

TITLE: The ICTS Dashboard: The GREGoR Data Model as an Operational Framework for Translational Genomics Workflows

AUTHOR BLOCK: Charles Hadley King (University of California, Irvine), Jonathan LoTempio (University of California, Irvine), Lightning Auriga (University of California, Irvine), Ivan De Dios (University of California, Irvine), Rebekah Barrick (University of California, Irvine), Seth Berger (Ambry Genetics), Emmanuele Delot (University of California, Irvine), Changrui Xiao (University of California, Irvine), Eric Vilain (University of California)

LEARNING OBJECTIVE: Examine practical implementation of schema-driven infrastructure for genomic, phenotypic, and biospecimen data integration.

Background: Large-scale rare disease genomics studies require systems capable of integrating diverse data sources. The Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR) Consortium developed a structured framework for these multimodal datasets across its five collaborating research centers. Each center submits validated quarterly data releases to the NHGRI AnVIL platform using the GREGoR-defined standards. To support all aspects of our UCI GREGoR workflow, from data intake to return of results, we developed the Institute for Clinical and Translational Science (ICTS) Dashboard. The ICTS Dashboard supports collaborative management of participant, phenotype, sequencing, genetic findings, and biospecimen metadata within translational genomics workflows.

Methods: The ICTS Dashboard is an open-source platform, governed by schemas generated directly from the GREGoR data model in support of rare disease genetics research. The platform uses a modular architecture: schemas, application-programming interfaces (APIs), database infrastructure, and user interfaces are maintained as independent but interoperable components. Database structures, validation rules, and interfaces were generated from the version-controlled GREGoR Data Model.

Results: The ICTS Dashboard architecture enables automated validation of submitted records, longitudinal phenotype tracking, biospecimen management, sequencing workflow coordination, reproducible data exports (for AnVIL upload), and integration of sequencing QA/QC reports through secure cloud-based APIs. The platform generated database structures, APIs, validation pipelines, and user interfaces directly from the version-controlled GREGoR data model releases, enabling continued alignment with evolving consortium standards. To date, it supports 37 GREGoR-derived schemas/tables, and hosts the metadata from 2,558 participants, 1,237 families, and 2,466 biospecimen sequences. Automated validation and export workflows enable the work of the analysts and research team as well as the quarterly data submissions to the NHGRI AnVIL platform by the bioinformatics team.

Conclusion: The GREGoR data model was designed to provide a blueprint for harmonization of datasets generated for rare disease genetics research. The modular, schema-governed infrastructure of the ICTS Dashboard provides an operational framework for other genomics programs to implement and adapt the platform for their own translational research workflows.

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