

# Researching, Recognizing, and Reaching the Rare

## Case Study on Global Patient Finding in Rare Disease

### CASE BACKGROUND



Condition is a rare disease that is **underdiagnosed** and **underdocumented** because of lack of ICD-10 code



**Heterogeneity in phenotype** makes it hard to establish clear guidelines on how to identify disease and target specialty



**Genetic testing needed** for diagnosis, but even if mutation is confirmed, it has limited impact on patient care

### Most Common Leakage Points in the Journey to Diagnosis and Treatment Include

#### HIDDEN DISEASE

HCPs/patients/caregivers do not recognize early symptoms as cause for concern

#### NO REFERRAL

Physician does not refer patient for further workup because of mild or unspecific symptoms

#### NO GENETIC TEST

HCP does not order genetic test to confirm underlying cause

#### INCONCLUSIVE TEST

Genetic test result comes back as a variant of uncertain significance

#### UNACTIONABLE DIAGNOSIS

There is currently no treatment beyond solid organ transplant

### EXAMPLE PORTFOLIO OF PROJECTS TO SUPPORT PATIENT IDENTIFICATION

FIND ALREADY DIAGNOSED PATIENTS

#### ICD-10 CODE SUPPORT

- Develop proposal to be submitted to the ICD-10-CM Coordination and Maintenance Committee
- Coordinate with KOLs and patient groups to ensure support for submission

#### PATIENT SURVEILLANCE SURVEY

- Use quantitative research with HCPs to estimate and confirm number of diagnosed patients in the US/EU
- Use broad approach to look for patients outside known expert centers based on claims data analytics

#### PATIENT FINDING ANALYTICS

- Develop patient profiles leveraging predictive analytics to create a model for identifying undiagnosed patients
- Leverage outputs from primary research to further train/refine model to maximize number of "true positives"

#### GENETIC TESTING LAB STRATEGY

- Aggregate genetic prevalence of mutations by surveying top genetic testing facilities
- Identify opportunities for partnerships with labs

#### ADVISORY BOARD & DISEASE EDUCATION WHITE PAPER

- Coordinate and facilitate meeting with top KOLs to build consensus about disease diagnostic process
- Develop consensus document (eg, white paper/publication)

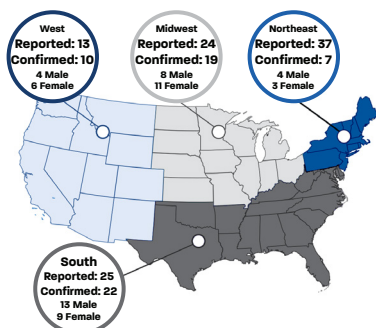
#### PATIENT JOURNEY

- Use qualitative research to map patient journey and identify pain points from a patient perspective
- Identify leakage points and develop recommendation

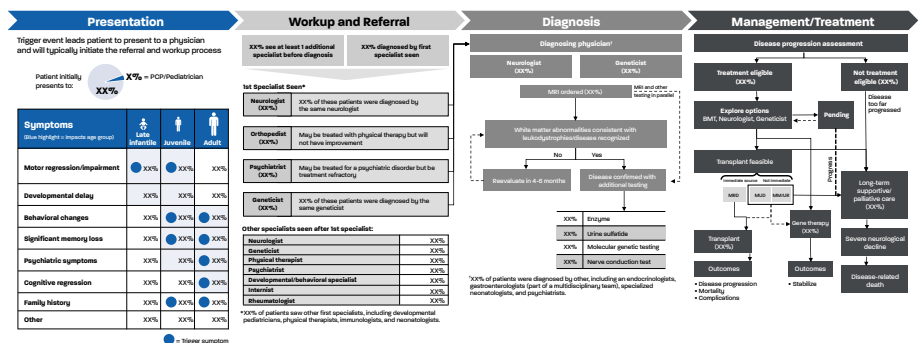
GROW NUMBER OF DIAGNOSED PATIENTS

### EXAMPLE OUTPUTS

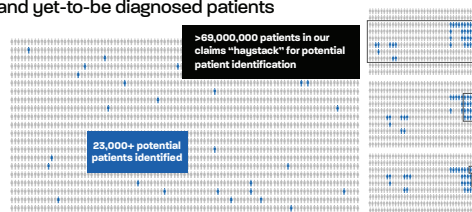
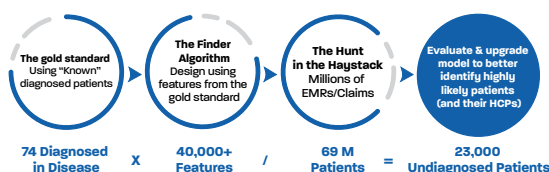
#### 1 Treatment center database and patient heat map



#### 2 Detailed map of patient journey



#### 3 Claims-based AI modeling to develop target lists with diagnosed and yet-to-be diagnosed patients



- A Perfect Recall; Low Precision
- B Balanced Recall and Precision
- C Low Recall; Perfect Precision

### APPLICATIONS

- A. Medical affairs: publish patient finder outcome
- B. Precommercial: plan strategy and tactics
- C. Clinical development: drive clinical trial enrollment by identifying relevant providers and patient clusters