1 Introduction

1.1 Purpose of Document
The Release Content Document (RCD) communicates information about Oracle Health Sciences Omics Data Bank (ODB) v2.5 release.

1.2 Release Overview
ODB v2.5 release includes significant extensions in omics data types and clinically-related annotations.

2 Product Overview
ODB is a flexible, cross-platform omics data management system that delivers meaningfully biological contents and the extreme performance required by today’s translational research studies. Moreover, it is fully integrated with the Oracle Health Sciences Cohort Explorer (OHSCE), bringing both phenotypic and genotypic data to researchers and clinicians.

The data model supports reference omics information and annotation, as well as result data from each individual sample. ODB 2.5 is packaged with a set of data loaders for reference and result data, thereby reducing your adoption barrier. It also includes Oracle SecureFile as an optional file storage mechanism.

ODB 2.5 includes the following components with indication of new components added in this release:

- Reference and annotation tables for biological context
  - Reference genome build, genome annotation, and variant annotation from Ensembl
  - Protein information from Swiss-Prot and UniProt
  - Gene information from HUGO
  - Pathway information from Pathwaycommons
  - SIFT and Polyphone functional annotations
- Multi-version reference support (new)
- Selected clinical annotations from BIOBASE’s GenomeTrax database, including HGMD and Drug Targets annotations (new)

■ Result tables for individual sample data across multiple omics datatypes
  - Simple variation including SNV, small insertion, and small deletion
  - Gene expression including both single-channel and two-channel microarray data
  - Copy number variation
  - RNA sequencing data
  - Full genotype support including both reference and mutant allele (new)
  - Structural variant including chromosomal re-arrangement, large insertion, and large deletion (new)
  - Data for experiment and analysis details (new)

Data Loaders

■ Reference data loaders for
  - Ensembl (for example, genome definition, sequence variant annotations from dbSNP and COSMIC, and so on)
  - Swiss-Prot and UniProt
  - HUGO
  - Pathwaycommons
  - SIFT and Polyphen functional annotations
  - HGMD and Drug Targets from BIOBASE’s GenomeTrax (new)

■ Result data loaders for
  - VCF v4.1 including both simple sequence variant and large-size structural variant with improved flexibility and usability (new)
  - GVCF for variant and monomorphic reference genotype (new)
  - Tab-delimited format for both single-channel and two-channel microarray expression data
  - Mutation annotation format
  - Copy number variant format defined in The Cancer Genome Atlas (TCGA) project
  - RNA-seq format defined in TCGA project

SecureFile (optional)

■ Oracle SecureFile as an option for all data files that should be kept in a secure environment

3 Features
Cross-Platform Omics Data Model: ODB supports omics data management and analysis regardless of the scientific approach (for example, next generation sequencing, gene expression, and so on) and technology platform (for example, Illumina, Life Technologies, CompleteGenomics, and so on).

Multiple Species: ODB can model reference DNA, gene, and protein information from multiple species.

Cross Reference Omics Information: ODB can handle cross reference information for DNA source, gene, gene component, variant, protein, and protein component.

Support of Multiple Versions of Reference Genomes: ODB can organize your result data based on the version of reference genome the data was aligned with. This design improves data management efficiently and reduces interpretation error.

Integration with Clinical Data: ODB is fully compatible with the Cohort Data Mart (part of OHSCE v2.0.2.1) to let end users integrate both clinical and omics information in their cohort selection.

Monomorphic Reference Genotype vs. No-call Region: ODB differentiates monomorphic reference genotype from no-call regions to improve genotype characterization.

Annotation Management: Bringing annotation data to the results, ODB efficiently manages ever improving omics annotations.

Scalability: Result tables are partitioned by study or gene based on your data distribution and usage patterns.

Data Loaders: Easy-to-use command line data loaders with comprehensive instructions.

Table 1 lists the terms used in this document.

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>COSMIC</td>
<td>Catalogue of Somatic Mutations in Cancer</td>
</tr>
<tr>
<td>DNA</td>
<td>Deoxyribonucleic Aid</td>
</tr>
<tr>
<td>GVCF</td>
<td>Genome Variant Call Format</td>
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<td>HGMD</td>
<td>Human Gene Mutation Database</td>
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<td>HUGO</td>
<td>HUGO Gene Nomenclature Committee</td>
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<tr>
<td>MAF</td>
<td>Mutation Annotation Format</td>
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<tr>
<td>ODB</td>
<td>Omics Data Bank</td>
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<tr>
<td>OHSCE</td>
<td>Oracle Health Sciences Cohort Explorer</td>
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<td>RNA</td>
<td>Ribonucleic Acid</td>
</tr>
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<td>SNV</td>
<td>Single-nucleotide Variation</td>
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<tr>
<td>TRC</td>
<td>Translation Research Center</td>
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<tr>
<td>VCF</td>
<td>Variant Call Format</td>
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