Quantifying the Burden of Rare Diseases in the Duke University Health System (DUHS)
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• To what extent do patients with rare diseases burden the DUHS?
• Do different types of rare diseases disproportionately burden the DUHS?
Data and methods

• Visits Data (DUHS 2012 – 2016)
• ~30 million rows, ~1.2 million patients
• Partitioned data set into patients with at least one rare disease ICD code and patients without any rare ICD code
  • 6627 Rare Diseases
  • 2913 Map to ICD-10
  • 2706 in the Duke Health System
• Compared the two cohorts
The Problem

• Poor specificity of ICD codes prevents an easy partition of rare vs. non-rare patients

• For example, our most common rare disease is Type 2 Diabetes because the rare disease Maturity Onset Diabetes of Youth (MODY) maps to the same ICD-10 code as ordinary Type 2 Diabetes.

• Future work includes:
  ▪ Repartitioning the data with the help of experts by throwing out non-specific ICD codes to find a lower bound burden
  ▪ Drilling down into ICD hierarchy to find which groups of rare diseases are responsible for more of the burden
    • Focus on more precise chapters and blocks
  ▪ Combining medication data with ICD codes to target specific rare diseases
  ▪ Using our methodology to explore the burden of undiagnosed patients on the DUHS