Oracle® Healthcare Translational Research User's Guide





Oracle Healthcare Translational Research User's Guide, Release 3.4

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Preface

This preface contains the following sections:

- Documentation accessibility
- Access to Oracle Support

Documentation accessibility

For information about Oracle's commitment to accessibility, visit the Oracle Accessibility Program website at http://www.oracle.com/pls/topic/lookup?ctx=acc&id=docacc.

Access to Oracle Support

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1

What can I do with Oracle Healthcare Translational Research?

Oracle Healthcare Translational Research enables integrating, controlling, and analyzing the clinical and omics data required to support the complete biomarker life cycle. This includes data acquisition, discovery, and research as well as clinical use of patient and specimen information and cohort maintenance in clinical trials.

Use Oracle Healthcare Translational Research (OHTR) to:

- Select cohorts of patients or subjects using 10 clinical dimensions and 4 types of omics data type criteria.
- Save cohort queries and lists and share them with colleagues.
- View complete clinical and genomic information for a single patient or subject.
- Use the built-in Visquick genome browser to view different types of genomic results in a circular plot.
- Search genomic data based on different genomic features like genes, genomic location, gene components, variants and so on.
- View information on patient or subject specimens linked to genes and variants.
- View and export specific clinical information for a selected cohort.
- View clinical activities on a visual timeline.
- View built-in cohort-driven reports on:
 - Demographic statistics
 - Clinical statistics
 - Genomic variants, copy number variation, gene expression, and structural variants
 - Genes compared to a sample matrix
 - Variants compared to a sample matrix
- Export various types of genomic data in VCF, SEG, GCT, and RES formats in synchronous and asynchronous mode.
- Schedule jobs for genomic data export.
- Drill in from one screen to another to further drill down and generate various reports.

For more information, see:

Regulatory Compliance



Regulatory Compliance

Oracle Healthcare Translational Research is developed with HIPAA regulations in mind. It includes obfuscation rules to protect any patient-identifiable information. Oracle Healthcare Translational Research development also follows the software development guidelines and requirements for FDA 21 CFR Part 11 compliant software.

The origin of any data stored in CDM must be traceable to its source, and all transformations applied to the data must be accessible.

The standard Oracle ADF logger is applied for logging, and by default, it is written in WebLogic diagnostic logs. Logging focuses on the following areas:

- SQL queries to retrieve cohort count or list and event data These are constructed dynamically based on user input and may need to be tracked for debugging and diagnostic purposes.
- DML operation of saved queries Unlike other parts of the UI that handle readonly data, saving queries writes data into database tables, and the operations are logged.



2

Modify criteria for queries using the Cohort Query

In this chapter you can find out how to:

- Before you begin building your query
- Build a cohort query
- · Combine inclusion and exclusion criteria
- View Patient or Subject Count and data
- View Specimen Count
- Save a query
- Load a saved query

Before you begin building your query

Before you start creating your query, let's go through these tips. Keep this section in mind, you will probably come back to it from time to time.

- Prepare the query definition in a structured format (line-by-line) before creating it in Oracle Healthcare Translational Research. Visualizing your definition helps you organize it in a simpler way.
- When you want to search for a criterion, but can't exactly remember the name or the code, just click **Search** without selecting any conditions, or adding a keyword. The entire list of codes and names appears.
- Every criterion you want to add in your query can be included as an inclusion or exclusion. When you select either option, it applies to all of the elements in that criterion.
- By default, most dialog boxes for choosing criteria use the **Equals** operator and list the first 100 types only. If there are more than 100 types for one criterion that you want to include in your query, you can search for them using other operators such as **Starts with**, **Contains**, and so on.

Build a cohort query

You can query on any combination of the following data:

- Patient Information
- Clinical Data
- Genomic Data

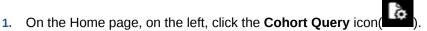


Patient Information

In this section;

- Demographics
- Consent

Demographics



- 2. In the **Cohort Query** screen, on the left, click the **Patient Information** category, then click **Demographics**.
- 3. Either enter the Patient ID or click the search icon ($^{\square}$).

To search for a **Patient ID**:

- a. Select a condition and enter part of a patient ID.
- b. Click Search.
- c. Select the patient ID that you want to use from the left side and move it to the right side using the arrow ().
- d. Click Submit.
- 4. Set any other criteria that you want to use in your query:
 - Gender: Select the gender from the drop-down list.
 - Marital Status: Select the status from the drop-down list.
 - Age: Select a condition, and then add the age in years, as a single value or a range.
 - Date of Birth: Select condition, and then enter the date.
 - Deceased: Select this check box, if needed.
 - Last Contact Date: Select a condition, and then add the date as a single value or a range.
 - Ethnicity: Select the appropriate ethnicity.
 - If in the Subject context, specify Age at Study Start. Select a condition, then add the Age as a single value or a range and select the unit of measure of the age, that is, Year(s), Month(s), Week(s), or Day(s). To match the Subjects based on this criteria, Cohort Query uses the Cohort Data Mart's database column Age At Study Start in Days (W_EHA_SUBJECT_D.AGE_AT_STUDY_START_DAYS or AGE_AT_STUDY_START_DAYS_OBF, depending on the data access privileges of the logged in user). The value in the database column Age At Study Start in Days is automatically populated based on the values populated in the columns Age at Study Start (AGE_AT_STUDY_START) and Age at Study Start Unit Of Measure (AGE_AT_STUDY_START_UOM_WID) using the following calculations and the same calculations are also used for parsing the search criteria defined for Age at Study Start.



If value of the database column Age at Study Start Unit Of Measure matches the parameterized value for:	Value of the database column Age At Study Start in Days:
Day	= Age at Study Start
Week	= Age at Study Start X 7
Month	= Age at Study Start X 365/12
Year	= Age at Study Start X 365
Any other value	=Null

Refer to the Oracle Healthcare Foundation Administrator's Guide for information on the Cohort Data Mart ETL's configurable parameters related to automatic population of the database column Age at Study Start in Days.

- Race: Select the appropriate race.
- Location: Enter either a city, state, zip code, county, or country.
- Data Source: Enter or search for a Data Source.
- Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- Click Submit.

Consent

- 1. On the left, click the **Patient Information** category, then click **Consent**.
- 2. Enter a **Consent Type** or click the search icon ($^{\square}$).

To search for a **Consent Type**:



Follow the same steps for the Consent Status.

- a. In the Select Consent Type window, select a condition for Consent Type
 Name or Consent Type Code and enter part of the consent name or type.
- b. Click Search.
- c. Select the consent type or code that you want to use from the left side and move it to the right side using the arrow ().
- d. Click Submit.
- 3. Set any other criteria that you want to use in your query:
 - Consent Code: Enter or search for a Consent Code.
 - Consent Status: Enter a consent status, or follow steps from 2a to 2d, to search for a consent status.
 - Consent Start Date: Select a condition, and then add the date.



- Consent End Date: Select a condition, and then add the date.
- Data Source: Enter or search for a Data Source.



To define search criteria for Consent, the criteria must include at least one of the following attributes: Consent Type or Consent Code.

- 4. Under Insert As select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 5. Click Submit.

For more information, visit:

"Build a cohort query"

"Before you begin building your query"

Clinical Data

In this section:

- Diagnosis
- Clinical Encounter
- Procedure
- Medication
- Patient History
- Test or Observation
- Specimen
- Study
- Relative Time Events

Diagnosis

- 1. On the left, click the **Clinical Data** category, then click **Diagnosis**.
- 2. Either enter the diagnosis or click the search icon ($^{\square}$).

To search for a **Diagnosis**:

- a. Select one of the **Search Modes**:
 - Classic Search. Select it to use more search conditions for Diagnosis Name, Code, and Code System.
 - Hierarchy Drill-in Search. Select it to search all of the codes in a hierarchy that are associated with a single Code System, and have only one parent code.



This feature does not support mixed hierarchy, meaning different levels of codes belonging to different Code Systems or multi-parent hierarchy, meaning one code having multiple parent codes.

- Select a condition for one or all: Diagnosis Name, Diagnosis Code, Code
 System ID or Code System Name.
- c. Click Search.
- d. elect the diagnosis codes that you want to use from the left side and move them to the right side using the arrow ().
- e. Click Submit.
- 3. Set any other criteria that you want to use in your query:
 - Diagnosis Status: Enter or search for a Diagnosis Status.
 - Diagnosis Severity: Enter or search for a Diagnosis Severity.
 - Onset Date: Select a condition, and enter a date.
 - Reported Date: Select a condition, and enter a date.
 - End Date: Select a condition, and enter a date.
 - Age At First Onset: Select a condition, and then add the age in years, as a single value or a range.
 - Anatomical Site: Enter or search for an anatomical site.
 See Sequence Variants for more details.
 - Hospital Acquired Concern: Select one or more values for Hospital Acquired status of the Concern/Diagnosis.
 - Cancer Grade: Enter or search for a Grade of diagnosis.
 - Cancer Stage: Enter or search for a Stage of diagnosis.
 - Staging System: Enter or search for a Staging System of the Stage.
 - Primary Tumor: Enter or search for a Primary Tumor value.
 - Node Extension: Enter or search for a Node Extension value.
 - Metastasis: Enter or search for a Metastasis value.
 - Data Source: Enter or search for a source of data.

Note:

To define search criteria for Diagnosis, the criteria must include at least one of the following attributes: Diagnosis or Anatomical Site.

- 4. Under Insert as select either:
 - Inclusion to include the criteria in the query
 - Exclusion to exclude the criteria from the query
- Click Submit.



Back to "Build a cohort query".

- 6. A Diagnosis search criteria can be linked to search criteria for Test or Observation, Procedure, Medication and Specimen to define advanced search criteria like "Diagnosis D1 with Test or Observation T1, with Procedure P1, with Medication M1, with Specimen S1" which allows searching for Patients who have:
 - Diagnosis D1 and Test or Observation T1 in association/linked to Diagnosis D1 and
 - Diagnosis D1 and Procedure P1 in association/linked to Diagnosis D1 and
 - Diagnosis D1 and Medication M1 in association/linked to Diagnosis D1 and
 - Diagnosis D1 and Specimen P1 in association/linked to Diagnosis D1
 In the above example the criteria D1, T1, P1 and M1 can be defined using
 multiple values of the selected data (for example: Diagnosis DA1 or DA2 or
 DA2...; Procedure PR1 or PR2 or PR3... and so on) and the criteria can
 include all of the attributes available for defining the search on a particular
 data (for example: Diagnosis, Diagnosis Status, Onset Date, Cancer Grade
 and so on)

To define a criteria linked to Diagnosis search criteria:

- Define a criteria on Diagnosis via Clinical Data category Diagnosis dialog and Submit.
- b. On the query builder page, where the query criteria is displayed under the
 - sections Inclusions and Exclusion, click the **Link** icon () displayed next to the Diagnosis search criteria. This action brings up the popup menu for selecting the clinical data for which search criteria is to be defined and linked to the Diagnosis search criteria. The menus has options to select the following clinical data: Procedure, Medication, Test or Observation, and Specimen.
- c. Click on the popup menu to select a clinical data. This action brings up the search dialog for the clinical data, which is similar to the search dialog invoked via Clinical Data menu on the left side of the screen. To indicate that this search dialog when submitted would create a linked criteria, a label 'Add Linked <clinical event>', for example 'Add Linked Procedure' is displayed in the search dialog.
- d. Define the search criteria for the selected clinical data and submit the search dialog for the clinical data. This action adds the search criteria for the linked clinical data to the query builder screen where the newly added criteria is displayed below the diagnosis search criteria and linked to it via the label 'WITH'.
 - For information on defining search criteria for a clinical data see section Clinical Data under Build a Cohort Query in this document.
- e. Similarly linked search criteria can be defined for the other clinical events.

A criteria defined using the Diagnosis dialog can be linked to criteria for Procedure, Medication, Test or Observation and Specimen to create a single set of linked criteria. Such linked set can be ANDed to any other criteria including another Diagnosis based linked set(s) or standalone criteria defined using any of the clinical or genomic data, for example:

Diagnosis (DA1 or DA2 or DA3...) OR (DB1 or DB2 or DB3...)



- WITH Procedure P1 or P2 or P3...
- WITH Medication M1 or M2 or M3...
- WITH Test or Observation T1 or T2 or T3...
- WITH Specimen S1 or S2 or S3...

AND

Diagnosis (DA2 or DA3 or DA4...) OR (DB2 or DB3 or DB4...)

- WITH Procedure P3 or P4 or P5...
- WITH Medication M3 or M4 or M5...
- WITH Test or Observation T3 or T4 or T5...
- WITH Specimen S1 or S2 or S3...

AND

Diagnosis D7 or D8 or D9...

AND

Procedure P10 or P11 or P12...

AND

Medication M10 or M11 or M12...

AND

History H1 or H2 or H3...

AND

Specimen S1 or S2 or S3...

...

AND

Criteria defined on Genomic data

Have you checked "Before you begin building your query" yet?

Clinical Encounter

- 1. On the left, click the Clinical Data category, then click Clinical Encounter.
- 2. Either enter the **Encounter Type** or click the search icon ($^{\square}$).

To search for an **Encounter Type**:

- a. In the Select Encounter Type window, select a condition for Encounter Name or Encounter Code and enter part of the encounter name or code.
- b. Select the consent encounter codes and names that you want to use from the left side and move them into the right side using the arrow ().
- c. Click Submit.
- Set any other criteria that you want to use in your query: Location, Time or Data Source.



- Location: Enter or search for a location.
- Time: Select a condition, and then enter a time in days or in the form of a
 date.
- Data Source: Enter or search for a source of data.



To define search criteria for Encounter, the criteria must include at least one of the following attributes: Time - in the past Or Time - Date.

- 4. Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 5. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Procedure

- 1. On the left, click the Clinical Data category, then click Procedure.
- 2. Either enter the **Procedure** and **Procedure Type** or click the search icon (). To search for a **Procedure** and **Procedure Type**:
 - a. In the new window, select a condition for Procedure Name, Procedure Code or Code System and enter part of the procedure name, code or code system.
 - b. Click Search.
 - c. Select the procedure code, name and code system that you want to use from the left side and move them into the right side using the arrow ().
 - d. Click Submit.
- 3. Set any other criteria that you want to use in your query: Procedure Start Date, Procedure End Date, Anatomical Site, Procedure Outcome or Data Source.
 - Procedure Start Date: Select a condition, and enter the date.
 - Procedure End Date: Select a condition, and enter the date.
 - Anatomical Site: Enter or search for an anatomical site.
 - Procedure Outcome: Enter or search for a procedure outcome.
 - Data Source: Enter or search for a data source.



To define search criteria for Procedure, the criteria must include at least one of the following attributes: Procedure, Procedure Type, Anatomical Site, or Procedure Outcome.

- 4. Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Medication

- In the Cohort Query screen, click the Clinical Data category, then click Medication.
- 2. Either enter the medication or click the search icon ($^{\square}$).

To search for **Medication**:

- a. Select one of the Search Modes:
 - Classic Search. More search conditions are available.
 - Hierarchy Drill-in Search. Select it to search all of the codes in a hierarchy that are associated with a single Code System, and have only one parent code.

Note:

This feature does not support mixed hierarchy, meaning different levels of codes belonging to different Code Systems or multi-parent hierarchy, meaning one code having multiple parent codes.

- Search for a medication either by Medication Name, Medication Code, Code
 System ID, or Code System Name and click Search.
- c. Select the medication codes that you want to use from the left side and move them into the right side using the arrow ().
- d. Click Submit.
- 3. Set any other criteria that you want to use in your query:
 - Medication Start Date: Select a condition and enter a date.
 - Medication End Date: Select a condition and enter a date.
 - Dosage: Select a condition and enter a date.
 - Dosage Units: Enter or search for dosage units.
 - Medication Outcome: Select a condition and enter a date.



Data Source: Enter or search for a data source.



To define search criteria for Medication, the criteria must include at least one of the following attributes: Medication or Medication Outcome.

- 4. Under **Insert as** select either:
 - Inclusion to include the criteria in the guery.
 - Exclusion to exclude the criteria from the query.
- 5. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Patient History

- In the Cohort Query screen, click the Clinical Data category, then click Patient History.
- 2. Either enter the **Patient History** code or click the search icon ($^{\square}$).

To search for **Patient History**:

- a. In the Select Patient History window, select a condition for History Name, History Code, History Type, Code System ID, or Code System Name and enter part of the History Code or History name.
- b. Select the history code, name, type, and code system that you want to use from the left side and move them into the right side using the arrow ().
- c. Click Submit.
- 3. Set any other criteria that you want to use in your query:
 - Patient History Start Date: Select a condition and enter a date.
 - Patient History End Date: Select a condition and enter a date.
 - Amount: Select a condition and enter a numeric value.
 - Amount Units: Enter or search for the amount units.
 - Frequency: Select a condition and enter a numeric value.
 - Frequency Units: Enter or search for frequency units.
 - Patient History Value: Enter or search for a patient history value.
 - History Applicable To: Then select whom is it applicable to.
 - Data Source: Enter or search for a data source.



To define search criteria for Patient History, the criteria must include the attribute Patient History.

- Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 5. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Test or Observation

- In the Cohort Query screen, click the Clinical Data category, then click Test or Observation.
- 2. Either enter the test or observation code or click the search icon ($^{\square}$).

To search for a **Test or Observation**:

- a. In the Select Test or Observation window, select a condition for Test or Observation Name, Test or Observation Code, Test or Observation Type, Code System ID, or Code System Name and enter part of either the code, the name or enter the Test or Observation Type.
- b. Select the test or observation code, name and type that you want to use from the left side and move them into the right side using the arrow ().
- c. Click Submit.

Note:

In the Search Test or Observation window, the Test or Observation Type by default uses the Equals operator and lists the first 100 types only. If there are more than 100 Test or Observation type values in your data, you can search using other operators like "Starts with", "Contains", and so on.

- Set any other criteria that you want to use in your query: Test Date, Result (Numeric) Search, Operator, Units, Coded Result, Text Result, or the Data Source.
 - Test Date: Select a condition, and enter a date.
 - Effective Start Date: Select a condition, and enter a date as a single value or a range.
 - **Effective End Date**: Select a condition, and enter a date as a single value or a range.



- Result (Numeric) Search: For Based on absolute values, select an Operator, and enter the units. For Based on Reference Range select an Operator, and an option for the Compare field.
- Coded Result: Enter or search for a Coded Result. In the Select Coded
 Result dialog, select a condition for Result Name, Result Code, Code System
 ID, or Code System Name to search for Coded Results.

In the earlier versions of the product the attribute Coded Result was labeled as Result String across all of the user interface.

- Text Result: Enter or search for a Text Result.
- Data Source: Enter or search for a data source.

Note:

To define search criteria for Test or Observation, the criteria must include at least one of the following attributes: Test or Observation, or Anatomical Site.

- Select an Anatomical Site.
- 5. Under Insert as select:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 6. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Specimen

- 1. In the Cohort Query screen, click the **Clinical Data** category, then click **Specimen**.
- 2. Either enter the codes in the mandatory fields (marked with a double asterisk) or click the search icon ($^{\square}$).

To search for a **Specimen Type**:

- a. In the Select Specimen Type window, select a condition for the Specimen Type Name, Specimen Type Code, Code System ID, or Code System Name and enter part of either the Name, Code, Code System ID, or Code System Name.
- b. Select the specimen code, name and code system that you want to use from the left side and move them into the right side using the arrow ().
- c. Click Submit.



Repeat the same steps when searching for a Specimen Number and Specimen Vendor Number.

- 3. Set any other criteria that you want to use in your query:
 - Anatomical Site: Enter or search for an anatomical site.
 See Sequence Variants for more details.
 - Specimen Collection Date: Select a condition and enter a date.
 - **Specimen Amount**: Select a condition and a numeric value.
 - Units: Enter or search for units.
 - Specimen Alias: Enter the Specimen Aliases in the input box or click the search icon () to invoke the Select Specimen Alias dialog.
 - In the dialog select a condition for Specimen Alias, Service Provider
 ID or Service Provider Name (Service Provider being the issuer of the Specimen Alias) and click the Search button.
 - Select the Specimen Alias(es) that you want to use from the left side and move them to the right side using the arrow icon ().
 - Click Submit.
 - Data Source: Enter or search for a data source.



To define search criteria for Specimen, the criteria must include at least one of the following attributes: Specimen Type, Specimen Number, Specimen Vendor Number, Anatomical Site, or Specimen Alias.

- 4. Under **Insert as** select:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 5. Click Submit.

Back to "Build a cohort query".

6. A Specimen search criteria can be linked to search criteria for Test or Observation to define advanced search criteria like Specimen S1 with Test or Observation T1 which allows searching for Patients who have Specimen S1 and Test or Observation T1 in association/linked to the Specimen S1.

In the above example, the criteria S1 and T1 can be defined using multiple values of the selected data (for example: Specimen S1 or S2 or S3...; Test or Observation T1 or T2 or T3... and so on) and the criteria can include all of the attributes available for defining the search on a particular data (for example: Specimen Type, Specimen Number, Specimen Vendor Number, Specimen Anatomical Site and so on).



To define a criteria linked to Specimen search criteria:

- Define a criteria on Specimen via Clinical Data category Specimen dialog and Submit.
- b. On the query builder page, where the query criteria is displayed under the

sections Inclusions and Exclusion, click the Link icon () displayed next to the Specimen search criteria. This action brings up the popup menu for selecting the clinical data for which search criteria is to be defined and linked to the Diagnosis search criteria, in this case, Test or Observation.

c. Click on the popup menu to select Test or Observation. This action brings up the search dialog for Test or Observation, which is similar to the search dialog invoked via Clinical Data menu on the left side of the screen. To indicate that this search dialog when submitted would create a linked criteria, a label 'Add Linked Test or Observation' is displayed in the search dialog.

Define the search criteria for Test or Observation and submit the search dialog. This action adds the search criteria for the linked Test or Observation to the query builder screen where the newly added criteria is displayed below the Specimen search criteria and linked to it via the label 'WITH'.

Have you checked "Before you begin building your query" yet?

Study

- 1. In the Cohort Query screen, click the Clinical Data category, then click Study.
- 2. Either enter the code for a study or click the search icon ($^{\mbox{\em Q}}$)

To search for a **Study**:

- In the Select Study window, select a condition for Study Name or Study Identifier and enter part of the name or identifier.
- b. Select the study identifier and study name that you want to use from the left side and move them into the right side using the arrow ().
- c. Click Submit.
- 3. Set any other criteria that you want to use in your query:
 - Study Start Date: Select a condition and enter a date.
 - Study End Date: Select a condition and enter a date.
 - Data Source: Enter or search for a data source.



To define search criteria for Test or Observation, the criteria must include the attribute Study.

- 4. Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the guery.



5. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Relative Time Events

You can search for patients who have a particular diagnosis, procedure, or medication combination in a particular chronological order. For example, you can search for patients who:

- Received a diagnosis of *X* disease any time after starting medication *A*.
- Had procedure K 30 days or more after receiving a diagnosis of Y.
- Started medication *B* at the same time as having procedure *M*.
- In the Cohort Query screen, click the Clinical Data category, then click Relative Time Events.
- 2. Under "Select Patients who have, Step 1" select **Diagnosis**, **Procedure**, **Medication**, **Test or Observation**, or **Genomic Variant**.
- Specify how the events in Step 1 and Step 3 are time-related: "at any time while" or specify a condition.
- **4.** Under Step 3, specify the related event: having Diagnosis, undergoing Procedure, or taking Medication.
- 5. Under **Insert as** selec eithert:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the guery.
- 6. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Genomic Data

Topics in this section:

- Sequence Variants
- Copy Number Variation
- Microarray Expression
- RNA-seq Expression
- Metadata Filters

Sequence Variants

 In the Cohort Query screen, click the Genomic Data category, then click Sequence Variants.



2. You can search on Specimen Type or Anatomical Site.

To search for a Specimen Type:

- a. Either enter part or all of the value for **Specimen Type** or click the search icon ($^{\square}$).
- b. In the Select Specimen Type window, select the condition for Specimen Type Name, Specimen Type Code or Code System and enter part of the code.
- Select the specimen code, name and code system that you want to use in your query from the left side and move them into the right side using the arrow ().
- d. Click Submit.

To search for an Anatomical Site:

- a. Either enter part or all of a value for **Anatomical Site** or click the search icon ($^{\square}$).
- b. In the Select Anatomical Site window, select one of the **Search Modes**:
 - Classic Search. More search conditions are available.
 - **Hierarchy Drill-in Search**. You can drill down to see the patient count for any sub-level.
- c. Search for an anatomical site either by Anatomical Site Name, Anatomical Site Code, Code System ID, or Code System Name and click Search.
- d. Select any Anatomical Site codes, names and code systems that you want to use from the left side and move them into the right side using the arrow ().
- e. Click Submit.
- 3. You can also search for patients by different genomic criteria. In the **Select Patients based on** section, select one:
 - having Variants in selected Genes: this enables you to search variants in specific genes.
 - having selected Genomic Variants enables you to search for variants by their Cosmic or dbSNP identifiers.
 - having Variants within specified Genomic Region enables you to search variants in specified genomic location.
 - having Zygosity enables you to search variants with specific zygosity: Any, Homozygous, Heterozygous or Hemizygous.
 - having Amino Acid enables to search for specific amino acid changes in selected genes or caused by specific genomic variant.



- Enter a Hugo Symbol with an optional parameter of amino acid change (such as K483Q,33_34insGA, R603*, 601Kdel, T1738lfs, K2137delinsRE, K224_D225delinsN) and you can see list of amino acid changes existing in the database.
- Enter a variant identifier like Cosmic dbSNP or HGVS identifier and you can see all associated amino acid changes.

Select the amino acids of interest and submit to search for patients or subjects having these amino acid changes. having Genotypes enables you to search for specific genotypes like AT, or AA, or wildtype (same as reference), and so on.

- **4.** After selecting the genomic criteria, expand the **Variant Location** section and specify a value for the selected option.
 - a. Select either at Genomic Variant or at Genomic Position. Add a value.
 - b. Select an Assembly Version.
 - c. Click View Available Genotypes.
- 5. Under **Insert as** select either:
 - **Inclusion** to include the criteria in the guery.
 - Exclusion to exclude the criteria from the query.



Tip:

This applies to all of the criteria specified above.

6. Click Submit.

(Optional) Step 2 Additional criteria

To add additional parameters to your query in the **Step 2 tab**:

- In the Sequence Variants window, click Step 2 (Optional).
- 2. Select any or all of the following parameters:

Variant Attribute:

- a. Specify **Variant Types** from the options below: Substitution, Insertion, Deletion, Indel, or Complex.
- b. Select a **Variant Impact**: Synonymous, Missense, Nonsense, Unknown.
- c. Select a Variant Status: Select Any, Known, or Novel.
- d. Strand: Select Any, + for forward, or for reverse.

Nonsynonymous Substitution Scores:

- a. Select either with Polyphen option or the with SIFT option.
- **b.** Select a condition for the **Version**.
- c. Either enter a **Transcript ID** or search for one by clicking the search icon



Variant Parameters Depending on the Sequencing File Type:

- a. Select either VCF, MAF or CGI master var.
- b. Select the condition for all active fields according to your choice in Step a.
- Under Insert as select:
 - Inclusion to include the criteria in the query.
 - **Exclusion** to exclude the criteria from the query.
- 4. Click Submit.

See "Metadata Filters".

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Copy Number Variation

 In the Cohort Query screen, click the Genomic Data category, then click Copy Number Variation.

To search for a Specimen Type:

- a. Either add the **Specimen Type** or click the search icon ($\stackrel{\square}{\longrightarrow}$).
- **b.** In the Select Specimen Type window, select the condition for **Specimen Type Name**, **Specimen Type Code** or **Code System** and enter part of the code.
- c. Select the specimen code, name and code system that you want to use from the left side and move them into the right side using the arrow ().
- d. Click Submit.

To search for an Anatomical Site:

- a. Either enter part or all of a value for **Anatomical Site** or click the search icon (\mathfrak{Q}) .
- b. In the Select Anatomical Site window, select one of the **Search Modes**:
 - Classic Search. More search conditions are available.
 - **Hierarchy Drill-in Search**. You can drill down to see the patient count for any sub-level.
- c. Search for an anatomical site either by Anatomical Site Name, Anatomical Site Code or Code System and click Search.
- d. Select any Anatomical Site codes, names and code systems that you want to use from the left side and move them into the right side using the arrow ().
- e. Click Submit.
- 2. In the CNV Attributes, specify the CNV Result Type using the two options: numeric or categorized.
 - a. If you select numeric, specify a time value for the SNP Log2 Ratio (Segment Mean).



- b. If you select categorized, specify a condition for the CNV Type, a value for the CNV Type Score, a condition for Called Ploidy and a value for the Ploidy score.
- Specify the CNV Location using any or all options from the in Genes from list: Ad-hoc List, Pathway or Gene Set.
- 4. Enter or search for a value for any or all of the options you selected above.
- 5. Select a value for in **Gene Region**.
- Specify the Assembly Version.
- 7. Specify the DNA Reference Version.

About the DNA Reference Version: it represents the Ensembl reference version for getting gene annotations. This is related to Assembly Version and by default displays the preferred DNA Reference Version, which is set in OHO. The sample value for this is Ensembl Version 70.

- 8. Select the radio button for at Genomic Position and enter a value for it.
- Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.



Tip:

This applies to all of the criteria specified above.

10. Click Submit.

See Metadata Filters.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Microarray Expression

- In the Cohort Query screen, click the Genomic Data category, then click Microarray Expression.
- 2. To select patients with Microarray Expression Results:
 - Either add the **Specimen Type** or **Anatomical Site** (both are optional) or click the search icon ($^{\square}$).

To search for a Specimen Type:

- In the Select Specimen Type window, select the condition for Specimen Type Name, Specimen Type Code or Code System and enter part of the code.
- ii. Select the specimen code, name and code system that you want to use in your query from the left side and move them into the right side using the arrow ().



iii. Click Submit.

To search for an Anatomical Site:

- i. Either enter part or all of a value for **Anatomical Site** or click the search icon (2).
- ii. In the Select Anatomical Site window, select one of the **Search Modes**:
 - Classic Search. More search conditions are available.
 - **Hierarchy Drill-in Search**. You can drill down to see the patient count for any sub-level.
- iii. Search for an anatomical site either by **Anatomical Site Name**, **Anatomical Site Code** or **Code System** and click **Search**.
- iv. Select any Anatomical Site codes, names and code systems that you want to use from the left side and move them into the right side using the arrow ().
- v. Click Submit.
- 3. Select the **Microarray Attributes**: the Array Type, the conditions and values for Intensity, P-value and/ or Call.
 - The Array Type: one-channel or two-channel.

If you select one-channel, specify the needed conditions and values for **Intensity**, **P-value<** and **Call**.

If you select two-channel, specify a condition for the **Log2Ratio**.

- n the Expression for Genes from select either: Ad-hoc list, Pathway or Gene Set.
 Then select the Assembly Version and the DNA Reference Version.
- 5. Under **Insert as** select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 6. Click Submit.

See Metadata Filters.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

RNA-seq Expression

1. In the Cohort Query screen, click the **Genomic Data** category, then click **RNA-seq Expression**.

To select patients with **RNA-seq Expression Results**:

• Either add the **Specimen Type** or **Anatomical Site** (both are optional) or click the search icon ($^{\square}$).



To search for a Specimen Type:

- In the Select Specimen Type window, select the condition for Specimen Type Name, Specimen Type Code or Code System and enter part of the code.
- ii. Select the specimen code, name and code system that you want to use in your query from the left side and move them into the right side using the arrow ().
- iii. Click Submit.

To search for an Anatomical Site:

- i. Either enter part or all of a value for **Anatomical Site** or click the search icon (2).
- ii. In the Select Anatomical Site window, select one of the **Search Modes**:
 - Classic Search. More search conditions are available.
 - **Hierarchy Drill-in Search**. You can drill down to see the patient count for any sub-level.
- iii. Search for an anatomical site either by Anatomical Site Name, Anatomical Site Code or Code System and click Search.
- iv. Select any Anatomical Site codes, names and code systems that you want to use from the left side and move them into the right side using the arrow ().
- v. Click Submit.
- 2. Select a type from Normalization Level: Gene, Exon, or Transcript.
- 3. Select a value for the Normalization Metrics: RPKM, TPM, FPKM, or FPKM UQ.
- **4.** Select Raw Counts, Median Length (Normalized), or Strand based on the chosen Normalization Level (Median Length is only available for Exon).
- 5. Select an option for the RNA-seq Location: Ad-hoc list, Pathway, and Gene.
- **6.** The Transcript IDs option is only available for Exon and Transcript Normalization Levels.
- 7. For each, either enter a value or click the search icon ($^{\square}$).
- a. Select if the location is available for a certain Transcript ID and select the Assembly Version and the DNA Reference Version.
 - **b.** Select at which Genomic Position the patient is (optional, is only available for Exon Normalization Level).
- 9. Under Insert as select either:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria in the query.
- 10. Click Submit.

See Metadata Filters.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?



Metadata Filters

This option is available at the bottom of each of the genomic criteria screens.

To add metadata filters:

- 1. Expand the **Metada Filters** option.
- 2. Click Add Metadata Filter to open the Select Metadata Attribute window.
- 3. Search for a metadata filter based on **Attribute Name**, **Scope** or **Category** to get a list of attributes associated with metadata.
- 4. Select the attributes from the list that you want to use and click **Add to Filter**. When you have added all the filters you need, click **Done**.
- **5.** For each attribute name from the table, select a condition in the **Operator** column, then enter a value for it in the **Value** column.

Example 2-1 Metadata Filters

Attribute Name	Operator	Value
ANALYSIS_ID	Starts with	TGTAID120

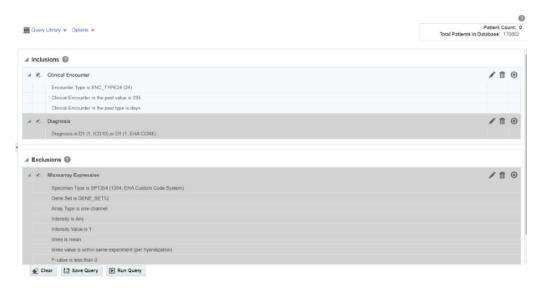
- 6. Under Insert as select:
 - Inclusion to include the criteria in the query.
 - Exclusion to exclude the criteria from the query.
- 7. Click Submit.

Back to "Build a cohort query".

Have you checked "Before you begin building your query" yet?

Combine inclusion and exclusion criteria

Figure 2-1 How inclusion and exclusion criteria work





For you to get the best results, you can use a combination of inclusion and exclusion criteria that can give the exact data that you need.

Once you have added several criteria to your query, you can:

- Expand or collapse the display for the Inclusions or Exclusions criteria.
- Select the arrow to the left of the criteria to expand or collapse their details.

For more information, see:

- Modify details of an existing criteria statement
- Understand the logic behind Inclusion and Exclusion criteria

Modify details of an existing criteria statement

You can continue to adjust the details of an existing statement using the icons on the right.

- The pencil icon () displays and edits your criteria selection.
- The trash can icon () removes any particular statement.
- The plus icon () adds another set of criteria from the same topic. For example, clicking the plus icon from within a Demographics tab prompts you to select additional Demographics criteria.

Understand the logic behind Inclusion and Exclusion criteria

Within an Inclusion statement, distinct criteria statements related to the same data element (Procedure, Medication, Gender, etc.) is considered with an OR criteria.

Example 2-1 Examples of how Inclusion and Exclusion criteria work

Within an Exclusion statement, distinct criteria statements related to the same topic will be considered with an AND.

INCLUDE: Diagnosis = Diabetes AND (Demographics = Male OR Demographics = Married)

EXCLUDE: Diagnosis = Diabetes OR (Demographics = Male AND Demographics = Married)

This function can be used to simplify your query structure, where you can address each data topic and category one at a time rather than having the same topic listed in both the Inclusion and Exclusion sections.

View Patient or Subject Count and data

With this functionality, you can see the number of patients or subjects who meet specified criteria and the total number of patients or subjects in the database. You can also see those patients' or subjects' data.

1. On the Home page, on the left, click the **Cohort Query** icon (





- 2. Select at least one row of criteria defined in the **Inclusions** or **Exclusions** options.
- 3. Click Run Query. The patient count is visible in the top right.
- 4. To view patient data, click Patient Count.

View Specimen Count

- 1. Along the top, click the **Options** drop-down.
- 2. Select Show Specimen Count.
- 3. The specimen count is displayed. This is the count of all the specimens in the cohort that match the selected criteria.



This count is shown for all specimens associated with a patient or subject, not just the ones specific to the selected criteria.

Save a query

After selecting the criteria for your query:

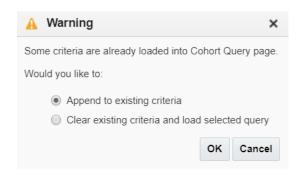
- 1. In the Cohort Query screen, click Query Library.
- 2. Click Save Query in the drop-down menu.
- 3. In the Save Query window, select:
 - create new to save it as a new query
 - search existing to save the query under an already existing name in the query library.
- 4. Click Submit.

Load a saved query

- 1. In the Cohort Query screen, click Query Library.
- 2. Click Load Query in the drop-down menu.
- 3. In the Load Query window, enter all or part of a Query Name.
- 4. Click Search.
- 5. Select the guery.
- 6. Click Submit.



Figure 2-2 Warning message for adding or clearing criteria



If an existing query is currently running, you can either add the loaded query to the newly created one or you can clear the existing criteria and use only your loaded query.



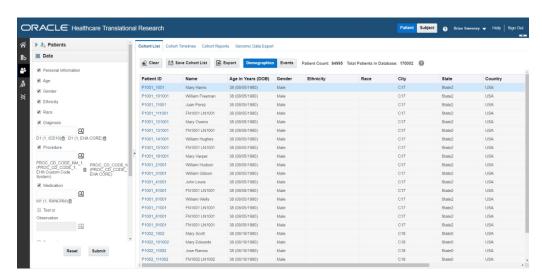
View and maintain cohorts

In this chapter, you will find out how to:

- View cohort lists
- View cohort timelines
- Align data by Patient or Subject Event
- View cohort reports
- Export genomic data

View cohort lists

Figure 3-1 A cohort list with Demographic and Events data



On the Home page, on the left, click the Cohort Viewer icon (



- 2. On the left side, in the **Patients** section, choose your option:
 - active query: to view the cohort list for a patient or subject query that is currently running
 - **library query**: to view the cohort list for a patient or subject query from the library
 - list: to view the cohort list for a saved patient or subject list
 - ad-hoc query: to view lists for specific patients or subjects you want to examine



3. Click Submit.

Use the Data pane on the left to select and filter the data you want to view for the cohort list. For details on filtering data, see Filter data in cohort lists.

After running the query, the patient or subject count is displayed above the table, and you can review the data of those patients or subjects.

Note:

Patient or subject count can also be viewed in the Cohort Query screen as well. For details, see View Patient or Subject Count and data. If you navigate back to Cohort Query screen to edit the active Query and run it, then after navigating back to Cohort Viewer, you must click Submit to refresh the data in this screen.

For more information, see:

- Filter data in cohort lists
- Display procedure associated with medication
- Display anatomical site details
- · Display specimen details

Filter data in cohort lists

- 1. On the left, below the Patients section, click Data.
- 2. Select or deselect any of the information that you want included in the cohort lists.
- Either click the radio button for Include criteria used in a query or set your custom criteria.



Note:

The **Include criteria used in a query** can only be used for active queries.

This option filters clinical records in cohort lists and cohort timelines only on the basis of the primary coded attributes from the Cohort Query screen. For example, it only uses Diagnosis Codes, Procedure Codes or Medication codes.

If the inclusion criteria of the active query is not based on any data topics supported in the cohort lists, a message is displayed to select at least one supported data element.

In the Cohort Viewer, criteria defined on any particular data (e.g. Diagnosis, Procedure etc.) using the OR option should be sufficient for filtering the clinical records in Cohort List. For example, if the Cohort Query criteria is "Diagnosis Code = D1 OR Diagnosis Anatomical Site Code = A1", then the Cohort Query will correctly fetch all the patients who have Diagnosis Code = D1 or any diagnosis associated with the anatomical site A1. However, selecting the Include Criteria Used in the Query checkbox will filter clinical records on the Cohort List such that only the Diagnosis records having Diagnosis Code = D1 AND Anatomical Site = A1 are displayed.

To set custom criteria:

- Select the criteria.
- b. Enter the criteria or click the search icon ($^{ exttt{Q}}$).
- c. In the new window, enter all required details and click **Search**.
- d. Select the elements that you want to use from the left side and move them to the right side using the arrow ().
- e. Click Submit.

4. Click Submit.

To remove data, clear the appropriate boxes and click **Submit** again.

To export the list to a Microsoft Excel sheet, click **Export** (support (support), along the top of the table. Keep in mind that additional information, such as address and email are only visible in the exported XLS file.

To save a cohort list, click the **Save** button (Save Cohort List). You can rename the list, or save it under an existing name. All cohort lists are saved as **Private**.

To switch between Demographic and Events data, use these buttons above the table:

Figure 3-2 Switching between Demographics and Events data in the Cohort Lists





Tip:

To display the date in the dd/mm/yyyy format, use the formatting option in Microsoft Excel.

Display procedure associated with medication

In the Cohort Viewer screen, Events tab, for the Medication records the column Relevant Data displays the Procedure in context of which the Medication was prescribed.

Display anatomical site details

In the Cohort Viewer screen, Events tab the column Anatomical Site displays the Anatomical Site associated with Diagnosis, Test or Observation, Procedure and Specimen records.

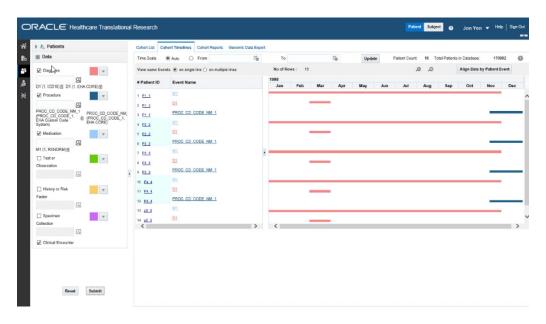
Display specimen details

In the Cohort Viewer screen, Events tab:

- For the Test or Observation records, the column Specimen Details displays the details of the Specimen associated with the Test or Observation.
- For the Specimen records, the column Specimen Details displays the identifier attributes associated with the specimen.

View cohort timelines

Figure 3-3 A cohort timeline containing data on diagnoses, procedures and medications of patients





For more information, see:

- Select patients or subjects
- Filter data in cohort timelines
- Display patient or subject data
- Select the Timeline Mode

Select patients or subjects

This step is similar to how you select patients or subjects for the **Cohort List**.

- Along the top, click the **Cohort Timelines** tab.
- 2. On the left, expand Patients or Subjects to select the source of your patients' or subjects' IDs from an active query, library query, list or ad-hoc query.
- 3. Click Make initial pool.

All patients or subjects from the selected query are added to the initial pool.

Select up to 20 patients or subjects and click the arrow () to move them from the initial pool to the display list.

The data displayed in the cohort timeline is of these 20 patients or subjects.



WARNING:

Don't click Submit yet! Wait until the next section.

Filter data in cohort timelines

- 1. On the left, click the **Data** section.
- 2. Change the colors of the data criteria, if needed.
- 3. Either click the radio button for **Include criteria used in a query** or set your custom criteria.



Note:

The **Include criteria used in a guery** can only be used for active queries.

This option filters clinical records in cohort lists and cohort timelines only on the basis of the primary coded attributes from the Cohort Query screen. For example, it only uses Diagnosis Codes, Procedure Codes or Medication codes.

If the inclusion criteria of the active query is not based on any data topics supported in the cohort lists, a message is displayed to select at least one supported data element.



To set custom criteria:

- Select the criteria.
- **b.** Click the search icon ($^{\square}$).
- c. In the newly opened window, enter all required details and click **Search**.
- d. Select the elements that you want to use from the left side and move them to the right side using the arrow ().
- e. Click Submit.
- 4. Now that you have your data set, click **Submit** again.

Display patient or subject data

The subject and patient data display is divided into two sections.

- The left side has attributes about the selected patients/ subjects: **Patient/ Subject ID** and **Event Name**.
- To see demographic data for a Patient or Subject, hover over their ID.
- The right side shows the colored layers of the selected clinical data, in a timeline view.
- To see more details of a patient or subject and their medical records, click on an **Event** link from the left side table. A pop-up appears with more data on the event.

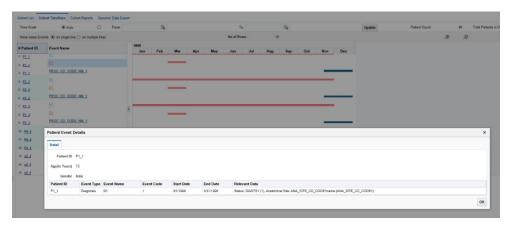


Figure 3-4 Event data shown in a pop-up window

- To navigate to the Single Patient Viewer, and see details of a specific subject or patient, click the Subject or Patient ID.
- To view data for a specific date range, along the top, use the Time Scale options:
 - To view data for certain date range, enter the From and To dates and click Update.
 - To view data without a specific time reference, select the **Auto** option.
- For plotting the Observation events, Observation Effective Start Date and Observation Effective End Date are used as the preferred dates. If these dates are not available then Observation Date is used. The following decision table is used for determining the Start Date and End Date for plotting the Observation event on the chart:



Table 3-1 Data for Plotting Observation Event

W_EHA_OBSV_PATIENT/SUBJECT_H_V			Start Date value in Timeline Plot	End Date value in Timeline Plot	Comment
OBSV_EFF_ START_DT	OBSV_EFF_ END_DT	OBSV_DT			
Available	Available	Available/Not Available	OBSV_EFF_ START_DT	OBSV_EFF_ END_DT	If difference between the two dates is less than 1 Day then for deriving End Date value OBSV_EFF_ START_DT plus 1 Day is used.
Not Available	Available	Available	* If OBSV_DT is one or more days(s) prior to OBSV_EFF_END_DT then OBSV_DT and OBSV_EFF_END_DT are on the same day then OBSV_EFF_END_DT minus 1 Day is used.* If OBSV_DT is one or more day(s) later than OBSV_EFF_END_DT then OBSV_EFF_END_DT then OBSV_EFF_END_DT minus 1 Day is used.	OBSV_EFF_ END_DT	
Available	Not Available	Not Available	OBSV_EFF_ START_DT	OBSV_EFF_ START_DT plus 1 Day	
Not Available	Available	Not Available	OBSV_EFF_ END_DT minus 1 Day	OBSV_EFF_ END_DT	
Not Available	Not Available	Available	OBSV_DT	OBSV_DT plus 1 Day	



Table 3-1 (Cont.) Data for Plotting Observation Event

W_EHA_OBSV_PATIENT/SUBJECT_H_V		Start Date value in Timeline Plot	End Date value in Timeline Plot	Comment	
Not Available	Not Available	Not Available	Null	Null	Event is not plotted

- Events are not displayed on the left-hand table or on the timeline in the following conditions:
 - When an event's Start Date is null.
 - When both Start Date and End Date for the event are null.
- If an event's End Date is null, then the System Date is used for displaying the
 event.

Select the Timeline Mode

To view events in different ways, go to **View same Events** along the top and select one of the radio buttons:

On single line

This is the default mode of displaying data in a timeline.

In this mode, all instances of a repeating event are displayed on the same line and separate events are displayed on separate lines.

On multiple lines

In this mode, all of instances of events are displayed on different lines, even if the events are repeating.

Align data by Patient or Subject Event

1. Click the Align Data by Patient or Subject Event button

- 2. In the new window, select a **Patient** or **Subject ID** from the drop-down list.
- 3. Click the search icon ($^{\mbox{\ \ }}$) next to the **Align by** field, and select the option you want to use.



When you select a particular diagnosis or medication code to align patients, all event codes at different hierarchical nodes are shown for the selected event code.

4. Click Submit.

Align Data by Patient Event * Patient ID P2 2 Result Summary: Data is plotted relative to event Test or Observation: OBSV1(1) for Patient P2_2 Date Range is between December 14, 1997 and November 2, 2004 and is highlighted. Gender (Age in Years) Event Details StartDate EndDate Male(73) Procedure: PROC... 11/14/1998 12/31/1998 P2 2 P2 2 Male(73) Test or Observatio... 6/22/2003 6/23/2003 P2 2 Test or Observatio... 11/22/2003 11/23/2003 P2_2 Test or Observatio... 11/1/2004 11/2/2004

Figure 3-5 How data is aligned by Patient Event

View cohort reports

The **Cohort Reports** tab lets you view various cohort reports.



In the Active Query mode, if no filters have been selected in Cohort Query, then the reports display data for only the first 10,000 patients or subjects. This limit is configurable and can be changed using the DEFAULT_ACTIVE_QUERY_LIMIT property in the TRC.properties file.

However, irrespective of the patient or subject selection option, if the count exceeds the value specified in GENOMIC.REPORTS_MAX_PATIENT_COUNT of TRC.properties file, a warning message is displayed. You can either continue to plot with a large number of patients or subjects (might impact performance) or change the selected cohort to a smaller number.

For more information, see:

- View demographic reports
- View clinical reports
- View genomic reports

View demographic reports

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.
- 3. Click Demographic Reports.



- 4. Select the check boxes for the data you want displayed in your list.
- 5. Click Submit.

Figure 3-6 Demographic reports in OHTR



For more information, see:

Handle unknown data

Handle unknown data

If any missing or unknown data is present, a check box is provided at the bottom of each graph to include missing or unknown data.

For the Age, Gender and Race graphs, if the check box is selected, the graph is refreshed with the unknown data. In this case, unknown data refers to age, gender or race details that are not defined for a particular patient.

For the Age and Gender graph, the unknown age for different genders is combined into one group and the known age is in another group. Both groups are shown in the graph.

View clinical reports

- 1. Along the top, click the **Cohort Reports** tab.
- 2. On the left, click the **Patients** or **Subjects** section, and select a source for your reports, if needed.
- 3. Click Clinical Reports.
- 4. Select the **Specimen Available** check box.
- 5. Click Submit.



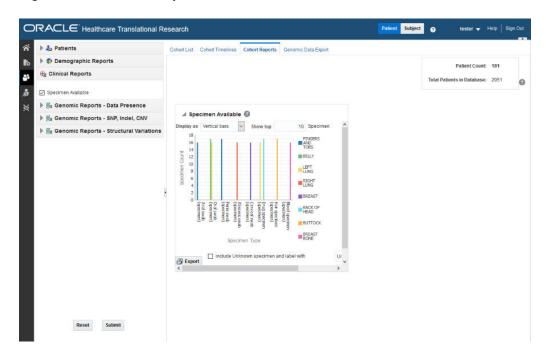


Figure 3-7 A clinical report in Oracle Healthcare Translational Research

For more information, see:

Handle Unknown Data

Handle Unknown Data

If any missing or unknown data is present, a check box is provided at the bottom of each graph to include missing or unknown data.

If Include Unknown Specimen and Label With or Include Unknown Anatomical site and Label With are selected, the graph is refreshed with the unknown data. In this case, unknown data refers to the specimen and anatomical site data that is not defined for a particular patient.

View genomic reports

In this section you will learn how to:

- · View genomic data presence reports
- View Genomic Reports SNP, Indel, CNV
- Structural Variations

View genomic data presence reports

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.





There is no upper limit on the number of patients or subjects to be selected, however the performance slows down as more and more patients are selected.

- 3. Click Genomic Reports Data Presence.
- 4. Select the **Genomic Data Presence** check box. An optional **Specimen Type** input field appears to allow for filtering the initial cohort on specimen type.
- 5. Click Submit.



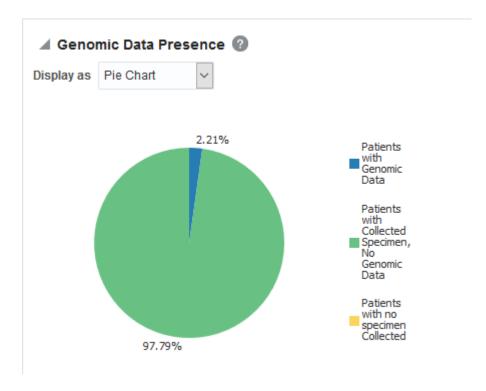
Tip:

To change the display type after generating the report, select the display option from the **Display as drop-down** list.

To view a breakup of present genomic data by data type, click View distribution of genomic data.

To export the graphical plot type as a PNG image, and the table type as an Excel spreadsheet, click the **Export** button(Export) for every graphic.

Figure 3-8 A Genomic Data Presence report in Oracle Healthcare Translational Research





Display as Horizontal Bars

Bequencing

RNA-Seq

Gene
Expression
Copy
Number
Variation

O 1 2 3 4

Number of Patients

Figure 3-9 Distribution of Genomic Data in Oracle Healthcare Translational Research

View Genomic Reports - SNP, Indel, CNV



Genomic Reports SNP, Indel and CNV display the available SNP Indel genomic reports based on the selected cohort of patients or subjects.

Mutated Gene Frequency and Gene Expression

To view these reports:

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.



There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.



- 3. Click Genomic Report SNP, Indel, CNV.
- 4. Select the radio button for **Gene Level Reports**.
- Select Mutated Gene Frequency and Gene Expression.
 Selecting Include All Specimens includes specimens without genomic data. Not selecting this option generates results with genomic data only.
- 6. Enter or search for at least one gene using one of the three search fields: Ad-hoc List, Pathway or Gene Set.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 8. Click Submit.

You can display the results as horizontal bars, vertical bars, or as a table. To export the bar results into PDF or the table results into Excel, click the Export button



Figure 3-10 A genomic report on Mutated Gene Frequency and Gene Expression



About the Mutated Gene Frequency and Gene Expression

Each bar represents a specimen, which is grouped patient-wise and in the order of the specimen collection date. The Sequence Variants and CNV information for each specimen is displayed. The percentage of specimens with mutation on the gene is also mentioned. Export functionality is provided at each specimen level.

There is a limit to the number of specimens that can be seen in the gene matrix plot. The default value of this parameter is 1000. If the number of specimens in the cohort used for this analysis is greater than the specified value, then the following warning message is displayed:



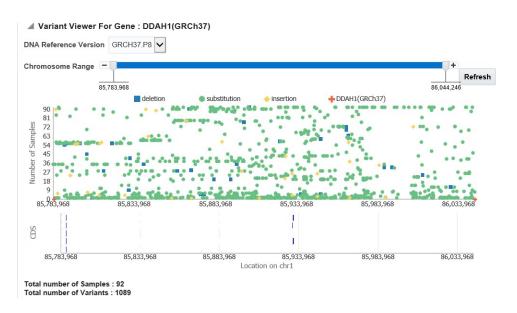
Figure 3-11 Warning message for reaching a limit on specimens



If you continue from this warning message, the report displays the summary statistics instead of the matrix plot. You may have to decrease the cohort size based on some criteria like specimen type or anatomical site or otherwise use cohort query to view the matrix plot. Alternatively, you can also change the default value of MAX_SPEC_REPORT parameter to a desired value and rebuild the plot. However, rebuilding maybe affect the performance of the plot generation.

You can further drill down to the sequencing results by clicking "Show Variant Viewer" in Figure 3-10. This will show data in the form of scatter plot depicting variants on the chromosome (Figure 3-12). This drilldown also shows all the variants in tablular format with reference information below the plot.

Figure 3-12 Scatter plot of all variants distributed on the chromosome for the selected gene



Copy Number Variation Frequency and Gene Expression

To view these reports:

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.



Note:

There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.

- 3. Click Genomic Reports SNP, Indel, CNV.
- 4. Select Gene Level Reports.
- Select CNV Frequency and Gene Expression.
 Selecting Include All Specimens includes specimens without genomic data. Not selecting this option generates results with genomic data only.
- **6.** Enter or search for at least one gene using one of the three search fields: Ad-hoc List, Pathway or Gene Set.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 8. Click Submit.

Note:

When opening the Excel file, you may receive a warning from Excel stating that the file is in a different format than specified by the file extension. This warning can be safely ignored. For more information, refer to http://docs.oracle.com/cd/E23943_01/web.1111/b31973/af_table.htm#autoId34.



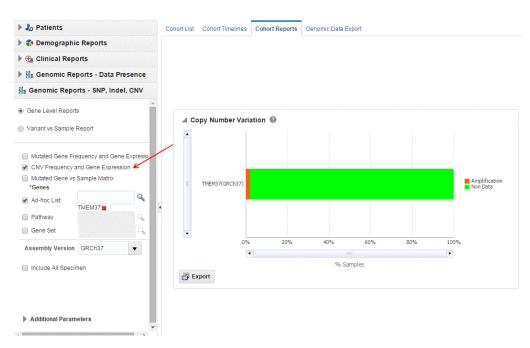


Figure 3-13 A genomic report on Copy Number Variation Frequency and Gene Expression in OHTR

To export the bar results into or table results into Excel by clicking **Export**



Select **Show Gene Expression** to display the Gene Expression details for **Single Channel** or **Dual Channel**. In the **Single Channel Expression**, you must provide hybridization details.

Click **Show CNV Viewer** to display necessary information about all CNV variants that are present in that gene in the samples belonging to the selected cohort of subjects or patients.



Figure 3-14 How to switch between views in a CNV report



Figure 3-15 CNV Sample Details for a selected gene

Mutated Gene vs Sample Matrix

To view this plot:

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.



There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.

- 3. Click Genomic Reports SNP, Indel, CNV.
- 4. Select Gene Level Reports.
- 5. Select Mutated Gene vs Sample Matrix.
- 6. Enter or search for at least one gene using one of the three search fields: Ad-hoc List, Pathway or Gene Set.
 - Selecting **Include All Specimens** includes specimens without genomic data. Not selecting this option generates results with genomic data only.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 8. Click Submit.



When opening the Excel file, you may receive a warning from Excel stating that the file is in a different format than specified by the file extension. This warning can be safely ignored. For more information, refer to http://docs.oracle.com/cd/E23943_01/web.1111/b31973/af_table.htm#autoId34.



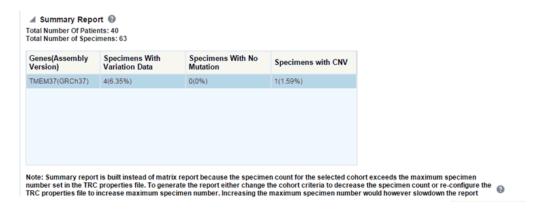
Figure 3-16 A Mutated Gene Vs. Sample Matrix Report in OHTR



Figure 3-17 A Mutated Gene Vs. Sample Matrix Report ready to be exported



Figure 3-18 A Summary Report for Mutated Gene Vs Sample Matrix Reports



Variant vs Sample Reports

To generate the plot:

- **1**. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.





There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.

- 3. Click Genomic Reports SNP, Indel, CNV, and select Variant vs Sample Report.
- Enter or search for a Variant ID.
 Selecting Include All Specimens includes specimens without genomic data. Not selecting this option generates results with genomic data only.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 6. Click Submit.

Figure 3-19 A Genomic Variant Vs Sample Report in Oracle Healthcare Translational Research

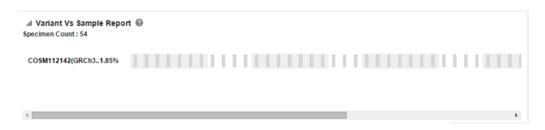
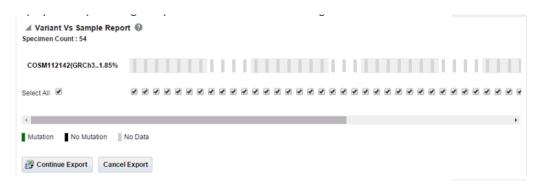


Figure 3-20 A Genomic Variant Vs Sample Report ready to be exported





When opening the Excel file, you may receive a warning from Excel stating that the file is in a different format than specified by the file extension. This warning can be safely ignored. For more information, refer to http://docs.oracle.com/cd/E23943_01/web.1111/b31973/af_table.htm#autoId34.



Structural Variations

Structural Variations in Genes

To view the structural variations in genes:

- **1**. Along the top, the click **Cohort Reports** tab.
- 2. On the left, click the **Patients** or **Subjects** section, and select a source for your reports, if needed.

Note:

There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.

- 3. Click Genomic Reports Structural Variations and select Structural Variations in Genes.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 5. Click **Submit**. By default, the top 10 genes are shown and you can increase or decrease the number of displayed genes.

To export the bar results in PDF or table results in Excel, click **Export** (Export).



🚄 Structural Variations In Genes 🔞 Display as Vertical bars Show top 10 genes 1 Specimen Count Count RHD(GRCh37) BID(GRCh38) FGGY(GRCh37) PLB1(GRCh37) AGPS(GRCh37) CRYZ(GRCh37) GBP3(GRCh37) MIB2(GRCh37) MXD1(GRCh37) TLK1(GRCh37) Genes

Figure 3-21 A Structural Variations in Genes Report in Oracle Healthcare Translational Research

Note:

When opening the Excel file, you may receive a warning from Excel stating that the file is in a different format than specified by the file extension. This warning can be safely ignored. For more information, refer to http://docs.oracle.com/cd/E23943_01/web.1111/b31973/af_table.htm#autoId34.

Structural Variations in Gene Pairs

To generate a Structural Variations in Gene Pairs report in Subject context:

- 1. Along the top, click the **Cohort Reports** tab.
- On the left, click the Patients or Subjects section, and select a source for your reports, if needed.

Note:

There is no upper limit on the number of patients or subjects to be selected. However the performance may slow down as the number of patients or subjects increases.



- 3. Click Genomic Reports Structural Variations and select Structural Variations in Gene Pairs.
- Expand the Additional Parameters section to add a Specimen Type or an Anatomical Site.
- 5. Click **Submit**. By default, top 10 gene pairs are shown and you can change the default number of bars shown.

To export the bar results in PDF or the table results in Excel, click **Export** (Export).



When opening the Excel file, you may receive a warning from Excel stating that the file is in a different format than specified by the file extension. This warning can be safely ignored. For more information, refer to http://docs.oracle.com/cd/E23943_01/web.1111/b31973/af_table.htm#autoId34.

Export genomic data

The **Genomic Data Export** page is used to export the genomic data for filtered patients or subjects based on **Study**, **Specimen type** and **Anatomical Site** in a specific file format.

Note:

Currently, exporting variation data from sequencing platform, single and double channel gene expression and Copy Number Variation data in VCF, SEG, RES and GCT file formats are supported. These formats are supported by the IGV browser.

For more information, see:

- Select results to export
- · Set the location
- Select file type
- Specify the export options

Select results to export

Currently only one version of data can be exported at a time.

To select results:

- On the left, click the Patients or Subjects section, and select a source for your results.
- Select the Assembly Version from the drop-down list.



- 3. Enter a **Specimen Type** or search for one using the search icon ($^{\square}$).
- 4. Enter an **Anatomical Site** or search for one using the search icon ($^{\square}$).

Set the location

You can export genomic data using three different methods:

- Click the radio button for In Genes from and select one or all options:
 - Pathway so you can select one or more pathways.
 - Gene Set so you can use the user-defined collection of genes.
 - Ad-hoc List so you can select one or more genes.
- Click the radio button for At Genomic Position and enter a specific chromosome region.

The following chromosome region formats are supported:

```
CHR15:10000-200000: Considers region between 10000 to 200000 in chromosome 15.

CHR15:1,200,000+5000 - Considers 5000 bases upstream from 1,200,000 position in chromosome 15.

CHR15 - Considers whole of the chromosome 15.

CHR15:1000 - Considers 1000th nucleotide position of chromosome 15.
```

 Click the radio button for All Data to export the complete genomic data of patients or subjects.

Select file type

Select one or all of the available file types:

- Mutation VCF
- Copy Number Variation SEG
- Microarray Expression RES
- Microarray Expression Dual Channel GCT



For the **At Genomic Position** location selection, the Gene Expression - RES and Gene Expression Dual Channel options are disabled.

About the file types:

- Mutation VCF
- Copy Number Variation SEG
- Gene Expression RES
- Gene Expression Dual Channel GCT



Mutation - VCF

This option exports the sequencing variation data for the selected patients or subjects for either the selected genes, pathway, gene set or for a given chromosome region as selected in the previous option. VCF supports multiple specimens' data in a single file.

The metadata header gives the following information that differs based on the search criteria:

- 1. ##fileformat=VCFv4.1
- 2. ##fileDate: Date and time of the VCF file generated.
- 3. ##source=Oracle Healthcare Omics (OHO, formerly known as Omics Data Bank)
- 4. ##Total Number of patients included in this VCF file
- 5. ##Total Number of samples included in this VCF file
- ##INFO=<ID=NS, Number=1, Type=Integer, Description=Number of Samples With Data>
- ##FORMAT=<ID=GT,Number=1,Type=String,Description=Genotype>
- 8. ##FORMAT=<ID=GQ, Number=1, Type=Integer, Description=Genotype Quality>
- ##FORMAT=<ID=GQVAF, Number=2, Type=Integer, Description=Genotype_quality_X>
- 10. ##FORMAT=<ID=DP, Number=1, Type=Integer, Description=Read Depth> ##FORMAT=<ID=AD,Number=.,Type=Integer,Description=Allelic depths for the ref and alt alleles in the order listed >
- 11. ##FORMAT=<ID=HQ, Number=2, Type=Integer, Description=Haplotype Quality>
- 12. ##FORMAT=<ID=BQ,Number=.,Type=Integer,Description=Average base quality >
- **13.** ##FORMAT=<ID=MQ,Number=.,Type=Integer,Description=Average mapping quality >
- **14.** ##FORMAT=<ID=SS,Number=1,Type=Integer,Description=Variant status relative to non-adjacent Normal,0=wildtype,1=germline,2=somatic,3=LOH,4=post-transcriptional modification,5=unknown>
- 15. ##FORMAT=<ID=SSC,Number=1,Type=Integer,Description=Somatic Score>

The following data types are imported to VCF file:

Data Type	Description
CHROM	chromosome
POS	position of the variation
ID	dbSNP ID or COSMIC ID associated with a variant
REF	reference allele
ALT	variant alleles
QUAL	not populated. Will have '.' specified in this column.
FILTER	is populated as PASS
INFO	Not populated. Will have '.' specified in this column.
FORMAT:GT	genotypic data for each specimen.



Data Type	Description
FORMAT:GQ	genotype quality. If not value available in DB, then '.' is specified in the file.
FORMAT:GQX	mapped to GENOTYPE_QUALITY_X column
FORMAT:DP	this stores the TotalReadCount for a specific variant
FORMAT:AD	this stores the reference read count and Allele read count for a specific variant.
FORMAT:HQ	not populated as of now. Will have '.' specified in this column.
FORMAT:FT	this stores GENOTYPE_FILTER column value
FORMAT:BQ	stores the RMS base quality
FORMAT:MQ	stores the RMS mapping quality
FORMAT:SS	stores the somatic status
FORMAT:SSC	stores the somatic status score value
Flex field format	If any custom formats are available, they are also included in the export.

1000 Genomes VCF 4.1 conventions are followed while exporting variation data, however certain data types, which are non-standard, like BQ and MQ, may differ in convention for some customers since there is no standard way to represent them.

Handle Non-variant and No-call Data

If NON_VARIANT and (or) NOCALL records exist for any given position, the zygosity is checked to determine if the information format from these tables is used.



For *het-ref* or *half* zygosity values, these other format fields are compared with the existing SEQUENCING information. This information is then used with zygosity to create the format string.

The NON_VARIANT data allows for GQ, GQX, MQ, BQ and the first reference read count of AD. The NOCALL data allows for all format fields to be compared. Both NON_VARIANT and NOCALL do not support exporting flex fields. The GT value of the format string reflects the stored zygosity as follows:

Zygosity	FORMAT string GT:GQ:GQX:BQ:MQ:AD:DP	
het-ref	1/0:99:98:38:45:20:10,10	
Half	1/.:99:98:34,34:45,45:20:10,5	
Het-alt	1/2:99:98:43,44:56,67:20:0,10,10	
Hom	1/1:99:98:34,34:45,45:20:0,19	

If there are no result records for any specimen, the export displays "." with no other information for the format.

Handle Duplicate Genetic Information Exports



There could be cases where users reload genetic information multiple times for the same specimen. This may create ambiguous values for the different fields that exist in the VCF export file. The export code deals with such ambiguous numerical values that represent the quality (that is, GQ, GQX, AD, BQ, MQ). This code now computes minimum values and ensure that the value of least confidence is reported. There could be more complex cases, for instance, if there are 2 different alleles for the same position belonging to the same specimen, or variants with same position for same specimen with different zygosity. The export code uses MIN functions on all values including all the text fields. This allows for VCF export to create a valid file that can be loaded into genome browsers.

Alternatively, you can choose not to consider data from a specific specimen or a specific file using following methods:

- Using DELETE_FLG A user may load results for a specimen more than once
 that can completely contradict previous results. Users can set the DELETE_FLG
 as 'Y' on W_EHA_RSLT_SPECIMEN and (or) W_EHA_SPEC_PATIENT or
 W_EHA_SPEC_SUBJECT to have previous loads excluded, and then reload the
 correct result files. When the user now exports the data, only the latest loaded
 specimen data is considered for export.
- Using FILE_URI Oracle recommends using this method since you need not reload the data again as opposed to the above method. When there are multiple files loaded with contradicting data for the same specimen, user can set some files as obsolete by changing the W_EHA_FILE_LOAD.FILE_WID column. For example, if you have loaded the same specimen data 3 times and would like to consider the latest file loaded for export, then you must first identify the latest FILE_WID from W_EHA_FILE_LOAD table. Then change the FILE_WID of two old files in W_EHA_FILE_LOAD table to the latest FILE_WID. Now, all the three records belonging to the three file loads contain same FILE_WID, which represents the latest file load and only the latest file export data is exported.

Represent AD Values

Allele depth values represented under the AD data type are in the order of the alleles represented in the GT. Refer to the following table with examples:

ALT	FORMAT	SAMPLE1	
G,C,T	GT:AD	1/2:0,4,6	0 represents reference_read_count
			4 represents allele_read_count of 'G'
			6 represents allele_read_count of 'C'
G,T	GT:AD	2/2:0,4	0 represents reference_read_count
			4 represents allele_read_count of 'T'
G,T	GT:AD	1/0:10,5	10 represents reference_read_count
			5 represents allele_read_count of 'G'

Copy Number Variation - SEG

The copy number variation data is exported in SEG format. Currently, CNV data from any array based system like Affymetrix Genome Wide SNP 6 array whose data is in SEG format while loading in OHO is supported. The main requirement for exporting CNV data is to have the SEG_MEAN value in the CNV table of OHO.

For exporting data that is not loaded from SEG files, for example, data from CGI CNV files or any other source of CNV data, users have to create their own loader.



The loader is expected to calculate the SEG_MEAN value since this value is most important for export.

- 1. ID: specimen ID of the reported CNV segment
- 2. chrom: chromosome name
- 3. loc.start: start position of the CNV segment
- loc.end: end position of the CNV segment
- 5. num.mark: for array based CNV data, this stores the number of probes details
- seg.mean: this stores the segment mean value from SEG_MEAN column in CNV table.

Gene Expression - RES

RES is one of the gene expression formats supported by IGV browser. Currently, only microarray gene expression data is exported to this format. Following data types are imported to RES format:

- 1. Description: hugo name of a specific probe
- 2. Accession: probe ID
- 3. Intensity: intensity value of the associated probe
- 4. Call: call of the associated probe

Gene Expression Dual Channel - GCT

GCT is one of the gene expression formats supported by IGV browser. Currently, only AgilentG4502A platform microarray gene expression data is exported to this format. Following data types are imported to GCT format:

- 1. Description: Gene symbol of a specific probe
- Accession: probe ID
- 3. Intensity: intensity value of the associated probe
- 4. Call: call of the associated probe

Note:

The GCT file takes its gene symbol for the probe from the 2-channel composite element of ADF file. This is input into the ADF composite table in OHO. This value may not match with HUGO name in certain cases as OHTR associates 2-channel records in the result table that has partial (which includes a flanking region set by the user) genomic coordinate. The coordinate overlaps between composite elements and gene segments in the reference. This may also result in some cases in more than one unique gene in the reference mapping to a gene composite element.

Specify the export options

Export data in three ways:



- Select the option to download last loaded file(s)
- **Immediately**, which is the default option
- Schedule

After selecting the **Export** option, click **Submit** to finalize the genomic data export. The data is generated and a separate link is provided in the bottom panel for each result type.

About Export Options

The **Immediately** option gives you the file link on the same screen and you can click to download it immediately. The link provided has a specific naming convention: *file type_OHO_date:MM-DD-YYYY_time:HH24*-MI-SS.file_type_extension*. For example, RES_OHO_09-14-2014_04-26.res. A short description of the file stating data type and advice on the expected count of features is displayed below the created link.

The **Schedule** option runs the process as a job. You can track the status of the job from the **Home**, **Jobs** tab. This option is best suited for exporting large data sets like **All Data** or whole chromosome variants. For the scheduling option, you must provide a job name and description.

There is a possibility of replicate and duplicate data in the database. This could be due to loading multiple files belonging to the same specimen_number. This can happen if the same library is sequenced multiple times or the data is reanalyzed. For example, the reads were realigned using the new reference version, so new VCF or gVCF files are created for the same sample. In this scenario, you can use the option to export VCF data only from last loaded files. For example, if variation data has been loaded for a specimen in Jan 2015, Mar 2015 and July 2015, then using this option you can export data from the file loaded in July 2015 and it would not consider variants from the file loaded in Jan and Mar 2015.

Note:

The **Schedule Jobs** option uses an asynchronous approach to store the file in DBFS. As an alternative to downloading the file using the link in the Oracle Healthcare Translational Research page, there are other ways to access DBFS. From a Linux OS, you can mount DBFS using *dbfs_client* application and then browse the directories. Windows OS does not support the FUSE interface and cannot mount DBFS directly. However, there is a *dbfs_client* application for Windows that can execute commands to access DBFS. The Windows version of *dbfs_client* lets you use the command line to execute normal directory commands. You can list the DBFS directories as well as copy data from DBFS to the local drive. The *dbfs_client* application is part of the standard Oracle client software.

For more information about using

dbfs_client, see http://docs.oracle.com/cd/E11882_01/appdev.112/
e18294/adlob client.htm#ADLOB0006.



4

View data of a single patient or subject

In this chapter you will find out how to:

- View records
- View circular genomic data

View records

To view a patient record:



- 1. On the Home page, on the left, click the Single Patient Viewer icon (
- 2. Enter the patient ID.
- 3. Select the Source:
 - All records is the default option. This allows you to search through all available patients in the CDM database.
 - Cohort
- 4. Select the clinical and genomic data you want to view for the patient.
- 5. Click Submit.

To sort patient or subject data, either click the phenotype column to reorder them, or enter a value in the blank field, at the top of any column, then hit **Enter**. In the Diagnosis table, the column Additional Diagnosis Details displays hyperlinked text for coded values of Grade, Stage, Staging System, Primary Tumor, Node Extension and Metastasis associated with the Diagnosis. The tool tip on the text displays the corresponding code names. To see detailed values of these attributes click the hyperlink to display the Diagnosis Details window.

In the Procedure, Test or Observation, Medication and Specimen tables the column Diagnosis Details displays the Diagnosis that is associated with these records. In the Medications Taken table the column Procedure Data displays the Procedure in context of which the Medication was prescribed.

In the Test or Observation table, use the drop down list 'Additional data to be displayed' to select the columns that should be displayed in the table. Irrespective of the columns selected for display, data from all of the Test or Observation columns is included in the exported data.

To save patient or subject data in an Excel file, click the **Export button** (Export to the category title. You can export either all data or just the selected criteria.

Variants can only be exported separately. To do so, follow these steps below:

- 1. In the Single Patient Viewer screen, on the left, select the **Variants** checkbox.
- Click Submit. The Variants hierarchy graph is displayed on the right side of the screen.

- 3. Click on any nodes in the graph to display the tables with Variants data and select it. These tables show 500 records only.
- Along the top, click Export to export the variants. Variants are exported in an XLS file for the selected node.

For more information, see:

- Display Variant Found
- Export Variant Found
- Navigate through selected patients or subjects
- View genomic data
- Select the Assembly Version

Display Variant Found

When the user clicks on one of the top 2 level nodes (Variants, Coding, Non-Coding, Unknown)

- If the count of variants is less than a threshold value, then the variant table is displayed.
- If the count is greater, then a message is displayed to select any lower level node.

Export Variant Found

While exporting the variants found, if the count of variants is higher than threshold value, user will be to choose a chromosome from drop down list to narrow down the variants.



If the number of variants that you want to export exceeds the limit of 50000, contact your database administrator. The limit can be modified within the database. For more details, see the *Oracle Healthcare Translational Research Administrator's Guide* on the Oracle Help Center.

Navigate through selected patients or subjects

To navigate through patients' or subjects' data in the **Single Patient Viewer**, keep in mind three things:

- If the selected patient ID generates a patient or subject count larger than 1, you
 can navigate through patients' or subjects' history using the Previous and Next
 buttons found next to the Patient ID field, on the left. This is also available for
 patient or subject counts generated in the Cohort Query or Cohort Viewer screens.
- Every patient has an ordinal number (his position within the selection) listed under the **Patient** field. If the selected source is a cohort, then the total number of available patients or subjects from that cohort is also displayed.



3. If you went from the Cohort Timeline screen to the Single Patient Viewer screen, then the total number of available patients or subjects in the cohort is the count of patients or subjects selected in the initial pool, in the cohort timeline.

View genomic data

Genomic data is displayed in four sections as follows:

- Specimen with Genomic Results
- Derived Files
- File Lineage
- Variants Found

Specimen with Genomic Results

Table 4-1 Specimens with Genomic Results

Column Heading	Definition	Sample Value or Values
Specimen Id	Specimen belonging to the selected patient or subject	HG00096
Specimen Vendor Id	Specimen Vendor for that specimen	Vcf
Version Label	Represents Assembly Version (DNA reference version against which this data was loaded)	GRCh37(V68)
Sequence Variants Results	Whether the sample has sequence variants results	Yes / No
Copy Number Variation Results	Whether the sample has copy number variants results	Yes / No
Single Channel Microarray Results	Whether the sample has single channel results	Yes / No
Dual Channel Microarray Results	Whether the sample has dual channel results	Yes / No
Rna-Seq Expression Results	Whether the sample has rna sequencing results	Yes / No
Genotyping Results	Whether the sample has genotyping results	Yes/No

Derived Files

Table 4-2 Derived Files

Column Heading	Definition	Sample Value or Values
Alternate Filepath	The FTP path of the file	
File Type, Version	Type of file and Version	Variant Call Format, 4.1
Result Type	Type of result data in the file	Sequencing, Copy Number Variation, Gene Expression (2-channel or single channel)
Alignment Version (DNA Reference Version)	Represents Assembly Version (DNA reference version against which this data was loaded)	GRCh37(v68)



Table 4-2 (Cont.) Derived Files

Column Heading	Definition	Sample Value or Values
Total Number of Specimen in File	Total number of specimen that the file contains where not all specimen belong to the selected patient	Numeric, positive integer

File Lineage



If you have appropriate permissions, and if files are present in the middle tier accessible location, the path listed in the **File Name** fields for Genomic data are enabled to let you click and download the files directly from Oracle Healthcare Translational Research.

Table 4-3 File Lineage

Column Heading	Definition	Sample Value or Values
Parent Filename	Parent Filename including path of the genomic file stored including path	C:/John_specimen01.BAM
Alternate Filepath	The FTP path of the file	
Alignment Version (DNA Reference Version)	Represents Assembly Version (DNA reference version against which this data was loaded)	GrCH37
Derived Child Files: File - Specimen Id, Vendor Id	Information about Derived Results files that have their lineage based on the particular Low Level file.	C:/John_specimen01.vcf - HG00096_1,HarvardLab1

Variants Found

Selecting this option displays different variants available for a patient. These variants are grouped and displayed in a hierarchical structure with the count of the variants displayed for each type of variants.



Variants can only be exported after they've been submitted.

Variant details are displayed in a table as follows:

Table 4-4 Variant Details

Column Heading	Definition	Sample Value or Values
Chromosome	Chromosome location of the variant	1
Position	The position of the variant within the chromosome	Numeric value



Table 4-4 (Cont.) Variant Details

Column Heading	Definition	Sample Value or Values
Reference Allele	The reference allele of the variant	С
Alternate Allele	The alternate allele of the variant	G
Gene	The gene containing the variant	BID
Transcript	The transcript name	ENSTXXX
Variant Name	The reference id of the variant	rs111
Variant Type	The type of variant	Substitution
Variant Status	Status of the variant	Known
Protein Name		
SIFT Impact	The SIFT impact of the variant	Intolerant
PolyPhen Impact	The polyphen Impact of the variant	Damaging
Drug	The related drug	Clofazimine
Associated Disease	The disease associated to the variant	Anaemia
Primary Histology	Histology information from COSMIC database	Carcinoma
Histology Subtypes	Histology subtypes from COSMIC database	Adenocarcinoma
Primary Site	Primary site from COSMIC database	Large_Intestine
Site Subtypes	Site Subtypes from COSMIC database	colon:NS:NS,NS:NS:NS
Specimen Id	The specimen containing the variant	HG00096
Alignment Version	Alignment version of a variant	GRCh37

Select the Assembly Version

By default, the last loaded assembly is displayed but this selection can be modified. The Genomic data is filtered out with this assembly version instead of the DNA reference version. One Assembly version can belong to multiple DNA reference versions.

View circular genomic data

- On the left, click the Circular Genomic Viewer tab. Select a Patient ID or Subject ID.
 - If an ID is selected in the **View Record** tab, it's inherited in the **Circular Genomic Viewer** tab.
- To filter your view, you can select a Specimen Type or an Anatomical Site. The DNA Reference Version selected is used to filter out the results and determine the cytoband to be used while rendering the circular genomic plot for any of the five data types.
- 3. Select an Assembly Version.
- 4. Click Submit.



- 5. Select only one specimen of different result types to plot the graph. By default, the cytoband of chromosomes is also plotted which is the outer most ring of the circular plot.
- 6. Click Plot Graph.



For the circular viewer to run properly in IE9, turn off compatibility mode.

About the Circular Genomic Viewer

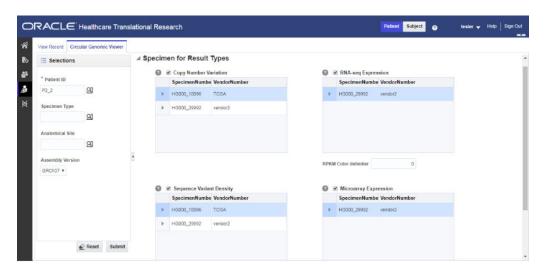
Circular genomic data viewer provides an interface for you to visualize the genomic data which includes variation, micro array expression, copy number variation, dual channel expression and RNA sequencing. The system uses the VisQuick tool, which is a Javascript library built specifically for genomic data visualization.

For more information, see:

· Select data to plot

Select data to plot

Figure 4-1 Selecting data to plot for the Circular Genomic Viewer



For more information, see:

- Microarray Expression
- · Sequencing-Variant Density
- RNA-Seg Expression
- Copy Number Variation
- Dual Channel Microarray Expression



Microarray Expression

The microarray expression panel displays all the specimens of the result type expression. You can select only one specimen at a time.

For the selected specimen from the panel, you can view the list of hybridizations available for that specimen. You can select a maximum of two hybridizations from the multiple choice box, for which the plot is rendered.

The color delimiter value is used to render color to the data points in the plot based on the intensity value. If the intensity value is above the value defined in color delimiter then the data points have green color Otherwise, the points have red color.

Sequencing-Variant Density

This option enables you to plot the density of variants for every 100-kb region for a specimen. The sequence variant density panel displays all the specimens of the result type sequence variants. You can select only one specimen at a time.

RNA-Seq Expression

This panel displays all the specimens of result type RNA Sequencing. Each level of normalization like Gene, Exon and Transcript appears as separate panel. You can select one specimen from each normalization level to view the plot. You also need to select a normalization metrics as RPKM, TPM, FPKM, or FPKM_UQ.

The color delimiter value is used to render color to the data points in the plot based on the normalization metrics value. For example, if the RPKM value is above the value defined in color delimiter then the data points have green color. Otherwise, the points have red color.

You can use the check boxes provided next to each of the result types to determine if the specimen selected is plotted or not. You can select one or all of the result types for plotting of the graph. Once the specimens are selected, the circular genomic plot is displayed. The outermost circle in the plot is the Cytoband. The supported Cytoband versions are: hg-18 and hg-19.

Note:

To display the cytoband in the Circular Genomic plot we need an entry mapping each DNA Reference Version to the Cytoband in the TRC_LOOKUP_CODE table in the APP_schema.

For each reference version we must insert a row with the following values:

- CODE_TYPE: TRC_REFVERSION_CYTOBAND
- CODE:cytobandHG19 (for HG19), cytobandHG18 (for HG18), or cytobandHG38 (for HG38)
- CODE NAME: Name of the loaded DNA reference version (for example, V69)

You can hover over the plot to get details such as:



- Microarray Expression: Chromosome, Start Position, End Position, Value and Gene
- Copy Number Variation: Chromosome, Start Position, End Position and Value
- Sequence Variants: Chromosome, start position, end position, and value. The density value is calculated by: Total number of variants for 100kb region / 100000
- Dual Channel Microarray Expression: Chromosome, Start Position, End Position, Value and Gene
- Rna Seq Expression: Chromosome, Start Position, End Position, Value and Gene

Copy Number Variation

The panel displays all the specimens of the result type copy number variation. You can select only one specimen at a time.

The value for copy number variation depends on the CNV result type selected. If the selected specimen contains data from Genome_Wide_SNP_6 array, then the value for CNV is taken from segment mean stored in the database. If the selected specimen has data from complete genomics, then the value is calculated based on the called Ploidy value stored in the database. The value to be plotted is calculated using the following formula for CNV data from complete genomics:

log2(called ploidy/expected ploidy)

where,

expected ploidy is 2 for chr1-22

expected ploidy is 2 for chrX for females.

expected ploidy is 1 for chrX outside the pseudo-autosomal region in males

expected ploidy is 2 for chrX inside pseudo-autosomal region in males

The pseudo autosomal regions on chrX for 'NCBI build 37' as reported by Complete Genomic are 60000 - 2,699,519 and 154,931,043 - 155,260,559.

The pseudo autosomal regions on chrX for 'NCBI build 36' as reported by Complete Genomic are 0 -2,709,519 and 154,584,237 - 154,913,753.

For called_ploidy zero, the log2 should be infinity, in which cases the final value is taken as -2.

Dual Channel Microarray Expression

This panel displays all the specimens of result type Dual Channel Microarray expression. You can select only one specimen at a time.

The color delimiter value is used to render color to the data points in the plot based on the Log2Ratio value. If the Log2Ratio value is above the value defined in color delimiter, then the data points have green color. Otherwise, the points have red color.

Here is how the graph looks.



Figure 4-2 How genomic data is visualized in the Circular Genomic Viewer



5

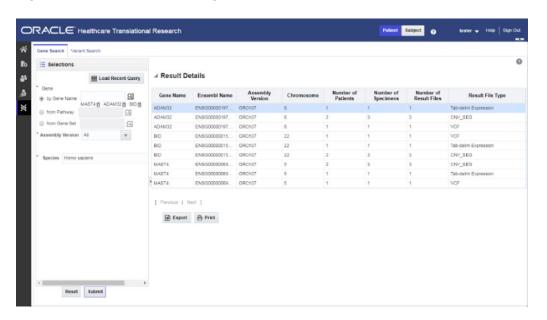
Search genomic results with the Genomic Query

In this chapter you will find out how to:

- Search for genes
- · Search for variants

Search for genes

Figure 5-1 How to search for genes



- 1. On the Home page, on the left, click the **Genomic Query** icon (Make sure that the **Gene Search** tab is open.
- 2. Either load a recently used query by clicking **Load Recent Query** or search for the gene by gene name, pathway and gene set.
- 3. Specify the Assembly Version, Species and DNA Reference version.
- 4. Click Submit.

The results are displayed on the right. You can see the following:

 Result Summary: displays how many genes were searched for, how many have results found and how many do not have any results present in OHO for the criteria selected, what types of results are found.



 Result Details: tabular list of metadata for each gene that is found to have genomic results in OHO.

To print the results, click Print.

To export the resulting table to a spreadsheet, click **Export**.

Click the **Number of Patients** column to navigate to different screens such as:

- View Record
- Circular Genomic Viewer
- Cohort List
- Cohort Timelines
- · Cohort Reports
- Genomic Data Export



If the pathway, gene set or genes are selected from the ad-hoc genes selection, the result details table is paginated based on gene selection. Each page displays the result for a 25 genes selection.

Search for variants

You can specify one or more variant identifiers to be searched across results in Oracle Healthcare Omics (OHO).

- 1. In the upper left corner, click the **Variant Search** tab.
- 2. Either load a recently used query by clicking Load Recent Query or search for the variant by:
 - a. Variant ID
 - b. Variant Attributes:
 - Variant status: select one or all options (All, Novel and Known)
 - Variant type: select one or all options (All, Complex, Indel, Substitution, Insertion, and Deletion).
- Enter the Species and Assembly Version.
- 4. Click Submit.

The results are displayed on the right. You can see the following:

- Result Summary: displays how many variants were searched for, how many have results found and, if applicable, how many do not have any results present in Oracle Healthcare Omics for the selected criteria, what types of result files are found for the variants, if any.
- Result Details: displays a tabular list of metadata for each variant that is found to have genomic results in Oracle Healthcare Omics. The columns list includes options such as...
- Perform a search by Variant ID



Perform a search by Variant Attributes

Perform a search by Variant ID

 Either use a recent query by clicking Load Recent Query or search for a variant by Variant ID.



You can specify Genomic Position by typing in chr#:from-to location.

Example: to find variants in base pair region 1-200 on chromosome 7, you would need to type in the chr7:1-200. If you want to search for any variants on chromosome 7, you can enter chr7.

Additional options that can be specified include known or novel status on the variant, variant type such as insertion, deletion, substitutions, indel, complex. The strand is the directionality of the variant and it can be either plus (+) or minus (-).

- Select the Species.
- 3. If in **Subject** context, enter a **Study** or search for one.
- 4. Select the Assembly Version.
- Click Submit.

Perform a search by Variant Attributes

Figure 5-2 Search for variants by attributes

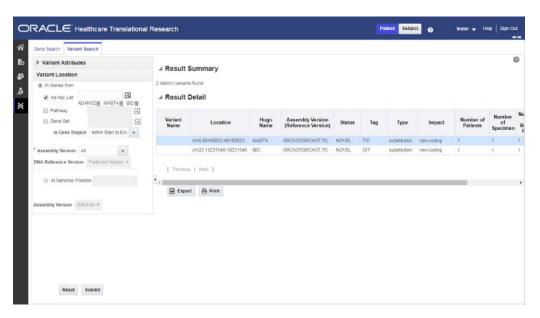
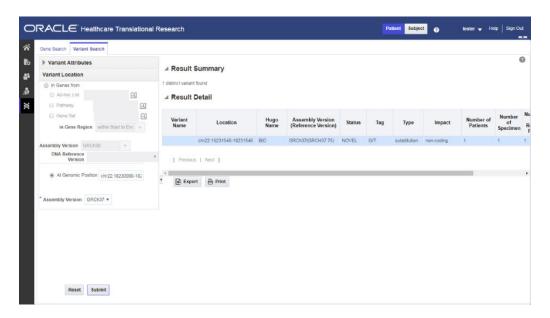




Figure 5-3 Select a variant location



- 1. Either use a recent query by clicking **Load Recent Query** or search for the variant by **Variant attributes**.
- 2. Select one or all options for a Variant status.
- 3. Select one or all options for a Variant type.
- 4. Select one or all options for a **Strand**.
- 5. If in **Subject** context, enter a **Study** or search for one.
- 6. Select the **Species**.
- 7. Expand the **Variant location** section and choose one of these options:
 - in Genes from: Ad-hoc List, Pathway or Gene Set. Also specify the Gene Region and the DNA Reference Version.
 - At Genomic Position



For both variant location options you need to specify the **Assembly Version**.

8. Click Submit.



6

Manage queries, gene sets and jobs in My Workspace



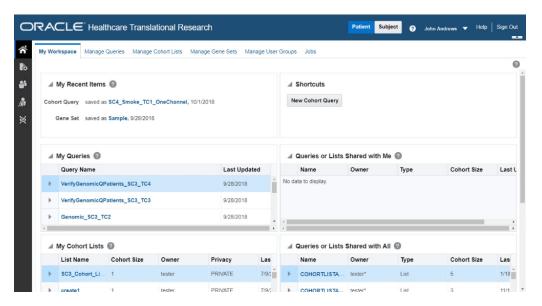
If you encounter issues resulting in Internet Explorer 11 incompatibility, see the *Oracle Healthcare Translational Research Installation Guide* for upgrade instructions.

In this chapter you will learn how to:

- What can I do in My Workspace?
- Manage queries
- Manage cohort lists
- Manage gene sets
- Administrate user groups
- Schedule and monitor genomic data export jobs
- Switch between Patient and Subject context

What can I do in My Workspace?

Figure 6-1 Oracle Healthcare Translational Research landing page and your workspace



The My Workspace page is the landing page for Oracle Healthcare Translational Research. Depending on the license purchased, you may have access to most or all of the sections on this web page. With a standalone Oracle Healthcare Translational Research license, you can only view the Cohort Queries and Cohort Lists details.

Besides managing queries, cohort lists, gene sets, user groups, and jobs, in here you can also get a constant overview of your work in the product. Your workspace features lists of your most recent items, shortcuts, queries and cohort lists.

To understand what you can see in every list, check the details below:

- My recent items
- My queries
- Queries or lists shared with me
- My cohort lists
- · Queries or lists shared with all
- · My gene sets

My recent items

This lists the most recent saved cohort query, cohort list, and gene set, which provides the quickest access to your recent work.

My queries

This lists the most recent saved patient cohort queries, sorted by the last updated date. Clicking the selected query will load it in the Cohort Query window.

Queries or lists shared with me

This lists all queries and lists that are shared with you. Clicking the selected query will load it in the Cohort Query window. Clicking the selected list will load it in the Cohort Viewer/Cohort List window.

My cohort lists

This lists the most recently saved cohort lists sorted by the last updated date. Clicking the selected list will load it in the Cohort Viewer/Cohort List window.

Queries or lists shared with all

This lists all public queries and lists. Clicking the selected query will load it in the Cohort Query window. Clicking the selected list will load it in the Cohort Viewer/Cohort List window.

My gene sets

The My Gene Sets section lists the most recently updated gene sets. Clicking the selected gene sets will load it in the Manage Gene Sets/Manage window.



Manage queries

What can I do here? In the Manage Queries tab, you can:

- Search through saved queries.
- View a list of queries, either created by or shared with you.
- · Modify queries.
- Set or modify restrictions to available queries.

For information on modifying criteria for queries, inclusions and exclusions, see Modify criteria for queries using the Cohort Query or the following topics:

- · Search for queries
- Modify queries

Search for queries

- 1. On the **Home page**, click the **Manage Queries** tab.
- 2. In the Search My Queries section, select a condition and criteria for your search.
- 3. Click Search.

To clear the search and enter new criteria, click **Reset**.

To view a query, select it from the table and click **Load**.

Example 6-1 Select the conditions and enter the criteria to search for queries

```
Query contains "cohort"
Privacy equals "shared"
Creation Date "On or Before" 21.05.2016
Last Update equals 23.07.2016
```

Modify queries

To change a query's name or description:

- Select it from the My Queries list.
- Click Edit and modify as needed.
- 3. Click Submit.

To change the privacy status of a query

- Select it from the My Queries list.
- 2. Change the access:
 - Click Private to prevent anyone else from seeing or using the query.
 - Click Shared with All to allow anyone to use the query.
 - Click Shared to share a private query with a user or a group of users.



If you chose **Shared**, follow the steps below.

- Click Add.
- **b.** Select a condition and enter part of a user name or a group name.
- c. Click Search.
- d. Select the user name or user group you want to include and click Add.
- e. Click Done.

To share the query with multiple user groups, repeat the steps above before clicking **Done**.

Manage cohort lists

What can I do here? In the Manage Cohort Lists tab you can:

- Search through available saved cohort lists.
- View a list of cohorts, either created by or shared with you.
- Modify cohort lists.
- Set or modify restrictions to available queries.

For more information, see:

- Search and view your cohort lists
- Modify cohort lists

Search and view your cohort lists

You can only search for cohort lists that you have access to.

- 1. On the **Home page**, click the **Manage Cohort Lists** tab.
- In the Search my Cohort Lists section, select the conditions and enter the criteria to search for your saved cohort list.

Example: How to enter criteria to search for your saved cohort list

```
List name = "cohort"

Privacy = Private

Create Date = "On or Before" 21.05.2016

Last Update = 23.07.2016
```

3. Click Search.

To clear the search and enter new search criteria, click Reset.

To view a cohort list, select it from the My Cohort Lists and click Load.

Modify cohort lists

You can modify your cohort lists in multiple ways, from editing their names to changing their privacy status.



To change a cohort list's name and description:

- Select it from the My Cohort Lists section.
- Click Edit.
- Click Submit.

To change the privacy status of a cohort list:

- Select if from the My Cohort Lists.
- Change the access:
 - Click Private to prevent anyone else from accessing the list.
 - Click Shared with All to allow everyone to access the list.
 - Click Shared to share a private list.

If you chose **Shared**, follow the steps below.

- a. Click Add.
- **b.** Select a condition and enter part of a user name or a group name.
- c. Click Search.
- d. Select the user name or user group you want to include and click Add.
- e. Click Done.

To share the cohort list with multiple user or user groups, repeat the steps above before clicking **Done**.

Manage gene sets

What are gene sets and what can I do in here? Gene set refers to groups or lists of genes. This set can consist of a couple of genes or hundreds of genes. This allows you to group genes into convenient *collections* for reuse. You can group a few genes for quick search retrieval or for use in a cohort query. In this tab, you can create, edit, and manage gene sets.

A few things to keep in mind about gene sets in Oracle Healthcare Translational Research:

- There are no restrictions on genes that can be included in a gene set. You can mix genes from multiple species or assembly versions.
- The same gene can be part of many different gene sets.
- Gene sets are private and cannot be shared among users.
- Gene set names are not case-sensitive.
- When genes are compared using the Contains or Starts With option, the limit is 512 characters.

For more information, see:

- Create a new gene set
- · Edit a gene set
- · Manage a gene set



Create a new gene set

- 1. Along the top, click Manage Gene Sets and go to the Create New or Edit section.
- Specify the genes you want to include in the gene set. This can be done in three ways:
 - Type in Gene Names Search for genes using their Ensembl or HUGO names. You can enter multiple gene names separated by space, comma or semicolon.
 - Add from existing gene set Search for genes based on an existing gene set.
 - Upload from a file Select a text file (CSV, TSV, TXT) from your computer
 where the genes are delimited by comma, space or tab. Click Choose File to
 browse for the file.



The file size limit is less than 5MB when genes are matched using Equals.

- 3. Select a value for Species.
- 4. Select an Assembly Version.
- 5. Click **Submit**. The matching genes appear in the Available Genes pane.
- 6. Use the arrow () to move available genes to the Final Gene Set panel.

Use the double arrow () to move all available genes to the Final Gene Set panel.

To remove genes from the Final Gene Set panel, use the **Remove** or **Remove All** buttons.

- 7. In the Save Gene Set section, enter the name and description of the gene set.
- 8. Click Submit.

Edit a gene set

To edit a gene set:

- Along the top, click the Manage Gene Sets tab, and go to the Create New or Edit section.
- 2. Search for the gene set you want to edit using the **Add from existing Gene Set** option.
- 3. Select a value for **Species**.
- 4. Select an Assembly Version.
- 5. Click **Submit**. The gene set appears in the Available Genes pane.
- 6. Use the arrow() to move available genes to the Final Gene Set panel.



Use the double arrow () to move all available genes to the Final Gene Set panel.

To remove genes, use the **Remove** or **Remove All** buttons.

7. Click Submit.



When you search genes using the **Contains** or **Starts With** option, the limit is **512** characters

Manage a gene set

To manage a gene set:

- 1. Click the Manage Gene Sets tab and go to the Manage section.
- 2. Enter the gene set you want to modify.
- 3. Click Submit.

To print the data, click **Print** on the top right.

To export the data in an.xls file, click **Export**.

To remove a gene set, click **Delete Gene Set** at the bottom.



When you delete a gene set, its record is still available in the database but it can only be restored by a system administrator.

Administrate user groups

Administrating user groups can be done directly from the Oracle Healthcare Translational Research UI.

To simplify sharing, create user groups and assign users to them. Instead of sharing queries or lists with each user individually, set up a list of users to share multiple items to multiple users at the same time.



Only the user that is the owner of a list can modify its members. However, any user can share any given lists with other groups and see members in other lists.

For more information, see:

- Create a user group
- Assign users to a group



- · Remove users from a group
- Manage user groups

Create a user group

- 1. Navigate to Manage User Groups, User groups.
- 2. Click Create.
- 3. In the Create User Group window, enter a name and description.
- 4. Click Submit.

Assign users to a group

Immediately after you create a group

- 1. Navigate to Manage User Groups, Group has members.
- 2. Click Add.
- 3. In the Add Users window, enter the user names or search for them.
- 4. Select the names you want to add.
- Click Add.
- 6. Click Done.

Search for a group you create earlier

- 1. Navigate to Manage User Groups, Search User Groups.
- Enter the details of the group you want to find.
- Click Search.
- 4. Navigate to Manage User Groups, Group has members.
- 5. Click Add.
- **6.** Enter the user name you want to add and select them.
- Click Add.
- 8. Click Done.

Remove users from a group

- 1. Navigate to Manage User Groups, Search User Groups.
- 2. Enter the details of the group you want to find.
- 3. Click **Search**. The user group and its details are displayed.
- 4. Navigate to Manage User Groups, Group has members.
- 5. Select the user you want to remove.
- 6. Click Remove.



Manage user groups

You can edit, duplicate or delete a user group using the buttons at the top of the **User Groups** section.

- Go to Manage User Groups, Search User Groups.
- 2. Enter the details of the group you want to find.
- Click Search.
- 4. Follow one of these steps:
 - Click Edit to change the name and description of the user group.
 - Click **Duplicate** to create a copy of a user group.
- Click Submit.

Schedule and monitor genomic data export jobs

Currently, jobs are scheduled only for **Genomic Data Export**. To schedule a job, navigate to the **Genomic Data Export** screen in the **Cohort Viewer** tab.



Any job executed prior to applying Oracle Healthcare Translational Research 3.1 will have its status changed back to '**Scheduled**'.

- 1. Select the patient IDs source from one of these options:
 - active query
 - library query
 - list
 - ad-hoc
 - Omics query
- Select the DNA reference version.
- 3. Select the location for the gene:
 - Ad-hoc List
 - Pathway
 - Gene Set
- **4.** Select one of the following file formats to export:
 - Mutation-VCF
 - Copy Number Variation-SEG
 - Microarray Expression-RES
 - Microarray Expression Dual Channel-GCT
- Select the Schedule export option.



6. Click Submit.

To view job information and progress, go to the Home page, and click the **Jobs** tab, along the top. All the jobs created by the Oracle Healthcare Translational Research UI are displayed. All users who created jobs can see job lists.

For more information, see:

- Manage jobs
- View job inputs
- View and download job outputs

Manage jobs

To search for a job:

- 1. On the Home page, along the top, click **Jobs**.
- 2. Scroll down, after the table, and either enter part of the job's name or slick the search icon ($^{\square}$).
- 3. In the Search User Job window, select the appropriate conditions and enter part of a **User Job Run ID** or a **User Job Name**.
- 4. Click Search.
- 5. Select the job you want to include in the list.
- Click Submit.To run that job, on the Jobs tab, click Submit again.

To view details of a particular job:

On the **Jobs** tab, in the first table column, click a job's name to get the details of the job.

Fo return to the **Jobs** tab, click the **Jobs** breadcrumb link.

Figure 6-2 Location of the Jobs link



To get back to the initial list of jobs, click Reset.

View job inputs

- To view details on the parameters provided when scheduling a job, in the **Details** page, click the **Inputs** button on the left.
- To load the job details in the Genomic Data Export page, click the Load input parameter values into Genomic Data Export page link.





You can only load these input parameters, when the job context matches the application's context.

View and download job outputs

This section shows the result files of a job. The following files are generated:

- Error log file contains the log file of the application if any exception or error occurred while running the job.
- **User log file** contains the log file, which shows the error occurred while generating the export files.
- Administrator log file is for the DB administrator.

To download any of these files, click the file link under the **Files** column.

You can download error and user log files, but cannot download the admin log file.



When you try to export a file that does not have any patients or subjects linked to the specimen, an empty file is generated in **Schedule** mode.

Switch between Patient and Subject context

Oracle Healthcare Translational Research can be run in either Patient or Study Subject context. In Patient context, all queries are directed at patient tables in the Cohort Data Model (CDM) schema, while in the Subject context, all queries are routed to the subject tables in the Cohort Data Model (CDM).

- To change context, click either one of the buttons (
 upper-right corner of the screen. By default, the context is set to Patient.
- The new context is available until the next time you change it.
- Switching context clears the data loaded in the Cohort Viewer tabs, but not the criteria of an active query in the Cohort Query screen. Only applicable criteria for the selected context is displayed.

Example

Encounter criteria is not supported in the Subject context and will be disabled. When you switch back to the Patient context, it is enabled again.

For a subject or a patient to be considered deleted and not visible for selection, a
given subject or patient's delete flag in CDM should be set to Y.



7

Oracle Healthcare Translational Research Next Gen (OHTR NG)

In this chapter you will learn about Oracle Healthcare Translational Research (OHTR NG):

- Overview
- Login
- Navigation
- Build Cohorts
- View Cohorts
- View Cohort Events
- View Patient Data
- Logout

Overview

Oracle Healthcare Translational Research Next Gen (OHTR NG) is an updated User Experience (UI) that is built on the same databases as of Oracle Healthcare Translational Research (OHTR). Oracle Healthcare Translational Research NG has been developed using modern technologies as Oracle JET and REST APIs and provides a fresh visual design that complements the modern layout approach.

The phase 1 of Oracle Healthcare Translational Research NG presented the Single Patient View in a more intuitive format retaining the ability to explore data associated with the patient using the same instances of the underlying data models of Oracle Healthcare Translational Research. Phase 2 introduces features to build cohort query criteria and view cohorts along with details of the patients In the cohorts.

Login

Oracle Healthcare Translational Research NG is available using the context path of / ohtr-nextgen/LoginScreen.html

Users of Oracle Healthcare Translational Research (OHTR) can access and login to Oracle Healthcare Translational Research NG using the same user name and password. The login user must be granted the ohf_cg_api_group role to use the application. The user has access to the same set of patients as in Oracle Healthcare Translational Research.

To login:

- 1. On the Login screen, provide your user name and password.
- Click Sign In. After successful login you will be directed to the Patient Viewer screen.

Navigation

The navigation bar on the left side of the screen provides links to:

- Cohort Builder screen
- Cohort List screen
- Patient Viewer Screen
- Oracle Parallel Graph AnalytiX (PGX) Data Studio environment

Build Cohorts

To build cohorts of patients, the Cohort Builder screen provides features to define cohort query which specifies the selection criteria for building cohorts of patients. Cohort query can be based on inclusion and exclusion criteria, each of which can be defined using a combination of demographic and clinical attributes.

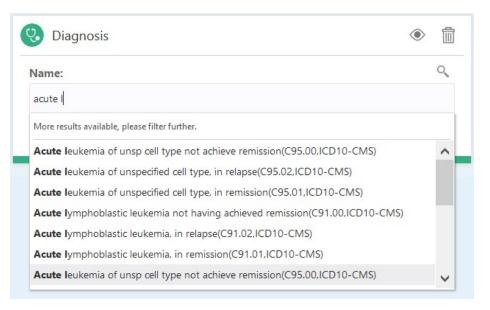
To create inclusion or exclusion criteria for a cohort query:

 Select the data category under the section Patient Info & Demographics or Clinical Events.

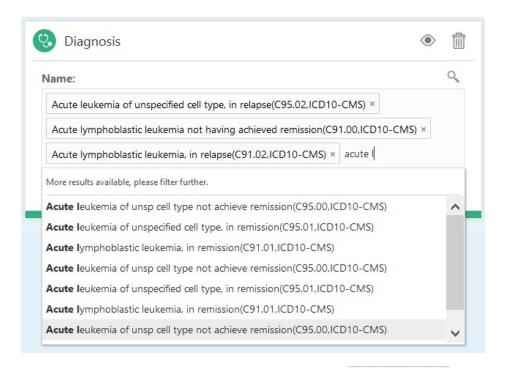


- 2. Click on the menu item for the data category (such as Demographics, Consent, Diagnosis, Procedure etc.) and drag it to the label 'drag items here' under the Inclusion or Exclusion section.
- The above action results in placement of the criteria tile in the Inclusion/Exclusion section.
 - a. In the criteria tile, the primary attribute of the data category (such as Patient ID, Diagnosis Code, Procedure Code, Medication Code, and so forth) can be searched by typing a text string which is likely match with the starting characters of Code Name of the primary attribute. As the text string is typed, the input box displays a list of items in which the typed text string matches with the initial characters of Code Name of the data attribute. For example, in the Diagnosis criteria tile, the text string 'acute I' lists all of the diagnoses in which the Code Name starts with the letters 'acute I'.





From the displayed list, select the desired items, one at a time, to add them to the selection criteria. The selected items are added in the input box one by one. Depending on the number of selected items scroll bar automatically appears in the input box. Note that an item from the drop down has to be selected. Keying in an item that is not selected (adding an item not in the list) would not work.



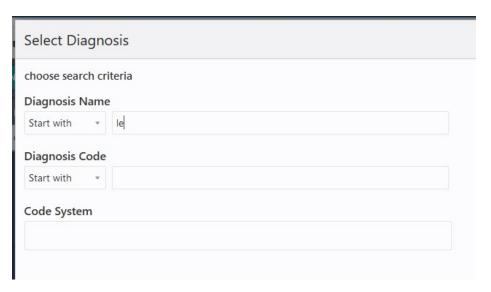
c. To remove the selected items click the 'x' icon (such as the respective its " the respective item.





d. To perform advanced search on the primary attribute or to list all of the

matching Codes click on the magnifying glass icon () in the criteria tile, which invokes the advanced search screen.



- e. In the above illustrated advanced search screen (for example, the Select Diagnosis screen) the attribute values can be searched using a combination of Code Name, Code and Code System. For all these three attributes the search string can be provided in the respective input boxes and a matching operator (such as, Starts With, Contains, Equals, or Not Equals) can be selected from the corresponding drop down list.
 - After specifying the search criteria, click the **Next** button, which displays a list of matching results.
- f. In the selection list screen, use the checkboxes corresponding to the list items to select one or more items from the list. In this screen:

 The label 'Search:' at the top provides information about the current search

To change the criteria use the hyperlink '**Change**' to navigate back to advanced search screen.

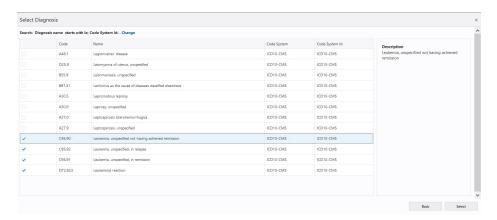
The 'Back' button navigates back to the advanced search screen.

criteria.

The 'Next' button navigates back to the criteria tile, which includes all of the items selected on this screen.

The 'x' icon at the top right section of the screen can be used to close this screen without applying the selected items in the selection criteria tile.

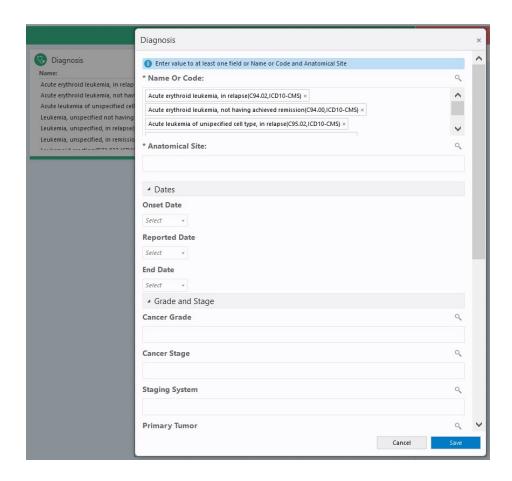




- g. All of the codes selected in a criteria tile are used in Patient search based on the OR condition. For example, in the Diagnosis criteria tile, if a Patient has any one of the selected Diagnosis Codes then the patient is considered a match for the selection criteria.
- h. More Details search screen: In the criteria tile, the More Details

(More Details) link invokes the criteria definition screen where additional attributes of the selected data category are available for definition of criteria. For example, for Diagnosis criteria definition, the dates associated with diagnosis, anatomical sites, and the Grade and Stage attributes are available. The input fields in this screen allow search by either Code Name or Code, wherever applicable. The search is done using the operator Starts With. Thus, for example, for the Diagnosis Grade and Stage field, the input value is searched against both (either) Code Name and Code of the diagnosis grade and stage.



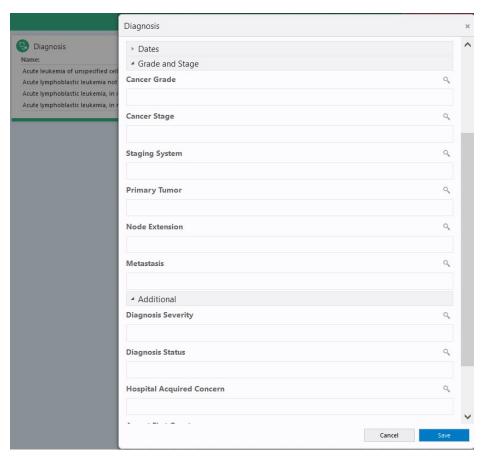


The More Details screen includes all of the attributes that can be used to define search criteria. The attributes are organized into different sections that can be expanded and collapsed by clicking on the respective section headers.

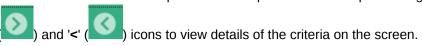


In any date fields, all dates before 1950 should be manually entered in the mm/dd/yyyy format to ensure correct retrieval of patient count.

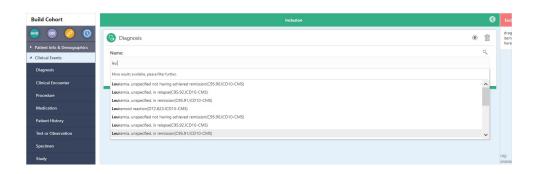


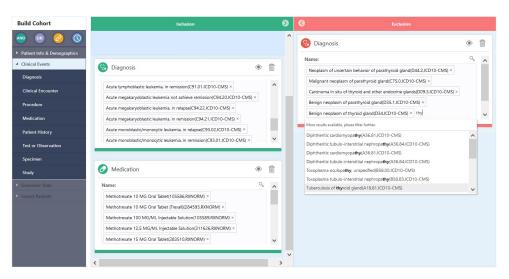


- i. All of the criteria a criteria tile can be enabled or disabled using the 'Eye?'
 - () icon. This action does not remove the criteria tile from Inclusion or Exclusion section but the criteria defined in the tile is not used in the cohort query while searching for the matching patients.
- j. A criteria tile can be deleted using the '**Delete**' () icon. This results in removal of the criteria tile from the Inclusion or Exclusion pan so use this icon with caution.
- 4. Similarly, criteria tiles for other data categories (such as Medication, Procedure, Test, or Observation and so forth) can be placed in the Inclusion or Exclusion sections to create complex criteria consisting of different categories of data.
- 5. The Inclusion and Exclusion pans can be expanded and collapsed using the '>'









- 6. The criteria across multiple criteria tiles is linked using AND operator to select Patients who match with all of the criteria. For example, if Inclusion criteria is defined using Diagnosis (D1, D2, D3...) and Medication (M1, M2, M3...) tiles then it will select all patients who have (Diagnosis D1 or D2 or D3...) AND (Medication M1 or M2 or M3...).
- 7. After a criteria tile is created or updated, clicking anywhere outside the criteria tile invokes the cohort query mechanism of the application to search for the matching patients and the count of matching patients in the cohort thus created is displayed in Number of Patients.





8. A cohort of patients thus created can be saved by providing a unique Cohort Name, Privacy (Public/Private) and optionally Description. After a cohort is saved,

the user who owns the Cohort (that is the logged in user who created the cohort) can update and save it with the same name. Users who have access to shared Cohorts created by other users cannot modify the cohort, but can save the Cohort with a different name. Cohorts created in Oracle Healthcare Translational Research Next Gen can be accessed and updated in Oracle Healthcare Translational Research and vice versa. In this release, Cohort Queries built and saved in Oracle Healthcare Translational Research cannot be imported into Oracle Healthcare Translational Research NG and vice versa.

List of all of the available Cohorts is available in the Cohort List screen.



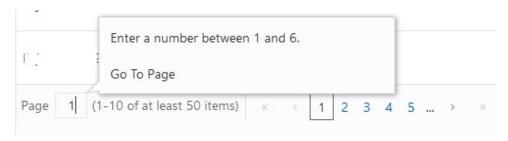
If the Code/Name column in the underlying terminology data in Cohort Data Mart (CDM) contains braces, that is (,) , or both, then the search may result incorrect number of matching Patients. For example, if the value of Diagnosis Name column in the terminology data contains the phrase "Injury (trauma)" then search criteria based on such Diagnosis values may result in incorrect number of matching Patients.

View Cohorts

To view and manage cohorts, the Cohort List screen is accessible using the application's navigation bar. The Cohort List screen provides information about the cohorts that are previously saved and available in the system. A user can only see the cohorts created by self, shared with the user or the public cohorts.

- The upper section of the screen displays the total number of cohorts and number of cohorts shared with other users.
- 2. The tabular cohort list includes the following attributes of the cohorts:
 - a. Name of the Cohort, displayed as hyperlink, double-clicking on which displays the Events List screen with details of the selected cohort.
 - b. Size: Total number of Patients in the Cohort.
 - **c.** Owner: The application User who created the Cohort.
 - d. Last Created Date
 - e. Last Updated Date
- The drop down list with value 'All Cohorts' can be used to selectively display all, Private or Shared cohorts.
- 4. The input box next to the above drop down supports filtering of Cohorts by Cohort Name. Search is based on the operator "Starts With" to match the Cohort Name with the input string. After providing the input string press 'Enter' on the keyboard
 - or click the magnifying glass icon () to refresh the tabular list.
- 5. Pagination available at the bottom of this screen allows navigation across multiple pages of the list. To directly navigate to a particular page, type the page number in the input box next to the Page label and press **Enter**. This is, however, limited to the pages that are already loaded and mentioned in the hover over of the input box. For example, the number between 1 and 6 in the image below.





6. Create Cohort provides navigation to the Cohort Builder screen.

View Cohort Events

The Events List screen provides information about the Patients in the Cohort which was selected in the Cohort List page.

1. The upper section of the screen, Cohort Details, which is closed by default and can be opened using the wedge icon next to the cohort name (such as

PT Test 001

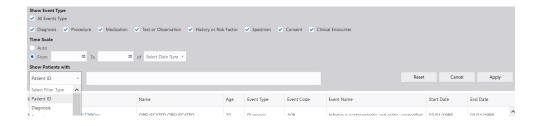
), provides the following information about the selected Cohort: Name, Patient Count, Description, Privacy, Owner, Created Date, and Last Updated Date.



The event list table provides a tabular display of the Patients and their clinical events. The table supports pagination and also direct navigation to the specified page.



3. The Filter/Funnel icon () when clicked, displays the sections Show Event Type, Time Scale, and Show Patients With.



a. In Show Event Type, checkboxes are available for each type of clinical event, which can be used to select the clinical events that should be displayed in



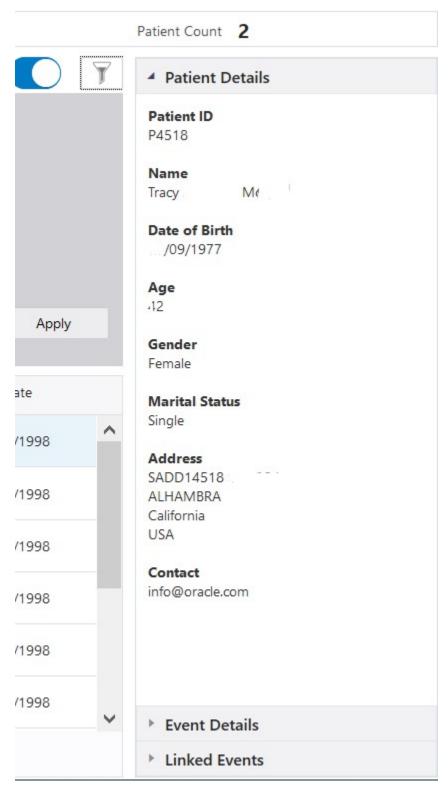
the event list table. The All Events checkbox can be used to display all types of clinical events. When this checkbox is selected, all other checkboxes are automatically selected and vice versa. If any of the event-specific checkboxes are deselected then All Events is automatically deselected. By default, all of the checkboxes are selected.

- b. In Time Scale section, the Auto option (selected by default) displays all of the selected event types without applying any filter on their Start Date or End Date attributes. The From option and To, along with the Select Date Type drop down list (with values Start Date and End Date) allow filtering and display of only those events that match with the specified time range, for example the events with Start Date between 01-January-2020 and 31-March-2020.
- c. The Show Patients With section allows filtering of Events that match with the filter criteria provided in the input box. Filter criteria can be defined based on Patient Identifier or the combination of Event Type (such as Diagnosis, Procedure, and so forth) and Event Name value(s). Multiple values can be provided in the above input box. Type ahead is provided in this text field. Only one filter type can be provided at a time for this filter.
- d. Click **Apply** to apply changes in the Show Event Type, Time Scale, and Show Patients With sections, refresh the information in the events list table, and close the above sections.

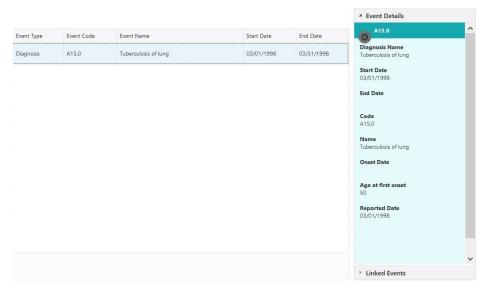


- Click Reset to restore all of the selections in the above sections to their default selection.
- f. Click Cancel to close the above sections without applying changes. The information displayed in the events list remains unchanged.
- 4. By default, the Latest Events Only toggle () is set to display only the latest date's events that are selected for display, wherein an event is defined by the combination of Event Type and Event Code. If a particular event has multiple instances on the same date (for example, Observation of Blood Pressure is performed multiple times in a day) then all of such instances are displayed. The toggle can be used to change the above filter and display all of the instances of the selected events.
- 5. Upon clicking a particular event record in the event list table, the collapsible/ expandable sections located at the right side of the events list table display the details of the patient and the selected event. Depending on the type of clinical event (such as Diagnosis, Medication, Specimen Collection, and so forth) the sections display the relevant details of the event using a unique background color for each section to provide visual cues about different types of events.
 - a. The Patient Details section displays the demographic and contact details of the selected patient.





b. The Event Details section displays the details of the selected event.

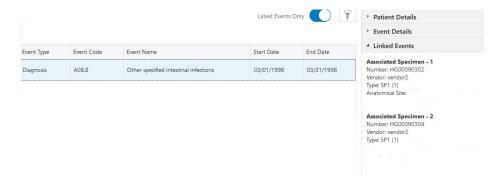


c. The Linked Events section is displayed for the clinical events which can be linked to other clinical events. This section displays details of all of the events that are linked to the currently selected event in the events list table. If a Diagnosis event is selected in the events list table then this section displays details of the Specimen Collection event(s) that are linked to the selected Diagnosis event.

For Medication, Procedure, and Observation events the linked Diagnosis event is displayed.

For Medication, Procedure and Observation events the linked Diagnosis event is displayed. For Medication events the linked Procedure event is also displayed.

For Test or Observation events the linked Specimen Collection event is also displayed.



6. Pagination available at the bottom of this screen allows navigation across multiple pages of the list. To directly navigate to a particular page, type the page number in the input box next to the label 'Page' and press 'Enter' key on the keyboard. However it is limited to the pages that are already loaded and mentioned in the hover over of the input box, for example the number between 1 and 21 in the image below.





- Click Back to Cohorts to return to the Cohort List screen.
- 8. Click Manage Criteria to go to the Cohort Builder screen.

View Patient Data

The Patient Viewer screen displays the clinical data of an individual patient.

To search for a specific patient:

1. In the search input box, (the input box with the text 'Search by patient ID...') provide the Patient Identifier.



Click the search icon () next to the input box or press Enter key on the keyboard.



The search for Patient Identifier is based on exact match and it is not case sensitive.

If the application finds a patient with matching Patient Identifier then the screen refreshes and displays the information about the patient, with the Patient ID and Name displayed just below the search input box. If no matching patient is found then a message about no matching results is displayed at the center of the screen.

- View Demographics
- View Clinical Data
- Filter Clinical Data
- View Details

View Demographics

To view demographic details of the patient:



1. Click on the triangular icon (ID label.

-) adjacent to the Patient

The demographics section expands to display the demographic attributes for the patient.

- 2. The Data Studio icon () provides navigation to the Oracle Parallel Graph AnalytiX (PGX) Data Studio environment.
- 3. Click the same icon to collapse this section.

View Clinical Data

Below the demographics section, the application displays clinical data of the patient in a tabular format in the center of the screen. By default, Diagnosis data is displayed.

To display other type of clinical data in the table, such as Test or Observation, Patient History, and so on:

1. Click the clinical data selection menu available on the left side of the screen.





Filter Clinical Data

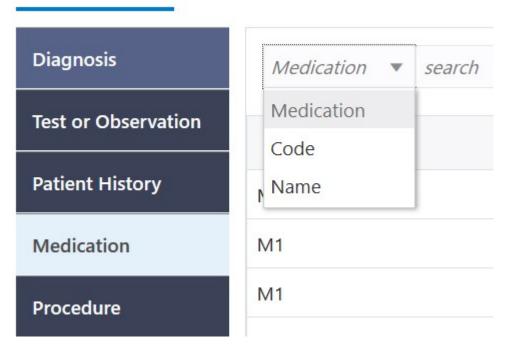
By default the tabular display lists the latest 100 records pertaining to the selected clinical event, sorted by the most recent events displayed first. You can further search or filter for any records for the patient in the system based on the values of specific attributes (columns in the table) and date range of clinical events.

To search and filter the records based on the value of attributes of clinical data:

 Select the column on which search is required. For example, in the Medication data, to search for the records with a specific medication, select Code or Name from the drop down list.



Clinical Data



- 2. In the search input box next to the column selection provide the search string.
- 3. Click the search icon ($^{\circ}$) or press Enter key on the keyboard.



In the tabular display, search is performed using the operator 'Starts With' and is case sensitive. For example, if the search string "Met" is provided for Code Name, then the tabular data is filtered to display only those records where Medication Name starts with the word "Met". A Medication Name starting with "met" would not be returned.

To filter the records based on dates of clinical events:

1. By default, the table display records without any filter on dates of clinical events, that is, the Time Scale is set to Auto. To filter the records based on the dates of clinical events, for example Medication Start Date or Medication End date, click on



2. In the time range selection dialog, select the desired date range and the clinical event's date column (such as Start Date or End Date) on which the filter is required.



For example, the filter on Medication can be specified to display only those records where Medication End Date is From 01-Jan-2018 To 31-Jan-2019. If only the From date is specified then the filter selects the records where the event date starts at the specified date, for example all records where Medication End Date is equal to or later than 01-Jan-2018.

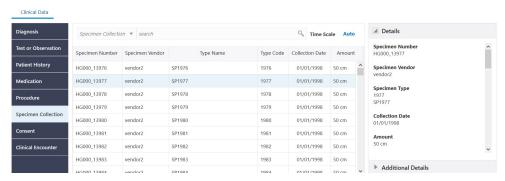
If only the End date is specified then the filter selects the records where the event date ends at the specified date, for example all records where Medication End Date is equal to or earlier than 31-Jan-2019.

View Details

For all types of clinical data (for example, Diagnosis, Procedure, Specimen Collection and so on) the application displays all of the clinical attributes available in Cohort Data Mart in the Details and Additional Details section.

To see the additional attributes available for any selected record in the tabular display:

- Select the record in the tabular display.
- 2. On the right side, click Details to expand it (if not already expanded).



- 3. The Details displays a vertical scroll bar depending on the number of attributes available in this section.
- 4. Similarly, below the Details section, click Additional Details to display the additional attributes available for the selected record.

The Additional Details section also displays details of all of the events that are linked to the event selected from the events list table. If a Diagnosis event is selected in the events list table then this section displays details of the Specimen Collection event(s) that are linked to the selected Diagnosis event. For Medication, Procedure, and Observation events, the linked Diagnosis event is displayed. For Medication events, the linked Procedure event is also displayed. For Test or



Observation events, the linked Specimen Collection event is also displayed.For Specimen Collection events, this section displays information about all of the Specimen Aliases and Specimen Metadata associated with the selected Specimen Collection record.

Logout

To log out from the application:

- 1. Click on the login user name at the top right corner of the screen.
- 2. Click on the **Logout** menu item.
- 3. To access online documentation, use the Help menu.





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