

Clinical and Translational Science Ontology  
Workshop 4/25/2012

# An Ontology for Informed Consents and Other Research Permissions

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# Outline

- The need for a Research Permissions Management System (RPMS)
- The need for an Ontology
- The RPMS pilot
- Current development
- The permissions ontology overview
- RPMS2 status
- Lessons learned

# Research Permissions Management System (RPMS): The Case For Change



## Current Environment

- **Paper-based** consents and permissions
- **Permissions data are unavailable** for analysis
- The number of patients who are generally **favorable to research participation is unknown**
- **Inconsistent process and semantics** in obtaining permissions across multiple hospitals
- **Poor comprehension** and satisfaction in subjects providing permission
- Research volunteer **subject lists are localized**



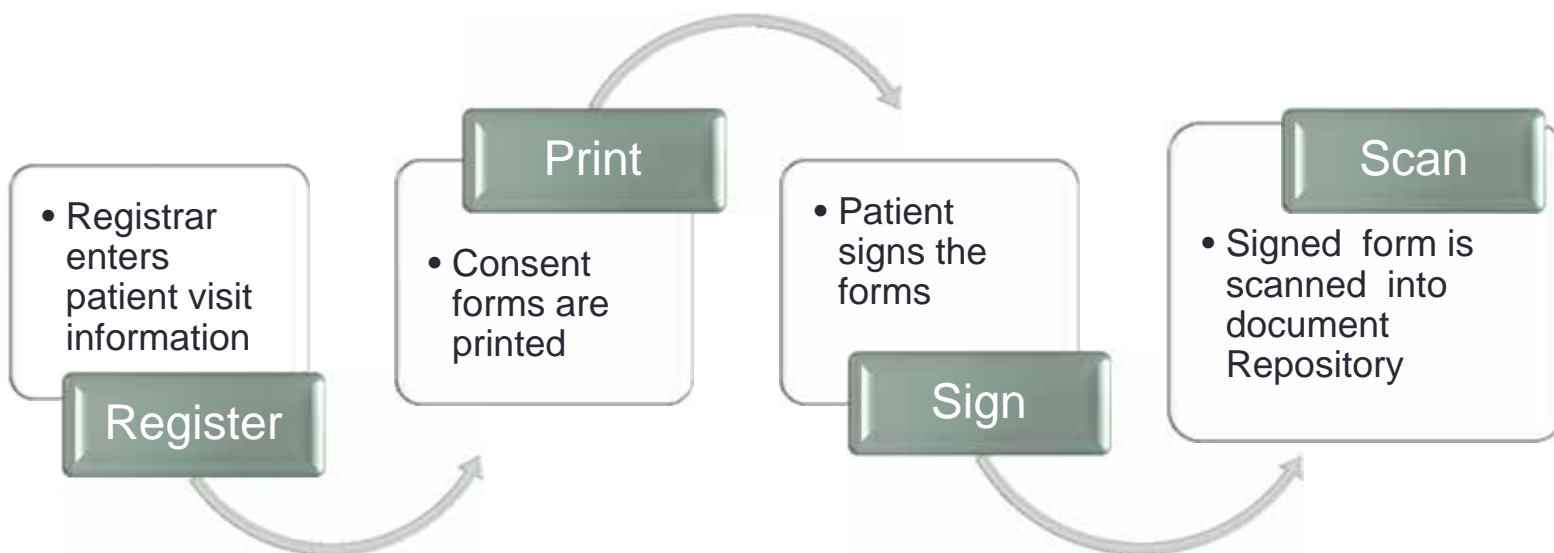
## New Permissions Management

- **Electronic recording** of consents and permissions
- **Access to permission** decisions for research
- Common **permissions terminology**
- **Facilitating research while enhancing compliance with patient wishes**
- Rich **educational opportunities** about the research process
- **Patients will have easy access** to their permission decisions (patient portal)
- **Tracking** of informed consents

# Informed Consents vs. Permissions

- Informed consent for research
- Consent to treat: regular patient registration
- Research permissions (can be opt-in or opt-out)
  - To contact for research
  - Use of tissues o/w disposed

# Typical Registration Process on Admission



Topaz Signature pad used in some hospitals

# The Need for an Ontology

- Standardize across institutions
- Make permissions and consent assumptions explicit
- Allow sharing of consistent data
- Allow analysis across institutions
- Reuse in other projects

Consent to the Disposal of any Blood, Body Fluids or Tissue which may be removed during my treatment. I consent to the disposal of any blood, body fluids or tissue which may be removed during my treatment. \_\_\_\_\_

**Permissions to Consent to Research Studies**  
I agree to be contacted about future research studies at MUSC for which I may be eligible. I understand that if I do not want to be contacted about future research studies, I need to check the box above again.

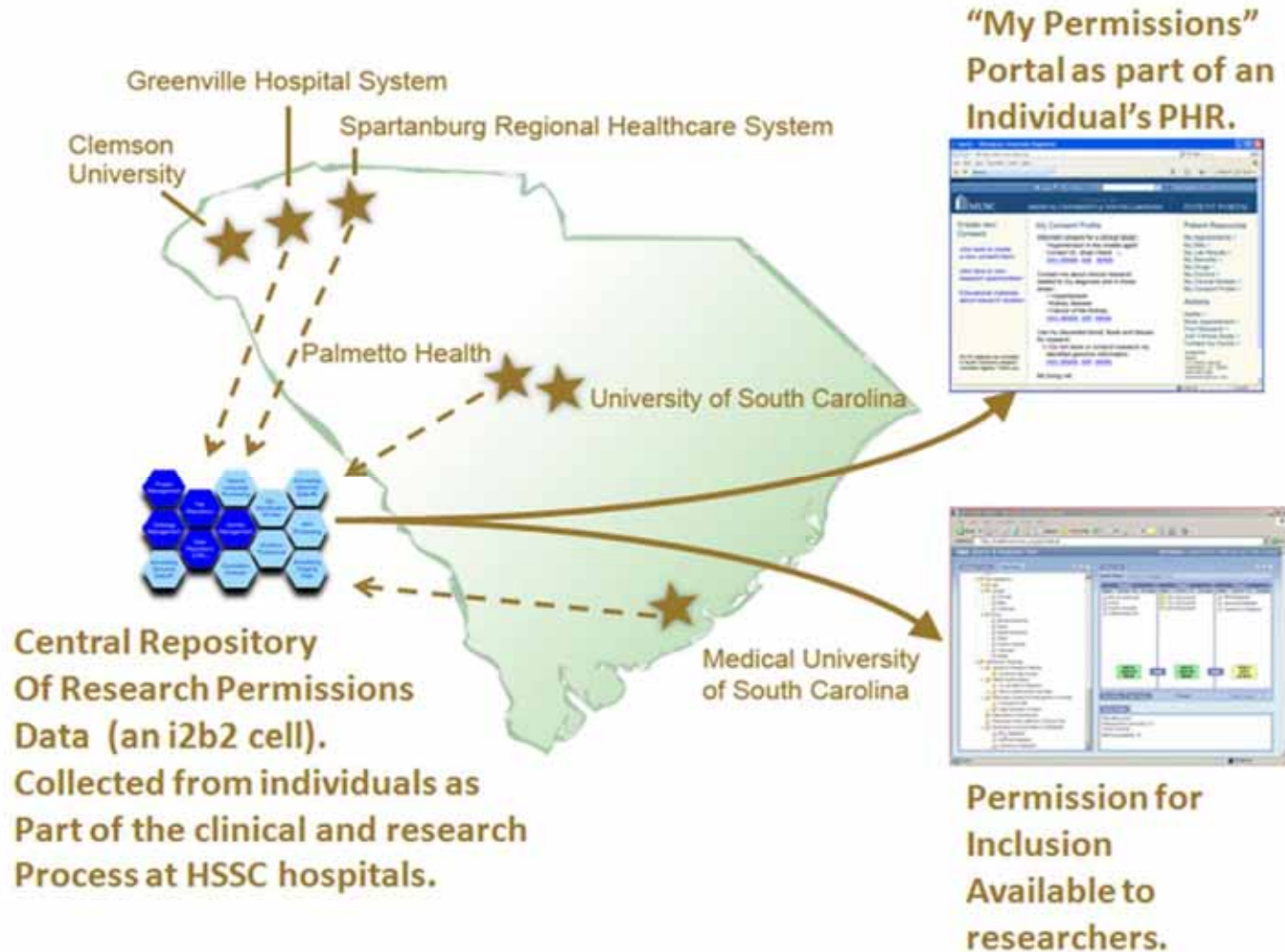
**Informed Consent for Disposal: I consent to the disposal by hospital authorities of any blood, tissue, body parts, fluids, or foreign objects as may be removed during the course of the procedure, I also consent to hospital authorities, at their discretion, saving for research or educational purposes all or part of the blood and/or tissue normally discarded. I understand that saved blood and/or tissue will not be stored, shipped, or otherwise distributed.**

If you do not give permission for any of the above, write through the parentheses and initial in the left of the paragraph.

**Agreement to Participate**  
I have read this consent form or it was read to me. I understand the information in this consent form and I have had my questions answered. I voluntarily agree to take part in this research.

1. May we collect and use your blood, fluid, urine (sp), and health information for use in future research?  Yes  No  
2. May we contact you in the future if we need additional samples or information?  Yes  No  
3. May researchers contact you in the future to invite you to participate in their research studies?  Yes  No

# A Research Permission Management System – GO grant Concept



# Permission as a Searchable Attribute in i2b2

The screenshot shows the i2b2 Query & Analysis Tool interface. On the left is a tree view of the ontology with categories like Demographics, Diagnoses, and Research Permissions. The 'Research Permissions' category is expanded, showing 'Future contact for Research'. In the center, a text box reads: 'Show me how many males over 65 have given permission to be contacted for research'. Three red arrows point from this text to the query builder: one to 'Male' in Group 1, one to '>= 65 years old' in Group 2, and one to 'Future contact for Research' in Group 3. The query builder shows three groups connected by 'AND' operators. Below the groups are buttons for 'one or more of these' and 'drop a term on here'. At the bottom, there are 'Run Query' and 'New Query' buttons, and a 'Query Status' section.



# RPMS Project – 5 Distinct tracks

with several partners:

- Discovery Phase
- Permissions Ontology Creation – **SAIC** then MUSC
- Duke ELSI Process and creation of the General Research Permissions Form
- Clemson Permissions research project and human subjects “Permissions UI Laboratory”
- Creation of the RPMS application initiated with **SAIC** and **Recombinant** – later brought in-house

Ontology



ELSI

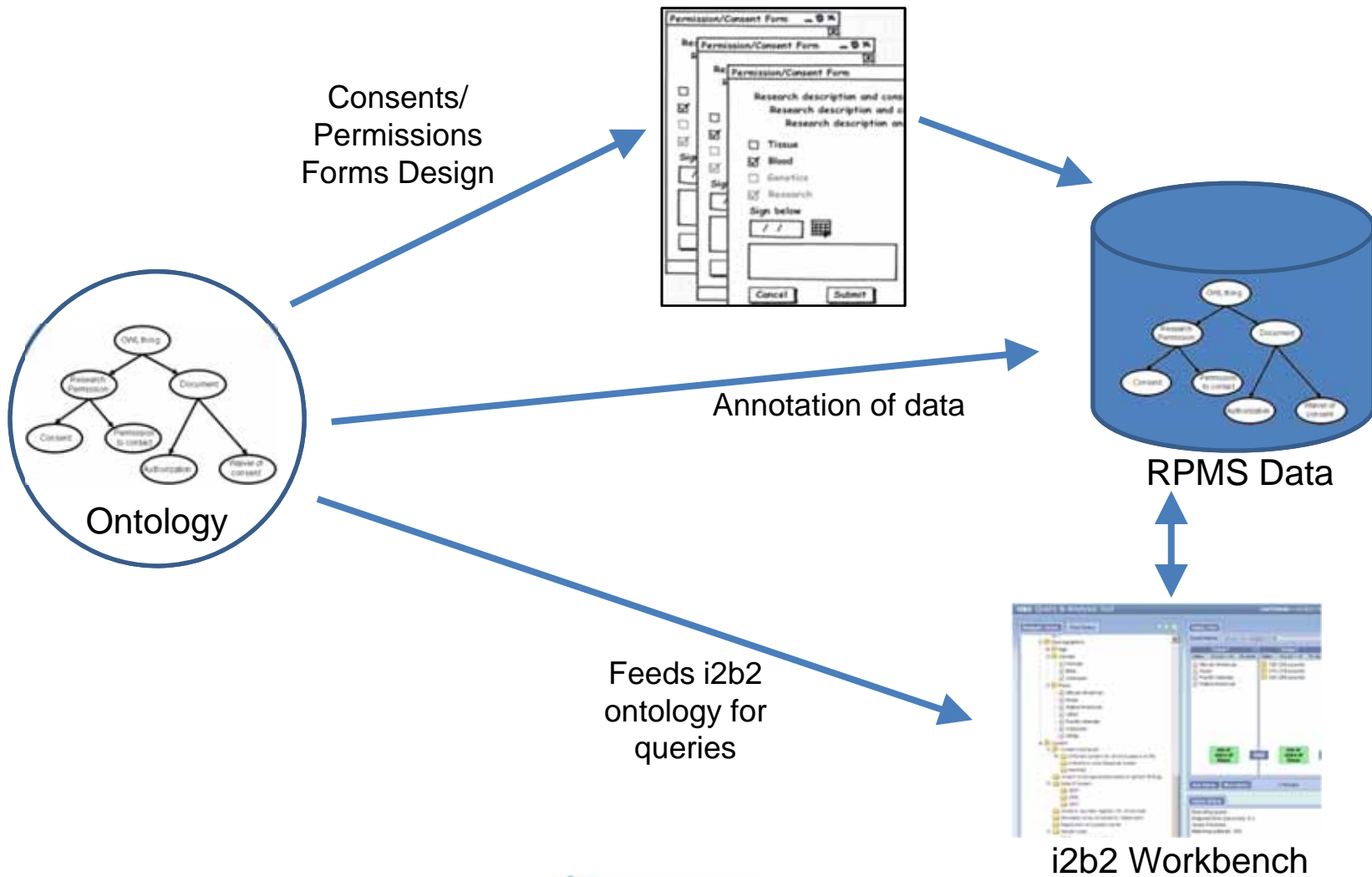


Clemson  
UI Lab



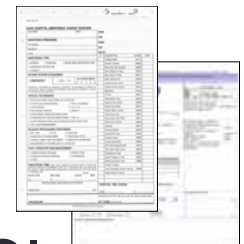
Design of the end-to-end RPMS application for Pilot

# Ontology Use Cases in RPMS

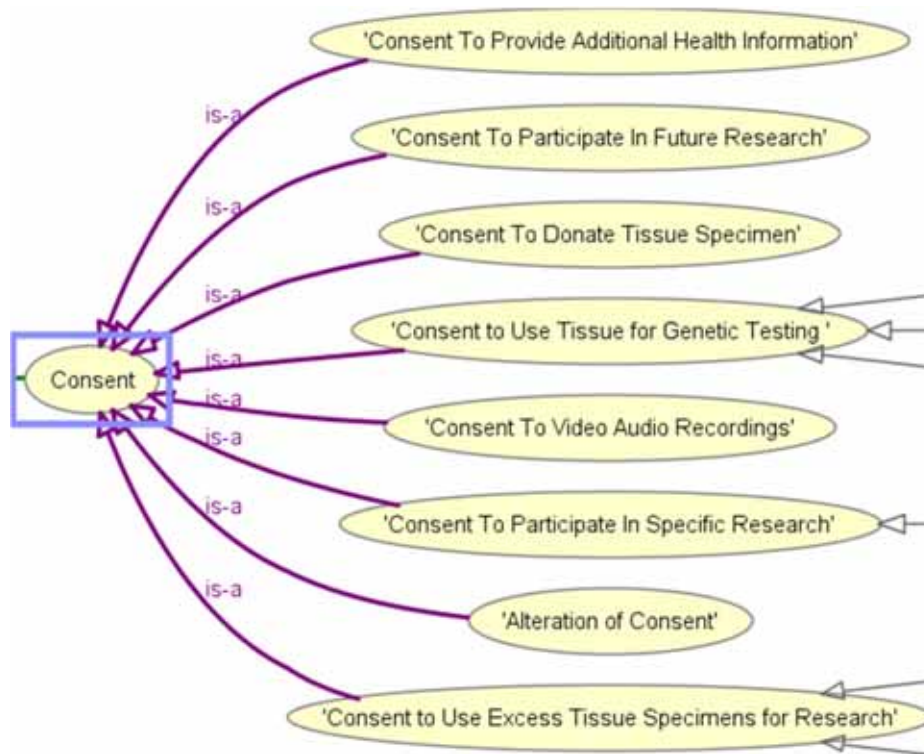


# Ontology Content: Domain Analysis

- Initial work done by collaborators from HSSC, Clemson and industry consultants (**SAIC**)
- Analysis of the permission processes at four institutions
- Reviewed various hospital forms, federal regs, and websites
- Reviewed national standards: HITSP, SNOMED, NCI
- Used NCI thesaurus for root classes
- Independent track from software architecture



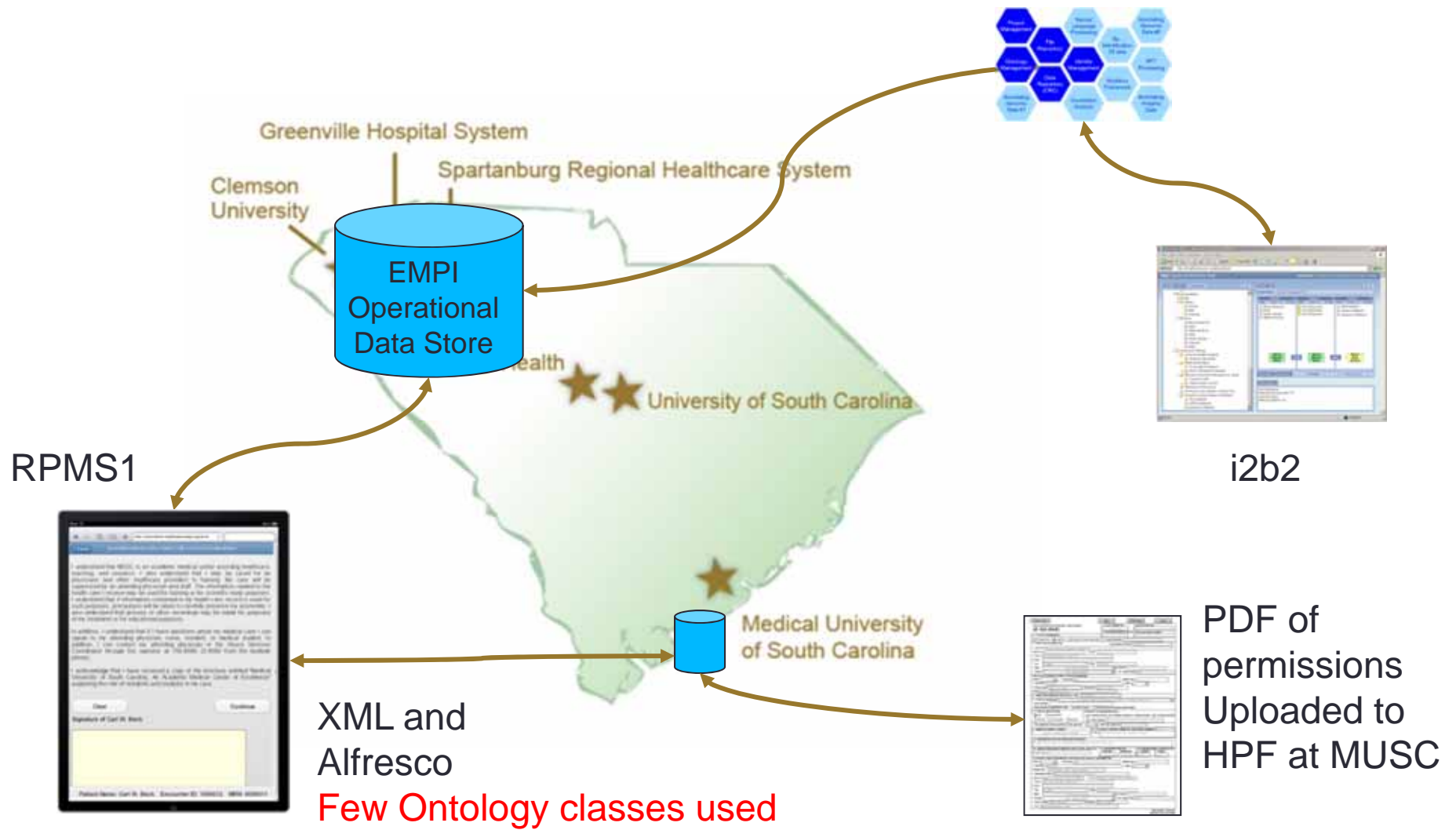
# RPMS Pilot Ontology Content



188 Classes  
(80 from NCI classes)

129 Properties (or interrelationships between classes)

# RPMS1 Pilot Architecture

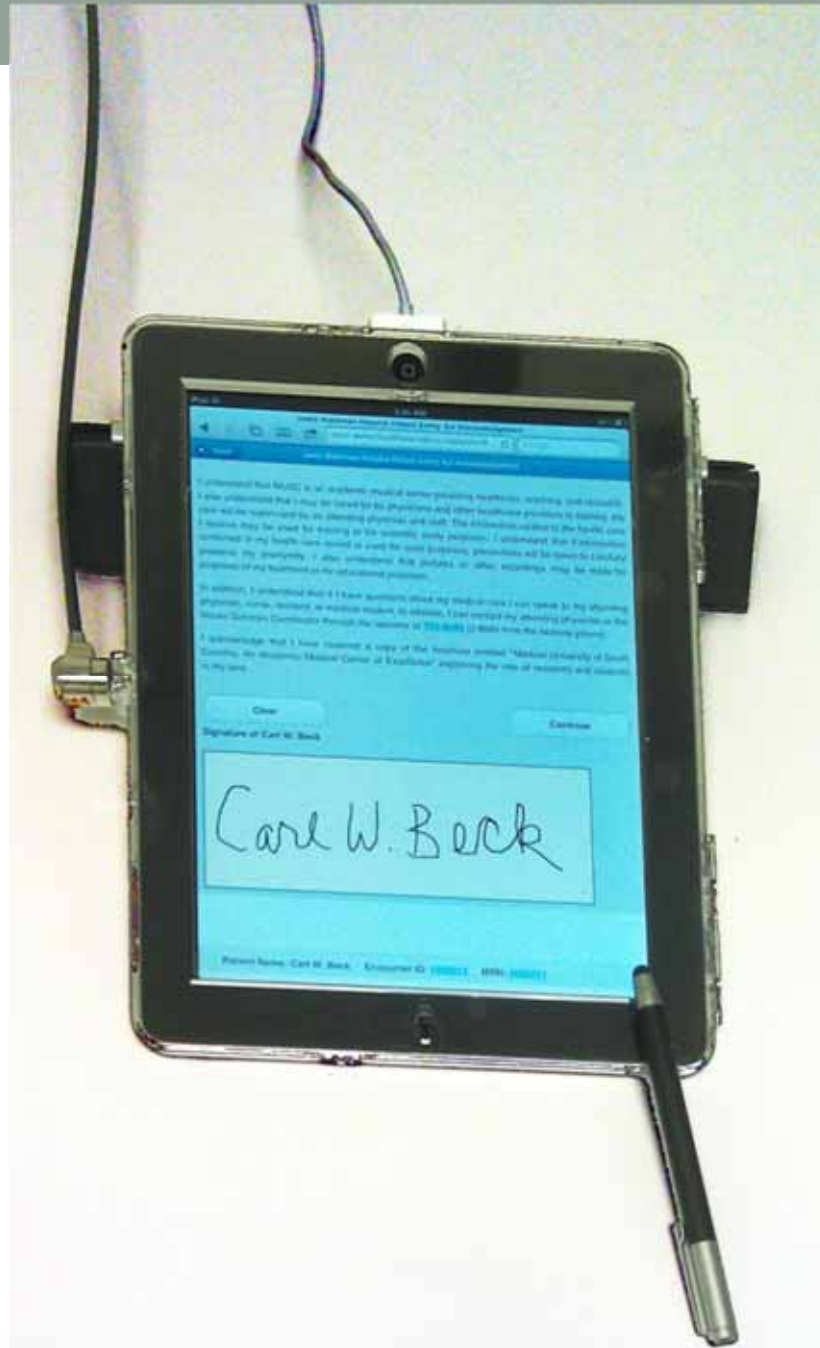


# RPMS1 Pilot Live at MUSC





Working pilot being tested at MUSC in select Patient registration areas.

# RPMS1 pilot: hardened iPad, accessories and stylus



# RPMS1: RPMS Generated Forms

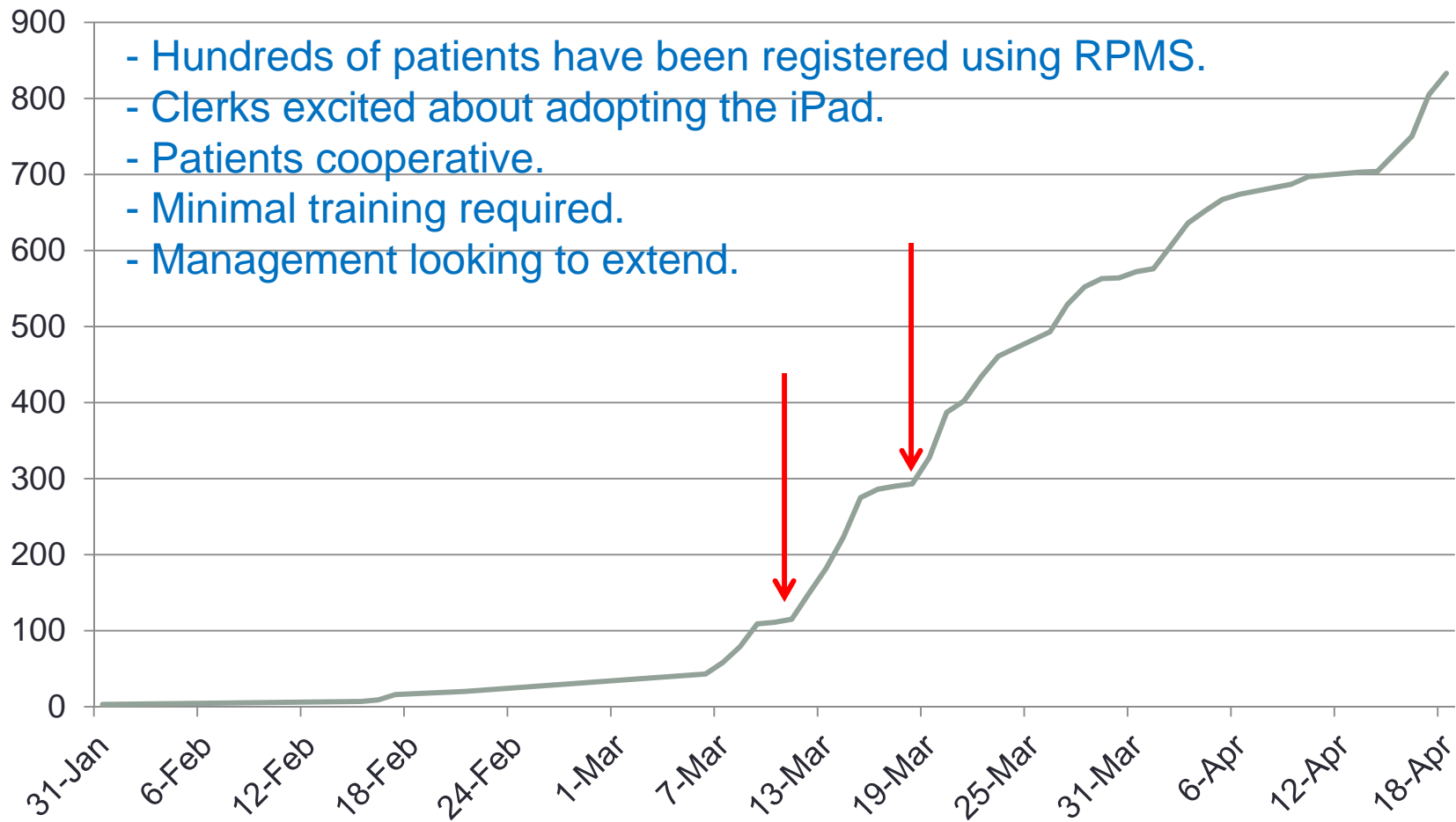
 <p><b>MUSC Health</b> UNIVERSITY Consent for Medical Treatment</p> <p>Form OTC0227 Rev. 5/11 Page 1 of 2 Version Date: 5/11</p>	<p>PATIENT IDENTIFICATION LABEL</p> <p>P-28437284 DOB: 01/01/2004 B F TEST ALEX ALEX Addr: 83 1912 M-001A2090</p>
<p><b>Your Permission is Needed</b> I (as patient, parent, guardian, spouse, guarantor, or other responsible party) consent to and authorize medical treatment and diagnostic procedures which may be ordered by my doctors and performed by MUSC Medical Center ("The Hospital"). I consent to have blood drawn and to be tested for infectious diseases, including but not limited to syphilis, AIDS, hepatitis and testing for drugs if my doctor orders these tests. <span style="float: right;">Agreement: agree</span></p> <p><b>Your Agreement to Pay for Treatment</b> I assign and transfer to The Hospital and / or my doctors all rights, and interest in benefits I may have under any insurance policy I may have, including but not limited to hospitalization, medical, third party liability insurance coverage, workers compensation benefits, or benefits paid by Medicare or Medicaid. This assignment is intended to include any interest in benefits that I may have relating to this date of service as well as any prior dates of service. I direct that any insurance company or other party make payment of such benefits to The Hospital or my doctor. I authorize The Hospital and / or my doctor to collect benefits from any responsible third party through whatever means may be deemed necessary, and to endorse benefit checks made payable directly to me.</p> <p>I understand that by signing below, I promise to pay all Hospital and doctor charges at the standard rates and terms of The Hospital or doctor including all charges not covered by my insurance or any other party. I promise to pay the patient's account at the rates stated in The Hospital's price list (known as the "Charge Master") and / or the doctor's fee schedule in effect on the date the charge is processed for the services provided. I understand that there will be a separate charge for the doctor and other professional services, but understand that The Hospital may bill for some professional fees.</p> <p>I understand that The Hospital and MUSC Physicians file insurance as a courtesy to me, and agree that I am responsible for payment of my bill, including any charges that are denied by my insurance or any other responsible party. I understand that care that is experimental as determined by my insurance company may not be covered and that I will be responsible for those charges. I agree that if this account is not paid, it may be turned over to a collection agency or attorney, and I must pay the amount due plus all costs of collection, including reasonable attorney's fees.</p> <p>I understand that any overpayments made to The Hospital or MUSC Physicians may be applied to any outstanding balance I may have with The Hospital or MUSC Physicians and will not be refunded to me unless all of my outstanding bills with The Hospital or MUSC Physicians are paid in full.</p> <p>I understand that if I am unable to pay my bills, I may speak with a Financial Counselor to determine whether I qualify for assistance or for a discount. I may call (843) 792-2311 for information or questions about my hospital bill and (843) 792-6200 for information about my doctor bill. <span style="float: right;">Agreement: agree</span></p> <p><b>Release of Information to My Referring Doctor</b> My referring doctor is (please print full name): <u>JOHN CUSACK</u> City: <u>Rock Hill, SC</u> My Primary Care Physician is (please print full name): <u>COSNER, ENF, RUTH S</u> City: <u>Charleston, SC</u></p> <p><b>Medicare Patient Certification (Medicare patients only)</b> I hereby certify that I have provided information about all insurance coverage available to me, including liability or worker's compensation insurance and that the information provided is correct and complete. I hereby authorize The Hospital and / or my doctors to release to the Social Security Administration, its intermediaries, or carriers any information needed for this or a related Medicare claim. I hereby authorize the payment of benefits to The Hospital or my doctors. <span style="float: right;">Agreement:</span></p> <p><b>Retention / Disposal and Use of Blood, Body Fluids, or Tissue</b> I understand and agree that any blood, body fluids or tissues normally removed from my body by MUSC in the course of any diagnostic procedures, surgery, or medical treatment that would otherwise be disposed of may be retained and used for research, including research on the genetic material (DNA) or other information contained in those tissues or specimens. I acknowledge that such research by MUSC may result in new inventions that may have commercial value and I understand that there are no plans to compensate me should this occur, regardless of the value of any such invention. I understand that any research using these leftover specimens or tissues will be done in a way that will not identify me. I also understand that if I do not want research to be done using my leftover blood, body fluids or tissue, I need to check the box shown below. If I have questions, I should call (843) 792-8300.</p> <p style="text-align: center;"><small>all other consent consentform</small> <span style="float: right;"><small>OTC 790227 Rev. 5/11</small></span></p>	

 <p><b>MUSC Health</b> UNIVERSITY Consent for Medical Treatment</p> <p>Form OTC0227 Rev. 5/11 Page 1 of 2 Version Date: 5/11</p>	<p>PATIENT IDENTIFICATION LABEL</p> <p>P-28437284 DOB: 01/01/2004 B F TEST ALEX ALEX Addr: 83 1912 M-001A2090</p>
<p style="text-align: center;"><input type="checkbox"/> I DO NOT agree to have my tissue or blood used for future research studies.</p> <p><b>Permission to Contact for Research Studies</b> I agree to be contacted about future research studies at MUSC for which I may be eligible. I understand that if I do not want to be contacted about future research studies, I need to check the box shown below. <input type="checkbox"/> I DO NOT agree to be contacted about future research studies</p> <p><b>Admission Photographs</b> I agree that my photograph may be taken for purposes of identifying me, or providing treatment to me. This photograph may become part of my medical record and may be disclosed if copies of my medical record are disclosed. <span style="float: right;">Agreement: agree</span></p> <p><b>Responsibility for Personal Items</b> I understand that The Hospital is not responsible for valuable items which I bring with me. I understand it is my responsibility to send any valuable items (such as medications, money, jewelry, electronics, etc.) home for safe keeping. Any items left at the hospital in excess of 30 days will be disposed of. <span style="float: right;">I understand</span></p> <p><b>I received a copy of the MUSC "Notice of Privacy Practices".</b> <span style="float: right;">Agreement: agree</span></p> <p>I certify that I have read or have had read to me this consent and agree to its terms. I also certify that I am the patient, or am duly authorized by the patient, or am duly appointed to sign this agreement. I accept and understand its terms.</p> <p style="text-align: right;"><i>Alex Test</i></p> <p>03/19/2012 5:13 PM Date and Time</p> <p style="text-align: right;">Signature of Patient Print full name <u>ALEX TEST</u></p> <p style="text-align: center;"><i>Randall Alexander</i></p> <p style="text-align: right;">Signature of Witness Print full name</p> <p>Advance Directives information given: Yes</p> <p style="text-align: center;"><small>all other consent consentform</small> <span style="float: right;"><small>OTC 790227 Rev. 5/11</small></span></p>	



# RPMS Pilot Status

## RPMS: Number of Patients Registered



- Hundreds of patients have been registered using RPMS.
- Clerks excited about adopting the iPad.
- Patients cooperative.
- Minimal training required.
- Management looking to extend.

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The Project is not over!



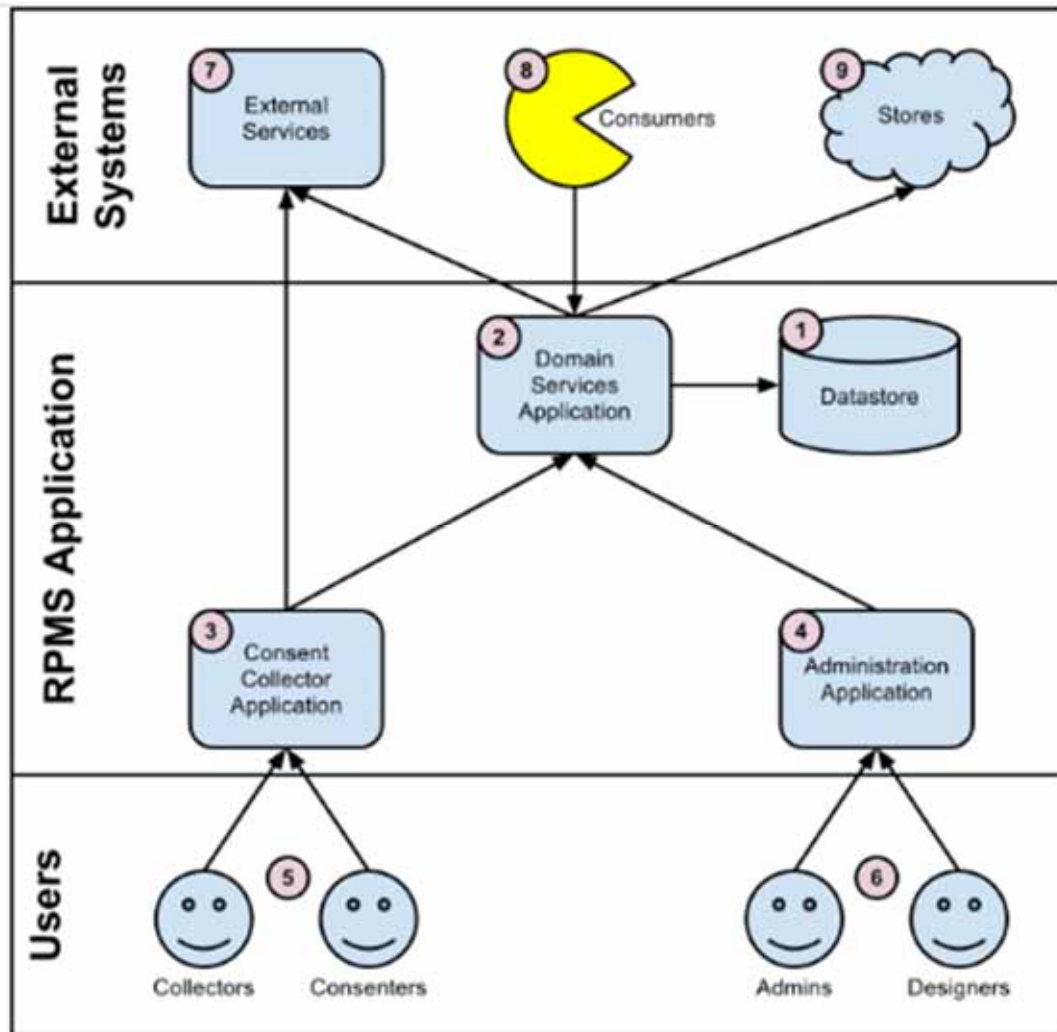
# RPMS2: Functional requirements

- Include RPMS1 functionality
- Add **Informed Consent** and **HIPAA Authorizations**
  - + general research permissions form and text from Duke ELSI group
- Include ability for **rich media** content (e.g. video)
- Architecture: **Ontology-based** (graph database at its core)
  - Nodes and relationships are flexible, extensible
  - Handles large and complex datasets (scalable)
  - Indexes to transverse relationships (performance much faster than RDBMS)
- Include consent forms **authoring module** tied to ontology
- Create a working application and publish it as the foundation of a community **open source** project

# RPMS2 Exploratory Phase

- Looked at VIVO, eagle-i
  - Established good collaborations with UF (used v1 code)
- Difficulty in applying to workflow in RPMS
- Collaborated with UCSD
  - Consent project at CTSA IKFC (Aziz Boxwala / Adela)
  - Teamed up on ontology foundation to avoid duplication
- Decided to balance between perfect ontology and pragmatic design
- Ontology development is hand-in-hand with architecture

# RPMS2: Architecture



Platform

Neo4j DB (graph db)

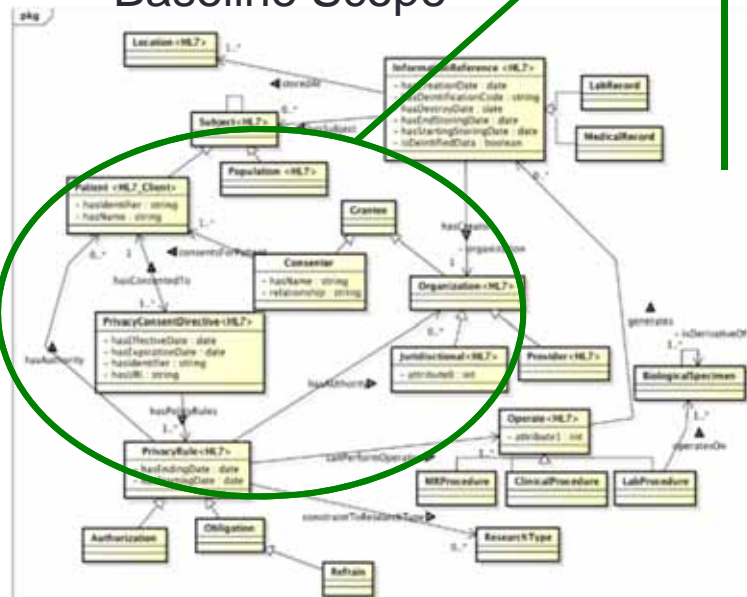
Clojure / JSON

Consumes OWL ontology

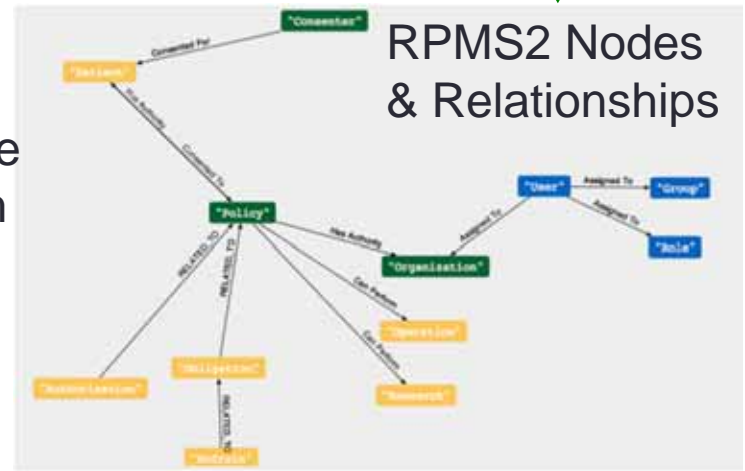
# RPMS2 Ontology Basis

RPMS2 graph structure extends UCSD's (based on HL7 Security and Privacy Ontology). Adds types and relationships needed for new requirements.

Baseline Scope



UCSD Ontology (Adela Grandio)



Shares some classes from original ontology

# Basic elements of informed consent (from 45 CFR 46) – to serve as IRB consent template

1. The study involves research
2. Risks
3. Benefits from research
4. Alternative options/treatments
5. Confidentiality of records
6. Availability of treatment for adverse results/compensation
7. Contact information
8. Voluntary participation
9. Other...

The ontology defines a well-formed consent.

# RPMS2 – Work in Progress

- Include RPMS1 functionality
- Add Informed Consent and HIPAA Authorizations
  - + general research permissions form and text from Duke ELSI group
- Include ability for rich media content (e.g. video)
- Architecture: Ontology–based (graph database at its core)
  - Nodes and relationships are (flexible, extensible)
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- Include consent forms authoring module
- Create a working application and publish it as the foundation of a community open source project



# Lessons Learned

- Ontology development should go hand-in-hand with software architecture design (e.g. VIVO/eagle-i)
  - No turn-key methodology for building ontology-based apps
  - Expertise has been hard to come by
- Pragmatic approach: don't get too hung up on ontology content – keep milestones and stakeholders in mind (we need to show a win and there is a real need for a functioning application)

# Acknowledgements

## RPMS core team

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## Duke ELSI

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Lawrence (Doc)  
Muhlbaier, PhD  
Kevin Weinfurt, PhD

## Clemson University

Anand Gramopadhye, PhD  
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And by



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Questions ?????