



## **Ethical and Privacy Principles in relation to Responsible Sharing of Genomic and Health---Related Data**

### **Preamble**

The sharing of genomic and health-related data for biomedical research is of key importance in ensuring continued progress in our understanding of human health and wellbeing. The challenges raised by international, collaborative research require a principled but nevertheless practical Framework that brings together regulators, funders, patient groups, information technologists, industry, publishers, and research consortia to share principles about data exchange. Such a Framework will facilitate responsible research conduct.

This Framework is developed under the auspices of the Global Alliance for Genomics and Health (GA4GH) and relates to Version 10 of their document “Framework for Responsible Sharing of Genomic and Health-related Data (Sept 2014). It is endorsed by InSiGHT for the purposes of InSiGHT’s responsibility for the management and curation of genes whose variants predispose to inherited gastrointestinal cancer. InSiGHT’s mission is to accelerate progress in human gastrointestinal health by helping to establish a common Framework of harmonized approaches to enable effective and responsible sharing of genomic and clinical data and to catalyze data sharing projects that drive and demonstrate the value of data sharing.

This Global Alliance Framework provides guidance for the responsible sharing of human genomic and health-related data, including personal health data and other types of data that may have predictive power in relation to health. In particular, it highlights, and is guided by, Article 27 of the 1948 *Universal Declaration of Human Rights*. Article 27 guarantees the rights of every individual in the world “*to share in scientific advancement and its benefits*” (including to freely engage in responsible scientific inquiry), and at the same time “*to the protection of the moral and material interests resulting from any scientific...production of which [a person] is the author.*” (As set out in Appendix 1, many other international conventions and national laws, regulations, codes and policies also guide responsible data sharing behavior).

### **The InSiGHT Framework**

This Framework is guided by the human rights of privacy, non-discrimination and procedural fairness. On the basis of this, InSiGHT annotates its ethical and privacy principles as they relate to InSiGHT’s mission of assembling a comprehensive database of variants of genes known to predispose to gastrointestinal cancer. At the same time, it considers all principles relevant, complementary and interrelated, based on respect for human dignity. Since science proceeds only with the broad support of society, respect for all persons is a primary driver underlying all other derived principles. In particular, this policy establishes a set of principles to underpin responsible research conduct and oversight of research data systems in the realm of genomic and health-related data sharing relevant to the genes in InSiGHT’s professional

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jurisdiction knowing that this may change over time in response to new developments in knowledge and practice.(see appendix).

This Framework takes as its basis the right of all people to share in the benefits of scientific progress and then sees this as being the duty of data producers and users to engage in responsible scientific inquiry and to access and share genomic and health-related data across the translation continuum, from basic research through practical applications. It also recognizes the rights of data producers and users to be recognized for their contributions to research, balanced by the rights of those who donate their data. Lastly, in addition to being founded on the right of all citizens in all countries to the benefits of the advancements of science, and on the right of attribution of scientists, it also reinforces the right of scientific freedom.

The value of this Framework, is that it:

- offers political and legal dimensions that reach beyond the moral appeals of bioethics
- provides a more robust governance framework for genomic and health-related data sharing;
- speaks to groups and institutions, not just individuals;
- stresses the progressive realization of duties; and
- urges action by governments, industry, funders, publishers, and researchers to create an international environment for responsible sharing data.

This InSiGHT Framework is elaborated by specific Policies (Appendix 2) on particular issues such as ethical governance, consent, privacy and security. The Policies should be seen in relation to this Framework rather than in isolation, however each policy can stand alone and can be used in projects around the world where InSiGHT “inspired” activities are undertaken. It is expected that they become the basis for developing locally agreed policy tools for entities operating in these various local jurisdictions Recognizing diversity of legal and ethical approaches and being responsive to emerging issues, this Framework, together with its linked policies, is intended to provide leadership for InSiGHT’s constituencies and members.

## I. Purpose and Interpretation

1. Purpose. The purpose of this document is to provide a principled and practical framework for the responsible sharing of genomic and health-related data. Its primary goals are to:
  - i. Protect and promote the welfare, rights, and interests of individuals from around the world in genomic and health-related data sharing, particularly those who contribute their data for biomedical research in the domain of inherited gastrointestinal tumours;
  - ii. Complement laws and regulations on privacy and personal data protection, as well as policies and codes of conduct for the ethical governance of research;
  - iii. Foster responsible data sharing and oversight of research data systems;
  - iv. Establish a framework for greater international data sharing, collaboration and good governance;
  - v. Serve as a dynamic instrument that can respond to future developments in the science,

Framework for Responsible Sharing of Genomic and Health-Related Data technology, and practices of genomic and health-related data sharing;

- vi. Serve as a tool for the evaluation of responsible research by research ethics committees and data access committees in the domain of inherited gastrointestinal tumours; and
  - vii. Provide overarching principles to be respected in developing legally-binding tools such as data access agreements.
2. **Interpretation.** Without ascribing legal meaning, this Framework should be interpreted in good faith and is to be understood as a whole. The Foundational Principles and Core Elements are to be understood as complementary and interrelated, as appropriate and relevant in different contexts, countries and cultures. This Framework will be supported by subsidiary Policies for guidance in particular issues such as, but not limited to, ethical governance, privacy and security, and consent. For the purposes of this Framework, “data sharing” includes data transfer or data exchange between data users, or where data are made available to secondary researchers, either openly or under specified access conditions.

## II. Application

This Framework is intended for all entities or individuals providing, storing, accessing, managing or otherwise using genomic and health-related data, including data donors, users, and producers pertaining to inherited gastrointestinal tumours. This includes, but is not limited to, researchers, research participants and patient communities, publishers, research funding agencies, data protection authorities, hospitals, research ethics committees, industry, ministries of health, and public health organizations.

## III. Foundational Principles

The Foundational Principles of this Policy guide the responsible sharing of genomic and health-related data. They also facilitate compliance with the obligations and norms set by international and national law and policies.

### **Foundational Principles for Responsible Sharing of Genomic and Health-Related Data**

- **Respect Individuals, Families and Communities**
- **Advance Research and Scientific Knowledge**
- **Promote Health, Wellbeing and the Fair Distribution of Benefits**
- **Foster Trust, Integrity and Reciprocity**

#### IV. Core Elements of Responsible Data Sharing

It is good practice for those involved in genomic and health-related data sharing to have core elements of responsible data sharing in place. The following Core Elements of the Framework aid in the interpretation of the Foundational Principles to individuals and organizations involved in the sharing of genomic and health-related data. The Core Elements should be interpreted in a proportionate manner that acknowledges different levels of risk and community cultural practices. **This Policy applies to use of data that have been consented to by donors (or their legal representatives) and/or approved for use by competent bodies or institutions in compliance with national and international laws, general ethical principles, and best practice standards that respect restrictions on downstream uses.** Endorsement of the Policy does not preclude the development of particular guidance via Policies for specific populations (e.g. children) or issues (e.g. ethical governance, privacy and security, and consent). InSiGHT endorses the principles developed by GA4H and incorporates them into the InSiGHT Framework expressing them in the following Core Elements.

## Core Elements of Responsible Data Sharing

### □ Transparency

*InSiGHT has appointed a Governance Committee to oversee the activities of the databases held in its jurisdiction of inherited gastrointestinal cancer, with terms of reference, and standard operating procedures for the curation and management of genes responsible for inherited gastrointestinal cancer, and the Variant Interpretation Committees that support those databases*

*The purpose of data collection is primarily to support interpretation of variants responsible for inherited gastrointestinal and other associated syndromic cancers, for diagnostic and treatment related purposes.*

*Transfer of data to other parties, other than government agencies (e.g. NCBI) requires submitter consent; commercial use of the data requires specific consent from submitters on an opt in basis, although access to the data for individual variant interpretation support is unrestricted; international data transfer is not considered outside these principles and is expected; data storage is for 15 years (subject to financial capacities of InSiGHT for same); stored data will be not identifiable but will be re-identifiable only through collaboration with the submitter (important for families worldwide in the case of life threatening information being uncovered prospectively in the course of the work of InSiGHT and its Variant Interpretation Committee); processes are in place in SOP of the InSiGHT databases to notify families of such eventualities; the governance committee is informed of regularities and irregularities of these processes; commercial involvement is an active independent interest of InSiGHT in this domain but only within the framework of this policy and about which Council is kept informed; no propriety claims are anticipated but this is a brief under continuing review by the InSiGHT Database Governance Committee and InSiGHT Council; withdrawal of data requests have not been the experience of InSiGHT but will be respected.*

*Use of the data for research purposes requires consultation with the relevant gene specific curator, and an approach to all submitters of data for their consent (and possibly authorship according to the Vancouver convention of authorship). This policy is articulated on the opening page of the InSiGHT LOVD databases.*

### □ Accountability

*Curators take responsibility for tracking the chain of data access and/or exchange and report anomalies to Council e.g comprehensive downloads of all, or a majority, of the data on the InSiGHT databases. This can be tracked through LOVD processes. Such requests are received from time to time from commercial and non-commercial sources, and are referred to the Governance Committee.*

*Conflicts of interest are referred to the Governance Committee.*

*The Governance Committee handles complaints related to data misuse, through their identification by curators, and reporting to Council as well as managing breaches and*

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*appropriate sanctions. The SOP for the databases articulates these.*

## □ Engagement

*Engagement with the genetics community, especially for hereditary gastrointestinal cancers, is the core business of InSiGHT, its databases and its Variant Interpretation Committees. Please refer to the SOP of the Variant databases. InSiGHT actively encourages scientific contributions representing these needs to its bi-ennial meetings, providing information enriching its databases. InSiGHT respects all scientifically credible information sources.*

*InSiGHT's broad membership captures, specifically, stakeholders professionally concerned with psychosocial, societal, ethical and political interests world wide*

## □ Data Quality and Security

*InSiGHT strives at the highest level of scrutiny to store and process the data collected, used and transferred in a way that is accurate, verifiable, unbiased, proportionate, and current, so as to enhance their interoperability and replicability and also preserve their long-term searchability and integrity. Data quality is inevitably limited by the accuracy of the data submitted to its database with which it holds no warranty. Nevertheless, in total, it warrants the credibility and clinical utility of the data it shares across all stakeholders. This is not to acknowledge the validity of individual data submission elements.*

*InSiGHT curators have processes for such feedback in place, limited by the contactability of submitters of relevant associated information worldwide through email addresses held by InSiGHT.*

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*InSiGHT databases are covered in this respect by the security processes embedded in LOVD*

*Issues relating to lawful requests for data based on law enforcement, public health, or national security concerns are elevated to the InSiGHT Governance Committee deliberation if escalation is required.*

### □ Privacy, Data Protection and Confidentiality

*InSiGHT is dependent on the submitters of data for its accuracy; InSiGHT holds the data confidentially and with privacy. InSiGHT ensures data is collected, stored, processed, and exchanged in a fashion which preserves the anonymity of the personal source of the data. Re-identification can only be done after passage of information back to the submitter*

*Such re-identification may be appropriate and important where information emerges through the scientific processes of the Variant Interpretation Committee facilitated by its international reach and scientific excellence, which identifies a substantial risk to the health of individuals represented on the database otherwise not evident to such individuals*

### □ Risk-Benefit Analysis

*InSiGHT, its Council, Governance Committee and curators strongly adhere to the following ethical principles:*

*Consideration of realistic harms and benefits of data sharing on and with individuals, families and communities, including opportunity costs associated with both sharing and not sharing of data. Potential realistic benefits may include development of new scientific knowledge and applications, enhanced efficiency, reproducibility and safety of research projects or processes, and more informed decisions about health care. Potential realistic harms may include invasions of privacy or breach of confidentiality and invalid conclusions about research projects*

*Interpretation of variants for diagnostic purposes is the core value of the InSiGHT databases. Scientific advances utilizing the data, though important, requires consent of InSiGHT curators and submitters insofar this is reasonably possible*

*Data sharing is conducted with a view towards minimizing harms and maximizing benefits to not just those who contribute their data, but also to society and health care systems as a whole, particularly where data pertains to disadvantaged people. Benefits arising from data sharing may not be uniformly distributed throughout communities around the world and may depend on the usability of data within a specified context, national priorities, as well as a specific community's concern about health and interpretation of wellbeing.*

- *InSiGHT is cognizant of, and adheres to the principle of proportionate assessment of the benefits and risks of harm in data sharing, which is periodically monitored according to the reasonable foreseeability of such harms and benefits. Such an assessment may also incorporate mechanisms that track subsequent harms, should they materialize, so as to help inform future policy*

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### □ Recognition and Attribution

*InSiGHT has developed a process of microattribution, supporting recognition and attribution of source data. InSiGHT is alert to developments in this field which it recognizes as critical to the facilitation of centralization of variant data. InSiGHT promotes the Vancouver convention for authorship of data including derived from its databases (submitter consent permitting)*

*InSiGHT is alert to developments in central databasing and data sharing and recognizes as critical to the facilitation of centralization of variant data. InSiGHT promotes the Vancouver convention for authorship of data including derived from its databases (submitter consent permitting). Local law is always respected by InSiGHT.*

### □ Sustainability

*Recognizing the humanitarian value of its genome-wide leadership in locus specific databasing, InSiGHT strives to ensure sustainability of curation and the relevance of its variant databases for human health in the gastrointestinal cancer, within the limits of its capacity.*

### □ Education and Training

*InSiGHT joins with the Human Variome Project and the Global Alliance to deliver education and training.*

### □ Accessibility and Dissemination

*The InSiGHT variant data are available in unrestricted fashion for individual variant enquiry through the InSiGHT website. This includes the deliberations of the InSiGHT Variant Interpretation Committees*

*Collaboration and sharing is a core function of the InSiGHT curators with networking across InSiGHT members, national variant databases and reaches into public and private diagnostic laboratories and specialized clinical consultative services.*

*Except as protected by copyright, All data is available publicly in the InSiGHT platforms*

## V. Implementation Mechanisms and Amendments

- 1. InSiGHT acknowledges that this Policy should be adopted by organizations and bodies involved in genomic and health-related data sharing. InSiGHT takes all reasonable and appropriate measures, whether of a regulatory, contractual, administrative or other character, to give effect to the Foundational Principles and Core Elements set out in the Global Alliance Framework on which this Policy is based, in accordance with the international law of human rights and should, by means of all reasonable and appropriate measures, promote their implementation.*



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This Global Alliance *Framework for Responsible Sharing of Genomic and Health-Related Data* is the result of the work of many people and committees. Developed under the auspices of the Global Alliance for Genomics and Health, the Framework was initially formulated by an international committee (Regulatory and Ethics Working Group) representing a wide spectrum of the bioethics, genomics, and clinical communities. Collaborative input was provided from individuals as well as biomedical, patient advocacy, and ethical, policy and legal organizations, committees, and projects from all regions of the world. These include, but are not limited to:

- [Biobank Standardisation and Harmonisation for Research Excellence project](#) (BioSHaRE);
- [Centre for Law and Genetics](#) (University of Tasmania);
- [Centre of Genomics and Policy](#) (McGill University);
- [ELSI 2.0](#);
- [H3Africa](#);
- [Health Research Authority](#) (UK);
- [HeLEX](#) (University of Oxford);
- [Human Variome Project](#) (HVP);
- [INSERM](#);
- [International Cancer Genome Consortium](#) (ICGC);
- [International Rare Disease Research Consortium](#) (IRDiRC);
- [International Society for Biological and Environmental Repositories](#) (ISBER);
- [Personal Genome Project](#) (USA);
- [PErsonalised Risk Stratification for Prevention and Early deteCTIon of breast cancer project](#) (PERSPECTIVE);
- [PHG Foundation](#) (UK);
- [Public Population Project in Genomics and Society-International Policy interoperability and data Access Clearinghouse](#) (P3G-IPAC); and
- other [Global Alliance for Genomics and Health Working Groups and Task Teams](#).

## Appendix 1

### Foundational Human Rights Instruments

- \* [Universal Declaration of Human Rights](#) (UN 1948) (Article 27)
- \* [International Covenant on Economic, Social and Cultural Rights](#) (UN 1966) (Article 15)

### Ethical and Legal Codes and Policies Guiding Data Sharing Behavior

- [Constitution of the World Health Organization](#) (WHO 1946)
- [Bermuda Principles on Human Genome Sequencing](#) (1996)
- [Universal Declaration on the Human Genome and Human Rights](#) (UNESCO 1997)
- [Convention on Human Rights and Biomedicine](#) (Council of Europe 1997)
- [Statement on DNA Sampling: Control and Access](#) (HUGO 1998)
- [Statement on Human Genomic Databases](#) (HUGO Ethics Committee 2002)
- [Declaration of Ethical Considerations regarding Health Databases](#) (WMA 2002)
- [International Ethical Guidelines for Biomedical Research Involving Human Subjects](#) (CIOMS, WHO 2002)
- [Budapest Open Access Initiative](#) (2002)
- [Sharing Data from Large-scale Biological Research Projects: A System of Tripartite Responsibility](#) (Fort Lauderdale Statement, 2003)
- [International Declaration on Human Genetic Data](#) (UNESCO, IBC 2003)
- [European Society of Human Genetics: Data Storage and DNA Banking for Biomedical Research](#) (ESHG 2003)
- [Universal Declaration on Bioethics and Human Rights](#) (UNESCO 2005)
- [Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research](#) (Council of Europe 2005)
- [Recommendation Rec \(2006\) 4 of the Committee of Ministers to Member States on Research on Biological Materials of Human Origin](#) (Council of Europe 2006)
- [OECD Principles and Guidelines for Access to Research Data from Public Funding](#) (OECD 2007)
- [International Ethical Guidelines for Epidemiological Studies](#) (CIOMS, WHO 2008)
- [Recommendations from the 2008 International Summit on Proteomics Data Release and Sharing Policy](#) (Amsterdam Principles, 2008)
- [Guidelines for Human Biobanks and Genetic Research Databases](#) (OECD 2008, 2009)
- [Toronto Statement on Prepublication Data Sharing](#) (2009)
- [Joint Statement by Funders of Health Research](#) (2011)
- [2012 Best Practices for Repositories: Collection, Storage, Retrieval and Distribution of Biological Material for Research](#) (ISBER 2012)
- [Responsible Conduct in the Global Research Enterprise: A Policy Report](#) (InterAcademy Council 2012)
- [Declaration of Helsinki](#) (WMA 2013)
- [Guidelines governing the Protection of Privacy and Transborder Flows of Personal Data](#) (OECD 2013)

## Appendix 2

# Global Alliance for Genomics and Health (GA4GH): Proposed Policy Template

### Standardized Preamble:

*The GA4GH Framework is founded on Principles and Core Elements, and further elaborated by Policies on particular issues. These Policies can become the tools and benchmarks that approved entities, recognized by different jurisdictions, turn or refer to for guidance. These Policies can help entities make improvements or adopt specific best practices for responsible data sharing and governance processes. Recognizing diversity of legal and ethical approaches and being responsive to emerging issues, both this Framework and its Policies are intended to provide leadership on these issues.*

### Context/Background: Describe Issues/Domain

### Specific Policy Principles particular to an issue

e.g.:  
Privacy / Security  
Consent  
Ethics and Governance

### Procedural Guidance

(with URL links to other documents or standards where appropriate/applicable)

### Implementation Mechanisms + Amendment Procedures

### Acknowledgements

