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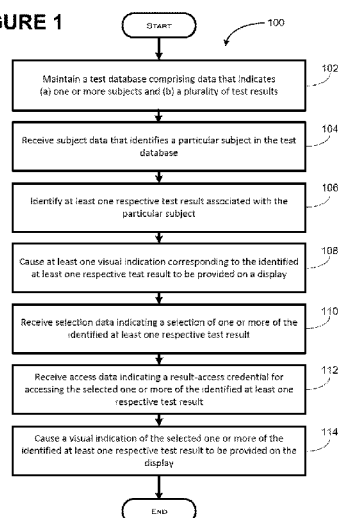
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(54) Title: METHOD AND SYSTEM FOR RETURNING PATIENT TEST RESULTS

FIGURE 1



(57) Abstract: Example systems and methods are provided for returning patient genome or exome sequencing results. In one example, a test database including data indicating one or more subjects and a plurality of test results may be maintained. Each subject may be associated with one or more respective test result from the plurality of test results. Returning patient genome or exome sequencing results from the database may involve receiving subject data identifying a particular subject in the test database, identifying at least one respective test result associated with the particular subject, causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display, receiving selection data, receiving access data, and causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display in response to receiving the access data.



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METHOD AND SYSTEM FOR RETURNING PATIENT TEST RESULTS

CROSS-REFERENCE TO RELATED APPLICATION

[01] This application claims the benefit of U.S. Provisional Application No. 61/692,646, filed August 23, 2012, which is incorporated herein by reference in its entirety.

BACKGROUND

[02] Whole genome sequencing is a process for determining a complete DNA sequence of an organism's genome. This process may involve sequencing all of the chromosomal DNA and the mitochondrial (or chloroplast) DNA of the organism. In some cases, a single biological sample from the organism may contain a full copy the organism's DNA, and may therefore be sufficient for full genome sequencing. Such samples may include, for example, saliva, epithelial cells, bone marrow, hair follicles, seeds, and plant leaves, among other examples.

[03] The amount of DNA sequence data produced from whole genome processing may be quite large. For instance, there are approximately six billion base pairs in each human diploid genome. As such, the computing power and data storage requirements necessary for whole genome sequencing may be quite large.

[04] Exome sequencing, also known as targeted exome capture, involves selectively sequencing the coding regions of the genome. Exons are generally short, functionally important sequences of DNA which represent the regions in genes that are translated into protein and the un-translated regions flanking them. There are approximately 180,000 exons in the human genome, and those exons constitute only about 1% of the human genome. Nevertheless, the protein coding regions of the human genome (i.e., those regions targeted by exome capture) have been estimated to constitute about 85% of disease-causing mutations. As such, the computing power and data storage requirements necessary for exome sequencing may be considerably less than that necessary for whole genome sequencing. Accordingly, exome sequencing may provide a less expensive but still effective alternative to whole genome sequencing, especially for purposes of detecting disease-causing mutations.

[05] Test results, whether from whole genome sequencing, exome sequencing, or other health and medical related tests such as blood tests, may be complicated and/or sensitive. The results are often sent from a laboratory, where analysis of a patient's test sample may have been performed, to a medical practitioner of the patient. According to some procedures, the medical practitioner may review the test results, and the patient and/or parents of the patient and the medical practitioner may subsequently schedule a time to meet and discuss the

results. This process, however, may be tedious and inefficient due to, for example, potential scheduling conflicts between the two parties involved and potentially limited time and attention available from either party. As such, systems and methods for helping improve the return of test results to a patient and/or other authorized individuals may be beneficial.

SUMMARY

[06] In one aspect, a computer implemented method is provided. The method includes maintaining a test database including data that indicates (a) one or more subjects and (b) a plurality of test results. Each subject is associated with one or more respective test result from the plurality of test results. The method also includes receiving subject data that identifies a particular subject in the test database, identifying at least one respective test result associated with the particular subject, causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display, and receiving selection data. The selection data indicates a selection of one or more of the identified at least one respective test result via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result. The method also includes receiving access data. The access data indicates a result-access credential for accessing the selected one or more of the identified at least one respective test result. The method also includes, in response to receiving the access data, causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display.

[07] In another aspect, a system is provided. The system includes a processor, a physical computer readable medium, and program instructions stored on the physical computer readable medium. The program instructions are executable by the processor to maintain a test database including data that indicates (a) one or more subjects and (b) a plurality of test results. Each subject is associated with one or more respective test result from the plurality of test results. The program instructions are also executable by the processor to receive subject data that identifies a particular subject in the test database, identify at least one respective test result associated with the particular subject, cause at least one visual indication corresponding to the identified at least one respective test result to be provided on a display, and receive selection data. The selection data indicates a selection of one or more of the identified at least one respective test result via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result. The program instructions are also executable by the processor to receive access data. The access data indicates a result-access credential for accessing the selected one or more of the identified at least one respective test result. The program instructions are also executable by the processor to cause a visual

indication of the selected one or more of the identified at least one respective test result to be provided on the display in response to receiving the access data.

[008] In yet another aspect, a physical computer readable medium having instructions stored thereon is provided. The instructions include instructions for maintaining a test database including data that indicates (a) one or more subjects and (b) a plurality of test results. Each subject is associated with one or more respective test result from the plurality of test results. The instructions also include instructions for receiving subject data that identifies a particular subject in the test database, instructions for identifying at least one respective test result associated with the particular subject, instructions for causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display, and instructions for receiving selection data. The selection data indicates a selection of one or more of the identified at least one respective test result via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result. The instructions also include instructions for receiving access data. The access data indicates a result-access credential for accessing the selected one or more of the identified at least one respective test result. The instructions also includes instructions for causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display in response to receiving the access data.

BRIEF DESCRIPTION OF THE DRAWINGS

[009] **FIGURE 1** shows a flowchart depicting an example embodiment for returning patient test results, in accordance with an example embodiment.

[010] **FIGURES 2A – 2L** each show aspects of an example interface for returning patient test results, in accordance with an example embodiment.

[011] **FIGURES 3A – 3L** each show aspects of an example interface for returning patient test results, in accordance with an example embodiment.

[012] **FIGURE 4** shows a physical computer-readable medium, in accordance with an example embodiment.

[013] **FIGURE 5** shows a simplified block diagram of an example communication network in which the present methods can be implemented.

[014] **FIGURE 6** shows a simplified block diagram of a network-access device arranged to implement aspects of at least one example embodiment of the methods.

[015] **FIGURE 7** shows a simplified block diagram of a server arranged to implement aspects of at least one example embodiment of the methods.

DETAILED DESCRIPTION

[016] The present application is directed to returning patient test results, such as patient genome or exome sequencing results. While the example embodiments described herein may relate particularly to returning patient genome or exome sequencing results, one having ordinary skill in the art will appreciate that the systems and methods disclosed in relation to the example embodiments may be implemented for a variety of different applications. The variety of different applications may include the return of other patient health and medical test results, or general academic or certification test results.

[017] Whether the test results indicate medical conditions or any other information, the test results may be both complicated and sensitive. As such, a suitable system and/or method may be provided for returning the test results in an efficient, secure, and meaningful way.

[018] Figure 1 is a flowchart depicting an example embodiment for returning patient test results, in accordance with at least some embodiments described herein. Method 100 shown in Figure 1 may include one or more operations, functions, or actions as illustrated by one or more of blocks 102-114. As shown, block 102 of the method 200 generally relate to maintaining a test database of data indicating test subjects and corresponding test results, blocks 104-106 generally relate to determining test results accessible by a user, blocks 108-112 generally relate to selection of test results a user may desire to view, and block 114 generally relates to providing the test results to the user. Although the blocks are illustrated in sequential order, these functions associated with these blocks may also be performed in parallel, and/or in a different order than those described herein. Also, the various blocks may be combined into fewer blocks, divided into additional blocks, and/or removed based upon the desired implementation.

[019] In addition, for the method 100 and other processes and methods disclosed herein, the flowchart shows functionality and operation of one possible implementation of present embodiments. In this regard, each block may represent a module, a segment, or a portion of program code, which includes one or more instructions executable by a processor for implementing specific logical functions or steps in the process. As will be further discussed below in connection to Figure 4, the program code may be stored on any type of computer readable medium, for example, such as a storage device including a disk or hard drive. The computer readable medium may include physical and/or non-transitory computer readable medium, for example, such as computer-readable media that stores data for short periods of time like register memory, processor cache and Random Access Memory (RAM). The computer readable medium may also include physical and/or non-transitory media, such as secondary or persistent long term storage, like read only memory (ROM), optical or magnetic

disks, compact-disc read only memory (CD-ROM), for example. The computer readable media may also be any other volatile or non-volatile storage systems. The computer readable medium may be considered a computer readable storage medium, for example, or a tangible storage device. In addition, for the method 100 and other processes and methods disclosed herein, each block in Figure 1 may represent circuitry that is wired to perform the specific logical functions in the process.

[020] Further, as will be further discussed below in connection to Figures 5-7, the method 100 and other processes disclosed herein may be carried out by a computing system in a network, e.g. in communication with a network, or locally. The computing system may include one or more of a personal computer, a laptop computer, a tablet computer, a cellular phone, or any type of user interface-capable device.

[021] As shown, block 102 may involve maintaining a test database including data that indicates (a) one or more subjects and (b) a plurality of test results. In one case, each subject may be associated with one or more respective test result from the plurality of test results. As suggested above, the one or more respective test results may be genome sequencing results, exome sequencing results, other types of medical test results, or general academic or certification test results. In some cases, patient genome or exome sequencing may be ordered for patients that may have a genetic condition. As such, the one or more respective test result associated with a particular subject may indicate one or more genetic conditions of the particular subject.

[022] Prior to or as part of maintaining the test database as discussed in connection to block 102, the one or more respective test result associated with the particular subject may be received at the test database as test-result data indicating at least one of the one or more respective test results associated with the particular subject. The test-result data may be received at the test database from laboratory where analysis of a sample of the particular subject was performed. In one case, the test-result data may be received over a network, such as the internet or a local access network. In another case, the test-result data may be received locally via input into a computing system coupled to the test database. In one example, the test-result data may be automatically received at the test database once the test-result data is available from the laboratory. In another example, the test-result data may be reviewed and annotated by a medical practitioner before being received at the test database.

[023] In one example, the received test-result data may include (a) an identification of the particular subject and (b) annotated variant data generated based on a test performed on the particular subject. In one example, the annotated variant data may indicate biological

information that has been identified within the sequenced genome or exome of the particular subject. In some cases, a genome or exome sequencing process performed on the particular subject may produce raw genome or exome sequence data. In this case, the raw or exome genome sequence data may be further processed and annotated to identify biological information represented within the genome or exome sequence data, producing the annotated variant data.

[024] Upon receiving the test-result data, a result-access credential for accessing the at least one test result may be generated based on the identification of the particular subject included with the test-result data. In one case, the result-access credential may be unique to the particular subject. In another case, the result-access credential may be unique to the specific at least one test result for the particular subject. In other words, the result-access credential provides credentials at least unique to the particular subject, and in some cases unique to the specific at least one test result. Ultimately, only users of the test database with the result-access credential may access the at least one test result for the particular subject. In one case, the result-access credential may be generated and provided to the particular subject such that particular subject can access the at least one test result. In another case, the result-access credential may be generated and provided to immediate family members or caretakers of the particular subject such that they may access the at least one test result.

[025] In one example, the result-access credential may be in the form of a textual access code. In one case, a single textual access code may be generated for any user authorized to access the at least one test result. In another case, different textual access codes may be generated for different users authorized to access the at least one test result. In some cases, different textual access codes may grant different degrees of access to the at least one test result.

[026] After generating the result-access credential, the received test-result data may be stored in the test database as part of maintaining the test database. In some cases, the generated result-access credential may also be stored in the test database along with the corresponding test-result data. In some other cases, the generated result-access credential may be stored in a separate database associated with the test database.

[027] As mentioned, only users of the test database with result-access credential for the at least one test result may access the at least one test result for the particular subject. In one example, a user may be required to set up an account to access the test database. Setting up the account may involve receiving from the user user-identification data indicating a database-access credential for accessing the test database. In one case, the user may setup an

account to access the test database via a user interface or network interface, such as web browser on a computer in communication with the test database. In one instance, the computer may be connected to the Internet and may access the test database over the World Wide Web. In another instance, the computer may be locally connected to the test database, such as over a local area network. As suggested above, the user-identification data may be associated with the identification of the particular subject. In some cases, a single user of the test database may be associated with more than one particular subject. For instance, the user may be a parent of children, each of who are subjects of genome or exome sequencing, and the parent may therefore be associated with each child.

[028] Block 104 of the method 100 may involve receiving subject data that identifies a particular subject in the test database. In this case, the user of the test database may provide the subject data when accessing the test database to indicate the particular subject for which the user wishes to access the test results. In one example, the subject data may be received from the user when the user is setting up an account to access the test database. Figure 2A shows an example interface 200 through which the user may setup an account, provide the subject data, and subsequently access the test database. In other words, the interface 200 as shown in Figure 2A may be provided such that the subject data identifying the particular subject in the test database may be received, as described in connection to block 104.

[029] The interface 200 may be provided on a display, which in some cases may be a touch-sensitive display. The display may be configured to provide access to the test database via the interface 200. In this instance, the interface 200 as shown in Figure 2A provides a “Create Account” box for receiving user-identification data 202 when the user sets up the account to access the test database. As shown, first and last names, an email address, a username and a password may be received to set up the account. In addition to the user-identification data 202, the “Create Account” box may also receive subject data 204 for linking or associating the particular subject with the account being setup. Upon entering the necessary information, the user account may be setup and the user may be automatically logged in to access the test database.

[030] In another case, as shown in Figure 2B, the example interface 200 may provide a login prompt 206 for the user to login if an account has already been set up. In this case, if the subject data was already received when the user account was setup, as discussed above, the user may not be prompted to provide the subject data again. In another case, which is not shown, the login prompt 206 may require the subject data to be provided each time the user attempts to login to access the test database.

[031] Block 106 of the method 100 may involve identifying at least one respective test result associated with the particular subject. After logging on to access the test database via the interface 200 shown in Figures 2A or 2B, the interface 200 may provide for display information on how to start accessing the at least one test result associated with the subject data provided. As shown in Figure 2C, the interface 200 may include a genetic counselor contact section 212 providing means to contact a genetic counselor if the user has any questions at any time. Further, a flow diagram 210 indicates steps for accessing test results in the test database. As shown in Figure 2C, the interface 200 may provide a “Get Started” icon selectable to proceed with accessing test results.

[032] As indicated above, subject data identifying the particular subject may have been provided when the user set up the account to access the database. Accordingly, identifying the at least one respective test result associated with the particular subject may be based on at least the user-identification data provided when setting up the account. As discussed above, the user-identification data may be associated with the identification of the particular subject. As such, test results associated with the particular subject may be identified through an identification of the particular subject associated with the user-identification data. In other words, in the case the user is a parent wishing to access the test database to view test results for a child, the test results for the child may be identified at least partially based on the identification of the parent, and data in the test database indicating the relationship between the parent and the child.

[033] In some cases, to more discriminately identify the at least one respective test result associated with the particular subject that the user wishes to access, additional information may be received from the user to identify the particular subject. For instance, as shown in the interface 200 of Figure 2D, a “Link Person Tested” box may be provided to receive additional information identifying the particular subject. In this example, the “Link Person Tested” box may be configured to receive information such as a first name, last name, and date of birth of the particular subject. In addition, an optional medical record number, if available, may be provided to further identify the particular subject. As shown in Figure 2D, a “Submit” icon may be selectable to submit the entered information and proceed to access the identified at least one respective test result.

[034] Upon selection of the “Submit” icon, the interface 200 as shown in Figure 2E may provide a “Results Preference” section 216 indicating that one non-selected test result is available for the identified particular subject. The interface 200 may further provide a “Set Preferences” icon selectable to proceed with setting preferences for viewing the at least one

respective test result associated with the particular subject, which in this case is the one non-selected test result. Upon selection of the “Set Preferences” icon, the interface 200 may provide preference setting options as shown in Figure 2F.

[035] Referring back to the method 100 of Figure 1, block 108 may involve causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display. In some cases, the identified at least one respective test result may be organized according to a category of the test. Accordingly, block 108 may further involve determining at least one result category associated with the identified at least one respective test result, and causing at least one visual indication corresponding to the at least one determined result category to be provided on the display.

[036] As shown in Figure 2F, causing the at least one visual indication corresponding to the at least one determined result category to be provided on a display may involve the interface 200 providing a result categories grid 218 showing a plurality of result category tiles. In this case, a “Genetic Syndromes” category tile may be highlighted (in this case, with accentuated borders) to indicate the identified at least one respective test result has been categorized under a “Genetics Syndromes” category. In some cases, as will be shown in examples below, a single test result may be categorized under multiple categories.

[037] In this example, the highlighted “Genetic Syndromes” category tile may further indicate that preferences for viewing the identified at least one respective test result may be set by selecting the “Genetic Syndromes” category tile. Accordingly, category-selection data indicating a selection of one or more of the at least one determined result category may be received via a selection of one or more of the at least one visual indication corresponding to the at least one determined result category. As mentioned above, the selected one or more of the at least one determined results category may be associated with the identified at least one respective test result.

[038] Upon selection of the “Genetic Syndromes” category style, the interface 200 may provide the at least one visual indication corresponding to the identified at least one respective test result, as shown in Figure 2G. In this case, as shown, the interface 200 may provide a result-viewing selection box 222 including selectable options 224 corresponding to the identified at least one respective test result. As shown in this case, the identified at least one respective test result may be a test result for “fragile x syndrome.” In one example that is not shown, a visual indication of the “fragile x syndrome” test results may be selectable to cause selectable option for viewing the “fragile x syndrome” test results to be displayed. In some cases, the at least one visual indication the identified at least one respective test result

may be organized by alphabetical order of a name of the identified at least one respective test result, or by the category or subcategory of the identified at least one respective test result. Other examples of visual indications may also exist.

[039] Referring back to the method 100 of Figure 1, block 110 may involve receiving selection data indicating a selection of one or more of the identified at least one respective test result. In one example, the selection data may be received via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result. As shown in Figure 2G, the received selection data may indicate as selection of “YES, I want results” for the “fragile x syndrome” test results. Subsequently, a selection of a “Save preferences” icon on the interface 200 may cause selection-preference data indicating the selection of the one or more of the identified at least one respective test result, which in this case may be the result for “fragile x syndrome,” to be stored in the test database. In one example, the selection-preference data may be associated with the user-identification data of the user to accessing the test database. As such, the result-viewing preferences for “fragile x syndrome” test results may be retained for when the user logs on to access the test database at a later time.

[040] Tests relating to genetic conditions or the human genome or exome in general may be complicated and daunting to some users. As such, in some cases, the selection-preference data may be received through a less explicit manner than that shown in Figure 2G. For instance, surveys configured to guide a user through setting result viewing preferences may be provided to help the user select results for viewing that best suits the user’s needs.

[041] Upon selection of the “Save preferences” icon, the interface 200 may provide a “Change Preferences” icon, a “View Results” icon, and/or a “Learn More” icon as shown in Figure 2H. Selection of the “Change Preferences” icon may bring the user back to the interface 200 as shown in Figure 2F, and selection of the “Learn More” icon may bring the user to a different interface that offers the user access to information on genetic testing, genome or exome sequencing, and the test database, etc.

[042] Upon receiving the selection data as described in connection to block 110, block 112 of the method 100 may involve receiving access data indicating the result-access credential for accessing the selected one or more of the identified at least one respective test result. Selection of the “View Results” icon provided by the interface 200 shown in Figure 2H may cause the interface 200 as shown in Figure 2I to provide a partial visual indication 234a of the selected one or more of the identified at least one respective test result, which in this case is the “fragile x syndrome” test result. In this case, the partial visual indication 234a only

identifies the test results and category of the test results without providing further information. The interface 200 as shown in Figure 2I may further provide the Results Preference section 216 indicating that one of the identified at least one test result has been selected, and a Results section 238 indicating that viewing of the selected one of the at least one test result is pending.

[043] The partial visual indication 234a and the Results section 238 indicating that viewing of the selected one of the at least one test result is pending may be because a result-access credential as discussed above may be required for viewing the selected one or more of the identified at least one test result. In other words, access data indicating the result-access credential for accessing the selected one or more of the identified at least one respective test result may be required.

[044] In one example, in response to receiving the selection data indicating a selection of one or more of the identified at least one respective test result and/or receiving the selection of the “View Results” icon shown in Figure 2H, access requirements such as the previously generated result-access credential for the selected one or more of the identified at least one respective test result may be determined. In one case, determining the access requirements for the selected one or more of the identified at least one respective test result may involve retrieving the result-access credential associated with the selected one or more of the identified at least one respective test result from the test database or a separate database associated the test database where the generated result-access credential was stored.

[045] As shown in Figure 2I, the interface 200 may further provide a visual indication of a prompt 236 to be provided on the display. The prompt 236 may request access data indicating the result-access credential to access the selected one or more of the identified at least one respective test result. As previously indicated, the result-access credential may be in the form of a textual access code. As shown in Figure 2I, access data indicating the result-access credential in the form of a textual access code may be provided to the prompt 236.

[046] In response to receiving the access data upon a selection of a “Submit” icon in the prompt 236 to submit the entered textual access code and subsequently matching the submitted access data with the previously generated, stored, and retrieved result-access credential, block 114 may involve causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display. The interface 200 of Figure 2J shows a full visual indication 234b of the selected one or more of the identified at least one respective test result, which in this case is the “fragile x syndrome” test result. As shown, the full visual indication 234b includes selectable “Summary” and “Report”

icons for viewing a summary and a report, respectively, for the “fragile x syndrome” test result.

[047] The full visual indication 234b further includes an importance indicator 228 indicating an inferred degree of impact of the identified at least one respective test result on a well-being of the particular subject. In one example, if the test result for a specific genetic condition is negative, then the inferred degree of impact on the well-being of the particular subject may be low, and the importance indicator 228 may indicate as such. As shown in Figure 2J, the inferred impact of the “fragile x syndrome” test results is low, and the importance indicator 228 indicates so. Also as shown in Figure 2J, the Results section 238 has been updated to indicate that new test results are available, and that no test results are pending.

[048] The interface 200 of Figure 2K provides an example summary 242 displayed if the “Summary” icon of Figure 2J is selected, and the interface 200 of Figure 2L provides an example report 244 displayed if the “Report” icon of Figure 2J is selected.¹² As shown, the result of the “fragile x syndrome” test is negative, hence the inferred low impact on the well-being of the particular subject. In general, the summary 242 and/or the report 244 may provide information indicating (a) a description of the genetic condition, (b) symptoms of the genetic condition, (c) health risks relating to the genetic condition, and (d) available options for managing the genetic condition. Other examples are also possible. In the context of block 114, the visual indication of the selected one or more of the identified at least one respective test result may refer to either or both of the full visual indication 234b provided by the interface 200 in Figure 2I or the summary 242 and report 244 provided by the interface 200 in Figures 2K and 2L, respectively.

[049] In one example, the visual indication of the selected one or more of the identified at least one respective test result may be selected based on the annotated variant data in the test result data, previously provided as discussed in connection to block 102. In other words, content of the summary 242 and/or report 244 may be compiled based on the annotated variant data associated with the “fragile x syndrome” test results. In one example, different content may be previously prepared for each available genetic test at each inferred level of impact and subsequently provided to a user based on the test result and inferred level of impact for the particular subject.

[050] As indicated by the interface 200 in Figures 2J-L, the result of the “fragile x syndrome” test is negative for the particular subject. In one example, additional genetic tests may have been ordered for the particular subject and updated test results may subsequently be

available. In one example, the user may receive a phone call or email informing the user that new test results are available in the test database.

[051] Figure 3A shows the example interface 200 when the user accesses the test database again, after being informed that new test results are available. As shown, the Results Preference section 216 may indicate that one result has been selected, and three results have not been selected. In this case, the one result that has been selected may be the previously selected “fragile x syndrome” test result. In this case, the “Set Preferences” icon on the interface 200 may be selected to proceed with setting result-viewing preferences for the three non-selected test results.

[052] Because the user has previously accessed the test database and may have provided subject data as discussed above in connection to blocks 102-106, the interface 200 as shown in Figure 3B may proceed directly to cause at least one visual indication corresponding to the identified at least one respective test result to be provided on the display, as discussed above in connection to block 108. As previously shown in Figure 2F, the interface 200 of Figure 3B may cause the at least one visual indication corresponding to the at least one determined result category to be provided on a display by providing the result categories grid 218. In this case, the three non-selected test results may fall under five different categories visually represented by a highlighted “Genetic Syndromes” tile, a highlighted “Metabolic Disorders” tile, a highlighted “Disease Risk” tile, a highlighted “Carrier Status” tile, and a highlighted “Newborn Screening Conditions” tile. From the result categories grid 218, the “Disease Risk” tile may be selected for further selection of test results to be viewed.

[053] The interface 200 of Figure 3C provides the result-viewing selection box 222 including selectable options 246 corresponding to a “sickle cell disease” test result so as to, as discussed above in connection to block 110 of the method 100, receive selection data indicating a selection of one or more of the identified at least one respective test result, which in this case is the “sickle cell disease” test result. As shown, a selection of “Yes, I want results” may be selected from the selection options 246 corresponding to the test result for “sickle cell disease,” and a selection of the “Save preferences” icon may cause the selection to be stored, as also described above in connection to block 110.

[054] Proceeding to Figure 3D accordingly, the Results Preferences section 216 of the interface 200 indicates that two results have been selected, and that two results have not been selected, and the Results section 238 indicates that one test result is pending. In this case, because there are test results that have not yet been selected, the interface 200 may be configured to cause the at least one visual indication corresponding to the at least one

determined result category to be provided on a display as discussed above in connection to block 108 by once again providing the result categories grid 218. In this case, the result categories grid 218 may show only four highlighted tiles - the “Genetic Syndromes” tile, the “Metabolic Disorders” tile, the “Carrier Status” tile, and the “Newborn Screening Conditions” tile, indicating that the two non-selected test results fall under the four categories visually represented by highlighted tiles.

[055] From the result categories grid 218, the “Genetic Symptoms” tile may be selected for further selection of test results to be viewed, and the interface 200 of Figure 2E may provide the result-viewing selection box 222 including selectable options 248 corresponding to test results for “classic galactosemia,” “fragile x syndrome,” “kabuki syndrome,” and “sickle cell disease” so as to receive selection data indicating a selection of one or more of the identified at least one respective test result as discussed above in connection to block 110. Because result viewing preferences for “sickle cell disease” and “fragile x syndrome” have been previously provided and stored, the selectable options 248 in the result-viewing selection box 222 may initially show the previously provided selections for “sickle cell disease” and “fragile x syndrome,” while no selections are shown for the selectable options 248 in the result-viewing selection box 222 for “classic galactosemia” and “kabuki syndrome.” Within the selection box 222, the selection options 248 for each of the four test results may be selected, including the two results for which selections have previously been made. As shown, a selection of “Yes, I want results” may be selected from the selection options 248 corresponding to each of the four test results, and a selection of the “Save preferences” icon may cause the selections to be stored, also similar to that described above in connection to block 110.

[056] Proceeding to Figure 3F, the Results Preferences section 216 of the interface 200 indicates that four results have been selected, and no results remain non-selected, and the Results section 238 indicates that three test results are pending. As shown, no category tile in the category grid 218 is highlighted, indicating that none of the visually represented categories include non-selected test results.

[057] From the interface 200 shown in Figure 3F, a selection of the “Go to Results Navigator” icon may cause the interface 200 as shown in Figure 3G to provide partial representations 250a, 252a, and 254a corresponding to test results for “classic galactosemia,” “kabuki syndrome,” and “sickle cell disease,” respectively, similar to that described above in connection to Figure 2I. In this case, the full visual representation 234b for the previously selected and displayed “fragile x syndrome” test result may also be provided on the interface

200 because “fragile x syndrome” remained selected on the interface 200 of Figure 3E. As discussed above in connection to Figure 2I and block 112, the interface 200 as shown in Figure 3G may provide the visual indication of the prompt 236 to be provided on the display so as to receive access data indicating a result-access credential for accessing the selected one or more of the identified at least one respective test result. In this example, the prompt 236 may request access data indicating the result-access credential to access the selected new results, which in this case are test results for “classic galactosemia,” “kabuki syndrome,” and “sickle cell disease.” As shown in Figure 3G, access data indicating the result-access credential for these new test results in the form of a textual access code may be provided to the prompt 236.

[058] In response to receiving and matching the access data, the interface 200 of Figure 3H may cause a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display as discussed above in connection to block 114 by providing full visual indications 250b, 252b, and 254b corresponding to test results for “classic galactosemia,” “kabuki syndrome,” and “sickle cell disease,” respectively, in addition to the full visual indication 234b corresponding to test results for “fragile x syndrome.” As shown, each of the full visual indications 250b, 252b, and 254b includes selectable “Summary” and “Report” icons for viewing a summary and a report on the selected one or more of the identified at least one respective test result, respectively. Also as shown in Figure 3H, the Results section 238 has been updated to indicate that three new results are available, and that no test results are pending.

[059] As shown, the “Summary” and “Report” icons for “classic galactosemia,” “kabuki syndrome,” and “sickle cell disease” are encircled by boxes indicating that the summary and report corresponding to each of these test results have not yet been displayed. Referring back to the method 100 of Figure 1, block 114 may in this instance further involve determining that the visual indication of the selected one or more of the identified at least one respective test result was not previously displayed before causing the visual indication of the selected one or more of the identified at least one respective test result to be provided on the display. Accordingly, the at least one visual indication corresponding to the identified at least one respective test result, when provided, may indicate that the visual indication of the selected one or more of the identified at least one respective test result was not previously displayed. In this case, the box encircling the “Summary” and “Report” icons may indicate that the corresponding results were not previously displayed.

[060] On the other hand, the “Summary” and “Report” icons for “fragile x syndrome,”

which have been previously displayed as shown in Figure 2K and 2L, are not encircled by a box, indicating that the summary and report corresponding to “fragile x syndrome” was previously displayed. Referring back to the method 100 of Figure 1, block 114 in this instance may further involve determining that the visual indication of the selected one or more of the identified at least one respective test result was previously displayed before causing the visual indication of the selected one or more of the identified at least one respective test result to be provided on the display. Accordingly, the at least one visual indication corresponding to the identified at least one respective test result, when provided, may indicate that the visual indication of the selected one or more of the identified at least one respective test result was previously displayed. In this case, the absence of a box encircling the “Summary” and “Report” icons may indicate that the corresponding results, in this case that of “fragile x syndrome” was previously displayed.

[061] As was the case with the full visual indication 234b, the full visual indications 250b, 252b, and 254b also include respective importance indicator indicating inferred degrees of impact of the corresponding test results on the well-being of the particular subject. In this case, the inferred degree of impact of the test result for “kabuki syndrome” is high, and the inferred degrees of impact of the test results for “classic galactosemia” and “sickle cell disease” are medium. Due to the high inferred degree of impact of the test result for “kabuki syndrome,” the “Summary” icon of the full visual indication 252b corresponding to the test result for “kabuki syndrome” may be selected. As shown in Figure 3I, a summary 256 of the test result for “kabuki syndrome” as applicable to the particular subject may be provided by the interface 200.

[062] Subsequent to displaying the summary 256 in Figure 3I, the interface 200 as shown in Figure 3J may provide full visual indications 234b, 250b, 252b, and 254b corresponding to test results for “classic galactosemia,” “sickle cell disease,” “kabuki syndrome,” and “fragile x syndrome” as previously shown in Figure 3H. In this case however, because the summary 256 for the “kabuki syndrome” test result has already been displayed, the “Summary” and “Report” icons of the full visual indication 252b corresponding to the “kabuki syndrome” test result may no longer be encircled by a box, indicating that summary and/or report corresponding to the “kabuki syndrome” test result was previously displayed. Also shown in Figure 3J is the Results section 238 updated to indicate that only two new results are now available.

[063] In some scenarios, a user accessing the test database may have previously selected a group of test results to be displayed before changing his/her mind and subsequently selecting

a different group of test results to be displayed. In other words, after receiving selection data and storing in the test database the selection-preference data, updated selection data indicating an updated selection of one or more of the identified at least one respective test result may be received via an updated selection of one or more of the at least one visual indication corresponding to the identified at least one test result, similar to that discussed above in connection to block 110.

[064] Before receiving the updated selection data, however, the interface 200 of Figure 3K may cause at least one visual indication corresponding to the identified at least one respective test result to be provided on the display as discussed above in connection to block 108 by providing the result-viewing selection box 222 as discussed previously. The result-view selection box 222 may be provided in response to the user selecting a “Return to Preferences Grid” icon of the interface 200 shown in Figure 3J, and subsequently selecting the “Genetic Syndromes” tile from the result categories grid 218. As shown in Figure 3K, updated selections may have been made to the selectable options indicating a selection of “No, I do not want results” for the “classic galactosemia” and “fragile x syndrome” test results, while the selection of “Yes, I want results” for the “kabuki syndrome” and “sickle cell disease” test results remains unchanged.

[065] Once these selections have been made, the “Save preferences” icon may be selected to cause the updated selection-preference data that indicates the updated selection of the one or more of the identified at least one respective test result may be stored in the test database similar to that described above in connection to block 110. In one case, the updated selection-preference data may override the previously stored selection-preference data.

[066] Upon receiving the updated selection data, if access data indicating a result-access credential for accessing any one of the updated selection of one or more of the identified at least one respective test result was not already received from the user, the user may be prompted to provide the necessary access data, as discussed above in connection to block 112. Upon entering the access data, or in the case access data for accessing the updated selection of one or more of the identified at least one respective test result was previously provided, a visual indication of the updated selection of one or more of the identified at least one respective test result may be provided on the display, as discussed above in connection to block 114.

[067] As shown, the interface 200 of Figure 3L provides a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display by providing the full visual indications 250b, 252b, and 234b corresponding to test results for

“sickle cell disease,” “kabuki syndrome,” and “fragile x syndrome,” respectively. In this case, because the stored updated selection-preference data indicates the selection of only test results for “kabuki syndrome” and “sickle cell disease,” the full visual indication 254b that corresponds to the “classic galactosemia” test result is not shown.

[068] On the other hand, the full visual indication 234b corresponding to the “fragile x syndrome” test result may still be shown because the summary 242 and report 244 for “fragile x syndrome” has already been displayed as shown in Figures 2K-L. In some embodiments, the full visual indication 234b corresponding to the “fragile x syndrome” test result may not be shown if the updated selection-preference data did not indicate a selection of test results for “fragile x syndrome,” whether or not the summary 242 or report 244 has previously been displayed. Other examples may also be possible. The interface 200 of Figure 3L also includes the Results Preferences section 216 indicating that two test results are selected and two test results are not selected. As shown, the Results section 238 may be updated to indicate that one new result out of the two selected, and in this case, three displayed test results are now available.

[069] As technology advances and further discoveries are made in the sequencing of the human genome and exome, additional information may be determined based on data acquired from previous tests. For instance, the raw genome or exome sequence data from a previous test may be re-processed and re-annotated to identify new, previously unavailable biological information represented within the genome or exome sequence data, producing updated annotated variant data.

[070] In connection to method 100 of Figure 1, updated test-result data indicating an updated result of the identified at least one respective test result may be received. As suggested above, the updated test-result data comprises updated annotated variant data generated based on the test performed on the particular subject. In this case, maintaining the test database as discussed in connection with block 102 of the method 100 may involve storing the updated test-result data in the test database.

[071] In one example, an updated result-access credential for accessing the updated result of the identified at least one respective test result may be generated in response to receiving from the laboratory updated test-result data at the test database. In another example, the same result-access credential generated for the original identified at least one respective test result may also be used to access the updated test result. Similarly, in one case, a new selection-preference data may be determined and stored for the updated result, while in another case the same selection-preference data stored for the original identified at least one respective test

results may be applied to the updated result. Other examples may also be possible.

[072] In some examples, because the updated result of the identified at least one respective test result may effectively be a new result that has not previously been displayed, the visual indication of the selected one or more of the identified at least one respective test result to be provided on the display may indicate that the updated result of the at least one test result is available. Referring back to Figure 3L for example, if an updated result corresponding to the “kabuki syndrome” test result is available, a box encircling the “Summary” and “Report” icons in the full visual indication 252b corresponding to the “kabuki syndrome” test result may re-appear. In some cases, to distinguish an updated result from a new result, the visual indication indicating the availability of the updated result may be different from the visual indication indicating a new result. For instance, the box encircling the “Summary” and “Report” icons in the full visual indication 252b corresponding to the “kabuki syndrome” test result may be of a different color depending on whether the available, not previously displayed test result is an updated test result, or a test result from a new test. Other examples may also be possible.

[073] As previously indicated, while the example embodiments described herein may relate particularly to returning patient genome or exome sequencing results, one having ordinary skill in the art will appreciate that the systems and methods disclosed in relation to the example embodiments may be implemented for a variety of different applications. The variety of different applications may include the return of other patient health and medical test results, or general academic or certification test results.

[074] As indicated above, in some embodiments, the disclosed methods may be implemented by computer program instructions encoded on a physical and/or non-transitory computer-readable storage media in a machine-readable format, or on other physical and/or non-transitory media or articles of manufacture. Figure 4 is a schematic illustrating a conceptual partial view of an example computer program product that includes a computer program for executing a computer process on a computing device, arranged according to at least some embodiments presented herein.

[075] In one embodiment, the example computer program product 400 may be provided using a signal bearing medium 402. The signal bearing medium 402 may include one or more programming instructions 404 that, when executed by one or more processors may provide functionality or portions of the functionality described with respect to Figure 1. In some examples, the signal bearing medium 402 may encompass a physical and/or non-transitory computer-readable medium 406, such as, but not limited to, a hard disk drive, a Compact

Disc (CD), a Digital Video Disk (DVD), a digital tape, memory, etc. In some implementations, the signal bearing medium 402 may encompass a computer recordable medium 408, such as, but not limited to, memory, read/write (R/W) CDs, R/W DVDs, etc. In some implementations, the signal bearing medium 402 may encompass a communications medium 410, such as, but not limited to, a digital and/or an analog communication medium (e.g., a fiber optic cable, a waveguide, a wired communications link, a wireless communication link, etc.). Thus, for example, the signal bearing medium 402 may be conveyed by a wireless form of the communications medium 410.

[076] The one or more programming instructions 404 may be, for example, computer executable and/or logic implemented instructions. In some examples, a processing unit of a computing device may be configured to provide various operations, functions, or actions in response to the programming instructions 404 conveyed to the processing unit by one or more of the computer readable medium 406, the computer recordable medium 408, and/or the communications medium 410.

[077] The physical and/or non-transitory computer readable medium could also be distributed among multiple data storage elements, which could be remotely located from each other. The computing device that executes some or all of the stored instructions could be a computing device such as any of those described above. Alternatively, the computing device that executes some or all of the stored instructions could be another computing device, such as a server.

[078] Figure 5 shows a simplified block diagram of an example communication network in which the method 100 described above can be implemented. It should be understood that this and other arrangements described herein are set forth only as examples. Those skilled in the art will appreciate that other arrangements and elements (e.g., machines, interfaces, functions, orders, and groupings of functions, etc.) can be used instead and that some elements may be omitted altogether. Further, many of the elements described herein are functional entities that may be implemented as discrete or distributed components or in conjunction with other components, and in any suitable combination and location. Various functions described herein as being performed by one or more entities may be carried out by hardware, firmware, and/or software. And various functions described herein may be carried out by a processor executing instructions stored in memory.

[079] As shown in Figure 5, example network 500 includes various network-access devices 502A-502D, public network 504 such as the Internet, and server 506. Note that additional entities not depicted in Figure 5 could be present as well. As an example, there

could be more network-access devices and more servers in communication with public network 504. Other network elements may be in communication with public network 504 as well. Also, there could be one or more devices and/or networks making up at least part of one or more of the communication links depicted in Figure 5. As an example, there could be one or more routers, switches, or other devices or networks on the communication links between network-access devices 502A-502D, public network 504, and/or server 506.

[080] Each of network-access devices 502A-502D may be any network-access device arranged to carry out the network-access device functions described herein. As such each of network-access devices 502A-502D, including network-access device 502A as shown in Figure 6, may include processor 602, data storage 604, and communication interface 610, all linked together via system bus, network, or other connection mechanism 612.

[081] Processor 602 may include one or more general purpose microprocessors and/or one or more dedicated signal processors and may be integrated in whole or in part with communication interface 610. Data storage 604 may include memory and/or other storage components, such as optical, magnetic, organic or other memory disc storage, which can be volatile and/or non-volatile, internal and/or external, and integrated in whole or in part with processor 602. Data storage 604 may be arranged to contain (i) program data 606 and (ii) program logic 608. Although these components are described herein as separate data storage elements, the elements could just as well be physically integrated together or distributed in various other ways. For example, program data 606 may be maintained in data storage 604 separate from program logic 608, for easy updating and reference by program logic 608.

[082] Communication interface 610 typically functions to communicatively couple network-access device 502A to networks, such as public network 504. As such, communication interface 610 may include a wired (e.g., Ethernet) and/or wireless (e.g., Wi-Fi) packet-data interface, for communicating with other devices, entities, and/or networks. Network-access device 502A may also include multiple interfaces 610, such as one through which network-access device 502A sends communication, and one through which network-access device 502A receives communication.

[083] Network-access device 502A may also include, or may be otherwise communicatively coupled to, user interface 620. User interface 620 may include input device 622 including, for example, buttons, a touch screen, a microphone, and/or any other elements for receiving inputs. User interface 620 may also include one or more elements for communicating outputs, for example, one or more graphical displays 624 and/or a speaker.

In operation, user interface 620 may be configured to display a graphical user interface (GUI) via graphical display 624 and may also be configured to receive inputs, via input device 622, corresponding to use of such a GUI.

[084] Server 506 may be any network server or other computing system arranged to carry out the server functions described herein including, but not limited to, those functions described with respect to Figure 1 described above. As such, as shown in Figure 7, server 506 may include processor 702, data storage 704 including program data 706 and program logic 708, and communication interface 710, all linked together via system bus, network, or other connection mechanism 712. Processor 702, data storage 704, program data 706, program logic 708, and communication interface 710 may be configured and/or arranged similar to processor 702, data storage 704, program data 706, program logic 708, and communication interface 710, respectively, as described above with respect to network-access device 502A.

[085] Data storage 704 may contain information used by server 506 in operation. For example, data storage 704 may include instructions executable by the processor for carrying out the server functions described herein including, but not limited to, those functions described above with respect to Figure 1. As another example, data storage 704 may contain various design logic and/or design data used for determining a test result, such as the logic and data described above with respect to Figure 1. Generally, data storage 704 may contain information used by server 506 to provide an e-commerce storefront that is accessible by various network-access devices, such as network-access device 502A, over public network 504.

[086] Returning to Figure 5, public network 504 may include one or more wide area networks, one or more local area networks, one or more public networks such as the Internet, one or more private networks, one or more wired networks, one or more wireless networks, and/or one or more networks of any other variety. Devices in communication with public network 504 (including, but not limited to, network-access devices 502A-502D and server 506) may exchange data using a packet-switched protocol such as IP, and may be identified by an address such as an IP address.

[087] While various aspects and embodiments have been disclosed herein, it should be understood that the embodiments are example embodiments and are described in connection to just some of many possible applications. Accordingly, it should also be understood that other aspects and embodiments are also possible. The various aspects and embodiments disclosed herein are for purposes of illustration and are not intended to be limiting, with the

true scope and spirit being indicated by the following claims.

CLAIMS

1. A computer implemented method comprising:
 - maintaining a test database comprising data that indicates (a) one or more subjects and (b) a plurality of test results, wherein each subject is associated with one or more respective test result from the plurality of test results;
 - receiving subject data that identifies a particular subject in the test database;
 - identifying at least one respective test result associated with the particular subject;
 - causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display;
 - receiving selection data, wherein the selection data indicates a selection of one or more of the identified at least one respective test result via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result;
 - receiving access data, wherein the access data indicates a result-access credential for accessing the selected one or more of the identified at least one respective test result; and
 - in response to receiving the access data, causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display.
2. The method of claim 1, further comprising:
 - in response to receiving the selection data, determining access requirements for the selected one or more of the identified at least one respective test result; and
 - causing a visual indication of a prompt to be provided on the display, wherein the prompt requests the access data indicating the result-access credential to access the selected one or more of the identified at least one respective test result.
3. The methods of claim 1 or 2, further comprising:
 - before receiving the subject data, receiving user-identification data that indicates a database-access credential for accessing the test database, wherein the user-identification data is associated with the subject data.
4. The method of claim 3, wherein identifying at least one respective test result associated with the particular subject comprises:
 - identifying the at least one respective test result based on at least the user-identification data.

5. The method of claims 3 or 4, further comprising:
storing in the test database selection-preference data that indicates the selection of one or more of the identified at least one respective test result, wherein the selection-preference data is associated with the user-identification data.
6. The method of claim 5, further comprising:
after storing in the test database the selection-preference data, receiving updated selection data, wherein the updated selection data indicates an updated selection of one or more of the identified at least one respective test result via an updated selection of one or more of the at least one visual indication corresponding to the identified at least one test result; and
storing in the test database updated selection-preference data that indicates the updated selection of the one or more of the identified at least one respective test result.
7. The method of any one of claims 1 – 6, further comprising:
before causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display, determining that the visual indication of the selected one or more of the identified at least one respective test result was not previously displayed,
wherein the at least one visual indication corresponding to the identified at least one respective test result indicates that the visual indication of the selected one or more of the identified at least one respective test result was not previously displayed.
8. The method of any one of claims 1 – 7, further comprising:
before causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display, determining that the visual indication of the selected one or more of the identified at least one respective test result was previously displayed,
wherein the at least one visual indication corresponding to the identified at least one respective test result indicates that the visual indication of the selected one or more of the identified at least one respective test result was previously displayed.

9. The method of any one of claims 1 – 8, wherein the at least one visual indication corresponding to the identified at least one respective test result indicates an inferred degree of impact of the identified at least one respective test result on a well-being of the particular subject.
10. The method of any one of claims 1 – 9, further comprising:
before causing the at least one visual indication corresponding to the identified at least one respective test result to be provided on a display, determining at least one result category associated with the identified at least one respective test result;
causing at least one visual indication corresponding to the at least one determined result category to be provided on the display; and
receiving category-selection data, wherein the category selection data indicates a selection of one or more of the at least one determined result category via a selection of one or more of the at least one visual indication corresponding to the at least one determined result category, and wherein the selected one or more of the at least one determined results category is associated with the identified at least one respective test result.
11. The method of any one of claims 1 – 10, further comprising:
before receiving the subject data, receiving test-result data indicating at least one of the one or more respective test result associated with the particular subject, wherein the received test-result data comprises (a) an identification of the particular subject and (b) annotated variant data generated based on a test performed on the particular subject; and
generating the result-access credential for accessing the at least one test result based on the identification of the particular subject,
wherein maintaining the test database comprises storing the received test-result data in the test database.
12. The method of claim 11, further comprising:
before causing the visual indication of the selected one or more of the identified at least one respective test result to be provided on the display, selecting the visual indication of the selected one or more of the identified at least one respective test result based on the annotated variant data in the test result data.
13. The method of any one of claims 1 – 12, further comprising:

before identifying at least one respective test result associated with the particular subject, receiving updated test-result data indicating an updated result of the identified at least one respective test result, wherein the updated test-result data comprises updated annotated variant data generated based on the test performed on the particular subject,

wherein maintaining the test database comprises storing the updated test-result data in the test database.

14. The method of claim 13, further comprising:

in response to receiving updated test-result data, generating an updated result-access credential for accessing the updated result of the identified at least one respective test result.

15. The method of claims 13 or 14, wherein the visual indication of the selected one or more of the identified at least one respective test result to be provided on the display indicates that the updated result of the at least one test result is available.

16. The method of any one of claims 1 – 15, wherein the one or more respective test result associated with the particular subject indicates one or more genetic conditions of the particular subject.

17. The method of claim 16, wherein the visual indication of the selected one or more of the identified at least one respective test result indicates one or more of the following: (a) a description of the genetic condition, (b) symptoms of the genetic condition, (c) health risks relating to the genetic condition, and (d) available options for managing the genetic condition.

18. A system comprising:

a processor;

a physical computer readable medium; and

program instructions stored on the physical computer readable medium and executable by the processor to:

maintain a test database comprising data that indicates (a) one or more subjects and (b) a plurality of test results, wherein each subject is associated with one or more respective test result from the plurality of test results;

receive subject data that identifies a particular subject in the test database;

identify at least one respective test result associated with the particular subject;
cause at least one visual indication corresponding to the identified at least one
respective test result to be provided on a display;

receive selection data, wherein the selection data indicates a selection of one
or more of the identified at least one respective test result via a selection of one or
more of the at least one visual indication corresponding to the identified at least one
test result;

receive access data, wherein the access data indicates a result-access credential
for accessing the selected one or more of the identified at least one respective test
result; and

in response to receiving the access data, cause a visual indication of the
selected one or more of the identified at least one respective test result to be provided
on the display.

19. The system of claim 18, the system further comprising program instructions stored on
the physical computer readable medium and executable by the processor to:

before causing the at least one visual indication corresponding to the identified at least
one respective test result to be provided on a display, determine at least one result category
associated with the identified at least one respective test result;

cause at least one visual indication corresponding to the at least one determined result
category to be provided on the display; and

receive category-selection data, wherein the category selection data indicates a
selection of one or more of the at least one determined result category via a selection of one
or more of the at least one visual indication corresponding to the at least one determined
result category, and wherein the selected one or more of the at least one determined results
category is associated with the identified at least one respective test result.

20. A physical computer readable medium having instructions stored thereon, the
instructions comprising:

instructions for maintaining a test database comprising data that indicates (a) one or
more subjects and (b) a plurality of test results, wherein each subject is associated with one or
more respective test result from the plurality of test results;

instructions for receiving subject data that identifies a particular subject in the test
database;

instructions for identifying at least one respective test result associated with the particular subject;

instructions for causing at least one visual indication corresponding to the identified at least one respective test result to be provided on a display;

instructions for receiving selection data, wherein the selection data indicates a selection of one or more of the identified at least one respective test result via a selection of one or more of the at least one visual indication corresponding to the identified at least one test result;

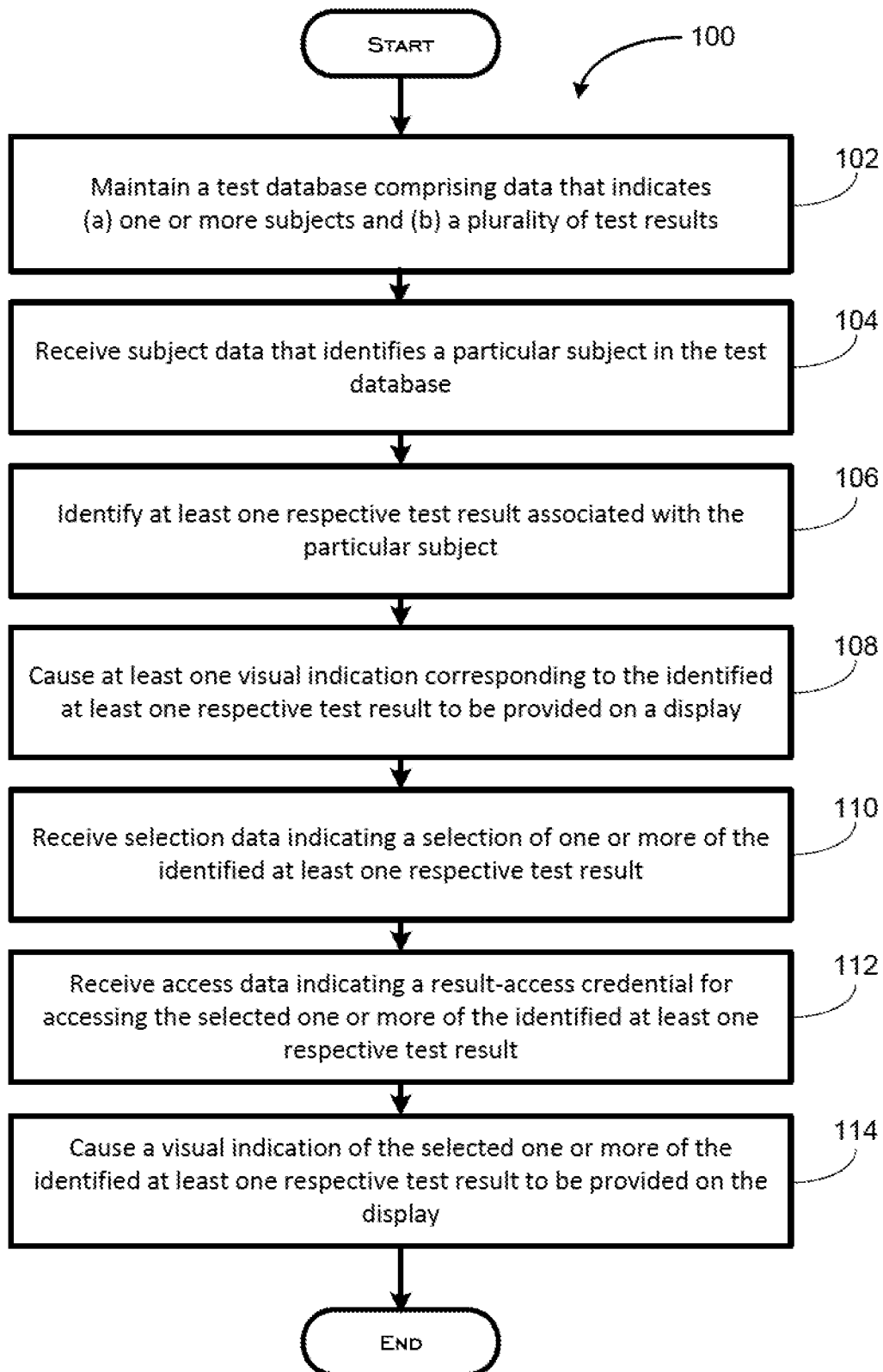
instructions for receiving access data, wherein the access data indicates a result-access credential for accessing the selected one or more of the identified at least one respective test result; and

instructions for causing a visual indication of the selected one or more of the identified at least one respective test result to be provided on the display in response to receiving the access data.

21. The physical computer readable medium of claim 20, wherein the instructions further comprise:

instructions for receiving user-identification data that indicates a database-access credential for accessing the test database before receiving the subject data, wherein the user-identification data is associated with the subject data; and

instructions for storing in the database selection-preference data that indicates the selection of one or more of the identified at least one respective test results, wherein the selection-preference data is associated with the user-identification data.

**FIGURE 1**

My46 Test Results Interface

My46 Username:
p1demo

Password:

SIGN IN

FIGURE 2B

My46 Test Results Interface

Create Account

First Name: Patent

Last Name: 1-demo

Email Address: 1demo@1demo.com

Username: p1demo

Password: *****

Re-enter Password: *****

Your My46ID: My46Patent0001

SUBMIT

FIGURE 2A

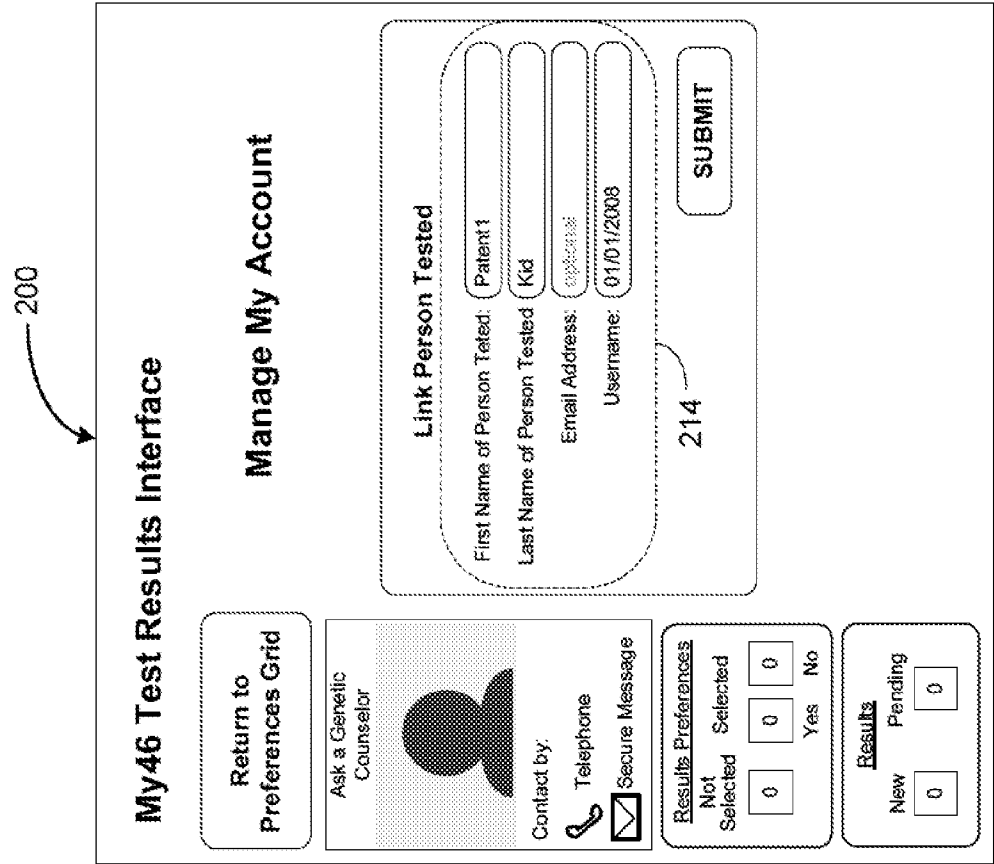


FIGURE 2D

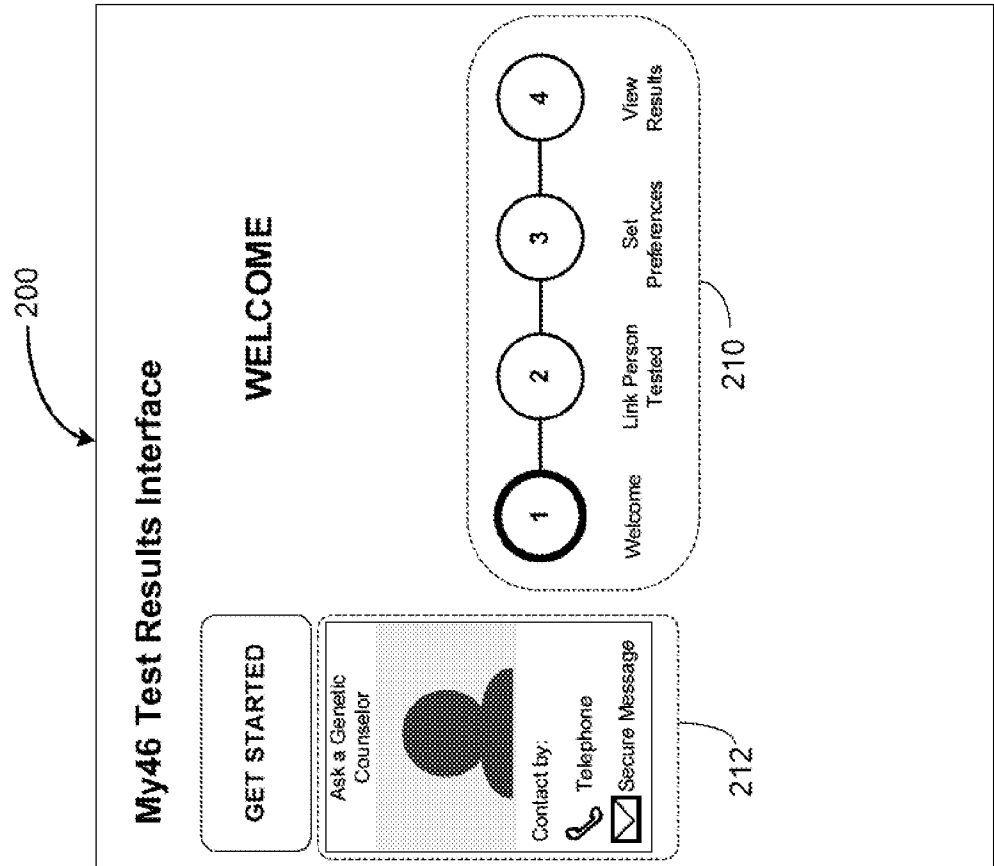


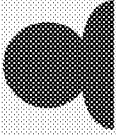
FIGURE 2C

200

My46 Test Results Interface

Go to Results Navigator

Ask a Genetic Counselor



Contact by:
☒ Telephone
☐ Secure Message

Preferences Grid
 Linked to Person Tested: Patient1 Kid

Genetic Syndromes	Metabolic Disorders	Disease Risk
Medication Response	Carrier Status	Newborn Screening Conditions
Ancestry	Prenatal Testing	Copy Number Variants

218

Select All

Results Preferences

Not Selected	Selected	Yes	No
☒ 1	☐ 0	☐ 0	☐ 0

Results

New	Pending
☐ 0	☐ 0

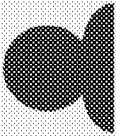
FIGURE 2F

200

My46 Test Results Interface

Return to Preferences Grid

Ask a Genetic Counselor



Contact by:
☒ Telephone
☐ Secure Message

Set Preferences

View Results

Results Preferences

Not Selected	Selected	Yes	No
☒ 1	☐ 0	☐ 0	☐ 0

216

Results

New	Pending
☐ 0	☐ 0

FIGURE 2E

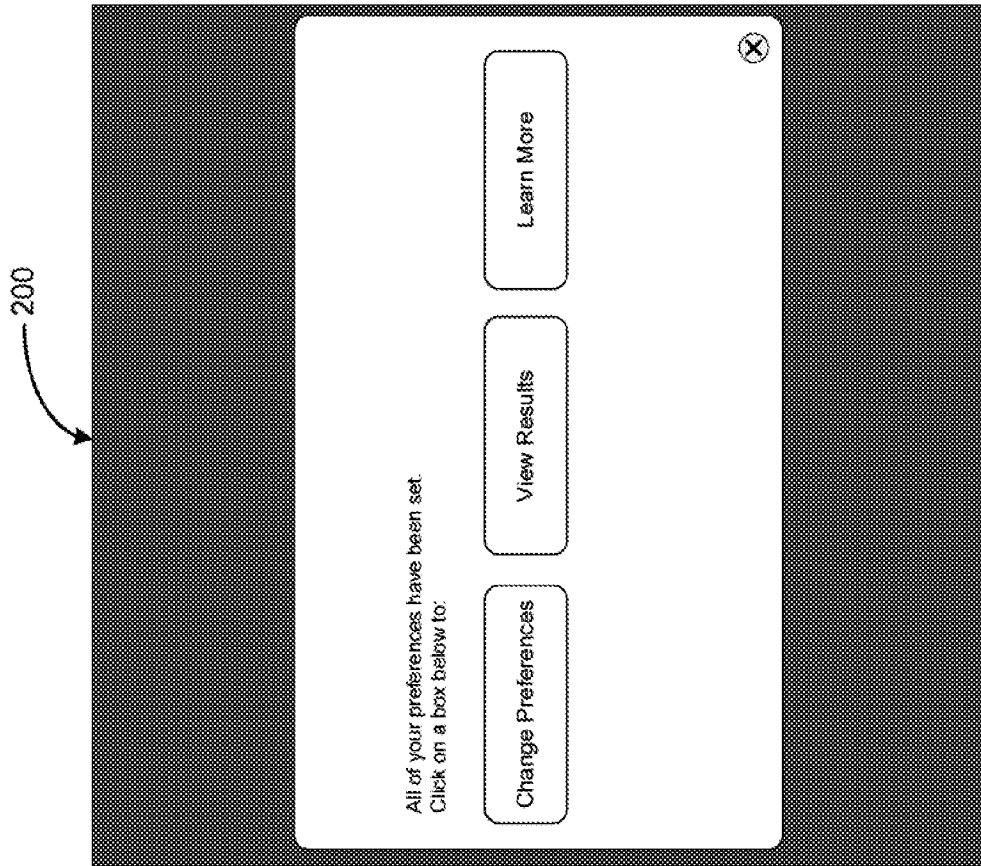


FIGURE 2H

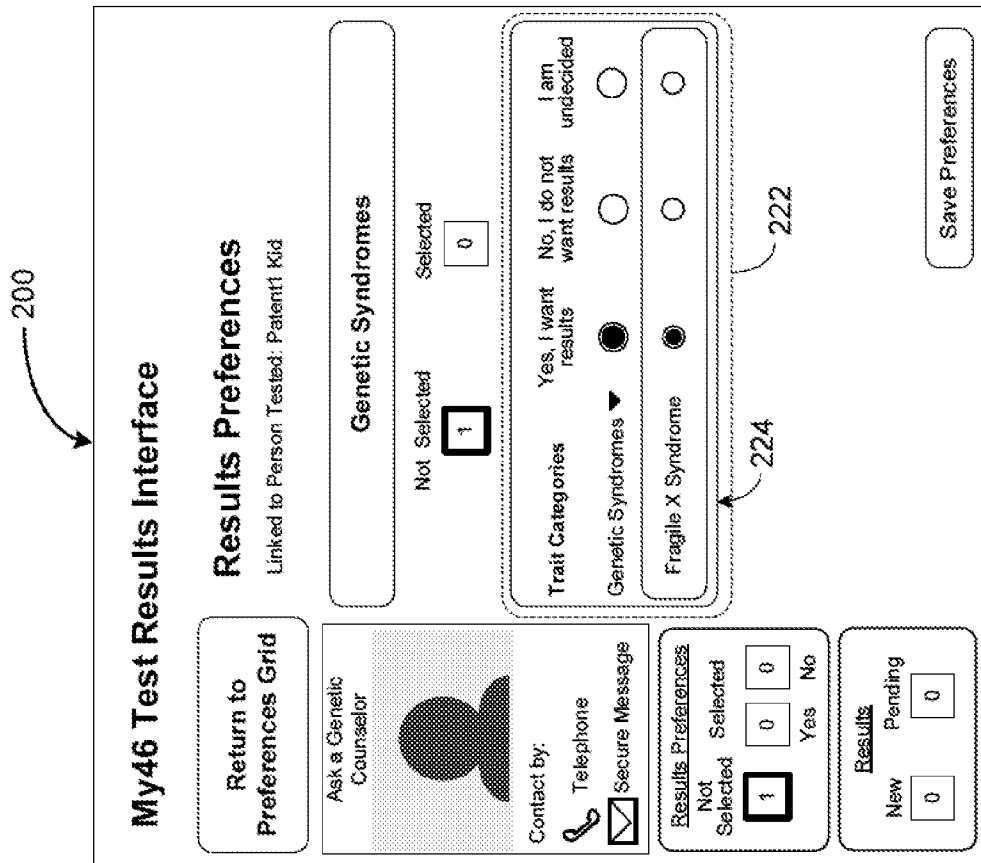


FIGURE 2G

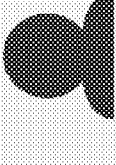
200

My46 Test Results Interface

Return to Preferences Grid

Results Navigator
 Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:
 SUBMIT

Ask a Genetic Counselor


Contact by:
☒ Telephone
☐ Secure Message

Results Preferences
 Not Selected: 0
 Selected: 1
 Yes: 0
 No: 0

Results
 New: 1
 Pending: 0

Trait Profile
 Category
 Summary
 Report
 Priority

Fragile X Syndrome
 Genetic Syndromes
 Report
 L

FIGURE 2J

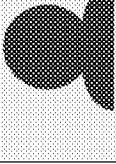
200

My46 Test Results Interface

Return to Preferences Grid

Results Navigator
 Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:
 SCHFRAX **SUBMIT**

Ask a Genetic Counselor


Contact by:
☒ Telephone
☐ Secure Message

Results Preferences
 Not Selected: 0
 Selected: 1
 Yes: 0
 No: 0

Results
 New: 0
 Pending: 1

Trait Profile
 Category
 Summary
 Report
 Priority

Fragile X Syndrome
 Genetic Syndromes
 Report

FIGURE 2I

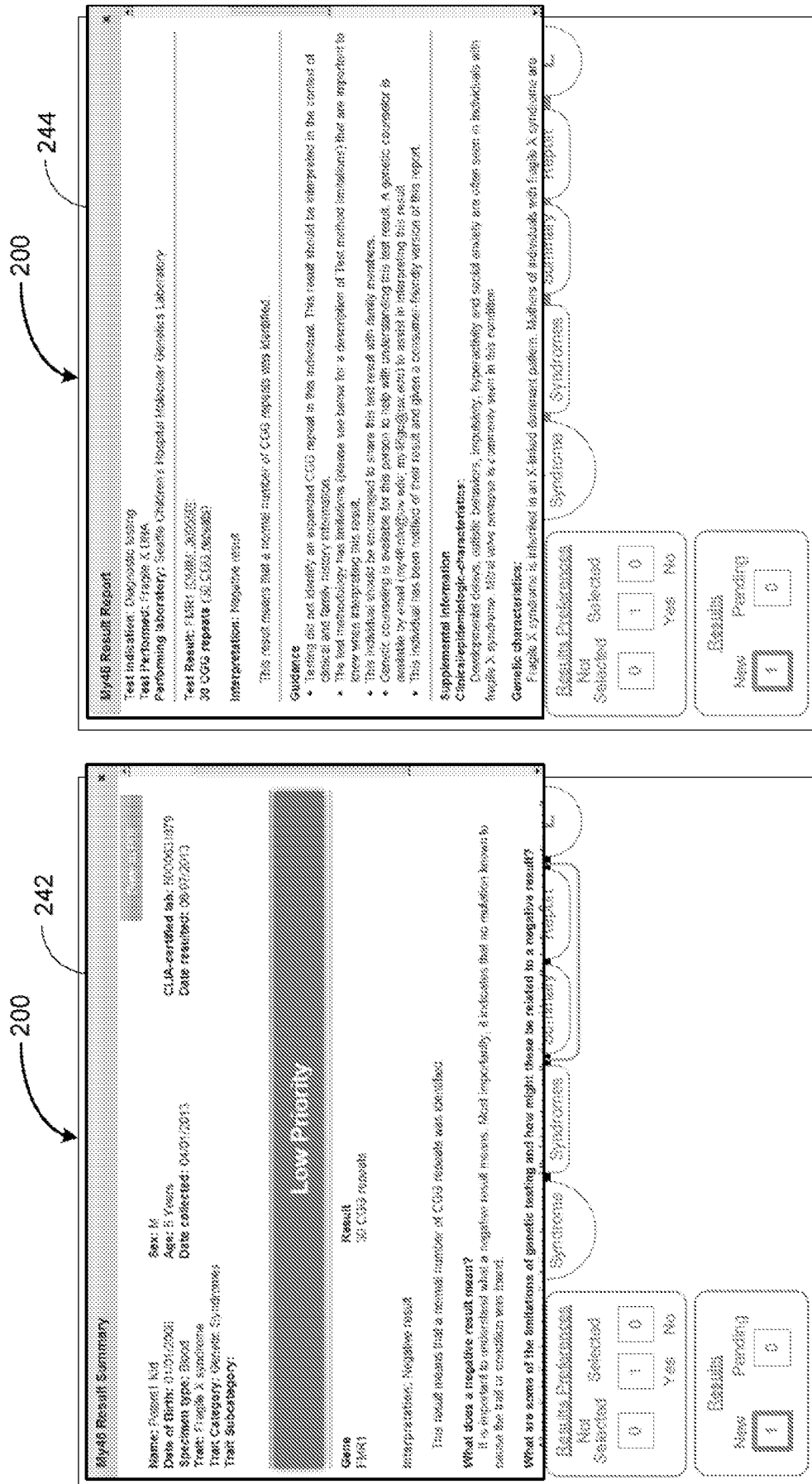


FIGURE 2L

FIGURE 2K

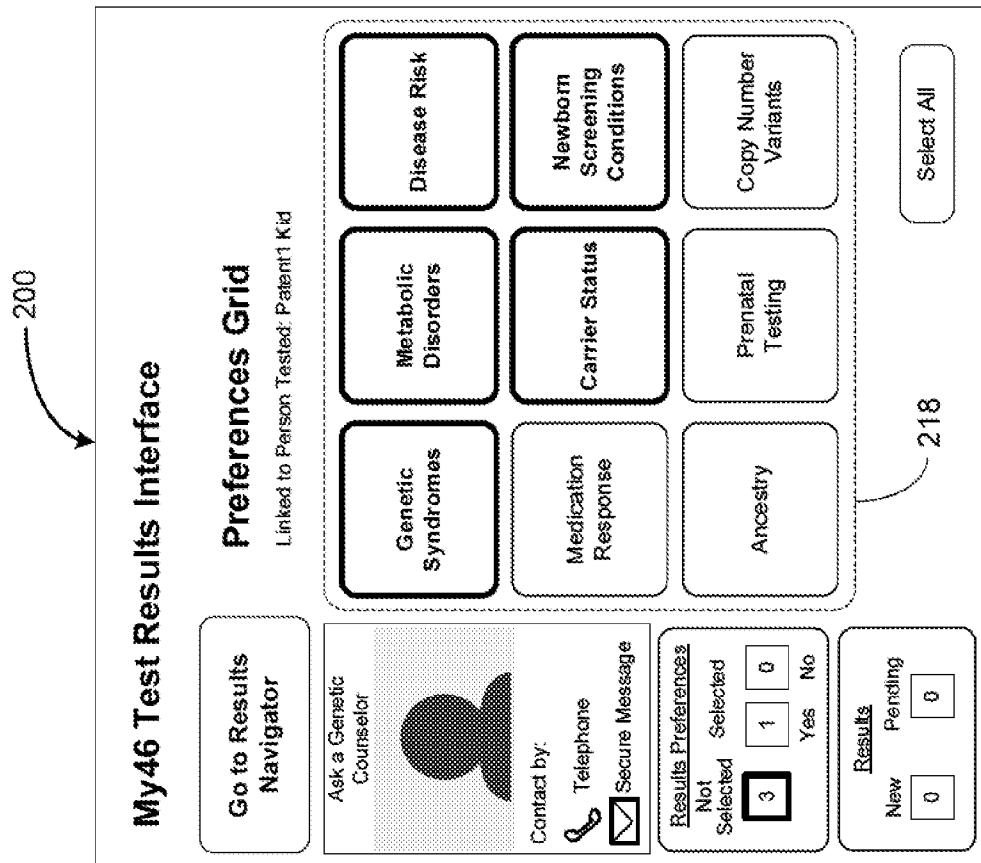


FIGURE 3B

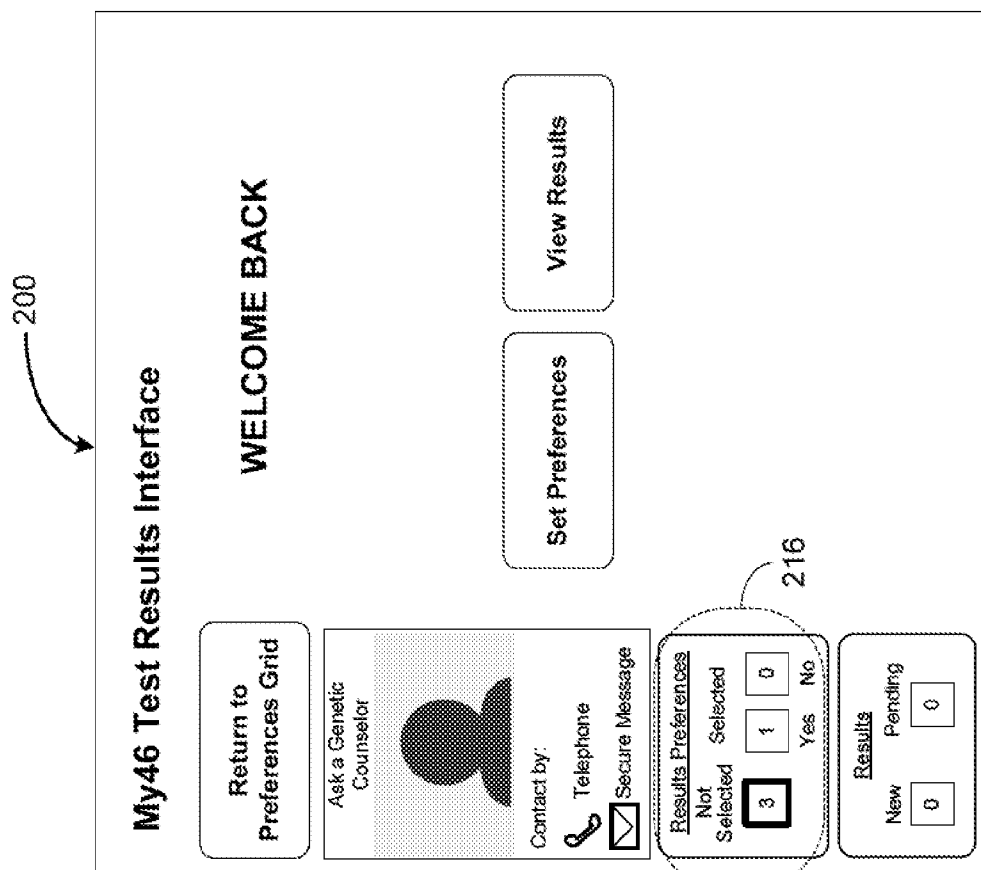


FIGURE 3A

200

My46 Test Results Interface

Go to Results Navigator

Preferences Grid
Linked to Person Tested: Patient1 Kid

Ask a Genetic Counselor

Contact by:
Telephone
☒ Secure Message

Results Preferences

Not Selected	Selected	Yes	No
☒	☐	☐	☐

Results

New	Pending
☐	☒

216

218

238

Select All

Genetic Syndromes

Metabolic Disorders

Disease Risk

Medication Response

Newborn Screening Conditions

Ancestry

Prenatal Testing

Copy Number Variants

FIGURE 3D

200

My46 Test Results Interface

Return to Preferences Grid

Results Preferences
Linked to Person Tested: Patient1 Kid

Ask a Genetic Counselor

Contact by:
Telephone
☒ Secure Message

Disease Risk

Not Selected ☒ Selected ☐

Trait Categories

Blood ☒ Yes, I want results ☐ No, I do not want results ☐ I am undecided

Sickle Cell Disease ☒ Yes ☐ No

Digestive System ☐ Yes ☐ No

Heart and Lungs ☐ Yes ☐ No

Nervous System ☐ Yes ☐ No

Results Preferences

Not Selected	Selected	Yes	No
☒	☐	☐	☐

Results

New	Pending
☐	☐

246

222

Save Preferences

FIGURE 3C

200

My46 Test Results Interface

Go to Results Navigator

Preferences Grid

Linked to Person Tested: Patient1 Kid

Ask a Genetic Counselor


Contact by:
Telephone
Secure Message

Results Preferences	
Not Selected	Selected
0	4
Yes	No

Results	
New	Pending
0	3

Disease Risk
Metabolic Disorders
Genetic Syndromes

Newborn Screening Conditions
Carrier Status
Medication Response

Copy Number Variants
Prenatal Testing
Ancestry

Select All

FIGURE 3F

200

My46 Test Results Interface

Return to Preferences Grid

Results Preferences

Linked to Person Tested: Patient1 Kid

Ask a Genetic Counselor


Contact by:
Telephone
Secure Message

Results Preferences	
Not Selected	Selected
2	2
Yes	No

Genetic Syndromes		
Not Selected	Selected	
2	2	

Trait Categories	Yes, I want results	No, I do not want results	I am undecided
Genetic Syndromes ▼	☐	☐	☐
Classic Galactosemia	☒	☐	☐
Fragile X Syndrome	☒	☐	☐
Kabuki Syndrome	☒	☐	☐
Sickle Cell Disease	☒	☐	☐

222
248

Save Preferences

FIGURE 3E

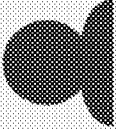
200

Results Navigator

Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:

Ask a Genetic Counselor



Contact by:

Telephone ☐

Secure Message ☒

Results Preferences

Not Selected	Selected	Yes	No
0	4	0	0

Results

New	Pending
3	0

238

FIGURE 3H

200

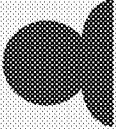
Results Navigator

Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:

236

Ask a Genetic Counselor



Contact by:

Telephone ☐

Secure Message ☒

Results Preferences

Not Selected	Selected	Yes	No
0	4	0	0

Results

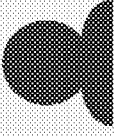
New	Pending
0	3

FIGURE 3G

200

Return to Preferences Grid

Ask a Genetic Counselor



Contact by:

Telephone

☒ Secure Message

Results Preferences

Not Selected

Selected

0

4

Yes

No

Results

New

Pending

2

0

Results Navigator

Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:

SUBMIT

Trait Profile

Category

Summary

Report

Priority

250b

Classic Galacto -semia

Genetic Syndromes

Summary

Report

M

254b

Sickle Cell Disease

Disease Risk

Summary

Report

M

252b

Kabuki Syndrom

Genetic Syndromes

Summary

Report

H

234b

Fragile X Syndrome

Genetic Syndromes

Summary

Report

L

FIGURE 3J

200

256

My MS Result Summary

Name: Patient1 Kid

Date of Birth: 01/01/2008

Specimen Type: Blood

Trait: Kabuki syndrome

Trait Category: Genetic Syndromes

Trait Subcategory:

Sex: M

Age: 5 Years

Data collected: 06/01/2013

CLIA-certified lab: 4300260200

Date received: 05/16/2013

High Priority

Gene

Mutation

Result

KMT2D

KMT2D:3 c.1058C>T

Unknown deleterious

Interpretation: This individual has a mutation in the KMT2D gene. This mutation changes the amino acid, R (arginine), to a stop codon, X, at position 2198. This result is suggestive of being affected with Kabuki syndrome.

What is the KMT2D gene?

The KMT2D gene makes a histone methyltransferase enzyme. This enzyme, along with other histone methyltransferases, are responsible for controlling the activity of certain genes. Mutations in the KMT2D gene may interfere with the enzyme's normal function and the ability to interact with certain genes that are important for normal development.

0

4

Yes

No

Results

New

Pending

3

0

Classic Galacto -semia

Disease Risk

Genetic Syndromes

Sickle Cell Disease

Disease Risk

Genetic Syndromes

Fragile X Syndrome

Genetic Syndromes

Genetic Syndromes

FIGURE 3I

200

My46 Test Results Interface

Return to Preferences Grid

Results Navigator
 Linked to Person Tested: Patient1 Kid

Please Enter Accession Number:

Trait Profile Category Summary Report Priority

Contact by:
☒ Telephone ☐ Secure Message

Ask a Genetic Counselor

Results Preferences
 Not Selected 0 Selected 2 Yes No
 Pending 0

Results
 New 1 Pending 0

254b
 Sickle Cell Disease Disease Risk Summary Report M

252b
 Kabuki Syndrome Genetic Syndromes Summary Report H

234b
 Fragile X Syndrome Genetic Syndromes Summary Report L

FIGURE 3L

200

My46 Test Results Interface

Return to Preferences Grid

Results Preferences
 Linked to Person Tested: Patient1 Kid

Not Selected 0 Selected 4

Trait Categories Yes, I want results No, I do not want results I am undecided

Genetic Syndromes ▼

Classic Galactosemia ☐ ☒ ☐

Fragile X Syndrome ☐ ☒ ☐

Kabuki Syndrome ☒ ☐ ☐

Sickle Cell Disease ☒ ☐ ☐

Save Preferences

Contact by:
☒ Telephone ☐ Secure Message

Ask a Genetic Counselor

Results Preferences
 Not Selected 0 Selected 4 Yes No
 Pending 0

Results
 New 2 Pending 0

FIGURE 3K

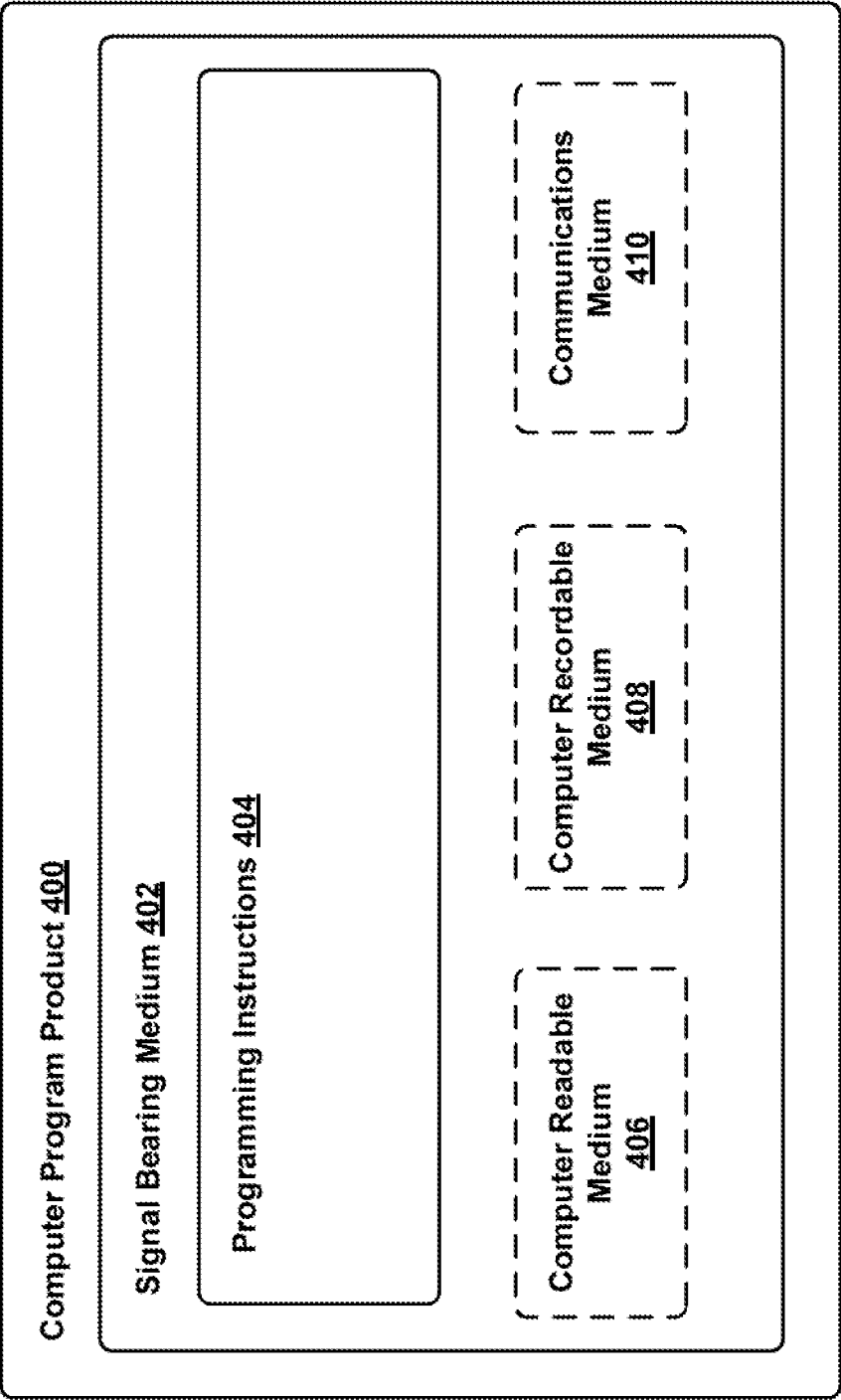


FIGURE 4

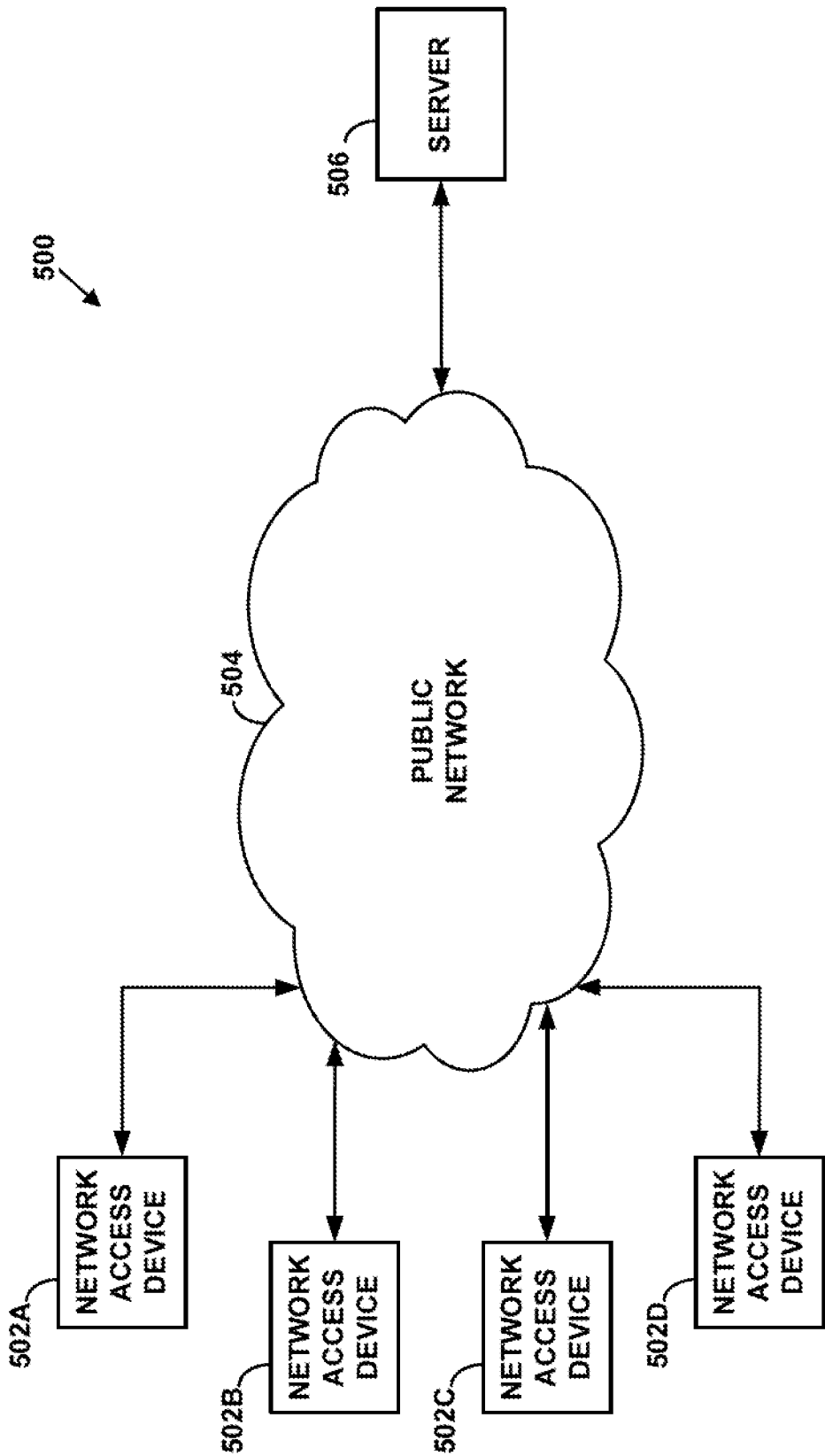


FIGURE 5

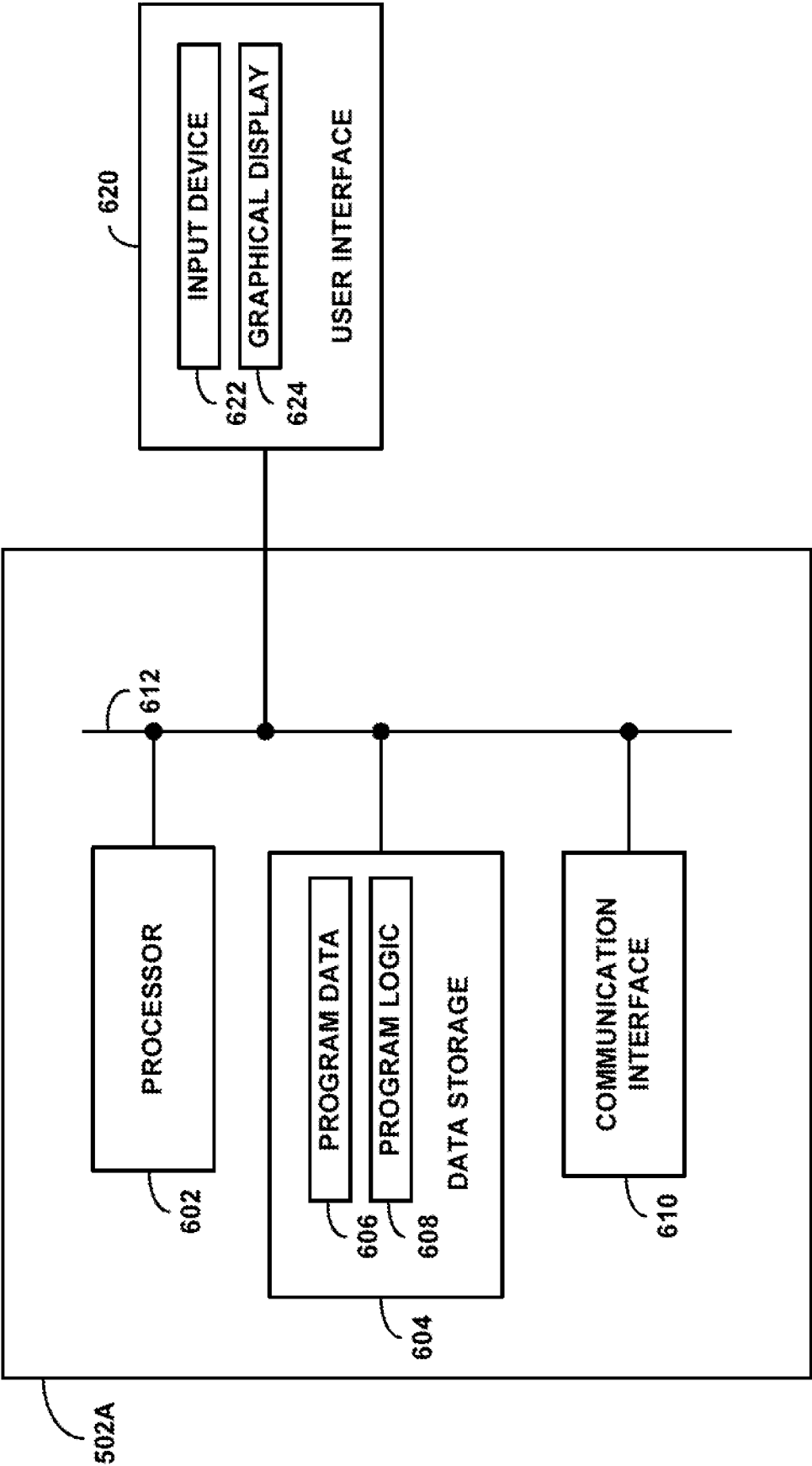


FIGURE 6

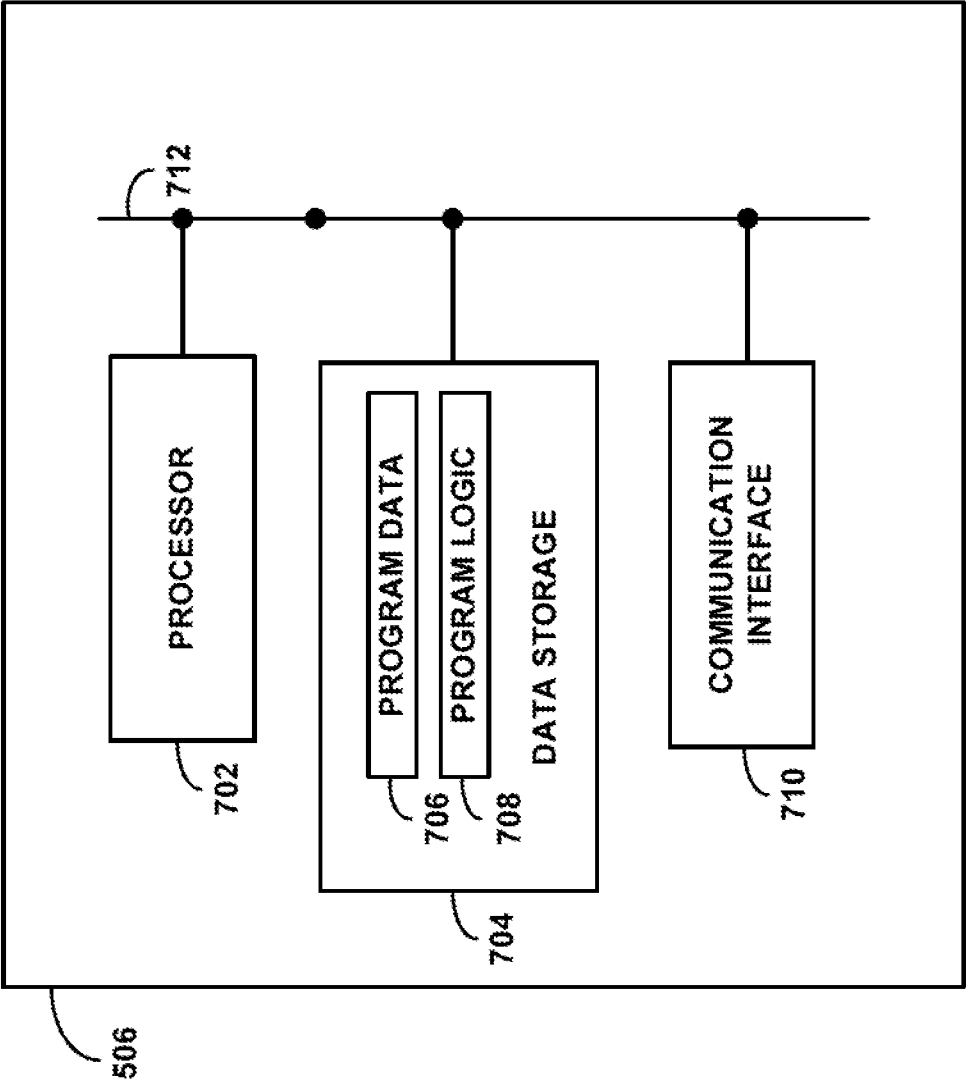


FIGURE 7

INTERNATIONAL SEARCH REPORT

International application No.
PCT/US2013/056445**A. CLASSIFICATION OF SUBJECT MATTER****G06F 19/00(2011.01)i, G06F 3/14(2006.01)i, G06F 3/048(2006.01)i**

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

G06F 19/00; H04K 1/00; G06Q 50/00; H04L 9/00; A61F 5/00; G06Q 10/00; G06F 3/14; G06F 3/048

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Korean utility models and applications for utility models

Japanese utility models and applications for utility models

Electronic data base consulted during the international search (name of data base and, where practicable, search terms used)

eKOMPASS(KIPO internal) & Keywords: biological, test result, authorization, display, database

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
Y	US 2011-0035237 A1 (SHUNSUKE ARIYOSHI et al.) 10 February 2011 See paragraphs [0063]-[0067], [0073]-[0076], [0086]-[0089], [0096]-[0099], [0120]-[0123]; figures 1, 7-9, 11; and claims 1, 15.	1-4, 18-21
Y	US 7237117 B2 (KENNETH P. WEISS) 26 June 2007 See column 6, lines 22-38, column 9, lines 65-67, column 10, lines 1-27; and figures 1, 5.	1-4, 18-21
Y	US 8112294 B2 (DAVID P. MCCALLIE, JR. et al.) 07 February 2012 See column 4, lines 64-67, column 5, lines 1-22, column 7, lines 34-67, column 8, lines 1-4, column 10, lines 35-60; and figures 1, 4.	19, 21
A		1-4, 18, 20
A	US 7734482 B1 (EARL D. VANCE et al.) 08 June 2010 See column 5, lines 63-67, column 6, lines 1-67; and figures 16-17.	1-4, 18-21
A	US 2011-0112865 A1 (ALAN J. YING et al.) 12 May 2011 See paragraphs [0063]-[0072]; and figures 4, 8.	1-4, 18-21

☐ Further documents are listed in the continuation of Box C.☒ See patent family annex.

* Special categories of cited documents:

"A" document defining the general state of the art which is not considered to be of particular relevance

"E" earlier application or patent but published on or after the international filing date

"L" document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)

"O" document referring to an oral disclosure, use, exhibition or other means

"P" document published prior to the international filing date but later than the priority date claimed

"T" later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention

"X" document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone

"Y" document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art

"&" document member of the same patent family

Date of the actual completion of the international search

05 December 2013 (05.12.2013)

Date of mailing of the international search report

05 December 2013 (05.12.2013)

Name and mailing address of the ISA/KR

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Authorized officer

YANG, Jeong Rok

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INTERNATIONAL SEARCH REPORTInternational application No.
PCT/US2013/056445**Box No. II Observations where certain claims were found unsearchable (Continuation of item 2 of first sheet)**

This international search report has not been established in respect of certain claims under Article 17(2)(a) for the following reasons:

1. ☐ Claims Nos.:
because they relate to subject matter not required to be searched by this Authority, namely:
2. ☒ Claims Nos.: 6, 12, 14, 17
because they relate to parts of the international application that do not comply with the prescribed requirements to such an extent that no meaningful international search can be carried out, specifically:
Claims 6, 12, 14 and 17 refer to multiple dependent claims that refer to other multiple dependent claims. Therefore, the meaning of the technical feature to which they refer is vague and unclear (PCT Article 6).
3. ☒ Claims Nos.: 5, 7-11, 13, 15-16
because they are dependent claims and are not drafted in accordance with the second and third sentences of Rule 6.4(a).

Box No. III Observations where unity of invention is lacking (Continuation of item 3 of first sheet)

This International Searching Authority found multiple inventions in this international application, as follows:

1. ☐ As all required additional search fees were timely paid by the applicant, this international search report covers all searchable claims.
2. ☐ As all searchable claims could be searched without effort justifying an additional fees, this Authority did not invite payment of any additional fees.
3. ☐ As only some of the required additional search fees were timely paid by the applicant, this international search report covers only those claims for which fees were paid, specifically claims Nos.:
4. ☐ No required additional search fees were timely paid by the applicant. Consequently, this international search report is restricted to the invention first mentioned in the claims; it is covered by claims Nos.:

Remark on Protest

- ☐ The additional search fees were accompanied by the applicant's protest and, where applicable, the payment of a protest fee.
- ☐ The additional search fees were accompanied by the applicant's protest but the applicable protest fee was not paid within the time limit specified in the invitation.
- ☐ No protest accompanied the payment of additional search fees.

INTERNATIONAL SEARCH REPORT

Information on patent family members

International application No.

PCT/US2013/056445

Patent document cited in search report	Publication date	Patent family member(s)	Publication date
US 2011-0035237 A1	10/02/2011	JP 2011-033536 A US 8209196 B2	17/02/2011 26/06/2012
US 7237117 B2	26/06/2007	US 2002-0178364 A1 US 2008-0005576 A1	28/11/2002 03/01/2008
US 8112294 B2	07/02/2012	US 2006-143043 A1 US 2011-225003 A1 US 7953608 B2	29/06/2006 15/09/2011 31/05/2011
US 7734482 B1	08/06/2010	None	
US 2011-0112865 A1	12/05/2011	CA 2434714 A1 US 2005-0065822 A1 US 7831449 B2 US 8548825 B2 WO 02-063541 A2 WO 02-063541 A3	15/08/2002 24/03/2005 09/11/2010 01/10/2013 15/08/2002 13/11/2003