# Physician Focused Genetic Visualizations

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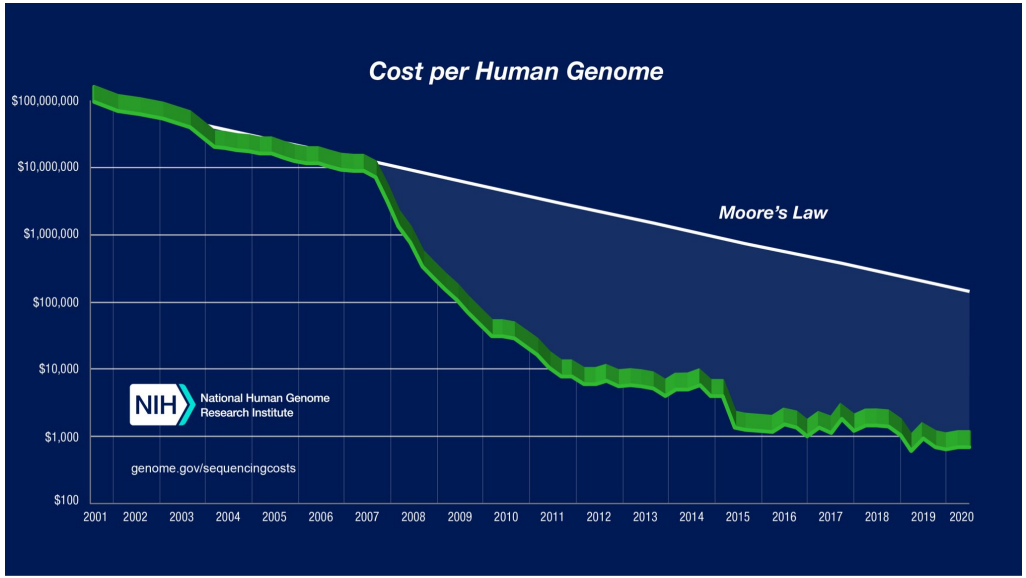
# Background

- Whole genome sequencing costs are dropping
  - Precision medicine will potentially become more widely available
- Existing systems are either proprietary or focused on researchers
- The genome is huge, requires specific knowledge to find useful information



# Design Principles

- Uncertainty
  - Having a mutation associated with a disease does **NOT** always mean you are guaranteed to have the disease
  - Not currently attempting to represent uncertainty
- User Behavior
  - Not adapting to user behavior
  - Expect user behavior is more important downstream of our system
- Top-Down Approach
  - Guide the user by attempting to pull out key insights up front

# Data

- Downloaded variant data for one family (~3 million variants per individual)
  - Courtesy of the Genome in a Bottle project
- Annotated with the Variant Effect Predictor (VEP) dataset
  - Provides information about the variants
  - Gene names, frequency of the variant, type of mutation, etc.
- Determined which variants likely affected the protein products
- Annotated with the DisGeNET dataset
  - Provides gene-to-disease mappings
- Manually added descriptions of diseases and severity rankings
- Ended with ~400 variants per individual

# Demo

# Limitations and Future Work

- More disease data, e.g. population level data showing outcomes
- Addressing uncertainty
- Family history
- Electronic health record (EHR) integrations
- User testing and expert opinions

Questions?