



**International Society  
of Nurses in Genetics**

## **Position Statement: Direct-to-Consumer Genetic/Genomic Testing (DTC-GT)**

### **Purpose**

This position statement is intended to provide nurses, as well as other healthcare providers, policymakers and the general public, with information and key factors relevant to informed decision-making about the use of Direct-to-Consumer health-related genetic/genomic testing (DTC-GT). The focus of this position statement is on DTC health-related genetic/genomic testing such as disease risk assessment, drug response genetic typing, and carrier testing for recessive conditions. It does not address reasons outside the parameters of health (e.g. ancestry analysis, wellness and trait characteristics, and paternity testing). This document reflects current knowledge and policies, but given the rapid evolution of applied genomics it also highlights issues relevant to future directions in healthcare.

### **Background**

Direct-to-consumer genetic/genomic testing is defined as marketing and/or provision of genetic/genomic tests directly to the public. Currently, DTC-GT for health-related reasons includes testing that detects variants in one or several genes (e.g. for a specific condition), targeted genome panels (e.g., cancer syndromes), and whole genome scans or sequencing (e.g., for broad-based risk assessments based on many genetic variants).

Within any clinical healthcare setting, there are sets of safeguards and regulations in place which aim to protect patients from harm. Regulation of clinical laboratory testing, and in particular DTC-GT, varies according to national policy and laboratory standards (Borry et al, 2012; Rafiq, Ianuale, Ricciardi & Boccia, 2015), as do consumer protections that oversee marketing to the public (National Institutes of Health, 2016). Safeguards include laboratory quality standards, provider involvement, restrictions on tests offered, clinical standards, informed consent and marketing regulations. Because DTC-GT companies differ in their approaches to health care provider involvement, genetic counseling services, testing methodologies and foci, and provision of information, these factors and safeguards should be assessed before proceeding with genetic testing (European Academies of Science Advisory Council, 2012; National Institutes of Health, 2016; National Pathology Accreditation Advisory Council, 2014). Regardless of the national strategy for provision, the DTC-GT environment shifts the driving force behind access to personal genetic information from the healthcare professional to the consumer (American College of Medical Genetics and Genomics [ACMG], 2016; American College of Obstetricians and Gynecologists [ACOG], 2008).

## **Benefits, Risks and Limitations**

Almost a decade since the inception of DTC-GT, a considerable amount of research has investigated the potential benefits and harms of this approach to testing. For example, a systematic review of the literature on direct-to-consumer genetic/genomic testing research identified 118 relevant studies and summarized findings into five categories (knowledge, attitude and perception of DTC-GT; impact of risk information on users; opinion of healthcare providers; content of DTC-GT websites; and scientific evidence and clinical utility of testing; [Covolo, Rubinelli, Ceretti, Gelatti, 2015]). Nonetheless, consensus on the impact of DTC-GT is still evolving and therefore definitive evaluations would be premature. In this context, providers of healthcare services should be up-to-date with potential benefits, risks and limitations of DTC-GT in order to promote informed professional and individual health-related decisions. The main points, and other relevant research findings, are presented below.

### **Potential Benefits**

Direct-to-consumer genetic/genomic testing empowers individuals to pursue genome testing and information independently, make autonomous decisions, and may support personal responsibility for health, healthcare and lifestyle choices (Wessel, Gupta, & de Groot, 2016). Public views and research exist that support DTC-GT as an individual right or freedom, whether it is pursued and used for health-related purposes or for personal reasons, values and utility (Farrell, Holaday, Evans, & Roberts, 2016; Su, Borry, Otte, & Howard, 2013). DTC-GT also provides consumers the privacy and control of personal genetic health information, including its entry into individual medical records (Carere et al., 2014; van der Wouden et al., 2016).

Direct-to-consumer genetic/genomic testing and marketing may provide public benefit through education about health risks, the relative contributions of genetics and/or environment to risks and their management, and the value of burgeoning genetic discoveries (Delaney & Christman, 2016; Turrini & Prainsack, 2016). In some cases, without the DTC route, genome testing may not otherwise be available. For example, in countries whose healthcare system restricts testing to persons with a positive family history, prospective parents not meeting eligibility criteria could not pursue carrier testing to identify potential risk to a child (Jackson, Goldsmith, & Skirton, 2014). The DTC-GT route is also valued by individuals concerned about medications and pharmacogenomic risk, as well as by persons with a positive family history that does not meet clinical criteria for testing (e.g., cancer in one relative).

### **Potential Risks and Limitations**

Informed consent: Genome testing and the interpretation of genetic test results are highly complex, technical and limited by the extent of knowledge for each specific variation in any of the thousands of human genes. The consumer should be fully informed regarding the purpose, extent and intent of genome testing (i.e., for diagnosis, carrier status, risk assessment, or screening), its scientific validity and clinical utility, and what the tests can and cannot determine about their health (ACMG, 2016). Concerns have been raised that online resources for consumers tend to lack essential information for proper informed consent (Niemi & Howard, 2016) and that written information is at a level inaccessible to most of the general public (Lachance, Erby, Ford, Allen, & Kaphingst, 2010). Furthermore, results may identify rare variants, or those of uncertain significance, with little available scientific or research data, thus posing challenges to knowledge, understanding and interpretation by individuals and healthcare providers (Jackson et al., 2014).

Misinterpretation of Test Results: Pursuit of genome testing, and information about genetic test results, delivered without consultation and advice of a certified and/or licensed healthcare provider with genomics expertise, may create additional risks for the consumer. These risks include misinterpretation of information (results), or distortion of the extent of its significance to the overall health or risk status of the person tested. Misinterpretation may lead to failure to engage in preventive behaviors because the risk is not adequately presented or understood, or alternatively, inappropriate and potentially harmful health-related action. However, these concerns may be more theoretical than actual based on current research findings that persons who pursue genome testing tend to be a more educated group and understand the limitations of genome test results (i.e., representing projected risk) as opposed to results being diagnostic or predictive (Roberts & Ostergran, 2013).

Psychosocial Concerns: The potential for psychological harm has also been raised. Although research in adults suggests some negative psychological impacts (e.g., anxiety) in some genome testing users, these effects appear to be short-lived (no significant differences by one year after testing), and personal value has been attributed to testing, regardless of results (Covolo et al., 2015). While the impact of genome testing on children is being explored, unless medical indications exist, it is generally recommended to defer testing until adulthood (Ross et al., 2013).

Confidentiality, Privacy & Integrity of Specimens: Confidentiality and integrity of genomic information is also a concern. Documentation, policies, regulations and security measures for the protection and privacy of consumers and their test samples, for assuring the quality and reliability of testing, for the handling, storage and reuse of samples, and for marketing, is imperative (ACMG, 2016). Appropriate oversight is being addressed, yet with recognized gaps (Borry et al., 2012; U.S. Food & Drug Administration, 2015, 2017).

## **The Role of Nurses in Direct-to-Consumer Genetic Testing**

Rapidly emerging genetic/genomic knowledge and technology relate to every area of health and disease. The International Society of Nurses in Genetics (ISONG) and American Nurses Association (ANA, 2016) support the integration of genetics/genomics nursing care at the basic and advanced-practice level in their joint publication *Genetics/genomics nursing: Scope and standards of practice*. Nurses recognize the influence of genes on the health of individuals and have a professional responsibility to update knowledge when involved in providing basic genetic services (Badzek, Henaghan, Turner, & Monsen, 2013).

Since consumers are now able to access some forms of genetic testing without previous discussion with health care providers nurses may be engaging with consumers as they contemplate DTC-GT or after they receive results. The dissemination of current research and information through nursing journals is vital to an informed nursing workforce in practice, education and research.

Therefore it is the position of ISONG that the professional nurse should:

1. Be informed about genetic/genomic testing and associated health, ethical, legal and social issues, together with updating knowledge of genetic evaluation, technology and services related to one's professional scope of practice.

2. Be receptive to open communication with consumers about DTC-GT to explore their expectations regarding the personal value of genomic information and its intended use.
3. Foster consumer understanding by educating about: (a) the role of genes and environment in health and disease, (b) the importance of family history, and (c) genetic risk assessment, benefits and limitations of testing, and risk reduction, disease prevention and health promotion options.
4. Facilitate access to appropriate genetic counseling services for those with unaddressed genetic risks or concerns, and for those planning to make changes in health behavior based upon information provided by DTC-GT.

In addition to the above recommendations, nurse educators are advised to provide a strong foundation in genetics/genomics at the undergraduate level and to foster more advanced genetics/genomics knowledge, understanding, assessment skills, and awareness of ethical, legal, professional, regulatory and policy issues at the graduate level. Nurses who are prepared at an advanced level should be able to integrate these considerations into healthcare practice, including for persons interested in or using DTC-GT. Nurse researchers are encouraged to investigate the physical, psychological, social, and economic effects of DTC-GT and to identify strategies that foster optimal delivery of information and genetic services.

## References

- American College of Medical Genetics and Genomics. (2016). Direct-to-consumer genetic testing: A revised position statement of the American College of Medical Genetics and Genomics. *Genetics in Medicine, 18*(2), 207-208. doi:10.1038/gim.2015.190.
- The American College of Obstetricians and Gynecologists Committee on Genetics & Committee on Ethics. (2008). ACOG Committee Opinion No. 409: Direct-to-consumer marketing of genetic testing. *Obstetrics & Gynecology, 111*(6), 1493-1494. doi: 10.1097/AOG.0b013e31817d250e.
- Badzek, L., Henaghan, M., Turner, M., & Monsen, R., (2013). Ethical, legal, and social issues in the translation of genomics into health care. *Journal of Nursing Scholarship, 45*(1), 15-24. doi: 10.1111/jnu.12000.
- Borry, P., van Hellemond, R. E., Sprumont, D., Jales, C. F., Rial-Sebbag, E., Spranger, T. M., . . . Howard, H. (2012). Legislation on direct-to-consumer genetic testing in seven European countries. *European Journal of Human Genetics, 20*(7), 715-721. doi:10.1038/ejhg.2011.278.
- Carere, D.A., Couper, M.P., Crawford, S.D., Kalia, S.S., Duggan, J.R., Moreno, T.A., . . . the PGen Study Group. (2014). Design, methods, and participant characteristics of the Impact of Personal Genomics (PGen) Study, a prospective cohort study of direct-to-consumer personal genomic testing customers. *Genome Medicine, 6*(12), 96. doi:10.1866s13073-014-0096-0.
- Covolo, L., Rubinelli, S., Ceretti, E., & Gelatti, U. (2015). Internet-based direct-to-consumer genetic testing: A systematic review. *Journal of Medical Internet Research, 17*(2), e279. doi:10.2196/jmir.4378.

- Delaney, S. K., & Christman, M. F. (2016). Direct-to-consumer genetic testing: Perspectives on its value in healthcare. *Clinical Pharmacology and Therapeutics*, *99*(2), 146-148. doi: 10.1002/cpt.287.
- European Academies of Science Advisory Council. (2012, June 29). *Direct-to-consumer genetic testing for health-related purposes in the European Union: The view from EASAC and FEAM*. Retrieved from [www.easac.eu/fileadmin/Reports/Easac\\_12\\_DTC\\_Web.pdf](http://www.easac.eu/fileadmin/Reports/Easac_12_DTC_Web.pdf).
- Farrell, C. E. K., Holaday, B., Evans J., & Roberts, J. S. (2016). Individual perceived health and health values as motivators of health action and priorities in DTC genome testing users: Insights with potential relevance to personalized healthcare. Manuscript submitted for publication.
- International Society of Nurses in Genetics and the American Nurses Association. (2016). *Genetics/genomics nursing: Scope and standards of practice* (2<sup>nd</sup> ed.). Silver Spring, MD: Nursesbooks.org.
- Jackson, L., Goldsmith, L., & Skirton, H. (2014). Guidance for patients considering direct-to-consumer genetic testing and health professionals involved in their care: Development of a practical decision tool. *Family Practice*, *31*(3), 341-348. doi: 10.1093/fampra/cmt087.
- Lachance, C. R., Reby, L. A. H., Ford, B. M., Allen, V. C. & Kaphingst, K. A. (2010). Informational content, literacy demands, and usability of websites offering health-related genetic tests directly to consumers. *Genetics in Medicine*, *12*, 304–312. doi:10.1097/GIM.0b013e3181dbd8b2
- National Institutes of Health, National Human Genome Research Institute. (2016). *Regulation of genetic tests*. Retrieved from <https://www.genome.gov/10002335/regulation-of-genetic-tests/>
- National Pathology Accreditation Advisory Council. (2014). *Provision of direct to consumer genetic tests guiding principles for providers*. Retrieved from [https://www.health.gov.au/internet/main/publishing.nsf/Content/F57C68E5E41DC946CA257EEC000396F2/\\$File/Genetic%20DTC%20Guiding%20Principles%20-%20May%202014](https://www.health.gov.au/internet/main/publishing.nsf/Content/F57C68E5E41DC946CA257EEC000396F2/$File/Genetic%20DTC%20Guiding%20Principles%20-%20May%202014).
- Niemiec, E. & Howard, H.C. (2016). Ethical issues in consumer genome sequencing: Use of consumers' samples and data. *Applied & Translational Genomics*, *8*, 23-30. doi: 10.1016/j.atg.2016.01.005. eCollection 2016 Mar PMID:27047756
- Rafiq, M., Ianuale, C., Ricciardi, W., & Boccia, S. (2015). Direct-to-consumer genetic testing: a systematic review of European guidelines, recommendations, and position statements. *Genetic Testing & Molecular Biomarkers*, *19*(10), 535-547. doi: 10.1089/gtmb.2015.0051.
- Roberts, J.S. & Ostergren, J. (2013). Direct-to-consumer genetic testing and personal genomics services: A review of recent empirical studies. *Current Genetics Medicine Reports*, *1*(3), 182–200.

Ross, L. F., Saal, H. M., David, K. L., Anderson, R. R. & the American Academy of Pediatrics; American College of Medical Genetics and Genomics. (2013). Technical report: ethical and policy issues in genetic testing and screening of children. *Genetics in Medicine, 15*, 234–245. doi:10.1038/gim.2012.176

Su, Y., Borry, P., Otte, I. C., & Howard, H. C. (2013). "It's our DNA, we deserve the right to test!" A content analysis of a petition for the right to access direct-to-consumer genetic testing. *Personalized Medicine, 10*(7), 729-739. <https://doi.org/10.2217/pme.13.69>.

Turrini, M., & Prainsack, B. (2016). Beyond clinical utility: The multiple values of DTC genetics. *Applied and Translational Genomics, 8*, 4-8. <https://doi.org/10.1016/j.atg.2016.01.008>.

US Food and Drug Administration [FDA]. (2015). *FDA permits marketing of first direct-to-consumer genetic carrier test for Bloom syndrome* [Press release]. Retrieved from <http://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm435003.htm>.

US Food and Drug Administration [FDA]. (2017). *FDA allows marketing of first direct-to-consumer tests that provide genetic risk information for certain conditions* [Press release]. Retrieved from <https://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm551185.htm>.

van der Wouden, C. H., Carere, D. A., Maitland-van der Zee, A. H., Ruffin, M. T., Roberts, J. S., Green, R. C.; & Impact of Personal Genomics Study Group. (2016). Consumer perceptions of interactions with primary care providers after direct-to-consumer personal genomic testing. *Annals of Internal Medicine, 164*(8), 513-522. doi: 10.7326/M15-0995.

Wessel, J., Gupta, J., & de Groot, M. (2016). Factors motivating individuals to consider genetic testing for type 2 diabetes risk prediction. *PLoS One, 11*(1), e0147071. doi:10.1371/journal.pone.0147071

**Original position statement prepared by** the Ethics and Public Policy Committee and approved by the ISONG Board on September 3, 2009.

**Revised by** Carolyn D. Farrell, PhD, MS, RN, WHNP-BC, CGC, Cheedy Jaja, PhD, MPH, MN, MSN, PMHNP-BC, RN, Heather Skirton, PhD, MSc, RGN, RGC, QMW, Dip Couns, PGCAP, Marie Twal, CPNP, DrPH and Christopher H. Wade, PhD, MPH (May 30, 2017).

**Approved by** the Executive Board of the ISONG on October 3, 2017.