81 qd Magnolia innovation

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

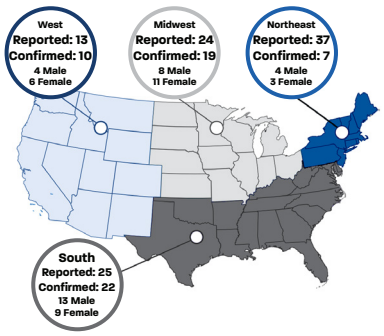
ADVISORY BOARD & DISEASE EDUCATION WHITE PAPER
- Coordinate and facilitate meeting with top KOLs to build consensus about disease diagnostic process
- Develop consensus document (eg, white paper/publication)

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

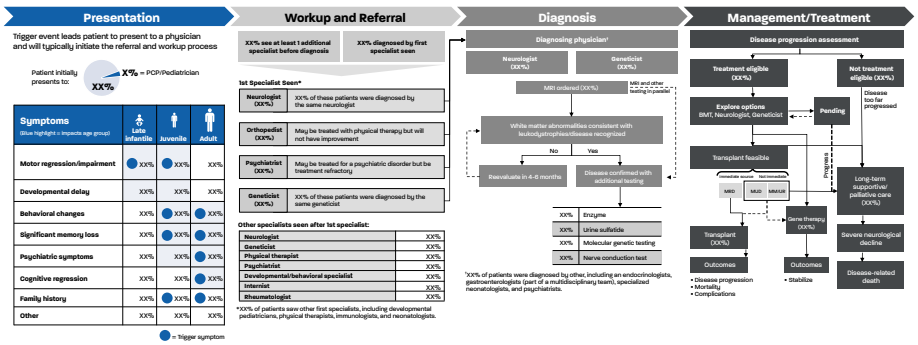
GROW NUMBER OF DIAGNOSED PATIENTS

EXAMPLE OUTPUTS

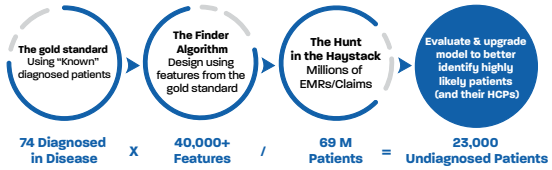
1 Treatment center database and patient heat map

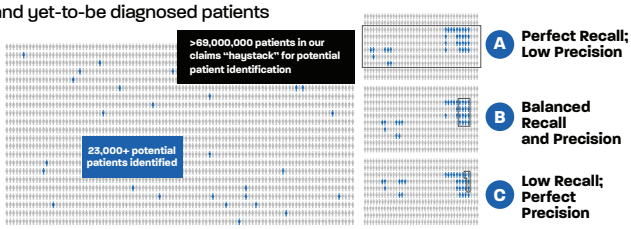


2 Detailed map of patient journey



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





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

AI, artificial intelligence; BMT, bone marrow transplant; EMR, electronic medical record; EU, European Union; HCP, health care professional; ICD-10, *International Classification of Diseases, Tenth Revision*; ICD-10-CM, ICD-10 Clinical Modification; KOL, key opinion leader; M, million; MM, multiple myeloma; MRD, minimal residual disease; MRI, magnetic resonance imaging; MUD, matched unrelated donor; PCP, primary care physician; UR, urgent revascularization; US, United States.