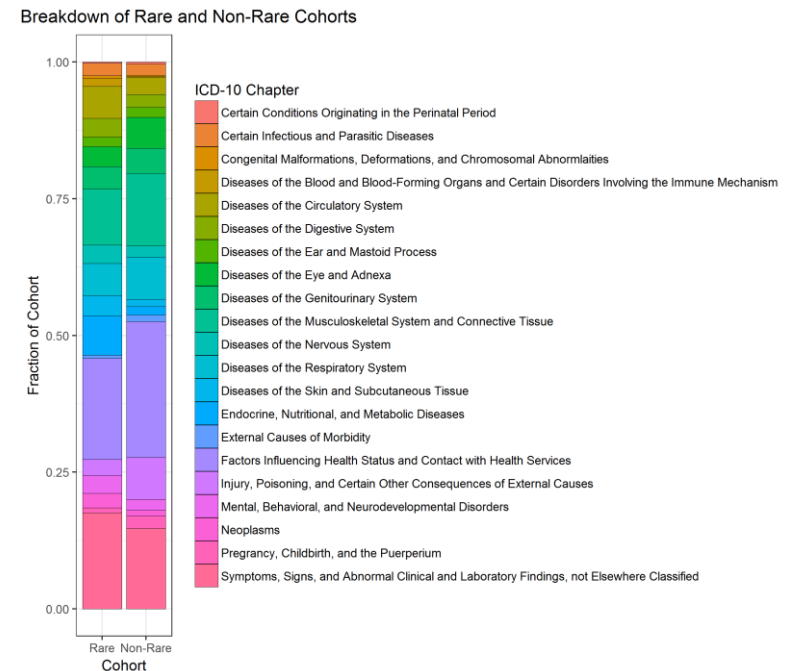


Quantifying the Burden of Rare Diseases in the Duke University Health System (DUHS)

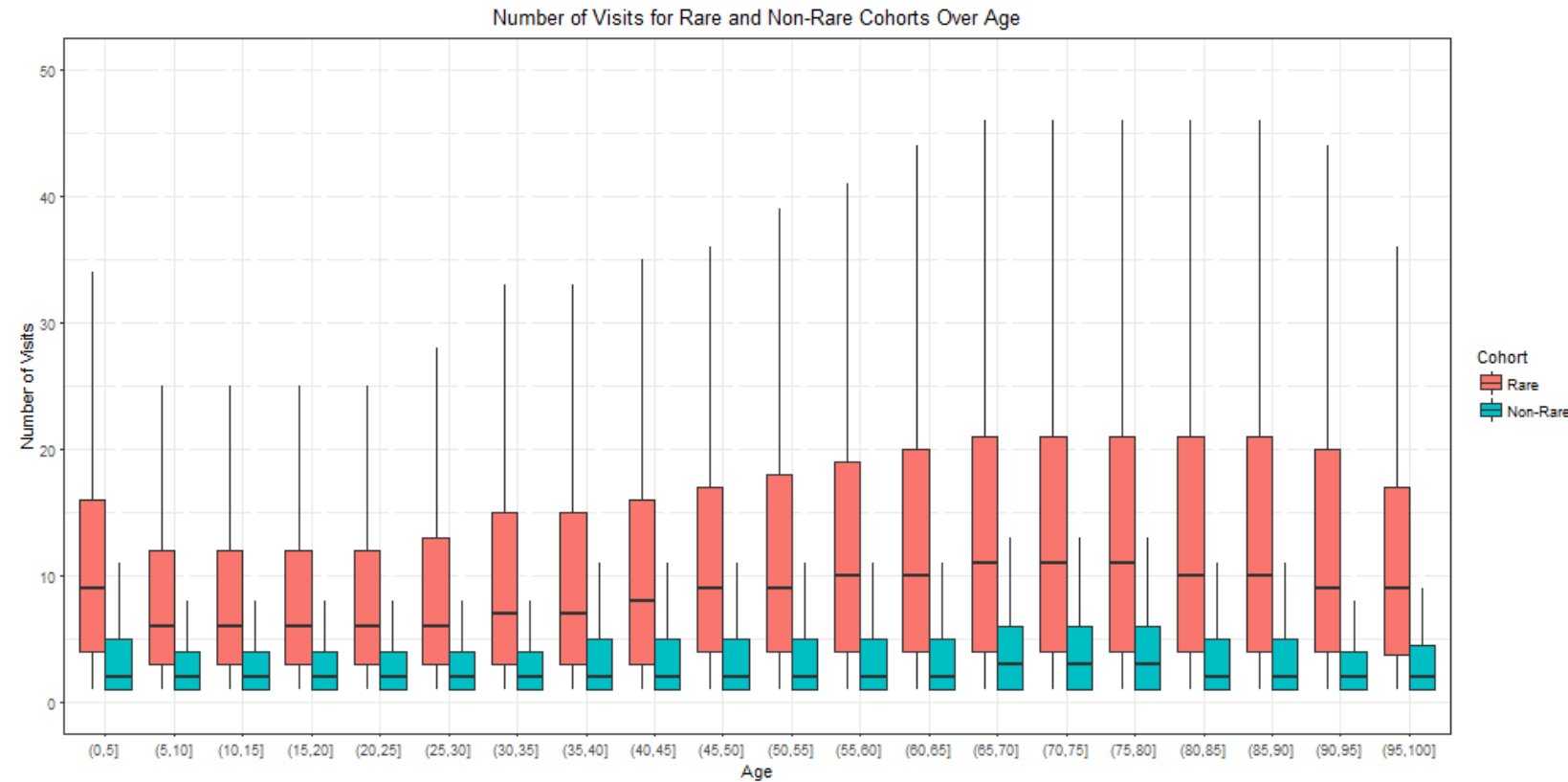
Gary Koplik, Isaac Lavine, Rachel Richesson, Matt Tribby

- 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

