Background

• Endometriosis (endo) is a chronic, reproductive disease in ~10% of childbearing-age women
• The disease is highly enigmatic
• Characterized through surgical findings with menstrual pain and infertility as primary symptoms
• Research gaps in endometriosis characterization
• Incomplete characterization of signs, symptoms, and comorbidities
• Lack of a well-validated phenotype definition of the disease

Methods

1. Define & Validate Endometriosis Phenotype for Electronic Health Record (EHR) Databases

• Four phenotype definitions implemented on ATLAS (available on ohdsi.org)

<table>
<thead>
<tr>
<th>Cohort</th>
<th>Initial Event</th>
<th>Inclusion Rules</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>endo-related procedures AND endo diagnosis OR endo-prevalent procedures AND endo diagnosis</td>
<td>Females ages 15-49</td>
</tr>
<tr>
<td>B</td>
<td>endo-related procedures AND endo diagnosis OR endo-prevalent procedures AND endo diagnosis</td>
<td>Females ages 15-49 AND 2 endo diagnosis after index date</td>
</tr>
<tr>
<td>C</td>
<td>endo-related procedures AND endo diagnosis AND endo-related imaging procedures AND endo diagnosis before index date</td>
<td>Females ages 15-49</td>
</tr>
<tr>
<td>D</td>
<td>endo-related procedures AND endo diagnosis AND endo-related imaging procedures AND endo diagnosis before index date</td>
<td>Females ages 15-49 AND 2 endo diagnosis after index date</td>
</tr>
</tbody>
</table>

• Relevant concept sets
  • Endo diagnosis: endometriosis diagnosis codes (including adenomyosis)
  • Endo-related procedures: guideline-based procedures for endometriosis diagnosis and treatment (e.g., pelvic laparoscopy)
  • Endo-prevalent procedures: procedures present >50% of patients with ≥1 endo diagnosis
  • Endo-related imaging procedures: guidelines-based imaging procedures (e.g. pelvic MRI)

• Validation
  • Patient records from all cohort definitions with index data between 1/1/2016 and 6/1/2018
  • Manual chart review from clinical experts: confirmed endometriosis diagnosis through histological analysis as reported in post-surgery pathology report at index time

2. Characterize Endometriosis Cohorts Across Different Types of Observational Health Databases (EHR, Claims)

• Using cohort with best precision/recall on Columbia EHR, characterized using ATLAS
• On four databases: Columbia EHR (Columbia EHR), Optum® Clinformatics® Extended DataMart (Optum), IBM MarketScan® Commercial Database (MCD), IBM MarketScan® Multi-State Medicaid Database (MMMD); All databases except Columbia EHR de-identified
• Report prevalent (>10%) conditions and drugs across databases

Results

• 1,406 patients over all phenotype definitions reviewed by clinical experts
• Cohort B had highest precision/recall and negative predictive value 0.84

Results (Continued)

<table>
<thead>
<tr>
<th>Cohort name on OHDSI.org</th>
<th>Database</th>
<th>Total records in cohort</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cohort D</td>
<td>Columbia EHR</td>
<td>1,248</td>
</tr>
<tr>
<td>Cohort C</td>
<td>Optum</td>
<td>24,725</td>
</tr>
<tr>
<td>Cohort B</td>
<td>MMMD</td>
<td>54,609</td>
</tr>
<tr>
<td>Cohort A</td>
<td>MCD</td>
<td>398,015</td>
</tr>
</tbody>
</table>

• Each of these cohorts was reviewed by clinical experts.
• * all other cohorts are a subset of this cohort so recall=1

Conclusions

• First study to develop a well-validated endometriosis phenotype based on EHR data
• Findings across databases consistent with what we know about the disease
• Patients experience pelvic pain and take a number of pain medications
• Most prevalent drugs known to be endometriosis treatments
• Conditions are known comorbidities or symptoms
• Results also point to new knowledge about the disease
• Recent literature shows link between endometriosis and migraines, coronary heart diseases