

COMPUTER-IMPLEMENTED DASHBOARD PROVIDING DYNAMIC DIGITAL HEALTHCARE DATA

TECHNICAL FIELD

5 This disclosure relates to computer-implemented methods, computer-readable media and computer systems to generate computational resources directed to health, wellness and other data of individuals such as patients and displaying the generated computational resources in display devices for consumption by patients or others.

BACKGROUND

10 Patients, medical care providers (e.g., clinicians, physicians), employers, health plans, life insurers and drug developers (e.g., pharmaceutical companies) each consumes information that can differ based on their specific needs and context. Often, an individual(s) viewing digital data, for example, on a display device of a computer system,
15 is interested in obtaining comparative information associated with the data and/or more details to allow the individual to make informed decision about the underlying healthcare issue, priorities, cost, risk and risk mitigation actions, management, interventions (e.g., treatment) and monitoring that the data addresses.

SUMMARY

20 This specification describes technologies relating to computer-implemented methods, computer-readable media and computer systems directed to generating and displaying a dashboard to provide digital data.

 In one aspect, methods are provided that include: generating, by a computer
25 system, a user interface for display on a display device operatively coupled to the computer system; generating, by the computer system, first data comprising genetic, epigenetic, and/or other biomarker test results for a patient to evaluate the presence or absence of a disease, disorder, or risk in the patient; displaying the user interface on the display device; and displaying the first data within the user interface.

30 In some embodiments, the data is selected from measurements (e.g., weight, blood pressure, etc.), test results (e.g., genetic, epigenetic, protein, metabolic assays, etc.), financial (e.g., cost of procedures, cost of medication, cost of claims, etc.), industry / prevalence (e.g., heart disease prevalence in a particular geographic region, number of

heart attacks a year among truck drivers, etc.), interventions (e.g., surgery, etc.), medical (e.g., ICD10 codes, medication usage, etc.), clinical (e.g., blood pressure, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), supplement usage (e.g., herbs, vitamin D, vitamin B12, iron, etc.), electronic data (e.g., imaging, electronic health records, publicly available data (e.g., repositories, etc.), publications, pharmaceuticals, clinical trials, etc.) and the diagnostic, intervention, management, monitoring and/or therapeutic implications thereof. In some embodiments, the data includes standalone data, aggregated data, imputed data, derived data, numerical data, text data, image data, and combinations thereof.

In some embodiments, the data is displayed in the user interface in one or more plots. In some embodiments, the data is compared to corresponding genetic, epigenetic, and/or other biomarker test results from a cohort population. In some embodiments, the data is superimposed on genetic, epigenetic, and/or other biomarker test results for a population.

In some embodiments, the user interface displays diagnostic information, prognostic information, uncertainty quantifications, clinical and/or business risk management or decision-making, regulatory and/or policy making, intervention, monitoring and/or therapeutic decisions and strategies related to a patient or a group of patients or a user or a group of users.

In some embodiments, the genetic, epigenetic, and/or other biomarker test results for the patient population comprises a bar chart showing ages of the patient population on an X-axis and probabilities that patients in different ages have the disease, disorder or risk on a Y-axis, wherein the test results for the patient comprise a probability that the patient has the disease.

In some embodiments, the patient is associated with an age, wherein the method further comprises displaying the probability that the patient has the disease adjacent to a probability of the patient population of the same age as the patient.

In some embodiments, the patient is associated with a gender, wherein the method further comprises displaying the probability that the patient has the disease adjacent to a probability of the patient population of the same gender as the patient.

In some embodiments, the methods further include: receiving a ranking of contributions of a plurality of genetic, epigenetic, and/or other biomarkers including the

genetic, epigenetic, and/or other biomarker to the presence of the disease in the patient; generating a chart comprising names of the plurality of markers on an X-axis and a normalized rank of the contributions of the plurality of genetic, epigenetic, other biomarkers for the patient; superimposing the chart on another chart showing normalized
5 ranking of contributions of the plurality of genetic, epigenetic, and/or other biomarkers to the patient population resulting in a second plot of rankings versus markers; and displaying the second plot adjacent the plot within the user interface.

In some embodiments, the methods further include: generating a plot of a genetic, epigenetic, and/or other biomarker measurement distribution associated with a specific
10 genetic, epigenetic, and/or other biomarker; superimposing on the plot, the genetic, epigenetic, and/or other biomarker measurement measured for the patient; and displaying the plot with the superimposed genetic, epigenetic, and/or other biomarker measurement within the user interface.

In some embodiments, the methods further include: determining an upper limit
15 and a lower limit of uncertainty associated with the genetic, epigenetic, and/or other biomarker measurement measured for the patient; and displaying, within the plot, the upper limit and the lower limit bounding the genetic, epigenetic, and/or other marker measurement.

In some embodiments, the methods further include: detecting a selection of the
20 data displayed within the user interface; in response to detecting the selection, displaying a window adjacent the data within the user interface; and displaying, within the window, hyperlinks to further information.

In some embodiments, the further information comprises literature related to the genetic, epigenetic, and/or other biomarker test results or literature related to the disease,
25 disorder or risk.

In some embodiments, the literature is selected from publications, clinical data, clinical trials and pharmaceuticals.

In still another aspect, computer-readable media storing computer instructions are provided, which, when executed by one or more computer processors, are configured to
30 cause the one or more computer processors to perform operations comprising any of the methods described herein.

In yet another aspect, computer systems are provided that include one or more computer processors; and a computer-readable medium storing computer instructions

which, when executed by the one or more processors causes the one or more processors to perform operations comprising any of the methods described herein.

The term “genome” as used herein, refers to the entirety of an organism's hereditary information that is encoded in its primary DNA sequence. The genome
5 includes both the genes and the non-coding sequences. For example, the genome may represent a microbial genome or a mammalian genome.

“Nucleic acid,” “oligonucleotide,” and “polynucleotide” refer to deoxyribonucleic acids (DNA) or ribonucleic acids (RNA) and polymers thereof in either single- or double-stranded form. Unless specifically limited, the term encompasses nucleic acids containing
10 known analogues of natural nucleotides that have similar binding properties as the reference nucleic acid. The term nucleic acid is used interchangeably with genetic material, cDNA, mRNA, and gene.

Reference to “DNA region” should be understood as a reference to a specific section of DNA. These DNA regions are specified either by reference to a gene name, a
15 set of chromosomal coordinates or Reference single nucleotide polymorphisms (SNPs). Both the chromosomal coordinates and the gene or genomic region would be well known to, and understood by, the person of skill in the art. In general, a gene or genomic region can be routinely identified by reference to its name, via which both its sequences and chromosomal location can be routinely obtained, by reference to its chromosomal
20 coordinates, via which both the gene or genomic region name and its sequence can also be routinely obtained, or by sequence, via which both the gene and genomic region can be routinely obtained.

Reference to each of the genes/DNA regions detailed herein should be understood as a reference to all forms of these molecules and to fragments or variants thereof. As
25 would be appreciated by the person of skill in the art, some genes are known to exhibit allelic variation. Allelic variations encompass single nucleotide polymorphisms, insertions and deletions of varying size and simple sequence repeats, such as dinucleotide and trinucleotide repeats. Variants include nucleic acid sequences from the same region sharing at least 90%, 95%, 98%, 99% sequence identity, i.e., having one or more
30 deletions, additions, substitutions, inverted sequences etc. relative to the DNA regions described herein. Accordingly, the compositions and methods described herein should be understood to extend to such variants. The compositions and methods described herein

should therefore be understood to extend to all forms of DNA which arise from any other mutation, polymorphic or allelic variation.

The term “sequencing” as used herein refers to sequencing methods for determining the precise order of the nucleotide bases (e.g., adenine, guanine, cytosine, and thymine) in a nucleic acid molecule (e.g., a DNA or RNA nucleic acid molecule). It includes any method or technology that is used to determine the order of the four bases in a strand of DNA.

The term “barcode” as used herein, refers to any unique, non-naturally occurring, nucleic acid sequence that can be used to identify a nucleic acid molecule.

Real time, derived, and predicted data (e.g., collected from samples, electronic health records, devices, etc.) can be collected and stored, and thus, become historic data for ongoing or future decision-making for a process, setting, or application.

A “computer-readable medium”, is an information storage medium that can be accessed by a computer using a commercially available or custom-made interface.

Exemplary computer-readable media include memory (e.g., RAM, ROM, flash memory, etc.), optical storage media (e.g., CD-ROM), magnetic storage media (e.g., computer hard drives, floppy disks, etc.), punch cards, or other commercially available media. Information can be transferred between a computer system and a medium, between computer systems, or between a computer system and a computer-readable medium for storage or access of stored information. Such transmission can be electrical, or by other available methods, such as IR links, wireless connections, etc.

Where a range of values is provided, it is understood that each intervening value, between the upper and lower limit of that range and any other stated or intervening value in that stated range is encompassed herein. It would be appreciated that test results, data, measurements, and biomarker (or marker) can be used interchangeably. For example, measurements can be measured or obtained from a patient (e.g., genetic biomarkers, epigenetic biomarkers, inflammation biomarkers, lipids, weight, etc.). For example, biomarkers can be measured (e.g., the percentage of methylation) or can be represented with binary data (e.g., yes or no; presence or absence; adenosine (A) or guanine (G)). For example, data can include one or more measurements from, e.g., test results, biomarker information (e.g., measured or binary), lifestyle information (e.g., lifestyle (e.g., smoker, drinker), medication or supplement use, etc.), and/or any number of other sources of

information (e.g., from public databases, publications, insurance claims, population statistics, costs or cost estimates, etc.).

The details of one or more implementations of the subject matter described in this specification are set forth in the accompanying drawings and the description below.

5 Other features, aspects, and advantages of the subject matter will become apparent from the description, the drawings, and the claims.

BRIEF DESCRIPTION OF THE DRAWINGS

FIG. 1 is a schematic diagram of a computer system configured to display the
10 dashboard described in this disclosure.

FIG. 2 is an exemplary user interface that can be displayed in the display device.

FIG. 3 is an exemplary user interface that can be displayed in the display device.

FIG. 4 is an exemplary user interface that can be displayed in the display device.

FIG. 5 is an exemplary user interface that can be displayed in the display device.

15 FIG. 6 is an exemplary user interface that can be displayed in the display device.

FIG. 7 is an exemplary user interface that can be displayed in the display device.

FIG. 8 is an exemplary user interface that can be displayed in the display device.

FIG. 9 is an exemplary user interface that can be displayed in the display device.

FIG. 10 is an exemplary user interface that can be displayed in the display device.

20 FIG. 11 is an exemplary user interface that can be displayed in the display device.

FIG. 12 is an exemplary user interface that can be displayed in the display device.

FIG. 13 is an exemplary user interface that can be displayed in the display device.

FIG. 14 is an exemplary user interface that can be displayed in the display device.

FIG. 15 is an exemplary user interface that can be displayed in the display device.

25 FIG. 16 is an exemplary user interface that can be displayed in the display device.

FIG. 17 is an exemplary user interface that can be displayed in the display device.

FIG. 18 is an exemplary user interface that can be displayed in the display device.

FIG. 19 is an exemplary user interface that can be displayed in the display device.

FIG. 20 is a block diagram illustrating an example computer system used to
30 provide computational functionalities associated with described algorithms, methods, functions, processes, flows, and procedures as described in the present disclosure, according to some implementations of the present disclosure.

Like reference numbers and designations in the various drawings indicate like elements.

DETAILED DESCRIPTION

5 This disclosure describes a platform that an individual (e.g., a patient), a clinician (e.g., a physician, a nurse practitioner) or another user (e.g., a pharmacist, a researcher, an employer, an insurer) can receive and understand standalone or aggregate information related to measurements (e.g., weight, blood pressure, etc.), test results (e.g., genetic, epigenetic, protein, metabolic assays, etc.), financial (e.g., cost of procedures, cost of medication, cost of claims, etc.), industry / prevalence (e.g., heart disease prevalence in a
10 particular geographic region, number of heart attacks a year among truck drivers, etc.), interventions (e.g., surgery, etc.), medical (e.g., ICD10 codes, medication usage, etc.), clinical (e.g., blood pressure, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), supplement usage (e.g., herbs, vitamin D, vitamin B12, iron, etc.), electronic data
15 (e.g., imaging, electronic health records, publicly available data (e.g., repositories, etc.), publications, pharmaceuticals, clinical trials, etc.) and/or the diagnostic, intervention, management, monitoring and/or therapeutic implications thereof.

 The platform can provide diagnostic information, prognostic information,
20 associated risk factors, uncertainty quantifications, etc. The platform can allow the individual (e.g., a patient), clinician, or another user to drill down into additional information that is deemed relevant based on the measurements (e.g., weight, blood pressure, etc.), tests (e.g., genetic, epigenetic, protein, metabolic assays, etc.), financial (e.g., cost of procedures, cost of medication, claims, etc.), industry/prevalence (e.g., heart
25 disease prevalence in a particular geographic region, number of heart attacks a year among truck drivers, etc.), interventions (e.g., surgery, etc.), medical (e.g., ICD10 codes, medication usage, etc.), clinical (e.g., blood pressure, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), supplement (e.g., herbs, Vitamin D, Vitamin B12,
30 Iron, etc.) and/or additional data (imaging, electronic health records, publicly available data (e.g., repositories, etc.), publications, pharmaceuticals, clinical trials etc.) to assist the physician or others with, for example, understanding and implementing diagnostic, clinical and/or business risk management or decision-making, regulatory and/or policy

making, intervention, monitoring and/or therapeutic decisions and strategies related to a patient or a group of patients or an user or a group of users. Furthermore, the platform can provide the ability to assist with drilling down, searching, interpreting, collating, communicating, and providing additional information (e.g., journal articles, clinical guidelines, raw or transformed data, etc.) with artificial intelligence or other algorithms (e.g., large language models). Although “patient” is used frequently throughout this disclosure, the term is not limited to individuals who have been previously diagnosed with a disease or a disorder and also is not intended to be mutually exclusive from “stakeholder” or “user”.

The compositions and methods described herein can be used to provide changes in management, outcomes (e.g., health, cost, etc.) or other decision-making for a user (e.g., clinician, payor, employer, researcher, insurer, government, etc.) for a patient or a group of patients or users with or without a disease, disorder, or risk. The compositions and methods described herein are discussed in the context of cardiovascular disease (e.g., coronary heart disease, stroke, etc.) and cardiovascular risk factors or co-morbidities (e.g., obesity, smoking, alcohol consumption, etc.), but the compositions and methods described herein can similarly be applied in the context of cardiometabolic disorders (e.g., hypertension, high lipoprotein, etc.), insulin-related health conditions (e.g., pre-diabetes, type 2 diabetes, etc.); dementia (e.g., Alzheimer’s disease, etc.), cerebrovascular disease, mortality, cancer (e.g., breast cancer, etc.), other complex disorders or diseases (e.g., autism, mental health, COPD, kidney disease, etc.), and any number of other diseases, disorders, or risks.

FIG. 1 is a schematic diagram of a computer system 100 configured to run and display the platform described in this disclosure. The computer system 100 includes one or more processors and a computer-readable medium (e.g., a non-transitory computer-readable medium) storing computer instructions which, when executed by the one or more processors, can perform operations described in this disclosure. The computer system 100 can generate a “dashboard,” comprised of one or more user interfaces (see, e.g., FIGS. 2-19) for display on a display device 105 that is operatively coupled to the computer system 100.

As described herein, genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein for a patient, a population of patients, or a user can be represented and displayed (e.g.,

graphically, textually) by the computer system 100 on the display device 105 via any number of user interfaces. For example, genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described herein for a population (e.g., a patient population) to which the patient belongs (e.g.,
5 based on age, gender, race, ethnicity, etc.) can be represented in standalone or aggregated form(s) and depicted for the patient alone, for a non-patient user (e.g., clinician, technician, etc.), or relative to a population (e.g., a population of cohorts). The same genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or data described herein for a population can be obtained,
10 and the genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or data described herein for the population can be categorized into cohorts of individuals (e.g., patients or non-patients) within the same or different demographic of the individual (e.g., age, gender, race, ethnicity, etc.). As used herein, test results or measurements refer to, without limitation, the results or data
15 obtained from genetic and/or epigenetic testing, but also can refer to the presence, absence or amount of a protein, features detecting during imaging, blood pressure, lifestyle (e.g., smoking, alcohol consumption, etc.), results from metabolic assays, the results of blood work, financial (e.g., cost of procedures, cost of medication, claims, etc.), industry / prevalence (e.g., heart disease prevalence in a particular geographic region,
20 number of heart attacks a year among truck drivers, etc.), interventions (e.g., surgery, etc.), medical (e.g., ICD10 codes, medication usage, etc.), clinical (e.g., blood pressure, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), supplement (e.g., herbs, Vitamin D, Vitamin B12, Iron, etc.) and/or additional data (electronic health records, publicly available data (e.g., repositories, etc.), publications, pharmaceuticals,
25 clinical trials etc.).

As described herein, the compositions and methods involve processing of a variety of different types of data (e.g. lifestyle, medical history, previous and current diagnoses (e.g., according to the International Classification of Diseases, Tenth Revision (ICD10)), occurrence of indication (e.g., by age, by gender, etc.)), which can be used for
30 risk assessment or quality control, or to create training, test validation data sets and/or iterative training for machine learning, train, test or validate a machine learning or other algorithm(s). Additionally, in some embodiments, the compositions and methods described herein are capable of rapid and/or high-throughput processing of samples and

data generated from sample processing, which can be used in the rapid generation of personalized care / intervention / management / monitoring / treatment program components.

5 *User Interfaces*

Examples of user interfaces that can be displayed are shown in FIGs. 2-19, however, these are not intended to be limiting. The computer system 100 can display the user interface in a static or dynamic form. The computer system 100 can display a number of different forms of data (e.g., standalone, aggregated, imputed, derived, numerical, text, 10 images, or combinations thereof), plots, test results, publications, or hyperlinks to any of the aforementioned, within one or more user interfaces to evaluate the presence or absence of a disease or disorder or risk in a patient or population. Using input devices 110, a user can interact with the plots in the user interface, e.g., to view different items of information included in the plots (e.g., in drop down menus, embedded links, etc.); to drill 15 down into the results, the data, the methods, the analysis, and/or the interpretation; to overlay or superimpose data, test results, etc., for a patient or user or group of patients or users onto corresponding data, test results, etc. for a population; and/or searching, interpreting, collating, communicating, and providing additional information (e.g., journal articles, clinical guidelines, raw or transformed data, etc.) with artificial intelligence or 20 other algorithms (e.g., large language models); provide artificial intelligence assistant (e.g., large language model) for providing references (e.g., journal articles, clinical guidelines), context, and/or query information, etc.

By employing the user interfaces described in more detail below, patients, physicians or other individuals or users or groups of users can review the patient's 25 information compared to the population as a whole or grouped by age and/or gender or other confounding variables. The patient information may be provided as de-identified matter. Furthermore, users can compare population information as a whole or grouped by age and/or gender or other confounding variables. Methods of determining the genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test 30 results and/or other data described herein for one or more patients or one or more users are known in the art or, if that information is already known for the patient(s) or user(s), that information can be input into the computer system 100.

The computer system 100 described herein can be configured to perform univariate biomarker analysis, which is primarily used to compare results for user(s) to a data source (e.g., an external source such as, without limitation, third party studies). The computer system 100 described herein also can be configured to perform multivariate risk and diagnostics predictions using a multitude of data sources (e.g., measurements, electronic health records, lifestyle, etc.). The system can perform both univariate and multivariate analysis on the same source for user(s).

FIGs. 2A and 2B show exemplary user interfaces that can be displayed by the display device. In a user interface, the computer system 100 can display patient information (e.g., patient ID, name, age, gender, etc.), user information (e.g., name, ID and contact information of a physician or other user requesting and/or reviewing the information), and/or sample information (e.g., type of specimen, date obtained from patient, etc.). While “clinician” is used in both of the user interfaces shown in FIGs. 2A and 2B, the same or different information can be configured similarly or differently in user interfaces for any number of users as discussed herein.

FIGs. 3A and 3B show exemplary user interfaces that can be displayed by the display device. In a user interface, different graphical formats can be used for stratifying patient information. For example, the computer system 100 can generate a plot as a vertical bar chart showing ages of the patient population on an X-axis and probabilities that patients in different ages have a disease or disorder on a Y-axis (FIG. 3A) or, conversely, as a horizontal bar chart showing probabilities that patients in different ages have a disease or disorder (e.g., a score) on an X-axis and ages of the patient population on a Y-axis (FIG. 3B). Genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein for the patient can include a probability value that the patient has the disease or disorder (or does not have the disease or disorder), and the computer system 100 can display the probability determined for the patient on a bar chart of populational probabilities. This allows a user to compare the probability of the patient having the disease or disorder relative to the age-related probabilities of the disease or disorder in the population.

In some implementations, the computer system 100 can generate a plot (e.g., a bar chart) showing a gender of the patient population on an X-axis and probabilities that patients of the same gender have the disease on a Y-axis, and the gender shown can be selected to match the gender of the patient. The computer system 100 can display the

probability determined for the patient relative to the population probabilities for individuals of the same gender as the patient. Similarly, the computer system 100 can display the probability determined for the patient relative to the population probabilities for individuals of the same race, ethnicity, or other feature as the patient.

5 FIG. 4 is an exemplary user interface that can be displayed by the display device. The user interface in FIG. 4 is a graph showing the risk of coronary heart disease (CHD) in a male patient relative to a population of male cohorts. For example, when the disease, disorder or risk relates to CHD, cohort data can be obtained from the Framingham Heart Study Offspring study, the Intermountain Healthcare (IM) study, and/or the Iowa study,
10 which are well-known in the art.

 FIG. 5 is an exemplary user interface that can be displayed by the display device. In some implementations, results can be obtained from more than one (e.g., multiple) genetic, epigenetic, and/or other biomarker from the patient. Different genetic, epigenetic, and/or other biomarker can have different levels of contribution towards association or
15 causation of a disease, disorder, indication, condition, or risk in a patient. The contribution can be obtained directly from model(s) or from other approaches such as SHapley Additive exPlanations. In some implementations, the computer system 100 receives a ranking of contributions of each of multiple genetic markers to the presence of the disease in the patient. The computer system 100 can generate a user interface that
20 includes names of each marker on the X-axis and a rank or score (e.g., normalized between 0 and 1) of the contributions of each biomarker disease, disorder or risk. For example, the computer system 100 can generate a graph that shows the biomarker that contributes most to the disease or disorder or risk and the genetic marker that contributes least to the disease, disorder or risk, with the remainder of the genetic markers arranged in
25 decreasing rank in between (FIG. 5). Generally, the contribution can be compared to a population as a whole or provided for a specific individual or aggregated in a group, regardless of age or gender. In some implementations, the computer system 100 can generate a Table 1 (see below), describing the relevant genetic and/or epigenetic biomarkers that are displayed in the user interface.

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Table 1

Associating to Risk Factor and Therapeutics	
Methylation	
cg1	site located in the body of the X gene. This CpG site, which is involved in the X pathway, has been shown to be strongly associated with cardiovascular health and risk of cardiovascular disease [reference]
cg2	has been shown to be associated with X drug [reference]
cg3	located between X bases upstream of the transcription start site (X) of the X gene. The X gene has been linked to X and has also been linked to CHG [reference]
cg4	site located in the body of the X is associated with X score [reference]
Genotype	
rs1	has been shown to be associated with risk factors for CHD, with this SNP being linked to obesity [reference]
rs2	is located within the X locus has been shown to be significantly associated with CHD [reference]

5 The computer system 100 superimposes the results of the tests of the genetic, epigenetic, and/or other markers for the individual patient on corresponding ranks of the tests of the genetic and/or epigenetic and/or other markers determined for a population as a whole. The computer system 100 displays the resulting plot in the user interface.

10 The plot shown in user interface shows the contribution of each marker to the disease in aggregate compared to the contributions in the individual patient. For example, if a first genetic and/or epigenetic and/or other marker (e.g., cg0) is ranked to have the greatest contribution towards a disease in the general population, but the same genetic and/or epigenetic and/or other marker has a comparatively low contribution towards the disease in the individual patient, then a physician is informed that the first genetic and/or epigenetic and/or other marker is likely of less importance to treating the individual patient. Conversely, if a different genetic and/or epigenetic and/or other marker (e.g., cg3)
15 is ranked to have the lowest contribution towards the disease in the general population, but the same genetic and/or epigenetic and/or other marker has the highest contribution towards the disease in the individual patient, then the physician is informed that the different genetic and/or epigenetic and/or other marker is likely of most importance to treating the individual patient. In this manner, the risk contribution can be used to
20 determine which of the markers significantly contribute to an individual patient's disease or a condition or indication.

As discussed herein, the computer system 100 can display each of the normalized ranks as a selectable object. Using an input device 115, a user can select the normalized rank for one of the genetic, epigenetic, and/or other biomarker. In response to detecting

the selection (e.g., a mouse click, a hover, a touch input, or any other input), the computer system 100 can display, e.g., information related to the genetic, epigenetic, and/or other biomarker (e.g., hyperlinks, publications, clinical data, clinical trial, pharmaceuticals, etc.).

5 FIG. 6 is another exemplary user interface that can be displayed by the display device. In some implementations, the computer system 100 can generate a box plot of a genetic, epigenetic, and/or other biomarker measurement distribution (e.g., for the whole patient population) associated with a specific genetic, epigenetic, and/or other biomarker. The computer system 100 can display the box plot with the superimposed genetic,
10 epigenetic, and/or other biomarker measurement for the patient within the user interface. For example, the computer system 100 can display the genetic, epigenetic, and/or other biomarker measurement for the patient as a dotted line across the box plot of the genetic, epigenetic, and/or other marker measurement distribution. The computer system 100 can similarly generate and display multiple box plots, each for a respective genetic,
15 epigenetic, and/or other biomarker, each displayed with the genetic, epigenetic, and/or other biomarker measurement for the patient. In some implementations, for each box plot and, specifically, for the genetic, epigenetic, and/or other biomarker measurement measured for the patient, the computer system 100 can determine an upper limit and a lower limit of uncertainty associated with the measurement. The computer system 100
20 can display the upper and lower limits as solid lines on either side of (e.g., above and below) the dotted line that represents the genetic, epigenetic, and/or other biomarker measurement for the patient. The computer system 100 can similarly generate and display uncertainty limits for the multiple box plots corresponding to the multiple genetic, epigenetic, and/or other biomarker measurement distributions. The user interface can be
25 used to ensure robustness of the measurements and allow the users to compare individual patient and patient population measurements.

FIG. 7 is an exemplary user interface that can be displayed by the display device. In some implementations, the computer system 100 can display a selectable object (e.g., a drop down box) using which a user can select a specific marker (e.g., biomarker “rs1”). In
30 response to the selection (e.g., using the input device 115), the computer system 100 can retrieve (from, e.g., publicly available databases or privately held databases) allele frequencies for different populations (e.g., European, Asian, East Asian, etc.) (see Table 2), and provided as part of the user interface. A patient, physician or other user can

interact with the user interface to learn additional context regarding, e.g., ethnicity, age, gender, etc. Within a user interface, the computer system 100 also can display publicly available genome-wide association study (GWAS) data, which can show association of biomarkers to risk factors and whether the disease, disorder or risk was detected in
5 independent studies.

Table 2

Allele Frequencies for Marker rs1				
Population	Group	Sample size	Ref Allele	Alt Allele
Other Asian	Sub	96	C=0.88	T=0.12
Latin American 2	Sub	3,064	C=0.8535	T=0.1465
European	Sub	41,124	C=0.84150	T=0.15850
Asian	Sub	488	C=0.838	T=0.162
East Asian	Sub	392	C=0.829	T=0.171
Other	Sub	4,930	C=0.8229	T=0.1771
Total	Global	57,260	C=0.81633	T=0.18367
Latin American 1	Sub	360	C=0.769	T=0.231
South Asian	Sub	162	C=0.704	T=0.296
African American	Sub	6,904	C=0.6543	T=0.3457
African	Sub	7,132	C=0.6541	T=0.3459
African Others	Sub	228	C=0.649	T=0.351

10 FIG. 8 is an exemplary user interface that can be displayed by the display device.
As shown in FIG. 8, the computer system 100 can produce cost analysis data (Y axis)
over time (X axis) for cardiovascular disease, displayed based on various populations
(disease type including high blood pressure, coronary heart disease, congestive heart
failure, stroke, other cardiovascular diseases, and high blood pressure) using national and
15 regional data or statistics on prevalence and other data or factors. As discussed herein,
data can be further aggregated based on, for example, demographic information (e.g., age,
gender, geographic location, etc.). In some embodiments, a user interface can display cost
analysis (e.g., cost-minimization, cost-effectiveness, cost-utility, cost-benefits, cost-
consequence, budget impact, value of information, pharma-economics, increment cost-

effectiveness ratio etc.) and/or test utilization information such as that shown in Table 3 below, which can include hyperlinks to further review the cost analysis data based on month, year, employee population, tested population, etc. The cost analysis displayed in a user interface can help estimate treatment cost and outcomes at an individual, group of
5 individuals and/or population level.

Table 3. Representative Cost Utilization

Investment in Employee Health	\$122,856.14	Ave \$12,000/mo
Heart Disease Tests	1,000	Amt of Tests Contracted
Employee Population	1400	Enroll new employee in outreach

10 FIG. 9 is an exemplary user interface that can be displayed by the display device in which the computer system 100 causes the user interface to display coronary heart disease prevalence in the US population based on age and gender. As discussed herein, the data used to create such a user interface can be obtained from national and regional statistics that are available and can be displayed based on any number of factors (e.g.,
15 demographic; disease, disorder or risk; ethnicity; geography, organization, etc.).

FIG. 10 is another exemplary user interface that can be displayed by the display device. FIG. 10 shows an example in which the death rate due to coronary heart disease is depicted geographically via a color-coded map based on age-standardized rate (per 100,000). FIG. 10 as shown is independent of race, ethnicity, gender, but the user
20 interface can be configured to display data in which the contribution of those factors is shown.

FIG. 11 is an exemplary user interface that can be displayed by the display device. In this example, uncertainty measurements (e.g., uncertainty quantification (UQ)) is graphically represented for various biomarkers, although uncertainty also can be assessed
25 for measurements, models, costs, etc. This type of user interface allows for a patient, a physician or another user to assess risk due to, for example, potential deviations or errors in measurements (due to, e.g., differences in manufacturing assays, devices, technicians, and/or sample collection and viability, etc.) or modeling. In some instances, the user interface can allow a user to review and evaluate the uncertainty for measurements, the

uncertainty for modeling, or the net uncertainty, defined as the aggregate of the measurements and modeling uncertainty. In some embodiments, uncertainty can be used as a gauge for assessing the reliability of measurements in quality control processes. In some embodiments, the uncertainty can be utilized in conjunction with data, metrics
5 and/or requirements provided by a user to ensure that targeted outcomes (e.g., cost, likelihood of event, etc.) fall within an acceptable range. For example, this approach can be employed to ascertain that treatment costs do not exceed a specified limit or that the probability of a coronary heart disease (CHD) event remains below a defined threshold.

FIG. 12A and 12B are exemplary user interfaces that can be displayed by the
10 display device to demonstrate quality control processes in the methods involved in obtaining the genetic, epigenetic, and/or other biomarker measurements (e.g., digital PCR (dPCR)) and/or modeling the resulting data. This type of user interface can demonstrate whether all samples measurement falls within an expected reference measurement (FIG. 12A) or falls outside of an expected reference measurement (FIG. 12B) and, therefore, is
15 considered a failed test.

FIG. 13 is an exemplary user interface that can be displayed by the display device. In this example, the user interface displays a line graph showing a change in a genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein (Y axis) over time (X axis). This type of user
20 interface can allow a patient, a physician, or another use to assess changes (e.g., significant changes) over time for an individual patient or for an individual patient relative to a particular designated population.

FIG. 14 is an exemplary user interface that can be displayed by the display device. In this example, the user interface displays a line graph showing a change in score (Y
25 axis) based on, e.g., one or more genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described above, over time (X axis). In this example, the graph in the user interface indicates a threshold score distinguishing patients between a low score and a high score, along with the trend observed from population data. As discussed herein, the computer system 100 allows for
30 the display to be manipulated and configured by the user to present the information in any number of different formats (e.g., displaying the low and high score ranges for different patient populations). Changes in a patient's score over time can identify when a patient is responding or not to a therapeutic strategy. Furthermore, the uncertainty (e.g.,

measurements, modeling, and/or aggregated measurements and modeling) could be used to further inform the user(s) about confidence on the risk group for the result(s). In some implementations, uncertainty may not be included.

FIG. 15 is an exemplary user interface that can be displayed by the display device.

5 FIG. 15 shows a score based on, e.g., genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described herein used to compute the score. This type of user interface allows a user to evaluate the contribution of individual genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described herein to a score
10 (e.g., risk probability, diagnostic probability, etc.).

FIG. 16 is an exemplary user interface that can be displayed by the display device. The user interface example shown in FIG. 16 demonstrates the change in the contribution of specific measurements and/or other data described data (Y axis) over time (X axis) for a user or group of users.

15 FIG. 17 is an exemplary user interface that can be displayed by the display device. This example depicts a graphical illustration of the contribution of genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein (e.g., SHAP value, model weights, etc.) (X axis) in a population for several different contribution of genetic, epigenetic, other biomarker(s)
20 (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein (Y axis). As discussed herein, this user interface can be integrated, overlayed, superimposed, etc. with one or more user interfaces that, for example, display a change in contribution of specific biomarkers over time.

FIG. 18 is another exemplary user interface that can be displayed by the display
25 device. FIG. 18 depicts a pie graph that, in conjunction with information such as that contained in Table 4 below, can be used to perform univariate or multivariate analysis on various disease pathways in individuals and/or populations. This type of user interface also can be used to inform treatment and/or intervention recommendations.

30

Table 4

Marker	Maps to disease pathways related to:
Marker 1	Diabetes, CHD [Reference 1, 2,3]
Marker 2	Inflammation [Reference 4]
Marker 3	Plaque [Reference 5]
Marker 4	BMI [Reference 5]

FIG. 19A and 19B is an exemplary user interface that can be displayed by the display device. In FIG. 19A, the computer system 100 generates a graph corresponding to an individual's treatment response (Y axis), based on, e.g., genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described above, over time (X axis). In FIG. 19B, the computer system 100 generates a graph corresponding to an individual's score (e.g., risk probability, diagnostic probability, etc.) (Y axis) over time (X axis). Furthermore, the uncertainty (e.g., measurements, modeling, and/or aggregated measurements and modeling) could be used to further inform the user(s) about confidence on the risk group for the result(s). In some implementations, uncertainty may not be included.

Example user interface shown in the figures referenced and described earlier show data associated with coronary heart disease measurements. The underlying data used to generate such figures was measured by testing genetic and epigenetic markers for coronary heart disease (CHD). In some implementations, the underlying data can be obtained by testing or collating or interrogating or aggregating appropriate genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described above for other diseases or disorders such as stroke, heart failure, diabetes, etc. User interfaces can be generated based on such underlying data. In this manner, the online platform described here can be used to present data with not only cardiovascular disease but also with other diseases as well as associated comorbidities. Genetic and epigenetic markers associated with CHD and many other indicators can be found, for example, in WO 2017/214397, WO 2022/051630 and WO 2022/051641, which are incorporated herein by reference.

As used herein, genetic biomarkers generally refer to single nucleotide deletions polymorphisms (SNPs) but can refer to any type of genetic changes (e.g., insertions, substitutions, etc.). As used herein, epigenetic biomarkers generally refer to the

methylation of a cytosine nucleotide, which is usually adjacent to a guanine nucleotide (CpG). As used herein, other biomarkers refer to proteins, metabolic assays, lipids, family history information, images, biochemistry, other data described herein, etc.

5 *Computer Systems*

FIG. 20 is a block diagram of an example computer system used to provide computational functionalities associated with the algorithms, methods, functions, processes, flows, and procedures described in the present disclosure, according to some implementations of the present disclosure. The illustrated computer is intended to
10 encompass any computing device such as a server, a desktop computer, a laptop/notebook computer, a wireless data port, a smart phone, a personal data assistant (PDA), a tablet computing device, or one or more processors within these devices, including physical instances, virtual instances, or both. The computer can include input devices such as keypads, keyboards, and touch screens that can accept user information. Also, the
15 computer can include output devices that can convey information associated with the operation of the computer. The information can include digital data, visual data, audio information, or a combination of information. The information can be presented in a graphical user interface (UI) (or GUI).

The computer can serve in a role as a client, a network component, a server, a
20 database, a persistency, or components of a computer system for performing the subject matter described in the present disclosure. The illustrated computer is communicably coupled with a network. In some implementations, one or more components of the computer can be configured to operate within different environments, including cloud-computing-based environments, local environments, global environments, and
25 combinations of environments.

At a top level, the computer is an electronic computing device operable to receive, transmit, process, store, and manage data and information associated with the described subject matter. According to some implementations, the computer can also include, or be communicably coupled with, an application server, an email server, a web server, a
30 caching server, a streaming data server, or a combination of servers.

The computer can receive requests over a network from a client application (for example, executing on another computer). The computer can respond to the received requests by processing the received requests using software applications. Requests can

also be sent to the computer from internal users (for example, from a command console), external (or third) parties, automated applications, entities, individuals, systems, and computers.

Each of the components of the computer can communicate using a system bus. In some implementations, any or all of the components of the computer, including hardware or software components, can interface with each other or the interface (or a combination of both) over the system bus. Interfaces can use an application programming interface (API), a service layer, or a combination of the API and service layer. The API can include specifications for routines, data structures, and object classes. The API can be either computer-language independent or dependent. The API can refer to a complete interface, a single function, or a set of APIs.

The service layer can provide software services to the computer and other components (whether illustrated or not) that are communicably coupled to the computer. The functionality of the computer can be accessible for all service consumers using this service layer. Software services, such as those provided by the service layer, can provide reusable, defined functionalities through a defined interface. For example, the interface can be software written in JAVA, C++, or a language providing data in extensible markup language (XML) format. While illustrated as an integrated component of the computer, in alternative implementations, the API or the service layer can be stand-alone components in relation to other components of the computer and other components communicably coupled to the computer. Moreover, any or all parts of the API or the service layer can be implemented as child or sub-modules of another software module, enterprise application, or hardware module without departing from the scope of the present disclosure.

The computer can include an interface. Although illustrated as a single interface in FIG. 20, two or more interfaces can be used according to particular needs, desires, or particular implementations of the computer and the described functionality. The interface can be used by the computer for communicating with other systems that are connected to the network (whether illustrated or not) in a distributed environment. Generally, the interface can include, or be implemented using, logic encoded in software or hardware (or a combination of software and hardware) operable to communicate with the network. More specifically, the interface can include software supporting one or more communication protocols associated with communications. As such, the network or the interface's hardware can be operable to communicate physical signals within and outside

of the illustrated computer. Any of the user interfaces described herein and/or other interfaces based on genetic, epigenetic, other biomarker(s) (e.g., protein, metabolic assays etc.), measurement(s), test results and/or other data described herein can be displayed, tracked and shared via a computer by way of an application environment (e.g., mobile application environment, web application environment, etc.), coaching, and/or connected devices.

The computer can include a processor. Although illustrated as a single processor in FIG. 20, two or more processors can be used according to particular needs, desires, or particular implementations of the computer and the described functionality. Generally, the processor can execute instructions and can manipulate data to perform the operations of the computer, including operations using algorithms, methods, functions, processes, flows, and procedures as described in the present disclosure.

The computer also can include a database that can hold data for the computer and other components connected to the network (whether illustrated or not). For example, database can be an in-memory, conventional, or a database storing data consistent with the present disclosure. In some implementations, database can be a combination of two or more different database types (for example, hybrid in-memory and conventional databases) according to particular needs, desires, or particular implementations of the computer and the described functionality. Although illustrated as a single database in FIG. 20, two or more databases (of the same, different, or combination of types) can be used according to particular needs, desires, or particular implementations of the computer and the described functionality. While database is illustrated as an internal component of the computer, in alternative implementations, database can be external to the computer.

The computer also can include a memory that can hold data for the computer or a combination of components connected to the network (whether illustrated or not). Memory can store any data consistent with the present disclosure. In some implementations, memory can be a combination of two or more different types of memory (for example, a combination of semiconductor and magnetic storage) according to particular needs, desires, or particular implementations of the computer and the described functionality. Although illustrated as a single memory in FIG. 20, two or more memories (of the same, different, or combination of types) can be used according to particular needs, desires, or particular implementations of the computer and the described

functionality. While memory is illustrated as an internal component of the computer, in alternative implementations, memory can be external to the computer.

The application can be an algorithmic software engine providing functionality according to particular needs, desires, or particular implementations of the computer and
5 the described functionality. For example, application can serve as one or more components, modules, or applications. Further, although illustrated as a single application, the application can be implemented as multiple applications on the computer. In addition, although illustrated as internal to the computer, in alternative implementations, the application can be external to the computer.

10 The computer can also include a power supply. The power supply can include a rechargeable or non-rechargeable battery that can be configured to be either user- or non-user-replaceable. In some implementations, the power supply can include power-conversion and management circuits, including recharging, standby, and power management functionalities. In some implementations, the power supply can include a
15 power plug to allow the computer to be plugged into a wall socket or a power source to, for example, power the computer or recharge a rechargeable battery.

There can be any number of computers associated with, or external to, a computer system containing computer, with each computer communicating over network. Further, the terms “client,” “user,” and other appropriate terminology can be used interchangeably,
20 as appropriate, without departing from the scope of the present disclosure. Moreover, the present disclosure contemplates that many users can use one computer and one user can use multiple computers.

Models for processing input(s) described and returning output(s) described, in order to achieve the performance characteristics described, can include classification
25 model(s) structured to receive input data, and to return indications of subsets of features having high predictive power (e.g., sensitivity, specificity, cost, etc.).

Different types of models (e.g., linear, non-linear, classification etc) can be built with one or more markers for varying objectives (e.g., cost, diagnostics etc.). Multiple models can be built with one or more markers for varying objectives (e.g., cost,
30 diagnostics, etc.).

Model architecture can include statistical (e.g., linear regression, logistic regression, proportional hazard models, etc.), machine learning (e.g., random forest, support vector machines, neural networks, bayes classifiers, etc.), deep learning (e.g.,

convolutional neural networks, recurrent neural networks, autoencoders, large language model, etc.), and time series (e.g., ARIMA, etc.), Bayesian models (e.g., Bayesian Networks, etc.), financial (decision tree, discrete event simulation, budget impact, etc.).

Prior to fitting model(s), input data can be conditioned or otherwise pre-processed, such that conditioned data elements (e.g., genomic reads associated with loci of interest, functional data, sensor data, other lifestyle data, etc.) are suitable for further processing. Conditioning, as described herein, can include filtering of data (e.g., sensor data outputs that have confidence values below a threshold etc.). Pre-processing step(s) can be taken on the model input(s) such as dimensionality reduction (e.g., principal component analysis, linear discriminant analysis, auto encoders, uniform manifold approximation and projection, partial least squares regression, etc.) before training model(s).

Transforming output data can be used to enhance the expandability of the models described previously. For example, SHapley Additive exPlanations (SHAP), Local Interpretable Model-agnostic Explanations, Integrated Gradients, Partial Dependence Plots, Global Surrogate Models, etc., can be used to enhance expandability of the output for users. One or more of these approaches can be utilized concurrently. In specific examples, SHAP can be utilized to identify the most significant contributing marker(s) to conditions or indication.

Transforming output data into digital objects, such as visualizations, in order to facilitate generation of characterizations, interventions for condition prevention, interventions for condition treatment, and/or other actions (e.g., computer-generated and machine-implementable instructions) for improving and/or maintaining metrics (diagnostics, risk score, cost etc.). In specific examples, digital objects and visualizations can be generated upon transforming input data, using various software (e.g., Rstats, ggplotz, matplotlib, etc.) in sequence and/or in parallel.

The model prediction(s) (e.g., risk score, diagnostics, etc.) uncertainty can be computed. The methods for estimating may include bootstrap methods, Bayesian methods, Monte Carlo dropout, ensemble methods, sensitivity analysis etc. The uncertainty of the method and/or model described herein can be aggregated to provide overall system uncertainty.

In some variations, methods described herein can further include refining the model(s): collecting a set of training data streams derived from a population of patients(s), the set of training data streams capturing data (e.g., genetic, epigenetic,

lifestyle, etc.) paired with intervention(s), treatment(s), management(s) information, from the population of patients(s), creating a training dataset derived from the set of training data streams and the set of transformation operations, and training the model(s) in one or more stages, based upon the training dataset.

5 Data/signal inputs and/or other inputs (e.g., contextual inputs, derivative inputs, combinatorial inputs, etc.) can be used for training the model(s). In more detail, markers may be transformed either individually or in combination before being processed by the model(s). Combinatorial markers can include genetics, epigenetics, metabolic assays, lipids, protein, lifestyle, demographic, and/or other data described herein.

10 Additionally or alternatively, dynamic aspects (e.g., changes over time in markers, changes in frequency between instances of respective features, other temporal aspects, other frequency-related aspects, etc.) of features derived from the samples can be used to predict or otherwise anticipate health condition statuses for generation of personalized intervention plan components.

15 Inputs can be aggregated from populations of patients associated with different demographic characteristics, health statuses, health conditions, lifestyles, and/or other suitable factors.

 In relation to model architecture, inputs to model(s) described herein can produce outputs that are subsequently used as inputs to an overarching model (e.g., classification
20 model having multiple layers, reinforcement learning models, etc.) that returns diagnostics, characterizations of the patient(s) (e.g., in relation to health state, disease state, etc.), personalized intervention plan aspects, and/or other aspects based upon processing data in stages. However, the model(s) can implement other suitable architecture having other suitable flow for processing data derived from the inputs.

25 Returned classification regression and/or other outputs of model(s) can include returned confidence-associated parameters in such classifications. In particular, confidence-associated parameters can have a score (e.g., percentile, other score) that indicates confidence in the returned output. The confidence may be estimated by aggregating measurement and or modeling uncertainties.

30 Furthermore, refined versions of the model(s) can be configured to process fewer inputs (e.g., only a subset of inputs described herein) in order to return accurate outputs for generating personalized intervention plan components. Furthermore, previous data

derived from inputs (e.g., new signals/signatures, interesting signals/signatures, etc.) can be returned by computing components during model refinement.

While embodiments, variations, and examples of model(s) (e.g., in relation to inputs, outputs, and training) are described herein, models can additionally or
5 alternatively include other machine learning architecture.

Statistical analyses and/or machine learning algorithm(s) can be characterized by a learning style including any one or more of: supervised learning (e.g., using back propagation neural networks), unsupervised learning (e.g., K-means clustering), semi-supervised learning, reinforcement learning (e.g., using a Q-learning algorithm, using
10 temporal difference learning, etc.), and any other suitable learning style.

Furthermore, any algorithm(s) can implement any one or more of: a regression algorithm, an instance-based method (e.g., k-nearest neighbor, learning vector quantization, self-organizing map, etc.), a regularization method, a decision tree learning method (e.g., classification and regression tree, chi-squared approach, random forest
15 approach, multivariate adaptive approach, gradient boosting machine approach, etc.), a Bayesian method (e.g., naïve Bayes, Bayesian belief network, etc.), a kernel method (e.g., a support vector machine, a linear discriminant analysis, etc.), a clustering method (e.g., k-means clustering), an associated rule learning algorithm (e.g., an a priori algorithm), an artificial neural network model (e.g., a back-propagation method, a Hopfield network
20 method, a learning vector quantization method, etc.), a deep learning algorithm (e.g., a Boltzmann machine, a convolution network method, a stacked auto-encoder method, etc.), a dimensionality reduction method (e.g., principal component analysis, partial least squares regression, etc.), an ensemble method (e.g., boosting, boot strapped aggregation, gradient boosting machine approach, etc.), and any suitable form of algorithm.

25 In an example, model training can be based on a cohort of patients who are taking drug(s) (e.g., statin) and/or lose >5% body weight while receiving personalized digital care for weight loss and present concomitant reduction of cardiovascular symptomatology and/or risk. A diagnostic signature can be built based on predictive variables identified in model(s) on the likelihood of a patient having a cardiovascular event. Example variables
30 include demographics, genomic (e.g., SNPs and/or methylation associated with inflammation, associated with obesity, associated with other factors described, etc.), and any other type of variable.

The intervention(s), treatment(s), management(s), and/or monitoring can also include modified variations of operations for training and refinement of other model architecture configured for improving other conditions.

A system / platform for intervention(s), diagnostics, treatment(s), management(s), and/or monitoring, includes: a computing platform comprising one or more processing subsystems comprising non-transitory computer-readable medium comprising instructions stored thereon, that, when executed by the processing subsystems, perform one or more steps of methods described herein; and one or more execution subsystems configured to execute components of personalized intervention plans informed by processes of the computing platform. In variations, the execution subsystems can be configured to execute control instructions generated by the computing platform, where control instructions can involve instructions for controlling operation modes of one or more of: application interfaces (e.g., mobile application interfaces, web application interfaces, etc.) for signal reception, data aggregation, and/or retrieval of other inputs to be processed by system architecture: sample processing architecture (e.g., by automated / robotic subsystems); communication interfaces for performing telehealth operations, providing group therapies, providing counseling (e.g., through human entities, through digital entities); interfaces for providing rewards and/or other incentives to patients; interfaces for providing and monitoring tasks provided to patients; interfaces for connecting devices (e.g., biometric monitoring devices) of the patient with an account of the patient within the system / platform; interfaces for providing medications to the patient; interfaces for providing dietary advice / tracking diet behavior of the patient; and/or other suitable functionality for delivering components of a personalized intervention plan.

Embodiments of the system are configured to perform one or more portions of methods described herein; however, variations of the system can be configured to perform other suitable methods.

The computing platform can include one or more processing subsystems comprising non-transitory computer-readable medium that contain instructions stored thereon, that when executed by the processing subsystems perform one or more steps of methods described herein; and one or more execution subsystems configured to execute components of personalized intervention(s), treatment(s), management(s), and/or monitoring plans informed by processes of the computing platform.

The execution subsystems can be configured to execute control instructions generated by a computing platform, where control instructions can involve instructions for controlling operation modes of one or more of: application interfaces (e.g., mobile application interfaces, web application interfaces, etc.) for signal reception, data
5 aggregation, and/or retrieval of other inputs to be processed by system architecture; sample processing architecture (e.g., by automated / robotic subsystems), communication interfaces for performing telehealth operations, for providing group therapies, for providing counseling (e.g., through human entities, through digital entities); interfaces for providing rewards and/or other incentives to patients; interfaces for providing and
10 monitoring tasks provided to patients; interfaces for connecting devices (e.g., biometric monitoring devices) of the patients with an account of the patient within the system / platform; interfaces for providing medications to the patient; interfaces for providing recommendations / tracking lifestyle / behavior or other measurement(s) or biomarker(s) of the patient; and/or other suitable functionality for delivering components of a
15 personalized intervention, treatment, management and monitoring plan.

In particular, the execution subsystems can be structured to implement a next-generation, prescription-grade, digital therapeutics or other program that uses artificial intelligence (AI) to analyze genetic, epigenetic, and/or other biomarker(s) (e.g., protein, metabolic assays, etc.), measurement(s), test results and/or other data described herein to
20 create evidence-based personalized care, intervention, treatment, management, monitoring program to improve targeted outcomes (e.g., risk reduction, cost reduction, etc.).

Uses and Benefits

25 The compositions and methods described herein provide systems and methods for generating, collating, aggregating, evaluating, analyzing, displaying, tracking and sharing standalone or integrated (genomic, epigenomic, proteomic, RNA, imaging, electrocardiogram, lifestyle factors, clinical markers, social determinants of health etc.) diagnostics, risk assessment, therapeutic pathways, cost estimation, prognosis, health
30 outcomes for care (e.g. with respect to prevention, diagnosis, treatment and/or management) or to understand, manage and mitigate risks (e.g. with respect to implementing cost reduction initiatives) for various conditions or indications. In examples, characterization can be used to provide actionable insights, with intervention(s)

including lifestyle changes and/or therapeutics (e.g., drug therapy, medical devices, nutritional therapy, stem-cell therapies) and/or interventions (e.g., gene-editing, base editing, epigenetic silencing, epigenetic editing). Additionally or alternatively, the data, analysis and interpretation of data from patients may also uncover more complex
5 diagnostic pathways and pinpoint potential treatments and/or intervention options. Additionally or alternatively, the systems and methods can be used to continuously monitor and assess the effectiveness of such treatments and/or interventions, providing data and actionable insights for further optimization of the condition, indication, health outcomes or cost.

10 In some embodiments, the compositions and methods described herein can be used for risk assessment, prevention, prognosis, management, intervention, treatment and/or monitoring of one or more diseases, disorders, or risks. In some embodiments, the compositions and methods described herein can be used for risk assessment, prevention, prognosis, management, intervention, treatment and/or monitoring of indication at more
15 than one time point.

The compositions and methods described herein can be used to recommend lifestyle change(s) and/or therapeutic(s) and/or intervention(s) and/or management strategy(ies) to optimize targeted outcomes (e.g., reduce cost, reduce likelihood of a heart attack). The compositions and methods described herein also can be used to monitor or
20 optimize the effectiveness of lifestyle change(s) and/or therapeutic(s) and/or intervention(s) and/or management strategy(ies) over time to optimize targeted outcomes (e.g., reduce cost, reduce likelihood of a heart attack).

The compositions and methods described herein also can estimate the benefit of new interventions such as a new pharmaceutical and/or other compositions including, but
25 not limited to, chemical compositions (e.g., small molecule modulators), biological compositions (e.g., pre-/pro-/syn-/post-biotics), gene editing, base editing, epigenetic silencing and epigenetic editing. Additionally, the compositions and methods described herein can be used to apply outputs of the analyses to effect one or more actions (e.g., recommend new health initiative) to achieve the intended outcome (e.g., tackle obesity
30 among employees).

The compositions and methods described herein encompass a comprehensive system designed to manage marker data (e.g., genomic, epigenomic, and/or other biomarkers) and other data or measurements derived from the processed sample(s). This

system also includes mechanisms for deriving insights by analyzing these biomarkers using models (e.g., linear regression, machine learning) and for facilitating personalized care / intervention / management / therapeutic strategies. As discussed herein, these processes can be integrated and data and insights delivered to patients or other users in a
5 customized manner, by way of a computer system 100 and a display device.

The compositions and methods described herein can be used to query, collate, aggregate, evaluate, analyze information from one or more of: financial (e.g. cost of procedures, cost of medication, claims, etc.), industry/prevalence (e.g., heart disease prevalence in a particular geographic region, number of heart attacks a year among truck
10 drivers, etc.), publications (e.g., journal articles, clinical guidelines, etc.), laboratory tests (e.g., epigenetic assessments, inflammation biomarker, etc.), imaging (e.g., FFR from CCTA, etc.), medical (e.g., ICD10 codes, electronic medical records, medication usage, etc.), clinical (e.g., blood pressure, weight, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), lifestyle (e.g., smoking, exercise frequency, etc.),
15 supplement use (e.g., vitamin D, vitamin B12, iron, etc.), data to display, track, derive insights for users in a customized manner.

The method can be used to generate, collate, aggregate, evaluate, analyze, display, track and share standalone or integrated (genomic, epigenomic, proteomic, RNA, imaging, electrocardiogram, lifestyle factors, clinical markers, social determinants of
20 health, etc.) diagnostics, risk assessment, therapeutic pathways, cost estimation, prognosis, health outcomes for care (e.g. with respect to prevention, diagnosis, treatment and/or management) or to understand, manage and mitigate risks (e.g. with respect to implementing cost reduction initiatives) for various conditions or indications. Consequently, these methods have the potential to enhance outcomes concerning one or
25 more health conditions or indications, offering a refined approach to precision medicine, personalized risk assessment, prevention, management, treatment, and monitoring.

The measurements received or generated can be data or information (e.g., a dataset) including, without limitation, clinical diagnoses; demographics (e.g., gender, race); lifestyle; costs; claims (e.g., health insurance); imaging (e.g., cardiac CT scan,
30 cardiac MRI, coronary angiogram); features derived from imaging (e.g., FFR from CT scan, percent stenosis); results from electrocardiogram or echocardiogram test; results from stress tests; blood tests (e.g., for metabolic assays, genetic, epigenetic, protein, etc.); blood pressure; results from a carotid ultrasound; etc. The data or information received or

generated also can include publications (e.g., journal articles) and, for example, clinical guidelines.

One or more dataset(s) can be received from a patient or those associated with a patient (e.g., clinician), which further functions to enable generation of a baseline state or a subsequent state and one or more measurements from which models for returning
5 diagnostics, risk assessment, personalized therapeutic pathways, cost estimation, prognosis, health outcomes, risk(s), other insights of interest, recommendations can be generated in subsequent portions of the method.

A dataset can include data derived from one or more of: body weight (e.g.,
10 receiving bodyweight values of the patients generated from a digital weighing scale), body fat percent, muscle mass, body water, height or other length measurements (e.g., via a ruler or measuring tape), other body mass index (BMI)- associated parameters, blood chemical and biochemical information, inflammatory markers, fasting blood sugar, high density lipids, low density lipids, blood interleukins, c-reactive protein, blood cell counts,
15 electrophysiology signals (e.g., electroencephalogram signals, electromyography signals, galvanic skin response signals, electrocardiogram signals, etc.), heart rate, body temperature, cardiovascular parameters, continuous glucose monitoring (glycemic response), respiration parameters (e.g., respiration rate, depth/shallowness of breath, etc.), blood oxygenation signals, motion parameters, and any other suitable physiologically
20 relevant parameter of the patient. Additionally and alternatively, a dataset can include data derived from one or more of: electronic health records, health plan claims, questionnaires, survey, wearables, public sources (e.g., repositories) and any other data relevant directly or indirectly to the patient or user. Additionally and alternatively, a dataset can include data that is raw, imputed, transformed, longitudinal, cross-sectional or
25 temporal.

A dataset can capture behavioral information for the patient pertaining to one or more of: energy levels (e.g., morning energy level, evening energy level, daytime energy level, etc.), dietary behavior, sleep behavior, stress levels, stress-associated events, cravings, exercise behavior, meditation behavior, perceived progress toward a health-
30 associated goal, actual progress toward a health-associated goal, state of symptoms, social determinants of health, familial determinants of health, and work determinants of health, and/or other behavioral information. In examples, the lifestyle dataset can capture one or more of the following lifestyle characteristics of the patient: energy levels, food intake

(e.g., through food photos which are then assessed and scored by a coaching entity or other entity, through dietary journal entries, through application programming interfaces (APIs) of diet-monitoring applications, etc.), sleep behavior, stress levels, cravings, exercise behavior, and weight loss progress. However, variations of the example can
5 alternatively capture other types of lifestyle information from the patient.

A dataset can include data derived from one or more of: blood chemical and biochemical profiles, saliva chemical and biochemical profiles, medication use, supplement use, fasting blood sugar, glycemic response, high density lipid values, low density lipid values, and any other similar parameters. A dataset can capture one or more
10 of: a morning energy level, dietary behavior, sleep behavior, stress levels, cravings, exercise behavior, meditation behavior, state of symptoms of the patient, medication use, social determinants of health, familial determinants of health, and work determinants of health.

A dataset can capture contextual and supplemental information at the level of one
15 or more of: population, location or geography, employer, health plan, government entity, non-profit organization, healthcare organization and any other similar parameters. Additionally or alternatively, a dataset can include or otherwise detect cyclic biometrics or biometrics that occur with some pattern (e.g., derived from user inputs, biometric monitoring devices worn by the user, or determined by an algorithm that tracks the user's
20 physical, emotional, or neurological states). A dataset can include data sampled from devices once (e.g., at a single time point, upon intake of a patient), or at a number of time points (e.g., at random points, at regular points, in relation to triggering events, with other frequency, etc.).

Other supplemental or contextual data relevant to generating actionable insights
25 can be received. For instance, the method can include capture of current therapeutic approaches the patient participates in (e.g., existing medications, existing supplements, etc.), trends in usage or adherence to current therapeutic approaches the patient participates in (e.g., increasing use, decreasing use, steady use, etc.), medical history, family medical history, and/or other information. Additionally or alternatively, additional
30 data can include information related to an environment of the user, such as a location of the user (e.g., as determined from a global positioning device, from a triangulation device), environmental temperature of the user, environmental audio of the user, and any other suitable environmental information of the user pertaining to potential stimuli

affecting health, and/or environmental devices that can be used for outputs (e.g. diagnostic, therapeutic, etc.) associated with subsequent blocks of the method. Additionally or alternatively, additional data can include information from assessments or procedures undergone by the user, such as imaging (e.g., information from angiogram or coronary calcium scan or CCTA, etc.), interventions (e.g., gene-editing, epigenetic silencing, etc.) and any other similar technologies or methodologies.

The compositions and methods described herein can be adapted for distinct populations (e.g. Americans in the Midwest, Native Americans, etc), varying demographics (e.g. age, gender, etc.), different states of health (e.g. diabetes, known risk factors, etc.), different lifestyles (e.g. smoking, diet type, etc.), different users (e.g., clinician, employer, insurance company, etc.), different interventions (e.g. drug A vs. drug B) and/or other factors (e.g., social determinants of health, family history, etc.). These adaptations are aimed at enhancing health outcomes, improving care, and/or understanding, managing, and mitigating risks.

Methods of Personalized Intervention

Once measurement(s) are received about a patient, the compositions and methods described herein can include: (A) performing uncertainty quantification of markers; (B) performing quality control of samples and/or markers; (C) performing risk assessment and/or diagnostics; (D) performing uncertainty quantification to evaluate confidence of the assessment; (E) determining the contribution of one or more biomarkers(s); (F) identifying casual or association of biomarkers to disease and treatment pathways; (G) generating personalized intervention(s), treatment(s), management and/or monitoring for the patient or user; (H) predicting estimated outcome(s) and/or costs associated with personalized intervention(s), treatment(s), management and/or monitoring; (I) and/or executing the personalized intervention(s), treatment(s), management and/or monitoring for the patient. See Table 5.

Table 5

A	Perform uncertainty quantification of markers(s)
B	Perform quality control of sample(s) and/or markers(s)
C	Assess baseline state (e.g., risk and/or diagnostic score)
D	Perform uncertainty quantification to evaluate confidence of the assessment(s)
E	Identify contribution of markers(s) for the assessment(s)
F	Identify casual or association of the markers to disease and treatment pathways
G	Generate personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient or user
H	Predict estimated outcome(s) and/or cost associated with the personalized intervention(s), treatment(s), management and/or monitoring
I	Execute the personalized intervention(s), treatment(s), management and/or monitoring

The order of operation may change, and some steps are not necessary for the system to operate. This process can be repeated a plurality of times to monitor and assess effectiveness of interventions, treatment(s), management and/or monitoring and update the intervention, treatment(s), management and/or monitoring recommendations. The following descriptions related to Table 5 are exemplary only and are not intended to be limiting.

Row (A) is related to uncertainty quantification, which can encompass all aspects of marker measurements. For instance, in epigenetic measurement, uncertainty quantification may include, but is not limited to, sampling error, reagent quality, instrumentation, and human error.

Row (B) is related to the uncertainty quantification mentioned herein, which can be used to perform quality control. For example, known reference sample(s) and/or measurement(s) can be compared against the new measurement(s). The difference between the known and new measurement(s), along with their uncertainty, can be used to evaluate error(s) and/or uncertainty(ies) and how they compare to a defined acceptable threshold. This process assists in identifying sources of errors and/or uncertainties and minimizes such error(s) and/or uncertainty(ies) to ensure the reliability of the measurement(s). For example, it can be used to determine if a sample(s) requires re-measurement(s) or re-collection(s) to meet a defined acceptable threshold for the measurement(s) and/or marker(s).

Row (C) is related to returning a baseline state(s) or subsequent state(s) and a set of signatures (e.g., diagnostic and/or therapeutic signatures associated with genomic and

demographic features of the patient(s)) upon processing the data (e.g., imaging, lifestyle, etc.) and/or measurements (e.g., DNA methylation, genetic biomarkers, etc.). Row (C) includes transforming data into various signatures that can be used to characterize the patient(s) and to generate a personalized intervention, treatment, management and/or
5 monitoring plan for the patient(s) based upon the characterization of the patient(s).

Transformation operation(s) can include one or more of: graph-based methods, linear and nonlinear dimensionality reduction, applications of supervised, semi-supervised and unsupervised machine or statistical inference methods to derive informative features from measurement(s) and/or data, imputing missing sites,
10 determining genetic ancestry of the patient, estimating genetic parameters comprising genetic diversity and homozygosity, estimating scores representing inherited risk, estimating scores representing acquired risk and estimated values of qualitative, quantitative, continuous and categorical traits which are normalized to biological gender, genetic ancestry, age, socioeconomic status, measurements, lifestyle variables and to any
15 other suitable parameter(s). In some instances, traits can be normalized to biological gender, genetic ancestry, age, socioeconomic status, measurements, lifestyle variables and to any other suitable parameter(s).

With respect to measurement(s) and/or monitoring devices described herein, Row (C) also can include signal processing operations including one or more of: denoising,
20 filtering, smoothing, clipping, transformation of discrete data points to continuous functions, and performing any other suitable signal conditioning process. For instance, some variations of Row (C) can additionally include performing a windowing operation and/or performing a signal cleaning operation. Signal cleaning can include removal of signal anomalies by one or more filtering techniques. In specific examples, filtering can
25 include one or more of: Kalman filtering techniques, bootstrap filtering techniques, particle filtering techniques, Markov Chain Monte Carlo filtering techniques; and/or another suitable technique. Signal cleaning can thus improve data quality for further processing, in relation to one or more of: noise, sensor equilibration, sensor drift, environmental effects (e.g., moisture, physical disturbance, etc.), and any other suitable
30 type of signal artifact.

The baseline state functions to establish a reference state against which progress is compared, as the patient participates in the personalized intervention, treatment, management and/or monitoring plan generated. The baseline state is preferably associated

with a state of the patient (e.g., epigenetic baseline state, coronary heart disease baseline state, etc.) prior to participation in the personalized intervention, treatment, management and/or monitoring plan, such that the baseline state characterizes a state of the patient prior to treatment or intervention or care according to the personalized intervention, treatment, management and/or monitoring plan. However, the baseline state can alternatively characterize another suitable state of the patient. Furthermore, the methods described herein can include re-establishment of a “baseline” state of the patient in coordination with provision of the personalized intervention, treatment, management and/or monitoring plan. As such, the baseline state can be updated as the patient progresses during participation in the personalized intervention, treatment, management and/or monitoring plan, at which point the data, prognostic, outcome, cost, risk, diagnostic and/or therapeutic signatures may lead the patient into a different personalized intervention, treatment, management and/or monitoring plan.

The baseline state can also characterize a physiological state of the patient, with respect to a condition or indication (e.g., health condition, disease state, etc.). As such, the baseline state can characterize a clinical diagnosis, a laboratory diagnosis, a radiological diagnosis, a tissue diagnosis, a principal diagnosis, an admitting diagnosis, a differential diagnosis, a prenatal diagnosis, a diagnosis of exclusion, and/or other diagnostic criteria. Additionally or alternatively, the baseline state can characterize aspects associated with a condition or indication (e.g., high cholesterol, etc.). In some embodiments, the baseline state can characterize risk assessment, therapeutic pathways, cost estimation, prognosis, health outcomes or any other suitable parameter(s).

In some embodiments, the diagnostic signatures generated in Row (C) can include one or more of: measurement(s) (e.g., genetic signatures, epigenetic signatures, epigenetic signatures associated with coronary heart disease, HbA1c, etc.), additional data (e.g., lifestyle, demographics, imaging, etc.) indicative of decreased or increased likelihood of developing a health condition or indication. In some embodiments, the signatures based on one or more of the factors and data outlined herein can be for risk assessment, therapeutic pathways, cost estimation, prognosis, health outcomes or any other suitable parameter(s).

In some embodiments, the diagnostic signatures generated in Row (C) can include one or more of: measurement(s) (e.g., genetic signatures, epigenetic signatures, epigenetic signatures associated with coronary heart disease, HbA1c, etc.), additional data (e.g.,

lifestyle, demographics, imaging, etc.) indicative of decreased/increased likelihood of improvement or remission of a health condition or indication. The signatures can be used to develop lifestyle change recommendations and/or therapeutics (e.g., drug therapy, medical devices, nutritional therapy, stem-cell therapies) and/or interventions (e.g., gene-
5 editing, base editing, epigenetic silencing, epigenetic editing) and/or care and/or understand, manage and/or mitigate risk(s). Additionally or alternatively, the intervention(s) can be used to generate the personalized intervention, treatment, management and/or monitoring plan, with respect to personalized and combinatorial therapeutic pathways, care, interventions and risk management implementing different
10 approaches.

Biomarker signatures and baseline state(s) or subsequent state(s), as well as diagnostics, risk assessment, personalized therapeutics, cost estimation, prognosis, health outcomes developed using the compositions and methods described herein, can target one or more health conditions or indications.

15 Row (G) indicates that a personalized intervention, treatment, management and/or monitoring plan can be included for the patient upon processing the baseline state(s) or subsequent state(s), the set of diagnostic signatures, and the set of therapeutic and intervention signatures with a model, which functions to transform signatures into a customized combination of therapeutic(s) and/or intervention(s) and/or management
20 and/or care approaches tailored to improve or maintain and/or improve a health condition or indication of the patient. Signatures can be that of risk assessment, therapeutic pathways, cost estimation, prognosis, health outcomes or any other suitable parameter(s) instead of diagnostic signatures.

One or more marker(s) and/or one or more model(s) could be used to provide
25 personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient(s) and/or user(s). One or more marker(s) and/or one or more model(s) could be used as a basis for designing and/or developing and/or optimizing personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient(s). For example, one or more marker(s) and/or one or more model(s) could be used for drug
30 discovery, drug development, understanding drug effectiveness in research or clinical or clinical trial settings, understand or predict side effect profile, define eligibility criteria for research or clinical trials.

One or more marker(s) can be used to measure impact (e.g., reduction in cost, improvement of health condition, etc.) for personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient(s) and/or user(s). One or more marker(s) impact(s) can be quantified for personalized intervention(s), treatment(s),
5 management(s), and/or monitoring for the patient(s) and/or user(s). The impact of one or more marker(s) can be re-evaluated over time by continuously monitoring (measuring and collecting data) for personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient(s) and/or user(s). One or more marker(s)' re-evaluated impact(s) can be used to optimize personalized intervention(s), treatment(s),
10 management(s), and/or monitoring for the patient(s) and/or user(s). For example, one or more marker(s)' re-evaluated could be used to optimize drug compound and/or dosage etc. One or more marker(s)' re-evaluated impact(s) can be used to optimize personalized intervention(s), treatment(s), management(s), and/or monitoring for the patient(s) and/or user(s). The re-evaluation of one or more marker(s) and/or one or more model(s) can be
15 done at regular intervals or random time interval(s).

The model is preferably constructed to process signatures and/or baseline state(s) and/or subsequent state(s) generated from Row (C), and to return the personalized intervention, treatment, management and/or monitoring plan that is tailored to the patient(s) and/or user(s). A model can return a set of measurement(s) (e.g., SNP,
20 methylation markers, etc.) and insight(s) (e.g., percent employees at high risk for a heart attack, etc.) for the patient and/or user, and generate their personalized intervention, treatment, management and/or monitoring plan from measurement(s), data and/or insight(s).

The personalized intervention, treatment, management and/or monitoring plan can
25 have sub-portions (e.g., modules, phases), which can include portions including one or more of: personalized medication regimens, personalized supplement regimens, personalized lifestyle recommendations, medical procedures, testing recommendations, preventative healthcare therapeutic approaches, and/or other suitable aspects. Furthermore, the personalized intervention, treatment, management and/or monitoring
30 plan can be characterized with a duration, such that the patient can complete the program and achieve one or more health goals. As such, generating the personalized intervention, treatment, management and/or monitoring plan can include returning recommendations (e.g., drug recommendations, gene editing, epigenetic silencing, target BMI etc.) and/or

care components, coaching components, risk management components delivered in person and/or digitally, based upon measurement(s) and/or data and/or insight(s) of the patient(s). However, the personalized intervention, treatment, management and/or monitoring plan can be otherwise configured.

5 Row (G) can include generation of instructions, that can be executed by systems having a computing element. For instance, one or more components of the personalized intervention, treatment, management and/or monitoring plan can be delivered digitally through a mobile device application, by way of an application-interface between care or coaching entities and the patient(s) being treated.

10 In one example, the personalized intervention, treatment, management and/or monitoring plan can have a set duration (e.g., 36 weeks, 24 weeks, 12 weeks, another suitable number of weeks, etc.), and can be configured for delivery using one or more interfaces (e.g., web interface, mobile device interface, wearable computing device interface, telephonic interface, in-person interface, interface with one or more robotic
15 devices, etc.) with the patient(s).

 The example personalized intervention, treatment, management and/or monitoring plan can have one or more phases configured to promote change in the patient(s)' markers or aggregate of markers, provide therapeutic interventions, and achieve desired outcomes with respect to diagnoses and characterizations generated (e.g., based on diagnostic
20 signatures processed by the model, based on therapeutic signatures processed by the model, etc.). In some variations, desired outcomes could be with respect to risk assessment, therapeutic pathways, cost estimation, prognosis, health outcomes or any other suitable parameter(s).

 In one example, the first phase has a duration (e.g., 4 weeks, another suitable
25 number of weeks) configured to guide the patient(s) in focusing on baselining health condition, indication, insights and/or habits, with functionality for tracking lifestyle through a mobile device application. In the example, the second phase has a duration (e.g., 12 weeks, another suitable number of weeks) focused on personalizing therapeutic, care, management approaches for producing transformations in behavior for the
30 patient(s). In the example, the third phase has a duration (e.g., 8 weeks, another suitable number of weeks) focused on personalizing therapeutic, care, management approaches for stabilizing the patient's life, with respect to maintenance of desired states (e.g., building and maintaining healthy habits, preventing relapse, maintaining remission, etc.).

With respect to the personalized intervention, treatment, management and/or monitoring plan, the platform can provide a kit (e.g., genetic and epigenetic sampling kit, connected devices, biometric devices, instructions, etc.), and interaction with the kit by the patient(s) facilitates establishment of a physiological baseline that can serve as a reference point for progress, with respect to the personalized intervention, treatment, management and/or monitoring plan. In the example, the kit can be mailed (e.g., as facilitated by the platform), to the patient's home or location. The kit also provides instructions for downloading an application with an interface to the platform, as well as functionality for receiving inputs associated with the lifestyle dataset described herein. In one specific example, the patient is prompted to download the mobile application, complete a lifestyle intake form, and schedule a counseling session as part of a personalized therapeutic, care, management approach.

Subsequently, with respect to the personalized intervention, treatment, management and/or monitoring plan, the platform can guide and support the patient using counseling entities (e.g., specialized counselors, clinicians, artificial intelligence counseling entities, etc.) across a period (e.g., weeks, months) with regular sessions (e.g., weekly and bi-weekly sessions) that can be scheduled and/or ad hoc. A counseling session of the specific example can be a 15-20 telephonic behavioral counseling session, but variations of the example can have another duration and/or be provided in another format. The personalized intervention, treatment, management and/or monitoring plan provides personalized therapeutic and intervention pathways configured to reconfigure the condition or indication of the patient to achieve a goal (e.g., 5-10% reduction of risk score). With respect to the personalized intervention, treatment, management and/or monitoring plan generated in Row (G), personalized and actionable insights are returned by the model and delivered to the patient(s) (e.g., through entities, through application interfaces, through other interfaces with the platform, etc.) as they complete phases of the personalized intervention, treatment, management and/or monitoring plan (e.g., complete tasks, interact with modules, interact with tools for monitoring diet (e.g., through upload of photo documentation of their meals/ diet/consumption), monitor their lifestyle vitals (e.g. smoking cessation, exercise, weight loss, etc., progress through the mobile application and wireless scale, etc.). The patient can engage with the personalized intervention, treatment, management and/or monitoring plan at any time or place via their mobile device application and/or web application, which allows for flexible participation.

Additionally or alternatively, the personalized intervention, treatment, management and/or monitoring plan can provide time-sensitive tasks and/or prompt interaction in a timely manner, with respect to more acute states of the patient (e.g., triggering events such as heart attack, etc.).

5 The application environment (e.g., mobile application environment, web application environment, etc.) can support one or more of the following: a dietary consumption log (e.g., food log, drink log, etc.) that can receive inputs from the patient and/or automatically track consumption by the patient (e.g., in coordination with applications supported by Apple Health™, Google Health™, etc.); drug usage;
10 personalized meal and fitness plans or any other parameters known or generated from the personalized intervention, treatment, management and/or monitoring plan. Additionally or alternatively, the application environment can include interfaces for connections (e.g., using Bluetooth™, using another protocol) with smart devices (e.g., as described herein, for automatic weight, cardiovascular parameter tracking, blood analyte parameter
15 tracking, motion tracking, etc.).

 The application environment can further support telehealth interactions between the patient and a counseling/healthcare-providing clinician. For instance, the application environment can provide one or more of: communication interfaces that connect the patient with a physician and/or facilitate delivery of care to the patient(s) (e.g., through
20 automated processing of insurance claims, through generation of appointments, through enabling consultations with a clinician, etc.); communication interfaces that provide constant or near constant access (e.g., 24 hour, 7 days a week access, etc.) to trained coaches, nutritionists, counselors, etc.; communication interfaces that enable telehealth group coaching; exercise regimen components (e.g., group fitness content, exercise
25 guidance content, such as yoga content, etc.); stress management material (e.g., provided to the patient in response to detected stress states and/or triggering events, provided to the patient such that the patient can access the content in a convenient manner, etc.);
interfaces to a private (e.g., invite-only) social network or community; tasks (e.g., healthy habits challenges); interfaces for reward provision (e.g., community celebration events,
30 incentives, other perks, etc.); and other suitable interfaces. The application environment can, however, support other suitable functionality associated with the personalized intervention, treatment, management and/or monitoring plan.

Row (I) refers to executing the personalized intervention, treatment, management and/or monitoring plan for the patient, where executing the personalized intervention, treatment, management and/or monitoring can involve executing components of the intervention, treatment, management and/or monitoring through interfaces described
5 herein and/or in relation to the system described below. As such, execution can involve mobile device application interfaces, web application interfaces, interfaces with an entity (e.g., human care-providing entity, digital care-providing entity, etc.), and/or other suitable interfaces.

Execution of the personalized intervention, treatment, management and/or
10 monitoring plan can be based on individual, multiple or aggregate of one or more of: financial (e.g., cost of care, cost of medication, etc.), prevalence (e.g., percent risk for a heart attack, etc.), laboratory tests (e.g., DNA methylation level, change in inflammation level, etc.), imaging (e.g., FFR from CCTA, etc.), medical (e.g., risk score, data from electronic medical records, etc.), clinical (e.g., blood pressure level, presence of
15 symptoms such as chest pain, etc.), co-morbidities (e.g., obesity, diabetes, hypertension, hypercholesterolemia, etc.) therapeutic (e.g., type of statins available, etc.), supplement (e.g., vitamin D, vitamin B12, iron, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), or other suitable data.

Execution of the personalized intervention, treatment, management and/or
20 monitoring plan produces outcomes for patients participating in their respective personalized intervention, treatment, management and/or monitoring plans and/or other users (e.g., employer, payor, etc.), examples of which include: improved engagement (e.g., enrollment of 93% of participants; consistent engagement by a significant percentage of participants after 60 days); improved outcomes (e.g., significant improved
25 weight loss, significant reductions in A1C levels associated with diabetes, significant reductions in cardiovascular symptoms, changes in DNA methylation biomarker(s) associated with cardiovascular disease risk, status or co-morbidities, reductions in mortality rate etc.); reduction in medication use/necessity; reduction in healthcare costs; reduction in events (e.g., reduction in the number of emergency room visits, reduction in
30 the number of heart attacks, etc.) and other suitable benefits.

Additionally or alternatively, improved outcomes can be measured at an individual level or at an aggregate number of patients within a cohort or relative to prior level, or in comparison to another cohort and can include: percent weight loss, weight loss

over a period of time, average number of months maintaining weight loss after completing the program, average reduction in HbA1C levels, average reduction of fasting blood glucose, percent change in DNA methylation, directionality of DNA methylation changes, among other improved outcomes.

5 Executing the personalized intervention, treatment, management and/or monitoring plan in Row (I) can include providing results presented one or more sections of one or more reports generated from model outputs (e.g., within an application environment), which were determined based on one or more markers of the same or different type. Reports can then be transmitted to the entities involved (e.g., patients,
10 caretakers, insurance companies, etc.) by mobile application and/or web application architecture. Executing the personalized intervention, treatment and/or monitoring plan in Row (I) can further implement the individual marker and/or aggregate marker profiles to guide the course of a patients' precision care and/or monitoring and/or management and/or treatment and/or coaching.

15 Executing the personalized intervention, treatment, management and/or monitoring plan in Row (I) can further include providing a personalized care program that implements body metrics, clinical metrics, lifestyle metrics, epigenetics and genetic profiles, and personalized health-coaching to manage target outcome (e.g., weight loss, reduce heart attack risk). In accordance with the personalized care program, patients can
20 be provided with digital tools for tracking lifestyle and wellness markers (i.e., blood pressure, heart rate, weight, sleep, hunger, cravings, stress, meditation, superfoods, energy, foods to avoid, exercise, etc.), documenting their habits and markers (e.g., through a photo journal, through a text journal, through a wearable device, through an app, etc.), and are assigned a health coach who works personally with the patient through
25 guided sessions as scheduled by the patient to interpret the personalized reports generated from the patients' tracking and data. In accordance with the personalized care program, the reports can provide a breakdown of metrics being tracked (e.g., obesity risk, etc.) based on an individual's data and measurement(s) (e.g., epigenetic profile, age, etc.). The program can be geared toward one or more goals (e.g., reducing 5% of their blood
30 pressure within 90 days of starting the program, reducing the number employee emergency room visits). To achieve this goal, example implementations of the program can include automated and manual tools for motivating participants to make incremental lifestyle changes, reminders to track progress, and tools to track progress.

In specific examples, implementation of the personalized intervention, treatment, management and/or monitoring plan, based upon outputs of models and/or data described, may not incorporate all elements such as a health coach or be substituted with similar elements (e.g., clinician instead of a health coach) or other elements can be added (e.g.,
 5 clinician in addition to the health coach). Additionally, outcomes measured upon the implementation of the personalized intervention, treatment, management and/or monitoring plan, can be used to iterate upon or optimize or change the personalized intervention, treatment, management and/or monitoring plan.

In specific examples, implementation of the personalized intervention, treatment,
 10 management and/or monitoring plan, based upon outputs of models and/or data described, can be used to evaluate individual, multiple, aggregate, comparative or simultaneous changes quantitatively or qualitatively at one or multiple time points in one or more of: financial (e.g., cost of care, cost of medication, etc.), prevalence (e.g., percent employees at risk for a heart attack, etc.), laboratory tests (e.g., DNA methylation level, change in
 15 inflammation level, etc.), imaging (e.g., FFR from CCTA, etc.), medical (e.g., changes in metrics reported in electronic medical records, etc.), clinical (e.g., changes in blood pressure, reduction of severity of symptoms such as chest pain, etc.), co-morbidities (e.g., obesity, diabetes, hypertension, hypercholesterolemia, etc.) therapeutic (e.g., type of statin associated with the largest changes to lipid level, etc.), supplement (e.g., vitamin D,
 20 vitamin B12, iron, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), or other suitable parameter(s) at baseline or in comparison to baseline state(s) for a patient or a group of patients or a user or a group of users on the personalized intervention, treatment, management and/or monitoring plan.

The set of cardiovascular symptoms can include one or more of: pain level (e.g.,
 25 chest pain level, extremity pain level, neck pain level, throat pain level, abdominal pain level, back pain level, etc.), chest tightness, chest pressure, angina, respiratory distress (e.g., shortness of breath, blood oxygenation, respiration rate, etc.), extremity numbness, extremity strength, vasculature narrowness, cardiovascular parameters (e.g., heart rate, heart rate variability, blood pressure, output, electrocardiogram signatures, stroke volume,
 30 cardiac index, etc.), cholesterol levels (e.g., HDL levels), blood glucose levels, HbA1c levels, percent stenosis, occlusion level, fractional flow reserve, DNA methylation values, genetic profile and other cardiovascular symptoms. Cardiovascular symptoms can be associated with one or more of: coronary artery disease, coronary heart disease,

hypertension, cardiac arrest, congestive heart failure, arrhythmias, peripheral artery disease, stroke, congenital heart disease, and/or other diseases or co-morbidities.

The methods described herein can produce changes to one or more of the factors outlined herein individually and/or simultaneously, within a duration of less than 30 days, 5 30-60 days, 60-90 days, 90-120 days, any intermediate number of days, or greater than 120 days. Variations of the methods described can produce changes to one or more of the factors outlined herein individually and/or simultaneously, by at least 0.1% in a patient.

Additionally or alternatively, the methods described herein can be used to evaluate changes to one or more of the factors outlined herein individually and/or simultaneously, 10 once (e.g., at a single time point), or at a number of time points (e.g., at random points, at regular points, in relation to triggering events, with other frequency, etc.)

While this specification contains many specific implementation details, these should not be construed as limitations on the scope of any inventions or of what may be 15 claimed, but rather as descriptions of features specific to particular implementations of particular inventions. Certain features that are described in this specification in the context of separate implementations can also be implemented in combination in a single implementation. Conversely, various features that are described in the context of a single implementation can also be implemented in multiple implementations separately or in any 20 suitable sub-combination. Moreover, although features may be described herein as acting in certain combinations and even initially claimed as such, one or more features from a claimed combination can in some cases be excised from the combination, and the claimed combination may be directed to a sub-combination or variation of a sub-combination.

While this document has presented certain realizations of the methods and 25 compositions described herein for illustrative purposes, it should be recognized by those with expertise in the field that these examples are not limiting. Those with the requisite knowledge may implement a range of modifications, changes, and substitutions without straying from the essence of the methods and compositions. It would be appreciated that various alternatives to the disclosed embodiments can be devised by those familiar with 30 the field.

Similarly, while operations are depicted in the drawings in a particular order, this should not be understood as requiring that such operations be performed in the particular order shown or in sequential order, or that all illustrated operations be performed, to

achieve desirable results. In certain circumstances, multitasking and parallel processing may be advantageous. Moreover, the separation of various system components in the implementations described herein should not be understood as requiring such separation in all implementations, and it should be understood that the described program
5 components and systems can generally be integrated together in a single software product or packaged into multiple software products.

Thus, particular implementations of the subject matter have been described. Other implementations are within the scope of the following claims. In some cases, the actions recited in the claims can be performed in a different order and still achieve desirable
10 results. In addition, the processes depicted in the accompanying figures do not necessarily require the particular order shown, or sequential order, to achieve desirable results. In certain implementations, multitasking and parallel processing may be advantageous.

WHAT IS CLAIMED IS:

1. A method comprising:
 - generating, by a computer system, a user interface for display on a display
 - 5 device operatively coupled to the computer system;
 - generating, by the computer system, first data comprising genetic, epigenetic, and/or other biomarker test results for a patient to evaluate the presence or absence of a disease, disorder, or risk in the patient;
 - displaying the user interface on the display device; and
 - 10 displaying the first data within the user interface.
2. The method of claim 1, wherein the data is selected from measurements (e.g., weight, blood pressure, etc.), test results (e.g., genetic, epigenetic, protein, metabolic assays, etc.), financial (e.g., cost of procedures, cost of medication, cost of claims, etc.),
15 industry / prevalence (e.g., heart disease prevalence in a particular geographic region, number of heart attacks a year among truck drivers, etc.), interventions (e.g., surgery, etc.), medical (e.g., ICD10 codes, medication usage, etc.), clinical (e.g., blood pressure, etc.), therapeutic (e.g., cost of statins, drugs that reduce inflammation, etc.), lifestyle (e.g., smoking, alcohol consumption, exercise frequency, etc.), supplement usage (e.g., herbs,
20 vitamin D, vitamin B12, iron, etc.), electronic data (e.g., imaging, electronic health records, publicly available data (e.g., repositories, etc.), publications, pharmaceuticals, clinical trials, etc.) and the diagnostic, intervention, management, monitoring and/or therapeutic implications thereof.
- 25 3. The method of claim 1 or 2, wherein the data comprises standalone data, aggregated data, imputed data, derived data, numerical data, text data, image data, and combinations thereof.
4. The method of any one of claims 1 to 3, wherein the data is displayed in the
30 user interface in one or more plots.
5. The method of any one of claims 1 to 4, wherein the data is compared to corresponding genetic, epigenetic, and/or other biomarker test results from a cohort

population.

6. The method of any one of claims 1 to 5, wherein the data is superimposed on genetic, epigenetic, and/or other biomarker test results for a population.

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7. The method of any one of claims 1 to 6, wherein the user interface displays diagnostic information, prognostic information, uncertainty quantifications, clinical and/or business risk management or decision-making, regulatory and/or policy making, intervention, monitoring and/or therapeutic decisions and strategies related to a patient or
10 a group of patients or a user or a group of users.

8. The method of any one of claims 1 to 7, wherein the genetic, epigenetic, and/or other biomarker test results for the patient population comprises a bar chart showing ages of the patient population on an X-axis and probabilities that patients in different ages have
15 the disease, disorder or risk on a Y-axis, wherein the test results for the patient comprise a probability that the patient has the disease.

9. The method of any one of claims 1 or 8, wherein the patient is associated with an age, wherein the method further comprises displaying the probability that the patient
20 has the disease adjacent to a probability of the patient population of the same age as the patient.

10. The method of any one of claims 1 to 9, wherein the patient is associated with a gender, wherein the method further comprises displaying the probability that the patient
25 has the disease adjacent to a probability of the patient population of the same gender as the patient.

11. The method of any one of claims 1 to 10, further comprising:
receiving a ranking of contributions of a plurality of genetic, epigenetic,
30 and/or other biomarkers including the genetic, epigenetic, and/or other biomarker to the presence of the disease in the patient;
generating a chart comprising names of the plurality of markers on an X-axis and a normalized rank of the contributions of the plurality of genetic, epigenetic,

other biomarkers for the patient;

superimposing the chart on another chart showing normalized ranking of contributions of the plurality of genetic, epigenetic, and/or other biomarkers to the patient population resulting in a second plot of rankings versus markers; and

5 displaying the second plot adjacent the plot within the user interface.

12. The method of any one of claims 1 to 11, further comprising:

generating a plot of a genetic, epigenetic, and/or other biomarker measurement distribution associated with a specific genetic, epigenetic, and/or other
10 biomarker;

superimposing on the plot, the genetic, epigenetic, and/or other biomarker measurement measured for the patient; and

displaying the plot with the superimposed genetic, epigenetic, and/or other biomarker measurement within the user interface.

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13. The method of any one of claims 1 to 12, further comprising:

determining an upper limit and a lower limit of uncertainty associated with the genetic, epigenetic, and/or other biomarker measurement measured for the patient; and

20 displaying, within the plot, the upper limit and the lower limit bounding the genetic, epigenetic, and/or other marker measurement.

14. The method of any one of claims 1 to 13, further comprising:

detecting a selection of the data displayed within the user interface;

25 in response to detecting the selection, displaying a window adjacent the data within the user interface; and

displaying, within the window, hyperlinks to further information.

15. The method of claim 14, wherein the further information comprises literature

30 related to the genetic, epigenetic, and/or other biomarker test results or literature related to the disease, disorder or risk.

16. The method of any one of claims 1 to 15, wherein the literature is selected

from publications, clinical data, clinical trials and pharmaceuticals.

17. A computer-readable medium storing computer instructions which, when
executed by one or more computer processors, is configured to cause the one or more
5 computer processors to perform operations comprising a method recited in any one or
more of claims 1 to 16.

18. A computer system comprising:
one or more computer processors; and
10 a computer-readable medium storing computer instructions which, when
executed by the one or more processors causes the one or more processors to perform
operations comprising a method recited in any one or more of claims 1 to 16.