Genomics, the NIH, and Health Informatics

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Deputy Director, NLM
NIH – Present at the creation of …

• Genomics

• Health/Medical/Biomedical Informatics
  – 1966 – NCI: Computer-assisted analysis of oligonucleotides
  – 1972 – NLM: Informatics training

• Beginning of convergence
  – 1988 – NCBI established by law at NLM;
    Office of Human Genome Research established by NIH Director
Research/Healthcare Benefits of EHRs = Rationale for Common Standards = Perpetual Informatics R&D Agenda

• Research data captured as a by-product of patient care
• More rapid translation of research findings into improved patient care & public health
• Best evidence delivered to patients & health professionals when, where, & how needed
Do more of what works, e.g.,

- Rapid release of research data, standards, results in public, maintained, connected databases
- Standardization & “analysis” at the data source to reduce cost, improve quality
  - e.g., NISO Z39.96 - JATS: Journal Article Tag Suite
- Use/improvement of existing standards for clinical research & genetics
  - e.g., SNOMED CT, LOINC, RxNorm, HL7
- New standards ONLY when needed, e.g., RefSeqGene
- Investigator-initiated informatics research
  - e.g., Personally-controlled health records, flexible anonymization techniques, explaining probability, NLP
New IHTSDO Policy Facilitates Inclusion of SNOMED CT Terms and Identifiers in International Research Databases, Other Health IT Standards

Copenhagen, Denmark: January 28th, 2011.

*International Genetic Databases to be early beneficiary*

The International Health Terminology Standards Development Organisation (IHTSDO) has announced a new policy to enable free use of English-language SNOMED CT terms and identifiers in international research databases, in complementary health IT standards, and in other projects and resources available worldwide. The new policy allows SNOMED CT to serve as a standard vocabulary for key data elements and value sets in international resources that accept input from - and are used in – both IHTSDO Member and non-Member countries.
Welcome to the UTS

The UMLS Terminology Services (UTS) allows you to:

- Request a UMLS Metathesaurus License and create a UTS account
- Search and display content from UTS Applications including:
  - Metathesaurus Browser
  - Semantic Network Browser
  - SNOMED CT Browser
- Download data files including:
  - UMLS Knowledge Sources
  - RxNorm weekly and monthly updates
  - SNOMED CT
  - CORE Problem List and Route of Administration Subsets of SNOMED CT
- Query data remotely via Web Services (see API Documentation)
- Complete UMLS Annual Report and SNOMED CT® Affiliate Reports

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The Genetic Testing Registry (GTR) is currently in development, with a projected launch in the latter part of 2011. Once operational, GTR will provide access to information about genetic tests for inherited and somatic genetic variations, including newer types of tests such as arrays and multiplex panels. GTR information about tests primarily will be based on voluntary data submissions by test developers and manufacturers.

A key part of the GTR development process has been engagement with stakeholders—such as genetic test developers, test kit manufacturers, health care providers, patients, researchers, and relevant federal agencies—for their insights on critical data elements to include in the database and the best ways to display test information. Further information about that is available on NIH’s GTR policy website, at http://oba.od.nih.gov/GTR/gtr_intro.html; the policy website includes background information, FAQs, Federal Register notices, public comments on the June 11, 2010 Request for Information, a transcript of the November 2, 2010 public meeting, and other information.

The GTR design is continuing to evolve based on feedback from advisory groups, stakeholders and other interested parties. However, it is sufficiently developed that NCBI is making available working documents about the design for further comment. Please see the README file for context and explanations of the other documents.

Send a comment
MedlinePlus Connect: Linking Electronic Health Records (EHRs) to MedlinePlus Health Information

Overview

MedlinePlus Connect is a free service of the National Library of Medicine (NLM) and the National Institutes of Health (NIH). This service allows electronic health record (EHR) systems to easily link users to MedlinePlus, an authoritative up-to-date health information resource for patients, families and health care providers.

How it Works

MedlinePlus Connect accepts requests for information on diagnoses (problem codes) and medications. It is available as a Web application or a Web service.
COMING SOON:
Lab test links in MedlinePlus Connect

LOINC:3067-6  Trypsinogen I Free [Mass/volume] in Serum or Plasma

Neonatal cystic fibrosis screening

Neonatal cystic fibrosis screening is a blood test that looks for increased levels of immunoreactive trypsinogen (IRT), an enzyme produced by the pancreas.

The test is performed on newborns to screen for cystic fibrosis (CF). Cystic fibrosis is an inherited disease that causes thick, sticky mucus to build up in the lungs and digestive tract.

How the Test is Performed

A sample of blood is either taken from the bottom of the baby's foot of a vein in the arm. A tiny drop of blood is collected onto a piece of filter paper and allowed to dry. The dried blood sample is sent to a lab for analysis.

How to Prepare for the Test

There is no special preparation needed.
Effects of CYP2B6 Genetic Polymorphisms on Efavirenz Pharmacokinetics

This study is ongoing, but not recruiting participants.

First Received on November 27, 2007. Last Updated on May 25, 2010  History of Changes

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<td>ClinicalTrials.gov Identifier:</td>
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Purpose

1. To see how the liver breaks down efavirenz by an enzyme called CYP2B6. It is suggested that when Efavirenz is taken repeatedly it may increase the amount of CYP2B6 in your liver and thus speed up your liver’s ability to get rid of efavirenz from your body. This may render efavirenz and other medications ineffective.

2. To acquire further participants with the aim to get statistically significant outcomes.
CLCNKA (Ka Renal Chloride Channel[CIC-Ka]) Polymorphism Effects on Hypertrophy Regression

This study is not yet open for participant recruitment.
Verified on January 2011 by Washington University School of Medicine

First Received on January 10, 2011. Last Updated on January 19, 2011  History of Changes

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Purpose

This study will consist of middle-aged Caucasian non-failing subjects with high blood pressure who are homozygous for a gene that confers increased risk of developing heart failure, the Glycine 83 variant of the Ka renal chloride channel (CIC-Ka Gly/Gly 83), or middle-aged Caucasian non-failing hypertensive subjects who lack the heart failure risk gene, the wild-type Arginine 83 Ka renal chloride channel (CIC-Ka Arg/Arg 83). Subjects on standard therapy for high blood pressure with an angiotensin converting inhibitor (ACEI) or angiotensin receptor blocker (ARB) will be randomized to additional treatment with eplerenone.