



Public Understanding of Risk in Direct-to-Consumer Genetic Testing

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Abstract

Direct-to-consumer genetic testing has expanded public access to information concerning genetic ancestry, carrier status, selected health predispositions, traits, and susceptibility to complex diseases. The accessibility of such testing changes the conventional pathway of genetic information because consumers may encounter probabilistic health-risk estimates without receiving pre-test or post-test interpretation from a genetics professional. This raises an important public-health question: whether consumers understand that a reported genetic predisposition represents one component of disease risk rather than a deterministic prediction of future illness. Earlier research demonstrated that members of the general public can perceive direct-to-consumer genetic reports as easy to understand while nevertheless interpreting their meaning incorrectly. Leighton, Valverde, and Bernhardt found significant differences between public and genetic-counselor interpretations of sample genetic-risk reports, particularly concerning the clinical meaning and usefulness of reported risks.

The present study develops a simulation-based framework for examining public understanding of genetic risk. Because no original participant dataset was supplied, 600 synthetic consumer profiles were modeled across three interpretation conditions: report-only disclosure, contextualized plain-language explanation, and genetics-professional-supported interpretation. Risk comprehension was conceptualized through understanding of probability, recognition of multifactorial disease causation, distinction between relative and overall risk, interpretation of negative results, and recognition of the need for confirmatory clinical testing when appropriate. Numerical findings are explicitly simulated and must not be treated as population estimates.

The study argues that public understanding of direct-to-consumer genetic testing should not be measured simply by asking whether reports appear clear or useful. Effective risk literacy requires the ability to recognize uncertainty, integrate family history and environmental influences, understand that tested variants may represent only part of relevant genetic variation, and resist both fatalistic interpretation of increased-risk findings and inappropriate reassurance from lower-risk findings. The paper concludes that direct-to-consumer genetic-risk communication should combine transparent limitations, absolute-risk context where scientifically justified, visual and plain-language explanations, access to qualified professional interpretation, and explicit guidance against making major medical decisions from unconfirmed consumer-generated results.

Keywords: direct-to-consumer genetic testing; genetic risk; risk communication; genetic literacy; public understanding; probabilistic reasoning; genetic counseling; consumer genomics



1. Introduction

Direct-to-consumer genetic testing has altered the relationship between the public and genetic information by allowing individuals to obtain certain genetic results without following the conventional sequence of referral, clinical testing, professional interpretation, and genetic counseling. Consumer genomic services have been used for ancestry exploration, health-related information, carrier status, traits, and assessments of susceptibility to selected diseases. Reviews of the field have consistently noted that this expanded autonomy is accompanied by concerns about the clinical validity of some interpretations, consumer understanding, privacy, psychological effects, and downstream healthcare use. The central public-health issue is therefore not whether people should have access to their own genetic information, but whether risk information is communicated in a form that supports proportionate and informed interpretation.

Health-related genetic results are especially challenging because most common diseases are not controlled by a single genetic variant operating independently of all other influences. Disease development may reflect interactions among multiple genetic factors, age, behavior, environment, family history, comorbid conditions, and other exposures. A consumer receiving an “increased genetic risk” result can therefore make at least two major reasoning errors. The first is genetic determinism, in which increased susceptibility is interpreted as equivalent to eventual disease. The second is false reassurance, in which the absence of a tested risk variant is interpreted as evidence that disease cannot occur. Consumer genetic-risk reports may also differ in which variants they include, and a test result may not capture an individual's complete genetic or clinical risk profile. These limitations make communication of uncertainty central to responsible interpretation.

Empirical evidence demonstrates that perceived report clarity does not guarantee accurate understanding. Leighton, Valverde, and Bernhardt presented members of the public with sample direct-to-consumer genetic-risk reports relating to colorectal cancer, heart disease, and skin cancer and compared their responses with those of genetic counselors. Although many members of the public described the reports as easy to understand, their interpretations frequently differed from those of genetics professionals, and they tended to perceive the reports as more helpful. This finding is particularly important because conventional usability questions such as “Was the result easy to understand?” may substantially overestimate actual risk comprehension.

Subsequent research has added nuance rather than confirming widespread catastrophic effects. Bloss, Schork, and Topol found that genome-wide consumer profiling did not produce major short-term changes in psychological health, dietary fat intake, or exercise among their study participants, showing that receipt of genetic-risk information does not inevitably produce severe anxiety or large behavioral reactions. Reviews and later consumer studies likewise suggest that many users perceive testing as interesting or personally useful and that severe regret is uncommon among self-selected consumers. Roberts and colleagues reported that many participants believed their results could influence future health management, whereas only small proportions reported regret. The important problem is therefore not that every consumer reacts badly but that specific misunderstandings can become consequential when genetic information is used to guide screening, treatment, or family decisions.

A further challenge arises when consumers download raw genetic data and submit those data to third-party interpretation services. Tandy-Connor and colleagues clinically reassessed variants identified



through direct-to-consumer raw data and found that 40% of the variants examined in their referral sample were false-positive calls; some variants described as increased risk were subsequently classified as benign by clinical laboratories. The study involved a selected clinical-confirmation sample and therefore should not be interpreted as a 40% false-positive rate for all direct-to-consumer tests. Nevertheless, it demonstrates why risk understanding must incorporate test scope, analytical confirmation, and the distinction between consumer raw data and clinically validated genetic findings. Against this background, the present simulation examines how different forms of interpretation support may influence public understanding of genetic-risk information.

2. Objectives of the Study

2.1 To Assess Public Comprehension of Probabilistic Genetic Risk

The first objective is to assess the conceptual dimensions required for accurate understanding of direct-to-consumer genetic-risk reports. These dimensions include recognizing that increased genetic susceptibility does not establish that disease will occur, understanding that lower genetic risk does not eliminate disease possibility, distinguishing the effect of a tested variant from overall clinical risk, and recognizing the contribution of non-genetic determinants.

The study specifically treats objective comprehension as distinct from perceived clarity. This distinction follows evidence showing that members of the public can believe that genetic reports are easy to understand while nevertheless interpreting their implications differently from genetics professionals.

2.2 To Examine Common Forms of Genetic-Risk Misinterpretation

The second objective is to examine several theoretically important interpretation errors: genetic determinism, false reassurance, confusion between relative and absolute risk, overgeneralization from one tested variant, and failure to recognize uncertainty or population-specific limitations.

These errors are clinically relevant because a misunderstood result can potentially influence screening expectations, healthcare utilization, lifestyle decisions, or requests for unnecessary medical intervention. Earlier consumer research has examined links between perceived genetic risk and subsequent healthcare use, while systematic reviews have assessed possible psychological, behavioral, and medical-consumption consequences of direct-to-consumer testing.

2.3 To Evaluate the Role of Contextual Explanation and Professional Interpretation

The third objective is to determine whether contextual risk communication and genetics-professional support represent plausible mechanisms for improving comprehension. Contextual explanation includes plain-language descriptions of probability, explicit statements concerning multifactorial causation, explanation of what variants were and were not tested, and differentiation between genetic contribution and overall disease risk.

Professional interpretation is modeled as the most supported condition because randomized evidence has shown that genomic counseling can improve objective understanding of genetic contributions to common complex diseases. The objective is not to argue that every consumer test must



require face-to-face counseling, but to identify the interpretive functions that need to be available when results are medically consequential or difficult to understand.

3. Review of Literature

3.1 Consumer Understanding, Genetic Literacy, and Risk Perception

Research into public understanding of direct-to-consumer genetic testing has repeatedly identified a gap between access to information and capacity to interpret that information. Leighton and colleagues' comparative study remains particularly influential because it directly tested interpretation of realistic risk reports rather than relying solely on attitudes toward genetic testing. Members of the public and genetic counselors interpreted the same reports differently, even though public participants frequently perceived them as straightforward. This discrepancy indicates that subjective confidence may be a poor proxy for genomic literacy.

Risk communication itself is cognitively demanding. Genetic-risk reports may contain percentages, comparisons with population averages, terms such as “elevated,” “typical,” or “reduced,” and explanations of variants with modest effect sizes. A person may correctly understand that a reported risk is “higher than average” while failing to appreciate whether the difference is clinically large or small. Earlier work examining literacy demands of direct-to-consumer genetic-testing websites showed that information complexity and usability need careful attention. Leighton and colleagues also grounded their study in the established risk-communication literature concerning numeracy and differences between verbal, numeric, and visual presentations.

Public awareness does not necessarily imply detailed knowledge. Agurs-Collins and colleagues reported that only a portion of surveyed adults were aware of direct-to-consumer genetic tests, with media and the Internet functioning as common sources of awareness. A later review of European public perspectives concluded that interest in direct-to-consumer testing could coexist with relatively low knowledge about the tests. These findings suggest that consumer markets may expand more rapidly than the public's understanding of variant interpretation, predictive limitations, privacy, and clinical relevance.

3.2 Risk Results, Behavioral Response, and Clinical Consequences

One early concern surrounding personal genomic testing was that genetic susceptibility information could create severe anxiety or encourage inappropriate lifestyle or healthcare changes. Evidence has been more moderate. Bloss, Schork, and Topol's prospective study did not identify major short-term adverse psychological effects or substantial changes in exercise and dietary fat intake after consumer genome-wide profiling. Stewart and colleagues' systematic review and meta-analysis similarly evaluated behavioral, psychological, and medical-consumption outcomes and found limited evidence that direct-to-consumer genetic testing produces large sustained behavioral effects.

This does not make misunderstanding unimportant. Kaufman and colleagues examined risk perception and use of medical services among direct-to-consumer personal genetic-testing customers, demonstrating that genetic-risk interpretation can enter subsequent healthcare behavior. Koeller and colleagues later showed that a subset of consumers sought genetic counseling after direct-to-consumer testing, illustrating demand for professional interpretation after results had already been received.



The clinical implications become more serious when raw data are interpreted as actionable variants. Tandy-Connor and colleagues' clinical-confirmation study provides a cautionary example: 40% of variants in their selected confirmation sample were false positives, and some variants assigned increased-risk labels were interpreted as benign during clinical review. The finding demonstrates that understanding genetic risk involves two different questions—what does this variant mean? and is the reported variant itself clinically confirmed?

3.3 Genetic Counseling, Consumer Autonomy, and Responsible Interpretation

The debate over direct-to-consumer genetic testing has often been framed as a tension between consumer autonomy and professional oversight. An autonomy-centered perspective emphasizes an individual's legitimate interest in accessing personal genetic information without unnecessary gatekeeping. A clinical-safety perspective emphasizes that probabilistic results, uncertain variants, incidental findings, and complex family implications may be difficult to interpret without professional support. Empirical research suggests that these positions need not be mutually exclusive.

Sweet and colleagues' randomized controlled trial found that genomic counseling improved objective understanding of the genetic contribution to several complex diseases and reduced overly genetic causal interpretations. Marzulla and colleagues subsequently interviewed people who sought genetic counseling after receiving direct-to-consumer genetic-testing results and documented the interpretive and decision-support functions consumers expected from counseling.

Genetic counselors themselves have expressed qualified concerns about direct-to-consumer testing while recognizing consumer interest. Hsieh and colleagues found that most surveyed counselors would be more accepting of direct-to-consumer testing if genetic counseling were incorporated into the process. These findings suggest that the central policy question is not simply whether testing should be direct-to-consumer, but whether consumers have access to accurate explanations, transparent limitations, confirmation pathways, and qualified professional support when the result could affect medical decisions.

4. Research Methodology

4.1 Research Design

The study adopts a simulation-based explanatory quantitative design. No individuals purchased a genetic test for this study, no biological samples were collected, and no actual consumer reports were analyzed. Simulation was selected because no participant dataset was supplied and because presenting invented survey results as empirical findings would be methodologically inappropriate.

A synthetic analytical population of 600 consumer profiles was generated and divided equally among three interpretation-support conditions. The report-only condition represents receipt of a standard genetic-risk report without additional interpretive intervention. The contextual-explanation condition adds plain-language probability framing and explicit limitations. The genetics-professional-supported condition represents interpretation with access to a qualified genetics professional.

4.2 Operationalization of Risk Understanding

Risk understanding was treated as a multidimensional construct rather than a single question asking whether a report seemed understandable. The model includes objective probabilistic comprehension, understanding of multifactorial causation, recognition of residual risk after a negative result, awareness of test scope, and recognition of when clinical confirmation may be appropriate.

Table 1: Operational Framework for Public Understanding of DTC Genetic Risk

Construct	Operational Definition	Proposed Scale	Analytical Role
Probabilistic Risk Comprehension	Correct understanding that genetic susceptibility changes probability rather than determining outcome	0–100	Primary outcome
Genetic Determinism	Tendency to interpret increased genetic risk as inevitability of disease	0–100	Misinterpretation outcome
False Reassurance	Tendency to interpret negative/lower-risk findings as absence of future disease risk	0–100	Misinterpretation outcome
Multifactorial Risk Understanding	Recognition that disease risk may include genetic, behavioral, environmental, age, and family-history factors	0–100	Secondary outcome
Test-Scope Awareness	Understanding that the test may examine only selected variants	0–100	Secondary outcome
Confirmation Awareness	Recognition that potentially actionable consumer findings may require clinical confirmation	0–100	Safety-related outcome
Perceived Clarity	Consumer-reported impression that the result is easy to understand	0–100	Comparative perception measure

4.3 Simulation and Analytical Procedure

The simulation intentionally allows perceived clarity and objective comprehension to diverge. Report-only cases can therefore report high confidence while still being assigned an interpretation error. This design reflects Leighton and colleagues' observation that consumers may describe genetic reports as easy to understand despite misinterpreting them.



Interpretation outcomes were classified as correct probabilistic interpretation, genetic-deterministic overinterpretation, false reassurance following a lower-risk result, or uncertain/mixed interpretation. Comparative support conditions were then modeled with progressively higher objective comprehension and lower deterministic interpretation.

Inferential significance tests are not presented because p -values calculated from deliberately simulated observations would not constitute empirical evidence. A future real-world study could use randomized report formats, pre- and post-result comprehension testing, validated genetic-literacy measures, numeracy assessment, and multivariable regression to determine which communication strategies improve understanding across demographic and educational groups.

5. Analysis

5.1 Simulated Risk Understanding Across Interpretation-Support Conditions

The simulation demonstrates substantial differences between perceived report clarity and objective risk comprehension.

Table 2: Simulated Genetic-Risk Comprehension Across Interpretation Conditions

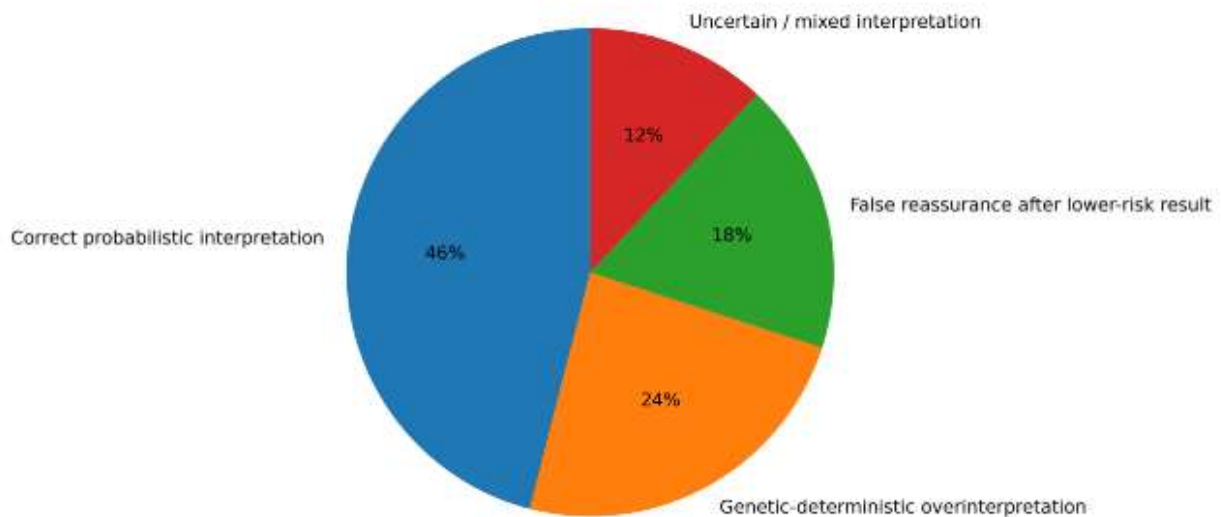
Outcome	Report Only	Contextual Plain-Language Explanation	Genetics-Professional Support
Simulated Cases	200	200	200
Correct Probabilistic Interpretation	46%	68%	84%
Multifactorial Risk Understanding	49%	74%	88%
Correct Interpretation of Negative Result	58%	78%	91%
Recognition of Limited Variant Coverage	41%	69%	86%
Awareness of Possible Need for Confirmation	44%	72%	92%
Perceived Report Clarity	76%	84%	91%

Source: Author-generated simulated data. Percentages are hypothetical and are not population estimates. The most notable simulated pattern occurs in the report-only group. Perceived clarity reaches 76%, but correct probabilistic interpretation reaches only 46%. The 30-percentage-point difference illustrates why subjective ease-of-understanding measures can substantially overstate genuine comprehension.

In the genetics-professional-supported condition, the difference narrows considerably, with modeled perceived clarity of 91% and probabilistic comprehension of 84%. This direction is consistent with evidence that genomic counseling can improve objective understanding of complex-disease genetic contributions.

5.2 Distribution of Risk-Interpretation Outcomes

Graph 1: Simulated Interpretation of Direct-to-Consumer Genetic Risk Results in the Report-Only Condition



The pie graph shows that only 46% of report-only cases are modeled as interpreting genetic susceptibility in an appropriately probabilistic manner. Genetic-deterministic overinterpretation accounts for 24%, while 18% show false reassurance following a lower-risk result and another 12% remain uncertain or demonstrate mixed understanding.

The graph does not represent the prevalence of misunderstanding among actual genetic-testing customers. Its purpose is to distinguish different errors that would otherwise be hidden if all incorrect responses were combined into a single “low comprehension” category.

5.3 Why Different Interpretation Errors Matter

Genetic determinism and false reassurance have opposite psychological directions but can both produce problematic decision-making. A deterministic interpretation may lead a person to experience a moderate increase in susceptibility as if diagnosis were inevitable. This can increase worry, distort perceived control, or prompt unnecessary healthcare requests.

False reassurance can be equally problematic. A lower-risk result may be treated as permission to ignore family history, routine preventive screening, behavioral risk factors, or symptoms. This is

particularly concerning when a consumer test examines only a subset of variants associated with a condition.

Raw-data interpretation adds another safety dimension. The Tandy-Connor study demonstrated that potentially actionable variants identified through consumer raw data should not automatically be treated as clinically established findings because false positives and classification disagreements can occur. The public therefore needs to understand both probabilistic uncertainty and analytical or interpretive uncertainty.

6. Discussion

6.1 The Difference Between Accessible Information and Understood Risk

Direct-to-consumer genetic testing has succeeded in making genetic information accessible to people who may never have sought conventional genetic services. That expansion of access has genuine value for autonomy and public engagement with genomics. Yet accessibility and comprehension are different outcomes. A visually polished report can reduce cognitive effort without necessarily conveying the conceptual limits of genetic prediction.

Leighton and colleagues' findings provide an especially important warning against equating confidence with understanding. Public participants often regarded reports as straightforward even when their interpretation differed from that of genetics professionals. This suggests that consumer-facing reports should be evaluated using objective comprehension tasks rather than satisfaction scores alone.

Risk communication should therefore make uncertainty visible rather than treating it as a disclaimer hidden beneath the main result. A statement such as “increased risk” should be accompanied, where scientifically appropriate, by the population reference, magnitude of the difference, limitations of the tested variants, and an explanation that genetic contribution represents only part of overall clinical risk.

6.2 Avoiding Genetic Determinism and False Reassurance

Genetic determinism is attractive because it converts uncertainty into a simple causal story: a risk allele becomes “the reason” a person will develop a disease. Common disease susceptibility rarely supports such a simple interpretation. Genomic counseling research suggests that professional discussion can reduce overly genetic causal beliefs and improve understanding of variant contribution.

Report design could reproduce some of these functions without requiring every user to attend an in-person counseling session. Plain-language explanations can explicitly distinguish predisposition from diagnosis. Visual representations can show the genetic contribution within a wider risk context. Prominent text can remind consumers that lifestyle, environment, age, family history, and additional genetic variants may remain relevant.

False reassurance requires equally explicit communication. A negative or reduced-risk result should not be framed as “all clear” when the test does not capture complete disease risk. Consumers should know precisely what has been tested and what remains unknown. Such communication is particularly important for selected-variant tests, where absence of the included variants cannot exclude other clinically important variants.

6.3 Genetic Counseling, Confirmation, and Responsible Consumer Genomics

The findings support a tiered interpretation model rather than mandatory counseling for every consumer interaction. Low-risk ancestry or recreational information may not require the same level of professional support as a potentially actionable health-related finding. By contrast, results involving serious disease predisposition, uncertain interpretation, strong family history, reproductive implications, or a raw-data variant presented as clinically actionable may justify genetics-professional involvement.

Consumers who voluntarily sought genetic counseling after direct-to-consumer testing have described a need for clarification, contextualization, and assistance interpreting implications. Genetic counselors themselves have also indicated greater acceptance of consumer testing when counseling is incorporated into the pathway. These findings suggest that professional interpretation can strengthen consumer autonomy by enabling better-informed decisions rather than simply restricting access.

Clinical confirmation is another essential safeguard. Tandy-Connor and colleagues' findings should not be generalized to every direct-to-consumer report, particularly because their sample consisted of results already referred for confirmation. Nevertheless, the discovery that 40% of variants in that selected sample were false positive demonstrates why major preventive, surgical, reproductive, or pharmaceutical decisions should not rest on unconfirmed raw consumer-genotyping results.

7. Conclusion

Public understanding of direct-to-consumer genetic testing depends on more than the ability to read a result. Meaningful genetic-risk literacy requires understanding that susceptibility is probabilistic, that common diseases are often multifactorial, that lower-risk findings do not eliminate disease risk, and that consumer tests may examine only selected portions of clinically relevant genetic variation. A report can appear simple while its implications remain conceptually difficult.

The simulation presented in this study illustrates these distinctions. Correct probabilistic interpretation was modeled at 46% under report-only disclosure, compared with 68% after contextual plain-language explanation and 84% with genetics-professional support. Within the report-only condition, genetic-deterministic overinterpretation, false reassurance, and uncertainty accounted for substantial proportions of modeled responses. These percentages are synthetic and must not be represented as actual population prevalence.

The pre-2022 empirical literature provides strong justification for testing this framework. Public respondents have been shown to misinterpret consumer genetic-risk reports despite perceiving them as understandable, genomic counseling can strengthen objective understanding, and selected raw-data findings can require clinical confirmation before they are treated as actionable. At the same time, consumer genomic testing does not appear to produce uniformly severe psychological harm, and many consumers perceive personal utility in the information they receive.

The appropriate public-health goal is therefore neither unrestricted genetic determinism nor unnecessarily restrictive gatekeeping. Consumers should retain meaningful access to their genetic information while receiving communication that accurately represents uncertainty, scope, and clinical limitations. Future empirical studies should compare numerical, verbal, and visual risk formats; test understanding before and after professional interpretation; examine disparities associated with health literacy and numeracy; and measure whether accurate risk understanding produces more appropriate



healthcare decisions. Responsible consumer genomics ultimately depends not simply on giving people more genetic information, but on ensuring that the information can be interpreted in proportion to what the underlying science actually establishes.

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