

# GTR Design

- Each record is a specific, orderable test from a specific laboratory.
- Each record has a stable, trackable accession and version.
- A record may be explicitly linked to one or more diseases and to one or more genome regions.
  - Supports large panels and arrays
  - Tests can be linked to specific mutations and defined genomic positions, not just gene names.
- A record may point to another record (a modified FDA test can simply refer to the FDA record)

## GTR Design

- Detailed discussion of schemas with GeneTests, ACLA HL7 eDOS, AMP Test Directory (new and old), McKesson Master Catalog for Diagnostics, DNAdirect/Medco Genetic Test Catalog, FDA Registry and Listing, CLIA database, Palmetto GBA, ACMG
- Correlation of GTR data items with CAP Molecular Pathology Checklist, CLIAC and SACGHS recommendations.
- Potential Bulk Upload discussion with ACLA, Quest, LabCorp, McKesson, DNAdirect/Medco.
- Automatic population of GTR from GeneTests.

NCBI



## GTR Design

- Experience supporting GeneTests
- Analysis of GeneTests web usage patterns
- Discussion with potential submitters and users

# Search for a Disease

Genetic Testing Registry - Windows Internet Explorer

http://dev.ncbi.nlm.nih.gov/sites/gtr@2.4/

Genetic Testing Registry

NCBI Resources How To My NCBI Sign In

**GENETIC TESTING REGISTRY**

All GTR **Diseases** Genes Tests Labs Clinics Resources

cardio

Search

Advanced

ABCC9: CARDIOMYOPATHY, DILATED, 10

ACTC1: CARDIOMYOPATHY, DILATED, 1R

ACTC1: Familial hypertrophic cardiomyopathy 11

ACTN2: CARDIOMYOPATHY, DILATED, 1AA

ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 1

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 10

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 11

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 12

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 2

**Genetic Testing Registry**

With an increasing number of genetic tests available, the National Library of Medicine is providing an online resource that will provide a centralized location for test information. This resource will have several key functions:

- Encourage providers of genetic tests to enhance transparency by publicly sharing information about the availability and utility of their tests;
- Provide an information resource for the public, including researchers, health care providers and patients, to locate laboratories that offer particular tests; and
- Facilitate genetic and genomic data-sharing for research and new scientific discoveries.

The overarching goal of the GTR is to advance the public health and safety of the United States by making information about these tests easily accessible to researchers, patients, health care providers, and consumers. This online resource that will provide a centralized location for test information.

Interacting with stakeholders—such as laboratory test developers, manufacturers, health care providers, patient and consumer groups, and researchers—will be a critical part of developing the GTR.

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Genetic Testing Registry is a service of the U.S. National Library of Medicine and the National Institutes of Health.

File last modified Tuesday June 15, 2010 09:22 EDT

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