HL7 Pedigree Model for Family History
Clinical Decision Support and Interoperability with EHRs

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HL7
In the age of the Human Genome Project

- We must identify patients at risk of hereditary disease and implement management strategies that will decrease morbidity and mortality
Adult hereditary syndromes ~188

- Benign 153
- Cancer 32
- Cancer plus benign 3

Clinical Decision Support

• Algorithms identify diagnosis/best treatment
• Visualizations help clinician better interpret data
• Visualizations help clinician take correct action
• Visualizations help patient make better choices
3 Levels of Clinical Decision Support
(Source: UK National Design Authority)

LEVEL 1
- **Information Support**
  Clinical user must navigate paper-based information & ascertain what content is relevant.

LEVEL 2
- **Action Rules**
  Clinical user supported by electronic data validation, clinical alerts & reminders.

LEVEL 3
- **Computer-Interpretable Guidelines**
  Clinical content is customized to individual patients or clinical trial subjects. Also referred to as integrated care pathways.
Clinical decision support is critical

- No PCP can
  - Identify all syndromes
  - Know best management for each

Patient entered data is critical

- Few PCPs
  - Have time to enter structured data
Functionality Lacking in EHR for FH

Interoperability
Patient data entry
Risk factors and demographics database
Complete family history database
Pedigree drawing
Risk algorithms
Letter generation
Stand alone programs to fill the void

My Family Health Portrait

CAGENE

My Generations

HughesRiskApps

Progeny

Jameslink

Family Healthware
2 Programs, 1 Translator

My Family
Health Portrait

HughesRiskApps
HL7 provides a standard intermediary

My Family
Health Portrait

HL7 Message

HughesRiskApps
Clinical Decision Support

Knowledge Base

Algorithms

HL7
Monolithic Approach
Vendor creates CDS from scratch

Vendor 1
Knowledge Base
Algorithms

Family History CDS

Electronic Health Record

My Family Link
Health Portrait
My Generations
Hughes Risk Apps
CAGENE
Monolithic Approach

150 Vendors create 150 different CDS approaches to the same problem.
... developing a **modular family history tool**, where collection of family health history is performed within the EHR, followed by **messaging of this information to a variety of richer family history tools that perform risk analyses**

The enhanced family history and results of these algorithmic calculations could then be returned **to the EHR**, allowing for ongoing **curation**
Clinical Decision Support

Knowledge Base

Algorithms

HL7

Vendor 1

Electronic Health Record

Vendor 2

Electronic Health Record

Vendor 3

Electronic Health Record
<table>
<thead>
<tr>
<th>Name</th>
<th>Relationship</th>
<th>Bloodline</th>
<th>Age</th>
<th>Status</th>
<th>Diseases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bill Harvard</td>
<td>Father</td>
<td>Paternal</td>
<td>67</td>
<td>Alive</td>
<td></td>
</tr>
<tr>
<td>Jack Harvard</td>
<td>Grandfather</td>
<td>Paternal</td>
<td>70</td>
<td>Dead</td>
<td></td>
</tr>
<tr>
<td>Lily Harvard</td>
<td>Grandmother</td>
<td>Paternal</td>
<td>72</td>
<td>Dead</td>
<td></td>
</tr>
<tr>
<td>Rick Harvard</td>
<td>Uncle</td>
<td>Paternal</td>
<td>62</td>
<td>Alive</td>
<td>Melanoma 45</td>
</tr>
<tr>
<td>Eric Harvard</td>
<td>Uncle</td>
<td>Paternal</td>
<td>60</td>
<td>Alive</td>
<td></td>
</tr>
<tr>
<td>Sue Doe</td>
<td>Aunt</td>
<td>Paternal</td>
<td>59</td>
<td>Alive</td>
<td>Breast Cancer 45; Ovarian Cancer 45</td>
</tr>
<tr>
<td>Amy Smith</td>
<td>Aunt</td>
<td>Paternal</td>
<td>47</td>
<td>Dead</td>
<td>Breast Cancer 35; Ovarian Cancer 45</td>
</tr>
<tr>
<td>Annie Harvard Proband</td>
<td>Self</td>
<td></td>
<td>37</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Janet Harvard</td>
<td>Mother</td>
<td></td>
<td>62</td>
<td></td>
<td>Uterine Cancer 50</td>
</tr>
<tr>
<td>David Jones</td>
<td>Grandfather</td>
<td>Maternal</td>
<td>82</td>
<td>Dead</td>
<td>Colon Cancer 61</td>
</tr>
<tr>
<td>Helen Jones</td>
<td>Grandmother</td>
<td>Maternal</td>
<td>85</td>
<td>Dead</td>
<td></td>
</tr>
</tbody>
</table>
Patient Name: Annie Harvard Proband

Unit Number: 99999990
Date Of Birth: 01/11/1971

Genetic Testing
Guideline: Consider testing a relative
Clinician's Recommendation: Consider testing a relative
Patient's Preference: agrees with recommendation

Synthesis of Mutation Risk:
- BRCAPRO: 18%
- Myriad: 12.2%

Pedigree
- Jack 70
- Lily 72
- Eric 60
- Rick 62
- Br 35
- Mel 45
- Amy 47
- Sue 59
- Br 45
- Bill 67
- Jane 62
- End 50
- Anni 37
- Davi 82
- Col 61
- Hele 85

Redraw

Exit
< Back
Next >
### Genetic Testing

**Guideline**
- Consider testing a relative

**Clinician's Recommendation**
- Consider testing a relative

**Patient's Preference**
- agrees with recommendation

### Synthesis of Mutation Risk

- **BRCAPRO**: 18%
- **Myriad**: 12.2%

### Patient's History

<table>
<thead>
<tr>
<th>Patient's History</th>
<th>No breast cancer or ovarian cancer, in any relative</th>
<th>Breast cancer &lt;50 in one relative; no ovarian cancer in any relative</th>
<th>Breast cancer &lt;50 in more than one relative; no ovarian cancer in any relative</th>
<th>Ovarian cancer at any age in one relative; no breast cancer &lt;50 in any relative</th>
<th>Ovarian cancer in more than one relative; no breast cancer &lt;50 in any relative</th>
<th>Breast cancer &lt;50 and ovarian cancer at any age</th>
</tr>
</thead>
<tbody>
<tr>
<td>No breast cancer or ovarian cancer, in any relative</td>
<td>2.8%</td>
<td>4.5%</td>
<td>8.7%</td>
<td>5.6%</td>
<td>9.6%</td>
<td>12.2%</td>
</tr>
<tr>
<td>Breast cancer ≥ 50</td>
<td>2.9%</td>
<td>5.3%</td>
<td>11.4%</td>
<td>6.4%</td>
<td>12.2%</td>
<td>15.9%</td>
</tr>
<tr>
<td>Breast cancer &lt;50</td>
<td>6.8%</td>
<td>15.8%</td>
<td>30.1%</td>
<td>16.9%</td>
<td>27.3%</td>
<td>39.2%</td>
</tr>
<tr>
<td>Male breast cancer</td>
<td>12.8%</td>
<td>21.8%</td>
<td>41.9%</td>
<td>20.0%</td>
<td>40.0%</td>
<td>61.9%</td>
</tr>
<tr>
<td>Ovarian cancer at any age, no breast cancer</td>
<td>8.8%</td>
<td>23.1%</td>
<td>42.3%</td>
<td>21.1%</td>
<td>33.2%</td>
<td>48.5%</td>
</tr>
<tr>
<td>Breast cancer ≥50 and ovarian cancer at any age</td>
<td>17.6%</td>
<td>26.1%</td>
<td>46.2%</td>
<td>30.3%</td>
<td>46.2%</td>
<td>60.0%</td>
</tr>
<tr>
<td>Breast cancer &lt;50 and ovarian cancer at any age</td>
<td>39.1%</td>
<td>53.9%</td>
<td>67.2%</td>
<td>66.0%</td>
<td>70.8%</td>
<td>79.0%</td>
</tr>
</tbody>
</table>
Risk of Mutation in Major Mismatch Repair Genes

<table>
<thead>
<tr>
<th></th>
<th>Any MMR Gene</th>
<th>MLH1</th>
<th>MSH2</th>
<th>MSH6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Probability</td>
<td>13.6%</td>
<td>6.1%</td>
<td>6.9%</td>
<td>0.6%</td>
</tr>
</tbody>
</table>

Risk of Colon Cancer

- 5-Year Risk: 0.2%
- Lifetime Risk: 7.5%

Risk of Endometrial Cancer

- 5-Year Risk: 0.6%
- Lifetime Risk: 6.9%