Kintalk/LSSN Collaboration to Facilitate Research on Screening for Lynch Syndrome

PI: Aparajita Singh, MD, MPH
Division of Gastroenterology

DISCLOSURES : None

Co-I: Amie Blanco, MS, LCGC
Director, UCSF Cancer Genetics and Prevention Program

DISCLOSURES: Spouse is an employee of Genentech

Co-I: Cecelia Bellcross, PhD, MS,CGC, LSSN Board

DISCLOSURES : None


Lynch Syndrome
Screening Network
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Promoting Universal Lynch Syndrome Screening
The Lynch Syndrome Screening Network promotes universal tumor screening (UTS) of all
"Empowering Families With Hereditary Cancers Through Communication and Education"
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Kintalk can help you understand
Hereditary Colon Cancer

Funds

- CGA - $20,000
  - Rebuilding database
- UCSF - $10,000
  - Annual hosting fee
- Omni Health Media - $7500
  - Data transfer & reporting tools

Overarching Goal

- To facilitate the multi-institutional collection of data on universal Lynch Syndrome screening, to evaluate screening and testing strategies and outcomes

Aim 1

Revise & rebuild LSSN database

Aim 2

Facilitate data collection and transfer by LSSN members

Aim 3

Answer initial questions, support further research by LSSN members

Database Content

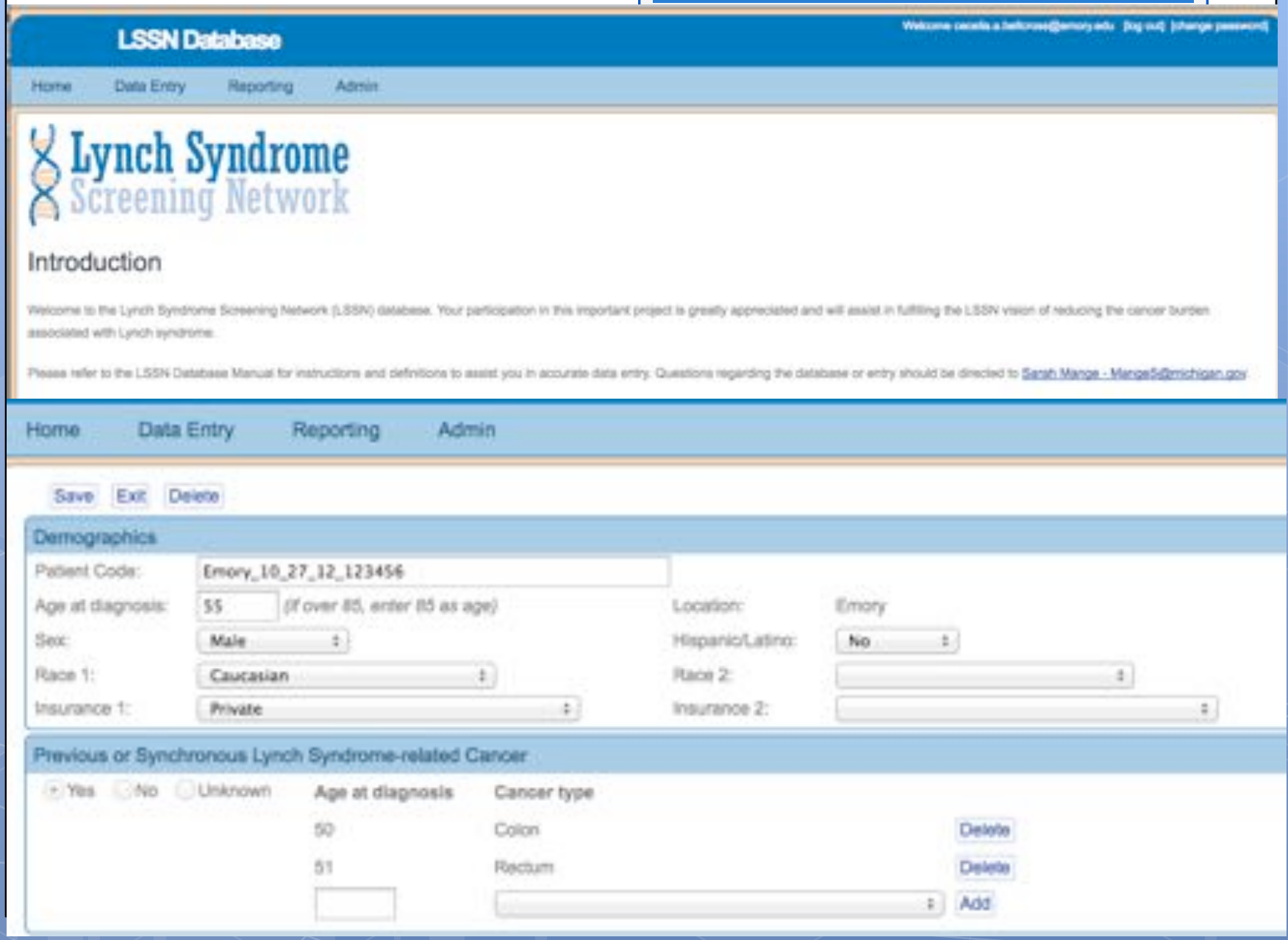
- Demographics: age, gender, race, insurance
- Index Cancer
  - Colon – site, specimen type, pathology
  - Endometrial – specimen type, pathology
  - Other LS-related cancers
  - TNM Pathological stage
- Tumor Screening results – MSI, IHC, BRAF methylation
- Previous CA or diagnosis of Lynch syndrome

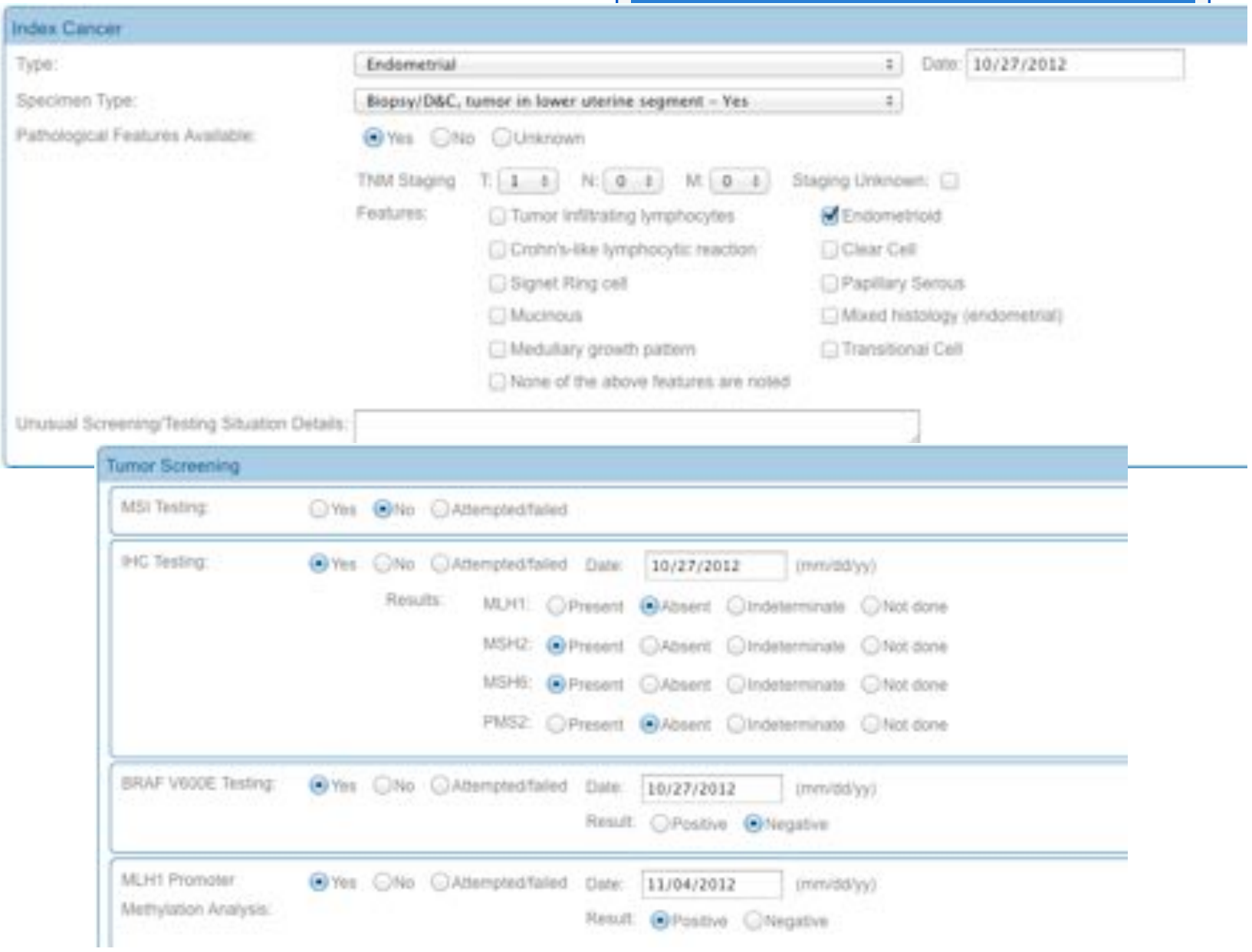
Database Content – Screen Positive

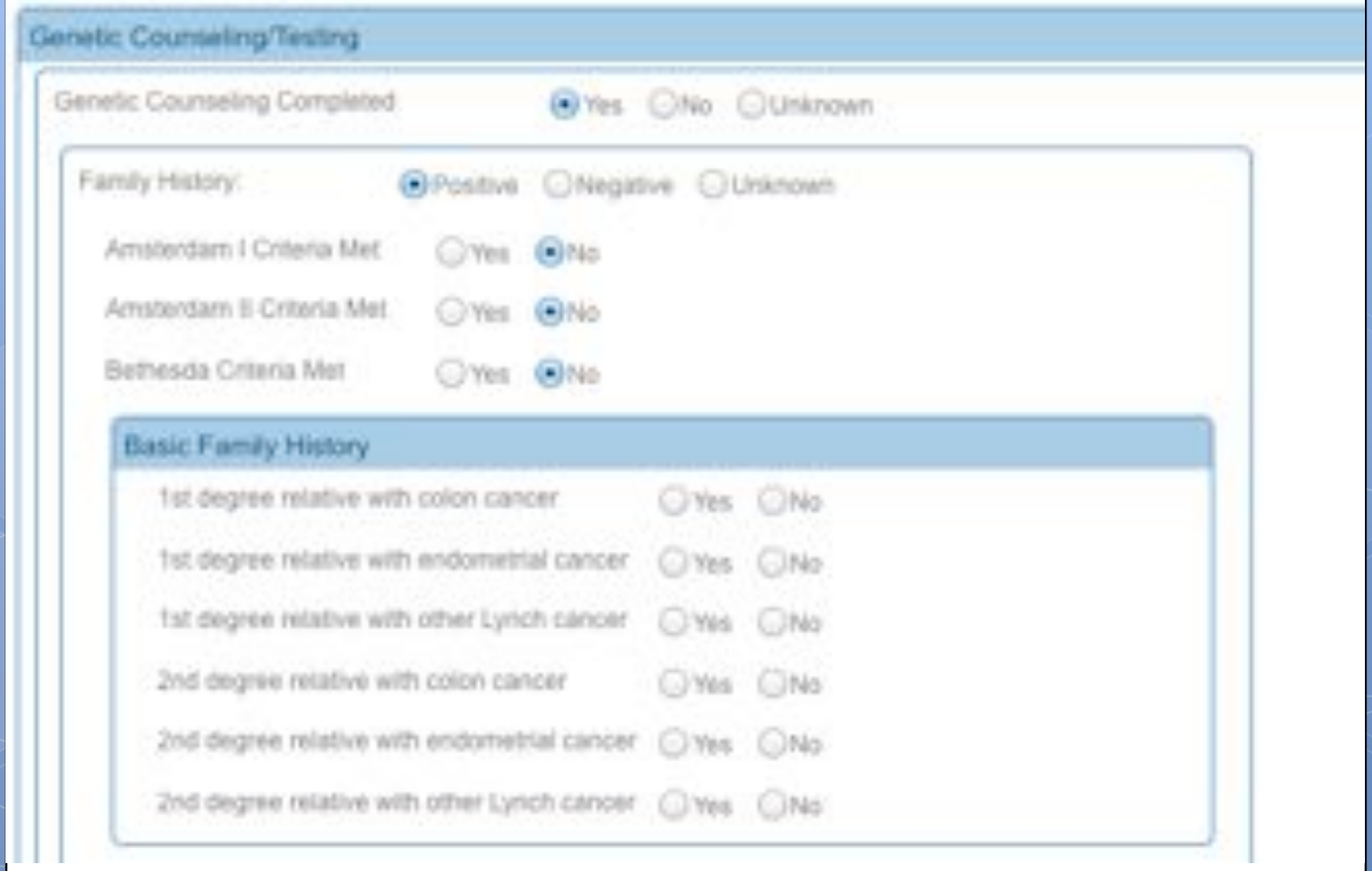
- Genetic Counseling – if no, reasons
- Family History – limited vs. extensive
- Genetic Testing Performed – germline and somatic, results (variant and pathogenicity)
  - If No - reasons
- If Pathogenic/likely pathogenic
  - Family members tested
  - Results
  - Screening recommendations

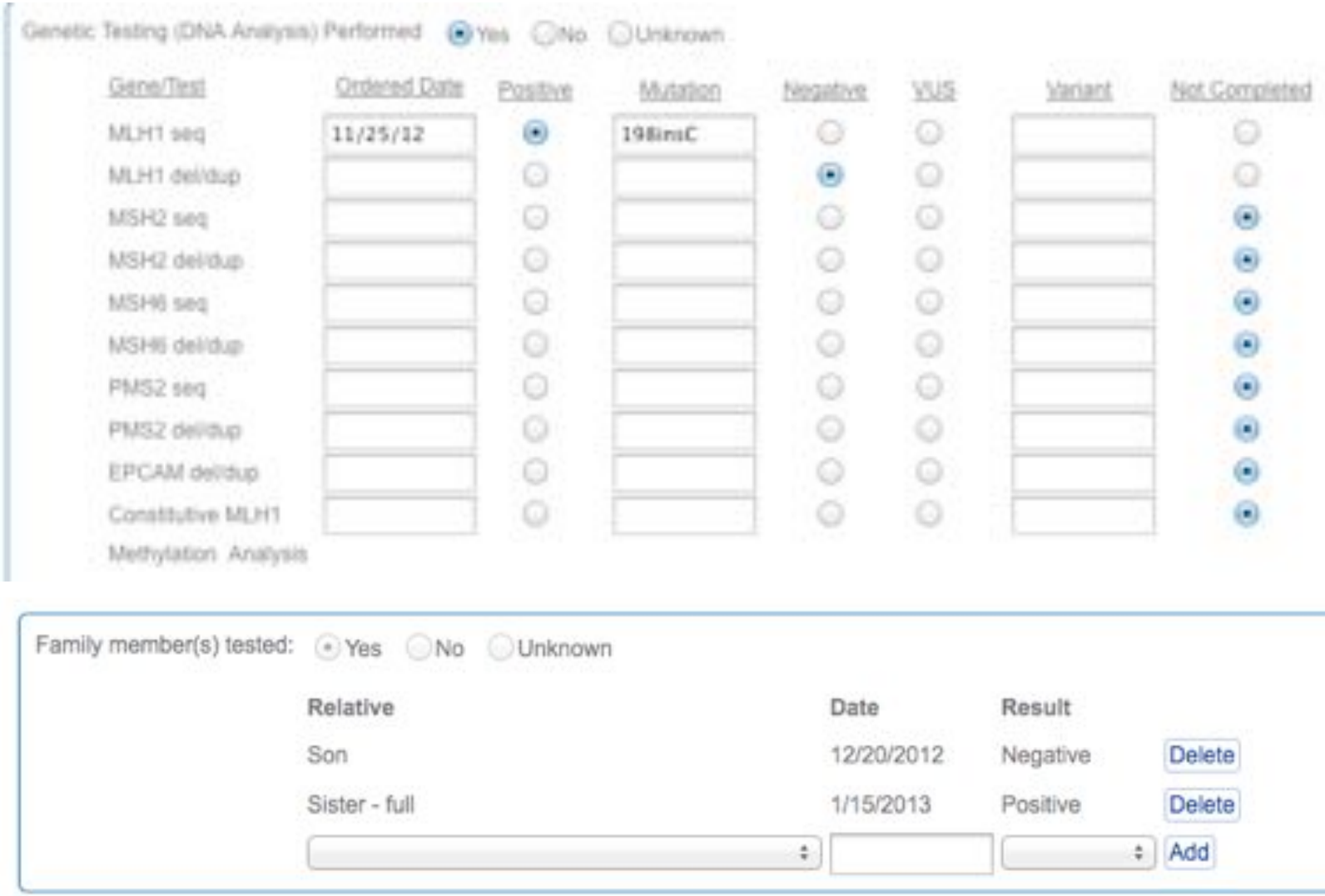
Database Features

- Participating institutions can assign data entry, data export, and site administrator privileges
- Access to aggregate data through LSSN research proposal and publication process
- Allows data dumping from existing LSSN member databases
- Use for primary LS Screening data collection
- Reporting tools


LSSN Database
Home
Data Entry
Reporting
Admin
Lynch Syndrome
Screening Network
Introduction
Welcome to the Lynch Syndrome Screening Network (LSSN) database. Your participation in this important project is greatly appreciated and will assist in fulfilling the LSSN vision of reducing the cancer burden associated with Lynch syndrome.
Please refer to the LSSN Database Manual for instructions and definitions to assist you in accurate data entry. Questions regarding the database or entry should be directed to Sarah Mange - MangeS@michigan.gov
Home
Data Entry
Reporting
Admin
Save
Exit
Delete
Demographics
Patient Code:
Emory_10_27_12_123456
Age at diagnosis:
55
(If over 85, enter 85 as age)
Location:
Emory
Sex:
Male
Hispanic/Latino:
No
Race 1:
Caucasian
Race 2:
Insurance 1:
Private
Insurance 2:
Previous or Synchronous Lynch Syndrome-related Cancer
Yes
No
Unknown
Age at diagnosis
Cancer type
50
Colon
Delete
51
Rectum
Delete
Add


Index Cancer
Type: Endometrial
Date: 10/27/2012
Specimen Type: Biopsy/D&C, tumor in lower uterine segment – Yes
Pathological Features Available: Yes No Unknown
TNM Staging T: 1 N: 0 M: 0 Staging Unknown:
Features:
Tumor infiltrating lymphocytes
Crohn's-like lymphocytic reaction
Signet Ring cell
Mucinous
Medullary growth pattern
None of the above features are noted
Endometrioid
Clear Cell
Papillary Serous
Mixed histology (endometrial)
Transitional Cell
Unusual Screening/Testing Situation Details:
Tumor Screening
MSI Testing: Yes No Attempted/failed
IHC Testing: Yes No Attempted/failed Date: 10/27/2012 (mm/dd/yy)
Results:
MLH1: Present Absent Indeterminate Not done
MSH2: Present Absent Indeterminate Not done
MSH6: Present Absent Indeterminate Not done
PMS2: Present Absent Indeterminate Not done
BRAF V600E Testing: Yes No Attempted/failed Date: 10/27/2012 (mm/dd/yy)
Result: Positive Negative
MLH1 Promoter Methylation Analysis: Yes No Attempted/failed Date: 11/04/2012 (mm/dd/yy)
Result: Positive Negative


Genetic Counseling/Testing
Genetic Counseling Completed
Yes
No
Unknown
Family History:
Positive
Negative
Unknown
Amsterdam I Criteria Met
Yes
No
Amsterdam II Criteria Met
Yes
No
Bethesda Criteria Met
Yes
No
Basic Family History
1st degree relative with colon cancer
Yes
No
1st degree relative with endometrial cancer
Yes
No
1st degree relative with other Lynch cancer
Yes
No
2nd degree relative with colon cancer
Yes
No
2nd degree relative with endometrial cancer
Yes
No
2nd degree relative with other Lynch cancer
Yes
No


Genetic Testing (DNA Analysis) Performed Yes No Unknown
Gene/Test
Ordered Date
Positive
Mutation
Negative
VUS
Variant
Not Completed
MLH1 seq
11/25/12
198insC
MLH1 del/dup
MSH2 seq
MSH2 del/dup
MSH6 seq
MSH6 del/dup
PMS2 seq
PMS2 del/dup
EPCAM del/dup
Constitutive MLH1
Methylation Analysis
Family member(s) tested: Yes No Unknown
Relative
Date
Result
Son
12/20/2012
Negative
Delete
Sister - full
1/15/2013
Positive
Delete
Add

Initial Research Questions

- Number of newly diagnosed CRC and EC screened and % positive by protocol
- Frequency of mutations in LS genes by: age of diagnosis, tumor characteristics, family history and screening protocols
- Average number of family members pursuing cascade testing

Status

- Database revisions completed
- Database under construction via OMNI
- Data use agreements – through OMNI
- IRB approval under review through UCSF
  - Linked and unlinked options for patient identifiers
- Data transfer template developed by UCSF
  - Several entities able/willing to dump large data sets – first step
- Finalization of data entry manual

Question?