



*Partnering for Electronic Delivery  
of Information in Healthcare*

# **MAXIMIZING THE POTENTIAL OF GENOMIC INFORMATION TO IMPROVE CARE COORDINATION AND HEALTH OUTCOMES**

**A White Paper by the Genomics Workgroup**

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***Workgroup for Electronic Data Interchange***  
*1984 Isaac Newton Square, Suite 304, Reston, VA. 20190*  
*T: 202-618-8792/F: 202-684-7794*

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# Maximizing the Potential of Genomic Information to Improve Care Coordination and Health Outcomes

## I. Introduction

Genomic medicine offers the potential to greatly improve medical practice by tailoring the preventive, diagnostic and therapeutic care available to each patient. However, this potential depends on high-quality data that can be readily accessed and applied in the patient care setting. Although improvements to electronic health records (EHRs) have been made, much work remains to optimize their ability to support genomically informed care, and in turn, for genomic information to contribute to improvements in care coordination. This white paper explores how genomic information could optimize clinical genomics care coordination, including how genomic data can be leveraged to improve health and care at both individual and population levels. It examines a number of clinical situations and life stages during which genomic information is generated, how health care providers access and use that data, the infrastructure needed for effective use of the data and the financial drivers of genomic data use for improved health care.

## II. Multiple Points of Entry That Generate Genetic/Genomic Data

### Primary Care

Genetic data is generated in multiple clinical situations, with varying levels of complexity depending on the type of service and the health professional overseeing it. In the primary care setting, physicians and other health professionals see patients with a broad number of health concerns, including those with chronic conditions needing ongoing care, those with newly emerging symptoms, and asymptomatic patients visiting for recommended preventive services. In most cases the collection of a family health history (a record of the health information of the patient and his or her close family relatives) is warranted. The family history can indicate whether a patient is at risk for a genetic condition by revealing patterns of inheritance and disease within the family, and often provides direction as to further evaluation that may be needed. It therefore generates a wealth of genetic data, which, in order to be most useful, must be documented and updated on a regular basis. Somewhat related to the genetic data generated by a family history is genetic data

generated by risk assessment and screening. For example, in the case of a patient whose family history shows a risk of a hereditary colorectal cancer syndrome, the risk of colorectal cancer in the patient must be evaluated, and if risk is elevated, the patient should undergo enhanced screening, such as a yearly colonoscopy beginning in his or her teens or twenties. The patient's risk profile and results of screening procedures are themselves genetic data since they are informing about the potential for genetic disease in the patient and must be readily accessible to the physicians and health professionals caring for the patient.

When a patient is identified as having a high risk for a hereditary syndrome or showing symptoms that may point to a genetic component to disease, genetic tests are often ordered to assist in diagnosis, the results of which generate varying amounts of data depending on the type of test. These can include data that are less complex (e.g., from a test of one genetic variant or one gene) or data sets that are large and extremely complex (e.g., from tests that sequence a large number of genes or even the whole genome). The interpretation of test results and the care management plan based on those results also generates large amounts of data. Included in management plans are therapeutic approaches, both pharmacologic and non-pharmacologic, that are informed by the results of genetic testing, such as drugs targeted to specific tumor characteristics that attack cancer cells but spare healthy cells, or doses chosen after the results of pharmacogenomic testing reveal whether patients metabolize drugs at slower or faster rates than normal.

### Specialty Care

The generation of genetic data can be magnified in the specialty setting. Patients are often referred to specialists, including medical geneticists and genetic counselors, when a genetic condition is suspected or when a primary care provider believes the patient would be better served by a specialist. When this is the situation, the processes of collecting a family history, assessing risk, ordering and interpreting tests, and developing management plans can generate large amounts of data as symptoms and differential diagnoses are considered and genetic disease is uncovered. The referral of patients to specialists highlights the need for care coordination. The referring provider, specialist, geneticist and genetic counselor all need to access genetic data to provide optimal coordinated care. In addition, some treatment plans may require review and prior authorization by insurers. Timely access to the necessary information for those who need it will support optimal and timely care management decisions.

### Decision Support and Genetic Testing

The assimilation of these multiple data sources and their application to care can be enhanced by clinical decision support (CDS) built into the EHR. CDS provides health professionals with knowledge and person-specific information, intelligently filtered or presented at appropriate times, to improve health care quality and outcomes. Examples of CDS tools are computerized alerts and reminders, clinical guidelines relevant to the case, focused patient data reports and summaries, and contextually relevant reference information. Automated messaging to each stakeholder when testing is ordered, results are reported or approval is granted will enhance the coordination of care utilizing the EHR

and CDS tools. In the genomics realm, CDS tools aim to provide guidance to health professionals on risk assessment, test ordering and interpretation, and management of patients by accessing and framing genetic information in the EHR.

#### Physicians Using Decision Support

CDS resources are especially helpful in care coordination, since multiple health professionals with access to the EHR will see the same data interpretation and guidance presented by the CDS. Additionally, CDS can ensure that tasks involving other stakeholders, such as pre-authorization requests to insurers, are automated. For example, in reexamining the patient with a family history that suggests a risk of hereditary colorectal cancer, a multi-variant panel test assessing several genetic variants may be ordered and pre-authorization granted. The treating physician might be primarily interested in the interpretation of overall colorectal cancer risk. To best answer that question, CDS related to the results of only those variants involved in colorectal cancer is presented. However, there is now a corpus of data related to other genes analyzed on that multi-variant panel that can be called on at a later point. CDS in the EHR removes the cognitive burden on the physician to recall which genes previously analyzed are related to future clinical situations for that patient. An actionable, systematic CDS process around genetic data has the potential to hasten the diagnostic process, accelerate therapeutic and non-therapeutic care planning, and better inform and engage all members responsible/accountable for adherence to such a care plan. Early detection and early treatment today for colorectal cancer might lead to an opportunity for better use of antiplatelet therapy in the future when heart disease is diagnosed in the same patient.

### **III. The Continuum of Health Care Providers Who Will Access the Data**

As we go beyond primary and specialty providers, we recognize that many ancillary providers also play a significant role in genetics-based care. Besides the genetic counselor (who may order genetic testing), these other providers likely will not order genetic or genomic tests, but will need to have access to the same patient data as the physicians. An integrated EHR capable of accepting and sharing data with each of these providers is essential to coordinated care in the expanding area of clinical genetics/genomics.

#### The Genetic Counselor

As genetic and genomic testing, data analysis and clinical interpretation become the standard of care, so will the demand for patients to visit with genetic counselors. The process starts with a referral from a physician, either primary care or specialty care. The physician has determined the need for genetic testing for the patient or seeks the consult of the counselor to help in deciding whether testing is clinically indicated. The collection and analysis of a family health history by counselors is an important tool to make risk assessment and testing recommendations. EHR systems and external third-party family health history tools are increasing the capability to manage and integrate this data between patients, patient families and their health providers.

The counselor then directs the test ordering and reviews the lab results with the patient and his or her physician. Counselors provide patient education, guide decision making and other patient management and follow-up activities. Besides just the patient, the counselor may visit with and test other family members. The workflow concludes with the counselor reporting back to the physician. This is the first step in a coordination of care and interspecialty team model. In order to manage this process, an integrated data system and targeted decision support for specific providers needs to be in place.

### Pharmacy

Medication therapy can be optimized based on pharmacogenomic test results. Pharmacists can provide assistance in test result interpretation and clinical guidance for return of results to providers and patients in collaboration with other health care professionals (e.g., physicians, laboratory professionals and genetic counselors). Pharmacists support and participate in research, consortia and networks that guide and accelerate the application of pharmacogenomics to clinical practice.

### Pathology/Laboratory

Although they work with many specialties, pathologists play a major role performing cytogenetic and molecular testing in the diagnosis of many conditions; for example, analysis of soft-tissue tumors. Oncology and cancer genomics are significant utilizers of genetic/genomic testing for both germline and somatic tests. Laboratory medical directors, besides signing off on test results, also are involved in the innovative process of genetic test development. Labs also contribute to variant databases, assisting in the continuing discovery of clinically relevant variant knowledge-bases. In an ongoing process, laboratory personnel notify health care providers when variants of unknown significance change to a new classification (e.g. benign or pathogenic).

### Nursing, Nurse Care Managers/Care Coordinators

Many diseases and the therapies used to treat them have a genetic element influenced by environmental, lifestyle and other factors. A responsibility to watch and understand all of these issues impacts the entire nursing profession. More than most other health care providers, nurses have an intimate knowledge of the patient's, their family's and the community's perspectives on the diagnostic and treatment process; an understanding of biologic underpinnings and experiences with genetic/genomic technologies and information; skills in communication and building coalitions as a patient advocate; and most importantly, the public's trust in their front-line care. The American Nurses Association has published genetics/genomics competencies for nurses.

### Physical Therapists

Studies have indicated that genetic factors influence many, if not most of the dysmorphic diseases commonly encountered in clinical practice by physical therapists. Genes appear to influence not only risk for disease, but also progression, outcomes and response to rehabilitation interventions.

## Emergency Care, Critical Care, Hospitalists

Heritable conditions can be chronic and severe, and may even be life threatening. As an example, clinically useful biomarkers can be used to identify a subset of difficult to treat asthmatic children who present in the emergency department. In other cases, genetic variations in adrenergic system function can influence the patient's vulnerability to post-traumatic pain.

## Nutritionists (Nutritional Genomics)

Nutritionists are involved in both prevention and intervention activities. First, they evaluate and recommend nutrition prevention strategies based on a health risk appraisal that includes assessment of genetic, environmental and ethnic risk factors, and cultural health beliefs and practices. Second, they may use genetic test results to inform the nutrition care process.

## Researchers and Large Studies

To take steps toward finding cures, physicians and patients need to be collaborators. Ensuring that patients can access their own health records, including their genetic/genomic data, will make it easier for patients to donate their data to research. Opportunities to share data exist with the many patients choosing to participate with registries and with emerging options for patients to donate clinical data from their EHR via the new programs such as, Sync for Science, a data-sharing tool developed as a resource for President Obama's Precision Medicine Initiative.

## Non-Provider Care Managers (Payer/Other Third Party)

Insurers have to understand genetic testing strategies, interpretations, outcomes, patient care and use that understanding in making sound payment policies and decisions regarding the health care services used by their insured. Analysis of the validity and utility of clinical genetic/genomic testing informs decisions in the economic utility of testing and treatment coverage by the health plan.

## **IV. The Patient View**

Consumers today have access to a vast array of health information without ever consulting a health care provider, from general internet resources on health and disease to more specific data points generated by wearable devices, home-based health monitoring tools and even results of genetic tests ordered directly by the consumer. When added to information that may be gained during a visit to a physician or other health provider, a need emerges for a better understanding of how to integrate these multiple sources of information to best guide health-related decisions. Accurate and reliable data must be available to those who can use it to improve care and health outcomes. This includes the consumers themselves (or patients, if seeking care) and their families and caregivers, if appropriate.



## Value of Genetic Services

In an increasingly value-based environment, the cost of health services and the benefits they provide will be an important consideration for health care consumers. This balance between cost and benefits is particularly relevant for genetic services; testing can sometimes be expensive, but results can yield information useful for a lifetime and beyond, for both the patient and family members. Some of the potential benefits of genetic testing from the patient's point of view are:

- helping to diagnose disease or identify risk for disease;
- providing information that will assist in choosing drug therapies that are more likely to benefit the patient and less likely to produce adverse reactions;
- enhancing health care providers' understanding about patients' lifestyle needs and socioeconomic realities, thereby informing preferences for treatment;
- contributing to the knowledge base about the benefits of precision medicine, potentially leading to better outcomes for specific disease states;
- reducing the overall cost of care by contributing information on risk factors and preventive care;
- providing information to assist health care providers and insurance payers in developing improved care models based on genetic variants and their related effects;
- and revealing eligibility for participation in clinical trials focused on the patients' genetic variation or condition.

Patients have complex and individual needs, with health outcomes being influenced by a host of factors. Longitudinal capture and storage of data such as test results, treatments and outcomes is important for informing care in both the near and long term. Enhanced knowledge and awareness on the part of consumers or patients often leads to better outcomes since they are empowered to control their own health and lifestyle behaviors. A historical health care record that can be easily updated and makes information available to patients contributes to this empowerment. The inclusion of genomic information in the health care record is essential since it can inform diagnoses, risks, therapies and preventive measures.

## The Family Perspective

The utility of genomic information extends beyond just the patient. Heritable genetic variations are shared among family members, meaning that the presence of a variant in one person indicates the possibility that other family members may also carry that variant. For example, the identification of a genetic variant that dramatically increases risk for breast cancer in a patient means that the patient's family members could be tested for that variant, with appropriate screening and intervention to follow. Similarly, the diagnosis of a genetic disease in a patient can yield powerful information for predicting and/or preventing the onset of that disease in family members. Collecting and updating family health history arms individuals with valuable information that can reveal red flags for possible genetic conditions in the family, assisting health care providers in identifying who is at risk and what kind of preventive action should be taken. For these reasons, the inclusion of genetic

information in a patient's medical record, including genetic conditions in family members, is valuable.

## The Longitudinal Perspective

Care coordination is particularly important in the crosscutting realm of genomic medicine. Heritable conditions affect not only individuals, their immediate families and the providers who deliver services, but also have broad implications for policymakers, public health decision-makers, public and private payers, employers, educational institutions and numerous other entities. Enhanced and longitudinal care coordination services are needed to ensure that patients affected by genetic conditions receive appropriate care as they transition through life stages. Conditions with a genetic basis occur across the lifespan, and at each stage, patients and their families experience different presentations and needs, including medical, psychological and social. Concerns about life years and quality of life, reproductive risks, and the effect on earnings potential are common in patients diagnosed with or at risk for genetic conditions, as are questions about costs and insurance coverage of genetic services. Genetic counseling is particularly important for addressing questions about therapeutic or preventive options, such as pregnancy termination or prophylactic surgery. Below is a brief discussion of genomic events that occur at predictable life stages, along with potential care coordination and support service needs.

### Prenatal

During the prenatal period, a number of tests that detect genetic conditions are recommended, often based on risk factors such as maternal age, ethnicity, conditions present in siblings and other family members, and previous pregnancy loss or complications. Test results that reveal a genetic condition should be accompanied by counseling and assistance in decision-making regarding pregnancy continuation or termination, preparation for the care needs of the affected neonate and future pregnancies. Couples who are carriers of genetic conditions may choose to perform pre-implantation genetic diagnosis, in which embryos generated through in vitro fertilization are screened for the genetic condition(s) of interest, so that non-affected embryos can be implanted.

### Newborn

All newborns in the U.S. undergo metabolic screening within the first days of life for dozens of genetic conditions. Early detection, diagnosis and intervention can prevent death or disability for these mostly rare conditions. When a condition is detected in a newborn, care coordination efforts are important for ensuring that the baby receives appropriate medical care and that the family receives appropriate support services to provide care for the baby as he or she matures. Coordination of social services, facilitation of home accommodations, arrangements for specialized transportation and acquisition of durable medical equipment are often needed to ensure proper care for the baby.

### Childhood

Some genetic conditions begin to manifest in early childhood; for example, those associated with intellectual and social development. Others, such as cancers that are more common in childhood, as well as rare genetic disorders that involve a complex diagnostic

odyssey before being identified, also are encountered at this stage. For parents and families, the diagnosis of genetic conditions in early childhood often sets off an intense and emotionally-fraught period of interaction with the health care system. Genetic testing of family members and consultations with many specialists are common, and depending on the patient's location, can involve long-distance travel, interruption to jobs and family schedules, and significant costs.

### Adolescence

Genetic services needed during adolescence tend to be those related to diagnosing early-onset diseases, such as cardiac conditions like hypertrophic cardiomyopathy or long-QT syndrome. During the teenage years, screening for hereditary cancer syndromes present in the family may be necessary, and older adolescents may contemplate genetic testing for adult-onset disorders for which they may be at risk, like Huntington's disease. Adolescents with genetic conditions may require support services as they transition out of pediatric-centered care and into adult health services. Similarly, if they are able to live on their own, they may need support transitioning out of the parent-run household.

### Adult

For young adults, genetic services often focus on reproductive planning. Carrier screening is common in at-risk populations. Also in adulthood, screening and diagnostic testing for hereditary cancer syndromes occurs, as well as diagnostic testing for adult-onset conditions that begin to manifest. While complex conditions such as cardiovascular disease and mental health disorders are not usually diagnosed using genetic testing, they have a strong hereditary component, and risk can be identified through analysis of family history. Cancers diagnosed during adulthood often are genetically analyzed to determine their molecular cause and to direct therapeutic plans. For many of the genetic services patients experience as adults, genetic counseling is helpful for understanding what the results mean for risk, future health complications and for passing on diseases to future generations.

### Elder Adult

As adults enter their elderly years, utilization of health services often increases. Neurodegenerative disorders such as Parkinson's disease, Alzheimer's disease and other dementias; cardiovascular complications like stroke and heart attack; and late-onset cancers affect a large number of people in this age group. Genetic testing for these and other conditions in the elderly is not common; however, many in this age group take multiple medications and are at risk for drug interactions and adverse events. Pharmacogenomic testing is sometimes used to assist in optimal drug choice and dosing, reducing the chances for adverse events. For families and caregivers of elderly patients, assistance is often needed in ensuring that their care needs are met and their living environments are amenable to their condition.

## V. Building the Genetic/Genomic Infrastructure

To achieve the promise of precision medicine – improved prediction, prevention, detection and treatment of disease – a computer-based infrastructure must be built. This is required to harness the power of clinical data linked with molecular data, to begin storing and utilizing genetic and genomic data from a centrally-managed resource. The construction of these ‘Omic data repositories will be key in support of genetic-based care coordination.

Just one file of genomic code is very large. A whole genome sequence can take the equivalent of 100 feature length movies or 150 gigabytes of data storage. The ‘Omic repository, however, will be just one of several components required build a comprehensive clinical genetic/genomic infrastructure. Other components will be:

- data pipelines run by bioinformatic tools that capture raw data from the sequencing machine to create clinically useful variant data files (called VCF files), which are specific to the person;
- Hadoop data lakes to store the larger pre-VCF files (BAM/SAM and FASTQ) for future research and analysis;
- databases that go beyond DNA storing variants that manage single-nucleotide polymorphisms, insertions/deletions, copy-number variants, alleles, and whole-genome/whole-exome sequences, but also other data like protein sequence, gene expression, methylation, and epigenomic data;
- interface and data integration capabilities that can link to data from multiple sources (sequencing machines, labs, electronic health records, clinical data warehouses, etc.);
- analytic capabilities that can be integrated as modular tools first for annotation, then clinical interpretation and application of genetic information for targeted patient treatment;
- and finally, external applications of all kinds to find, display, understand, and act on this data, for doctors, patients and researchers, made possible via application programming interfaces (APIs).

With this infrastructure in place, the complex process of discovery, treatment, and cure requires that genomes from a great many individuals—perhaps millions— must be collected, stored, and analyzed before the full extent of human variation becomes known and understood.

### Integrating with Critical Clinical Systems

With the ‘Omic repository playing a central role, the key requirement is to link the data to the electronic health record. When a clinician orders a drug, needs genetic information for a diagnosis, or a risk analysis to begin early screening, the EHR can query the repository and get the genotypic data it needs and copies the data locally. In fact, with just these two components and one clinical genetic interpretation service, the basics of precision

medicine are supported. Standardized data models, standards-based data transmission and APIs are being developed and adopted by many system vendors to achieve this goal.

### Patient-Generated Genetic/Genomic Data

With the fear of online genetic testing services causing harm having been largely unproven, more and more health care consumers could be generating their own genetic/genomic data in this manner. Many technology start-ups will be offering apps and other services that will be patient-centered and patient-driven. These consumers will likely demand that their data be in their EHR. Even traditional research protocols that produce data from studies and clinical trials will be required to give access to the study participants. This new model will be widely adopted because of the influence of the Precision Medicine Initiative's decision to require data access.

### Genetic-Based Clinical Decision Support

One of the major goals of building a clinical genomic infrastructure is to combine clinical, genetic, family health history, occupational and environmental information, to achieve optimal risk assessment and advanced clinical decision support. The generation of a patient genomic profile will become the standard of care. As mentioned before, this will include data beyond DNA - such as proteomic, gene expression, epigenomic and microbiome data, to complete a metabolic pathway.

### The Critical Role of Data Standards

There are several national and international activities happening concurrently that are developing and testing data capture and storage, data access and transmission, and data analysis and processing of genetic/genomic data. Health Level Seven® International (HL7) has created standards for the interoperability of family history and genetic sequence data between systems, apps and repositories. Besides the previously developed HL7 Version 2, Version 3 and Clinical Document Architecture (CDA)-based standards, new efforts are focusing on translating these same data structures into HL7 FHIR® (Fast Healthcare Information Resources). Other groups like the Global Alliance for Genomics and Health have created application-programming interfaces (APIs) to access data in any genomic database. The Healthcare Services Platform Consortium (HSPC) is leading an effort to standardize these APIs. ClinGen is working to develop a variant nomenclature that is more robust and computable. The National Academies have brought together health care systems, EHR vendors and labs to pilot sending coded genetic lab results using HL7 and LOINC standards to an EHR for use in CDS. Finally, the SMART on FHIR effort will result in a standards-based, interoperable apps platform for electronic health records, including the use of family health history and genetic data.

## VI. Data Privacy and Security

### Protections Against Genetic Discrimination

Given the rapid advance of genomic technologies that are transforming health care, consistent, robust data privacy protections are needed and will help to foster patient trust and engagement in care. The Genetic Information Nondiscrimination Act of 2008 (GINA) is intended to protect individuals from improper use of genetic information by health insurers and employers. Title I of GINA prohibits group and individual health insurers from using a person's genetic information in determining eligibility or premiums and prohibits health insurers from requesting or requiring that a person undergo a genetic test in order to collect genetic information on that person for underwriting decisions. Title II of GINA prohibits employers from using a person's genetic information in making employment decisions such as hiring, firing, job assignments or any other terms of employment. Title II also prohibits employers from requesting, requiring or purchasing genetic information about a person or their family members. However, GINA does not extend to life, long-term care or disability insurance, and certain populations are not protected by its provisions.

In addition to GINA, other laws provide partial privacy protections in the health insurance realm. The Health Insurance Portability and Accountability Act of 1996 (HIPAA) specifically lists genetic information as protected health information and explicitly states that a genetic risk factor for disease cannot be considered a preexisting condition. HIPAA prevents health insurers from increasing the cost of an individual's insurance discriminatorily, but insurance companies may raise an employer's group premiums based on the genetic information of its employees as a whole. Other federal and state laws provide a patchwork of varied protections. Slightly fewer than half of U.S. states have laws providing additional protection in aspects of life, long-term care and disability insurance, as well as in other areas, that are not present in GINA. For example, California law prohibits genetic discrimination in such areas as housing, mortgage lending, education, life insurance and elections. Arizona statute prohibits the use of genetic information in the underwriting of life and disability insurance policies. In contrast, many states' protections are no stricter than those afforded by GINA. Importantly, GINA does not preempt state law in states that provide more comprehensive protections than those provided by GINA.

### Ensuring Data Security

As genetic and genomic data is generated for the patient's benefit, and as care coordination requires the sharing of that data, security issues must be addressed. Security policies should be in place to identify and track who gets access to the information. Policies should also govern the safe transmission of data for risk assessment, analysis and interpretation, diagnosis, and decision making for treatment, and finally, to secure the data, as with other patient longitudinal record data, in a safe environment. Emerging investment in this space includes the use of block chain technology and strong private key encryption to maintain digital DNA privacy and individual anonymity, allowing for even more secure data sharing.

## Patient-Controlled Consent

A key component of data security in care coordination is the ability for the patient to control how the data is accessed and used. A consent management system would not only allow the management of current medical data, but also family health history and genetic test result data. Patient control of data is still in its infancy, but growing with recently released Meaningful Use and MACRA regulations.

## VII. Financial Issues

As payers and providers move further down a path away from fee-for-service reimbursement and towards value-based payment models, the need for coordinated care management will continue to be more important. Both payers and providers have complimentary capabilities in this area that when offered in concert with one another, can move toward optimizing the health of individuals. A key aspect of the harmonious synchronization of these activities is access to perfect data sets.

### Genetic Data for Analytics

While perfection is a lofty goal, more well-codified data to serve as a foundational basis for analytics is desirable. As data become more readily available and well structured, it becomes easier to parse data into meaningful analytics that can better predict the onset of health events and better target individuals for intervention before issues arise. Further, with better access to data and robust analytics, care can be streamlined without duplication of diagnostic resources, for an optimal approach to quality, cost and ultimately patient outcomes.

Readily available and well-codified genomic data to integrate into analytic models presents the next great land grab opportunity for health care as an industry. As both the cost of testing decreases and the scientific knowledge of the genome increases, actionable insight on available data is more often able to be part of the data and analytic foundation necessary for care coordination.

Payers, for some time now, have relied on well-structured data from medical, behavioral health, pharmacy and laboratory claims to better understand early opportunities for intervention with their members. Access to germ-line genomic data for the sole purposes of health care operations and care coordination will undoubtedly make payer models better at supporting individuals. Partnering with providers using genomic data, especially in a reimbursement model that is based on value where providers are best aligned with a need to not duplicate diagnostic tests and leverage all information and analysis currently available, will also foster a greater push toward health care's triple aim of optimizing quality, outcomes, and cost.

## Costs and Population Health

One common question will be who pays for genetic testing. Again, as the price point continues to fall and as consumers are more often provided opportunities outside of the traditional payer-provider provision of services, this question becomes less relevant. However, more relevant questions might be where results of genetic testing are stored, how is that cost managed, how is the data accessed and where value can be both captured and compensated. These questions are all being tackled today by the testing industry and payer-provider collaboratives interested in the data and analysis that ensues; they will need to recognize the importance of such data and the value thereof in order to open access for a large population. Undoubtedly, there is a role to play for many players in the health care rent chain. Pharmaceutical manufacturers will continue to grow more interested in the support and payment for testing, as better research will yield more targeted therapy molecules to treat disease. Payers and providers should be equally interested in supporting both data collection and well-structured data storage for access in order to target intervention with individuals for the proactive management of disease, to stave off or get in front of management of issues. There can be a great benefit to this, not only economically, but most importantly for the quality of life individuals will have if proactive self-management ensues.

The industry must move toward a common standard for genomic data (testing, markers and results interpretation), not dissimilar to procedural coding, diagnostic coding, drug coding, etc. The ability for payers and providers to leverage this data for a more complete picture of the individual and to run this data against a larger set of tens of millions of individuals will only improve the predictive power of analytics and, in the process, create a more robust care coordination offering for the benefit of individuals and the nation at large.

## **VIII. Call for Deeper Research, Publications and Conference Sessions**

### Summary and Conclusion

The presence of a broad set of genetic information for the health care community (patients, providers, payers and others) to access for shared decision support is no longer a promise, but a reality of the day in which we live. As both cost and access to testing barriers continue to be removed, now, more than ever, is the time for the industry to not only consider, but move aggressively toward a common standard for genetic information exchange. Supporting a common standard for all parties will allow for better access, more seamless integration and an informative position on infrastructure, architecture, and security requirements for housing data. As this paper outlines, such a standard will benefit patients, providers, payers and others, irrespective of the entry point of genetic information into the ecosystem and throughout the lifetime of an individual. As more genetic variations on the human genome become known, with respect to clinical significance, the common standard will allow for a distinct framework to receive further scientific breakthrough for the benefit of patient care and healthier living. As such, this paper should serve as the



continuation of a dialogue and a call for deeper research and discussion between and among all stakeholders that can influence and benefit from this process.

## **IX. Further Reading**

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## **X. Authorship**

The following Genomics Workgroup members authored this white paper:

Grant M. Wood (Workgroup Co-Chair)  
Intermountain Health

Katherine Johansen Taber (Workgroup Co-Chair)  
American Medical Association

Adam Scott (Workgroup Co-Chair)  
Active Health Management

M'Lynda Owens  
Cognosante

Rosalyn Ryan  
Dell

Virginia Daniel  
NASCO

The following Genomics Workgroup members contributed to the ideas that are included in this white paper:

Allison Keenan  
Xerox Care and Quality Solutions, Inc.

Simon Lin  
Nationwide Children's Hospital

David A. Stumpf  
Northwestern University, Woodstock Health Information & Technology

Scott Whyte  
ClearDATA