when the standards deemed appropriate relative to the duty of care are breached and patient harm ensues.[38] At common law, such breaches occur when the healthcare practitioner has failed to take reasonable care, taking into consideration whether the risk was foreseeable, not insignificant, and whether a reasonable person would have taken precautions considering, for example, both the probability and seriousness of harm.[39] Historically determining whether standards were breached was determined with reference to medical opinion.[40] In 1992, the Australian High Court determined that while 'evidence of acceptable medical practice is a useful guide', ultimate adjudication was the responsibility of the courts.[41] In 2002, legislative reform codified common law principles, with each

[32] The National Law establishes a National Health Practitioner Privacy Commissioner and the Office of the National Health Practitioner Ombudsman <http://www.nhpopc.gov.au>; States also provide services e.g. Health Complaints Commissioner for Tasmania <http://www.healthcomplaints.tas.gov.au>.
[33] See HGCA 'Guidelines for Training and Certification in Genetic Counselling' <http://www.hgsa.org.au/documents/item/1593>.
[34] Australian Government National Health and Medical Research Council, *Medical Genetic Testing Information for Health Professionals* (April 2010), viii <https://www.nhmrc.gov.au/guidelines/publications/e99>.
[35] Legal duty of confidence is inherent in doctor-patient relationship. See *Coco v AN Clark (Engineers) Ltd* [1969] RPC 41, parties realise or should information is of confidential nature and should remain private.
[36] Patients can voluntarily use Australia's eHealth system, deciding which providers are allowed access to particular information. See <http://www.ehealth.gov.au>. Alternatively, information may be housed with individual providers, with consent required for sharing.
[37] *Donoghue v Stevenson* [1932] AC 562 established general duty to take *reasonable* care to avoid *foreseeable* injury to a 'neighbour'. Civil liability laws such as *Civil Liability Act 2002* (NSW) s5O specifically address the standard of care expected of professionals. *Rogers v Whitaker* (1992) 175 CLR 479 established that the standard is raised to that of a competent practitioner in the particular field of specialty if specific skill or expertise is professed. See also *Wilsher v Essex Area Health Authority* [1988] 1 All ER 87 (UKHL).
[38] Healthcare professionals owe a concurrent duty of care in tort and contract although most actions are based in tort. *AAA v BBB* [2005] WASC 139 and *Rosenberg v Percival* (2001) 205 CLR 434.
[39] See *Tame v New South Wales* (2002) 211 CLR 317 and *Gett v Tabet* [2009] NSWCA 76.
[40] Referred to as the Bolam test – see *Bolam v Friern Hospital Management Committee* [1957] 1 WLR 582.
[41] *Rogers v Whitaker* (1992) 175 CLR 479. In 2015, the UK also overruled *Bolam* in *Montgomery v Lanarkshire Health Board* [2015] UKSC 11 moving their position on information and advice nearer the AU position.

state and territory enacting their own medical negligence laws, including type and quantum of remedies available and limitation periods.[42]

Financial gatekeepers: What's on offer and who pays?

Medicare is illustrative of an organisation performing a dual role as both a quality and a financial gatekeeper, functioning as a quality gatekeeper by determining which forms of CGT should be subsidised and a financial gatekeeper by determining levels of subsidisation. For CGT to be subsidised, it must be included on the MBS, which only occurs after evidence-based analysis to determine analytic validity, clinical validity and clinical utility.[43] Analytica validity measures whether tests identify the presence or absence of specific gene variants, while clinical validity determines whether mutations are related to specific diseases.[44] Tests for polygenic or multifactorial conditions are predictive, indicating susceptibility and therefore have lower clinical validity compared to the definitive diagnoses of monogenic tests.[45] Tests with clinical utility provide prevention, diagnosis or treatment information of use in healthcare decision-making.[46] While tests for conditions with no known cure or treatment may have low clinical utility they may have personal utility, assisting in family, lifestyle/quality of life and financial decision-making, equally relevant metrics.[47]

Figure 6 outlines the organisations involved in deciding which forms of CGT are listed on the MBS and therefore eligible for subsidisation for *all* Australians regardless of location.[48]

[42] Commonwealth of Australia, Ipp Committee, *Review of the Law of Negligence: Final Report*, September 2002; See, for example, *Civil Liability Act 2003* (Qld). Despite aiming to achieve a uniform approach to personal injury claims this has not transpired.

[43] See W Burke, "Genetic Tests: Clinical Validity and Clinical Utility" (2014) 81 *Current Protocols in Human Genetics* DOI: 10.1002/0471142905.hg0915s81; ACCE model developed by US Centre for Disease Control and Prevention's Office of Public Health Genomics <http://www.cdc.gov/genomics/gttesting/ACCE/>; Genetics Home Reference, 'How can consumers be sure a genetic test is valid and useful?" <https://ghr.nlm.nih.gov/primer/testing/validtest>.

[44] Genetics Home Reference <http://ghr.nlm.nih.gov/handbook/testing/validtest>; Genetics Home Reference <http://ghr.nlm.nih.gov/handbook/testing/validtest>.

[45] See Royal College of Pathologists of Australasia, "What Should I Know About Direct-To-Consumer Genetic Testing?" <https://www.rcpa.edu.au/getattachment/c7768ade-842d-4c9d-852a-6e38656964f8/FctSht-9-DrctConsumerGenTesting.aspx>.

[46] See M Foster, J Mulvihill and R Sharp, "Evaluating the Utility of Personal Genomic Information" (2009) 11 *Genetics in Medicine* 570; D Hunter, M Khoury and J Drazen, 'Letting the Genome Out of the Bottle – Will We Get Our Wish?' (2008) 358 *The New England Journal of Medicine* 105.

[47] See M Turrini and B Prainsack, "Beyond Clinical Utility: The Multiple Values of DTCGT Genetics" (2016) 8 *Applied & Translational Genomics* 4; E Bunnik, C Janssens and M Schermer, "Personal Utility in Genomic Testing: Is There Such a Thing?" (2014) *Journal of Medical Ethics* DOI: 10.1136/medethics-2013-101887.

[48] A similar system is in place for pharmaceuticals, which must be on the PBS to attract subsidisation and must be dispensed by registered pharmacists. The PBS Advisory Committee of experts advises the Commonwealth Department of Human Services of therapeutic benefits and cost-effectiveness compared to alternative therapies (<www.pbs.gov.au/pbs/home>).

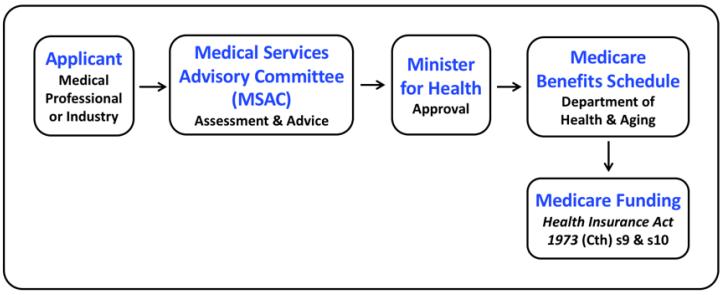

Figure 6: How CGT gets on the MBS

The process for obtaining Medicare approval is time-consuming and often contentious given escalating costs, budgetary constraints, and patient/practitioner demand. The Medical Services Advisory Committee (MSAC) is the sole source of expert appraisal advising the Minister of Health as to comparative safety, benefits (e.g. clinical utility) and cost effectiveness of CGT based on available evidence. If approved, the particular test is added to the MBS, making it eligible for subsidisation.[49] The MBS also controls access and funding for medical or counselling interventions required post-testing.[50] In reality, the MBS subsidises comparatively few types of CGT; most are subsidised by state and territory governments from their Commonwealth-allocated health budgets for *selected* patients from their respective jurisdictions.[51] Testing is conducted through specialised clinics in each state or territory's public health system, generally on referral from GPs, with criteria for access varying by state or territory. Ability to access non-subsidised CGT depends on individual financial capacity, as private health insurance only covers CGT for patients already admitted to hospital.[52]

Safety and quality gatekeepers: Tests and testing laboratories

Two quality gatekeepers are of particular note: The Therapeutic Goods Administration (TGA), ensuring CGT quality and safety and the National Association of Testing Authorities (NATA) accrediting laboratories.

The Therapeutic Goods Administration

The *Therapeutic Goods Act 1989* (Cth), as administered by the TGA, governs the safety, quality and performance of therapeutic goods either supplied within Australia or imported and exported.[53] The TGA applies scientific and clinical expertise to ensure benefits to individuals outweigh any risks associated with use. Therapeutic use is defined as use in or in connection with (a) preventing, diagnosing, curing or alleviating a disease, ailment, defect or injury (*disease diagnosis*); (b)

[49] *Health Insurance Act 1973* (Cth) s9 and s10. The MBS sets subsidisation rates for GP, specialist and laboratory services and conditions under which tests will be funded such as pre-test counselling. See <www.mbsonline.gov.au> Group 7 Genetics.
[50] Access and funding of pharmaceutical interventions is controlled by the PBS.
[51] For a discussion of the complexity of state and territory and federal funding of CGT, see D Nicol, J Nielsen and V Dawkins, "The Genetic Diagnostic Testing Industry in Australia", in *D'Arcy v Myriad Genetics: The Impact of the High Court's Decision on the Cost of Genetic Testing in Australia* (Hobart: Centre for Law and Genetics Occasional Paper No. 9; 2018), Chapter 4. See overview of CGT and counselling services on a state and territory basis at <http://www.genetics.edu.au/Genetics-Services>.
[52] Personal communication with three of Australia's main private health insurers. An example of a non-subsidised test is for Cystic Fibrosis - <http://www.cysticfibrosis.org.au/vic/carrier-screening>.
[53] Therapeutic goods are defined as those 'represented in any way to be, or that are … likely to be taken to be for therapeutic use'. *Therapeutic Goods Act 1989* (Cth), s3 definition of therapeutic goods.

influencing, inhibiting or modifying a physiological process (*pharmacogenomics*); (c) testing the susceptibility of persons to a disease or ailment (*disease susceptibility*); and (d) influencing, controlling or preventing conception (*carrier status*).[54] While these definitions are quite broad, covering everything from tongue depressors to medicines and programmable pacemakers, they also clearly capture a range of CGT (as noted in parentheses).

The *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) provide the regulatory framework for in vitro medical devices defined as 'any instrument, apparatus, appliance, material or other article …used alone or in combination …' for 'diagnosis, prevention, monitoring, treatment or alleviation of disease' or 'control of conception' amongst others.[55] This is the *only* Australian legislation specifically capturing manufacture and supply of genetic tests used in CGT. Amendments to these regulations passed in 2010 established a regulatory regime specifically for in-vitro diagnostic medical devices (IVD medical devices), as a subset of medical devices.[56] IVD medical devices are pathology tests and related instrumentation such as reagents and specimen receptacles used to test human samples for clinical diagnosis or management.[57] IVD medical devices must be intended by manufacturers to be used *in vitro* for examination of specimens originating in the human body solely or principally for the purpose of providing information about a physiological or pathological state, a congenital abnormality or to monitor therapeutic measures.

The IVD medical device regulatory regime clearly captures genetic tests used in CGT, ensuring regulatory scrutiny prior to supply and ongoing monitoring. IVD medical devices are classified according a four-tier risk based system based on the risk posed to health from incorrect results (false positives or false negatives) with Class 1 representing the lowest and Class 4 the highest risk to individual or public health, such as infectious diseases.[58] The level of initial assessment and ongoing monitoring varies with risk classification.[59] Genetic tests used in CGT are deemed to be Class 3, indicating moderate public health and high personal health risk, where incorrect results could lead to significant detriment relative to patient management decisions, including the potential for patient distress.[60]

It is the responsibility of manufacturers of tests to ensure proper classification and adherence to specific requirements for their class of IVD medical devices, or face fines or imprisonment.[61] All therapeutic goods, including genetic tests used for CGT, must, as a general rule, be entered on the

[54] Ibid s3 definition of *therapeutic use* a, b, c and d.
[55] Ibid Division 2 s41BD definition of *medical device* (i) and (iv).
[56] In vitro refers to testing conducted outside an organism's body cf. in vivo conducted inside the body.
[57] *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) Dictionary 161 for full definition of IVD medical device.
[58] Ibid Schedule 2A for classification rules for IVD medical devices.
[59] The TGA is a member of the International Medical Devices Regulators Forum, a voluntary association of regulators whose aim is to facilitate international medical device regulatory harmonisation and convergence. See <http://www.imdrf.org> for full details.
[60] See NHMRC, *DNA Genetic Testing in the Australian Context: A Statement from the National Medical Health Research Council* (2012) <https://www.nhmrc.gov.au/about-us/publications/dna-genetic-testing-australian-context>. See also Australian Government, Department of Health, Therapeutic Goods Administration, 'IVD medical devices: Definitions & links' <https://www.tga.gov.au/ivd-medical-devices-definitions-links>.
[61] *Therapeutic Goods Act 1989* (Cth) Chapter 4 Division 2 s41BG. For non-compliance details see Chapter 4 Division 1 s41JB (3)

Australian Register of Therapeutic Goods (ARTG)[62] as a condition of legal supply. They must also comply with the essential principles and conformity assessment procedures as required by the Act.[63] It should be noted, however, that IVD medical devices falling within classes 1 to 3 that are developed or modified by a laboratory for use specifically within the laboratory (or laboratory network), with no intention to supply them outside of the laboratory (or laboratory network) are exempt from inclusion in the ARTG, subject to conditions.[64] As such, genetic tests developed and used in-house for the purpose of CGT are not required to be entered onto the ARTG, provided that they satisfy the required conditions.

The National Association of Testing Authorities (NATA)

To ensure analytic and clinical validity, Medicare and state/territory subsidisation is only provided for CGT deemed necessary by treating practitioners and conducted by approved pathologists at accredited laboratories.[65] NATA is recognised as the national authority for laboratory accreditation for all Medicare and state/territory CGT.[66] Figure 9 outlines the NATA accreditation process.

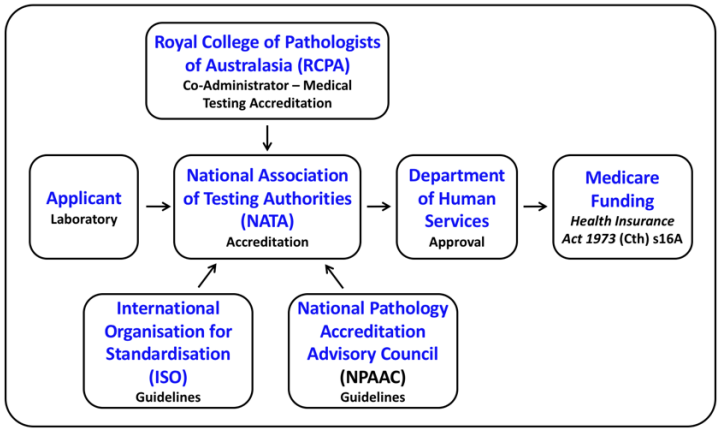

Figure 7: NATA accreditation

NATA and the Royal College of Pathologists of Australasia co-administer the accreditation process with NATA conducting all accreditation assessments and audits for medical testing laboratories, recommending approval to the Department of Health. Accredited laboratories conducting subsidised CGTs must comply with international standards, in particular ISO 15189, which outlines quality control measure for sample collection, analysis and training.[67] The National Pathology Accreditation Advisory Council develops and maintains standards required under The *Health Insurance (Accredited*

[62] See *Therapeutic Goods Act 1989* (Cth) Chapter 4 Part 4-5 for details.
[63] The essential principles are designed to ensure safety and performance with the conformity assessment procedures ensuring quality assurance and post-market monitoring including reporting of adverse events. *Therapeutic Goods Act 1989* (Cth) Chapter 4, Division 1 s41BA. See also *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth), Schedule 3 for details on conformity assessment.
[64] These are referred to as in-house tests. See *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth), Schedule 3, Part 6A.
[65] *Health Insurance Act 1973* (Cth): deemed necessary s16A(1a); approved pathology practitioner at approved laboratory s16A (2b) (a) and (b).
[66] See Memorandum of Understanding <http://www.nata.com.au/nata/phocadownload/publications/government/MoU-Cwlth-NATA.pdf>.
[67] See <http:www.iso.org>.

Pathology Laboratories – Approval) Principles 2002 for pathology accreditation.[68] NPAAC has also developed specific minimum standards for laboratories conducting human nucleic acid testing such as independent processing of duplicate samples for predictive CGTs.[69]

A complex web of protection … but can it keep pace with genetic discoveries?

This overview has identified *some* of the gatekeepers charged with keeping Australia's CGT patients safe. Protection is provided by a complex labyrinth of rules, regulations, policies and procedures influenced by intergovernmental agreements, international conventions and agreements, as well as professional oversight - all relying on the input of 'experts' in each field who it can be assumed find current levels of protection adequate for CGT. Gatekeepers and regulators, in attempting to keep up with advances in the field of genetics, have tended to 'graft on' new provisions to existing legislation and regulation rather than creating *sui generis* legislation, or have taken a 'wait and see' adverse outcomes approach.[70]

As CGT becomes increasingly integrated into healthcare, the rate of genetic discoveries accelerates, and general awareness of testing benefits develops, Commonwealth, state and territory governments will undoubtedly come under patient and practitioner pressure to offer and fund more CGT.[71] However, the complexity involved in offering more CGT has significant time and cost implications, potentially resulting in *protracted* translation of genetic discoveries into genetic testing, clinical applications and patient management.

Protections for DTCGT *Consumers*

As noted previously, the potential exists for the CGT and DTCGT spheres to merge when there is consumer or company-initiated engagement with healthcare. In both of these instances, once DTCGT *consumers* enter their doctors' surgeries, they enter as *patients*, afforded all of the protections previously discussed. But what if an individual chooses not to go to their doctor with their DTCGT test results? What protections are they afforded?

The commercial nature of DTCGT means it naturally falls within the ambit of consumer protection legislation. The definition of consumer has broadly been held to include what are often referred to as 'health consumers' (an increasingly popular term used for patients). For example, in *E v Australian Red Cross Society* (1991) 27 FCR 310, a hospital patient receiving nursing care was deemed a consumer. However, the nature of what DTCGT offers also brings it within the regulatory ambit of

[68] See NATA, "Medical Testing Field Application Document: Requirements for Accreditation" November 2013 for interpretation of NPAAC and ISO requirements <www.nata.com.au>.
[69] See Australian Government Department of Health, NPACC *Requirements for Medical Testing of Human Nucleic Acids* (2013) NPAAC <http://www.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-docs-nad2.htm>.
[70] See TGA explanation and procedures for reporting adverse incidents by both individuals and medical professionals <https://www.tga.gov.au/reporting-adverse-events> and <https://www.tga.gov.au/publication/reporting-medical-device-adverse-incidents>.
[71] The Royal College of Pathologists of Australasia, "New Nationwide Research on Genetic Testing in Australia" Media release, 22 February 2019 <https://www.rcpa.edu.au/News-and-Media-Releases/Media-Releases/Docs/New-nationwide-research-on-genetic-testing-in-Aust>.

the TGA. Other heads of law that might provide protection to Australia's DTCGT consumers such as general contract are beyond the scope of this article.

The role of the TGA

DTCGT for disease diagnosis, disease susceptibility, pharmacogenomics and carrier status is clearly captured by the therapeutic use definition in the *Therapeutic Goods Act 1989*.[72] DTCGT undertaken for these purposes falls within the distinct category of self-testing IVD medical devices, given that samples are collected by the individual at home or similar environment and, as a general rule, results are returned directly to them without the direct supervision of a health professional.[73] While most IVD medical devices used for self-testing are classified as Class 3, there are exceptions for tests that are not used for determining a 'serious condition, ailment or defect' (such as pregnancy testing or where testing is preliminary).[74] It is doubtful that DTCGT disclaimers that results are provided for 'research, information and education only' and not for diagnosis would exempt tests that are clearly related to serious conditions.[75] However, it could be argued that if a DTCGT provider merely reports on disease risk factors relative to the rest of the population, based on the set of markers within their database, this does not amount to diagnosis as such.

Importantly, the *Therapeutic Goods Act 1989* (Cth) provides that certain IVD medical devices cannot be certified or included on the ARTG on the basis that their use would be exclusively for an 'excluded purpose'. [76] The *Therapeutic Goods (Excluded Purposes) Specifications 2010* (Cth) (the Excluded Purposes Specification), which ceased in October 2020, provided direction on what constituted an excluded purpose with regard to self-testing IVD medical devices. Excluded purposes included those used to 'determine the presence of, or predict susceptibility to, diseases in humans', 'diagnose, aid in diagnosis or indicate the presence of a serious disease or condition...' and 'test for the presence of markers that are precursors to a serious disease or condition...' amongst others.[77] Serious disease was defined a disease that (a) may result in death or long-term disability; and (b) may be incurable or require major therapeutic interventions; and (c) must be diagnosed accurately, to mitigate the public health impact of the disease.'[78] Manufacturers seeking to list self-testing IVD medical devices on the ARTG so that they could be sold in Australia were required to certify they were not intended for any of these excluded purposes.[79]

While the it appears that the intention of the Excluded Purposes Specification was to make it illegal to sell DTCGT in Australia, there were points of ambiguity.[80] In particular, self-testing IVD medical devices did not fall within s41BE of the Act if 'the device is *also* to be used for another purpose'[81]

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[72] *Therapeutic Goods Act* (Cth) 1989, s3 definition of *therapeutic use* a, b, c and d.
[73] *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) Dictionary, s(b), 161.
[74] Ibid Schedule 2A s1.4(a) and (b).
[75] D Nicol and M Hagger, "Direct-to-Consumer Genetic Testing – A Regulatory Nightmare?" (2013) 198 *Medical Journal of Australia* 501.
[76] *Therapeutic Goods Act 1989* (Cth) s41BEA.
[77] Ibid 4(2)(b)(c) and (d). Cancer and myocardial infarction are mentioned specifically as to diagnosis exclusion and Pap smear and Prostate cancer tests for presence of markers exclusion.
[78] *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) Dictionary, 175.
[79] *Therapeutic Goods Act 1989* (Cth) ss41FD(ia) and 41FF(1A). s41BEA provides for specification, by legislative instrument, of excluded purposes.
[80] D Nicol and M Hagger, "Direct-to-Consumer Genetic Testing – A Regulatory Nightmare?" (2013) 198 *Medical Journal of Australia* 501.
[81] *Therapeutic Goods (Excluded Purposes) Specification 2010* (Cth) 4 s2.

including for Commonwealth or state/territory public health screening, for self-testing to monitor a diagnosed disease or condition, or for export only.[82] Given the bundled nature of DTCGT testing, many companies include trait and ancestry in addition to health-related testing which might arguably have exempted them from Excluded Purposes Specification prohibitions. It was also unclear what would be covered – sample receptacles, tests, results, or interpretation.

In 2019, the TGA undertook a review of the Excluded Purpose Specification[83] and in 2020 this Specification was replaced with the Therapeutic Goods (Medical Devices – Excluded Purposes) Specification 2020. The re-made Specification continues to prohibit a range of self-testing, including DTCGT. However, questions still remain about what aspects of DTCGT are included in the excluded purpose, and what the effect is of bundling health-related testing with other genetic reports on the requirement that the device is used 'exclusively' for the excluded purpose.

In sharp contrast to this prohibition on the supply of DTCGT for health-related purposes in Australia, overseas providers appear to be at liberty to deliver specimen receptacles to Australian consumers and consumers are at liberty to return their samples to those providers. Although it is an offence to import IVD medical devices that are not entered on the ARTG,[84] this offence does not apply where the IVD medical device is 'for use in the treatment of the importer, or a member of importer's immediate family, or for use in the in vitro examination of a specimen obtained from the importer or a member of the importer's immediate family'.[85] Devices exported for non-commercial purposes are similarly protected.[86] As such, not only can Australians purchase from DTCGT companies operating offshore, but such tests are exempt from the quality, safety and monitoring provisions required of CGTs. Instead, reliance has to be placed on the regulatory requirements imposed on DTCGT providers in their home jurisdictions.

The role of Australian consumer protection law

From 2011, *all* Australian consumers are provided with the *same* protections and *all* businesses are under the *same* obligations under the *Australian Consumer Law* (ACL), contained in Schedule 2 of the *Competition and Consumer Act 2010* (Cth).[87] The ACL replaced all existing national and state consumer laws,[88] an important step towards ensuring consistency of protection for *all* Australians.

The law reflects a combination of three general prohibitions and rule-based regulation prohibiting specific types of conduct.[89] General prohibitions against misleading and deceptive conduct, unconscionable conduct, and unfair contract terms provide Australian consumers with a basic 'safety-net' of minimum standards and businesses with the basic norms of conduct expected in

[82] Ibid 4 s3.
[83] Therapeutic Goods Administration, *Review of the Regulation of Certain Self-testing In Vitro Diagnostic Medical Devices (IVDs) in Australia* (Consultation Paper, September 2019) <https://www.tga.gov.au/consultation/consultation-review-regulation-certain-self- testing-ivds-australia>
[84] *Therapeutic Goods Act 1989* (Cth) s41BMI.
[85] *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) Schedule 4 part 1 (1.1).
[86] *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) Schedule 4 part 1 (1.2).
[87] Replacing the *Trade Practices Act 1974* (Cth).
[88] For example, *Fair Trading Act 1989* (Qld); *Door to Door Trading Act 1986* (Tas); *Lay by Sales Agreements Act 1963* (ACT).
[89] See JM Paterson and G Brody, "Safety Net Consumer Protection: Using Prohibitions on Unfair and Unconscionable Conduct to Respond to Predatory Business Models" (2015) 38 *Journal of Consumer Policy* 332.

Australia's marketplace. The ACLs general prohibitions were developed using a principles-based approach, allowing for flexible application without seeking to provide exhaustive lists. Being broad provisions, these general provisions lack clarity and certainly, thus requiring judicial interpretation. Rules-based regulation prohibiting specific behaviours such as pyramid selling provide certainty and clarity; however also provide unscrupulous businesses with opportunities to operate outside tightly defined definitions and prohibitions.

While the ACL does not specifically address genetics, its provisions *could* apply to DTCGT. However, whether they *would* apply would depend on ACCC enforcement action and/or judicial interpretation. The ACL's general provisions prohibiting misleading or deceptive conduct (s18)[90] and unfair contract terms (s23)[91] have general application so would apply to the DTCGT offering and its marketing messaging.[92]

Enforcing the ACL: The role of the ACCC

The Australian Competition and Consumer Commission (ACCC) and each of the state and territory consumer law agencies are responsible for enforcement of the ACL.[93] The ACCC is *selective* in the matters investigated and sectors where their own market analysis and education programs is undertaken, focusing on those with the greatest potential to harm the competitive process or result in widespread consumer or small business detriment. It rarely becomes involved in individual consumer-initiated complaints, leaving that to state and territory agencies or industry-specific regulators.

The ACCC's efforts are guided by its enduring priorities as well as those set annually. One of its ensuring priorities is the protection of vulnerable and disadvantaged consumers. Disadvantaged consumers have ongoing attributes or circumstances such as poor education while vulnerability may occur because of personal characteristics or context such as purchasing at times of emotional stress or where quality is difficult to ascertain. Given the DTCGT offering with its high credence qualities[94] and the emotive nature of DTCGT results, it is arguable all DTCGT consumers are vulnerable to a certain degree.

The ACCC uses a range of tools to ensure compliance, such as consumer education and enforces the non-compliance provisions in the ACL, determining which tools will achieve the best results for the community while managing risk and assessing proportional remedies. The ACCC operates

[90] Misleading or deceptive conduct is determined using an objective test considering ordinary or reasonable prospective buyers and focuses on effect not intent. See *Campomar Soiedad Limitada v Nike International Ltd* (2000) 202 CLR.
[91] Unfair contract terms are considered void in standard form contract such as those used by DTCGTs. 'Unfair' terms cause significant imbalances in parties' rights and obligations, are not reasonably necessary to protect legitimate interests of advantaged parties, and could cause detriment (financial or otherwise). DTCGT contracts are non-negotiable by consumers.
[92] Chapter 3, Part 3-1 s29(1)a, b and l, s33 and s34 cover misleading conduct concerning suitability for purpose and quality of any goods or services. This might not just cover marketing messages but possibly also results.
[93] The ACCC is an independent Commonwealth statutory authority charged with enforcing all aspects of the *Competition and Consumer Act 2010* (Cth), promoting competition, fair trading and regulating national infrastructure.
[94] Credence qualities cannot be evaluated by consumers pre or post consumption e.g. DTCGT consumers cannot determine analytic and clinical validity, accepting test results 'on faith'.

transparently, publicising all enforcement activities and, while independent, is ultimately accountable to Parliament and the courts.

Unlike the TGA, whose jurisdiction is limited to Australia, the ACL has extraterritorial effect given inclusion of 'between Australia and places outside Australia' in its definition of 'trade and commerce',[95] and the ACCC's international agreements with several overseas regulators.[96] Even though Australia has legislation concerning enforcement of foreign judgements, the lack of consistent rules for determining jurisdiction suggest it may not be straightforward to enforce Australian protections for online DTCGT activities.[97]

There had not been a lot of enforcement activity in Australia's DTCGT space, but what there has been shows a willingness to address DTCGT claims and enforce the ACL.[98] For example, in 2016, the ACCC accepted an administrative undertaking from pharmacy chain Chemmart regarding 'representations regarding the effectiveness of a myDNA genetic test in identifying an individual's response to certain drugs'. In response, Chemmart withdrew all promotional materials and agreed to refrain from making future statements that might mislead consumers.[99]

The ACL is designed to protect *all* consumers in *all* marketplace activities whether what is purchased is a chocolate bar, haircut or insurance policy, with mandatory consumer guarantees assumed to provide adequate baseline protection.[100] Consumers themselves are assumed able to judge if there is an issue e.g. item ordered as blue arrives as red. And it is consumers themselves who are required to initiate complaints and remedy procedures. Standard remedies under these guarantees of 'replace, repair or refund' are relatively straightforward to calculate, and deemed adequate to return consumers to their original state as expected in torts e.g. a $5 refund for an item costing $5.[101]

However, the core question remains as to whether the protections afforded Australians under the ACL are sufficient if the purchase is for DTCGT for breast cancer or Alzheimer's? DTCGT operates outside the traditional healthcare protections afforded CGT patients. As individuals must self-interpret, DTCGT also has the potential for negative consumer outcomes, especially so if DTCGT

[95] *Australian Consumer Law* Schedule 2, Schedule 2, Chapter 1 Section 2 Definitions.
[96] ACCC, *Treaties and Agreements* (2013) <https://www.accc.gov.au/about-us/international-relations/treaties-agreements>.
[97] See Australia's *Foreign Judgments Act 1991* (Cth). See *Dow Jones & Company Inc v Gutnick* (2002) 194 ALR 433 (place of damage = place of publication), *Calder v Jones* 465 US 783 (1984) (effects plus website targeting) and *Zippo Manufacturing v Zippo.com* 952 F Supp 1119 (WD Pa 1997) (sliding scale of interactivity).
[98] See, for example, Katherine Fenech, 'DNA test scam warning' *WA today* 5 January 2011 <https://www.watoday.com.au/national/western-australia/dna-test-scam-warning-20110105-19fjz.html>.
[99] ACCC, EBOS Group Ltd, on behalf of its subsidiary Symbion Pty Ltd, the owner of the Chemmart pharmacy franchise, MR 166/16 <https://www.accc.gov.au/media-release/chemmart-agrees-to-improve-its-promotion-of-%E2%80%9Cmydna%E2%80%9D-tests>.
[100] While businesses cannot opt-out of these consumer guarantees, they are free to supplement e.g. 'change of mind' return policies.
[101] For example, in tort the purpose of compensatory damages is to place the plaintiff as far as possible in the position they would have been had the wrong not occurred. See *Livingstone v Rawyards Coal Co* (1880) 5 App Cas 25.

results are used to make significant treatment, prevention and lifestyle decisions.[102] Additionally, the legal duty to take reasonable care to avoid causing emotional distress is well established in tort, with accidental infliction, if negligent, sufficient to support a cause of action in the courts.[103] Emotional distress, however, is much more difficult for consumers to determine and is a lot less straightforward to both quantify and determine remedies by regulators.

Conclusion

This article modelled the pathways, processes and protections afforded to *patients* pursuing CGT and *consumers* pursuing DTCGT. While the paradigm shift from *medical* to *consumer* is obvious, modelling identified the potential for CGT and DTCGT pathways and processes to merge, which would mean that some of the protections provided for CGT would also applying to DTCGT in Australia. When these pathways and processes merge through consumer or company-initiated engagement with healthcare, individuals' roles change from *consumer* to *patient*, providing traditional protections afforded in the CGT pathway, with a primary focus on safety, analytical and clinical validity and clinical utility.

In the commercial space, protections focus primarily on either marketing or distribution of tests. The analytic and clinical validity of DTCGT is dependent on the science the day, with clinical validity the responsibility of laboratories, which may or may not be accredited. Clinical utility is the responsibility of individuals who firstly decide to purchase tests and ultimately interpret results determining what, if anything, they will do in response. During the DTCGT process, consumers make significant assumptions – assuming tests conducted and results provided are accurate, and that they have interpreted and actioned them in appropriate ways. Self-interpretation of genetic results, however, has the potential to generate 'false positives' and 'false negatives' each leading to potential personal harm.

Modelling of the CGT space suggests a complex web of regulators, regulations, gatekeepers and advisory parties, with expert opinion sought at every juncture, all focused on ensuring the efficacy of genetic testing, laboratory analysis and transmission of results and action plans to patients. Such a complex web, however, is both time and cost inefficient, leading to potential protracted translation of genomic breakthroughs into therapeutic tests and treatments. Modelling of the DTCGT space, in contrast, reveals the main gatekeeper to be the commercial imperative – adding tests to bundled offerings as soon at the science is available. This commercial imperative is precisely what they *should* and *would* do – in the absence of anything saying they *can't* ... regardless of whether tests have ultimately been validated and proven useful.

At its core, the overarching issue relative to consumer protection is DTCGT's commercial nature. Unlike the time and resource intensive processes designed to protect CGT patients, one of DTCGT's key strengths is its ability to move quickly, driven by its players' commercial imperative of profitability. However, while the CGT processes have the potential for *protracted translation* of

[102] See N Bansback et al, "The Effect of Direct-to-Consumer Genetic Tests on Anticipated Affect and Health Seeking Behaviors: A Pilot Survey" (2012) 16 *Genetic Testing and Molecular Biomarkers* 1165.
[103] The tort action is for wilful infliction of nervous shock but requires proof of actual damage such as psychiatric illness and is available under limited circumstances − *Wilkinson v Downton* [1897] 2 QB 57.

genetic discoveries into accessible genetic testing and flow-on treatment, DTCGT has the potential for *premature translation*. 'As soon as someone publishes a paper with an association, someone can start testing the next day', meaning time from lab to market may be faster than the rigours of the science can be tested and replicated.[104]

DTCGT companies have also structured their operations to limit their responsibility well beyond disclaimers and contract terms. The genetic associations and markers used in developing tests derive from the peer-reviewed science of the day, placing responsibility for analytic and clinical validity on scientists. In many instances, although DTCGT companies make use of the same genome-wide association studies (GWAS) and scientific evidence as CGT providers, they move on their results much faster. Provided accredited laboratories are used, these labs bear responsibility for clinical validity. In terms of interpretation and resulting actions, this is left to individuals or the medical profession if individuals choose to share. As such individuals and/or their doctors bear responsibility for determining clinical utility.

In Australia, the TGA has taken the step of imposing a significant bar on the provision of DTCGT where it is exclusively for disease-related purposes. However, there are still some remaining questions about whether the full gamut of health-related DTCGT services are included in this bar, and about how bundling of services plays out vis a vis exclusivity. Moreover, the fact remains that the provision of DTCGT services from overseas providers is beyond the jurisdictional oversight of the TGA. The ACL is not tied to such jurisdictional restrictions and may provide useful recourse to consumers should the need arise. However, given that consumers are masters of their own fate when it comes to DTCGT, genetics education ultimately remains the most powerful regulatory tool.

When Justice Windeyer referred to 'Law, marching with medicine but in the rear and limping a little', it was well before the completion of the Human Genome Project in 2003.[105] However, add science and technology to medicine and the quote still has resonance today. Law, by its very nature advances slowly in a generally reactive rather than proactive manner.[106] Science and technology, especially in genetics, moves fast and at a rapidly accelerating pace in today's intensely competitive commercial space.

While this article focuses on the medical and commercial spheres relative to health-related genetic testing, it can also function as a lens with which to view what will be undoubtedly be continued incursions into the medical sphere by commercial players as the public healthcare sector struggled under escalating burdens.

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[104] B Kuehn, "Risks and Benefits of Direct-to-Consumer Genetic Testing Remain Unclear' (2008) 300 *Journal of the American Medical* Association, 1503, 1503. Quoting Lawrence Brody then of the National Human Genome Research Institute.

[105] *Mount Isa Mines Ltd v Pusey* (1970) 125 CLR 383 at 395.

[106] See James Farrell, ' Slow, expensive, complicated legal system must be improved' *The Conversation* April 10, 2014 <http://theconversation.com/slow-expensive-complicated-legal-system-must-be-improved-25382>; Rosalind Croucher, 'A window on law reform for government lawyers', *Australian Law Reform Commission* August 28, 2012 <https://www.alrc.gov.au/news-media/2011-2012/window-law-reform-government-lawyers>.